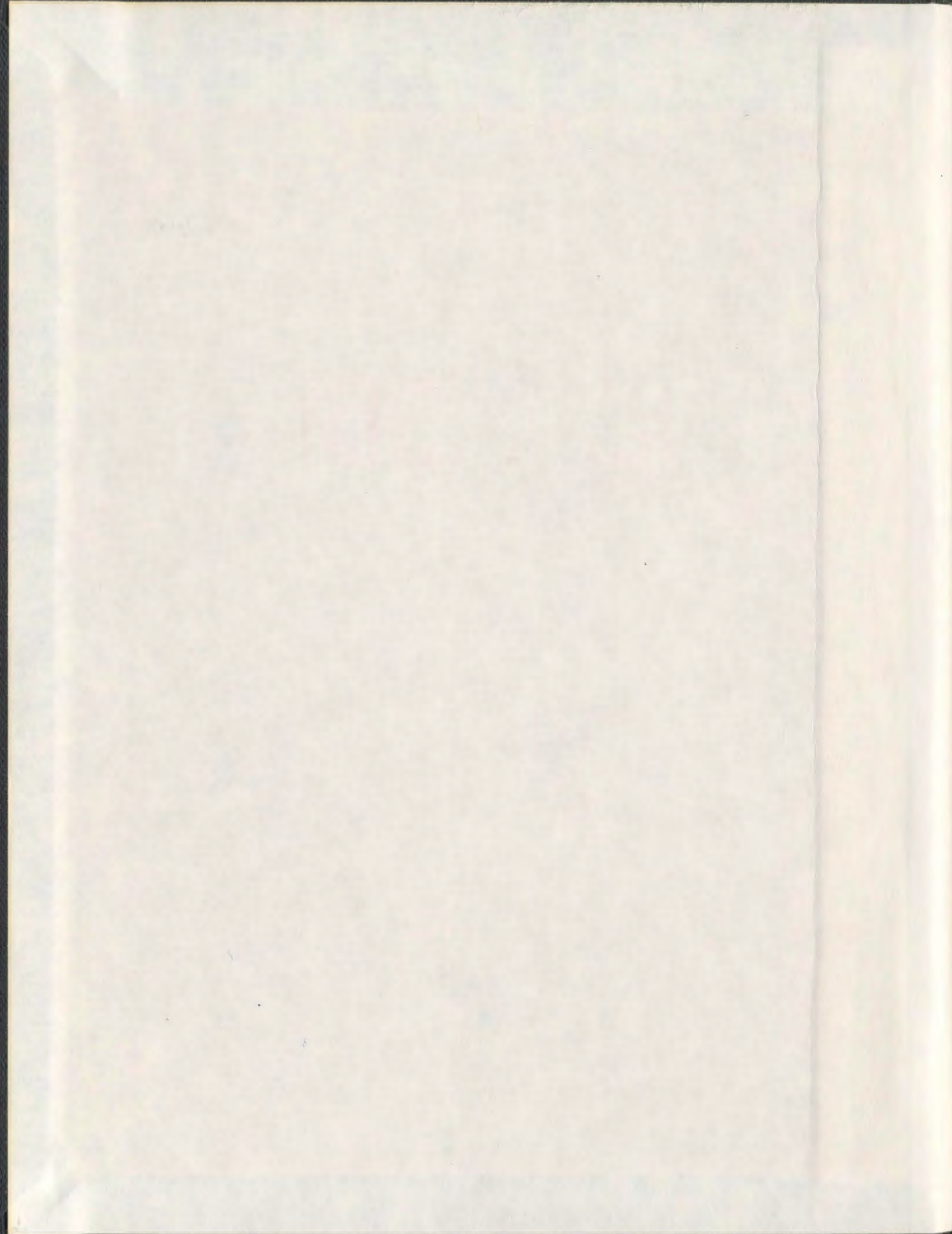


CONSTRUCTING THE MEANING OF BEING AT-RISK:
THE EXPERIENCES OF INDIVIDUALS LIVING IN
FAMILIES AT RISK FOR ARRHYTHMOGENIC RIGHT
VENTRICULAR CARDIOMYOPATHY IN THE
PROVINCE OF NEWFOUNDLAND AND LABRADOR

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**CONSTRUCTING THE MEANING OF BEING AT-RISK: THE EXPERIENCES
OF INDIVIDUALS LIVING IN FAMILIES AT RISK FOR ARRHYTHMOGENIC
RIGHT VENTRICULAR CARDIOMYOPATHY IN THE PROVINCE OF
NEWFOUNDLAND AND LABRADOR**

by

© April D. Manuel

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ABSTRACT

The past decade has seen a proliferation of available predictive genetic tests. These are the result of one of the most significant scientific advances of the 21st century: the Human Genome Project. Social scientists have examined how the availability of genetic testing shapes the lived experience of at-risk people as well as subsequent health decision-making. Little attention has been paid to how the embodiment of risk is (re)shaped in light of changing genetic technologies or how experience of risk may in turn shape the development of genetics. A grounded theory approach was used to gain a fuller understanding of how 29 individuals living in a family at risk for Arrhythmogenic Right Ventricular Cardiomyopathy, in the province of Newfoundland and Labrador, construct the meaning of being at-risk prior to, during, and following genetic testing in relation to the various stages of gene discovery and test availability. Three phases of constructing meaning were identified: (1) Awakening to a New Meaning of Being At-Risk, (2) Deciphering the Meaning of Being At-Risk, and (3) Embodying a New Meaning of Being At-Risk. This study found that at-risk individuals' understandings of the meaning of being at-risk both shapes and is shaped by the "lived experience" of the genetic testing process and also impacts (and is impacted by) health care decisions. The meaning assigned to being at-risk is *pragmatic*, *transient*, and *fluid*. It is *pragmatic* in that the participants juxtapose three types of **contextual dimensions** (scientific knowledge, experiential knowledge, and phase of the genetic testing process) against the existing **conditions**, or specific factors that influence risk perception, as they assign meaning to being at-risk and make decisions, a process that is ongoing throughout the genetic testing process. The meaning assigned to risk is *transient*, in that as one's risk perception

fluctuates so do the contextual dimensions and conditions that influence participants' choices. It is *fluid*, in that the meaning of being at-risk is shaped and reshaped (and the decisions change), with each new experience and coincident with the particular stage of gene discovery. These findings lead to recommendations for genetic service providers, health policy makers, and genetic scientists on best practices for health care in the context of novel gene discovery.

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Table of Contents

Abstract	ii
Acknowledgements	iv
Table of Contents	v
List of Tables	x
List of Figures	xi
List of Abbreviations	xii
List of Appendices	xiii
 Chapter 1: Introduction	14
Purpose of the Study	16
What is ARVC?	16
Prevalence of ARVC	18
Diagnosis of ARVC	19
Symptoms and Management of ARVC	20
History of ARVC	21
History of ARVC Gene Discovery in NL	22
Overview of Chapter Contents	25
 Chapter 2: Theoretical Approaches to Understanding Risk	29
Cultural Theory and Risk	30
Cognitive (Psychological) Theory and Risk	32
Sociological Theory and Risk	34
Governmentality and Risk	37
Chapter Summary	39
 Chapter 3: The Construction of Risk	41
Rational for Literature Review	42
Shaping the Meaning of Risk: Risk Assessment	44
How “Accurate” is Risk Perception	45
Representations of Risk: Laypersons and Experts	49
Laypersons’ Construction of Risk	50
Who are the “Experts”?	55
Expert and Lay Knowledge of Genetics	56
Laypersons, Experts and Technology: Who Trusts Whom?	58
Cardiovascular Literature: Being At-Risk for a Genetically Linked Cardiovascular Disease	59
The Coronary Candidate	60
Using Biomedical Knowledge to Construct Risk	63
Predictive Genetic Testing Decision-Making Process	68
Why Do People Participate in Predictive Genetic Testing?	69
Why Do People Refuse to Participate in Predictive Genetic Testing?	71
The Decision-Making Process in Predictive Genetic Testing	73

Risk Communication	79
Health Care Providers and Risk Communication	80
Family and Risk Communication	84
How Does Risk Perception Influence Behaviour?	87
Risk Perception and Behavioural Responses	88
Hypervigilant and Hypovigilant Behaviours	91
Cardiovascular Literature.....	93
Chapter Summary	94
Chapter 4: The Psychosocial Aspects of Predictive Genetic Testing	97
Psychological Distress: Quantitative Literature	98
Psychological Distress:	99
Breast Cancer	100
Huntington Disease	101
Colorectal Cancer.....	103
Factors Influencing Psychological Distress	104
Baseline Level of Psychological Distress	104
Inconclusive Predictive Genetic Testing.....	105
Demographics and Psychosocial Factors.....	106
Genetically Linked Cardiovascular Disease	107
Individuals' Experiences with Predictive Genetic Testing: Qualitative Literature:	108
Genetically Linked Conditions	108
Cardiovascular Disease and Genetics	111
Psychosocial Impact of Living with LQTS.....	111
Psychosocial Impact of Predictive Genetic Testing on Family	112
Young Children, Young Adolescents, Parents, and Partners.....	113
Young Adolescents and Parents.....	113
Young Children and Parents	116
Family Members and Partners	120
Being a Partner or Spouse of an Individual at Risk for a Genetically Linked Condition.....	120
Chapter Summary	121
Chapter 5: Methodology.....	124
Grounded Theory	124
Symbolic Interactionism	126
Pragmatism	127
Situation of the Researcher and the Centrality of Postpositivism.....	127
Data Collection	130
The Sample	131
Recruitment of Participants.....	135
Interview Process	139
Data Analysis	142
Constant Comparative Method.....	142
Coding.....	145

Substantive Coding	147
Theoretical Coding.....	150
Grounded Theory Research Strategies.....	151
Theoretical Memoing	151
Diagramming	154
Theoretical Sensitivity	155
Rigour in Grounded Theory.....	156
Fit	156
Work	158
Relevancy.....	160
Modifiability	160
Ethical Considerations.	161
Chapter Summary	164

Chapter 6: Construct 1: Findings and Discussion: Awakening to a New Meaning of Being At-Risk165

The Rubik's Cube: The Puzzle and Shifting Faces of Risk.....	167
Construct 1: Awakening to a New Meaning of Being At-Risk	172
Category 1: Making Sense of Numerous Losses	173
Property 1: Living in a Family Familiar with Loss.....	173
Condition One: Loss	174
Property 2: The Struggle to Understand the Meaning of Being At-Risk for Oneself and Others	178
Conditions 1 and 2: Age and Gender	180
Condition 3: Physical Signs and Symptoms	181
Condition 4: Family History	184
Condition 5: Modifiable Lifestyle Factors.....	187
Condition 6: Concern for Children	189
Category 2: Struggling to Break the Cycle of Uncertainty	190
Property 1: Making Sense of and Living Through Early Clinical Investigations and Prescribed Treatments.....	191
Condition 1: Experts' Knowledge and Care	191
Condition 2: Available Technology.....	195
Condition 3: At-Risk Relative	198
Property 2: Acknowledging a Possible Genetic Origin to Risk	199
Condition 1: Interactions with Experts	199
Condition 2: Family History	200
Chapter Summary	201

Chapter 7: Construct 2: Deciphering the Meaning of Being At-Risk.....204

Category 1: Taking the First Steps of the Genetic Testing Process.....	206
Property 1: Being Offered a Predictive Genetic Test	206
Condition 1: Availability of Predictive Genetic Test	206
Condition 2: Interactions with Health Care Providers	211
Property 2: Making the Decision to Participate in Predictive Genetic Testing	213

Condition 1: Availability of Predictive Genetic Test	218
Condition 2: Numerous Losses	221
Condition 3: Physical Signs and Symptoms	222
Condition 4: Gender	224
Condition 5: Test Relevancy.....	225
Condition 6: Relational Responsibility	227
Condition 7: Family Support	231
Category 2: Building One's Risk Portfolio.....	233
Property 1: Waiting for Predictive Genetic Test Results.....	233
Condition 1: Waiting Period	233
Condition 2: Coping Mechanism	236
Avoidance	236
Gathering Information	236
Surveillance of the Body.....	237
Modifying Lifestyle Factors	237
Initiating an At-Risk Event	238
Property 2: Receiving Genetic Test Results	239
Condition 1: Genetic Test Results	239
Condition 2: Mode of Receiving Result	242
Chapter Summary	243
Chapter 8: Construct 3: Embodying a New Meaning of Being At-Risk.....	247
Category1: Adjusting to Living with or without a Genetic Condition	248
Property 1: Accepting and Assigning Meaning to Treatment Regimes	248
Conditions 1 and 2: Genetic Test and Physical Symptoms	249
Condition 3: Believed Efficacy of Prescribed Treatments: Pharmacological Management.....	251
Condition 4: Believed Efficacy of Prescribed Treatments: ICD Management....	255
Positive Aspects of Having an ICD	256
Negatives of Having an ICD.....	259
Accepting ICD and Anticipating First Shock	260
Understanding Why ICD Fired	262
Preventing ICD from Firing.....	263
Modifiable Factors	264
ICD Function	265
Body Image	266
Impact on Spouse.....	268
Property 2: Questioning the Accuracy of the Predictive Genetic Test	268
Condition 1: News of a More Definitive Test	269
Condition 2: Discrepancies in ARVC Management	271
Category 2: Recognizing the Reality of Living in a Family at Risk for a Genetic Disease	273
Property 1: Facing Everyday Worries and Challenges	273
Condition 1: Living with Restrictions Imposed on Oneself and Others.....	273
Modifiable Lifestyle Factors.....	274

Driving	276
Social Restrictions.....	278
Employment and Educational Opportunities	279
Condition 2: Being a Spouse.....	279
Property 2: Coping with Barriers to Resources	281
Condition 1: Human Resources	281
Condition 2: Physical Resources.....	286
Condition 3: Financial Resources	286
Overcoming Barriers.....	288
Condition 4: Informal Supports	290
Category 3: Looking Towards the Future Realities of Living in a Family at Risk for a Genetic Disease.....	291
Property 1: Trying to Maintain a Positive Outlook	292
Condition 1: Predictive Genetic Testing: Positive Aspects	292
Being Able to Focus on the Good Quality of One's Life	294
Condition 2: Being Able to Handle an Adverse Event	296
Property 2: Living with Concern for At-Risk Children	297
Condition 1: Physical Signs and Symptoms	297
Condition 2: Children's Knowledge of ARVC.....	300
Chapter Summary	301
Chapter 9 Conclusion	305
Findings.....	305
Limitations	316
Sample.....	316
Method	317
Final Reflection.....	318
References.....	320
Appendices.....	375

List of Tables

Table 5.1 Participant Sociodemographics.....	134
Table 5.2 Focus Groups Characteristics	138

List of Figures

Figure 1.1: TMEM43 Protein Placement	17
Figure 1.2: Triangle of Dysplasia	18
Figure 1.3: History of ARVC in NL	23
Figure 5.1: Grounded Theory: Theoretical Coding	146
Figure 6.1: Rubik's Cube: Generic Model.....	170
Figure 6.2: Construct 1: Awakening to A New Meaning of Being At-Risk.....	172
Figure 6.3: Rubik's Cube Model: Summary of the Conditions that Influence the Psychosocial Process of Awakening to a New Meaning of Being At-Risk.....	203
Figure 7.1: Construct 2: Deciphering the Meaning of Being At-Risk	205
Figure 7.2: Making the Decision to Have Predictive Genetic Testing For ARVC.....	215
Figure 7.3: Rubik's Cube Model: Summary of the Conditions that Influence the Psychosocial Process of Deciphering the Meaning of Being At-Risk.....	246
Figure 8.1: Construct 3: Embodying a New Meaning of Being At-Risk.....	247
Figure 8.2: Relationships between Predictive Genetic Test Result, Disease Symptoms, and Treatment	250
Figure 8.3: Rubik's Cube Model: Summary of the Conditions that Influence the Psychosocial Process of Embodying a New Meaning of Being At-Risk	304
Figure 9.1: Rubik's Cube Model: Summary of the Conditions that Influence the Psychosocial Process of Constructing the Meaning of Being At-Risk.....	319

List of Abbreviations

AIDS: Acquired Immune Deficiency Syndrome

ARVC: Arrhythmogenic Right Ventricular Cardiomyopathy

ARVC5: Arrhythmogenic Right Ventricular Cardiomyopathy Type 5

BRCA1/2: Breast Cancer Gene One and Gene Two

CHF: Congestive Heart Failure

CHD: Coronary Heart Disease

CRC: Colorectal Cancer

DNA: Deoxyribonucleic Acid

FAP: Familial Adenomatous Polyposis

FH: Familial Hypercholesterolemia

HBOC: Hereditary Breast and Ovarian Cancer

HCM: Hypertrophic Cardiomyopathy

HIV: Human Immune Deficiency Virus

HD: Huntington Disease

HNPCC: Hereditary Non-Polyposis Colorectal Cancer

HREB-MUN: Health Research Ethics Board Memorial University of Newfoundland

ICD: Implantable Cardioverter Defibrillator

LQTS: Long QT Syndrome

NL: Newfoundland and Labrador

TMEM43: Transmembrane Protein

VT: Ventricular Tachycardia

List of Appendices

Appendix A: ARVC Diagnostic Criteria	375
Appendix B: The Theory: Constructing the Meaning of Being At-Risk	376
Appendix C: Amendment for Ethical Approval.	377
Appendix D: Recruitment Script.	379
Appendix E: Research Study Cover Letter	380
Appendix F: Summary of the Research Study	381
Appendix G: Consent Form	382
Appendix H: Sample Interview Schedule/ Questions.....	386
Appendix I: Ethics Approval	390
Appendix J: Research Approval: Eastern Health.....	392

CHAPTER 1

INTRODUCTION

Mary¹ was born and raised in outport Newfoundland, one of six children. She has personally witnessed the sudden deaths of three of her brothers, each from a heart condition, each by their early 50s. As well, her father died suddenly at the age of 32 while fishing. Her grandmother was only 43 when she collapsed one spring while hanging clothes on the line. Mary has three sons who have taken up the family tradition of fishing for a living. As they approach their late twenties, she wonders if they will live to see their children grow up. This dissertation shares the stories of women and men like Mary, whose entire lives have been consumed with losses related to a particular form of hereditary sudden cardiac death, as they engage in the predictive genetic testing process in order to identify who is at-risk and as a consequence construct what it means to be at-risk in an ever changing landscape of new genetic discoveries.

The past decade has seen a proliferation of available predictive genetic tests. These state-of-the-art tests are the result of one of the most significant scientific advances of the 21st century: the Human Genome Project, a ten-year 2.7 billion dollar, project that identified approximately 20,000-25,000 genes in human deoxyribonucleic acid (DNA), and the sequences of the 3 billion chemical base pairs, that make up human DNA (Venter et al., 2001). Given the magnitude of the scientific advances, social scientists have begun the process of examining how individuals living in families at-risk for a fatal genetic

¹ This is a pseudonym. Throughout this dissertation identifying gender has been removed except when it is necessary for understanding the context of the discussion.

condition assign meaning to their risk and make subsequent decisions about their health throughout the genetic testing process.

A large body of research has explored the psychosocial and behavioural impacts of participating in predictive genetic testing for several genetically linked conditions (e.g., breast cancer [BRCA1/2], Huntington disease [HD], and colorectal cancer [CRC]) (e.g., Cox & McKellin, 1999; d'Agincourt-Canning, 2005; 2006b; Etchegary, 2006a, 2006b, 2009, 2010; McAllister, 2003). This has done much to advance the understanding of how genetic technologies shape individual lives in terms of the lived experience of being at-risk. That research focuses on how the availability of a genetic test shapes everyday experiences and health decisions. Little attention has been paid to how a lack of available technology, or sudden advances in an available technology, may shape and reshape the meaning of risk in relation to everyday lives and health decisions; virtually no attention has been paid to the important question of how those embodied experiences of risk may in turn to some extent shape the development of genetics.

This study examines how individuals construct their ideas about risk alongside new gene discovery. It examines how the experience of being at-risk and meanings assigned to “risk” for a genetic linked condition are shaped by, and to some degree help to shape, the science of gene discovery and genetic testing.

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) is an ideal genetic condition through which to examine how the experience of risk is shaped by and reshapes genetic science. ARVC is a highly penetrable and fatal condition. ARVC is not susceptible to most lifestyle modifiable factors (e.g., diet). The gene mutation was isolated in NL; individuals and families living at risk for ARVC in this province were

directly engaged in the scientific developments that led to increasingly accurate risk assessments for the condition. A focus on ARVC in the province of NL therefore provides a unique opportunity to gain insights into how ideas about being at risk for a genetically linked condition are shaped and reshaped in light of one's experiential knowledge and evolving scientific knowledge.

Purpose of the Study

The purpose of this study is to examine the experiences of individuals living in a family at risk for ARVC as they move through the genetic testing process at different historical periods of gene discovery. It examines how individuals living in families at risk for a fatal genetic condition assign meaning to their risk and make subsequent decisions about their health at the intersection of science and life context. The intent is to highlight for health care professionals, health policy makers, and genetic scientists the potential health care needs facing at-risk individuals in the context of novel gene discovery.

What is ARVC?

ARVC is an autosomal dominant heart condition found primarily in young males that can cause sudden cardiac death (Hodgkinson et al., 2009). Offspring of affected parents have a 50% chance of inheriting the condition. Although women can be affected with ARVC, they seem to experience symptoms to a lesser degree and later in life (Hodgkinson et al., 2012). ARVC is genetically heterogeneous; the form of ARVC that one inherits depends on the gene affected, the location of the gene on the chromosome, the type of mutation, and the protein involved. At the time of this writing, twelve types of autosomal dominant ARVC and one type of autosomal recessive ARVC have been

identified. The participants in this study have ARVC5. The gene for ARVC, as shown in figure 1.1, is located on chromosome 3p25 and is caused by a mutation in a transmembrane protein (TMEM43) (Merner et al., 2008).

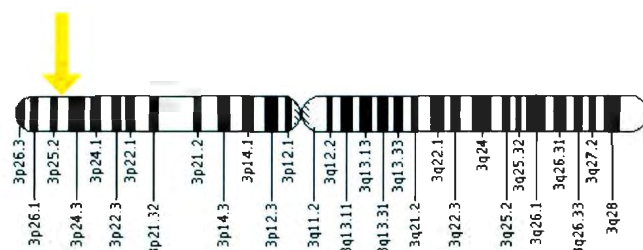


Figure 1.1. Diagram of the position of the TMEM43 protein. National Library of Medicine. (2012). *Where is the TMEM43 gene located?* Diagram retrieved from <http://ghr.nlm.nih.gov/gene/TMEM43>

In ARVC normal heart muscle is replaced by fibro-fatty tissue on the wall of the right ventricle. This fibro-fatty tissue impedes the cardiac cells' or myocytes' ability to generate normal electrical impulses throughout the heart, predisposing individuals to lethal ventricular arrhythmias that cause a sudden cardiac death (Cox et al., 2010; Fontaine, Fontaliran, & Frank, 1998; Hodgkinson et al., 2009; Marcus et al., 1982; Merner et al., 2008; Theine, Nava, Corrado, Rossi, & Pennelli, 1988). The extension of this fatty fibrous tissue throughout all layers of the myocardium tissue leads to a thinning of the ventricular wall, the development of aneurysms, and creates what has been referred to in research literature as the "triangle of dysplasia" (see Figure 1.2) (Basso, Corrado, Marcus, Nava, & Thiene, 2009). As the disease progresses, the left ventricle can also be affected (Corrado et al., 2000; Gerull et al., 2004). The loss of myocyte adhesions during times of mechanical stress on the heart (e.g., exercise) explains the occurrence of ARVC during exertion and in athletes (Gerull et al.).

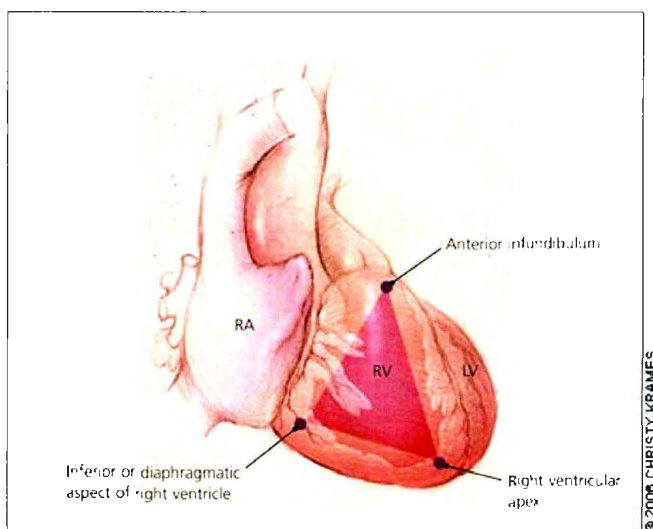


Figure 1.2. An illustration of the Triangle of Dysplasia in the right ventricle .Reprinted from Anderson, E (2006). Arrhythmogenic right ventricular dysplasia. *American Family Physician*, 73(8), p.1392. 2006 by Christy Krames. Reprinted with permission.

Prevalence of ARVC

The precise global prevalence of ARVC is not known because ARVC is difficult to diagnose (Corrado et al., 2000) and a definitive predictive genetic test for ARVC has only been available since 2007. The worldwide prevalence of ARVC has been estimated to be in the range of 1:1,000 to 1:5,000 (Gollob et al., 2011; Thiene, Corrado, & Basso, 2007). A familial history of ARVC has been noted in 30% to 50% of documented cases of ARVC (Corrado et al., 2000; Merner et al., 2008).

At the time of this research study, there were 15 families in NL with a documented history of ARVC. Of these 15 families, there were 284 suspected individual cases of ARVC (Hodgkinson, 2007). Although the prevalence of ARVC in NL remains unknown, estimates suggest it to be approximately 1:1,000 (K. Hodgkinson, personal communication, October 29, 2012). What is known about ARVC in NL is that the

median life expectancy of affected and unaffected males is 41 and 83, respectively, and 71 and 83 in affected and unaffected females (Merner et al., 2008). ARVC is 100% penetrance in males by age 63 and females by the age of 76, which means that if one has the gene they will have clinical signs of the disease regardless of gender and will eventually develop heart failure in the long-term (Hodgkinson et al., 2009; Merner et al., 2008). Of those identified as having ARVC in NL, a sudden cardiac death occurred in 86% of affected males and 42% in females. Thus, affected males were 6.8 times more likely in comparison to females to die from ARVC (Merner et al., 2008).

Diagnosis of ARVC

Prior to the availability of a definitive predictive genetic test in 2007, clinical diagnosis of ARVC was based either on the presence of two major criteria, one major and two minor, or four minor criteria². The diagnosis was made using multiple measures including electrocardiograms, signal averaged electrocardiograms, Holter monitors, cardiac ultrasound, and family history (see Appendix A³). Despite the availability of diagnostic criteria, the clinical diagnosis of ARVC prior to 2007 was difficult as it relied on physiological and pathological testing for structural changes in the right ventricle, the presence of myocardial fatty fibrous tissue, and the presence of ventricular arrhythmias and electrocardiogram changes (e.g., T wave inversion, extended QRS, and epsilon waves) (Hodgkinson et al., 2009; Merner et al., 2008).

² ARVC Diagnostic Task Force Criteria were proposed in 1994; however these have been modified to improve diagnostic sensitivity for early disease and detection in children.

³ Appendix A: ARVC Diagnostic Criteria.

Added to this difficulty with diagnosing ARVC prior to 2007 was that the diagnostic criteria were “biased due to the skewed ascertainment of those presenting with severe disease and because many require tertiary-level cardiac testing. Subjects in NL often presented with death in rural areas, with no previous medical history” (Hodgkinson et al., 2012, p.7). As well, patients may have “concealed” or had absent clinical signs of the disease even with structural changes in the right ventricle (Nava et al., 2000). In the case of children, absent or limited phenotypic expression of ARVC has been noted, because ARVC is a progressive disease that manifests itself predominantly in young men in their mid-forties (Basso et al., 2009).

The discovery of a definitive genetic test in 2007 enabled health care practitioners to diagnose ARVC with certainty. A predictive genetic test refers to the examination of genetic material through blood analysis in order to predict the likelihood that an individual will or will not develop the condition in question (Davison, 1996). Predictive genetic testing is unique in that it can foretell the health outcomes or lifetime risk of acquiring a disease of an otherwise healthy, asymptomatic person.

Symptoms and Management of ARVC

ARVC penetrance is age-related. Clinical signs and symptoms of ARVC can occur from adolescence onwards (Basso et al., 2009; Nava et al., 2000); however, symptoms get progressively worse with age, peaking in the forties for men and much later in life in women (Basso et al.; Hodgkinson et al., 2012). The physical signs and symptoms of ARVC are variable, ranging from no symptoms to palpitations, chest pain, syncope, ventricular arrhythmias, biventricular heart failure, and a sudden cardiac death (Fontaine et al., 1998). Anatomically, ARVC can cause structural changes in the right

ventricle, which over time may involve the left ventricle, causing ventricular arrhythmias (Marcus et al., 2010).

Recent research (Hodgkinson et al., 2012) on the history of ARVC in NL reported that affected men were hospitalized four times more frequently than affected women. Affected women not only lived longer, but also did not present with symptoms as serious as those in affected men. The men also had higher incidences of congestive heart failure and sudden cardiac deaths in comparison to the women.

Primary management of ARVC includes the insertion of an implantable cardioverter defibrillator (ICD) to prevent arrhythmias; medications to control blood pressure, arrhythmias, and cholesterol; and restricted physical activity (Gollob et al., 2011). The ICD is a small device inserted under the chest wall. In the event of an arrhythmia, it delivers an electrical shock to restore the heart to a normal sinus rhythm.

History of ARVC

The first historical reference to ARVC was noted by Lancisi in 1736 (Thiene, et al., 2007; Thiene, Nava, & Marcus, 1997). Lancisi described a family whose members over four generations experienced symptoms similar to ARVC: heart palpitations, heart failure, and in some cases, a sudden cardiac death (Basso et al., 2009). In 1961 in Padova, Italy, researchers reported on two clinical cases wherein heart disease had affected the right ventricle, impairing its ability to effectively pump blood (Dalla-Volta, Battaglia, & Zerbini, 1961). A group of French researchers coined the term “ARVC” as an arrhythmic disorder when, in 1977, they identified six patients who had sustained ventricular tachycardia (VT) resistant to antiarrhythmic drugs with signs of heart disease (Fontaine et al., 1977). Formal credit for the first clinical description of ARVC has been

given to Marcus et al. (1982) who described 24 cases of right ventricular dysplasia. Shortly thereafter, ARVC was defined as something familial (Nava et al., 1988) that caused a sudden cardiac death in young people with little or no warning (Thiene et al., 1988).

History of ARVC gene discovery in NL

A history of sudden cardiac death was first noted in the 1970s in a NL family whose origins traced back to a couple born in 1799 and 1800 (see Figure 1.3⁴) (Hodgkinson et al., 2012); however, it was not until 1988 that NL researchers reported on a local study wherein five patients had received a diagnosis of ARVC (Marshall et al., 1988). Included within the family cited in the study by Marshall et al. was one individual who had a surgical dissection of the right ventricular free wall as a treatment for ventricular tachycardia caused by right ventricular dysplasia in 1983 (Guiraudon et al., 1983).

In 1994, following funding for the Human Genome Project in 1990 (Anderson, 2004), a US-based team began to conduct research on ARVC in NL. Three years later, local NL researchers began conducting research similar to that of the US team. Unfortunately, the blood samples collected by the initial NL research in the 1980s were destroyed due to a lack of storage facilities. This coupled with the fact that many of the original family members who had given blood samples had died, left researchers with no source of DNA upon which to complete further analysis (Hodgkinson et al., 2009). Using the blood samples attained in NL, the US research team completed DNA haplotype

⁴ Figure 1.3: The History of ARVC in Newfoundland and Labrador

analysis of the blood samples obtained in NL and identified the ARVC gene as being on the short arm of chromosome three in position 25 or 3p25 (Ahmad et al., 1998).

Haplotype analysis refers to the identification of a series of genes inherited as a unit and not a definitive genetic test.

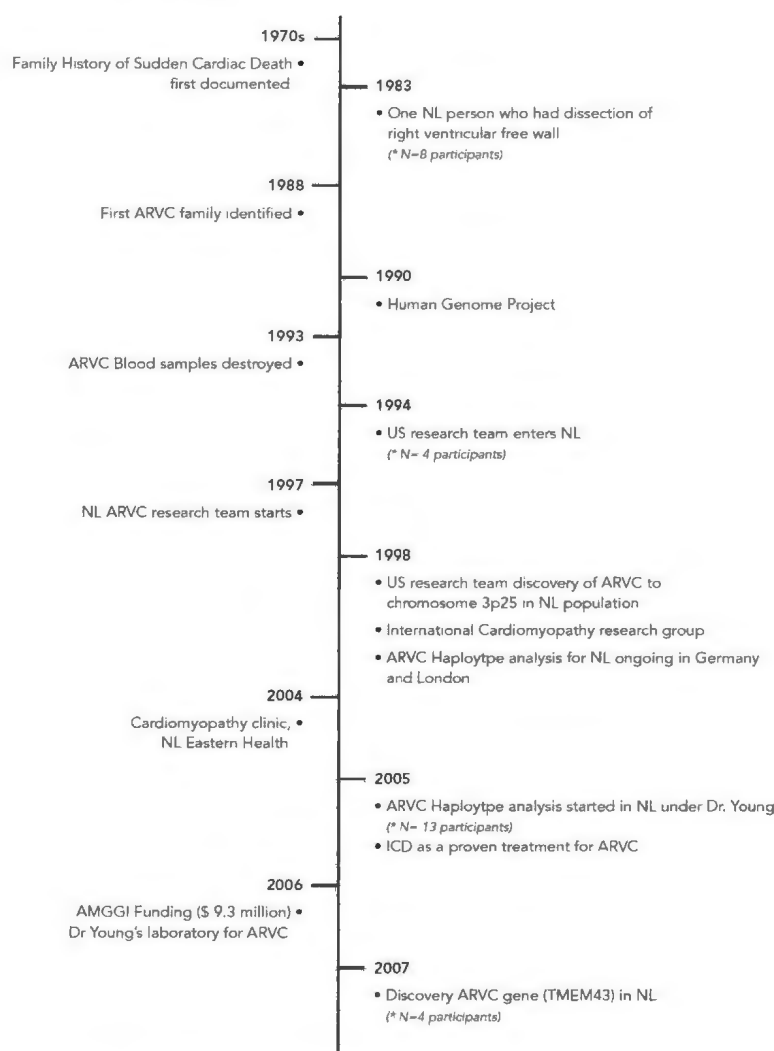


Figure 1.3 History of ARVC Newfoundland and Labrador (NL). This figure provides a chronological representation of ARVC gene discovery in NL. (* represents the number of participants in this study who entered the genetic testing process at the time of discovery.)

Research on ARVC in the late 1990s was the impetus behind the start up of an international genetic cardiomyopathy research group based in NL, whose goal was to isolate the gene responsible for ARVC. Haplotype analysis for ARVC was done in Germany until 2005, when a genetics lab was started in NL under the direction of Dr. Terry Lynn Young (Hodgkinson et al., 2012). Haplotype analysis enabled researchers to assign at-risk individuals a status of either “high risk” or “low risk,” depending on if they had the disease-associated haplotype. A high-risk status meant that the likelihood of having the ARVC gene was 98% (Hodgkinson et al., 2009). There were still cases, however, where haplotype analysis could not assign risk status or where individuals were assigned a 50% risk of being positive or negative. In 2006, the Atlantic Medical Genetics and Genomics Initiative (AMGGI) supported Young’s lab, as part of a four-year, \$9.3-million project launched by Genome Canada, to discover and identify the causative mutation for the ARVC gene (Genome Canada, 2009). In 2007, the causative gene for ARVC was isolated in 15 NL families (Merner et al., 2008).

Parallel with the discovery of the haplotype for ARVC was the introduction of the ICD as first line treatment for the lethal cardiac arrhythmias causing death from this disease. Research on the efficacy of the ICD as a treatment for ARVC confirmed its effectiveness in young males in NL, as the five-year mortality rate post-ICD implantation in males was zero (Hodgkinson et al., 2005).

The heightened awareness of the presence of ARVC within NL following haplotype analysis, the knowledge of the potentially sudden outcome of ARVC, the start of the international genetic cardiomyopathy research group in NL, the development of a local research lab dedicated to ARVC, and the knowledge that the ICD provides a

effective treatment led to the development of a cardiac genetics clinic in 2004 within the Eastern Health Regional Health Authority of NL. The cardiac genetics clinic provides an array of services using a multidisciplinary approach to care and includes genetic counsellors, an ethicist, nurses, and a cardiologist. Any individuals living in families at risk for developing ARVC are referred to this clinic for assessment and counselling (Hodgkinson et al., 2012).

This research study is significant in that it examines how the embodiment of risk is (re)shaped in light of new genetic discoveries and treatments. To date, there is no research that has explored the “lived experience” of living in a family at risk for ARVC. This study not only fills this gap but also offers recommendations for future health policy, education, research, and practice in order to meet the health care needs of this population.

Overview of Chapter Contents

Chapter two provides an historical overview of the concept of risk, and it also reviews four key theoretical approaches to understanding risk within the context of genetics: cultural theory, cognitive (psychological) theory, sociological theory, and governmentality. A common theme throughout this chapter is the contextual nature of risk construction.

Chapter three is a review of the literature that examines how risk for a genetically linked condition is assessed and understood by laypersons and experts, how decisions about predictive genetic testing are made, how risk is communicated, and how the perception of risk shapes behaviour in those classified as at-risk. This discussion takes place under the umbrella of three genetically linked conditions: BRCA1/2, CRC, and HD.

These three conditions are used for comparative purposes throughout the dissertation, along with references to general (non-ARVC) cardiovascular diseases where relevant. This chapter highlights the fact that perceptions about risk do not develop solely in reference to numerical values but also in relation to subjective experiences. Therefore, individuals living in at-risk families take a pragmatic approach to assigning meaning to risk and to understanding and coping with their risk.

The purpose of chapter four is to discuss the body of literature that has explored the psychosocial impact of predictive genetic testing. The review of this literature includes a discussion of the presence and duration of psychological distress experienced by carriers and non-carriers of genetically linked conditions. This includes the factors that influence psychological distress throughout the genetic testing process, and the impact of testing on families and individuals who have testing.

Chapter five begins with an overview of grounded theory and the particular grounded theory method used in this study. Details are provided as to how I situate myself within the research process, drawing on the tenets of symbolic interactionism and pragmatism. Concluding this chapter is an overview of the ways in which I ensured rigour in the study and a description of the ethical considerations.

Chapter six is the first of three chapters that focus on the research findings from this study. Chapter 6 starts with an introduction into the substantive theory, *Constructing the Meaning of Being At-Risk* (see Appendix B⁵). It describes the Rubik's Cube as a model to understand how at-risk individuals juxtapose the three contextual dimensions

⁵ Appendix B: The Theory: Constructing the Meaning of Being At-Risk

(scientific knowledge, experiential knowledge, and phase of the genetic testing process) against relevant conditions to construct ideas about risk (see Figure 6.1⁶). The first theoretical construct, *Awakening to a New Meaning of Being At-Risk*, captured participants' experiences as they awakened to the idea that they may be at risk, prior to predictive genetic testing. The two categories that capture this experience are (a) *making sense of numerous losses* and (b) *struggling to break the cycle of uncertainty*. Evident within this chapter is how ideas about risk are understood in relation to social and scientific contexts.

Chapter seven examines the second phase of the psychosocial process, *Constructing the Meaning of Being At-Risk*. The theoretical construct *Deciphering the Meaning of Being At-Risk* captures participants' experiences during genetic testing. This includes being offered a predictive genetic test, waiting to receive test results, and receiving the test results. The two categories (a) *taking the first steps of the genetic testing process* and (b) *building one's risk portfolio* reflect this experience. This chapter describes how individuals make a decision to have genetic testing or not. Part of making this decision includes the juxtaposition of one's scientific knowledge against one's experiential knowledge, and the conditions that hold relevancy at the time. Threaded throughout this chapter is the idea that objective risk alone is not enough to help at-risk individuals decipher their meaning of being at-risk. Furthermore, the psychosocial process of *Constructing the Meaning of Being At-Risk* is fluid, transient, and pragmatic.

⁶ Figure 6.1: The Rubik's Cube: Generic Model

Chapter eight is the last of the results chapters and represents the last phase of *Constructing the Meaning of Being At-Risk*. The theoretical construct *Embodying a New Meaning of Being At-Risk* describes participants' experiences post-genetic testing. This chapter describes how participants start to adjust to and cope with their experiences of the genetic test results in their everyday lives. The three categories (a) *adjusting to living with or without a genetic condition*, (b) *recognizing the reality of living in a family at risk for genetic diseases*, and (c) *looking towards the future* capture this experience. As in the other two phases of the genetic testing process (pre-testing and during testing), participants rely on their experiential and scientific knowledge to help them embody a new meaning of risk, cope with their experiences of living in an at-risk family, and overcome barriers to resources (e.g., human, physical, and financial) and restrictions on their daily lives (physical activity, social activity, driving, education, and employment). This chapter highlights participant concerns for how their offspring will cope and manage being at risk for ARVC.

Chapter nine concludes this dissertation with a discussion of the findings and implications of this research study for education, research, and practice. Limitations of the study are addressed.

CHAPTER 2

THEORETICAL APPROACHES TO UNDERSTANDING RISK

The first documented illustration of risk is noted in the Tigris-Euphrates region of Mesopotamia in 3200 BC amongst the Asipu (Covello & Mumpower, 1985; Molak, 1997). The meaning of risk has shifted throughout history from a neutral term that inferred the mathematical probability that an event would take place to a term that described harmful or adverse outcomes synonymous with a danger or hazard (Douglas, 1990, 1992; Douglas & Wildavsky, 1982a; Fox, 1999; Lupton, 1993; Skolbekken, 1995). Over the last three decades research on risk and genetics has shifted from a quantitative approach of measuring risk to qualitative methods that acknowledge the intersubjective nature of how risk is embodied by individuals, families, and communities (Cox, 2003; d'Agincourt-Canning, 2005; Etchegary, 2009, 2006a, 2006b; Gifford, 1986; Lock & Nguyen, 2010; Lupton, 1993, 1995; McAllister, 2002, 2003).

I take the approach, following Lupton (1995) and other critical scholars of risk, that risk is a socially constructed concept with multiple meanings depending on the perspective of the one “at” risk and the one “measuring” risk. The meaning of risk is developed in relation to historical, social, and cultural contexts, and that holds true for individual perceptions of risk as well as for academic theories of risk. In this chapter I examine four theoretical perspectives that have made the largest contribution to our understanding of risk: cultural theory, sociological theory, cognitive-psychology theory, and governmentality.

Cultural Theory and Risk

The earliest writings on risk in cultural theory are found in Douglas's (1966) work on *Purity and Danger*, Douglas examined how risk avoidance in ancient civilizations was a means to construct cultural boundaries and maintain social order. That is, objects or events thought to be contaminated or polluted were considered dangerous or "risky" to the social order and deemed taboo. According to Douglas (1985), risk acted like a "forensic resource" that provided explanations as to why mishaps occur.

Cultural theorists view risk as a social process that varies according to context (Douglas & Wildavsky, 1982a; 1982b; Rayner, 1992; Wildavsky & Dake, 1990). The meaning of risk is constructed in relation to the values, belief system, ideologies, and structure of the social organization within which an individual interacts. Ideologies about risk are developed as collective beliefs of all members of a certain social group and not individually. Hence, people who do not share the same cultural belief patterns may not define the same behaviours as "risky" (Krimsky & Golding, 1992). Furthermore the identification of a specific risk factor and the meaning assigned to the risk is fluid, as it can change in response to its usefulness to an existing social system and the perceiver's social affiliation (Douglas 1985; Rayner, 1992).

Douglas (1970, 1978), and later, in collaboration with Wildavsky (Douglas & Wildavsky 1982a), developed the grid-group approach to understanding risk perception and risk management in relation to the sociocultural context in which individuals find themselves situated. This grid-group approach describes the degree of social affiliation to a particular group and the strength of social interactions within that group. The grid represents the nature, the expectations, and the constraints (e.g., gender, age) of these

social interactions (Krimsky, 1992). A high group-grid infers a cohesive group that perceives and responds to risk in a uniform manner reflective of the social group. A low group-grid lacks group solidarity in lieu of a more individualistic self-regulatory approach to risk (see also Lupton, 1999).

Douglas (1996) described this social process of assigning risk as similar to creating a taboo. A taboo is a practice where boundaries are set up as to how one interacts with the object considered taboo. Interactions with the taboo result in danger to oneself and can cause contamination of the community. The creation of a taboo is evident in the stigmatization of homosexual males living with Acquired Immunodeficiency Syndrome (AIDS) (Farmer, 1992). The process of labeling something a taboo represents a visible attempt to provide some sort of structural dimension to at-risk populations in order to justify our collective rational thoughts and maintain societal order. As noted by cultural theorists, the identification of something “risky” within society serves to construct cultural boundaries between individuals, social groups, and communities (Douglas, 1996; Lupton, 1999). Groups that are assigned a social status of “risky,” such as homosexual males, are therefore singled out because of their symbolic threat to society. Cultural theory helps us understand the contextual nature of risks, that is, how individuals’ social relationships influence perceptions of risk and subsequent decisions.

Cultural theory provides insight into how individuals might understand risk. However, it fails to fully explain why some individuals living in a family with a predictive genetic disease decline to participate in the predictive genetic process. Drawing on cultural theory, one might assume that individuals growing up in a family at

risk for a genetic condition would have comparable beliefs and therefore would respond to predictive genetic testing in a similar manner. This is not the case, as individuals living in these at-risk families do not consistently react in the same manner and engage willingly in genetic testing. Although cultural theory provides good insights into how individuals construct and respond to risk, it does not explain resistors to technology and why some individuals do not conform to collective beliefs of a social institution or group.

Another assumption of cultural theory is that one's actual risk is not known until after the incident happens (Tansey & O'Riordan, 1999). This is not the case for individuals at risk for a genetic condition such as ARVC. These individuals may know their risk status and outcomes years prior to having any risk symptoms. Thus, the experience of being at risk for a genetic condition and the responses to that risk can be significantly different from being at risk for another chronic disease, as the element of uncertainty is removed in the former.

Cognitive (Psychological) Theory and Risk

Starr's (1969) paper, "Social Benefit Versus Technological Risk," was critical in setting the stage for the development of a new sub-discipline of risk perception, that is, technology and risk. Looking at what constitutes an acceptable technological risk, Starr's work sparked interest in understanding how individuals conceptualize risk. A conceptual framework that acknowledges the subjective nature of risk perception guides the cognitive paradigm. That is, individuals who are influenced by an assortment of psychosocial, organizational, and cultural factors subjectively define risk. It is through the use of psychometric instruments that these foregoing factors and their interrelationships are quantified and modeled in order to understand and predict

perceptions of risk and coinciding rationale responses (Slovic, 1992, 2000). Researchers have challenged the reliability of psychometric instruments to explain and predict risk responses, noting a significant difference between laypeople's perception of risks and that of experts when presented with the same information (Slovic, Fischhoff, & Lichtenstein, 1979). Heuristics is thought to influence the meanings that laypeople assign to risk, particularly if these meanings differ from that of experts and existing quantitative research (Lupton & Tulloch, 2002).

Heuristics are mental road maps that describe the process that individuals experience as they configure responses to risk (Lupton & Tulloch, 2002). An individual's road map is dependent upon his or her life experiences and personal frames of references that is then identified as risk attributes. These risk attributes help individuals then to understand and make decisions about their risk under certain circumstances (Krimsky & Golding, 1992; Tversky & Kahneman, 1973, 1974; Zajonc, 1980). Acknowledgement that decisions surrounding risks rarely occur in an emotionally neutral context (Johnson & Tversky, 1983), but are often in response to contextual variables in one's life, has contributed to the popularity of the heuristic paradigm.

A key critique of cognitive theory in general is that it is guided from an individualistic paradigm and thus lacks generalizability at a population level. This individualistic approach makes it difficult to translate findings into healthy public policy. Others (e.g., Lupton, 1999; Slovic, 1992) criticize the psychometric paradigm in that does not take into account the broad contextual nature of risk. Some scholars (e.g., Slovic) challenge the ability of cognitive theory to explain why individuals define certain events as "risky" but ignore those factors labeled "risky" by others.

Cognitive theory provides insight into how individuals living in a family with a genetic condition understand their risk; however, it often fails to capture the fluid nature of risk, that is, how individuals' perception of risk is shaped and reshaped over time. As well, it cannot account for many diverse contextual factors (or variance in these contextual factors) that influence risk perception, many of which cannot be quantified.

Sociological Theory and Risk

According to some sociological theorists, risk is a social process that evolves in response to the meanings that individuals assign to social relationships and interactions. It takes into consideration the lens through which individuals view the world and how individuals give meaning to their experiences, interpret knowledge, and respond to social relationships (Krimsky & Golding, 1992; Lupton, 1999; Lock & Nguyen, 2010). Lupton (1999) proposed that knowledge about risk is mediated through discourses or social and cultural frameworks of understanding. That is, the meaning an individual attributes to risk is in a constant state of flux and is historical and contextual in nature. Lupton forgoes the argument on rationality or irrationality of risk decisions to focus on how the meaning of risk operates as part of one's social relations and subjectivity.

A key theoretical approach influencing sociological theory is that of a world risk society noted by Beck (1992) in the book *Risk Society: Towards a New Modernity* and threaded throughout Giddens's (1991) earlier work. This perspective examined the social construction of the concept of risk in the light of social and technological advancements in modern society, which have increased to such a magnitude as to be impossible to calculate, impossible to manage, and impossible to avoid (Beck, Bonss, & Lau, 2003; Lupton, 1999; Wilkinson, 2010). According to Beck (1992) the concept of risk can

become tantamount with large-scale hazards that are difficult, if not impossible, to quantify. These hazards lack accountability or compensation, and they can be considered the by-products of manufactured uncertainties. The label of being “risky” evolves as a result of discourse between producers of risk knowledge, disseminators of knowledge, and receivers of knowledge. These factors have led to what Beck refers to as the globalization and individualization of risk.

Beck (2006) argued that risk has become so “global” that it impacts everyone’s life to some point. Globalization is the impetus to society’s constant state of risk that compels its members to engage in collaborative public forums on the future of society and risk management strategies characteristic of what Beck describes as a cosmopolitan society (Beck, 1992, 1996; Fox, 1999; Lupton, 2006; Wilkinson, 2010). In contrast to this cosmopolitan society, individuals at risk for a genetic condition construct their own personal theories of risk in order to develop a more concrete meaning of risk for themselves and their families through the process of what Beck refers to as “becoming individual”: an individual who is in a constant state of risk awareness, disequilibrium, and experiences a non-linear existence that is at the interface of the social and technological world (Beck & Beck-Gernsheim, 2002; Beck et al., 2003). This is a difficult task given the lack of control associated with risk and the breadth of its meaning (Zinn, 2008). As in the case of the Human Genome Project, the proliferation of genetic knowledge because of technological and scientific advancements upon which one had little or no control has led individuals to question their biological certainty and also sparked discussions regarding technological determinism, social reductionism, and the meaning of being at-risk.

Beck's idea of a risk society has influenced how the concept of risk is perceived in the 20th–21st century of environmental, political, and technological disasters. Disasters such as nuclear plants accidents of Chernobyl (1986) and Three Mile Island (1979); the chemical spill in Bhopal (1984); the explosion of methane gas in the Westray Mine disaster (1992); the spill of petroleum in the Love Canal disaster (2008); the Tsunamis of Thailand (2007), Japan (2010); and most famously the “911” terrorist attack on the World Trade Centre (2001) have shifted the meaning of risk to include the need to prepare for the unpreparable. This has caused individuals to be in a constant state of risk anxiety, which is impossible to calculate, impossible to manage, and impossible to avoid (Beck et al., 2003). Risk is no longer in relation to a specific object but linked to a variety of factors such as human error, technological malfunction, cultural and religious beliefs, and the unpredictability of “mother nature.” These events have led to a flurry of activity to identify, decipher, and cope with potential risks in a broader global sense in areas such as acid precipitation, depletion of the ozone layer, global warming, second-hand smoke, biodegrading of waste, and genetic discovery.

Sociological theory provides a good foundation upon which to understand how individuals living in families with a genetic condition conceptualize their risk. First, it accounts for the fluid and contextual nature of risk at a population and individual level. Second, it provides a lens upon which to understand how individuals juxtapose competing ideas about risk in order to develop personal theories of risk. Third, it captures those factors influencing risk that are neither quantifiable nor static. Finally, sociological theory helps to explain how at-risk individuals make choices about their health and how these decisions fluctuate in response to social interactions.

Governmentality and Risk

The concept of governmentality emerged from the work of Foucault (1977, 1991), Castel (1991), Ewald (1991), and O'Malley (2004). According to this perspective, the impetus behind how risk is understood and the template against one's conduct is measured is determined by those in positions of power (e.g., government experts, policy makers, and researchers). It is the experts' varied perspectives on risk that have led to the proliferation of risk definitions and the development of institutions dedicated to the construction, reproduction, dissemination, and practices of varying theoretical approaches to risk (Foucault, 1977, 1991; Lupton, 1999). Implicit within this perspective is the notion that risk is controllable, as long as experts can continue to identify what are the risk factors and society can employ methods to cope with the risk or at least the risk factors.

According to Dean (1999a), in response, individuals interpret risk and the suggested health promotion and prevention activities as something that they want to voluntarily participate in as responsible moral agents of society. It is this sense of volunteerism and moral responsibility, not social pressures, gained through conceptualizing the social world in terms of risk that allows government to govern through society and guide the actions of self-regulating individuals (Foucault, 1991; Lupton, 1995; Rose, 1993; Wilkinson, 2010). Thus, to be identified as "at-risk" is to be the object of constant self-surveillance (Castel, 1991; Foucault, 1977, 1991), because of the development of a risk consciousness (Lupton). This voluntary sense of surveillance and self discipline is salient within many health promotion and screening programs that individuals have grown to accept as routine or a normal part of everyday life (Bratich,

Packer, & McCarthy, 2003; Nadesan, 2008), such as prenatal screening (Lupton, 1999). If individuals fail to engage in preventive regimes, that failure can lead to punitive negative consequences, such as poor health (Nadesan, p.109).

This neoliberal approach to health care management (Dean, 1999b) has existed in Canada since the 1970s with the introduction of a government-driven health promotion participation programs (Boyce, 2002). Experts in the field of clinical genetics have done a fantastic job in identifying genes that put one at risk for future development of a disease (e.g., CRC, BRCA1/2, and cardiovascular disease) with predictive genetic testing. Simultaneously, health care professionals offer solutions to prevent the phenotypic expression of the disease, such as healthy eating guidelines, stress and coping techniques, and physical education. However, this approach has limitations, in that the responsibility to participate in these health programs is on the individual; this is often not feasible for those who are socially and economically disadvantaged. Furthermore, it does not address those cases where individuals have a genetic condition that has 100% penetrance regardless of participation in health promotion activities, such as in the case with ARVC. It also does not take into account how one's experiential knowledge can shape scientific knowledge or the implications that the rise of new discipline-driven biotechnologies (e.g., nanotechnology, genetic engineering) may have on at-risk individuals, as noted by other scholars (Duster, 2003; Rose, 2007).

Governmentality offers insights into risk perception and risk management; however, it does not explain how resisters to technology and health promotion programs conceptualize risk or why they do not engage in health promotion activities. Thus, the layperson's subjective interpretation of what constitutes risk or deviant behaviour is not

addressed. Also, if experts' opinions were the predominant factor shaping the meaning of risk, then one would expect that individuals would participate in surveillance programs and genetic blood sampling readily; however, volunteerism in predictive genetic testing programs is not always the case, regardless of how well representations of the risk factor are orchestrated by experts and government. Finally, many autosomal dominant conditions, such as ARVC, do not fit comfortably under the health promotion umbrella. That is, despite engaging in health promotion activities, modifying one's lifestyle, and taking prescribed treatments, physical signs of the ARVC persist. One would argue that theory of risk is satisfactory, if the context of risk was predictable, measureable, and static, and laypersons were passive individuals. However, this is not the case.

Chapter Summary

There are four theoretical approaches to risk that shed light on the ways risk is constructed from a social, cultural, cognitive, and political stance. Cultural theorists link risk perception to the cultural patterns, values, and beliefs of one's allied social group. Cultural theorists refer to the grid-group framework as a method to explain how individuals conceptualize risk in relation to one's social interactions. The analogy of a taboo is used in cultural theory to describe how at-risk individuals assign meanings to being at-risk. Cognitive theorists describe risk in terms of a subjective experience, which can be measured using psychometric tools. Cognitive theorists believe that heuristics plays a significant role in conceptualizing risk and in identifying risk attributes. From a sociological perspective, risk perception is a social process that evolves in response to the nexus of one's social relationships and interactions within society. Risk perception is viewed as something non-static, something not quantifiable, and something that can be

both individualized and global. Governmentality holds that, although those in positions of power or disciplines drive ideas about risk, it is the responsibility of the individual to govern their health through self- surveillance and voluntarily engaging in healthy activities.

CHAPTER 3

THE CONSTRUCTION OF RISK

Even though risk is a concept evident in the literature since the 14th century, ideas about the meaning of “risk” for individuals and families living with hereditary genetic conditions continue to be discussed, debated, and contested by scholars. Despite the strong presence of the rationalist perspective in the risk literature, risk perception is a deliberate, logical, and conscious process based on one’s appraisal of scientific evidence and events (Lupton, 1999; Slovic, Finucane, Peters, & MacGregor, 2004). Recent research highlights the subjective nature of risk and proposes an “experiential paradigm” for understanding risk (Cox, 2003; Cox & McKellin, 1999; d’Agincourt-Canning, 2005; Etchegary, 2010; Lock & Nguyen, 2010; McAllister, 2002, 2003). Within this paradigm, one’s life experiences and social interactions influence how ideas about risk are constructed.

The “experiential paradigm” does not supplant the notion of risk as being somehow real in the sense of greater or lesser degree of certainty but rather is used as a complementary framework to conceptualize risk. This shift is due to a growing awareness of the multifactorial nature of many genetic conditions resistant to traditional Mendelian theories of inheritance (Rose, 2007), inconsistencies in quantitative literature on what constitutes being at-risk (Sivell et al., 2008), and a lack of understanding of the contextual factors that shape and reshape risk perception. Thus, there is a growing body of literature that suggests risk perception is a social process that is best understood using a pragmatic lens. That is, ideas about risk are shaped and reshaped in response to the nexus of one’s social interactions, interpretations of these interactions, and available information

(Jeon, 2004; MacDonald, 2001). Throughout this literature review I draw on the tenets of symbolic interactionism (SI) (Blumer, 1969; Mead, 1934) as my framework to discuss this body of literature. The meaning of being at risk for a genetic condition is understood in relation to the social interactions individuals engage in. The meaning assigned to being at-risk is in a state of flux; it is constantly being shaped and reshaped to reflect one's worldview, one existing reality, and the interpretive process.

The following discussion includes five bodies of literature that facilitate an understanding of how risk perception is assessed, how representations of risk are understood by laypersons, how risk perception influences decisions surrounding predictive genetic testing, how risk is communicated, and, finally, how risk perception affects behavioural outcomes.

It is only with a thorough examination of how individuals and families construct their notions about risk in light of competing subjective realities that health care providers can begin to tailor programs to meet clients' needs and facilitate their movement through the predictive genetic testing process in a positive manner. First, I provide a brief rationale for my selection of the content of this literature review.

Rationale for Literature Review

To my knowledge, this is the first study to investigate the experience of living in a family with a history of Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC). As such, there is no existing body of literature on the psychosocial aspects of predictive genetic testing in this population. To contextualize my research I have drawn on two types of relevant research. First, I reviewed literature on patient and family experiences of living with genetically linked cardiovascular diseases, (that is, not specific to ARVC),

in order to gain insight into the experience of predictive genetic testing among individuals and families with ARVC. Second, I examined the quantitative and qualitative literature primarily on three well documented genetically linked diseases: (a) HD, a degeneration of the nerve cells in the brain causing marked cognitive, affective, and motor impairments; (b) BRCA 1/2, which includes two mutated genes isolated on chromosomes 13 and 17; and (c) CRC, which includes hereditary non-polyposis colorectal cancer (HNPCC) and familial adenomatous polyposis (FAP).

These three conditions were selected because they represent the majority of research in the field of predictive genetic testing, all have a predictive genetic test, and all are autosomal dominant conditions with a fifty percent chance of passing the disorder to one's offspring. HD, BRCA 1/2, and CRC all provide an excellent comparison group to ARVC. HD, like ARVC, is a fatal disease with high penetrance, early onset, and no cure. As with ARVC, most individuals with a positive genetic test will develop the disease independent of modifiable factors such as diet and exercise (Etchegary, 2009). By comparison, BRCA 1/2 and CRC are genetically linked diseases but are not necessarily fatal; they have varied penetrance, and treatment options are available. Furthermore, BRCA 1/2 and CRC are multifactorial in nature and less determinant; as a result, having the gene does not necessarily mean that one will develop the disease. For example, the life-time risk of developing CRC with an identified mutation is estimated to be between 30-95% (Esplen et al., 2007; Schneider, Kloor, Furst & Moslen, 2012). For women, the lifetime risk of developing BRCA 1/2 with an identified mutation is 39-85% (van Oostrom et al., 2003). Modifiable factors such as diet, environment, smoking, exercise, stress, and medications may prevent the manifestation of the gene or prevent the onset of

the disease. The reverse is also true; not having the gene does not necessarily mean one will not develop the cancer, as non-gene carriers remain at risk for cancer comparable to the general population.

Shaping the Meaning of Risk: Risk Assessment

Risk perception is the subjective assessment of the likelihood that an event will take place and the meaning assigned to the consequences of the event (Slovic, 1987). Given this definition one can easily understand how confusion about what constitutes risk exists. That is, depending on the paradigm or theoretical approach of the researcher (interpretive or empirical), risk has been framed primarily as either an objective or subjective concept in the literature, when in fact both approaches influence perceptions of risk. This study goes beyond this dichotomous approach to understanding risk; it examines how individuals at risk for ARVC juxtaposed objective and subjective risk. The meaning assigned to being at-risk, in this study, as a result of this juxtaposition, was significant in guiding participants' health care decisions. The following discussion provides an overview of the research that examines the accuracy with which individuals assess their risk status.

Limited evidence suggests that at-risk individuals correctly estimate their risk status (Grover et al., 2009; Himmelstein, 2010; van Dooren et al., 2004). Most of the literature suggests that at-risk individuals overestimated their risk (Braithwaite, Sutton, Mackay, Stein, & Emery, 2005; Codori et al., 2005; Croyle & Lerman, 1999; Esplen et al., 2001; Katapodi, Lee, Facione, & Dodd, 2004; Lerman et al., 1995; Ozanne, Wittenberg, Garber, & Weeks, 2010; Sheinfeld, Gorin, & Steven, 2003; Shiloh, Petel, Papa, & Goldman, 1998). By contrast, there is some evidence to suggest that those at risk

underestimate their risk (Katapodi et al., 2004) but this research is limited (Dorval et al., 2005; Grover et al., 2009; van Maarle, Stouthard, & Bonsel, 2003). These accounts presume that risk is somehow value-free and that risk perception can be measured against a norm and be quantified as correct or incorrect compared to the “real” measure of risk. However, many critical theorists have argued that such measures of risk perception are inappropriate, because one’s perception of being at risk for a genetically linked disease is influenced by many factors such as variability in disease patterns, treatment options, family history, and those variables that cannot always be quantified (Cameron, Sherman, Marteau, & Brown, 2009; Katapodi et al., 2004; Sivell et al., 2008). More importantly, an assessment of whether one’s perception of risk is higher or lower than what it really is (in terms of percentage of qualifying as “at-risk” in a numerical sense) is beside the point. It is the *meaning* of risk that is important, not the degree to which the assessment of risk is higher or lower than the scientific clinical assessment. What is considered “risky” for one person may not be judged risky for another, because risk, being subjective, is only meaningful in relation to the riskiness of life (Rapp, 1988; Brunger & Bassett, 1998). These two points are supported in the body of literature that has examined the inconsistencies in what constitutes an accurate risk perception.

How “Accurate” is Risk Perception?

Two systematic reviews have examined inconsistencies in the evidence on risks perception. These reviews support the position of critical theorists that risk is a subjective experience and not easily quantifiable (Katapodi et al., 2004; Sivell et al., 2008). Katapodi et al. conducted a meta-analysis (n=42 studies) in which they explored measurements of women’s perceptions of being at risk for BRCA 1/2. The consensus in

the literature they reviewed was that women do not have an accurate perception of their actual risk; in fact they often underestimated their risk when compared to assigned objective risk. This finding was contradicted in some studies that reported that a perception of high risk was associated with having a family history of BRCA1/2, having an affected relative, engaging in genetic counselling, and having a prophylactic mastectomy. A positive relationship was observed between perceived risk and intensity of emotional response to BRCA1/2 concern. Of those studies that examined the relationship between social demographics and risk, 12 studies reported a weak correlation between risk perception, age, and income; five of these found that young women and those with less than a high school education were either unaware or overestimated their risk. In all of the 12 studies, white women perceived their risk as higher than did women of minority ethnic communities.

Although the review by Katapodi et al. (2004) provides insights into women's risk perception, the authors noted many methodological concerns with the reviewed literature, particularly in relation to measurement scales. Some studies (n=6) used numerical scales while others used verbal Likert-type scales (n=7) or single-item scales (n=2), which can bias results. Second, scales measured perceived risk as a one-dimensional concept (low or high) that remains static over time, which is not the case. Third, only eight of 42 studies addressed reliability and validity of psychometric instruments. Fourth, the review included only cross-sectional studies, which are not representative of the transient and fluid nature of risk perception throughout the genetic testing process but are, rather, a snap shot of risk perception at a specific time. Finally, findings are limited in their generalizability, since the focus was on one population and one disease.

Sivell et al. (2008) completed one of the most comprehensive systematic reviews located (n=19 studies), using a multitude of genetically linked diseases and that included both quantitative and qualitative methodologies. The goal of the review was to illustrate how individuals perceive, construct, and interpret genetic risk and thus make corresponding decisions about prevention and care. Although Sivell et al. cited evidence that supported the findings of Katapodi et al. (2004), that risk perception is not easily quantifiable due to its variability, they emphasized the multidimensional nature of risk perception and concluded that risk is a social process influenced by many factors.

The majority of studies reviewed by Sivell et al. (2008), in contrast to Katapodi et al. (2004), reported that individuals overestimated their risk, particularly if they had a relevant family history. Of these 16 studies, six found that participants had difficulty with understanding objective risk, with only three studies reporting good recall of objective risk. A more accurate perception of risk was noted post-genetic counselling (n=12 studies). In studies it was noted that risk perception did not change over time. By comparison to the general population, participants in three studies perceived themselves as being at higher risk, with only one study reporting that participants perceived their risk as similar to that of the general population.

Sivell et al.'s (2008) review also included nineteen studies that explored how individuals constructed their meaning of risk and made decisions accordingly. Evidence cited strongly suggested that the perception of risk is transient in nature and contingent on subjective interpretations of risk and heuristics, or "common-sense practical knowledge," rather than on objective estimates. Similar observations were noted in other studies (e.g., Cameron et al., 2009; Shedlosky-Shoemaker et al., 2010;). The consensus of these

studies is that perception of risk is shaped and reshaped in relation to personal theories of inheritance: the factors that help individuals construct their perception of risk include past experiences, disease patterns, environment, occupation, diet, stress, physical likeness of other affected persons, and family history. This review provides a glimpse into how individuals understand their risks. It also, however, solidifies the point that examining why risk perception is high or low may not be as relevant as examining the meaning those individuals assign to being at-risk, a point that is illuminated in qualitative studies of risk perception.

Although the review conducted by Sivell et al. (2008) provides valuable insights into the subjective nature of risk, the authors have acknowledged its limitation. Firstly, despite the inclusion of other genetic conditions, the majority of cited studies ($n=38$) were on BRCA 1/2, which limits the generalizability of findings to other genetic conditions (Sivell et al.). Secondly, methods and instruments that were used to measure risk were inconsistent and employed at varying intervals (Sivell et al.). Thirdly, the review contained limited longitudinal studies that would provide valuable insights into evolving perceptions of risk (Sivell et al.). Fourthly, as with the review by Katapodi et al. (2004), this meta-analysis included cross-sectional studies that do not give a full account of the genetic testing process; this may account for the inconsistencies in the literature on risk perception.

Some researchers offered insights beyond the preceding methodological critiques as to why inconsistencies in risk perceptions exist. Some suggest that, because many of the participants in these studies have lived in these families for years, they have been exposed to an affected relative. Thus, they do not perceive themselves as being high risk;

having the disease is part of their expected life trajectory (Katapodi et al., 2004). Others may have grown accustomed to the uncertainty in their lives and have accepted this as a part of their everyday norm. Meanwhile, some may be falsely reassured with an indeterminate genetic test result (Dorval et al., 2005; Grover et al., 2009). And, finally, as was the case with the review by Katapodi et al., the findings fail to take into account how individuals understand and attribute meaning to being at-risk (Sivell et al., 2008). The lack of attention to this critical social constructivist framework supports the further need for research to explore how risk perception and coinciding decisions regarding genetic testing, disease prevention and management are influenced by laypersons' everyday life experiences and interactions, particularly with experts.

Representations of Risk: Laypersons and Experts

Emerging literature on genetics suggests that-risk perception is an evolving social process that is socially constructed. Risk is not a concept that exists in a silo, or one that is easily quantifiable, but one that is influenced by interactions with other at-risk individuals and experts in the field of genetics. Thus, in order to understand how representations of risk are conceptualized and subsequent responses formulated, it is important to review the literature that examines how laypersons and experts perceive risk. Only by understanding how individuals construct their own ideas about risk will health care providers be able to develop collaborative and effective plans of care. This section starts with a review of the literature on laypersons' construction of risk, and how these constructions are followed by discussion of how laypersons' ideas about risk are influenced by that of experts' constructions of risk. The section concludes with an overview of the cardiovascular literature on risk construction.

Laypersons' Construction of Risk

Laypersons construct their ideas about risk in reference to factors outside the realm of numerical labels, drawing on the subjective nature of risk and their personal beliefs about inheritance (Cameron et al., 2009; McAllister, 2002; Norris, Spelic, Synder, & Tinley, 2009; Shiloh & Saxe, 1989; Smith, Michie, Stephenson, & Quarrell, 2002). Thus, in order to plan care for at-risk populations that will be reflective of their needs, health care professions need to have as full an understanding as possible of the varied methods in which laypersons construct, communicate, and perceive risk, and their coinciding behavioural responses (d'Agincourt-Canning, 2001; Cameron et al., 2009; Cox, 2001, 2003; Cox & Starxomski, 2004; Etchegary, 2006a, 2006b; Frich, Ose, Malterud, & Fugelli, 2006; Hallowell, 1999; Klitzman, Thorne, Williamson, & Marder, 2007; Lerman, Croyle, Tercyak, & Hamann, 2002; McAllister, 2003; Murray, Manktelow & Clifford, 2000; Rees, Fry, & Cull, 2001; Sivell et al., 2008).

Evidence suggests that personal beliefs about inheritance and risk are constructed by drawing on factors that create mental representations of one's health, such as past experiences, disease patterns and causation, and growing up in an at-risk family (d'Agincourt-Canning, 2005; Etchegary, 2006b, 2010; Etchegary & Perrier, 2007; Hall, Suakko, Evans, Qureshi, & Humphries, 2007; Hallowell et al., 2006; Hunt, Davison, Emslie, & Ford, 2000; Hunt, Emslie, & Watt, 2001; Marteau, Kinmonth, Pyke, & Thompson, 1995; McAllister, 2002, 2003; Ponder, Lee, Green, & Richards, 1996; Senior, Smith, Michie, & Marteau, 2002; Weiner, & Durrington, 2008). Some studies have found that individuals perceive their risk in relation to multiple lifestyle, environment, and psychosocial factors, and not as having a solely genetic basis (Shiloh & Saxe, 1989;

Walter, Emery, Braithwaite, & Marteau, 2004). Others have noted specifically that ideas about risk are related to the varying penetrance of genetic conditions, the knowledge regarding modifiable factors on genetics, the ability to control the trajectory of the disease, the difficulty with understanding objective risk, and the efforts to cope and manage the disease (Cameron et al., 2009; Etchegary, 2010). In keeping with these research findings, many scholars acknowledge the importance of having a good understanding of how laypersons construct their meaning of risk in order to provide individualized patient and family care (d'Agin-court-Canning, 2005; Cox, 2003; Cox & McKellin, 1999; Etchegary, 2006a, 2006b; Ozanne et al., 2010; Parsons & Atkinson, 1992; Richards, 1993; Shiloh, Gerad, & Goldman, 2006).

D'Agin-court-Canning (2005) found that experiential knowledge (empathetic and embodied) was critical in shaping risk perception in individuals at high risk for breast and ovarian cancer (n=28 carriers; n=11 non-carriers). Empathetic knowledge (knowledge derived due to close social relations with individuals who have experienced a particular phenomenon) and embodied knowledge (knowledge obtained by actually living through the phenomenon) were described by d'Agin-court-Canning as co-existing in a symbiotic relationship. That is, both types of knowledge are shaped or reshaped in response to each other and variable experiences. An individual's perception of their own risk is dependent on the components of their knowledge at a particular point in time, reflecting the transient and contextual nature of risk as noted by other researchers (Shedlosky-Shoemaker et al., 2010; Sivell et al., 2008). These findings support previous research that highlights the subjective nature of risk, the impact that living in a family with a genetic linked disorder has on shaping one's perception of risk, and the how one makes life decisions (Binedell,

Soldan, & Haper, 1998; Cox, 2003; Cox & McKellin, 1999; Etchegary, 2006b; McAllister 2002, 2003).

Similarly, Cox, and McKellin (1999) argued that objective risk alone is not sufficient to explain the complexity of risk perception. Health care providers need to understand laypersons' habitual as well as rational understandings of risk by moving beyond traditional Mendelian theories of inheritance to incorporate the social, biographical, and temporal factors that exist within the context of one's everyday life. The authors' prospective qualitative study examined how individuals living in a family with a history of HD construct risk. Participants' narratives described individuals' sense of risk as being transient in nature, going from being non-existent to a heightened state of awareness or relevancy. Relevancy of risk is attributed to a critical event such as a sibling being diagnosed, a relative dying from the disease, or the person approaching the age of onset. This transient nature of risk has also been reported for patients living with cardiac disease (e.g., Brorsson, Troein, Lindbladh, Selander, & Widlund, 1995) and in other studies on HD (Etchegary, 2006a, 2006b, 2010).

Recent work by Etchegary (2010) examining living at risk for HD referred to the conditions upon which laypersons' ideas about risk are constructed as "zones of relevancy." These zones of relevance sparked participants to rethink their personal perception of being at-risk. Zones of relevancy included such things as stage of life (e.g., nearing age of disease onset, marriage, and reproduction choices); family history of HD; news of another affected relative; one's intuition or belief as to their genetic status; and the experience of a critical event in one's life (e.g., onset of symptoms, starting school).

It is the transient and subjective nature of the zone of relevancy as described by Etchegary that helps us understand how risk construction is an evolving social process.

The role of heuristics, subjectivity, and life-relevancy in shaping one's risk perception is also explicit in Sanders, Campbell, Donovan, and Sharp's (2007) study of the general public's accounts of hereditary risk perception. While participants were open to health education messages, they drew upon multiple sources of information that were personally relevant and accessible at the time in order to construct, control, and rationalize their personal sense of risk (that is, rather than relying solely on genetic information). Key sources of relevant information included family history, personal theories of inheritance, existing social relationships and, at times, existing scientific knowledge. Noteworthy is the fact that although participants were aware of the function of genetics in disease causation, they often downplayed its role or rejected genetic causes in favor of lifestyle and environmental factors in order to gain some sense of control over their risk. Despite this, many did not state that they would make the suggested behavioural changes. The authors suggested that this might be attributed to the fact that irrelevant knowledge or that which may still be considered abstract causes a sense of disembodiment, having no personal meaning and thus requiring no action.

A study by Kenen, Arden-Jones, and Eeles (2003) on women at risk for hereditary breast and ovarian cancer (HBOC) supports Sanders et al. (2007), as to the significant role heuristics plays in understanding one's risk and disease management. Kenen et al. also suggested that laypersons' perceptions of risk are strongly shaped by observing similarities amongst family characteristics such as shared phenotypic features and shared personality traits, which has been reported by other scholars (Finkler, 2001, 2005). Finkler

(2001) refers to the role of shared familial characteristics in defining risk as the medicalization of family and kinship. That is, people are considered “at-risk by association”; living in a family at-risk means one will most likely develop the condition. The participants in the study by Kenen et al. (2003) also considered pieces of family stories as ingredients to formulate their personal theories of inheritance and coinciding sense of risk. Woman referred to physical characteristics or circumstances of an affected person that was representative of their own current state of affairs, regardless of statistical probability, to shape their risk perception. For others, ideas surrounding risk were interpreted in relation to a critical event, such as the death of a family member that stood out in one’s mind. For those with a heightened sense of fear, the illusion of having some control over their risk and disease management was an essential coping mechanism regardless of scientific merit. As had been previously reported by Marteau et al. (1999), Kenen et al. concluded that information about genetics is not processed in a silo but is superimposed on pre-existing perceptions of disease causation. That is, newly acquired information is understood in light of pre-existing personal frameworks in the lay construction of risk. Understanding lay constructions of risk therefore can be challenging, given the complex nature of many diseases, as well as competing media and familial perceptions of disease causation. Kenen et al. (2003) and Sanders et al. (2007) support the work of other studies on HNPCC (McAllister, 2002, 2003) and HD (Cox, 2003, Etchegary, 2006a, 2006b, 2010; Smith et al., 2002) as to the vital role that lay models of inheritance play in the construction of risk.

Who are the “Experts”?

The burst of technological advancements in recent decades has resulted in a proliferation of disciplines-deemed “experts” (Foucault, 1977). These experts are considered to have the most up-to-date knowledge and experience within a designated field. In the context of genetics, this is a role that has traditionally been held by the scientific community and, more recently, other health care providers, such as genetic counsellors. Lippman (1994) intentionally moves away from the traditional scientific description of experts to depict them as storytellers who draw upon experiences, information, and knowledge to construct explanations and stories for diseases of interest, in a way that reflects not only their own views but also the current social and cultural context. Most of the literature that supports a social construction of risk perception perspective draws on Lippman’s definition of experts and extends it to include the layperson as expert. Thus, the quandary that exists in the literature is how to account for the knowledge, experiences, and stories of laypersons that are critical to understanding how people’s perception of risk is shaped and managed, given that a layperson’s perception of risk is often different from that of experts (Slovic, Fischhoff, & Lichtenstein, 1980). I argue, following the work of critical theorists such as Lock and Nguyen (2010), that by using a social constructivist lens, health care providers can appreciate and merge the varying sources of expertise in order to understand how risk is constructed. To facilitate this discussion, an examination of the literature that highlights the existing contentions surrounding laypersons’ versus expert’s knowledge, as well as how laypersons “trust” in technology and experts influence their risk perception, is helpful.

Expert and lay knowledge of genetics. One of the foremost differences between lay compared to expert knowledge surrounding genetics is, of course, the layperson's basic knowledge of the science of genetics. Some researchers have described laypersons as lacking an understanding of the core concepts used in genetics such as basic anatomy and physiology of genetic material, relevant terminology, and probability of transmission (Condit, 2010; Lanie et al., 2004; Richards, 1996).

Research that emphasizes the lack of knowledge of genetic science on the part of non-experts unfortunately supports the assumption that laypeople who fail to follow the advice of experts do so because of a lack of understanding of the science that guides the decision or advised behaviour. Some critical theorists refer to this as the "knowledge deficit" framework for understanding lack of uptake of genetic testing services (Hansen, Holm, Frewer, Robinson, & Sandoe, 2003). Of those who adhere to the knowledge deficit understanding of genetic testing decisions, education of laypeople by experts is proposed to resolve the knowledge deficit (Hansen et al., 2003).

Although the literature depicts a clear discrepancy between laypersons' and experts' understanding of the concept of risk, this does not equate a knowledge "deficit" of the layperson. Rather it reflects the diversity of the meaning assigned to risk based on competing types of knowledge; scientific versus embodied and practical knowledge. In fact, some researchers persuasively argue that laypersons do have a practical knowledge of genetics and are able to make decisions surrounding predictive genetic testing (e.g., Binedell et al., 1998). It is this practical knowledge that leads to the development of lay theories of inheritance in lieu of traditional biomedical models of inheritance (Richards, 1993) and that also disputes the proposed knowledge deficit model (Einsiedel, 2000).

In contrast to the knowledge deficit theory, other researchers have emphasized that discrepancies between lay and expert perceptions of risk are not strictly dictated by a deficit in knowledge, but linked to how one assimilates presented knowledge into existing personal frameworks of beliefs and values about genetics (Cox & McKellin, 1999; Ozanne et al., 2010; Parsons & Atkinson, 1992; Richards, 1993; Shiloh et al., 2006). Given the fact that individuals have different representations of disease causation and what it means to be at-risk, it does not suffice to provide information; rather, we need to explore further how these representations evolve in order to decipher and clarify any misconceptions (Shiloh et al.), to move towards a more collaborative understanding of risk, and in order to facilitate informed decision-making (Cox, 2003; Cox & McKellin, 1999; Etchegary, 2006a; Parsons & Atkinson, 1992; Richards, 1993; Ozanne et al., 2010). Joffe (2003) further critiques the knowledge deficit theory in that it fails to take into consideration the diverse social contexts (i.e., symbolic, organizational, and political), in which laypersons construct their realities in lieu of the idea that they are a rational actor who will always engage in risk prevention activities.

Ozanne et al. (2010) offer some insights into how experts influence laypersons' beliefs about being at risk for BRCA1/2. They found that although physicians communicated risk information in a way that coincided with client preferences, the physicians had difficulty estimating the participants' perceptions of lifetime risk. The findings questioned the abilities of experts to tailor risk information to reflect individuals' objective risk and facilitate informed decision-making. Also, having determined that counselling had no effect on women's preferences for preventive interventions, the authors queried whether experts indeed clearly understood the pre-existing values and

ideas that guide participants' decisions about engaging in healthy behaviours and predictive genetic testing, as had been previously suggested by Sivell et al. (2008).

Another qualitative study noted that risk communicated by experts was more meaningful when family histories were taken into account during risk assessments (Hall, Saukko, Evans, Qureshi, & Humphries, 2007). In that study, participants' ideas about risk and subsequent management of their condition was found to be more in alignment with experts when health care providers took the time to listen to and explore participants' concerns about being at-risk.

Laypersons, experts, and technology: Who trusts whom? Research on differing perceptions of risk between laypersons and experts has demonstrated that there is a disconnect between the two sets of ideas about risk that has led to a sense of mistrust, predominantly in relation to new, innovative technologies, such as predictive genetic testing. Giddens (1991) argued that lay perceptions of modern technologies swing between support and doubt that are shaped by the discourse surrounding the risks and benefits of the technology under question. It is, therefore, no surprise that a difference between lay and expert understandings of risk leads to mistrust, given that new technologies such as predictive genetic testing and subsequent practices are controlled by the experts (Giddens, 1991), which in itself can spark a sense of uneasiness due to a lack of control for those designated to be at-risk.

Jallinja et al.'s (1998) poll on attitudes towards genetic testing (n=1 169) within the general population in Finland supports Giddens' perspective on lay perceptions of modern technologies. Participants spoke of ambiguity regarding the long-term repercussions of predictive genetic testing, such as eugenics. This uncertainty about

eugenics and the long-term implications of the technology fostered mistrust in the general population towards the advice of experts. Some participants in the study by Jallinja et al. disagreed with this opinion and took the approach that laypersons need to have confidence in the abilities of experts and technology if engaged in the predictive genetic testing process, in keeping with Giddens' (1991) description of modernity that holds that trust is a process of mutual disclosure amongst laypersons and experts (p.6).

Cardiovascular Literature: Being At-Risk for a Genetically Linked Heart Disease

Little attention has been paid to how individuals at risk for a genetically linked cardiac disease understand and manage their own risk status. The majority of research related to coronary heart disease (CHD) and genetics is quantitative in nature, emphasizing gene identification, symptom management, and behavioural modifications. The management of CHD is discipline driven and comes from several theoretical standpoints: a genetics framework, a focus on the specific factors that cause CHD (e.g., diet and exercise), and a consideration of a broader range of risk factors that lead to CHD (e.g., access to resources) (Weiner & Martin, 2008). In addition to using competing frameworks about CHD, each disciplinary approach focuses on management approaches, prevention strategies, and the different causes of heart disease. Finally, each approach has projected different visions for the future of CHD reflective of the disciplinary knowledge that has shaped the particular model (Weiner & Martin, 2008). All of the above factors are apparent in the literature that describes the "coronary candidate" (that is, the typical individual that develops cardiovascular disease, i.e. obese, unemployed, smoker, and inactive) and the use of biomedical knowledge to construct risk.

The coronary candidate. While most of the literature on CHD does not attend specifically to the social construction of risk among patients or families living with cardiac disease, messages about “risk” that have been constructed within the medical model of cardiac disease are fairly clear. In particular, an analysis of the notion of a “coronary candidate” is a predominant thread in the literature, and illustrates how the concept of being at risk for coronary disease is constructed, conveyed, and taught to patients. Critical theorists have commented on how the notion of a “coronary candidate” in literature conveys specific meanings regarding being at risk for and managing heart disease (Davison, Frankel, & Smith, 1989, 1992; Davison, Smith, & Frankel, 1991; Emslie, Hunt, & Watt, 2001; Hunt et al., 2000; Walter & Emery, 2005)

Davison et al.’s (1991) study was the first major study to explore the social and cultural context of CHD in relation to the notion of a coronary candidate. According to that study, the typical coronary candidate is one who is obese, unemployed, a smoker, and inactive. For laypersons, the mental representations of the coronary candidate function to provide retrospective explanations of heart illness for one’s self and others as well as a means to predict others’ and one’s own risk of illness and death from heart disease. In order to assess one’s candidacy for heart disease, the layperson assumes the role of the lay epidemiologist, deciphering various sources of evidence, such as direct observation of cases of heart disease, stories about heart disease, scientific knowledge, and the media in relation to their visible factors (Davison et al., 1991, 1992). Davison et al. (1991) explain that the coronary candidate has four opposing outcomes regarding the status of being a candidate for heart disease: (1) the victim of heart trouble; (2) the lucky survivor, who has characteristics of coronary candidacy but no heart disease; (3) the survivor; and (4) the

unlucky victim or the last person you would expect to have heart disease. Individuals draw on these outcomes in order to rationalize existing beliefs surrounding coronary candidacy and anomalies.

In his description of the coronary candidate, Davison et al. (1989, 1991, 1992) refer to the idea of fatalism to describe how individuals conceptualize anomalies or poor cardiac outcomes that are not explained by behavioural outcomes. This idea can be utilized when considering the case of the unlucky victim who is not obese, eats healthy, does not smoke, and exercises routinely but who has a myocardial infarction versus the lucky survivor: a 70-year-old who smokes, drinks, and eats a diet high in salt and fat but who has no medical problems. The role that fatalism plays in understanding being at risk for FH has been explored in several studies (Hunt et al., 2000; Senior, Marteau, & Peters, 1999; Weiner, 2009).

Senior et al.'s (1999) study of parents who participated in neonatal screening for familial hypercholesterolemia (FH) found that those who interpreted the test as only identifying an elevated cholesterol level perceived the disease as familial but modifiable with diet and hence less threatening. In contrast, when the results were thought to be genetic in nature, then the elevated cholesterol was viewed as more threatening and not modifiable. The authors suggested that the latter view could lead to a fatalistic approach that negatively affects one's adherence to behavioural changes. Senior et al. concluded that when presenting genetic information, it is important that health care providers take time to understand laypersons' models of disease causation in order to plan realistic management strategies and avoid a fatalistic approach.

Weiner's (2009) study on participant experiences of living with coronary heart disease (CHD) reported findings similar to those of Davison et al. (1989, 1991, 1992) in terms of participants' descriptions of models of inheritance and outcomes. Participants were found to oscillate between various accounts of disease causation for FH and CHD that fit within their existing personal theories of inheritance at the time. That is, CHD was not framed purely from a genetic standpoint but in regards to laypersons' understanding of the coronary candidate. While some participants did recognize that staying healthy was their responsibility, they did not see themselves as being responsible for having high cholesterol. Hence, having a genetic disposition for FH did not necessarily infer a fatalistic attitude. This pragmatic approach gave participants the option to relinquish responsibility for their carrier status and negative health outcomes while at the same time maintaining their dedication to a healthy heart lifestyle, congruent with heart disease management and defiant of the vision of the coronary candidate.

The notion of coronary candidacy has also been gendered as a male concept (Emslie et al., 2001). Emslie et al. (2001) examined the relationship between perceptions about heart disease, gender, and social class. Sixty-one men and women from various social economic situations were included. Participants' narratives revealed three key themes: (1) the mechanical versus the emotional heart, (2) the gendering of coronary candidacy, and (3) accounts of heart problems amongst women. The first theme summarized participants' perceptions of the heart as serving both a mechanical and emotional role. The gendered nature of coronary candidacy was highlighted in participants' stories that depicted men as the typical coronary candidacy victim in keeping with Davison et al., 1991, 1992. The final theme described participants' accounts of heart

disease in the family. Recollections of men with heart disease tended to focus on graphic accounts of mortality, whereas descriptions by women with heart disease centered on morbidity and framed it in light of a chronic illness that limited productivity. The authors concluded that laypersons' perceptions of heart disease are framed and continue to be framed within the strong historical context of a male disease.

Using biomedical knowledge to construct risk. Another common thread throughout the literature, similar to but outside discussions of the coronary candidate, is that laypersons and experts draw upon traditional biomedical models of disease causation to inform themselves and others about the nature of CHD and subsequent management. These biomedical models are a reference point for risk construction and provide the framework through which many individuals begin to develop their sense of risk; however, it is the context of one's experiences that shapes and reshapes the meaning of being at-risk. Furthermore, ideas about risk are intermittently superimposed on biomedical models of cardiovascular heart disease as an individual gives meaning to their risk status, makes lifestyle changes, and/or dismisses ideas about risk. This is evident in the literature that explores the construction of risk in other genetic linked diseases (Braithwaite, Emery, Walter, Prevost, & Sutton, 2004; Frich et al., 2006; Hunt et al., 2000; van Maarle et al., 2003; Walter & Emery, 2005; Walter et al., 2004; Weiner, 2009; Weiner & Durrington, 2008).

Walter et al. (2004), in a systematic review of qualitative research that examined laypersons' understandings of familial risk, included one study on FH (the study by Brorsson et al., 1995). Their review of the study by Brorsson et al. supported previous findings that laypersons and experts do have varying perceptions of risk and, more

importantly, that risk perception is something that develops parallel to ongoing assessments of one's vulnerability to the disease, as was supported by subsequent studies (Farrimond, Saukko, Qureshi, & Evans, 2010; Finch et al., 2006; Walter & Emery, 2005). In other words, a layperson's risk assessment, as described by McAllister (2002, 2003), is informed by and grounded in a personalizing process that includes an awareness of the saliency of one's own family history, a reflection on personal experiences, and the development of personal theories of inheritance. For some, risk perception coincides with a sense of fatalism or luck as described by Davison et al., 1989, 1991, and 1992, but for others risk is defined in terms of the consequences of and success in managing the disease.

In a follow-up study to the original (2004) review, Walter and Emery (2005) used a comparable cohort to that of Walter et al. (2004). Although they reported related results to the 2004 study, several new themes emerged that strengthened their previous theories about risk—themes that were similar to results that had been reported in an earlier study by Hunt et al. (2001). Walter and Emery attributed participants' ideas about risk as being influenced by the number of affected relatives, whether the participants had witnessed the illness, the strength of their personal relationships with the affected person, and their perceived likeness to the affected person (i.e., personality, mannerisms, and physical appearance). Participants also described a type of bargaining to decrease their own sense of vulnerability and to justify not adopting a healthy lifestyle by the use of counter examples. This finding is similar to Davison et al's (1991, 1992) descriptions of the lucky survivor. According to Walter and Emery, participants believed they could control their risk for the disease by modifying their behaviour and lifestyle.

Finch et al. (2006) also described layperson's perceptions of vulnerability for FH. For these participants, their sense of vulnerability was shaped in relation to ideas about genetics and inherited risk. Participants identified a two- step process undertaken to tease out their susceptibility for heart disease. This approach taken by the participants in the study by Finch et al. supports the findings of Walter et al. (2004). The first step taken by participants included an appraisal of factors that would serve as a reference point to guide one's risk assessment, such as family history, patterns of disease (i.e., morbidity and mortality rates), and personal experiences. These factors were all critical in order to configure mental representations of the families' coronary candidacy. These mental representations were then used as a framework to construct participants' own evolving sense of vulnerability in response to biographical and social contexts. In contrast to earlier studies (Walter & Emery, 2005), participants did not make reference to physical or mental similarities to relatives as being imperative to risk construction. Age of onset of disease was the primary factor drawn upon to solidify vulnerability to heart disease. The significance of age as an appropriate evaluative measure of cardiac mortality and risk has been noted in two earlier studies (Emslie et al, 2001; Weiner & Durrington, 2008).

A Canadian study (Angus et al., 1995) explored the experiences of individuals as they tried to assess their own risk for coronary heart disease in lieu of visible physical signs of heart disease. Participants described difficulty understanding how they could be labeled "at-risk" for coronary heart disease when they had no visible symptoms for the disease. Accepting the label of being at-risk meant modifying lifestyle factors thought to cause CHD. In order to deal with this "abstract" sense of risk, participants sought evidence to support the fact that they were indeed at-risk. Hence, many looked to their

family histories, lifestyle habits, and characteristics of the coronary candidate to negotiate ideas about risk. Others used medical hermeneutics to conceptualize their risk. That is, they drew on the knowledge of physicians and diagnostic tests in an effort to quantify their risk. Finally, they measured their risk according to whether they had had a myocardial infarction or not. For some, they saw this critical event (the myocardial infarction) as the impetus for behavioural changes and as altering their future. These findings might explain why individuals who have no outward physical signs of a genetic condition do not engage in behavioural management regimes. That is, one's "genes" are something invisible and abstract, requiring no immediate intervention. It is only with the onset of physical symptoms (e.g., chest pain, shortness of breath) that these at-risk individuals start to reconceptualize what it means to be at-risk. This reliance on visible physical factors as a measurement of one's risk has been reported in several other studies (Chan, Lopez, & Chung, 2012; Marteau et al., 1995).

Hunt et al's (2000) study, which spanned nine years, found that having a family history of heart disease was a significant factor in the construction of a layperson's perceptions of risk. Over time the meaning of living with heart disease in these families was continuously juxtaposed against other risk factors in order to make behavioural decisions. Family history has been noted by other researchers as being a more important indicator of coronary candidacy than epidemiological trends and/or invisible measures such as blood pressure or cholesterol (Marteau et al., 1995; Ponder et al., 1996). By comparison, one systematic review of the literature on perceptions of risk factors for heart disease identified lifestyle, stress, and heredity as having the biggest impact on one's perception of being at risk for heart disease (French, Senior, Weinman, & Marteau, 2007)

How individuals cope with the news that they are at high risk for CHD has also been examined in the literature. Farrimond et al. (2010) explored how individuals conceptualized and responded to the news that they were at high risk for CHD. A variety of coping strategies were employed to decrease the sense of susceptibility to heart disease and to retain health status. Participants compared themselves to the traditional coronary candidate. Some evaluated their risk of getting heart disease in light of more pressing factors that required their attention. Others normalized their risk and prescribed treatments (i.e., cholesterol-reducing medications) in terms of the aging process or made downward comparisons, where they compared themselves to others less fortunate in order to judge their own situation. The contextual nature of one's vulnerability was another key theme. Participants emphasized the symbolic representations of aging and CHD in reference to their sociocultural environment. Vulnerability to CHD was framed in reference to the physical context of their wellbeing, identified risks status, and expected life trajectory such as age, in order to make decisions surrounding their risk. The final theme reflected participants' efforts to retain and embody a healthy identity by adhering to any recommended behavioural and lifestyle changes such as diet and exercise. Findings suggested that heart prevention programs needed to be context-sensitive to the aging process and the sociocultural context in which individuals at risk for heart disease exist. The interplay of social and cultural meanings of CHD disease was also evident in several other cardiovascular studies (Emslie, Hunt, & Watt, 2003; Hunt et al., 2001; Murray et al., 2000).

From this discussion it is evident that ideas about being at risk for CHD are contextual in nature. In order to personalize the meaning of being at-risk, individuals take

a pragmatic approach. That is, they juxtapose various sources of scientific knowledge against their own experiences. It is this juxtaposition that facilitates individuals in decision-making as they move through the predictive genetic testing process.

Predictive Genetic Testing Decision-Making Process

There have been copious studies that have attempted to identify key factors influencing decisions surrounding the predictive genetic testing process. While some studies reported that a positive correlation exists between risk perception and making the decision to participate in predictive genetic testing (Armstrong et al., 2000), others suggested that this is not always the case (Cameron, et al., 2009; Shiloh et al., 1998), arguing instead that a heightened sense of risk could in fact lead to disengagement or refusal to participate in the process (McAllister, 2002). There is a large body of literature that explores the factors that impact the decision to participate in predictive genetic testing or not. However, the literature does not focus on the social process of decision-making in genetic linked conditions but instead looks at how specific variables (e.g., age) impact the decision-making experience. This gap fragments our understanding of the predictive genetic testing decision-making process. The following discussion begins with an overview of the literature that has explored why people engage or disengage in predictive genetic testing, and it is followed by an examination of existing qualitative research that highlights the contextual issues that influence the predictive genetic testing decision-making process.

Why Do People Participate in Predictive Genetic Testing?

There is a large body of research that attempts to discover key factors that motivate people to participate in the predictive genetic testing process. Findings suggest that one of the primary motivators is an increased awareness of one's risk status as a result of growing up in a family with a history of a genetic linked disease (Armstrong et al., 2000; Cox, 2003; d'Agincourt-Canning, 2005; Etchegary, 2006a; McAllister, 2002; Norris et al., 2009) or having a first-degree relative affected with the disease under investigation (Lerman, Daly, Masny, & Balshem, 1994). Another key motivating factor is a sense of relational responsibility (d'Agincourt-Canning, 2001; 2006a, 2006b; Etchegary, 2006a; Etchegary & Fowler, 2008; Hallowell, 1999; Hallowell et al., 2006; Klitzman, Thorne, Williamson & Marder, 2007) that extends to include concern for other family members' risk status, particularly that of children (Andersen, Oygén, Bjorvatn, & Gjengedal, 2008; Armstrong et al., 2000; Binedell et al., 1998; Bruno et al., 2004; Chivers Seymour, Addington-Hall, Lucassen, & Foster, 2010; d'Agincourt-Canning, 2006a, 2006b; Decruyenaere et al., 1997, 2003; Douma, Aaronson, Vasen, & Bleiker, 2008; Evers-Kiebooms et al., 2002; Esplen et al., 2001; Etchegary, 2006a; 2009; Haddow, 2009; Lerman et al. 1994; Meiser et al., 2007; Norris et al., 2009; Smart, 2010; Smith et al., 2002; Weiner & Durrington, 2008).

Critical events, such as the onset of disease symptoms or the death of a family member, have also been found to increase risk awareness and thusly motivate individuals to participate in the predictive genetic testing process (Armstrong et al., 2000; Etchegary, 2006a, 2010; Klitzman, Thorne, Williamson & Marder, 2007; Smith et al., 2002). This has been explained as relieving feelings of uncertainty about carrier status (Armstrong et

al, 2000; Binedell et al., 1998; Christiaans et al., 2009; d'Agincourt-Canning, 2006b; Decruyenaere et al., 1997, 2003; Douma et al., 2008; Evers-Kiebooms & Decruyenaere, 1998; Evers-Kiebooms et al., 2002; Lerman et al., 1994; Meiser et al., 2007). Some theorists describe participant choices to participate in predictive genetic testing as a means of obtaining information about disease management and gaining easier access to resources such as screening protocols, prophylactic surgeries, and lifestyle management (Armstrong et al., 2000; d'Agincourt-Canning, 2006a, 2006b; Esplen et al., 2001; Lerman et al., 1994; Meiser et al., 2007). Other theorists have reported that knowing one's risk status is in itself a coping mechanism—an opportunity to acquire a sense of control over life and to plan for future life experiences such as marriage, reproduction, finances, and employment (Binedell et al., 1998; Decruyenaere et al., 1997; Etchegary, 2009; Evers-Kiebooms & Decruyenaere, 1998; Evers-Kiebooms et al., 2002; Lerman et al., 1994). Some studies found that the decision to participate in predictive genetic testing was based on recommendations of physicians (Esplen et al., 2001; Madlensky, Esplen, Gallinger, McLaughlin, & Goel, 2003), in response to genetic counselling (Christiaans, Birnie, Bonsel, Wilde, & van Lange, 2008), and to advance research (Esplen et al., 2001).

From this overview, it is quite evident that decisions to undergo predictive genetic testing are contingent upon a multitude of factors, some of which have strong historical roots in biomedical models of inheritance, and others that reflect the contextual and experiential nature of risk. Unfortunately, biomedical and social motivators for participating in predictive genetic testing have tended to be discussed in isolation from each other in the literature. As a result, health care providers are neglected a full understanding of how biomedical models of disease causation are superimposed and

integrated into one's everyday "practical" knowledge and life experiences in order to understand, cope, and make decisions about their own risk.

Why Do People Refuse to Participate in Predictive Genetic Testing?

Despite the abundance of research that explores factors influencing an individual's willingness to engage in the predictive genetic testing process, there is a scarcity of literature asking whether and how individuals make the decision to forego predictive genetic testing. The principal reasons cited as to why at-risk people do not participate in predictive genetic testing are as follows: concerns about being stigmatized, concerns over the financial impact of having tests, and concerns about loss of health insurance (Armstrong et al., 2000; Binedell et al., 1998; Klitzman, Thorne, Williamson, & Marder, 2007; Landsbergen, Verhaak, Kraaimaat, & Hoogerbrugge, 2005; Lerman & Shields, 2004; Meiser et al., 2007; Quaid, & Morris, 1993; Weiner & Durrington, 2008).

Additionally, distress anticipated with the predictive genetic testing process was noted by some studies to be a deterrent to participation (d'Agincourt-Canning, 2006b; Klitzman, Thorne, Williamson & Marder, 2007; Landsbergen et al., 2005; Lerman, et al., 1998; Meiser, 2005; Smart, 2010). In several studies participants were apprehensive about their ability to cope with the uncertainty of the test result (Binedell et al., 1998; d'Agincourt-Canning, 2006b; Decruyenaere et al., 1997; Evers-Kiebooms & Decruyenaere, 1998; Lerman & Shields, 2004), as well as the ability of other family members to cope with the genetic test result (Binedell et al., 1998; d'Agincourt-Canning, 2006b; Evers-Kiebooms & Decruyenaere, 1998; Klitzman, Thorne, Williamson, Chung & Marder, 2007; Lerman & Shields, 2004; Meiser et al., 2007). Thus, in order to avoid or defuse the distress associated with knowing the test result, many participants declined to

engage in the predictive genetic testing process (d'Agincourt-Canning 2006b; Decruyenaere et al., 1997; Evers-Kiebooms & Decruyenaere, 1998).

Lack of participation in predictive genetic testing has also been attributed to the fact that knowing one's risk status does not benefit certain individuals, including those at risk for diseases considered deadly, with high penetrance and having no known cure, such as HD (d'Agincourt-Canning, 2006b; Binedell et al., 1998; Evers-Kiebooms & Decruyenaere, 1998; Quaid & Morris, 1993), BRCA1/2 (d'Agincourt-Canning, 2006b) and this would also be the situation for ARVC. Anticipated difficulties with disease management was also identified as a deterrent to testing, such as access to resources (Klitzman, Thorne, Williamson, & Marder, 2007) and adherence to screening protocols (Evers-Kiebooms & Decruyenaere, 1998). Other deterrents included having a negative perception of the benefits of technology (d'Agincourt-Canning, 2006b), lack of trust in predictive genetic testing (Meiser et al., 2007), and lack of relevancy at the time—for example, those who have no children to whom to pass the gene on, those who are asymptomatic or those who are young and do not see predictive genetic testing as something important for them at that particular point in time (Landsbergen et al., 2005; Smith et al., 2002; Weiner & Durrington, 2008). For the participants in one study, the fact that life decisions were not contingent on predictive genetic testing results made it a non- priority at the time (Decruyenaere et al., 1997). In order to fully appreciate the contextual nature in which decisions about predictive genetic testing are made, it is imperative to examine qualitative literature that takes a closer look at the decision-making process as opposed to literature that looks mainly at the reasons for choices in isolation from the process of decision-making (Etchegary, 2006a). This includes exploration of the

broader social, political, and familial context in which decisions about genetic testing take place (d'Agincourt-Caning, 2006).

The Decision-Making Process in Predictive Genetic Testing.

The preceding discussion highlights the multitude of factors that may influence one's decision to engage in the predictive genetic testing process at any point. Although research indicates that experiencing a critical life event or having to make life decisions (i.e., marriage, reproduction) triggers engagement in the predictive genetic testing process (Armstrong et al., 2000; Binedell et al., 1998; Decruyenaere et al., 1997; Evers-Kiebooms & Decruyenaere, 1998; Evers-Kiebooms et al., 2002), there is limited research addressing how decisions about genetic testing evolve over time. There is also a dearth of literature that takes a closer look at the contextual nature of the decision-making process for predictive genetic testing. Qualitative research that explores the decision-making process in genetics provides a good lens through which to gain a fuller understanding of how decisions related to predictive genetic testing are made. Threaded throughout much of the qualitative literature are the impacts that personal stories have on the decision-making process and the diverse social contexts in which decisions are undertaken (Boenik & Vander Burg, 2010; Cox, 2003; Etchegary, 2006a; Smith et al., 2002; Taylor, 2005).

Qualitative studies on HD conducted by Cox (2003), Etchegary (2006a), Smith et al., (2002) and Taylor (2005) bring to light how at-risk individuals develop a sense of readiness to participate in the predictive genetic testing process in relation to the context of their everyday lives. In Cox's study participants described the decision to have predictive genetic testing as an evolving conscious awareness that happened over time. For others, this decision was something unconscious and in many regards a non-event—

something that required little thought or discussion but that was accepted as being necessary at the time. This was also reported in Smith et al's (2002) study on HD and Taylor's (2005) research on HD. Unique to Taylor's (2005) study is the inclusion of individuals who made the decision not to be tested at that time (i.e., those who made the decision not to having genetic testing right now but who did not exclude it as an option later in life, and those who were undecided). Participants describe an emergent sense of test readiness that reflects the emotional readiness, informational readiness, and circumstantial readiness of oneself and others to participate in the predictive genetic testing process.

Etchegary's (2006a) research in the province of Newfoundland and Labrador offers many excellent insights into the meaning of being at risk for HD and how decisions to participate in predictive genetic testing, or not, are made. Similarly to Cox (2003), several participants in Etchegary's study described their decision as "a non-decision," requiring little conscious effort. Other participants spoke of the decision to have predictive genetic testing as being self-constraining. That is, the decision to engage in testing was not perceived as an opportunity or choice for them but rather a moral duty to their children. Some participants were described as constantly "re-evaluating the decision." For these participants the decision oscillated between wanting the genetic test and deciding against it, to refusing to have testing and making the decision to be tested. Finally, "test-triggers" such as onset of what was perceived by participants as signs of HD prompted some to request testing.

Scully, Porz, and Rehmann-Sutter (2007), similar to Cox (2003), found that the decision to participate in genetic testing for BRCA1/2, CRC, and HD is a conscious

process that evolves over time. The researchers found that individuals either engage in incremental decision-making for late onset conditions such as HD or make a conscious decision to narrow the temporal depth of field of the issue, particularly when faced with decisions that are time sensitive, such as prenatal testing. Scully and colleagues (2007) noted that these coping mechanisms are strategies not only to maintain a sense of control over the decision process but to manage segments of the decision that participants are emotionally prepared to address and to provide an opportunity to closely examine the situation from a moral standpoint. Hence, when providing care for individuals faced with the decision to have genetic testing or not, it is important to recognize the context of the decision-making process and have resources to support the individuals throughout the entirety of the process. As suggested by Scully et al. (2007), counselling programs need to have time built into them for individuals to reflect on the meaning and consequences of their decisions so their consent is truly informed. Finally, health care providers need to support individuals to align decisions with their personal construction of reality at the time. This means appreciating the complexity of factors influencing the predictive genetic testing decision-making process.

The multifactorial nature of making the decision to engage in the predictive genetic testing process for HNPCC has also been described in McAllister's (2002) Theory of Engagement. Engagement "reflects the degree of cognitive and emotional involvement with cancer risk in individuals from these families, and models the psychosocial process of engaging with cancer risk" (McAllister, 2002, p. 421). The level of engagement is not static; it is contingent upon one's personal perceptions of risk and one's life experiences. The level of engagement oscillates between partial, intense, and disengagement. In that

study, individuals who perceived their risk as low were partially engaged in the process of predictive genetic testing, and hence only engaged at the cognitive level. Participants who perceived their risk as high described some distress as to their carrier status but were intensely engaged at the cognitive and affective levels. Disengagement describes intensely engaged participants who, due to some critical event nonetheless felt it more appropriate to withdraw from the genetic testing process. Critical events and distress were found to be either the impetus to engagement or disengagement. These findings support the fact that in order to support decisions made throughout the process of predictive genetic testing, it is imperative that health care professionals understand the varying personal experiences of those involved in the process and factors that impede engagement in this process.

McAllister (2002) further identifies three interacting moderating factors that at any point can individually or collectively influence risk perception and the level of engagement: (1) casual conditions that facilitate the process such as previous knowledge, personal experience of family history, stage of adult development, gender, family talk, family history of diagnosis delay, and risky age; (2) intervening conditions that impede the process such as life stressors, ignorance of family history, impersonal knowledge of family history, geography, experience with sporadic cancers, and cancer taboo; and (3) individual psychological factors such as personal theories of inheritance, ideas about luck, confidence about carrier status, and coping strategies. This study is significant in that it challenges the assumption that a perception of high risk will result in desirable health behaviour. That is, an increased sense of risk can result in disengagement in the genetic testing process.

Bombard et al.'s (2008) research draws on McAllister's (2002) concept of engagement in order to understand individuals' experiences of being at risk for HD. Participants described doing a mental survey of contextual variables to assess their risk status, consequences, and coinciding decisions. Engagement with the predictive genetic testing process was found to occur in two phases: (1) making meaning of genetic disease and defining it; and (2) validating its threat and personalizing the risks and consequences to oneself and one's family.

Shiloh and Ilan's (2005) work on risk perception and BRCA1/2 is significant in that it reinforces McAllister's (2002) Theory of Engagement. In order to understand how individuals make decisions surrounding predictive genetic testing, one needs to be aware of the collaborative role of moderating factors on this process. A heightened risk perception alone did not predict interest in predictive genetic testing; however, in conjunction with moderating variables that focus on illness prevention, health orientation, and decreasing anxiety level, a willingness to participate in predictive genetic testing was evident. This study clarifies inconsistent findings surrounding the correlation between risk perception and engaging in predictive genetic testing. For example, if the moderator is "emotional reassurance," despite being at low risk, one may engage in health behaviours such as screening to receive this reassurance. Without an understanding of individuals' moderating factors, it is difficult to anticipate responses to predictive genetic testing and to develop counselling services. This study suggests that in order to increase uptake for predictive genetic testing, health care providers need to spend time with these families and identify moderators that are barriers to positive health outcomes. Included in this is an understanding of the moral and sociocultural factors that impact risk perception.

Although limited, there are several studies that specifically draw attention to the moral and sociocultural contextual factors that influence engagement in the genetic testing process (d'Agincourt-Canning 2006a, 2006b; Etchegary, 2006a; Etchegary & Fowler, 2008; Hallowell et al., 2006; Ho, Ho, Chan, Kwan, & Tsui, 2003; Klitzman, Thorne, Williamson, & Marder, 2007). D'Agincourt-Canning's (2006b) narratives of individuals living in families at risk for hereditary breast and ovarian cancer (HBOC) described how they made the decision to engage in genetic testing. For these people the meaning of being at-risk was found to take shape in relation to the self, which includes (1) the embodied self, (2) the familial-relational self, and (3) the civic self. The embodied, or physical, self emphasizes how one constructs ideas surrounding risk in relation to factual information and relationships with other at-risk family members. The familial-relational self reflects the sense of responsibility and duty to get tested for the health of other family members, which has also been noted in several studies on HD (Etchegary, 2006a; Etchegary & Fowler, 2008), on HBOC and HNPPC (Etchegary et al., 2009), and BRCA 1/2 (Hallowell et al., 2006). The civic self refers to the benefits to society at large. It is this interconnectedness between the embodied self, the familial-relational self, and the civic self that shapes the meaning of illness and its significance, and frames one's decisions surrounding the predictive genetic testing process.

There was one cardiovascular study that contributed to our understanding of the factors that can influence participation in predictive genetic testing for individuals at risk for two genetic linked heart diseases hypertrophic cardiomyopathy (HCM) and long QT syndrome (LQTS) (Smart, 2010). Making the decision to participate in predictive genetic testing consisted of weighing the benefit of knowing one's risk status versus the

psychosocial impact that the knowledge will have on their lives. Uncertainty surrounding the reliability of accuracy of the test was evident in those with an inconclusive test and in non-carriers. Despite these reservations, the majority of participants described predictive genetic testing as a way of obtaining family health information and something they would be willing to do. Imperative throughout this process was appropriate communication of risk.

It is clear from the above discussion that making the decision to engage in predictive genetic testing is complex. For some the decision evolves over time; however, for others, it is something that is seemingly more instantaneous in nature, perhaps requiring less thought. Regardless of how the process evolves in order to make the decision to have genetic testing or not, it is clear that this decision reflects the meanings that one assigns to their at-risk status, social interactions, acquired knowledge, life experiences, and ability to cope with the news of their genetic test result. A large part of making this decision is how risk is understood and communicated between health care providers and laypersons.

Risk Communication

There is a large body of literature that has addressed how risk communication influences risk perception. Researchers have noted that obtaining health information about and communicating an individual's risk status is generally understood to be the responsibility of the health care provider (e.g., genetic counsellor). Despite this, there is an appreciation of informal methods of risk communication as the "unsaid" responsibility of the proband, or the first family member identified as being at-risk.

Health Care Providers and Risk Communication

Discussions about using heuristic devices to aid in the communication of risk statistics were pioneered in the 1970s within the field of psychology and were the impetus for health care providers to reevaluate their own methods of communicating risk in appreciation of the subjective nature of “risk” in the context of health (Sjoeberg, Moen, & Rundmo, 2004). Hence, there was a shift from the traditional sender-receiver model of communication, wherein the health care provider simply disclosed the results, to a more holistic model of disclosure (Wilson et al., 2004). Another key shift was the introduction of professional genetic counsellors who, in Canada, are prepared with a Master’s degree in the field of genetics and counselling (Cowan, Morales, Dagua, & Ray, 2008). Given that the role of the genetic counsellor in communicating risk accounts for the largest body of literature on risk communication in genetic conditions, this section will focus on the impact that genetic counselling has on risk.

Genetic counselling refers to the interactive process that occurs between the counsellor and the individual and family at risk for a genetic linked disease. It is the process that provides education, support, and knowledge about one’s at-risk status in relation to family history, inheritance, genetic testing, disease patterns, disease symptoms, and available treatment options. Its overall goal is to facilitate the informed consent process (Biesecker, 1998; Braithwaite, Emery, Walter, Prevost, & Sutton, 2004; Wiseman, Dancyger, & Michie, 2010).

A large amount of literature has explored the role genetic counsellors play in shaping risk perception. A synthesis of this literature is presented in a brief summary of six systematic reviews spanning the last 16 years (Butow, Lobb, Meiser, Barratt, &

Tucker, 2003; Braithwaite et al., 2006; Kaphingst & McBride, 2010; Meiser & Halliday 2002; Sivell et al., 2008; Smerecnik et al., 2009)

Meiser and Halliday's (2002) meta-analysis of 16 prospective and randomized controlled trials (RCT) in women at risk for BRCA1/2 found that genetic counselling decreased anxiety related to perceived risk and led to a 23.4% increase in risk accuracy. A systematic review by Butow et al. (2003) of quantitative research on risk perception and psychological outcomes, post-genetic counselling of women at risk for BRCA1/2, reported results similar to those of Meiser and Halliday (2002), in that improvements in risk perception were noted post-genetic counselling; however, these were found in the short term. In fact, longitudinal studies reported that 22% to 50% of women continued to overestimate their risk up to one year post-counselling. Results varied in relation to psychological outcomes ranging from some reduction in psychological distress to no reduction at all. Unlike the findings of Meiser and Halliday, the study by Butow et al. did not find that psychological distress was related to a change in risk perception.

Braithwaite et al. (2006) conducted a meta-analysis and systematic review of 21 studies on BRCA1/2, HBOC, and CRC, of which 5 were randomized controlled trials and 16 were prospective studies. They examined the impact of genetic counselling on three outcomes: cognitive (i.e., level and accuracy of risk perception and knowledge), affective (i.e., distress, anxiety, depression, and cancer specific worry), and behavioural (i.e., appropriate screening and surveillance). Findings suggested that genetic counselling increased knowledge of cancer genetics and did not cause adverse affective outcomes. The randomized controlled trials reported in this review contradicted earlier findings by Meiser and Halliday (2002), in that the former did not find any reduction in some

affective outcomes post-counselling. Prospective studies up to one year post-genetic counselling offered a different picture; not only was there an increase in the accuracy of risk perception, but an intermittent reduction in cancer worry and anxiety. Sivell et al. (2008) also noted these contradictory findings—that in some cases genetic counselling did facilitate an understanding of one's risk—but this was not always the case.

A more recent systematic review (n=19), by Smerecnik et al (2009), builds on earlier reviews of both Meiser and Halliday (2002) and Butow et al. (2003) to include other genetic conditions. Prospective and randomized controlled studies were included in the review. Most studies supported previous research, in that counselling was found to cause a 25% increase in the number of individuals who had an accurate risk perception up to one year. However, some studies still found that an average of 25% of individuals continued to overestimate their risk, 19.5% underestimated their risk, and only 8% of participants reported a decrease in overestimation of risk post-genetic counselling. The authors concluded that the varied findings as to the effect of genetic counselling on producing accurate risk perception may be attributed to numerous factors: discrepancies between one's subjective versus objective risk, the content of counselling session, the fact that the majority of existing research is on BRCA1/2 and HNPCC, a lack of consensus on what the appropriate measurements of risk perception and accuracy should include, and variations reporting terminology.

The most recent systematic review by Kaphingst and McBride (2010) of research on HBOC (n=12) and HNPCC (n=5) concluded that although genetic counselling appears to improve participants understanding of being at risk for a genetic linked disease, it is of concern that participants still do not consistently interpret their predictive genetic testing

results appropriately. The literature on cardiovascular disease sheds some light on this concern, proposing that it is the inconsistency of information that exists amongst health care providers—and the dissemination of such inconsistencies to the public—that contributes not only to inaccurate risk perceptions but also to the reluctance of at-risk individuals to engage in health promoting behaviours such as diet (Goldman et al., 2006). That is, in a kind of snowball effect, these misconceptions are spread in communication amongst family members and across generations. This warrants a close examination of how families communicate risk amongst themselves.

The above six systematic reviews, while offering insights into understanding how risk is understood, presuppose the knowledge deficit model of risk perception. That is, laypersons lack an understanding of the science surrounding genetics, and, once the experts educate them, this deficit will resolve (Hansen et al., 2003). I agree with other scholars (Cox & McKellin 1999; d'Agincourt-Canning, 2005; Einsiedel, 2000; Ozanne et al., 2010; Parsons & Atkinson, 1992; Shiloh et al., 2006) in that laypersons do have a unique body of knowledge that is subjective and contextual in nature and that does not rely on objective measures of risk as the primary source of their knowledge and that it is important to their decision-making process. Furthermore, being that the majority of studies included in these systematic reviews are quantitative in nature, they do not capture the complexity of laypersons' knowledge. My research study has the potential to fill this gap and offer a plausible explanation for how laypersons draw on and develop diverse sources of knowledge in order to understand their risk and to make decisions about their health.

Family and Risk Communication

There are three recent systematic reviews that provide further insights into the challenges faced by at-risk individuals as they make the decision to communicate genetic risk information to family members (Gaff et al., 2007; Chivers Seymour, Addington-Hall, Lucassen, & Foster, 2010; Wiseman et al., 2010).

Gaff et al. (2007) conducted a systematic review of 29 peer-reviewed papers of 26 studies that investigated the process and outcomes of communicating genetic risk in inherited cancer syndromes. Of these studies, 17 were qualitative. In terms of three phases, the authors described the challenges that at-risk individuals faced as they prepared to communicate risk information to other family members: (1) deliberation before communication, (2) communication strategies, and (3) outcomes of communication.

Deliberation before communication (that is, how individuals collect and organize their thoughts prior to disclosing risk information) included factors such as being clear about one's own personal risk, making the decision as to what information needs to be communicated and when, and anticipating potential outcomes of disclosure. During this phase, individuals attempted to balance their desire to avoid harm with the need to inform a relative of their risk status. This required an assessment of their family member's ability to understand and cope with the news. Participants also described being selective in the information they disclosed in terms of family versus personal information. Timing of the disclosure was contingent on what participants sensed was the right time—a critical event or a social interaction. Communication strategies were described as being spread across the continuum of no disclosure, partial disclosure, and cautious disclosure, to open and supportive communication. At times disclosure is delegated implicitly or explicitly

to intermediaries deemed responsible to disseminate the information. There are, however, times that this communication does not happen. The final theme discussed by Gaff and colleagues—outcomes of communication—considers the impact of disclosure on participation in predictive genetic testing, the knowledge level of recipients, and the impact of participation in predictive genetic testing on familial relationships. In light of their review, the authors suggested that genetic counsellors do play a more active role in supporting the sender and receiver of at-risk information throughout the entire disclosure process.

The systematic review by Chivers Seymour et al. (2010) was the first to look at factors that facilitate or impede family communication post-predictive genetic testing for cancer. The qualitative research reviewed (n=14) was analyzed in terms of six core themes: (1) reaction to being the informant, (2) relevancy of information and anticipated responses of relatives, (3) closeness of familial relationships, (4) timing and content of disclosure, (5) the role of health care providers in disclosure, and (6) family rules and patterns of disclosure. These themes support the significance of having a good understanding of the contextual factors that influence communication between at-risk family members (e.g., a sense of relational responsibility) and being able to communicate knowledge about risk in a confident and appropriate manner. These points have also been made in studies on individuals living with genetic linked heart conditions such as FH, (Weiner & Durrington, 2008) HCM, and LQTS (Smart, 2010). Thus, when planning care for individuals living with ARVC, health care providers need to consider the social and moral context of family relationships.

The third systematic review (Wiseman et al., 2010) on the dissemination of at-risk information and the factors that influence the process of risk communication builds on the two previously cited reviews. Peer-reviewed articles ($n=33$) from 1985–2009, using a variety of methodologies, were included in their review. Wiseman et al. reported that there are several consistent threads in the literature on risk communication. First, dissemination of risk information is influenced by a combination of individual, relational, and contextual factors. Second, desirability to communicate risk information depends on the closeness of familial relationships. Findings, however, were inconsistent as to the impact of genetic risk information on family relationships and the family unit, warranting further research.

By comparison to the three above systematic reviews, a recent study took a somewhat different approach and looked at the effects of the process of disclosure of at-risk information on family members' risk perception (Vos et al., 2011). This is the first study to examine the impact of communicating genetic testing information by a proband to an at-risk relative in a population with BRCA1/2. Participants had received either an unclassified variant test, a finding that they had a genetic mutation for which the meaning is not known, or an inconclusive test. Vos and colleagues found that relatives of the proband did not make their health decisions primarily based on the information communicated by the proband but rather on the "art" of communication by the proband. That is, the less understandable and reassuring the communication from the proband was, the higher the relative's perception of being at risk for cancer was. This supports the position that other critical theorists and I hold: that "objective" risk is not the most significant concern when constructing ideas about risk.

It is evident from this discussion that risk communication is complex, and its effectiveness is contingent upon a multitude of factors. This body of literature strongly suggests the need for more research that illuminates challenges in the dissemination of risk information across the continuum of the genetic testing experience. Furthermore, in order to plan for and predict psychosocial and behavioural responses, it is imperative that health care providers first take time to explore how risk is being communicated and received amongst family members. Finally, given the dearth of literature that looks at risk communication in genetic linked cardiovascular disease, more research is critical in order to plan for psychosocial and behavioural responses that will add to the existing body of knowledge about risk communication.

How Does Risk Perception Influence Behaviour?

There is an inference within the literature that knowing one's genetic risk status leads to increased participation in behavioural and lifestyle changes that can minimize one's risk (Senior, Marteau & Weinman, 2000), however, this relationship still remains unclear (van Maarle et al., 2003). This ambiguity is partly due to the lack of understanding of the relationship between objective risk and subjective risk (Kaphingst & McBride, 2010; Pilarski, 2009). The literature that has examined individuals' behavioural responses to living in a family at risk for a genetic condition has also examined the correlation between risk perception and behavioural responses of carriers and non-carriers, including hypervigilant and hypovigilant behaviours. There is also a small body of cardiovascular literature that has addressed behavioural responses to being at risk for a genetically linked cardiovascular disease.

Risk Perception and Behavioural Responses.

Although some studies report that an increase in risk perception, or a positive test, motivates behavioural changes (Beery & Williams, 2007; Codori, Petersen, Miglioretti, & Boyd, 2001; Collins, Meiser, Gaff, John, & Halliday, 2005; Halbert et al., 2004; Johnson, Trimpath, Petersen, Griffin, & Giardiello, 2002; Sheinfeld Gorin & Steven, 2003; Van Roosmalen et al., 2004; Watson, Foster, et al., 2004), others report that this is not always the case (Lerman et al., 2000, 2002; Marteau et al., 2004; Marteau & Lerman, 2001). In fact, the findings from studies suggest that a positive correlation between risk perception and levels of psychological distress compromises compliance to surveillance programs and increases the likelihood that one will disengage in the genetic testing process (MacDonald, Doan, Kelner, & Taylor, 1996; McAllister, 2002). Others argue that knowing one's genetic status results in fatalism due to a sense of lack of control over the disease and its outcomes, and hence a feeling that there is no point in making behavioural changes if one's destiny is, in fact, predetermined (Davison et al., 1989, 1991, 1992; Marteau & Lerman, 2001; McAllister, 2002, 2003; Senior et al., 1999; Senior et al, 2000).

Sivell et al.'s (2008) systematic review (n=12) found that research that examined the correlation between risk perception, screening and surveillance uptake, and willingness to engage in other health-related behaviours (e.g., diet and exercise) is inconsistent. Although several studies in this review supported earlier research that an increase in stress can lead to disengagement in prescribed treatment regimes (MacDonald, Doan, Kelner, & Taylor, 1996; McAllister, 2002) and surveillance (Heshka, Palleschi, Howley, Wilson, & Wells, 2008), this was not always the case.

Rees, Martin, and Macrae's (2007) literature review, which included research that spanned over 12 years (n=30), examined participation in screening by individuals with a family history of CRC and supported the observations of Sivell et al. (2008), in that research is inconsistent in its findings about at-risk individuals' adherence to recommended screening behaviours. Rees and colleagues suggest that there are key predictors of adherence to recommended screening: the advice of a health care professional, a strong family history of disease, and easy access to screening. In contrast, a more recent study that looked at BRCA1/2 noted that although women who had an elevated risk perception were more likely to participate in health preventive behaviours, there is a threshold: the more invasive the procedure, the less likely one is to engage in the activity (Ozanne et al., 2010). This finding might explain some of the inconsistencies in behavioural responses noted in the literature.

Living in a family with a genetic linked disease has been found to have a significant behavioural impact on all members of the family unit. Taoqiet, Ingrand, Beauchant, Migeot, and Ingrand (2010) conducted a cross-sectional survey of siblings (n=172) of patients who had undergone surgery for CRC, examining their willingness to undergo a colonoscopy. Sixty-six percent of siblings declared they would participate in a colonoscopy. Factors influencing this decision were similar to those cited by Rees et al. (2007), including reduced barriers to screening, ease of access to resources, recommendations of the physician, and discussions with siblings and with health care providers. Social support and a sense of pressure to undergo screening were other factors identified that impacted screening behaviours.

Findings are also controversial as to the correlation between carrier status and adherence to recommended management regimes. Some of these findings suggest that individuals continue to participate in appropriate screening regardless of carrier status (Kaphingst & McBride, 2010; Meiser, 2005; Meiser et al, 2004). Other studies found that behavioural responses to predictive genetic testing change over time and between carriers and non-carriers. This may be linked to the changing contextual nature in which one constructs their sense of risk, the trajectory of the disease, and relevancy of predictive genetic testing (Brorsson et al., 1995; Sanders et al., 2007; Chivers Seymour, et al., 2010). Foster et al. (2007) measured behavioural changes at baseline and at three years in women at risk for BRCA1/2 (n=53 carriers; n=101 non-carriers). They found no differences in behavioural management at baseline, regardless of genetic status, but at three years mammography rates were higher in carriers (89%) compared to non-carriers (47%). Carriers also reported an increase in breast self-exams and opted for more risk reducing surgeries such as oophorectomy (43%) and mastectomies (34%). In comparison, 36% of non-carriers did not have a mammogram post-testing. Others have also reported similar delays in screening up to one year with HNPCC (Bleiker et al., 2005). Some studies found that 80% of unaffected carriers tested for HNPCC had a colonoscopy within 1-2 years post-testing (Ponz et al., 2004), with 65% following recommended screening guidelines over the immediate 12-month period post-testing (Hadley et al., 2004).

Carriers and non-carriers both have huge variations in terms of their behaviours post-disclosure of predictive genetic test. Wainberg and Husted (2004) conducted a systematic review (n=7) on the choices of women post-disclosure of a positive BRCA1/2

test. They noted that between 0–54 % of unaffected BRCA 1/2 carriers opted for a prophylactic mastectomy, with 13–53% having an oophorectomy. Of those who did not have surgical intervention, 57–93% complied with suggested mammographic screening. Meanwhile, non-carriers post-BRCA1/2 screening reported no changes in their screening practices (Lerman et al., 2000). Similarly, other studies have reported that BRCA 1/2 carriers had more prophylactic surgery in comparison to non-carriers and in some cases valued it higher (Beery & Williams, 2007; Van Roosmalen et al., 2004)

Hypervigilant and hypovigilant behaviours. A body of literature has reported that in some cases individuals living in at-risk families exhibit hypovigilant or hypervigilant behaviours in response to the uncertainty of predictive genetic testing results and beliefs that the disease is not solely due to genes but a combination of modifiable factors (Collins et al., 2005, 2007; Hadley et al., 2004; Michie, McDonald, & Marteau, 1996; Michie, Weinman et al., 2002;).

The systematic review by Heshka et al. (2008) of 30 studies reported in 35 published articles examined the impact of carrier status on risk perception and psychological and behavioural outcomes in HNPCC, HBOC, and Alzheimer disease. Most studies in the review concluded that carriers exhibited some limited changes in behaviour. There were only two studies on HBOC that reported carriers and non-carriers making lifestyle changes to reduce risk (e.g., diet, exercise and smoking cessation). Adherence to suggested surveillance regimes increased in carriers up to one year post-disclosure of test results compared to pre-disclosure and compared to non-carriers; however, they were not as high as expected, and they varied. For example, rates for mastectomy and oophorectomy amongst carriers ranged from 0–51% to 13–65% amongst

those at risk for HBOC. One study on HNPCC reported that 20% of all participants were non-adherent with recommended screening practices up to 12 months after receiving results. As well, 35% of carriers did not adhere to screening programs post-testing, with only 50% having recommended colonoscopy. In non-carriers 13% did not adhere to recommended screening and 40% did not have recommended colonoscopy. A few carriers (18%) and non-carriers (8%) were hypovigilant and hypervigilant in screening behaviours.

Hypervigilant behaviour was noted in Michie, Weinman et al.'s (2002) cross-sectional quantitative study. They found that FAP non-carriers (n=127) still perceived themselves as being at high risk for developing FAP. Of those, 42% wanted to continue with screening despite their identified low risk. This was attributed to the beliefs that the predictive genetic testing was inaccurate and that the FAP disease trajectory was not solely due to genetics but to diverse lifestyle factors, as well. These findings were noted in earlier research (Michie et al., 1996; Reeve, Owens, & Winship, 2000).

High levels of psychological distress associated with an elevated sense of risk have been linked to what some might consider extreme, or hypervigilant, behaviours. In one study, 23% of individuals at risk for HBOC elected to have prophylactic surgery prior to predictive genetic testing; subsequently only 47% were found to be a carrier. In contrast, another study that measured psychological consequences of waiting for test results for women at risk for HBOC at various intervals post-testing up to one year reported no change in frequency of their monthly breast exam (Broadstock, Michie, Gray, Mackay, & Marteau, 2000). Of these women, only three entertained the idea of having a prophylactic mastectomy. Only one woman had planned a mastectomy. Ten women felt

that prophylactic mastectomy was a very drastic measure and would only consider it if they knew their test results.

Cardiovascular Literature. In the cardiovascular literature, behavioural responses have also been linked to one's sense of control over the disease and model of risk communication. In a quantitative study by Senior et al. (2000), participants were asked to envision how they would respond to the news that they were at risk for heart disease or arthritis. Findings reported that participants were more likely to perceive the disease as being less preventable through behavioural and lifestyle factors when disease causation was framed in a genetic perspective. These findings suggest that providing genetic risk information may lead to fatalism due to a lack of control over the disease, resulting in a lack of participation in health prevention behaviours. Other studies have reported comparable results (Marteau & Lerman, 2001; Senior et al., 1999, 2000). In keeping with research by McAllister (2002, 2003), Senior et al. (2000) found that individuals have pre-existing models to explain the factors influencing risk of heart disease, including personal behaviour (e.g., diet, lifestyle, stress), contextual factors (e.g., environment, character), and chance (genes) that they draw on in order to develop their own sense of risk.

In a follow-up study to that of Senior et al. (2000), Senior and Marteau (2007) conducted a study to measure the relationship between illness perceptions and behaviours of individuals with FH at one week and at six months post-assessment. The authors supported the earlier (2000) study in that individuals who perceived FH as having a genetic basis held behavioural risk-reducing strategies, such as diet, as less important in lieu of more traditional biomedical approaches, such as cholesterol-reducing drugs. In

comparison, those who attributed FH as having a behavioural influence were more inclined to participate in behavioural activities.

One study specifically measured behavioural changes in carriers of FH seven months post-genetic testing (van Maarle et al., 2003). In this study carriers were noted to have used more medication in comparison to baseline (58% up to 77%); however, there were no changes found in smoking or body mass index regardless of carrier status.

It is quite evident from the above literature that risk perception does impact one's behavioural response, but there is no consensus in the literature as to the strength of this correlation. For some participants, a high perception of risk motivates one to engage in healthy behaviours for a while, but the longevity of this relationship is not fully understood. Furthermore, there is evidence that participants do not necessarily adhere to recommended behavioural treatments regardless of their carrier status. There are also cases of hypovigilant and hypervigilant behaviours that are not fully explainable. Evident throughout the literature is the fact that one's responses to being at-risk are constructed in a pragmatic way.

Chapter Summary

The literature discussed in this chapter highlighted the five key bodies of research on risk perception, which includes risk assessment, laypersons' understanding of risk, risk and decision-making, risk communication, and behavioural responses to risk. A critical look at this research revealed that individuals living in families at risk for a genetic linked disease do not conceptualize their risk solely in relation to numerical values, but as a subjective experience that is socially constructed. Researchers have proposed the "experiential paradigm" as a complementary framework to understand how risk is

conceptualized. Threaded throughout the literature reviewed is the fact that risk perception is something transient, fluid, and contextual in nature, which cannot be captured fully with objective measures. This explains the many inconsistencies noted in the literature surrounding risk perception and behaviour responses.

Being that ideas about risk are shaped and reshaped in relation to one's "lived experiences," recent research has used qualitative methods to examine this phenomenon. This body of literature makes evident that the knowledge acquired through living and witnessing the dynamics of being in a family at-risk is a key factor that influences ideas about risk, one's willingness to participate in the genetic testing process, and how one copes with the genetic test result. This experiential knowledge gained through these "lived experiences" is transient in nature; it gets reshaped in light of new knowledge, new experiences, new interactions, and over time. My study goes beyond existing research and captures this transient nature of risk—that is, how individuals at risk for ARVC juxtapose scientific knowledge against changing experiential knowledge, phase of the genetic testing process (pre-testing, during testing, and post-testing), and the specific conditions that explain the variations in one's experiences at a particular moment. Out of this juxtaposition emerges an understanding of the meaning of being at-risk that participants rely on to make decisions about their health.

Although the literature acknowledges the positive influence that experts in the field of genetics (such as the genetic counsellor) has on helping individuals understand and cope with their genetic status, the longevity of these effects remains unclear. It is clear that laypersons draw on the expert-provided information and superimpose it upon their own beliefs, experiences, and knowledge about risk in a pragmatic manner (such as

in the case of the coronary candidate) in order to understand their personal risk. In doing so, the layperson takes on the role of the expert, dismissing the knowledge deficit model. Research also remains controversial as to the behavioural responses of carriers and non-carriers. Although hypervigilant behaviours have been noted in both carriers and non-carriers, the opposite is also true; hypovigilant behaviours exist in carriers and non-carriers.

One's family history, growing up in a family at-risk, a sensed relational responsibility, how risk is communicated, the relevancy of the factor considered "risky" to one's life, critical events (e.g., onset of physical signs of disease, death of a family member), and the belief that one is able to cope with and manage the outcomes of the genetic test result have all been noted in the literature as significantly influencing risk perception, behavioural responses, and the decision to engage in the genetic testing process or not. This research captures this decision-making process and how laypersons draw on diverse sources of knowledge in order to understand their risk.

CHAPTER 4

THE PSYCHOSOCIAL ASPECTS OF PREDICTIVE GENETIC TESTING

The Human Genome Project has crystallized the paramount role that genetics plays in the health of individuals, families, communities, and populations. Although predictive genetic testing has led to increasingly rapid translation of genomic information into clinical application, there has been limited opportunity to examine how individuals living in these families assign meaning to being at-risk as they move through the genetic testing process. Despite the large number of studies that have attempted to describe, explain, and predict the variables that influence one's psychosocial responses to the genetic testing process, we still do not have an adequate understanding of how the experience of being offered and undergoing genetic testing shapes the meaning of the illness experience. The many inconsistencies in the literature describing the various psychosocial responses to predictive genetic testing, as well as the inability of existing quantitative studies to fully capture these experiences, reflect this lack of understanding. In order to develop a plan of care reflective of the needs of individuals at risk for a genetically linked condition, it is critical that health care providers have an understanding of how individuals cope with and assign meanings to being at-risk.

This discussion is framed in relation to three prevalent genetic linked conditions (previously discussed in chapter three): (CRC) which includes HNPCC and FAP, BRCA1/2, and HD. Relevant literature on the meanings of risk in relation to cardiac disease will be threaded throughout the discussion. Given the obvious similarities and significant differences between these three diseases and compared to ARVC, the literature surrounding these genetically linked conditions provides a fertile ground to explore the

psychosocial impact of predictive genetic testing on individuals living through the genetic testing process. It will also be useful to inform my analysis of the psychosocial experiences of living with ARVC and how these individuals assign meaning to being at-risk as they engage in predictive genetic testing.

This chapter provides an overview of the literature that explores the psychosocial impact of the predictive genetic testing process. This literature review is divided into three sections: (1) quantitative literature that explores psychological distress in carriers and non-carriers, (2) qualitative research that explores the individuals' experiences with predictive genetic testing, and (3) the psychosocial impact of predictive genetic testing on members of the family unit.

Psychological Distress: Quantitative Literature

Quantitative studies hold strength in identifying trends, patterns, and correlations between variables. The largest body of quantitative literature that reports on the impact of the predictive genetic testing process is in relation to the magnitude and duration of psychological distress in carriers (that is, individuals who have a positive predictive genetic test) in comparison to non-carriers (individuals who have a negative predictive genetic test). That research indicates that there is a wide variation in the psychosocial impact of predictive testing and a wide range of psychological reactions to being informed of one's hereditary risk. While some studies report no difference between carriers' versus non-carriers' responses to notification of genetic status, others make a clear distinction. There are varied findings as to whether psychological distress increases, remains the same, or decreases across time following disclosure of predictive genetic

testing in comparison to baseline pre-test measurements between carriers and non-carriers' responses to learning the test result.

Below, I examine the body of quantitative research on the presence and duration of psychological distress in carriers and non-carriers. Following that, I review the factors that influence psychological distress throughout the predictive genetic testing experience. Literature that focuses on BRCA1/2, HD, and CRC are presented under each subsection. The section concludes with a brief overview of relevant quantitative cardiovascular literature.

Psychological distress. The literature on responses to genetic testing from a quantitative perspective has established that the predictive genetic testing process does not cause significant psychological distress (Aktan-Collan, Haukkala, Mecklin, Uutela, & Kaarianen, 2001; Arver, Haegermark, Platten, Lindblom, & Brandberg, 2004; Braithwaite et al., 2004; Broadstock, Michie, & Marteau, 2000; Claes et al., 2004; Collins et al., 2007; Decruyenaere et al., 1995; Evers-Kiebooms & Decruyenaere, 1998; Gritz et al., 2005; Kinney et al. 2005; Meiser, Collins et al., 2004; Meiser et al., 2004; Murakami et al., 2004; Reichelt, Heimdal, Moller, & Dahl, 2004; Schwartz et al. 2002). That research indicates there is little difference in the level of psychological distress (e.g., anxiety, depression, and worry) throughout the genetic testing process between carriers and non-carriers; that is, regardless of genetic status, predictive genetic testing did not cause any significant adverse psychological outcomes. Several studies also reported that unaffected carriers and affected carriers had no significant changes in psychological distress in measures taken pre-test and post-disclosure (Croyle, Smith, Botkin, Baty, &

Nash, 1997; Lerman & Croyle, 1996; Lodder et al., 2001; Reichelt et al., 2004; Schwartz et al., 2002).

The literature indicating that predictive genetic testing does cause some distress is inconsistent in its illustration of how this distress manifests itself in carriers and non-carriers, particularly in the context of onset and duration of the disease. Some researchers have noted that even though psychological distress was found to increase immediately post-disclosure of test results in carriers (but not in non-carriers), this difference subsided within a year (Aktan-Collan et al., 2001; Almqvist, Brinkman, Wiggins, & Hayden, 2003; Arver et al., 2004; Claes et al., 2004; Codori, Slavney, Rosenblatt, & Brandt, 2004; Collins et al., 2007; Gritz et al., 2005; Heshka et al., 2008; Meiser & Dunn, 2000; Meiser, et al., 2002, 2004; Shaw, Abrams, & Marteau, 1999; Watson et al., 2004). However, for some individuals, the psychological distress experienced after genetic testing continues to extend further than the first year—up to and beyond five years (Taylor & Myres, 1997; Timman, Roos, Maat-Kievit, & Tibben, 2004; van Oostrom et al., 2003). Of these, there were only three longitudinal studies: one on BRCA 1/2 and two on HD.

Breast cancer (BRCA1/2). The impact and duration of predictive genetic testing has been explored with individuals at risk for BRCA1/2 (Foster et al., 2007; Schwartz, 2002; van Oostrom et al., 2003; van Roosmalen et al., 2004). Schwartz (2002) measured pre- and post-levels of psychological distress in women at risk for BRCA 1/2 (n=79 carriers; n=58 non-carriers; n=143 inconclusive) at one month and six months. Results suggested that these women did not exhibit any increased psychological distress up to six months post-disclosure of genetic testing. Those who had a negative test, however, described reduced feelings of risk and distress. Other researchers noted that individuals at

risk for BRCA 1/2 could experience psychological distress for upwards of one year (van Roosmalen et al., 2004), and in some cases, up to three years (Foster et al., 2007). Foster, et al. (2007) reported no differences between BRCA 1/2 carriers and non-carriers at baseline. Notably, at three years post-predictive genetic testing, mammography rates were higher in carriers (89%) in comparison to non-carriers (47%). Also, carriers reported a higher level of distress than non-carriers, had engaged in more risk management strategies, and had opted for more risk-reducing surgeries (e.g., oophorectomy and mastectomy) (Foster, et al., 2007).

The one longitudinal study on BRCA 1/2 (van Oostrom et al., 2003), which extended up to five years post-genetic testing, reported that there is little or no difference in overall levels of psychological distress scores in carriers and non-carriers for cancer worry, avoidance, general anxiety, depression, and cancer-related intrusions. Furthermore, any increase in anxiety and depression reported was triggered by critical events such as having a family member diagnosed with cancer.

Huntington Disease (HD). The quantitative literature on HD has shown that long-term psychological reactions to predictive genetic testing process does differ between carriers and non-carriers and can last up to one year (Decruyenaere, Boogaerts, Cloostermans, & Demyttenaera, 1999; Foster et al., 2004; Gargiulo et al., 2009; Timman, et al., 2004; van Oostrom et al., 2003) and returns to baseline measurements at varied intervals (Decruyenaere et al., 2003; van Oostrom, 2003).

Decruyenaere et al. (1999, 2003) conducted two consecutive studies measuring the psychological impact of participating in the genetic testing process for HD. The initial study (n=29 carriers; n=40 non-carriers), reported that 10% of participants tested

for HD, regardless of carrier status, experienced mild depression, a high score for anxiety, or both at one year (Decruyenaere et al., 1999). In the follow-up study participants' (n=24 carriers; n=33 non-carriers) mean test scores were within normal range five years post-testing. Carriers did report having some negative feelings post-testing (Decruyenaere et al., 2003). In a similar vein, Codori et al.'s (2004) retrospective chart review of individuals tested for HD (n=50 carriers; n=103 non-carriers) found that up to one year post-disclosure of test results, depression occurred more frequently in carriers.

Other studies on HD that provided evidence of the predictive genetic testing process being responsible for long-term adverse psychological impact also noted that this impact varies. Gargiulo et al. (2009) used self-reporting scales and structured interviews in asymptomatic individuals (n=62 non-carriers; n=57 carriers), post-disclosure of predictive genetic testing results within a mean of 3.7 years. Depression was reported in both asymptomatic carriers (58%) and asymptomatic non-carriers (24%). Scores for social adjustment remained within the normal range for both groups. Similar results were reported by Cordi and Brandt (1994); although high risk individuals described feelings of worry and guilt, those at low risk also experienced some stress related to adjusting to their new risk status up to two years post-predictive genetic testing.

Only one Canadian study, by Almqvist et al. (2003), measured the psychological effects of being tested for HD (n=202) at baseline and post-disclosure of test results at one to two weeks, two months, six months, one year, two years, and five years. Despite the fact that psychological distress had decreased when compared to pre-test disclosure measurements, participants who had been assigned a high-risk status did experience higher incidences of adverse psychological events (e.g., depression, suicide, substance

abuse, etc) within the first two years. Of those affected adverse events, 21.8% occurred within the first year. In comparison, an older study reported the presence of psychological distress post-genetic testing at six years post-predictive genetic testing (Taylor & Myres, 1997). In that study, those considered at low risk for developing HD experienced fewer feelings of uncertainty, anxiety, and worry about their children's genetic status than their high-risk counterparts, who experienced chronic depression.

Colorectal Cancer (CRC). Quantitative studies that have explored the psychological impact of predictive genetic testing on individuals at risk for CRC support the majority of research done on BRCA 1/2 and HD; that is, individuals who engage in predictive genetic testing for CRC may experience some transient psychological distress, however, it does not persist or cause any significant adverse psychological effects within the first three years post-testing (Aktan-Collan et al., 2001; Arver et al., 2004; Claes et al., 2004; Collins et al., 2007; Gritz et al., 2005; Meiser et al., 2004; Murakami et al., 2004). Furthermore, any difference in psychological distress between carriers and non-carriers was noted to subside within the first one to three years (Claes et al., 2004; Collins et al., 2007).

Several studies have shown that individuals at risk for CRC may have an initial increase in distress (e.g., anxiety, depression) immediately post-disclosure of test results, which resolved within the first year (Aktan-Collan et al., 2001; Collins et al., 2007; Gritz et al., 2005; Meiser et al., 2004).). Included within this population, however, were several subgroups of individuals who may be more vulnerable to psychological distress and who might require more intense psychological screening and support both pre- and post-testing (Landsbergen, Prins, Brunner, Kraaimaat, & Hoogerbrugge, 2009). These were

asymptomatic carriers (Gritz et al., 2005), who experienced depression prior to predictive genetic testing, reported a lower satisfaction with social supports, had an escape-avoidance coping style, had a family history of CRC, and predicted that they would be depressed if they had a positive test (Esplen et al., 2003, 2007).

Despite the fact that debate exists in the literature as to the presence and duration of psychological distress experienced by carriers and non-carriers, the consensus is that psychological support is equally important for both groups throughout the genetic testing process. All agree that, in order to provide this support, health care providers need to be cognizant of the many factors that influence the predictive genetic testing experience.

Factors Influencing Psychological Distress.

Although predictive genetic testing results have been noted to influence the presence of and duration of psychological distress experienced by individuals at risk for a genetically linked disease, there are several other factors that impact how one experiences the predictive genetic testing process. These include the individual's pre-testing level of distress, having an inconclusive test, demographics, and psychosocial factors.

Baseline level of psychological distress. Regardless of the inconsistencies noted in the literature as to the existence, duration, and clinical manifestations of psychological distress in response to predictive genetic testing, there is overwhelming evidence that the best predictor of psychological distress is the pre-test psychological state. People who experienced distress throughout the genetic testing process reported higher levels of pre-test distress at baseline measurements (Bleiker, Han, & Aaronson, 2003; Broadstock, Michie, & Marteau, 2000; Codori, Slavney, Young, Miglioretti, & Brandt, 1997; Croyle, Smith, Botkin, Baty, & Nash, 1997; Decruyenare et al., 1999; Gargiulo et al., 2009;

Hendriks et al., 2005; Lodder et al., 2001; Meiser & Dunn, 2000; Murakami et al., 2004; Reichelt et al., 2004; van Oostrom et al., 2003; van Roosmalen et al., 2004). These studies suggest that individuals found to have psychological challenges in pre-genetic testing may benefit from intensive counselling services prior to testing in an effort to facilitate a smooth transition through the genetic testing process (Meiser & Dunn, 2000). Knowledge of the pre-test psychological state can help plan for resources to manage any arising issues, particularly in the case of those who are trying to cope with an inconclusive predictive genetic test result.

Inconclusive predictive genetic testing. There is a paucity of literature examining the psychosocial impact of receiving an inconclusive test result. It is unclear if being in a continuous state of uncertainty contributes to one's level of distress, given that for many individuals the rationale for participating in genetic testing is to decrease uncertainty (Claes et al., 2004). It is also unclear if clients are misinterpreting inconclusive test results. Researchers who have studied how individuals react to receiving an inconclusive or uncertain test result (Hallowell et al., 2002; Meiser, 2005) have found those individuals to be falsely reassured that they do not have the condition under investigation. Others have reported that such individuals feel frustrated (Frost, Venne, Cunningham, & Gerritsen-McKane, 2004) and distressed while waiting for the results (Broadstock, Michie, Gray et al., 2000; Meiser, 2005).

A pilot study by Broadstock, Michie, Gray, et al. (2000) measured the long-and short-term psychological consequences of predictive genetic testing in individuals at risk for hereditary breast and ovarian cancer at one week, six months, and twelve months. Noteworthy is the fact that study participants did not report any increase in outcome

variables (e.g., general anxiety, general distress about cancer, and cancer specific worriers) up until twelve months. It was only after 12 months that the women reported an increase in general anxiety. The authors suggested this might be credited to an escalating concern about the uncertainty of their results. Thus, this group may be vulnerable to psychological distress (Bleiker et al., 2003) when the results are not available immediately or are inconclusive (as also is typically the case with ARVC). It is in cases such as this that it is important to be aware of the demographic and psychosocial factors that have been noted in the literature to compound stress.

Demographics and psychosocial factors. The most common individual characteristics that have been cited in the literature that correlate with higher levels of psychological adverse responses include being a middle-aged female, having poor social support networks, poor communication skills, anticipating a positive result, having an affected relative, a low social economic status, a low self-esteem, having children, a poor coping style, and developing the disease prior to testing (Bleiker et al, 2003; Esplen et al., 2003, 2007; Gritz et al., 2005; Hendriks et al., 2005; Landsbergen, Prins, Brunner, Kraaiaamt, & Hoogerbrugge, 2011; Michie, Bobrow, & Marteau, 2001).

Other research has shown that high levels of psychological distress are related to worry over a loss of income, lack of eligibility for health insurance coverage (Lynch et al., 2006; Watson et al., 2004), reluctance to disseminate genetic risk information to other family members (Holt, 2006; Riedijk et al., 2005), guilt about passing the gene on to children (Lynch et al., 2006), feelings of uncertainty about one's risk (Cordi & Brandt, 1994; Michie, French, & Marteau, 2002), and worry regarding children's risk status (Aktan-Collan et al., 2000; Lynch et al., 2006; Marteau & Croyle, 1998).

Genetically linked cardiovascular disease. Individuals at risk for developing Hypertrophic Cardiomyopathy (HCM) have been noted to experience psychosocial responses to genetic testing similar to those at risk for other genetic conditions (e.g., BRCA 1/2, HD, and CRC). HCM, like ARVC, is an autosomal dominant genetically linked heart disease that results in hypertrophy of the ventricle wall. HCM has no cure and can cause sudden cardiac death at a young age. Treatment is based on a combination of pharmacological interventions and an ICD. Christiaans et al. (2009) did a cross-sectional study using questionnaires that compared quality of life, anxiety, and depression in individuals (n=228) who participated in genetic testing for HCM to the general population. Participants in this study were classified into three groups: symptomatic HCM carriers who had diagnosis confirmed with predictive genetic testing, HCM carriers who developed symptoms post-testing, and asymptomatic HCM carriers. Findings reported that, overall, quality of life and distress did not differ significantly in HCM carriers in comparison to the general population. Quality of life and distress were, however, found to be worse in symptomatic HCM carriers who had symptoms prior to genetic testing but better in asymptomatic carriers of HCM. Thus, knowing one's genetic status may instill a sense of control and reassurance in lieu of feelings of uncertainty that often lead to psychological distress (Aatre & Day, 2011; Christiaans et al., 2009).

Quality of life was also explored in another cross-sectional study in adults (n=174) with Marfan syndrome (n=174), an autosomal dominant genetically linked condition that affects connective tissue including the cardiovascular system (Peters, Kong, Hanslo, & Biesecker, 2002). Findings are somewhat consistent with other previously cited studies on HCM, in that, overall, participants did not describe a lower quality of life in

comparison to similar population surveys that looked at quality of life in those with cardiovascular disease. Participants' mean scores on the psychological/spiritual subscale of the QLI-Cardiac III, however, were lower than other populations with cardiovascular disease. Concerns raised were in relation to their personal health such as sexual wellness and reproductive choices.

Individuals' Experiences with Predictive Genetic Testing: Qualitative Literature

Predictive genetic testing is a social process; quantitative methodologies are not well suited to capture the lived experience of predictive genetic testing. Qualitative research extends beyond examining the psychosocial impact of the predictive genetic testing process; it provides an understanding of the meaning individuals assign to being at risk for a genetic condition. Here I examine (a) qualitative literature on individuals' experiences of living in a family at risk for a genetically linked condition and (b) qualitative literature on cardiovascular disease and genetics.

Genetically Linked Conditions.

The grounded theory study by McAllister, Davies et al. (2007) provides excellent insights into the emotional effects experienced by individuals who have multiple genetic conditions such as HD, Marfan Syndrome, and Duchene Muscular Dystrophy. Eight core emotional effects were identified: anxiety, worry regarding risk of offspring, guilt, anger, uncertainty, redemptive adjustment, sadness and grief, and depression. The degree of psychological distress and ability to cope was shown to depend upon the meaning assigned to the variability of genetic condition (e.g., penetrance and expressivity), availability of a diagnosis, and perceptions of care. As found in two other studies (McAllister, 2002; van Oostrom et al., 2003), participants spoke of a continuous state of

anxiety that fluctuated and became more intense during critical life events (e.g., becoming symptomatic and family planning). Then, ability to cope with having a genetic disease correlated with the amount of control participants felt they had over the disease (van Oostrom et al., 2003). This correlation was also reported in other research on BRCA 1/2 (Howell et al., 2006; Vodermaier, Esplen, & Maheu, 2010) and in HD (Sobel & Cowan, 2003).

The importance of being able to instill a sense of control over one's environment is clear in Bandura's social cognitive theory (2004). According to Bandura a high level of self-efficacy or the belief that one possesses the attributes to achieve a desired outcome motivates one to reach goals or change behaviours. The reverse is also true in that a low level of self-efficacy results in a lack of confidence in one's abilities, contributing to feelings of depression, low self-esteem, anxiety, and lack of motivation. Thus, if individuals believe they can control some aspects of their health, then they will engage in healthy behaviours and cope better with challenges they may encounter (Shaw et al., 1999).

The concepts of coping and uncertainty described by McAllister, Davies et al. (2007) were core themes in other qualitative studies. Sobel and Cowan (2003) examined the impact of predictive genetic testing on families with a history of HD. Two themes were identified: (a) the nature of the loss and (b) coping with the loss and ambiguity. Nature of the loss refers to how participants felt about the loss of the assumption they were positive. Some participants had assumed they were positive, had planned their lives around this expectation, and had experienced depression as a result of their negative status. Although non-carriers described a sense of relief with a negative test, they also

felt guilty about their predictive genetic testing result and found themselves being segregated by the other members of the family who were positive. In contrast, carriers described feelings of anger, ambiguity regarding their future and changing roles, and uncertainty as to the meaning of being at-risk for themselves and their offspring. Similar feelings were described in other studies (Cordi & Brandt, 1994; Cox & McKellin, 1999; Decruyenaere et al., 1999; Gargiulo et al., 2009).

The second theme identified by Sobel and Cowen (2003), coping with loss and ambiguity, highlights the behavioural, cognitive, and emotional responses of family members. Family members spoke of being pressured to get testing and feeling helpless because of their lack of control over the disease. Others described their genetic risk as bringing the family either together or further apart; family rituals were either formed or given up. Reactions of carriers were divided; some spoke about embracing life. Meanwhile, others described having a sense “anticipatory loss,” similar to that described in the work of McAllister, Davies et al. (2007).

Howell et al. (2006) conducted research on males’ experiences of living with BRCA 1/2. Participants spoke about feelings of guilt because they could pass the BRCA 1/2 gene on to their children and grandchildren. Individuals in that study described the decision to engage in the genetic testing process as part of their familial responsibilities and roles, similar to what d’Agincourt-Canning (2006b) referred to as a sense of familial relational responsibility (p. 106).

Several authors have discussed the phenomenon of survivor guilt—the feelings of guilt that accompany a negative test result, knowing that other family members have tested positive (Cordi & Brandt, 1994; McAllister, Davies et al., 2007; Duncan et al.,

2008; Hallowell et al., 2006; Mireskandari et al., 2009; Murakami et al., 2004; Sobel & Cowan, 2003; Tibben et al., 1993). In several studies, participants spoke of not being able to celebrate their negative status when other family members had received a positive test result (Hallowell et al., 2006; McAllister; Davies et al., 2007 Tibben et al., 1993). Scholars (Finkler, 2005; Rose, 2007) argue that survivor guilt has the potential to disrupt traditional meanings of the family, as the focus turns to biological rather than social relationships; hence, family members can find themselves segregated into a subgroup defined by their carrier status.

Cardiovascular Disease and Genetics

There are limited qualitative studies that have explored the psychosocial impact of predictive genetic testing on individuals living in a family with a history of a genetically linked cardiovascular disease. The largest body of relevant cardiovascular literature that sheds some light on the potential challenges facing this population is LQTS.

Psychosocial impact of living with LQTS. LQTS is an autosomal dominant genetically linked cardiac disease, identified primarily by a long QT interval due to interruption in ion channels. Signs and symptoms of this disease vary from nonexistent to sudden cardiac death depending on the specific ion channel affected (e.g., sodium or potassium). As with ARVC, there is a predictive genetic test for LQTS. The condition can be diagnosed at an early age, has varying levels of penetrance, and can be fatal if untreated. Its main treatment modality is the ICD. LQTS has a mortality rate of 6–13% because of sudden cardiac death prior to 40 years old (Farnsworth, Fosyth, Haglund, & Ackerman, 2006; Modell & Lehman, 2006). Thus, this population provides a good

reference for examining the potential challenges that individuals living with ARVC may encounter.

Andersen et al.'s (2008) study provides an in-depth examination into the lives of seven individuals living with LQTS. Participants' narratives highlighted four themes: (1) positive responses to the diagnosis; (2) causes of anxiety, worry, and risk; (3) limitations and loneliness; and (4) risk and existentiality. Participants described being relieved in knowing their risk status. For many, knowing their carrier status was seen as an opportunity to take responsibility for their own health and that of their children. Despite restrictions imposed by the disease (e.g., challenges with work, sleep, reproduction, and activity), many have grown to accept this condition as part of their everyday normal life. This sense of normalcy transcended to carriers' children, as parents described efforts to balance over-protectiveness with concern for the child's wellbeing.

Psychosocial Impact of Predictive Genetic Testing on Family

Living in a family with a genetically linked condition can have lasting psychosocial effects on the entire family unit. This section provides an overview of the qualitative and quantitative research that has examined (a) the psychosocial impact of predictive genetic testing in young adolescents, young children, and parents, and (b) the psychosocial impact of predictive genetic testing on partners of at-risk individuals.

Young Children, Young Adolescents, Parents, and Partners.

Given the transgenerational impact of predictive genetic testing, it is imperative to have a good understanding of the psychological impact of testing on young children and young adolescents. There are few studies that explore what life is like for children in families with a genetically linked disease and even fewer studies on children with hereditary cardiovascular disease. The majority of existing literature that does exist highlights the psychosocial impact on children from the perspectives of the parents or young adolescents (Andrews et al., 2006; Codori, Petersen, Boyd, Brandt, & Giardiello, 1996; Codori et al., 2003; Duncan et al., 2007, 2008; Michie et al., 2001; Smets et al., 2008). That research concluded that predictive genetic testing has had adverse psychological consequences on young adolescents and their parents (Andrew et al., 2006, Duncan et al., 2007, 2008; Farnsworth et al., 2006; Hendriks et al., 2005; Mireskandari et al., 2009). However, the testing led to little or no psychosocial distress on young children (Cordori et al., 1996, 2003; Michie et al., 2001; Smets et al., 2008). The duration and magnitude of any distress noted remains a contentious issue in this literature.

Young adolescents and parents. Andrew et al. (2006) used self-administered questionnaires to assess the views about genetic testing and information and support needs amongst young adolescents (n=88) ages 18-35 who had either been diagnosed with FAP or were at high risk for the disease. The findings of that study are significant in that they provide insights into the challenges faced by young adolescents as they mature. Findings noted participants, as they matured, experienced anxiety, including that related to not knowing the risk status of their young children (39%), fear of developing cancer (28%), dealing with the uncertainty of the impact of living with FAP (23%), difficulty

with obtaining information about FAP (20.5%), and difficulty with understanding the information given (18.2%).

Using the same FAP cohort as Andrew et al. (2006), another qualitative study found that eleven individuals at risk for, or diagnosed with, FAP had diverse experiences depending on whether they were clinically unaffected or clinically affected (i.e., had bowel surgery) (Mireskandari et al., 2009). Those who were clinically unaffected spoke of concerns related to infertility and how to enjoy life while healthy. Those clinically affected raised concerns about their body image, reproduction, maintaining employment, their ability to engage in social activities and sustain relationships, their ability to disclose their diagnosis to their partners, their body image, and about reproduction. The findings of Mireskandari et al. (2009) support those of Andrew et al., suggesting that long-term psychological support is imperative for addressing the ongoing challenges with knowing one's genetic status and living with FAP.

Duncan et al. (2007, 2008) conducted two studies using a grounded theory approach to examine young peoples' experiences with predictive genetic testing for HD and FAP. These studies provide much needed information about how young people construct their meaning of the genetic testing process in the broader context of growing up in families with hereditary illness. In the first of the two studies, Duncan et al. explored the genetic testing experience and its impact on young adolescents (n=2 carriers; n=6 non-carriers) who had predictive genetic testing for HD. At the time of testing, participants were between 17–25 years of age. Findings were focused on themes pre- and post-genetic testing. Prior to genetic testing, young adolescents described living their life as if they were a carrier. Many spoke of engaging in risky behaviours (e.g., drugs) as

they tried to assign some meaning to and cope with living in a family at risk for HD. For many, this was not easy, as they witnessed first-hand family members becoming ill, had difficulty understanding information about HD, and experienced frustration with not being able to have testing performed earlier. In post-genetic testing the focus was on balancing one's new identity of having a positive or negative genetic test with living life. This meant moving forward with life in a positive manner regardless of one's genetic status.

Building on that previous work, Duncan et al. (2008) conducted a second study with the same participants but included an additional ten individuals who were asked to reflect on their earlier experiences with genetic testing for FAP (n=5 carriers; n=5 non-carriers). Results highlighted the harms and benefits of participating in the genetic testing process in relation to test results. Harms, described in relation to having a positive test, included psychological distress, a negative impact on family and friend relationships, distress of parents, feelings of regret knowing their risk status, anxiety related to others' responses to news of the test results, concern for future employment, an awareness of future health issues, and a conscious awareness of disease potential at difficult points in one's life. Harms associated with having negative test result included survivor guilt, worry about other family members and the impact on family relationships, and dealing with unexpected negative emotions. Harms related to the overall genetic testing process included family stress, being irritable, feeling anxious while waiting for results, lack of control over the process, anxiety related to the actual blood retrieval, and having to confront the issue of genetic status.

Benefits associated with a positive test included the following: relief from uncertainty, the ability to move forward with life, a sense of clarity, strengthened friendships and family relationships, and a sense of control in the context of disease management (FAP only). Benefits of a negative test result included the following: knowledge of not having the disease, relief for themselves and their parents, and a feeling of being able to move on with their lives in a positive manner. Benefits related to the overall process included feelings of empowerment, benefit related to accessing genetic counselling, and enhanced family relationships (Duncan et al., 2008).

Young children and parents. Few studies in the literature examine the psychological distress experienced by parents and youth. One prospective study explored the psychological effects (e.g., anxiety, depression, behavioural problems, and competency), on genetic testing for FAP in young children (n=19 carriers; n=22 non-carriers), ages 6–17 years (Codori et al., 1996). At three months there was little change from baseline levels of outcome measures; however, children who were carriers and had mothers who were carriers experienced the highest level of anxiety and depression. At varied intervals over three years, no psychological distress in the children was reported, regardless of risk status or sex of affected parent. Those children who did have some anxiety had a sibling who tested positive (Codori, 2003).

The impact of predictive genetic testing on young children in relation to parents has been noted in other research studies. Michie et al. (2001) measured the emotional state (e.g., depression and anxiety) in children tested for FAP (n=29 non-carriers; n=31 carriers) compared to adults tested for the mutation (n=125 non-carriers; n=23 carriers), in two studies—a cross-sectional and a prospective study. Results of both studies

supported the findings of Codori et al. (1996, 2003) in that children did not show clinically significant distress during the first year post-genetic testing nor did they have more anxiety or depression than adults. Children's self-esteem remained within the normal range regardless of test result; their perception of health was ranked high regardless of test results, as well. Noteworthy is the fact that the study by Michie et al. (2001) found that children who tested negative for FAP had a momentous decrease in distress compared to pre-testing, while the study by Codori et al. (2003) reported no major changes in the level of pre-testing and post-testing distress. This decrease in stress noted by Michie et al. may reflect a transient sense of relief felt by the children as they were told they were negative for FAP. This may mean that the children did have some psychological distress that was not captured in data analysis.

Several studies have investigated the psychosocial impact of living with a genetically linked cardiovascular disease. Smets et al. (2008) measured health-related quality of life in young children (n=35) ages 8–18 with genetically linked cardiac diseases including FH, HCM, or LQTS. Findings showed that there were no significant differences in the scores when compared to the general population, suggesting that genetic testing does not have a big impact at this stage. A limitation of this study, however, is that 80% of this sample was asymptomatic; thus, the experience of these participants may be significantly different than those individuals who become symptomatic with HCM, as described in other studies of adults who exhibit signs of HCM (Christiaans et al., 2009).

There are two studies that have looked at the impact of having a young child with LQTS syndrome (Farnsworth et al., 2006; Hendriks et al., 2005, 2008) that used

completed semi-structured interviews with parents of children who were HCM carriers at 18 months post-disclosure of genetic testing results. Distress, anxiety, and depression in parents were measured within two weeks of first consultation, at four weeks post-test disclosure, and at 18 months post-test disclosure amongst both carriers (n=24) and non-carriers (n=12). Findings showed that parents of HCM carriers do have difficulty adjusting to the news of their child's genetic test result in comparison to non-carriers. Parents' anxiety was correlated to their experiential knowledge such as being familiar with HCM, having experienced a sudden cardiac death in the family, being dissatisfied with knowledge provided about HCM, and having previous distress pre-testing.

Responses of parents in these two aforementioned studies provide insight as to how parents cope with the news of having a young child with a genetically linked condition who is asymptomatic (Hendriks et al., 2005). Although all children were asymptomatic and receiving prophylactic treatment, 75% of parents remained focused on the clinical manifestations of the disease, with only 20% reporting full confidence in the prescribed treatment. Of those parents, 55% described prophylactic treatments as a burden, and 30% thought it was beneficial to the child. Parents also voiced concerns about the future of their children with regard to relationships (54%), career choices (46%), impact on puberty (54%), and stigmatization (46%). Another area of contention was the lack of information about the disease, as well as the lack of support and knowledge of attending physicians. In keeping with those findings, congruent in other studies, parents did not regret participating in the genetic testing process despite these challenges (Anderson et al., 2008; Duncan et al., 2007, 2008; Smets et al., 2008).

Farnsworth et al. (2006) did a secondary analysis of interviews (n=31) that explored parents' perceptions of LQTS. Findings echoed those of Hendriks et al. (2005). Three core themes were identified: (a) concerns for their offspring, (b) quality of life of the family unit, and (c) uncertainty of their children's health. In order to cope with their concerns, many lifestyle adjustments were made to ensure the child's safety such as giving the child a cell phone, instituting treatment regimes, having a baby monitor in the child's room while sleeping, bringing a portable defibrillator to events, and educating others (i.e., teachers, primarily physicians, and counsellors) and the child regarding symptoms of the disease requiring management. Concerns about quality of life were addressed by making behavioural changes and restrictions to one's daily routines. Parents initially attempted to keep children safe by controlling factors that might trigger the disease (e.g., physical activity); however, these restrictions dissolved as parents became more knowledgeable and experienced in LQTS management. Living with and managing LQTS, for the parents, became a normal part of daily life.

The two studies discussed above show similar findings to others. That is, in order to understand the genetic testing experience through the lens of those receiving care and to ensure that people move through the genetic testing process in a positive manner, health care providers need to co-situate themselves within the social context of individuals and families (Cox & McKellin, 1999; d'Agincourt-Canning, 2006a, 2006b; Decruyenaere et al., 1999; Etchegary, 2006a, 2006b). Such research supports the perspective that health care providers must attend to the needs of parents of children who are tested; parents are not only the core decision makers in relation to the child's psychosocial development, but they also experience psychological distress related to their

child's experience of testing. Given the dearth of literature exploring what life is like to live in a family at risk for ARVC from the perspective of the child and the parent, this body of literature sheds light on the potential challenges facing this population.

Family Members and Partners.

A dominant theme in the predictive genetic testing literature is the fact that individuals rarely go through the process of testing in isolation. There is a small body of literature that demonstrates that family members do experience psychosocial distress as they move through the predictive genetic testing process adjacent to their significant others. An understanding of this perspective is important for several reasons. First, family members are generally called upon to be key psychosocial supports for these individuals. Second, one of the key sources of distress facing many in this population is the decision to have children. Third, the spouse or partner is quite often the caregiver for the affected individual. Fourth, the spouse or partner is often the initial responder to any medical crisis, which can be very distressful not only at the time of the event but in the period pre- and post-event. Finally, family members' lives are often dictated by the illness trajectory of their family member's chronic disease.

In the following section I give a brief overview of how the predictive genetic testing process impacts at-risk individuals' partners and spouses. In conducting this review I anticipated it would help conceptualize how individuals living in a family at risk for ARVC assign meanings to their experience.

Being a partner or spouse of an individual at risk for a genetically linked condition. Research supports the fact that partners of carriers with a genetically linked disease experience a high level of psychosocial distress (Douma et al., 2010; Hendriks et

al., 2008). Hendriks et al. (2008) conducted a prospective study over 18 months that measured disease-related anxiety and depression in individuals ($n=77$) and partners ($n=57$) who engaged in predictive genetic testing for LQTS. Partners of carriers reported higher levels of disease-related anxiety than non-carriers over the long term. This is not surprising given that psychological distress has been noted to extend up to periods of five years post-predictive genetic testing (Codori & Brandt, 1994; Decruyenaere et al., 2003; Foster et al., 2004; Gargiulo et al., 2009; Schwartz et al., 2002; Timman et al., 2004; Meiser et al., 2002; van Oostrom et al., 2003).

Other studies report similar results to those of Hendriks et al. (2008). The cross-sectional study by Douma et al. (2010) used self-reported surveys to measure psychological distress and quality of life of partners ($n=129$) of individuals with FAP. They found that 30% of participants experienced distress equal to that of their affected partner, including distress over having children and feelings of guilt warranting professional services. There was little difference between the participants and the general population in measures of quality of life; however, 9–21% of participants reported that their work, leisure time activities, and relationships were affected. These findings are significant given the fact that partners are often a key part of these individuals' support networks; if the partner is experiencing anxiety, then it may have a negative impact on the affected individual. Thus, it is important to include the partner throughout the entire predictive genetic testing process, particularly in genetic counselling.

Chapter Summary

The majority of quantitative research has shown that predictive genetic testing does not cause significant adverse psychological distress, regardless of carrier status. The

few quantitative studies that have reported the presence of psychological distress post-genetic testing remain inconsistent as to its clinical manifestations (e.g., onset, duration, and magnitude). What is known, however, is that any psychosocial distress experienced throughout the predictive genetic testing process appears to be transient in nature and often sparked by a critical event such as having to make reproduction choices. Pre-testing levels of psychological distress, having an inconclusive test, demographics, and psychosocial factors have been noted to significantly influence how individuals experience this phenomenon.

Qualitative literature examining individuals' experiences with predictive genetic testing shows quite a different picture than the quantitative research. Qualitative research has found that these individuals do experience psychological distress throughout the predictive genetic testing process. The degree of psychological distress is related to the meaning assigned to being at-risk and one's ability to cope with the news of their genetic test result. Key factors identified that precipitated psychological distress included practical everyday challenges such as employment, pressure to have testing, feelings of guilt linked to having a negative test while others were positive, passing the gene on to their offspring, and, finally, concern for their health and that of their offspring. For others, knowing one's test result was critical in relieving distress.

Research has also shown that the predictive genetic testing process does have an adverse psychological impact on young adolescents and their parents. However, this is not the case for young children. Research found that psychological distress in young children becomes more evident as they mature into young adolescents and are faced with life decisions (e.g., employment, disclosing risk to partners, reproductive decision-

making). As noted in research on adults, the clinical manifestations of any distress found remains unknown; however, evidence does suggest that long-term psychological support is needed to address the ongoing challenges these children face as they move into adulthood. The parents' primary sources of distress are related to the overall quality of life of their children, including the children's physical and mental health. This concern has caused many parents to constantly monitor their children for any signs of the disease, which is psychologically challenging.

Evident in the research on genetics and cardiovascular disease is that predictive genetic testing is a family affair. Partners or spouses of these at-risk individuals do experience psychological distress arising from changes in roles and practical everyday challenges of life.

This study is significant in that it delves into how individuals juxtapose scientific knowledge against experiential knowledge and the phase of the genetic testing process. It is out of this juxtaposition that ideas about risk are constructed and understood. This is a complex and fluid process—one that existing research has failed to capture. This research not only fills this gap but also provides insights as to how laypersons' meanings of being at-risk are shaped and reshaped alongside new scientific knowledge and experiences of everyday life. It is only through a fuller understanding of the psychosocial process of constructing the meaning of being at-risk can health care resources reflective of this populations' needs be addressed.

CHAPTER 5

METHODOLOGY

A modified grounded theory approach as outlined by Glaser and Strauss (1967) was used to generate a substantive theory that explains how individuals living in families at risk for Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) construct their meaning of risk as they move through the predictive genetic testing process. This chapter provides an overview of grounded theory, its philosophical underpinnings, and how I situate myself within the research process. It outlines the process of data collection, data analysis, and specific research strategies used. The explanations of my method draw on detailed examples from my research, thus serving the purpose of foreshadowing the results. This chapter concludes with a discussion of strategies to ensure rigour of the research as well as ethical considerations

Grounded Theory

Grounded theory was first introduced by Glaser and Strauss (1967) in *The Discovery of Grounded Theory: Strategies for Qualitative Research*. Grounded theory is a methodological approach that allows the researcher to discover and gain an understanding of a psychosocial process as it unfolds. This methodology proposes that the emerging theory is grounded in the data, and that emerging concepts are linked to the data and embedded in the context of the participants' lives (Morse, 2001). According to Glaser and Strauss, grounded theory is inductive in that the theoretical explanations evolve as data are collected and analyzed simultaneously. Throughout the research process, and as the theoretical constructs emerge, an inter-weaving between data

collection, analysis, and review of the literature coincides in effort to gain a better understanding of individuals' experiences (Glaser & Strauss, 1967).

The four key features of grounded theory outlined by Morse (2001) are the use of gerunds—words ending in “*ing*” that signify an action change or concept—to code data; an emphasis on a process and trajectory described in a phase or stage; the presence of a core category that describes and explains the relationships, and variations, among categories, phases, or stages; and the generation of a theory, often in an area where little is known.

Grounded theory generates either substantive or formal theory that is grounded in the data (Glaser & Strauss, 1967). Substantive theory is focused in one particular area. For example, in my research, the theory is focused on ARVC. The generation of a substantive theory is achieved by the comparison of groups within the same focus area, or, in the case of my research, individuals at risk for a genetically linked condition. A substantive theory provides the foundation as to potential categories and constructs that need further exploration to develop a more abstract or formal theory.

Formal theory is broader comparative analysis of several diverse substantive groups (Glaser & Strauss, 1967). For example, if I wanted to develop a formal theory on risk construction, I would look at other situations in which individuals may be at-risk. This might include other chronic diseases or life situations (e.g., risk of failing an exam). The philosophical underpinnings of grounded theory stem from symbolic interactionism (SI) and pragmatism.

Symbolic Interactionism (SI)

The birth of grounded theory emerged from the tenets of SI as initially laid out by George Herbert Mead (1934), a social psychologist, and developed further by his student, Herbert Blumer (1969). SI is a micro-sociological theory that explores individuals' relationships to interactions with the natural world (MacDonald, 2001), the meaning individuals attribute to events, and the symbols they use to convey that meaning (Annels, 1996; Baker, Wuest, & Stern, 1992). It is through interpretations and meanings given to social interactions that individuals come to understand the self and others (Blumer, 1969). Blumer (1969) identifies the three critical aspects of SI:

...human beings act towards things on the basis of the meanings the things have for them...the meaning of such things is derived from, or arises from the social interaction that one has with one's fellow... meanings are handled in, and modified through an interpretative process used by the person in dealing with the things he encounters. (p. 2)

Essential to SI is the process of active interpretation, whereby one interacts with others and "the self" through human conduct (Blumer, 1969). A sense of "the self" is acquired through reflection and the ability to take on the role of "the other", which is the ability to envision oneself from the perspective of others (Mead, 1934). In order to capture the meaning of participants' experiences on a behavioural and symbolic level, the researcher must be actively engaged in the participant's world and their interactions (Baker, Wuest, Stern, 1992; Chenitz & Swanson, 1986). The researcher is then posed with the task of acquiring and understanding the meanings of participants' interactions

and coinciding behaviours through the lens of the participants (Blumer, 1969; Chenitz & Swanson, 1986).

Pragmatism

SI draws on the philosophy of pragmatism and the works of Pierce (1878) and Dewey (1922). Pragmatism emphasizes the idea of transformation through interaction, wherein one's thoughts, ideas, and coinciding choices are continuously adapting in response to one's social interactions and interpretations (Jeon, 2004; MacDonald, 2001). Pragmatism emphasizes the individual's ability to cope with changing environments by being flexible and innovative (Johnson, 1991). Pragmatism proposes that there is no absolute truth but truths that evolve in response to human interaction (Jeon, 2004).

Throughout the predictive genetic testing process, at-risk individuals and their families experience many social interactions that shape and reshape their ideas about what it means to be at-risk. These interactions are diverse; hence, individuals' responses are varied, pragmatic, and reflect multiple truths and realities. The assigned meaning of the interaction is assimilated into one's existing reality through an interpretation process, in order to cope with the meaning of the interaction, its consequences, and to formulate a response. Drawing on the ontological and epistemological stance of SI and pragmatism, I situate myself within a postpositivist perspective.

Situation of the Researcher and the Centrality of Postpositivism

Throughout this research study I have drawn upon the tenets of symbolic interactionism and pragmatism as my interpretative framework to discover how individuals living in families at risk for ARVC construct their meaning of risk. This interpretive framework enables me to conceptualize how interpretations of risk are

understood in relation to one's social interactions, and how the meaning one attributes to an interaction influences one's behaviour and decisions related to the predictive genetic testing process. Thus, in believing that human behaviours are influenced by one's interpretations or meanings, I needed to get the perspectives of those living in families at risk for a genetically linked condition in order to understand and explain how individuals experience the predictive genetic testing process. This was achieved using a qualitative research approach and a postpositivist framework (Clarke, 1998; Popper, 1968).

As a researcher I acknowledge that my personal ontological and epistemological stance itself is also being continuously shaped. From an ontological perspective and in keeping with a realist postpositivist paradigm, I believe that objective truths exist; but I also acknowledge that these truths can change in response to interactions with the world, one's social context, and science. That is, reality exists but it is "imperfectly and probabilistically apprehendable" (Lincoln & Guba, p. 168), or an estimation of what is true rather than the exact truth. I use truths in the plural to signify that I believe there are multiple truths or realities that individuals draw upon in order to assign meaning to interactions and make decisions surrounding genetic testing. Depending on the situation at hand, individuals juxtapose existing realities with available truths in order to understand their risk and respond in a pragmatic manner.

As a postpositivist, from an epistemological stance I hold that ideas about risk are constructed in relation to objective and subjective knowledge. Therefore, while I acknowledge that the stories of participants are an existing truth as they see it, my nursing background allows me to also acknowledge that health care providers at times understand, explain, and communicate from within an empirical or objective paradigm.

Thus, throughout this study I was cognizant that my knowledge and approach may not be neutral nor value-free. That is, my perceptions of what it means to be at risk are not completely detached from the inquiry. This awareness allowed me to appreciate, discover, understand, and be sensitive to how existing ideas about risk, including those of health care providers, influence and shape how individuals assign meaning to being at risk for a genetically linked condition. Furthermore, this awareness guided me to employ a variety of strategies (e.g., memoing, diagramming) to ensure my objectivity as much as possible in order to let the stories of the participants be heard without my input.

I view genetic testing as a reflective social process. It is reflective in that it challenges some participants to continuously think about what they have experienced or learned in order to interpret and create meaning to being at-risk as they move through the genetic testing process. It is a social process in that it starts with an awareness of the possibility of a genetic condition at an early age, which continues to evolve and be influenced by one's social interactions. Unique to the genetic testing process is its cyclic nature; due to its genetic component, ideas about risk are widespread across generations. That is, the meaning assigned to being at risk for a genetically linked condition and subsequent decisions will continue to influence others living in these at-risk families for many years to come. Moreover, the experiences of these "future" at-risk individuals, and how they understand risk, will differ from that of their ancestors in light of new interactions, different contextual situations, and scientific advancements.

Finally, I believe that humans are not passive but are active participants in shaping the social processes that they encounter. They are able to make choices as to the meanings of their perceptions and coinciding behaviours. Individuals interpret the

meaning of being at-risk; they then make decisions about testing in complex ways—ways that are neither rational nor irrational but pragmatic and shaped by the interactions that are part of their everyday life. The philosophical tenets of grounded theory provide an appropriate framework to discover and generate substantive theory that describes, predicts, and explains how individuals construct their sense of risk.

Given that the primary focus of grounded theory is to discover basic psychosocial processes that exist within human interactions over time within the context of the situation (Morse, 2001), it is a suitable methodological lens to understand how individuals construct a meaning of being at risk for ARVC, as this phenomena takes place over an extended period of time. Secondly, given that grounded theory holds strong ties to SI, it is an appropriate lens to explore how this phenomenon is experienced individually and collectively within family interactions. Finally, grounded theory is an appropriate method to guide research in areas in which there has been little research done, such as the case with ARVC, because theory generation, as opposed to testing, is the focus in grounded theory (Chenitz & Swanson, 1986; Glaser & Strauss, 1967; Strauss & Corbin, 1998).

This research study was made to discover the psychosocial processes that individuals living in families for ARVC experience as they move through the predictive genetic testing process. The first step in this method is data collection.

Data Collection

The key steps in data collection in grounded theory are sample selection, recruitment, and the interview process. Data collection in grounded theory is based on theoretical sampling. Theoretical sampling is the purposeful selection of research participants based on their experience with the psychosocial process under examination,

the needs of the emerging concepts, and the exploration of the similarities and differences of the concepts under examination (Glaser, 1978). In theoretical sampling the researcher simultaneously collects, codes, and analyzes data. What data are collected and where depends on the needs of the emerging theory. Thus, it is only as the researcher starts to determine codes and tries to saturate them by looking for a comparison group, while comparing existing and emerging data within and between interviews, that a framework for generating substantive theory is generated and the meaning of codes and their properties start to take shape (Glaser, 1978, p.37).

In grounded theory sample selection and size is not predetermined but continues until no new theoretical categories emerge (Glaser & Strauss, 1967; Glaser, 1978). Morse (1994) suggests that an adequate sample size consists of 30–50 data sources. In this study I had 29 participants and 34 data sources (nine individual interviews; 3 focus groups; 5 follow-up interviews).

The Sample

Inclusion criteria for participants in this study included individuals who (a) were over the age of 18, (b) able to fluently communicate in English, (c) capable of understanding the purpose of the study, and who (d) had been tested positive or negative for ARVC or had an inconclusive genetic test or had refused testing for ARVC, despite an identified risk. The inclusion criteria enabled me to collect a range of stories that represent the phenomenon of inquiry, ensuring richness of the data.

Inclusion criteria were eventually broadened to include spouses, other significant family members, and older youths. A key reason for this broadening of the inclusion criteria was to meet the theoretical needs of the evolving theory. It had been apparent

from my initial literature review on the psychosocial impact of predictive genetic testing that individuals' experiences of being at-risk for a genetically linked condition were influenced by their support network. The significance of a support network in individuals living with cardiovascular disease was also a common theme in the cardiovascular literature. In addition to this, my early data analysis led to the discovery that the experience of genetic testing was so strongly influenced by interactions with family members, such as spouses and children, that interviewing participants as if they thought about this process independently of their key family members was inappropriate. Indeed, participants spoke of the support of their spouse as being essential to their ability to cope and make decisions throughout the genetic testing process. Moreover, as many of the decisions faced throughout the genetic testing process affected not only the individual being tested but also the whole family unit, spouses were frequently an active part of the decision-making experience. Thus, in order to fully analyze how individuals living in at-risk families construct their meaning of risk, spouses or other key support people were invited to participate in focus groups.

The inclusion of the two older youths was initiated by four parents who had been interviewed. The two older youths presented themselves as part of one family focus group, with their parents requesting me to have their children tell their stories as part of the family unit. In this case, children and their parents signed the consent form and the Health Research Ethics Board was informed of the departure from the approved protocol (see Appendix C). These participants' stories provide a glimpse into the experiences of youths and, as will be discussed later, point to the need for further research into how young adolescents experience this phenomenon.

This study took place in the province of Newfoundland and Labrador (NL). The sample was comprised of 29 participants, as shown in table 5.1⁷. Twenty-four of these participants lived in rural NL, and five participants lived in urban NL. Of these 24, 17 individuals lived in areas that were serviced by small health centers; seven of the 24 individuals lived near a larger center. Nine participants had completed post-secondary education. Seventeen had either completed high school or had some high school education, and three were currently in high school. Nine individual interviews and three focus groups were completed. There were 15 ARVC positive participants and five ARVC negative participants. One participant was awaiting genetic testing at the time of the interview. Despite efforts, I was unable to recruit any participants who had refused genetic testing. The remaining eight participants were spouses of ARVC-positive individuals; of those, five were spouses of the individuals comprising the ARVC-positive sample in this study, two were surviving spouses of those who had died from ARVC, and one was the spouse of the individual who had a heart transplant. The mean age of ARVC-positive participants was 41, and the mean age for ARVC-negative participants was 51. The mean age of the spouses was 45. Two young adolescents, ages 15 and 16, presented at the focus group to be interviewed who were both ARVC positive. Fourteen ARVC positive participants had an ICD, and one ARVC positive male had received a heart transplant. Of the participants, 20 were married, one was divorced, five were single, and three were widowed. Fourteen participants were employed; fourteen were unemployed with eight retired and three in high school.

⁷ Table 5.1 Participant Sociodemographics

Table 5.1 Participant Sociodemographics

Characteristic	Number of Participants
Age	
15–20	3
21–30	0
31–40	8
41–50	8
51–60	7
61–70	2
71–80	1
Marital Status	
Single	5
Married	20
Widow	3
Divorced	1
Spouse	8
Employment Status	
Employed	14
Unemployed	4
Retired	8
Students	3
ARVC Status	
ARVC positive (males)	8
ARVC negative (males)	1
ARVC positive (females)	7
ARVC negative (females)	4
Pending Testing (males)	1
Pending Testing (females)	0
Inconclusive Test	0
Treatments	
ICD	14
Heart Transplant	1
Location ⁸	
Smaller Rural Center (health center)	17
Larger Rural Center	7
Urban Center	5
Education	
In High School	3
Completed Some High School	17
Post-Secondary School	9

⁸ Description of Populations: Small rural center (less than 400); Larger rural center (between 9,000–15,000); Urban Center (200, 000).

Recruitment of Participants

Participants were recruited through two methods: (1) a clinical geneticist and (2) using *snow ball sampling*, whereby individuals heard about the study from participants and self-referred to participate. Individuals who had participated in the predictive genetic testing experience were recruited by a clinical geneticist who had been involved in clinical research with this population. A standardized script was provided for the clinical geneticist to use when recruiting participants (see Appendix D⁹). The clinical geneticist informed the individuals of the study and of its purpose; those who were interested gave permission to have their names and contact information released to me. I then contacted each potential participant and explained the study and its purpose. A study information package was then forwarded to all potential participants. This included a cover letter, a summary of the study, and a consent form (see Appendices E, F, & G¹⁰). One month after the information package was mailed, I contacted the individuals to confirm participation in the study and to arrange the first interview.

Throughout the course of this study, individual interviews revealed the centrality of family, rather than individuals, in the genetic testing process. That is, I found that the genetic testing process is something that occurs within families rather than being something that happens *to* individuals. Therefore, I revised the methodology to accommodate this new way of understanding the process from the participants' perspectives. I added focus groups as a way of ensuring that the narratives reflected the

⁹ Appendix D: Recruitment Script

¹⁰ Appendix E: Research Study Cover Letter
 Appendix F: Summary of Research Study
 Appendix G: Consent Form

voices of the participants of genetic testing in an authentic way. Furthermore, the use of focus groups fit well conceptually within the framework of symbolic interactionism; individuals at risk for ARVC assign meaning to being at-risk in relation to their interactions with family members. As many participants spoke of the meanings attributed to interactions with other family members, I wanted to explore how these relationships shaped the predictive genetic testing process. Thus, as noted by Glaser and Strauss (1967), I needed to “employ the best method of data collection to obtain the desired information” (p.66).

Upon consultation with the clinical geneticist, three families were approached to participate in focus groups, as shown in table 5.2.¹¹ These particular families were selected because they represented a broad range of families and experiences with genetic testing in order to provide a rich and varied account of the predictive genetic testing process experience. The same protocol was followed for the recruitment of focus group participants as was followed for the individual interviews. The clinical geneticist identified a key contact within each of the three families, and the name of each was given to me. I advised the participant that they could invite other family members to participate in the focus group, using the snowball method of recruitment. An informational package was sent to the key contact, with additional packages for other family members who might express interest in telling their story as part of the family group. If they were not

¹¹ Table 5.2 Focus Groups Characteristics

comfortable participating in a focus group the option to have an individual interview was given.

As with the individual interviews, data analysis and collection of focus groups was simultaneous. That is, members of the focus group were chosen based on the theoretical needs of the study. For example, individual interviews suggested that the genetic testing experience is contextual in nature and that individuals living in these families may have significantly different experiences. As well, one individual interview spoke of the support of his wife as being critical in making the decision to have genetic testing. Another participant talked about how family members' experiences were similar and different depending on the availability of a reliable predictive genetic test for ARVC.

I recruited individuals who had participated in the gene testing process at all phases of genetic discovery (that is, prior to and after the discovery of the ARVC gene in 2007). Overall, there were eight participants who had experienced or engaged in clinical investigations in the early 1980s, seven who had experienced or engaged in clinical and genetic testing in the early 1990s, ten who had participated in genetic testing post-haplotype analysis around 2005, and four who had undergone genetic testing post-discovery of the ARVC gene.

Table 5.2 Focus Groups Characteristics

	Focus Group 1 N=4	Focus Group 2 N=12	Focus Group 3 N=5
ARVC positive men	2	2	0
ARVC positive women		1	2
ARVC negative men		0	
ARVC negative women	1	1	
Youths (15-20)		3	
Spouses	1	4	3

I met with the clinical geneticist multiple times over the course of data collection and analysis; recruitment of participants took place over a five-month period. Using the combination of snowball sampling and purposeful selection via the clinical geneticist, I was able to expand the participant pool to include a comparative analysis to existing data, in order for me to examine the diverse properties of conceptual codes and to saturate categories. In this way, the first participants were chosen because they met the inclusion criteria; as data collection and analysis continued, participants were selected based on the theoretical needs of the data.

Only one person who had participated in an individual interview participated in the focus group and a subsequent third interview, as well. A single participant made the decision, for personal reasons, to forgo the group interview in lieu of a private interview. Four of the participants in the individual interviews were interviewed a second time to

clarify points in the first interview and to provide them with the opportunity to confirm that the categories captured thus far reflected their experience.

Interview Process

Prior to beginning the interviews, I explained the purpose of the study, provided an opportunity for participants to ask questions, and obtained written informed consent. All participants were informed that they were not obligated to participate in the interview and could withdraw at any time. No participants withdrew from the study. I collected demographic information prior to the start of the study. In the case of focus groups, I invited participants to introduce themselves in relation to the other members of the group. This facilitated my understanding of the context of individual and family experiences.

The audio component of interviews was digitally taped. A professional transcriptionist who had signed an oath of confidentiality transcribed the audio files. The written transcriptions were coded and any identifying information was removed to provide anonymity and thus ensure confidentiality. The audio files were stored on a password-protected computer file. Interview transcripts, consent forms, and recorded interviews were stored in separate locked cabinets in my private, locked office.¹²

In addition to audio taping, I took notes during the interviews. These notes helped ensure that I would revisit any questions I might have during an interview, reminded me to clarify any misconceptions in the interview, and facilitated the development of subsequent questions for upcoming interviews. Written notes provided an audit trail to record my views, thoughts, and decisions as to recruitment and methodology. This tool

¹² In keeping with Memorial University policy, this information will be retained for five years following the completion of the project, at which point it will be destroyed.

enabled me to be vigilant about observing my emerging theory, identify patterns in the data, and allow transparency in the research process. Inserted within the written text of the audit trail were numerous diagrams that represented my thought process and decisions such as participant selection.

Immediately following each interview, an interpretative summary, or memo, was completed. In addition to this, I sketched detailed family trees, or *genograms*, following the interviews. These family trees allowed me to gain a fuller understanding of the social and historical context of the data collected. This was important as participants frequently drew upon stories of relatives in order to understand their own risk. A family tree was found to be beneficial for quick reference and understanding of biological relationships, particularly in the focus groups (for example, in one focus group a grandmother, her children, their spouses, and grandchildren participated). Finally, on several occasions, in consultation with the clinical geneticist, the family trees were useful for recruitment choices as I sought to ensure a broad range of perspectives in order to reach saturation of data.

I did all of the individual interviews. I conducted focus groups with the assistance of a researcher in the area who helped with observation and contextual note-taking. Interviews were semi-structured, consisting of open-ended questions and probes (see Appendix H¹³). Given the nature of grounded theory, the research questions were redefined as data was generated, and analyzed. Additional questions were included in further follow-up interviews to clarify concepts, expand on emerging themes, and ensure

¹³ Appendix H: Sample Interview Schedule/Questions

data saturation (Speziale & Carpenter, 2007). For example, building on the idea that the implantable cardiac defibrillator (ICD) was for many “a life saving device,” I asked subsequent participants about their perceptions of the ICD. In another four individual interviews I asked the participants their thoughts about the idea of “constructing one’s sense of risk” as being a core category. Their consensus validated that I had indeed captured the experience. In fact, one participant looked at me with a sense of surprise and relief and said, “I never thought of it like that, but you have got it. Yes! This is it, after all those years...Now it makes sense...That is what I was doing!” Throughout data collection and analysis of the interviews, I took time to simultaneously reflect on the transcriptions, review the literature, and reflect on emerging constructs prior to conducting further interviews. I also listened to the interviews on multiple occasions to ensure I had captured the context of the participants’ narratives.

Individual interviews were conducted in participants’ homes or in another private space convenient to the participant. Two of the focus groups took place in participants’ homes; the third was held in a rented space at a local hotel. All of the focus groups were done in rural NL. Individual interviews lasted approximately 45 to 60 minutes; focus group interviews ranged from 60 to 90 minutes in duration. Data analysis commenced immediately post-interview, either by reviewing the taped interviews, the transcript, my notes, and/or discussing the interview with one of the members of my supervisory committee. All interviews were reviewed several times.

Of the second interviews, two were carried out in person in a private office and the other two were done over the phone at the participants’ request. Only one person in

the focus group had a second interview over the phone. Second interviews lasted from 20 to 30 minutes. All interview data were included in data analysis.

Data Analysis

A professional transcriptionist transcribed the audio-filed interviews, and I checked the transcription for accuracy and content. Two members of my supervisory committee facilitated data analysis. One member confirmed the interpretative summaries or memos prior to them being sent out to the participants (to be discussed below). The other member reviewed and confirmed emerging codes, properties, conceptual categories, and theoretical constructs that depict the basic psychosocial process of constructing one's sense of risk throughout the predictive genetic testing process.

Data analysis methods used in this research included the constant comparative method, substantive and theoretical coding, theoretical memoing, and diagramming, as outlined by Glaser (1978).

Constant Comparative

The constant comparative method was used in order to jointly code and analyze data. The constant comparative method adds to the credibility or trustworthiness of the data; data are repeatedly being compared throughout the entire research process in an effort to construct theory grounded in the data (Jeon, 2004). Throughout data analysis, time was taken to reflect on and compare coded categories. Isolated incidents within and between interviews were compared to each other in order to code for and develop concepts, conceptual categories, and their properties. This process was repeated for all subsequent interviews. Conceptual categories and their properties were also compared within and between interviews. Throughout this time, the focus was on the diverse

conditions or the factors that make up a category, how these factors present themselves, how they are maximized or minimized, and their consequences on a category. As the theory started to emerge through constant comparison methods, category reduction was done in order to create parsimony of the categories and generalizability of the emerging theory (Glaser & Strauss, 1967). That is, links between categories that explain how individuals construct ideas about risk throughout the genetic testing process are clearly presented on a broad theoretical level (Cutcliffe & Hader, 2009).

In the case of the focus groups, I initially compared focus group interviews to individual data within the same focus group. I then compared this data to other individual and focus group interviews. I also compared subgroups within and between the focus groups. For example, I compared the experiences of spouses with each other and to other participants' experiences. I also compared the experience of different family units. Thus, varied experiences were compared to ensure the richness of the evolving theory. Throughout this process, conceptual categories and their properties were integrated or collapsed as the core category was discovered. As gaps in the evolving theory became evident, I searched for participants who would help me gain a fuller understanding of what was happening.

Throughout this process I started to "think about the full range of types of continua of the category, its dimensions, the conditions under which it is pronounced or minimized, its major consequences, its relationship to other categories and its other properties" (Glaser and Straus, 1967, p. 106). For example, while I was comparing incidents surrounding one's initial awareness that they might be at risk for a genetic condition, I noticed that, for many participants, this initial risk awareness was sparked by

story of an unexpected death of a relative. After several interviews, it became apparent that, although many participants had similar stories of unexpected deaths in their families, for some it was only when they had tangible signs of having heart trouble themselves such as a “racing heart” that they started to think that their symptoms may have a genetic cause. In fact, for many participants, physical signs of heart problems or witnessing a critical event, such as the illness of a close relative, had a significant impact on shaping their meaning of risk throughout the entire genetic testing process. Also, even though some participants were ARVC negative, the news of another relative’s death caused them to rethink their own personal perception of risk. In addition to this, it was evident that conditions that contribute to one’s understanding of risk were also varied. While some participants understood their risk for ARVC as being analogous with their lifestyle habits and gender, for others this was not the case. Thoughts about risk were also linked to treatment modalities. Many participants held varying perspectives surrounding the efficacy of the ICD as a treatment for ARVC. For some, the ICD was seen as a “lifesaver” and in some way suspended their risk for the disease, but for others the ICD was a temporary solution. This led me to believe that the meaning of being at-risk emerges at different points in an individual’s life, often sparked by a meaningful interaction with oneself, others, and objects. It is how one interprets and assigns meaning to that interaction that shapes one’s behaviour and beliefs about the interaction.¹⁴

By comparing data it also became clear that there was an obvious core category, *constructing the meaning of being “at-risk,”* threaded throughout all narratives that

¹⁴ This finding will be discussed further in Chapters 6, 7, and 8. I provide this information here as a means of illustrating the process of ongoing analysis during data collection.

linked all of my various categories together in a logical manner and could account for variations in experiences. The constant comparison method helped me to code for the core category and saturate other categories; that is, as I compared data, new thoughts and questions came to light, and I was able to develop questions to gather more data in order to address those questions and to help me understand and explain the experience of constructing a sense of risk. For example, as I interviewed participants I asked them to tell me about growing up in their family, to describe their understanding of ARVC, and to talk about their experience with the ICD. This simultaneous data analysis and collection continued throughout the coding process until I felt that I had saturated all categories and their properties, and that no new information was being heard.

Coding

Theoretical coding as outlined by Glaser (1978) was used in conjunction with the constant comparative method to analyze data, as shown in figure 5.1. In grounded theory, theory emerges as relationships between participants' narratives or indicators (I) are discovered, compared, and clustered together to form concepts that reflect similarities in the indicators. Indicators are raw data: words, phrases, or sentences from the participant narratives. The hypothetical relationships noted between indicators and concepts are merged into properties. Properties describe conditions, dimensions, and consequences under which conceptual categories occur (Corbin, 1986). Conditions refer to the specific factors that influence the property (e.g., age). Dimensions are the social context under which the phenomenon under investigation unfolds (e.g., pre-genetic testing) (following Glaser & Strauss, 1967). Properties are then combined to form conceptual categories. A category is reflective of the variability and similarities of properties. A category is

considered “a conceptual element of a theory” (Glaser & Strauss, 1967 p.36). These conceptual categories reflect a core category, a main theme that links all categories together in a logical manner. Categories are further refined to represent core theoretical constructs that describe the psychosocial process under examination, in this case the social process of constructing one’s sense of risk. Significant throughout this process is the fact that as properties and categories emerge it is their interrelation that guides data collection and analysis (Glaser & Strauss, 1967). Theoretical coding occurs on two levels in order to generate a substantive theory: (1) substantive coding and (2) theoretical coding (Glaser, 1978).

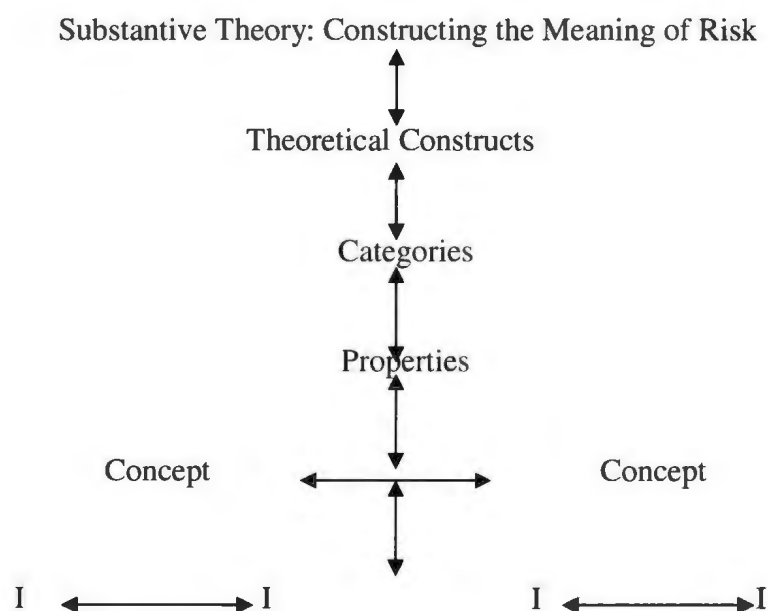


Figure 5.1. Ground Theory: Theoretical Coding. Schematic of coding used to guide data analysis. The letter I indicates the participants’ quotes, or “indicators.” The arrows represent the constant comparison of the data as the indicators are collapsed into concepts; concepts are compared and collapsed into properties and so forth. (Adapted from Glaser, B. G. [1978]. *Theoretical sensitivity*, Sociology Press Mill Valley: California.).

Substantive coding. Substantive coding is comprised of both open and selective coding. Open coding, according to Glaser (1978), starts with line by line coding of the interview transcript in order to ensure full theoretical coverage of data. These codes or conceptual labels, which identify the phenomena indicated by the data, are merged into similar concepts and properties and then collapsed into conceptual categories. The researcher “codes for as many categories that might fit, coding different incidences into as many categories as possible. New categories emerge and new incidences fit existing categories” (Glaser, 1978. p. 56). It is through this process that the data are “opened up” or “fractured” into analytical pieces that can be coded to fit into properties and categories as patterns amongst data are recognized. As open coding continues, core categories start to emerge. As one starts coding for core variable(s), categories become saturated and their properties confirmed (Glaser, 1978).

This study included line by line coding of participants’ narrative accounts. This approach to coding forces the researcher to verify and saturate categories, minimizes missing an important category, produces a dense rich theory and gives a feeling that nothing is left out” (Glaser, 1978. p. 58). For ease of coding, all transcripts were placed in a table with two columns, one for the data and one for the codes and any theoretical memos. Gerunds were used to code data throughout this process (e.g., understanding, knowing, and trusting). NVivo software was used for data management, providing a method to link indicators to concepts, properties, categories, and constructs. Throughout the coding process, codes and memos were written in the margins of the transcripts prior to being inputted into NVivo. The corresponding indicators were linked to the assigned code for ease of retrieval and data analysis; therefore, at any point during the data analysis

process, the “raw data” to support properties, categories, and constructs was readily available in an organized fashion. Theoretical memos were also inputted into NVivo and again linked to data. Multiple codes were constantly being assigned to the data as I attempted to discover the relationships between data sources.

As I engaged in open coding, I asked myself several key questions that Glaser (1978) suggested as being important to the proper use of open coding. Initially, I asked “What is this data the study of?” As I moved through the narratives, it became apparent that the data were telling me that I was not simply looking at the psychosocial and behavioural outcomes of genetic testing, as initially proposed in my research proposal, but rather at how participants construct a personal meaning to being “at-risk.” Secondly, I asked, “What category does this incident indicate?” This question allowed me to remain focused on the generation of codes that are interrelated and grounded in the data. The final question asked was, “What is actually happening to the data?” As I conducted more interviews, the answer to this question became more apparent: the data were taking me through the process of how one’s meaning of risk is shaped and reshaped at various stages in the genetic testing process *as well as* in relation to the state of scientific knowledge at that point in time. Also, the significance of social interactions and relationships in formulating one’s construction of risk was obvious.

Open coding continued until it was evident that the participants’ narratives were telling the story of how ideas about risk are shaped and reshaped throughout the predictive genetic testing process. To facilitate an understanding of how individuals assign meaning to being at risk for a genetically linked condition the core variable, “constructing one’s sense of risk,” emerged, revealing how risk is understood, and the

conditions and consequences that influence its meaning in the genetic testing process (Glaser, 1978). A core category is one that is identified as being central to the psychosocial process, accounting for variation in a pattern of behaviour; it occurs frequently, it reflects meaningful relationships amongst properties, it takes time to saturate due to the nature of its variability and relationships, and it can stand alone as a separate dimension to the process. A core category, however, is relevant in understanding the entire social process (Glaser, 1978). A core variable explains contradictory cases such as one's reluctance to engage in genetic testing or varied responses to prescribed treatments, such as the ICD (Strauss & Corbin, 1998).

In this study, participants' experiences with genetic testing were shaped and reshaped in relation to their social interactions and existing beliefs about risk. Their ideas about the meaning of being "at-risk" accounted for the variations in their behaviours and influenced individual coping strategies; regardless of the life context of each participant, all interviews emphasized the process of conceptualizing risk for either themselves, their families, or their children. For example, in the one interview, a key factor that influenced the decision to have the reinserted ICD was the reconstruction of risk post-cardiac arrest. Similar constructions and reconstructions of the meaning of being "at-risk" are evident throughout the major categories found in the analysis and reflect all of the data in some manner,

As I continued selective coding for the core variable, I started to saturate categories that related to the construction of one's risk (that is, no new properties or categories emerged from the data). This process led me to select additional participants whose alternate perspectives would help me to develop the emerging theory. I continued

to ask myself questions such as, “How do other individuals adjust to living with a positive ARVC result?” “Is this experience the same or different for those participants?” “Under what conditions do people successfully cope with having a positive test?” Once I had established a sufficiently diverse range of perspectives (that is, once saturation of types of perspectives had been gained) I began the task of theoretical coding.

Theoretical coding. Theoretical coding of data is the process of making theoretical links between categories. The selection of a theoretical code constitutes continuous reflection on the data and forces the researcher to conceptually analyze the social process at hand on a higher theoretical level (Glaser, 1978). It is at this stage that one starts to expand and pull conceptual categories into a basic psychosocial process (Stern & Porr, 2011).

In this study, the categories generated from the data were linked together as a process that evolves over time and in response to one’s interactions. Throughout data collection and analysis it was evident from participants’ stories that their construction of risk was a psychosocial process that sparked at an early age with the awareness that something was just “not right” in the family. This awareness was quite often linked to the unexpected death of a family member or stories about the demise of a distant relative(s). Following the early recognition of risk in this vague sense, many participants started to face the presence of ARVC in their families as they engaged in social interactions with health care providers, were exposed to more deaths and were given the opportunity to have predictive genetic testing. It is during this time that participants start to tease out and assimilate, for themselves and others, the meaning of being at risk for ARVC. Participants’ narratives end with a recollection of how they continue to reconstruct the

meaning of being “at-risk” in light of predictive test results. For many, it is a process of adjustment as they look towards a future of trying to balance their concern for their offspring with a positive outlook.

The psychosocial process that represents how participants construct their personal meaning of risk is captured by three key theoretical constructs as they move through the three phases of the predictive genetic testing process (pre-testing, during testing, and post-testing). Theoretical constructs are the highest level of codes (Hutchinson, 1986). Although each theoretical construct has its own categories and properties, they overlap as individuals move through the phases of the predictive genetic testing process and construct their own sense of risk. The three theoretical constructs identified in this study were: (1) *awakening to a new meaning of being “at-risk,”* (2) *deciphering the meaning of being at-risk,* and (3) *constructing a new meaning of being “at-risk”*¹⁵. It is the interaction of these three theoretical constructs that represents the core category, “constructing a sense of being at-risk.”

Grounded Theory Research Strategies

In order to develop emerging theory, several strategies common to grounded theory were employed, including theoretical memoing, diagramming and theoretical sensitivity.

Theoretical Memoing

Memoing is critical to grounded theory. It consists of a candid account of the researcher’s idea about the codes and their relationships (Glaser, 1978). It has three core

¹⁵ These will be further discussed in chapters 6, 7, and 8.

goals: (1) to identify any pre-existing ideas and assumptions of the researcher, (b) to record methodological decisions throughout the research process, and (c) to reflect on and analyze the data in an open and candid manner (Glaser, 1978; Glaser & Strauss, 1967). It is through the writing of memos that the researcher “analytically interprets the data and discovers emergent patterns” (Lempert, 2007). Memoing allows the researcher to develop codes in a free manner; it also creates a venue for the researcher to describe the rationale for the code itself and its placement within the coding process (Glaser, 1978). Furthermore, memoing creates a “safe place” for the researcher to tease out the meaning of the code, to discuss conceptual depth of emerging patterns and themes, and to capture speculations about relationships between indicators, properties, and categories. It can provide a forum to guide theoretical sampling or a place to “store” ideas for future reference while one delves into thoughts about the emerging theory and concepts (Morse & Field, 1995).

Memos were used to note my perceptions of the data and to write down ideas that needed further exploration. For example, many participants’ stories comprised woven recollections of factors that influenced their decision to engage in the genetic testing process. As I interviewed more participants, I was more cognizant of these factors, and at times asked direct questions to determine whether these factors were a common denominator in other participants’ stories and to discover other potential factors.

Memoing also included as the following: why I thought the code fit the data, what relationships exist between codes, and questions surrounding the variability of the emerging concepts and my own personal perceptions. In this study, I inputted memos into NVivo and conducted my coding of transcripts in conjunction with the memos.

Memos were also linked to codes for ease of retrieval and development. Also, as I started to further develop the core category, I reviewed the memos and sorted them to put the “opened” or “fractured” data back together in a meaningful way that would capture the social process of genetic testing (Strauss & Corbin, 1998). I started sorting using the core category and organized the memos to reflect the process of constructing risk throughout the various phases of the genetic testing process.

Memos, or interpretative summaries, were written about every participant to reflect their stories and to facilitate development of the emerging theory. Another researcher (a member of my supervisory committee) confirmed the interpretative summaries; all identifying information was removed and the summaries were mailed to all participants. A stamped self-addressed envelope was included for participants to return the signed form stating whether the interpretative summary captured their experience. Participants were asked to confirm that I had indeed captured their experience. The summaries provided another opportunity for participants to add to the experience or clarify any misconceptions. For some, the interpretative summary was a springboard for the second interview. Two participants did call me to request minor changes in the summary to better reflect their experience. Four summaries included minor editorial changes, such as dates, and one participant added several more comments. Two reminders were sent to each participant requesting return of their comments on the summary, including one follow-up telephone call. Twenty interpretative summaries were returned signed, confirming that I had indeed captured the essence of their experience. One interpretative summary was returned without having been reviewed, due to the fact that the participant had moved. I followed up on this participant and mailed the summary

to the new address; however, it was not returned. This interview was included in data analysis as the participant had provided informed consent.

Diagramming

In addition to memos, I drew diagrams throughout the multiple stages of data analysis in order to illustrate variations within conditions, relationships amongst categories, to reflect on my thoughts, and to identify any areas that required theory development (Corbin, 1986; Strauss & Corbin, 1998). As a visual learner, diagrams were a place where I “parked” questions about the data itself and its relationships with other data; at times the use of diagrams enabled me to identify places where categories should be collapsed. Throughout this process I again asked myself several key questions as suggested by Corbin (1986), including: “What is the name of this category?” “Under what conditions or context does it occur?” “What are its properties?” “How does it happen?” “Who experiences this situation?” and “What are the consequences?” For example, as it became more apparent that the core category centered on one’s understanding of risk, I constructed a diagram that represented how the meaning of risk developed as participants moved through the genetic testing process, in relation to the above questions. I used the diagram to discuss emerging categories, theory, and phases of this social process with my supervisory committee. As I reworked the diagram, I could easily see gaps in emerging theory that guided sampling and prompted me to explore these gaps in the literature. These diagrams made me more aware of my own preconceived ideas about genetic testing.

I also drew a visual representation of the historical evolution of the theories of risk, the history of genetics, and the history of the Canadian health care system. Through

data analysis I constantly juxtaposed what the participants' narratives were telling me with existing theory and sociocultural shifts that influenced the concept of risk in society in general. Thus, these visual representations were another data source that facilitated my understanding of the meaning of risk and made me sensitive to emerging theory.

Theoretical Sensitivity

Throughout the process of coding and category development, theoretical sensitivity is essential (Glaser & Strauss, 1967; Kelle, 2007). Theoretical sensitivity refers to the researcher's ability to have theoretical insights into the substantive area of research, the ability to make sense of these insights (Glaser & Strauss, 1967), and the ability to think inductively and build theory that reflects the data (Schreiber, 2001). Glaser (1978) notes that this level of sensitivity can be obtained by remaining open to emerging ideas and by reviewing literature "that deals with both the kinds of variables and their associated general ideas that will be used" (p. 3). In order to be theoretically sensitive to the data, I did a brief overview of the literature that explored common issues with genetic testing across multiple disciplines. This overview was essential given the fact that, to date, there is no literature that focuses on the experience of living with ARVC. Also, as noted by Morse (2001), one cannot ignore the literature but rather it can be "bracketed and used for comparisons with emerging categories providing a meaningful springboard into analytical data analysis" (p. 9). Noteworthy is the fact that this initial literature review was very brief; however, as I started to see a core category emerge, I once again turned to the literature as another data source and compared my findings to the existing data (Glaser, 2004). As I became more familiar with the data and started to see

the emergence of the core category, I turned my attention to literature surrounding theories of risk to further understand the meaning of the data.

Another way of facilitating theoretical sensitivity is to acknowledge my own personal theories about the meaning of predictive genetic testing (Scheiber, 2001). This was captured in memos, in the margins of transcripts throughout the coding process, and in diagrams. I labeled these thoughts as “ME.” Also, given the fact that I had previous experience with conducting qualitative research at the graduate level, I was able to remain open to emerging ideas while being cognizant of my own thoughts and ideas.

Rigour in Grounded Theory

Throughout the study, I employed many strategies to ensure rigour of the research as outlined by Glaser (1978). Rigour, or credibility, relates to the trustworthiness of findings in qualitative research (Sandelowski, 1986). Glaser (1978) identified four criteria essential to ensure rigour in grounded theory: fit, relevance, work, and modifiability.

Fit

Fit refers to the fact that the categories must correspond with the data without being forced to fit preconceived categories or match preconceived ideas (Glaser, 1978; Stern & Porr, 2011). Fit was ensured in this study in several ways. Throughout data analysis, I was cognizant of the fact that I needed to continuously examine the data until I could see how individuals living in at-risk families conceptualized this process. Initially, I had thought that the challenge for my participants might be in terms of understanding one’s objective risk; but as I reflected on the narratives, this was actually a small part of what the data were saying. It became evident that, for participants, the construction of

one's sense of risk and coinciding psychosocial and behavioural responses was challenging. As I coded for the core category of constructing risk there was an emergent fit between the data and this core category (Glaser, 1978).

Stern and Porr (2011) explain that to ensure fit, researchers should be guided by their interpretative framework, that is, the underlying set of ideas and principles that informs and guides one's perspective and research methodology. Drawing on the tenets of symbolic interactionism, pragmatism, and postpositivism, I was able to understand how participants' different beliefs and interpretations about risk were symbolically reflective of their existing reality and relationships. It became more apparent through data collection and analysis that at-risk was an evolving concept that is shaped and reshaped throughout the testing process, reflecting the changing realities of life. Correspondingly, individuals seem to cope with their risk in a pragmatic fashion. Relationships with others and responsibility to others are key to his processual nature of risk perception.

Although the literature suggests that risk is related to one's social relationships (in the sense of relational responsibility as discussed by d'Agincourt-Canning, 2001; d'Agincourt-Canning, 2006a, 2006b; Etchegary, 2006a; Etchegary & Fowler, 2008; Etchegary et al., 2009; Hallowell, 1999; Hallowell et al., 2006; Klitzman, Thorne, Williamson, & Marder, 2007), I did not raise questions about relational responsibility in the early interviews. At the time, it seemed to me that the notion of relationships being important to decision-making had been well enough established in the literature to be taken as a "given," rather than intentionally explored. It was only through comparison of the data, when it became more evident that participants' ideas about risk evolved over time and were rooted in more than just a sense of relational responsibility that I started to

probe further into this area. And, in probing further, I discovered that the way risk functions is far more complex than I had imagined based on my readings of existing theory. Thus, by ensuring that the “fit” was there in relation to my interpretive framework, my theory was able to emerge more fully.

A final way that fit can be ensured is when participants recognize in the reported research findings their own experiences (Sandelowski, 1986). Theory fits the data, according to Glaser (1992), when the categories and properties reflect the realities of the participants. van Manen (1997) suggests that findings of qualitative data are credible only when they are returned to the participants to confirm the interpretation and to ensure that the researcher(s) have an understanding of the importance of each category. In my research, interpretive summaries were provided to each participant and participants were asked to confirm that the summaries reflect their experiences. As mentioned above, this was most evident in the comment of one participant: “Yes! This is it, after all those years...now it makes sense...that is what I was doing!”

Work

The second criterion for rigour in grounded theory is “work.” This means that “the theory should explain what happened, predict what will happen, and interpret what is happening” (Glaser, 1978, p. 4). In order to make theory work, one must capture what is going on and the results must be relevant to the substantive area of research (Glaser, 1978). “Work” was achieved by travelling to many of the homes of participants and listening to their stories within the context of their everyday lives. I strived to obtain the facts by asking questions that delved into the breadth of their experience, such as, “Tell me what life was like growing up in your family?” I also asked participants to explain

their interpretations of what it means to have a genetic linked condition or to take me through the time they found out they were negative or positive for ARVC.

Secondly, throughout the interview process, I invited participants to include significant others in the interview. The inclusion of significant others “worked” well, enriching the participants’ narratives and facilitating an understanding of the experience through the lens of another family member. Additionally, focus groups provided an opportunity for me to observe social interactions and understand how relationships impact one’s construction of risk and patterns of behaviour within the predictive genetic testing process. As I moved through theoretical sampling and data analysis it became clear that kin relationships were a significant factor in determining how one constructs their meaning of risk; this enabled me to predict how individuals draw upon family relationships to understand and cope with being at-risk.

In order to arrive at a theory that worked, I began looking for variation in the cases. I included individuals who had a positive or negative test result, those who had lived with inconclusive knowledge of their test result until recently, and one who was waiting for testing. Data were constantly compared in an effort to identify commonalities and differences.

Finally, my preliminary findings have been presented at several local, national, and international conferences. Several members of the audience who have expertise in the field of genetics have commented that this study holds much value in understanding how the genetic testing process impacts at-risk individuals and their families. I have also presented to Genome Atlantic researchers who have supported this research study in light of its contribution to the substantive area. As a result of this study I was approached by

Dr. Brugada, a leading cardiologist who discovered Long QT Syndrome, and was asked to write a chapter in a book regarding the psychosocial implications of genetic testing (Manuel, Brunger, & Hodgkinson, 2010). Finally, throughout the research process, I have had multiple conversations with my thesis committee supervisors and colleagues as the theory emerged. I found this dialogue particularly beneficial when I was attempting to diagram the model and to ensure that the categories fit.

Relevancy

Relevancy, the third criteria for rigour, is achieved by allowing the core process and problems to surface while avoiding the influence of preconceived ideas and theories (Glaser, 1978). Constructing the meaning of risk was a core process that emerged quickly during data analysis and was confirmed through follow-up interviews and participants' commentaries on the interpretative summaries. In order to facilitate the discovery of this social process I used an audit trail and memoing as a place to "park" any preconceived ideas. Reflecting on these memos I was able to ensure transparency of the research process and note any potential bias.

Modifiability

The final criterion for rigour in grounded theory is modifiability. This means that although the construction of one's sense of risk is central to understanding the predictive genetic testing process, the relevance and variation of contributing factors are continuously changing (following Glaser, 1978). Throughout the research process I sought variations in cases and used a variety of interview approaches to generate a multitude of ideas and perspectives about the genetic testing process. I also remained open to new and emerging ideas. For example, early in the study I noted that the initial

awareness that one might be at-risk seemed to be different for participants; thus, in subsequent interviews, I delved into the process of how one becomes aware that one might be at-risk. Open ended questions such as, “tell me what it was like to receive your genetic test results,” were used to elicit participants’ perspectives on risk awareness. I found that even though the conscious awareness of being at-risk might be articulated differently by participants, what was clear was the fact that on a subconscious level many participants were aware that something was “just not right” in the family. In fact, for many it was only when they became aware of the significance of having a positive genetic test that many revisited earlier thoughts about being at-risk. Thus, the process of constructing one’s risk is general allows for variation and relevancy, which makes it fit the criterion of being modifiable.

Finally, I constantly used diagrams as a method to modify categories and to identify variations in the data that warranted further examination. For example, in an illustration that outlined the meaning of the ICD, it came to light that one participant thought the ICD was a cure for ARVC. This notion of the ICD as curative was not something that I had considered in my data collection and analysis to date; therefore, I revisited all of the transcripts to see if this was a recurring theme.

Ethical Considerations

Prior to the start of the proposed study, ethical approval was obtained from the Human Investigation Committee of Memorial University of Newfoundland (now the provincial Health Research Ethics Board) (see Appendix I.¹⁶) Approval was also

¹⁶ Appendix I: Ethical Approval

obtained from the Research Proposals Approval Committee of Eastern Health (see Appendix J.¹⁷)

Written informed consent was obtained prior to the start of the interviews. At the beginning of each interview the potential risks and benefits of participating in the study were discussed to ensure that participation was voluntary and informed. Each participant was given the opportunity to ask questions. They were also notified that they could withdraw from the study at any time (at which point the audio-file would be erased), take a break from the interview, or reschedule the interview, regardless of reason. Focus group participants were informed of the importance of confidentiality of the information discussed within the group and that given that the researcher has no control over what other individuals in the group might say outside of the group, confidentiality could not be guaranteed. Focus group participants were informed that individual information could not be extracted from the focus group transcription, and therefore, once the focus group had been conducted, individuals would not have the option to withdraw their data.

As with any study of this nature there was a possibility that participants might find the interview psychologically distressing. In anticipation of any psychological distress I was prepared to assist participants to gain access to their genetic counsellor; in fact for each interview I carried with me the telephone numbers of the genetic counsellors. Secondly, the Director of the Provincial Medical Genetics Program in Newfoundland agreed to be available to discuss any concerns that might arise on behalf of the

¹⁷ Appendix J: Research Approval : Eastern Health

participants or myself from the interviews.¹⁸ Also, given that I am a nurse and have experience dealing with clients in a state of emotional distress, I felt confident in my abilities to address any potentially distressing situation. Finally, I had a toll-free telephone line installed in my private office so that participants could call to ask me questions at any time.

There was one participant who took a break from the interview for five minutes but decided to continue the interview. Two participants did become tearful when discussing their children; however, both participants returned to conduct a second interview as well, and one of them also participated in the focus group. To my knowledge no participants required access to a genetic counsellor post-interview. Participants were informed prior to the interview that there was no direct personal benefit from the interview except that they may find it therapeutic to talk about their experience.¹⁹

Every effort was made to ensure confidentiality for all of the participants. All interview audio-files and transcriptions were coded, and all identifying information removed from the transcribed interview. The written transcriptions and CDROM backups of transcriptions were locked in a cabinet separate from the consent forms and will be held for five years, then destroyed. I was the only person who had access to the locked cabinet(s) containing research information and to the password access to the computer storing the audio-filed interviews and transcriptions. Any future publications or

¹⁸ At no time was that assistance required.

¹⁹ Following the interviews several participants stated that they found the interviews beneficial and particularly liked being able to discuss concerns as a family.

presentations of this research will ensure that the anonymity and confidentiality of participants is maintained.

One ethical concern that arose during data collection was the presence of two individuals under the age of 18 who attended one focus group interview. Prior to their participation, I consulted with my supervisory committee member, who was present at the focus groups. The purpose of the study was explained to these participants, and then written consent was obtained from both the participant and their parent. An amendment was sent to the research ethics board informing them of the event and requesting permission to include the data in the study (see Appendix C.²⁰)

Chapter Summary

This chapter provided an overview of grounded theory, as the research methodology used in this study suggests. The philosophical underpinnings of pragmatism and symbolic interactionism were discussed. Situation of the researcher and the centrality of post-positivism were addressed. This was followed by a description of data collection, data analysis, and grounded theory research strategies. The chapter concluded with synopsis of the ethical considerations of the study.

²⁰ Appendix C: Ethics Approval: Amendment

CHAPTER 6

FINDINGS AND DISCUSSION: AWAKENING TO A NEW MEANING OF BEING AT-RISK

The substantive theory generated by participants' interviews centered on *Constructing the Meaning of Being At-Risk* (see Appendix B²¹). This theory emerged strongly in the narratives of 29 individuals living in families at risk for Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) who were moving through the three phases of the predictive genetic testing process (pre-testing, during testing, and post-testing). In order to assign a personal meaning to risk, participants continuously juxtapose ideas about risk against their existing beliefs about genetics and heart disease. This embodied sense of risk is pragmatic, transient, and fluid in nature. It is pragmatic in that when individuals construct ideas about risk they draw on contextual factors to shape or reshape risk perception, using a "bricolage"²² approach to make decisions; risk is transient in that the meaning one assigns to risk fluctuates in relation to the contextual dimensions in one's life, specifically one's stage of the predictive genetic testing process, one's experiential knowledge, the current state of scientific knowledge, as well as relevant conditions (e.g., age, gender, number of deaths in family, etc); and finally, risk is fluid in that risk perception is not static but an evolving process that changes with each new

²¹ Appendix B: The Theory: Constructing the Meaning of Being at Risk

²² Anthropologist Claude Levi-Strauss uses the term "bricolage" to describe the improvisational recombination of a fixed series of elements. These elements, he argues, are a key dimension of the "science of the concrete," a modality of human thought which he contrasts with formal or abstracted scientific thought. Following Levi-Strauss' sense of bricolage, the at-risk individuals are continually refashioning "new worlds from fragments" without a prior blue-print or overarching plan (Strauss, C.L. (1968). *The Savage Mind*. London: Weidenfeld & Nicolson.)

experience, advancement in science, phase of the genetic testing process, and other conditions.

In the first part of this chapter I present a generic model of a Rubik's Cube (see Figure 6.1²³) to illustrate the situational context in which the theory *Constructing the Meaning of Being At-Risk* unfolds. Following this is a discussion of the research findings. The findings of each subsequent chapter begin with the introduction of the theoretical construct under discussion. There are three theoretical constructs, and each construct corresponds to one phase of the genetic testing process (i.e., pre-testing, during testing, and post-testing). Each theoretical construct captures a category(s)—a group of similar concepts that describe a central idea. Embedded within each category are descriptive properties that depict specific conditions (or dimensions of the category) that influence how one assigns meaning to risk. The three contextual dimensions (i.e., the phase of the genetic testing process, scientific knowledge, and experiential knowledge) represent the broad social context under which the construction of risk unfolds at any particular point in time. These conditions and contextual dimensions emerge as participants' narratives are constantly compared to each other and, ultimately, become integrated to generate the substantive theory that captures the experience of *Constructing the Meaning of Being At-Risk* for individuals living in a family at-risk. Participants' narratives or indicators that shed light on their experiences are threaded throughout this discussion.

²³ Figure 6.1. Rubik's Cube: Generic Model

The Rubik's Cube: The Puzzle and Shifting Faces of Risk

The Rubik's Cube is a three-dimensional, six-faced puzzle created by an architect, Erno Rubik, in 1974. Each face of the Rubik's Cube, which is in the shape of a square, is made up of nine smaller individual blocks or cubes that are the same colour when the puzzle is solved. The cube is held together by a single core structure that has three intersecting axis, which hold the six faces of the cube in place. The design of the cube allows each face of the cube to move independently; thus, as the face of the cube moves the colours become intertwined. In order to solve the puzzle all faces must be restored to one complete colour (e.g., blue, red, or yellow). There are numerous ways to solve the Rubik's Cube using mathematical algorithms; however for the novice individual, attempts to solve the puzzle generally involve trial and error (www.rubiks.com, 2012).

As participants in this study move through the three phases of the predictive genetic testing process experience (pre-testing, during testing, and post-testing), their ideas about risk are constantly being shaped and reshaped in relation to experiential and scientific knowledge analogous to solving a puzzle. Experiential knowledge refers to the cumulative knowledge obtained by assigning meaning to the conditions or factors that influence one's perception of risk (e.g., growing up or living in an at-risk family, witnessing firsthand numerous losses, listening to accounts of relatives' deaths passed down through many generations, experiencing the effects of ARVC, etc.). Scientific knowledge, in the context of this theory, refers primarily to the stage of gene discovery within the short history of ARVC genetic research and existing biomedical models of inheritance and disease causation. As the scientific "reality" of knowledge about, testing

for, and clinical care for ARVC changes, that in turn reshapes the individual's perception of their own risk status.

These three contextual dimensions (phase of genetic testing, experiential knowledge, and scientific knowledge) all shape perceptions of the conditions that impact one's risk status. Conditions are the key factors that influence how one embodies the genetic testing process, how one assigns meaning to being at-risk, and how one copes with new meanings of risk. A condition captures the cause of the experience and the consequence (Glaser, 1978). Conditions include, but are not limited to, age, gender, modifiable lifestyle factors, and the onset of signs and symptoms. For example, the phase of testing, experience, and science may intersect to lead a particular individual to hold as salient the notion that they are not as at-risk because of their gender, or may lead them to perceive that they are more at-risk because of their advanced age or because they smoke. The reverse is also true; perceptions of the influence of conditions (age, gender, etc.) on risk in turn (re)shape the ways in which the contextual factors (phase of testing, experience, and science) impact one's risk assessment. The key conditions are, therefore, in a constant of flux. The conditions are not the same for everyone, and the meanings assigned to these conditions are transient as individuals move through each phase of the genetic testing process in relation to existing scientific and experiential knowledge.

The Rubik's Cube represents how individuals living in a family at risk for ARVC construct ideas about risk. Using the Rubik's Cube as an analogy one can visualize the relationship between the three broad contextual dimensions and specific conditions impacting the process of *Constructing the Meaning of Being At-Risk* as providing the components for the puzzle to be solved. Each axis of the three-dimensional structure of

the Rubik's Cube symbolizes one contextual dimension. As the meaning assigned to each contextual dimension changes (the axis moves direction), the meaning assigned to the conditions change (the face of the cube changes colour), and the overall meaning assigned to the experience of being at-risk shifts (physical shape of the cube changes).

The physical shape of the cube, or one's "risk reality," is made up of the conditions that influence risk perception during each phase of genetic testing. Each individually coloured block that makes up a face of the cube represents a condition. These conditions will differ from person to person and will change for each individual as they enter different phases of the genetic testing process, and as scientific and experiential knowledge changes. Subsequently, as the conditions change (the colours on the face of the cube are intertwined), the way one experiences each phase of the genetic testing process changes; as the meanings assigned to scientific and experiential knowledge changes (the axis moves direction), so does the shape of the cube and the meaning assigned to risk. Hence, the cube reflects individuals' diverse meanings of being "at risk" depending on the contextual dimensions and conditions that influence it. The cube's axis that is shared by the three contextual dimensions and the conditions within the six faces of the cube captures the influence that each has on shaping the other throughout the psychosocial process of *Constructing the Meaning of Being At-Risk*. These meanings assigned to risk are transient, fluid, and pragmatic in nature.



Figure 6. 1. Rubik's Cube: Generic model. ® used by permission of Seven Towns Limited., www.rubiks.com

This Rubik's Cube illustrates the theory *Constructing the Meaning of Being At-Risk* and explains why and how concepts of risk vary. For example, the experience of an elderly woman, who has witnessed many of her family members die suddenly and who has engaged as a research participant in the process of gene discovery since the early 1980s, may be significantly different than the experience of a 20-year-old male who has only heard stories of relatives who have died suddenly and who engaged in the genetic testing process in 1997, when haplotype analysis could provide a strong indication of risk

for ARVC. Furthermore, this theory explains how those two experiences would be different in comparison to that of a 10-year-old boy in 2010 (given the discovery of the ARVC gene in 2007, the availability of the ICD as an effective treatment for ARVC, and the lack of exposure to loss). Although each of these individuals pass through the three phases of the genetic testing process, their experiences, and the meaning each assigns to being “at-risk” would be different because of the differences in available scientific knowledge of the time, type of experiential knowledge, and beliefs about the conditions that influence risk.

This theory also explains how conditions influence, and are influenced by, the three broad contextual dimensions that shape risk construction; that is, just as the three contextual dimensions influence the meaning assigned to conditions, the reverse is also true. For example, the science surrounding ARVC frames it as a disease affecting men in their 40s; however, as women start to witness or experience signs of the disease they start to advocate on their own behalf to be tested for the gene and to receive an ICD. Thus, the experiences of women were influential in shaping scientists’ perceptions about the meaning of risk for women living in families with a history of ARVC and prescribed treatment. Hence, it is important that health care providers understand not only the conditions that influence risk perception (such as age and gender) but also how the conditions shape contextual dimensions that influence risk construction.

In the following chapters I will discuss, using relevant literature on genetics, risk, and heart disease, how participants in this study constructed their sense of risk as they moved through the three phases of the predictive genetic testing process (pre-testing, during testing, and post-testing). The first theoretical construct that describes the

psychosocial process of *Constructing the Meaning of Being At-Risk* is *Awakening to a New Meaning of Being At-Risk* (see Figure 6.2).

Construct 1: Awakening to a New Meaning of Being At-Risk

This theoretical construct examines participants' earliest experiences growing up and living in a family at risk for ARVC prior to receiving genetic testing. This construct describes the process by which at-risk individuals become aware of their risk; it describes the conditions and contextual dimensions that impact this awareness; and explicates the psychosocial and behavioural responses to the new meaning of risk. The chapter also highlights how individuals living in at-risk families juxtapose their understandings about heart disease and genetics against their experiential knowledge, against existing science, and against personal beliefs, to assign a personal meaning to being at-risk. Within this construct of *Awakening to a New Meaning of Being At-Risk*, there are two categories: (a) *making sense of numerous losses* and (b) *struggling to break the cycle of uncertainty*.

<i>Category 1: Making Sense of Numerous Losses</i>
<ul style="list-style-type: none"> • Property 1: Living in a Family Familiar with Loss. • Property 2: The Struggle to Understand the Meaning of Being At-Risk for Oneself and Others.
<i>Category 2: Struggling to Break the Cycle of Uncertainty</i>
<ul style="list-style-type: none"> • Property 1: Making Sense of and Living Through Early Clinical Investigations and Prescribed Treatments. • Property 2: Acknowledging a Possible Genetic Origin to Risk.

Figure 6.2. Construct 1: Awakening to a New Meaning of Being At-Risk. Emerging properties and categories that describe the first phase of the psychosocial process *Constructing the Meaning of Being At-Risk*, prior to predictive genetic testing.

Category 1: Making Sense of Numerous Losses

This category explores participants' recollections of growing up in a family familiar with loss. It describes their struggles to assign meaning to their experience of loss within the context of that experience. That is, depending on the chronological time of the experience within their life history (*experience*), and within the history of scientific knowledge (*science*), perceptions differ. Listening to family members' stories of death and illness, living through the illness experience, and witnessing firsthand the loss of family members, most study participants had started to construct some ideas about their risk at an early age. Participants' reports of their responses to being "at-risk" were diverse: some recalled that they had tried to normalize their risk; other reports indicated that participants drew upon their experiential knowledge and their knowledge of science to create a mental representation of the *at-risk relative*, creating a framework for understanding what being at-risk meant, in a way that enabled them to juxtapose competing realities about risk. These two, divergent, approaches will be explicated in the following analysis.

The two properties: (a) *living in a family familiar with loss* and (b) *the struggle to understand the meaning of being at-risk* capture participants' experiences as they attempt to make sense of and cope with the numerous losses in the family. Evident within these two properties are the conditions and contextual dimensions that influence participants' early ideas about risk.

Property 1: Living in a family familiar with loss. Participants described being enmeshed from an early age in a family culture of loss passed on through family stories, firsthand experiences of the deaths of close relatives, and personal experiences of illness

as they hoped for some answers as to why these losses were occurring. This property, *living in a family familiar with loss*, captures how experiences with loss influence how participants awakened to a sense of being at-risk and their psychosocial and behavioural responses. Variations of the meaning assigned to the condition of loss were evident in participants' descriptions of living in a family with a history of loss, witnessing first-hand repeated accounts of loss, the actual number of losses, the events surrounding the loss, and age of exposure to loss.

Condition One: Loss. As part of an at-risk family, participants knew from an early age that heart problems “ran in the family,” as these two statements describe: “The first time I heard of heart problems was when I was eight years old, when my grandfather died,” and, “We’ve lived with it all our lives, knowing that there was something wrong.” The sense of knowing that something was wrong was precipitated by the numerous accounts of illness and death in the family. Watching history repeat itself time and time again was at times difficult and, over time, caused several participants to worry about their own health at an early age. This is evident in the narrative of one participant who described becoming aware of his risk in his teens, “My favorite uncle died in his 30’s. Then my cousin’s brother died at 26, and that’s when it started. From that time on I think that the worry was in my mind that it was going to happen to me.”

Often it is the sheer number of deaths in the family that spark one’s awareness that something is just not right in the family, that they are living with something that places family members at risk. These feelings of awareness are particularly noteworthy in the older participants who, over the years and prior to gene discovery, have witnessed firsthand numerous accounts of illness and loss, as this foreboding comment notes, “I

have three brothers and three of them have passed away.” The recurring pattern of death in the family caused many participants to start to seriously think that they were dealing with something more than simply heart disease, as this participant summarized: “there is something [happening]; people are dying around me.”

For the younger participants, the awareness that they were living in an at-risk family was something that they, too, grew up with; however, younger participants did not witness numerous losses. One reason for this is that the ICD had already been introduced as a viable treatment option. Their awareness to being at-risk was, instead, acquired through stories told second-hand by family members, most often the parents. This is evident in the narrative of a young participant currently waiting for genetic testing, who briefly described being more aware of his risk status through his mother. He stated that he “never really knew about it until recently.”

For some, the awareness that something was going on in the family was similar to what Etchegary (2006b) described as “coming out of the blue” (p. 109), sparked by the shocking news of a relative’s sudden death, as this participant noted:

The first indication that there was any sickness was my uncle. One day he stopped working. I knew he was having some trouble; he didn’t like to talk too much about it... and then during Christmas he died. He was 42.

Quite often it is the events surrounding death that leave a lasting impression. This was the case in the death of one participant’s brother who died at their sister’s wedding. Reflecting on the incident, the participant recounted, “It came to light when my brother died. My sister got married... my brother was the father-giver, and he dropped dead on the floor at the wedding.” It was hard for participants to grasp how someone could die so quickly, without any warning, with no window of opportunity to prevent the death.

Incidences such as this caused many participants to re-trace their family history of loss, as the idea that they were dealing with something more than heart-disease started to come to light. What they found useful as a framework for assigning meaning was the circumstances surrounding the deaths, such as age and gender. The following narrative illustrates how individuals used knowledge of the numerous deaths in the family to assign the label of at-risk to young males:

My first recollection was hearing that my grandfather had passed away at a young age. He was 49 at the time.... As time went on, my uncle passed away. I was only a couple of years old.... I got close to my cousin and he died of heart-related problems. I can remember Mom describing exactly what happened to him when he passed away. Their son passed away at 27. I can remember another uncle, he went out fishing, and he never did come back. He had drowned in about a foot of water. Then my brother, he was 26 when he died with a heart attack.

Similar to other studies of hereditary conditions, including HD (Cox & McKellin, 1999; Etchegary, 2006b; Taylor, 2005), HBOC (d'Argincourt, 2005), and HNPCC (McAllister 2002, 2003), participants in this study drew on experiential knowledge, such as awareness of family history or personal interaction with an at-risk relative, to formulate ideas about risk. As noted in Etchegary's (2006b) study on HD, participants were aware at an early age that something was wrong in the family; however, they could not pinpoint the exact cause. Older family members knew they were at risk for something cardiac in nature, something prevalent in young males and something with a fatal outcome. This knowledge, as Etchegary noted in relation to HD, is acquired gradually. This knowledge was not hidden but became more pronounced as participants interacted with other at-risk family members and started to assign meanings to these interactions.

Exposure to the sudden loss of young male relatives, did not instantly translate into a heightened sense of risk in all cases. That said, these family members still knew

they were at risk for something familial. Their exposure sparked a beginning awareness that something was just not right in the family, as this narrative notes: “I remember Mom saying that she always knew that Dad was not going to live to be a very old age. I think she sensed there was something seriously wrong with him.”

For some participants, knowing that family members die young and accepting this as normal were two different things. That is, although participants knew that the men in their family did not live to be old, they experienced difficulties with trying to rationalize why this was the case. Several participants’ recalled conversations with other family members who had expressed their concerns with health care providers, as heard in this narrative of an older ARVC positive participant:

My brother approached the cardiologist and said he felt there was something wrong; so many people dying so young with heart problems ... but he passed away in ‘89. We agreed between us that there seemed to be something wrong.... He discussed several things with me, and it was always the men that were involved. It seemed like Mom’s brothers all passed away young. I knew there was something going on, but not until my brother really sat down and talked about it. I lived every single day knowing there’s something wrong, something not right.

Eventually, as they continued to experience numerous losses within the family, it became clear that these deaths--or as one participant calls it, the “unnatural course of events”--was not a coincidence. One participant’s narrative highlights this, stating that, “When my uncle passed away ... I think it really kind of opened everybody’s eyes a little bit that there was something here.” Similar experiences such as this sparked awareness for participants that they may be dealing with something genetic at a time when the technological means to substantiate the belief did not exist.

In response to the emerging awareness that “there was something here,” participants tried to integrate the numerous family losses as an existing part of their reality. This is similar to what Bourdieu (1977) refers to as “habitus,” or something accepted as a norm because it supports an understanding of everyday life at the time (Wildavsky & Dake, 1990). For many participants, trying to normalize something that they knew was not normal (the numerous losses within the family) was not an easy task as described in this statement:

You’re kind of wondering about it [reason for deaths], so you don’t acknowledge it. It’s like being in a house and you don’t know if this place is haunted, but there’s weird stuff going on so I won’t look.

Similar sentiments were shared by another participant, who described trying to continue on with everyday life knowing that his current reality may be subject to change with little or no notice. The participant said, “It was just always there. You were always wondering is it going to happen to me, or how many more.”

Over time, participants’ ideas about risk begin to shift from being a suspicion to a belief that they are at risk for a genetic heart condition. These psychosocial and behavioural responses to a new meaning of risk are further examined in the following property that describes participants’ struggles to understand the meaning of being at-risk.

Property 2: The struggle to understand the meaning of being at-risk for oneself and others. This property captures participants’ struggles to assign a meaning of risk for them and their children and the conditions that influenced this experience. Building on experiential knowledge (that is, the sense of knowing that they are dealing with a fatal genetic condition), and knowing that science in its current state could not

answer their questions, participants spend a considerable amount of time constructing ideas about risk. Other studies have also referred to the difficulties experienced by participants as they try to decipher and assign meaning to competing ideas about risk in order to cope and make decisions about their health (Braithwaite et al., 2004; Cox, 2003; Cox & McKellin, 1999; d'Argincourt-Canning 2005; Etchegary, 2006a, 2006b, 2009; 2010; Frich et al., 2006; Hall, Suakko, Evans, Qureshi, & Humphries, 2007; Hallowell et al., 2006; Hunt et al., 2000, 2001; Marteau et al., 1995; McAllister 2002, 2003; Ponder et al., 1996; Walter et al., 2004; Walter & Emery, 2005; Weiner & Durrington, 2008, Weiner, 2009; van Maarle et al., 2003).

In contrast to what some other researchers have found (Hunt et al., 2001; Emslie et al., 2003; Walter et al., 2004), participants did not emphasize physical characteristics or personality as being significant in how they framed their own risk perception. Similar to Marteau et al. (1995) and Ponder et al. (1996), age, gender, physical signs of the disease, family history, and modifiable lifestyle factors were the most influential in risk assessment. These conditions are evident in all three phases of the genetic testing process, and used by participants as a framework to juxtapose with competing realities about risk, and to decipher and to assign meanings to risk; however, the relevancy of the conditions and the meanings assigned to them change in relation to the contextual dimensions. That is, participants drew upon the condition(s) that were most relevant and were in alignment with their beliefs about heart disease (and, later, about genetics) at different times throughout the genetic testing process. Depending on the contextual dimensions, new conditions deemed more relevant or "risky" emerge while other conditions assigned a status of being "less risky" are put on "the back burner."

As participants struggle to understand the meaning of being at-risk for themselves and others, the six conditions (age, gender, physical signs and symptoms of the disease, family history, modifiable lifestyle factors, and the concern felt for children in the family) were the most important conditions that shape the construction of risk and were the impetus to participants' creation of the *at-risk relative*.

Conditions 1 and 2: Age and gender. Age and gender²⁴ are two key conditions that form the image of what I am calling the "*at-risk relative*." The at-risk relative refers to participants' mental representation of the individual at risk for ARVC (similar to the construction of the coronary candidate depicted in the cardiovascular literature [Davison et al., 1989, 1991, and 1992]). The criteria for the *at-risk relative* that reoccurs throughout participants' narratives includes: being over 40, being male, exhibiting physical signs of heart disease, having a poor diet, lacking physical activity, living in a family with a history of heart disease, and, as science progressed, having a positive genetic test. This mental image of the *at-risk relative* is used as a reference point to construct ideas about risk, to make decisions throughout the genetic testing process in times of uncertainty (such as while waiting for genetic test results), and to predict the likelihood of a cardiac event.

Participants in this study frequently referred to age as a condition that influenced their risk perception and caused psychological distress. For younger participants,

²⁴ "Gender" as a condition is, specifically, the condition of "being male." Laypersons, as well as clinical experts, tend to emphasise the association of ARVC with being male, such that even when scientific knowledge provides evidence that women too are affected, the connotation of ARVC as being a "male" disease is dominant in the discourse about ARVC and shapes the perception of risk.

observing their older relatives experiencing distress as their age of “being at-risk” drew closer was, in turn, highly stressful for the young observers, as illustrated in this participant’s recollection:

I remember my father used to be always worried. I can remember seeing him sitting on a chair with his head down on his arm. I think it was always there in his mind that something was going to happen to him; because at that time his brother had died; and then he had another brother that died, and they were too young.

Once these young observers themselves approached the age of 40, or knew of another relative who was nearing 40, they too experienced a heightened sense of risk-anxiety. This finding supports the work of Cox and McKellin (1999), who reported that as at-risk individuals approach age of disease onset, there is a shift from the abstract sense of risk to an embodiment of risk, or what they refer to as an “intersubjective awareness” of risk.

Participants also identified gender as a condition that increased one’s risk status. One participant reported that it was years of observing a large number of young males dying in the family, including the recent death of a brother at age 33, which prompted the family to request the father’s autopsy report. He explained how the information from the autopsy solidified for him that he was at risk for a fatal cardiac disease prevalent in young males:

The coroner’s report was listing off all the things that had happened to [my father]. When I read that it was like reading about me. I knew. By the time they got around to telling me [*in the 1990s*] I had something and I had to go on medication, I pretty well knew I was a shoo-in. So I wasn’t totally surprised.

Condition 3: Physical signs and symptoms. During this pre-test phase of the genetic testing process, it is primarily the onset of physical signs and symptoms that are thought to be cardiac in nature that marks the entry into the illness experience and causes a tremendous amount of stress. As one participant explained, “I was scared to go

somewhere by myself because I used to just drop down, pass out. My heart used to go that fast.” Similarly another participant noted:

I recall the first time I was aware that I probably had this heart condition was that syncope incident in the fall of 1993, when I almost blacked out. For some reason, I knew that that would've been symptomatic of this heart condition.

In a similar vein, this same participant describes how the onset of physical symptoms sparked his awareness that he might be at risk for a heart disease similar to that to which his brother had succumbed:

Two months after my brother died I was working and I started to get electric shocks in my arm...and different things like dizzy spells. I kind of clued in then that something was up. So I went to the doctor to see what was wrong.

It is evident in these narratives that participants knew they were at risk for some kind of heart disease and in many regards had already started to identify and assign meanings to key conditions that placed them at-risk. As they started to exhibit physical signs, they immediately attributed this to the suspected heart condition that had claimed the lives of their family members. This awakening to the idea that they were at-risk was, for some participants, was not a shock, but rather something expected given the strong family history of loss. Still, for some, this awakening period was a time of much distress as they struggled to construct and attribute some meaning to the numerous losses.

A lot of time and psychological effort went into reconfiguring the mental representation of the *at-risk relative* to encompass the individual who was asymptomatic and who did not meet the *at-risk relative* profile, as many relatives did not have any symptoms prior to death. For most, this was not easy as they continued to spend a tremendous amount of time monitoring themselves and others for any visible signs of the disease. Any signs and symptoms that bore a resemblance to those of the *at-risk relative*

were perceived as “risky,” caused a heightened sense of risk, resulted in psychological distress, and marked the onset of the illness experience, which then required immediate management. These feelings are nicely summarized in this one parent’s narrative, “I am so afraid to take a chance that it’s just chest congestion and not something else, because [my son] said he feels tightness in his chest.”

Despite witnessing and listening to numerous stories of people who had succumbed to heart disease, some participants still struggled with the question of whether this was something that they should be overly concerned about. This struggle to assign a meaning to being at-risk is captured in this participant’s recollection (in the early 1980s, prior to gene haplotype analysis) of awakening to the fact that he may be at-risk and yet not overly concerned:

Nobody really paid much attention to it [heart troubles]. There is nothing wrong with us. We are all fine. I remember watching as a program on TV over in Italy, and there was this family; young fellows all dying, about 40 years old, and it was the men it seemed to be affecting. They were all dying off, and I remember them saying something about a study to see if it’s a heart problem that runs in the family. At the time I was saying maybe we got the same thing. Because by then it was Mom and my cousin; and where everything was happening in my family, maybe someone will do a study on us some day.

Responses such as this can be attributed to the fact that research investigating genetic heart conditions in the early 1980s was in its nascent stage. At that time, science did not hold any answers as to what was going on in these families. Some participants took the stance that although there is some kind of a heart condition in the family that affects young males, there was no point in dwelling on it because there were no answers. Still others took a different approach to normalizing their at-risk families, expressing that their

families' heart troubles were not dissimilar from many other Newfoundland families.

This comment summarizes that method of normalizing the experience:

We had no sickness like heart, no more than anybody else, probably less than some. We used to say we thought we were pretty good; pretty well off because we didn't have any major problems or anything. There was nothing wrong with us.

Similarly, even though other participants described physical symptoms of heart disease (episodes of being dizzy, light-headedness, etc.) they were thought to be of little concern.

"It was just once or twice a year and could happen to anybody," one participant rationalized. For these participants, in order to cope, they compared themselves to the traditional cardiac population. This strategy of normalizing the experience was a pragmatic approach, given the lack of available scientific technology in the 1980s to explain what was happening.

Condition 4: Family history. For those participants who awakened to a sense of being at-risk around the same time as haplotype analysis in 1998, the struggle to understand that they were at risk for a genetic heart condition was not as difficult. They had the support of relatives who had been through some clinical testing for what was, by 1998, thought to be a genetic heart condition; there was a viable genetic test that could provide some answers as to their risk status. For the younger participants who engaged in genetic testing in the early 2000s, awareness of their risk emerged as a result of a conversation with their parents. As one participant noted, "I didn't really find out until a couple of years later when they found out Mom had it. I'd say I was 16 when I figured out that we could have a problem with this too." Thus, the mental representation of the *at-risk relative* for these two groups of individuals expanded to include the genetic test

result. What would become an issue later, as these individuals moved through subsequent phases of the genetic testing process, was assigning a *meaning* to the genetic test result. This will be addressed in the next chapter, in the discussion of phase two of the predictive genetic testing process.

As participants struggled to make sense of (and assign meaning to) the idea of being at-risk, they continuously reflected on family stories of loss and on their own observations and experiences to understand what conditions predispose one to cardiac disease within the family. They made mental notes as to the number of cases of heart disease; they searched for predisposing events; and they collected data on the array of health outcomes within the family, similar to that done in property one, *living in a family familiar with loss*.

For some, the outcome of this familial information gathering and analysis was the prioritization of physical activity as a condition that precipitates the onset of symptoms, as is evident in this participant's account:

The last time I was out in the garden making rabbit snares, and I felt it coming on. I remember I saw everything going away from me, the house moved, and I started to go out.

It is noteworthy that the identification of the conditions labeled "risky" was often a family affair, being the result of many family discussions reliving the events of their family members' deaths and questioning the cause of numerous deaths in the family, as this commentary illustrates:

My brother discussed several things with me about the deaths in the family. It was always men that were involved. It seemed like Mom's brothers had all passed away young. She had a sister who was 49 when she passed away suddenly; and her other two brothers were on their way back from a funeral, had an accident and they were killed. So we wondered if the one who was driving had

a heart attack. I knew something was going on; but not until my brother really sat down and talked about it did I start to make the connections.

It is conversations such as this that highlight the importance of family story-sharing for constructing ideas about risk and the conditions that influence what is deemed “risky” or not.

What was particularly stressful for participants was the ever-present knowledge that they too could have a fate similar to that of their family members. One participant emphasized this concern, stating, “I knew that I could drop down dead having this condition ... I knew that my father had died with this same condition, so I knew it was a possibility.” Thus, the identification of any factors that could provide any insights into this disease and its management was important, particularly for those individuals who lived through the pre-test phase at a time when scientific knowledge could not yet explain what was going on.

Participants who lived through phase one (prior to the existence of genetic testing and prior to the discovery of the gene responsible for ARVC) are engaged in what Davison et al. (1992, 1991) describe as the work of a lay epidemiologist. Lay epidemiologists are those who gather information through personal observations or reports about the conditions and circumstances that predispose one to be at risk for a heart disease using lay epidemiology. Using lay epidemiology, similarities are noted that generate an explanatory framework for one’s risk of heart disease. This framework also serves to challenge or support other conditions thought to impact one’s risk for heart disease. The explanatory models created by the lay epidemiologist, which are shaped by the specific contextual dimensions (phase of the genetic testing process, experiential

knowledge, and scientific knowledge), are juxtaposed against existing beliefs about the conditions that influence risk and *the at-risk relative* profile, such as modifiable lifestyle factors.

Condition 5: Modifiable lifestyle factors. Based on the conclusion of the “lay epidemiologist,” decisions are then made to either ignore or dismiss the conditions that are understood to shape one’s risk. For example, they may decide not to adjust their lifestyle. Depending on the decision made, one either continues to live as they had before, now with the knowledge that they may have a fatal condition, or one alters their lifestyle in reaction to the new risk knowledge. For several participants who chose the “normality” path, the knowledge of being “at-risk” was very stressful, as is reflected in this comment: “the stress that I’d gone through trying to go out and live life knowing that I had something that could kill me, where I could drop dead. I think that was more stressful.” As noted by Etchegary (2009) in her research on HD, participants modified their diet and exercise regimes in an attempt to decrease their risk. This was evident in the narrative of one participant who gradually began to alter his lifestyle once he began to experience subtle signs of heart disease. “I gave up drinking, I gave up coffee, and I just tried to be physically fit, without overdoing it,” he recounted. Similarly, another participant notes that, being aware of his strong family history of heart disease, he made the decision early on to engage in a healthy lifestyle: “Even before I knew that ARVC existed, I knew there was heart disease. I said I have got to get myself to a place where I don’t smoke, don’t drink, eat well.”

Of those who emphasized the potential for modifying lifestyle as a means of managing or altering risk status, a healthy diet and a regular exercise regime were cited as

critical to avoid the fate of the *at-risk relative*. However, physical activity had two very different connotations in relation to modifying risk status. For most of the participants in this study, regular physical activity was associated with lessening risk. For some, however, physical exercise gradually became assigned as something “risky” when participants became aware of family members who had died during physical activity. Equipped with this new knowledge, the mental representation of the at-risk relative shifted to include exercise as something that may put individuals at increased risk. This new understanding of risk was hard for many to assimilate into their existing framework of risk, given that physical activity tends to be viewed as an activity synonymous with the prevention of heart disease. Hence, responses to the proposed link between physical activity and heart disease vary from person to person and can fluctuate over time for any one person, depending on whether and when a given activity is assigned a meaning of being “risky.”

The following narratives capture how the meaning assigned to physical activity is shaped and reshaped in relation to one’s experiences, knowledge about heart disease, and risk perception. This first quotation illustrates the fact that knowledge about being part of a family at risk for heart disease does not instantly translate to a heightened sense of risk, but rather causes one to look at modifiable lifestyle factors (such as exercise) as a health prevention activity. In such cases, exercise is not deemed a “risky” behaviour.

Growing up you kind of think to yourself there is heart disease in the family so you take care of yourself. I eat lots of oily fish, exercise, play basketball, volleyball, and try to stay in shape. I walk my dog and things like that.

Later, as the same person became aware that this type of heart disease manifests itself in young males during physical exertion, even causing death, the meaning of physical

activity was reassigned as being something “risky” that must be stopped. The participant noted that he then “stopped playing basketball.”

Condition 6: Concern for children. As participants awaken to and struggle with the meaning of being at-risk for themselves, they also experience an awakening to the fact that their children are at-risk as well. Although this awakening to being at-risk (for themselves and their children) happens at different times, the parents’ experiences in both instances are quite similar (except that parents reported that their concern for the wellbeing of their children was greater than the concern they had felt for their own wellbeing). While all participants had gradually come to terms with the fact that the males in their families die at a young age due to heart disease, they were surprised when symptoms of heart disease started to be noticed in their children. One parent described this feeling as follows:

I took my son to have a lymph node removed and he had an EKG done prior. The girl was checking him in and going over his file prior to surgery. My goodness the cardiograph does not look good. He was sent to a pediatrician; and hearing the history of the family, the pediatrician did say to me this is ARVC.

For many parents, this re-awakening to the notion that their children could be at-risk was distressing. As one parent said, “Every time one of the youngsters said they felt weak or sick I wondered, have they got [heart disease]?” Parents’ descriptions of this process indicate that as they juxtapose ideas about *the at-risk relative* against this new knowledge, they feel an overwhelming sense of anxiety as the concern about their own wellbeing is quickly replaced by concern for their children. This concern results in diligent monitoring of the children for physical signs and symptoms of ARVC, as evident

in this parent's account: "Every morning I got up, opened the door, and went in [to the children's room]. As long as I saw the chest rising or they were breathing, I was fine."

The psychosocial and behavioural reactions of parents to the news that the onset of ARVC is much earlier than what was known scientifically reflects what has been reported in the cardiovascular literature (Andersen et al., 2008; Farnsworth et al., 2006; Hendriks et al., 2005). The concern for the children is threaded throughout the genetic testing process and is addressed further in chapter 7 and chapter 8.

The experience of growing up in an at-risk family and witnessing the numerous losses due to ARVC shapes parents' beliefs about and behavioural responses to the risk status of their children.

Category 2: Struggling to Break the Cycle of Uncertainty

This category captures participants' experiences as they took part in multiple diagnostic screening tests and prescribed treatment regimes in hopes of gaining an understanding of the causative factor that was putting them at-risk. Participants in this study reported that they had agreed to participate in any clinical investigations that could provide insights into what was going on, knowing that the numerous deaths in the family were not a coincidence (that is, given their experiential knowledge of being at-risk).

Even though receiving a professional's suggestion that they may have a genetic-linked disease solidified what participants already knew (the experiential knowledge of being at-risk), the genetic testing process was a difficult time for all of the participants in this study. Many struggled with trying to give meaning to being at-risk, from a genetic stand point, in the absence of a definitive diagnosis, consistent treatment, or visible signs of the disease. Hence, participants' ideas about risk fluctuated between biomedical

models of disease causation, experiential knowledge, and the evolving mental representation of the *at-risk relative*. The two properties, (a) *making sense of and living through early clinical investigations and prescribed treatments* and (b) *acknowledging a genetic origin to risk*, describe participants' experiences as they struggled to understand what being at-risk meant for them and their families.

Making sense of and living through early clinical investigations and prescribed treatments. This property examines participants' experiences and the conditions that influence their experiences as they continued to search for answers about their risk status and the meaning of risk. The conditions that describe the variations in participants' experiences as they live through early clinical investigations and treatments are: (1) experts' knowledge and care, (2) available technology, and (3) the at-risk relative.

Condition 1: Experts' knowledge and care. Participant narratives reveal that as they engaged in diagnostic clinical testing (such as electrocardiograms, echocardiograms, and Holter monitor testing), they experienced a shift in risk perception as they looked towards traditional biomedical models of disease causation and to the opinions of experts to help them (re)construct their "reality" of their risk status. This is evident in the narrative of one participant who became aware of his risk for heart disease in his discussion with a specialist in the early 1980s, prior to haplotype analysis:

I remember asking him, "What do you think I should do?" He said "what, what you are asking me is if you were my wife, what would I tell you to do. I honestly don't know." I then said, "Well, is there anywhere or anything I can do to find out more about this particular heart condition?"

The significance of using clinical diagnoses to explain increased risk is evident in the fact that some participants travelled outside of the province in hopes of finding some

answers. Several participants recalled trips, made themselves or by others, to a neighboring province to undergo further clinical investigations, to get the opinions of experts, and to avail of new technology to manage their risk for heart disease. The following narratives illustrate this:

In 1988 he [brother] went to an institute in Ottawa, and he came back and he drew a picture for me that described how they were doing defibrillators.

I went to arrhythmia clinic in Ontario in 1994. I went up there thinking they might do some electrophysiology. I saw cardiologist he did a bunch of things: wall motion, signal allergy, EKG, and a stress test, I think. They did a 48-hour Holter, an adrenaline surge and an echo.

The desire to understand what was putting them at-risk was so strong that when a US research team approached some of the participants in the early 1990s to participate in a study investigating heart disease, they welcomed the opportunity. "I really would do anything out there, whatever the researchers wanted," one participant noted. For some, that research experience was the first indication that they may be dealing with something more than traditional heart disease that runs in NL families.

Participants described being both surprised and disappointed on a number of levels, about the use and role of the clinical and genetic testing process in helping them to understand their risk status. First, although they participated in a myriad of clinical tests, results were inconclusive and did not bring participants any closer to understanding their risk in most cases. This was emotionally draining, as evident in the narratives of several participants:

I had seven tests done [...] because they could not stimulate an arrhythmia [...] [the cardiologist] told me he didn't see any need of putting me on medication or doing electrophysiological studies [...] Just go on home and live your life as normal.

My brother and I was basically a couple of miles away from each other, and this was two months after another brother died. We were told at the time they couldn't do a whole lot for us; put us on medication and hope for the best.

Second, the lack of consistency in treatments offered and knowledge amongst experts about the management of ARVC contributed to feelings of ambiguity and frustration, similar to the reports of McAllister, Payne et al. (2007). The sense of frustration was evident in this participant's narrative:

If you're going to three or four doctors you are getting different things from all of them. You really don't know which one to believe; you just kind of got to hope for the best, really. I think it's all a luck thing.

Although participants described being disappointed with the inability of experts to provide definitive answers as to the cause of the numerous deaths within the family, they did appreciate the fact that gene discovery does take a long time, that health care professionals were providing the best treatment they could given their current state of knowledge, and that they were facing the possibility of a rare cardiac condition which challenges current biomedical models of heart disease. These sentiments are nicely summarized in this participant's statement:

I think the medical community does have a good handle on it [ARVC], from reading about it and having the knowledge and seeing so many people and dealing with people who succumb to health conditions. I think they are empathetic and understanding.

In contrast to the above, several participants were not as understanding: they could not comprehend why they were not being taken seriously given their strong family history, as this woman's commentary describes: "The first time I went to the hospital [...] I told them I was having a heart attack, the doctor looked at me and laughed and said, 'You're too young for that.'" It is understandable that a health care provider (drawing on their

own experiential and scientific knowledge about the “coronary candidate”) would make such a comment, as young women were not considered to be at high risk for heart disease. Without a definitive diagnosis of ARVC or any of the diagnostic criteria for heart disease, these comments seem reasonable. It is incidences such as this that highlight the fact that beliefs about risk that are strongly grounded in biomedical models of disease causation, such as the age of onset of heart disease, are very difficult to reshape for both experts and laypersons. Thus, when reconstructing ideas about risk, as in cases of new or rare conditions, it is important that educators acknowledge the origins of existing competing explanatory frameworks. As Gifford (1986) points out, knowing that health care providers cannot diagnose all conditions nor prescribe effective treatment 100% of the time, they must recognize lay experiential knowledge as significant in managing care (p.240). The reverse is also true: laypersons need to accept that experts do not have all the answers all of the time.

Despite difficulties in understanding the meaning of clinical investigations, participants still continued to attend scheduled tests in hopes of finding some answers. According to participants, however, with each subsequent testing visit, the wait became increasingly stressful: “When I’d get the letter for the appointment for the Holter monitor, the electrocardiogram, and the echocardiogram, I’d be sick for three days. I would be a state. I couldn’t sleep.”

In terms of reactions to the results of clinical diagnostic tests some participants reported that receiving a negative result was a relief because it implied that everything was fine. Other participants reported that receiving a negative result did little to alleviate the distress associated with feeling “at-risk.” As one participant noted, “Still in the back

of my mind I was thinking that if this could happen to my brother then it could happen to me.” This response was not surprising, given that these participants had watched many family members fall ill to the disease, that definite diagnostic tests were still being developed at the time that most participants were receiving testing, and that there continued to be a lack of general clinical knowledge about this relatively rare condition and related inconsistency in treatment.

Condition 2: Available Technology. The experiences of participants who had become aware of their risk for heart disease post-haplotype analysis were slightly different than the experiences of those who had been living with the sense of unknown risk for years. Reflecting the transient and contextual nature of risk, as cited by Shedlosky-Shoemaker et al. (2010) and Sivell et al. (2008), participants’ beliefs and initial awareness about risk were gradually shaped and reshaped in light of new technological advancements and research on ARVC. Thus, the experiences of individuals living in a family at-risk 50 years ago were very different from individuals living in these families during the 20 years of rapid advancement in ARVC genetic research (1980s to 2000s), and will differ again from the experiences of children growing up in these families today. As noted in the literature, once participants in this study were aware of advancements in gene discovery and technology, beliefs about risk were no longer contingent purely on experiential knowledge but developed analogous to scientific knowledge (Farrimond et al., 2010; Frich et al., 2006; McAllister, 2002, 2003; Walter & Emery, 2005). Participants are continually juxtaposing what might seem to be “competing realities” – experiential knowledge and scientific or expert knowledge – and

out of that juxtaposition, successfully (re)shaping for themselves a coherent sense of what being at-risk means.

It is noteworthy that at times of distress and when science could not answer their questions, regardless of genetic status or phase in the genetic testing process, and regardless of whether scientific advances and /or individual results could now offer scientific answers, participants drew on their experiential knowledge. The experiential knowledge in turn reshapes the understanding of the scientific knowledge (and the reverse is also true). This active use of experiential knowledge in combination with scientific knowledge has been found in other studies as being essential to how individuals construct their perceptions about their own risk (Cox, 1999; Hunt et al., 2000; McAllister 2002, 2003). It is the juxtaposition of the two sources of knowledge that enables participants—as bricoleurs (Strauss 1968)—to evaluate their own risk factors for heart disease and to make decisions about genetic testing. This narrative illustrates one participant's use of experiential knowledge to assign physical activity as something "risky":

He [father] went home and shoveled snow, and after when he came inside he just dropped in the porch. It was recorded as a massive cardiac attack. His father [participants' grandfather] died at 42 in the same way, even died in the porch in their house coming inside from doing work.

The same participant used this knowledge to make choices about his own health:

So I've always known that my father was 44 [when he died]; my grandfather was 42 [when he died]. And as you start to inch up I told myself, "I'm going to go and get physically tested when I am 40."

This group of participants drew on their experiential knowledge to formulate ideas about risk and at the same time focused on the hope that the experts and science could answer their questions about the causative factor behind this heart disease and offer options as to

its management. For participants for whom clinical testing became available during the “pre-test” phase, scientific knowledge was momentarily given precedence over experiential knowledge. Many participants wanted to participate in clinical investigations as early as possible. This is obvious in the narrative of a spouse who advocated for her husband to have clinical testing: “So I called one evening, about ten minutes after that genetic counsellor returned my call; three or four days after, they had appointments set up. Just as quick as that.”

Although participants knew that in order to find out what was putting them at-risk they needed to continue to participate in clinical investigations, having the clinical testing done was stressful. This is evident in one participant’s account of giving consent for a cardiac catheterization and the experience of actually going through the procedure:

So, I balled up the paper and threw it in the garbage and said, “I’m not getting that done.” I was frightened to death. And I just sat there and said, “Well I have to get it done,” so I took the paper out, smoothed it out, and signed my name.”

The only time I was really stressed was when I went in and the cardiologist was going up through the groin [to do the procedure]. He was late that morning with his surgeries so I had more time to be there in the bed and just worrying and getting so stressed out. I did not think I was going to be able to stay. Yeah that’s the most stressful time, having that done.

Awakening to the fact that they were at-risk, most participants wanted to participate in any clinical investigations that would inform them about their risk, even though at times doing so was particularly stressful. This momentary shift in knowledge acquisition and disease management has also been reported in the literature (Beery & Williams, 2007; Codori et al., 2001; Collins et al., 2005; Esplen et al., 2001; Halbert et al. 2004; Heshka et al., 2008; Johnson et al., 2002; Sheinfeld Gorin, & Steven, 2003; Watson et al., 2004; Van Roosmalen et al., 2004). When participants realized that they might not get definitive

answers as to the cause of their illness from experts, they referred to their personal knowledge and experiences acquired growing up in a family familiar with loss to try and make sense of what was going on.

Condition 3: At-risk Relative. The narratives of participants recounting their experiences of being tested reveal the complex ways in which risk is (re)constructed during the pre-test phase. Equipped with the framework of the *at-risk relative*, individuals being tested are eager to confirm that their mental representation is correct and reflective of a person at risk for heart disease. By being tested, they hope to discover any qualities absent from *the at-risk relative* framework that would place them outside of the “at-risk” group. That is, in the process of being tested, they hope to both distance themselves from, and reaffirm the validity of, the *at-risk relative* framework. This approach was somewhat unique in comparison with other literature that has looked at the construction of risk in other genetic conditions, (Braithwaite et al., 2004; Frich et al., 2006; Hunt et al., 2000; van Maarle et al., 2003; Walter et al., 2004; Walter & Emery, 2005; Weiner & Durrington, 2008, Weiner, 2009), and the cardiac literature that describes the coronary candidate (Davison et al., 1989; 1991; Emslie et al., 2001; Hunt et al., 2001; Walter & Emery, 2005), in that some participants in this study describe their experiential knowledge as the initial reference point for risk assessment, rather than the biomedical model as being the starting point.

What was disappointing for many participants was that despite engaging in a multitude of clinical testing they were no closer to answering their questions, as this narrative highlights, “They worked on ultrasounds and hooked me up to the heart machine and stuff like that, but they couldn’t pick anything up.” As participants in this

study were quick to realize that science in its current state on many occasions did not have the knowledge to provide the answers they wanted, they drew on their own experiences and knowledge acquired thorough years of living in at-risk families to cope and manage their risk. Once again they found themselves re-using and prioritizing lay stories of loss and the mental representation of the *at-risk relative* to create their own subjective interpretations of the meaning of risk and disease management. This is similar to the findings of other studies (d'Agincourt, 2005; Condit, 2010) and is an example of what Lippman (1994) depicts as the layperson as expert.

Property 2: Acknowledging a possible genetic origin to risk. This property captures participants' experiences as they come to terms with the fact that they are dealing with a genetically linked heart disease. The two conditions that describe the variations in participants' experiences are (1) their interactions with experts and (2) their family history

Condition 1: Interactions with experts. This moment of reconstructing risk out of the juxtaposition of experiential and scientific knowledge typically happened at the time when the genetic counsellor or cardiologist first approached the individual or family and addressed the possibility of a genetic link to the condition. For many, this solidified what many of them had started to entertain as a possibility for the numerous losses in these families, as these narrative noted:

“He [relative] did say at the time it could be genetic; and then a couple of years later I received the call from [genetics counsellor]. I think that's really when I realized it could be genetic and there could be something to it.”

For one particular family it was the comment put forth by the investigating pathologist that planted the idea that the recent deaths of two brothers were not a coincidence: "It's got to be genetic. There is something to this."

Based on that comment, this participant subsequently advocated for an investigation into the deaths of other relatives.

Condition 2: Family History. For several participants, the moment of knowing that they may have a genetic condition was a result of discussions with other relatives under investigation for heart disease, as one participant recalled:

I found out about ARVC when my aunt started showing some kind of symptoms. She didn't know what they were so she spoke to the cardiologist and he identified it as ARVC; from right there it sort of traced back.

For others, the reshaping of risk as being genetic in nature evolved slowly with the news of more relatives falling ill in a pattern that became too obvious to ignore, as this narrative described: "While mother was in the hospital, her cousin and her nephew were in there, and somebody had the foresight to say there might be something going on with genetics there."

In one family, the awareness of the possibility of a genetic condition presented itself serendipitously during a friendly neighborhood telephone conversation where two women were discussing heart disease and comparing relatives' stories of illness. By coincidence, the discussion lead to the story of a mutual friend who was under investigation for a heart condition similar to what the spouse of one of the women had been experiencing. Given the striking similarity in symptoms between the mutual friend and the woman's spouse, the woman immediately contacted the research nurse and the spouse began to be investigated for a genetic condition.

In comparison, some participants did not require reconfirmation that they were dealing with something genetic, as the large number of unexpected deaths was enough to suggest a genetic cause, as this narrative suggests: "Ten days after we buried the first brother on Friday, the second brother died." It is encounters such as these that highlight how participants juxtapose contextual dimensions (experiential knowledge, scientific knowledge, and phase of the genetic testing process) with conditions, as they assign meaning to their risk.

With the individual's introduction to the possibility of a genetic linked condition, the *at-risk relative* framework begins to reshape to include a genetic component. Participant narratives indicate that at this time of reconstruction of the concept of risk, individuals begin to try to cope with the awareness of this new genetic reality, taking time to reflect and re-shape their experiential knowledge, through a genetic lens, to see whether and how the new conception of risk resonates as meaningful.

Chapter Summary

In relation to the first phase of the genetic process, the construct *Awakening to a New Meaning of Being At-Risk* captures how one's social and historical context shapes risk perception. The first category, *making sense of numerous losses*, examines how the experiences of living in a family familiar with loss and efforts to understand the meaning of being at-risk for oneself and others influence early perceptions of risk. Participants' reactions to the news that they may be at-risk were varied; some participants had chosen to ignore this information, while others sought out the advice of health care providers. Others tried to normalize risk as part of their everyday lives. This approach was somewhat effective as it allowed them to carry on with daily activities as if normal;

however, in the back of their minds, many could not help but wonder what this news meant for them and their family members. Many participants used the mental representation of the at-risk relative as a reference point to construct ideas about risk. Others spent a considerable amount of time reflecting on their family history and gathering information that solidified the criteria of the at-risk relative such as gender, age, signs and symptoms of ARVC, and lifestyle factors. Underlying participants' efforts to make sense out of these numerous losses was concern for the children.

The second category, *struggling to break the cycle of uncertainty*, describes how participants try to make sense of and live through clinical investigations and prescribed treatments as they start to acknowledge a genetic origin to risk. Part of breaking the cycle of uncertainty is making sense of and living through existing clinical investigations and prescribed treatments as dictated by the experts, available technology, and the risk relative framework. In response, participants juxtaposed the knowledge of experts and science against their own experiential knowledge as they revisited lay stories of loss, challenged the criteria of the at-risk relative, engaged in available technology, and critiqued the care provided. For many, acknowledging a possible genetic origin to their risk was the only pragmatic response to break the cycle of uncertainty. That is, as participants revisited their family histories, connections between familial losses were so obvious that many felt they had to have a genetic basis. Hence, participants advocated for investigations into their family as to why so many were dying suddenly at such a young age.

As participants juxtapose the three contextual dimensions (scientific knowledge, experiential knowledge, and phase of the genetic process), the conditions that influence

and explain the varied experiences of participants throughout this process are loss, age and gender, physical signs and symptoms, family history, modifiable lifestyle factors, concern for children, experts' knowledge and care, available technology, and interactions with experts and the at-risk relative (see Figure 6.3).



Figure 6.3. Rubik's Cube Model: Summary of the Conditions that Influence the Psychosocial Process of Awakening to a New Meaning of Being At-Risk. Rubik's Cube ® used by permission of Seven Towns Limited, www.rubiks.com.

CHAPTER 7

DECIPHERING THE MEANING OF BEING AT-RISK

This chapter examines participants' experiences as they move into the second phase of the genetic testing process and continue the psychosocial process of *Constructing the Meaning of Being At-Risk*. The theoretical construct *Deciphering the Meaning of Being At-Risk* describes participants' experiences during predictive genetic testing as they are offered a predictive genetic test, make the decision to have genetic testing, wait to receive their result, and receive the test result (see Figure 7.1). This construct highlights how specific conditions and the three contextual dimensions (phase of the genetic testing process, scientific knowledge, and experiential knowledge) influence and are influenced by each other.

It is important to emphasize that the particular situatedness of any individual's story within the time frame of gene discovery shapes participants' experiences during the "testing" phase of the genetic testing process. That is, the experience of those who participated in clinical testing (e.g., electrocardiograms, echocardiograms) may be significantly different from those who were offered genetic testing in 1998 and those being offered genetic testing since the discovery of the causative gene for ARVC in 2007. The identification of the causative mutation for ARVC in 1998 provided some participants with a risk status of either "high" or "low" based on whether they had inherited the disease-associated haplotype. A "high" risk status meant they were 95% at-risk of having the gene; a "low" risk meant they were only 5% at-risk of having the ARVC gene. For others haplotype testing was inconclusive. In subsequent years, some were assigned a 50% risk of either being positive or negative. It was not until nearly a

decade later, in 2007, that the gene responsible for ARVC was identified and a somewhat definitive test available. Thus, for individuals who were being tested during that decade (the 1990s), the decision to have genetic testing was made in intervals corresponding to each scientific step towards gene discovery. Furthermore, the experiences of a person who had been living in an at-risk family for 50 years and witnessing numerous losses will be different from an individual who was born later and had not experienced the loss of immediate family members to ARVC. Hence, for any two individuals moving through the same three phases of the genetic testing process, experiences and decisions about engaging in genetic testing will be shaped by different contextual factors depending on the state of the science at the time.

The two categories, (a) *taking the first steps of the genetic testing process* and (b) *building one's risk portfolio*, describe the variations in participants' experiences as they decipher the meaning of being at-risk.

<i>Category 1: Taking the First Steps of the Genetic Testing Process</i>
<ul style="list-style-type: none"> • Property 1: Being Offered a Predictive Genetic Test. • Property 2: Making the Decision to Participate in Predictive Genetic Testing.
<i>Category 2: Building One's Risk Portfolio</i>
<ul style="list-style-type: none"> • Property 1: Waiting for Predictive Genetic Test Results. • Property 2: Receiving Genetic Test Results.

Figure 7.1. Construct 2: Deciphering the Meaning of Being At-Risk. Emerging properties and categories that describe the second phase of the psychosocial process Constructing the Meaning of Being At-Risk, during predictive genetic testing.

Category 1: Taking the First Steps of the Genetic Testing Process

This category examines participants' accounts of being offered a genetic test and making the decision to have genetic testing. It captures the conditions and contextual dimensions that shape how participants assign meanings to risk as they make the decision to have genetic testing. Threaded throughout this discussion are the psychosocial and behavioural responses of participants. The two properties, (a) *being offered a predictive genetic test* and (b) *making the decision to participate in predictive genetic testing*, describe participants' experiences as they face a new reality—the availability of a predictive genetic test for ARVC.

Being offered a predictive genetic test. All participants were offered a predictive genetic test for ARVC or were present when a family member was offered testing (as in the case of the spouses of at-risk individuals whom I interviewed). Some were offered haplotype testing in 1998; others were offered a more definitive test, as researchers moved closer to identifying the causative mutation for ARVC. It is with each offer of a genetic test that the influence of the contextual dimensions and specific conditions that shape risk perception are elucidated. The two conditions that capture the variations in being offered a predictive genetic test are (1) the availability of the predictive genetic test and (2) interactions with health care providers.

Condition 1: Availability of the predictive genetic test. Even though many participants had become aware on a cognitive and emotional level that they were living in a family at risk for a genetic condition (as described in chapter 6), it was not until they were approached by a genetic counsellor or cardiologist to have the predictive genetic test that the notion of being “at-risk” came to be translated into an objective reality. That is,

despite having had experiential knowledge of being at-risk (such as growing up in an at-risk family, experiencing the loss of close relatives, and participating in clinical diagnostic tests, all of which suggest risk for a genetic heart condition), they did not yet embody the notion of being “at-risk.” It was the interaction with the genetic counsellor that prompted participants to reconceptualize their experiential and scientific knowledge as noted in these comments:

Probably a couple of years later [following the visit with the pediatrician], I received a call from [Genetic Counsellor]. I think that’s really when I realized it could be genetic and there could be something to it.

I never associated it with a genetic disorder until I met [Genetic Counsellor].

As found in Cox and McKellin’s (1999) study, which explored how risk is socially constructed in families at risk for HD, being offered a genetic test often was the catalyst that caused participants to reassess existing ideas about risk, as they made the shift from experiencing an abstract sense of risk to embodying an intersubjective awareness of being at-risk for a genetic linked heart condition that could be identified by a predictive genetic test. This re-examination and reframing of risk in light of the availability of a predictive genetic test was serendipitous in that it provided to them an opportunity to rationalize the numerous losses in the family and to finally “know” (in a sense that is as embodied as it is rational) one’s objective risk. This is reflected in the statement, “I think as soon as I found out it was something that someone could tell me about, I said, ‘I need to know; I can’t sit here and think, “Do I or don’t I?””

Responses to the availability of a predictive genetic test were both optimistic and pessimistic. Many of the adult participants in this study who were presented with the

option to have genetic testing were eager to avail of this opportunity.²⁵ As one participant explained in discussing his eagerness to have genetic testing, “Whoever is doing [genetic testing], please find a cure, or find something that can be done to help.” Saliently embedded within participants’ narratives is an overwhelming sense of relief that their experiential knowledge was accurate: something was going on in their family that was putting them at-risk -- something that affected young males suddenly, and something that was fatal.

One of the first things that [Genetic Counsellor] told me is that with the men the first and last symptom is the same: sudden death. Knowing that there is not much of a warning genetic testing is an opportunity [to try and prevent these deaths from happening].

Participants expressed being thrilled when they found that the technology finally existed that would rid them of the ambiguity that they had lived with for so many years of not knowing the reason for the numerous illnesses and deaths within the family. Participants spoke of a feeling of relief, of feelings that something was finally being done, of feelings of hope that there would be a viable treatment, and of an overwhelming desire to know their risk status. Genetic testing was seen as the next pragmatic step in deciphering their risk status. These sentiments are reflected in the following comments:

It is a huge relief when you get contacted by the genetic counsellor.... You know someone is out there. Before that, it was this obscure disease, this condition that sits in the dark. You don’t know anything about it.

I don’t know if there is any cure for this disease [ARVC] but as technology improves and our knowledge of ARVC increases, it’s my hope that something

²⁵ As discussed in Chapter 5, Methods, I was unable to recruit participants who had declined genetic testing: their perspectives are therefore noticeably absent from this discussion.

will be found to help my grandchildren or perhaps my sons. They [participant's sons] are still only in their twenties and thirties.

I wanted to know.

I must say it was good to know that somebody was starting something, and [to] realize that probably down the road some answers about this disease [ARVC] could materialize.

When the test [became] available there was hope that at least we can delay this [onset of ARVC] with treatments.

In contrast to the positive responses to being offered genetic testing, there were a few participants who did not see the opportunity in the same light. For them, being offered a genetic test elicited a somewhat ambivalent approach, as they tried to understand and assign meaning to the opportunity to have genetic testing:

I'd go on the website and look it [ARVC] up. I was reluctant to talk to my wife about it. I read about it quietly, but I didn't discuss it a lot. There is reluctance; you don't even want to think about this condition because it is like this thing out there that you can't control.

If I don't talk about it [ARVC] then there is no confirmation right?

For one participant, growing up knowing that there was something wrong in the family that caused relatives to die suddenly at a young age, the news of a genetic test was at first welcomed; however, this was short-lived. Despite having genetic testing on several occasions, his genetic test continued to be inconclusive until 2007. For this person predictive genetic testing was described as the beginning of a long and stressful process. "I didn't find out or come to terms with it as a family curse.... until [Genetic Counsellor] appeared in '96, and it's been awful since." For others, the news of a genetic test caused them some unease, as they momentarily questioned their own theories about risk and gravitated towards biomedical models of inheritance to explain their risk. "They [family

members] started to talk about testing and how [ARVC] was genetic, that it was passed on, it could be 50/50.” This sparked participants to juxtapose their objective knowledge and experiential knowledge about risk.

The juxtaposition of objective and experiential ways of knowing risk inevitably led experiential knowledge to overshadow numerical measures of risk, especially for participants who felt that they were being offered a test that in their eyes was not 100% accurate. As found by d’Agincourt-Canning (2005) and Cox and McKellin (1999), experiential knowledge took precedence over objective estimates of risk. Thus, even at the preliminary stages of entering the genetic testing process, experiential knowledge and the conditions (gender, age, availability of a genetic test, and treatment) that account for the variations in risk are used to construct ideas about being at risk for ARVC, including ideas about its expressivity, its penetrance, and its available treatment options:

Before my sister died, she found out that her son had ARVC. He was diagnosed at [the] age of 21, and he had to have a transplant. He passed away in 2005. My sister, who also died from ARVC, had seven kids. Five of the kids were positive for ARVC.

I thought to myself that [the] defibrillator is only going to work for so long. My understanding of the disease was the heart itself is going to deteriorate. It [function of the heart] is going to get worse and worse. I knew there were treatments, but I was thinking [that], as of right now, a heart transplant is the ultimate treatment.

I came from a generation of all girls. My great-great grandmother had all girls. It [ARVC] was never thought of as a problem because most females don’t have any problems with this disease. There were no boys in the family until I had my son. Next thing you know he’s thirty-something years old and he drops down dead because no one ever knew that it [ARVC] could be passed on through the women. It was only then we started to realize that ARVC [could] be passed down through the women in the family to the men.

The above excerpts reflect the fact that individuals living in at-risk families continuously juxtapose competing realities about risk (scientific knowledge and experiential knowledge) in order to construct meanings of risk. This is a fluid process, in that the meanings assigned to being at-risk fluctuate in response to one's everyday life and the specific conditions that are relevant at the time of being offered a genetic test. Objective risk is not sufficient to assign meaning to being at-risk, as it is not possible to quantify that which is "felt" and therefore unquantifiable. Objective risk alone is not enough to understand one's risk; there are many competing contextual dimensions and conditions that influence risk perception, and objective risk is only a small piece of deciphering the meaning of risk. That is, as noted by Sivell et al. (2008), one's perception of risk is transient in nature and may be more contingent on one's subjective interpretations of risk and heuristics than objective estimates. It is experiences such as those highlighted in the previous narratives that elucidate the psychosocial distress endured while juxtaposing one's experiential knowledge with evolving scientific knowledge.

Condition 2: Interactions with health care providers. Offers of genetic testing occur during a face-to-face meeting with a genetic counsellor. It is during this preparatory phase that the genetic counsellor provides clients with information about the genetic test, information on available services, and an opportunity to ask questions. As one participant described it, "It was a lot of information, but [Genetic Counsellor] kept saying, 'do you have any questions?' If it was not clear [Genetic Counsellor] was available to answer any questions." Participants spoke of the emotional support provided at these encounters, as well. "I felt like the genetic counsellor cared and wasn't just doing

this as part of a job or whatever; you could see the sincerity.” Similar accounts of therapeutic relationships between at-risk individuals and genetic counsellors have been noted in the literature as being beneficial in decreasing the anxiety related to perceived risk (Meiser & Halliday, 2002).

The information provided in this preparatory period reflected the portrayal of the coronary candidate described in the literature by Davison et al., 1989, 1991, 1992, and participants’ representations of the “*at-risk relative*” described in chapter 6. This information was easily assimilated into pre-existing cultural beliefs or frameworks about risk and cardiovascular disease and thus did not cause a significant amount of psychological distress (as has been discussed by Douglas & Wildavsky, 1982b; Wildavsky & Dake, 1990). It was only when there was not a “good fit” between what the experts were telling them and what participants understood about genetics and heart disease, what they had observed going on in the family over the years, and what they had envisioned as the *at-risk relative*, that they experienced some anxiety.

Although participants generally reported understanding the information provided to them during this preparatory phase, that was not always the case. One participant, despite having had multiple conversations with the attending cardiologist, still apparently did not have a clear understanding of the disease:

When [cardiologist] told me I had hypertrophic cardiomyopathy he said its sudden cardiac death syndrome—you could drop down and you could die. That was really upsetting. I was kind of hoping ARVC wasn’t as bad as hypertrophic cardiomyopathy.

Experiences such as this remind us that laypersons may not have an understanding of the basic anatomy and physiology of genetics, might struggle with core concepts of genetics,

and may have misconceptions about the scientific facts when assigning meaning to their risk (see also Richards, 1996; Lanie et al., 2004).

Similarly, other participants, despite being aware of the signs and symptoms of ARVC, still found it hard to grasp how one could have such a fatal disease without any physical signs. Therefore, asymptomatic participants spoke often of being disinterested in learning more about the disease because they did not believe that they were at-risk. This was particularly evident in younger participants who had not experienced as many losses:

I thought that I didn't have it [ARVC]. I'm very active and never had any heart palpitations or any adverse effects at all. So it made me think there's nothing wrong with me.

I have no signs and symptoms that would make me think that I had anything wrong.

Incidences such as this can lead to individuals becoming disengaged in the genetic testing process or being reluctant to participate in recommended screening as noted in McAllister's (2002, 2003) Theory of Engagement. The Theory of Engagement describes the outcomes of one's cognitive or emotional beliefs that they are at risk for a disease. These participants were partially engaged; they were cognitively aware of their risk but did not have any significant anxiety related to this knowledge. Their understanding of their risk and subsequent decisions about genetic testing was based on the fact that they did not have any signs of the disease; hence, several had entertained the idea of disengaging in predictive genetic testing.

Property 2: Making the Decision to Participate in Predictive Genetic Testing.

Although the purpose of this research was not to discover the decision-making process

that occurs throughout the genetic testing process, as I started my interviews I discovered that a significant part of understanding one's risk was "teased out" as participants made the decision to have genetic testing. Thus, the focus of this property is to discuss how the decision to have genetic testing unfolds.

All of the 29 participants in this study, at some point, were involved in making the decision to have predictive genetic testing. Among the participants, six at-risk individuals and two spouses lived through early clinical investigations for ARVC in 1980s, up to and including haplotype testing in 1998, and through to the discovery of the causative mutation for ARVC in 2007; two at-risk individuals and their spouses entered testing between 1994 and 1997; and eleven at-risk individuals and two spouses entered the genetic testing process with haplotype testing after 1998. Only three participants had engaged in testing after 2007, and one participant was awaiting testing at the time of the interview (see Figure 1.3).

The property *making the decision to participate in predictive genetic testing* describes participants' experiences of making the decision to have genetic testing for themselves and for their children. I found that there were two distinct approaches to this, as the decision to be tested either (a) develops gradually over time or (b) happens so quickly that it is felt as a non-decision (see Figure 7.2).



Figure 7.2. Making the Decision to Have Predictive Genetic Testing For ARVC.

As I have argued previously in this work, the decision to have genetic testing is pragmatic, transient, fluid, and contingent upon the meaning that one assigns to being at-risk at any given time. It is pragmatic in that participants juxtapose the two contextual dimensions (scientific knowledge and experiential knowledge) against the conditions or specific factors that influence risk perception in order to make the decision to engage in genetic testing. It is transient in that as one's risk perception fluctuates so do the contextual dimensions and conditions that influence it. It is fluid in that the decision evolves with each new experience, with gene discovery, and it is contingent upon each reassignment of the meaning of being at risk for ARVC.

Participants' experiences when making the decision to have genetic testing are variable, and yet there are some underlying commonalities. They are variable in that participants' decisions either develop gradually over time (as the individual reflects on the pros and cons of having testing), or the decisions are made quickly with little to no conscious effort (that is, the correct decision is so immediately obvious to the decision maker that it is felt as a non-decision). However, no matter how quickly the decision-

making process unfolds, all participants juxtapose the two contextual dimensions (scientific and experiential knowledge) with the specific conditions that influence their risk perception.

While participants typically follow one process of decision-making (developing the decision slowly or deciding so quickly that it is a non-decision), for some decision makers the two types merge momentarily or change completely. For instance, for some who have been engaged in their decision-making process over time, their decision can shift to become a non-decision in light of new contextual information or conditions influencing their risk perception (for example, onset of signs of the disease). Importantly, this is not the same as an individual simply completing the decision-making process and arriving at a decision: rather, the decision-making process transforms – the pros and cons that were being weighed have now changed and the decision-making process shifts from being gradual to being sudden. Similarly, the person who had quickly decided one course of action or inaction may re-enter the decision-making process due to a shift in the contextual dimensions and conditions that influence risk perception (for example, new knowledge about the average age of onset of symptoms). The decision-making process, then, is dynamic, because the contextual dimensions or conditions that shape the meaning of being at-risk are constantly in flux and continually reshaping the perception of risk.

In figure 7.2, the bidirectional arrows represent the potential sharing of the contextual dimensions and conditions that influence risk perception. That is, when making the decision about whether to engage in genetic testing, participants draw on similar contextual dimensions and conditions. It is, however, the meaning that each individual assigns to these contextual dimensions and conditions that influences one's

risk perception and the speed at which one makes the decision. For example, for those participants whose decision developed over time—in the absence of a definitive predictive genetic test—considerable time had been spent trying to understand the meaning of the numerous losses within the family in order to assign a personal meaning to being at-risk. Although those individuals who took the non-decision approach in making the decision to have genetic testing do reflect on the meaning of the numerous losses within the family, they also do not engage in as in-depth analysis of this meaning, because they know that there is genetic test that will do this for them. This information is processed quickly and used as evidence to rationalize the merits of the predictive test in deciphering their risk. The state of scientific knowledge (availability of a predictive genetic test) alters how experiential knowledge (numerous losses) is understood and how the decision to have genetic testing is made for these two groups of participants (those who took a “developing decision” approach and those who took a “non-decision” approach), regardless of when they became aware of their genetic risk (1980s, 1998, 2007).

The dashed lines of the bidirectional arrows in figure 7.2 represent the fluid nature of risk perception and the varied contextual dimensions and conditions that influence it. The figure reflects the transient, fluid, and pragmatic nature of decision-making around genetic testing. The two approaches to making the decision to have genetic testing (developing and non-decision) are further described in this property *making the decision to participate in predictive genetic testing*. The seven conditions that were of most significance for all 29 participants as they made the decision to engage in genetic testing are as follows: (1) available predictive genetic test, (2) numerous losses or

deaths within the family, (3) physical signs and symptoms of disease, (4) gender, (5) test relevancy, (6) one's sense of relational responsibility or moral obligation to other family members to have testing, and (7) family support.

Condition 1: Availability of a predictive genetic test (comparing 1980s, 1998, and 2007). For those eight at-risk individuals and two spouses who became aware of the family's risk for heart disease prior to haplotype testing in the late 1990s, the decision to have genetic testing in 1998 and for some again in 2007 was something that evolved with gene discovery and with each offering of a more definitive genetic test. For these participants the decision to have genetic testing developed over time, similar to the process of decision-making described in Cox's (2003) work on HD.

Of the six at-risk individuals and two spouses who took part in early clinical testing (e.g., echocardiograms, cardiac stress tests, blood analysis) in the 1980s, when there was no predictive genetic test for ARVC, many knew they were at risk for something that "ran in the family." However, this awareness did not immediately translate into a heightened sense of risk, partly due to the lack of scientific evidence to support the experiential knowledge. Some participants took the approach that they were not at risk for a specific genetic condition but rather were simply living with something that they constructed as being normal for the typical Newfoundlander (that is, normal within a province that is well known to have a high incidence of cardiac disease). The sense of normalizing one's risk, initially sparked in the pre-testing phase and summarized through the construct *Awakening to a New Meaning of Being At-Risk*, continues to resonate in participants' narratives about their decision to have testing:

Well, we [family members] thought we might have something wrong. We were just thinking it was an arrhythmia; nobody really paid much attention to it back then. The fact that it might be genetic was something way out there. We thought there is nothing wrong with us.

According to many participants, their attitude in the early 1980s had been not to worry about something that was not tangible, not treatable, and that did not have a definitive diagnostic test. Thus, as noted by Esplen et al. (2001) in their research on HNPCC, the decision to engage in blood analysis prior to having any predictive genetic test was simply altruistic in nature for several participants in the early 1980s:

We were just happy to help out; any kind of information that they [researchers] could get from our blood, go for it... They [researchers] just wanted to do some testing, and it wouldn't affect anything in our lives; but we didn't really think about stuff like that back then.

This sense of altruism continued for participants as they made the decision about whether to have testing later in 1998 and again in 2007, as one participant explained, "Well, even if I die of this [ARVC], hopefully somewhere someone will stop it [ARVC] before the next generation is over."

Coinciding with each offering of a more definitive genetic test for ARVC (in 1998 and again in 2007), was a heightened sense of anxiety, as is summarized in this participant's description of weighing the pros and cons of knowing his risk status:

I started to think about it ... the problems of knowing and of not knowing such as if you don't have life insurance and medical insurance. Do you want to know? Here we [at-risk family members] had the chance to be tested ... but then we wondered what happens if there is something wrong. What do we do? What are they [health care providers] going to do? They are just learning now,, just starting to find out about this condition.

Although some participants agree that the decision to have genetic testing can be stressful, in contrast to other studies (e.g., d'Agincourt-Canning, 2006b; Klitzman,

Thorne, Williamson, & Marder, 2007; Landsbergen et al., 2005; Meiser, 2005; Smart, 2010), this anxiety was not a deterrent to making the decision to have testing. This may be explained by the fact that for participants who engaged in the process of decision-making over time, the period of contemplation prepares them emotionally and intellectually to make the decision to have testing and to deal with the potential outcome of the test.

The experiences of those participants who entered the genetic testing process in the early 2000s are similar to those who entered it in the early 1980s, with two exceptions. First, participants making the decision in 1998 and again in 2007 were presented with the option of a somewhat definitive genetic test immediately; thus, they did not have to endure the same level of ambiguity regarding their risk status as those who became aware of their potential risk in the 1980s when there was no genetic test for ARVC. Second, those making the decision in 1998 and in 2007 did not experience a prolonged period of contemplation when making the decision to have testing.

For these participants, accounts of making the decision to have genetic testing were vague: "I don't even remember going in and getting tested for the first time." This vagueness suggests the decision to have genetic testing did not cause a large amount of psychological distress but was rather, as Cox (2003) noted, something they knew they wanted, something self-evident, something requiring little conscious effort, and something believed to be the next logical step to deciphering their risk for ARVC. These sentiments are evident in the following statements:

I wanted to know. I don't remember any anxiety or stress of problems making the decision to get it done [genetic testing]. It wasn't a big decision.

It's just something you just got to do. You can go get tested and do something that may save your life, or you can just hope and pray that nothing happens to you.

I vaguely [recall making decision to have testing]. I had no hesitation about being tested because I felt, as long as there was a cure somewhere on the horizon, I would do anything.

For those who made the decision immediately, their convictions about knowing their risk were so strong that they believed that having the genetic test was the only thing that could relieve them of their distress.²⁶

Despite the fact that participants entered the genetic testing process at various phases of gene discovery, there was a similarity to the contextual dimensions and conditions when deciding whether to undergo genetic testing. The meaning that individuals assign to these contextual dimensions and conditions is what makes the decision-making process dynamic, fluid, and unique for each person. The meaning assigned to being at-risk can shift from the development of a decision to a non-decision or from a non-decision to one that requires more thought in order to decide in light of new contextual information or conditions that influence risk perception. Depending upon the context wherein the decision to engage in genetic testing occurs (e.g., year and state of the science), the influence of the contextual dimensions and the conditions on risk perception varies.

Condition 2: Numerous losses. As found by other researchers, growing up in a family culture enmeshed with stories of loss and witnessing first-hand the loss of a family

²⁶ The argument can be made that it is the time lapse between the making of the decision and the interview with me that contributed to participants' vague accounts of making the actual decision and giving consent. I do not believe this is the case, given participants' vivid accounts of the circumstances pre- and post-decision.

member were key factors that motivated participants to have genetic testing (Armstrong et al., 2000; Cox & Mckellin, 1999; d'Agincourt-Canning, 2006a, 2006b; Lerman et al., 1994; McAllister, 2002; Norris et al., 2009). The impact that a loss had on one's perception of risk and willingness to undergo testing was determined by the individual's psychological closeness to the loss. This is evident in these accounts of losing a relative:

My third brother was 43 when he passed away suddenly...In the back of my mind I was thinking if this could happen to him [brother] it could happen to me.

I didn't want to see my life turn out the same as my mother's -- burying two of her children before herself. I didn't want that.

Observing and listening to numerous accounts of illness and loss, many participants knew there was something "going on" in the family. These participants viewed a predictive genetic test as an opportunity to confirm or dispute their suspicions. Supporting the work of Lock and Nguyen (2010), participants' desire to know their risk status, coupled with the availability of a genetic test, supplemented their experiential knowledge rather than replacing it. As reported in the literature, in order to assign meaning to risk and to make the decision to have genetic testing, participants juxtaposed their experiential knowledge against the science of genetics, and it is out of this juxtaposition that risk was constructed (Boenik & Vander Burg, 2010; Cox, 2003; Sanders et al., 2007; Smith et al., 2002; Taylor, 2005).

Condition 3: Physical signs and symptoms. As found in other studies (Armstrong et al., 2000; Klitzman, Thorne, Williamson, & Marder, 2007), the onset of physical signs of ARVC was a key factor in heightening one's sense of risk, and thus prompting participants to engage in genetic testing. This is similar to what Cox (2003) describes as "taking the decision" (p.269). That is, the onset of physical signs of heart

disease raises one's conscious awareness that genetic testing is something that must be done in order to decipher one's risk. This decision was difficult for those who felt that the inclusion criteria of the coronary candidate or the *at-risk relative* did not apply to them— that is, they were young, physically active, and, most importantly, had no signs of cardiac disease. This soon changed as perceptions about risk were reshaped in light of overt physical signs and symptoms of heart disease that could not be denied as noted in the comments of two participants: "I think there were a couple of times I went and got checked out. I thought I was having shortness of breath," and "I had some pain in my chest... I went to see what was wrong." This supports Nelkin (1992) and her observation of it being the onset of physical signs and symptoms that represents the onset of the illness experience that causes participants to engage in the genetic testing process. The onset of physical signs and symptoms is an excellent example of how those participants whose decision to have genetic testing was "developing" would suddenly switch decision-making approaches to that of "a non-decision" (that is, suddenly knowing that one *would* be tested), as the meaning assigned to risk was quickly reshaped to reflect the new risk reality.

For some participants the onset of physical signs and symptoms, despite efforts to maintain a healthy lifestyle (diet and exercise), was frustrating. The only means by which participants felt that they could have some control over and be able to deal with their feelings of anxiety was to have testing. The benefits of taking a proactive approach to knowing one's risk is evident in several participants' comments, such as, "I find it's better to know [genetic status] than not to know because [ARVC] is not going to go away," and,

“I took a proactive approach. There is something lurking in the dark, and I went looking for it.”

The role that anxiety caused by uncertainty plays in making the decision to engage in genetic testing is evident throughout the genetic literature (Armstrong et al., 2000; Binedell et al., 1998; Christiaans et al., 2009; d’Agincourt-Canning, 2006b; Decruyenaere et al., 1997, 2003; Douma et al., 2008; Evers-Kiebooms & Decruyenaere, 1998; Evers-Kiebooms et al., 2002; Lerman et al., 1994; Meiser et al., 2007; Smart, 2010). As in the cardiovascular literature (Aatre & Day, 2011; Christiaans et al., 2009) and as argued by Lupton (1995), the sense of control acquired with making the decision to have genetic testing displaces feelings of uncertainty and psychological distress about not knowing one’s genetic status.

Condition 4: Gender.²⁷ For the women in the study who lived through gene discovery in the early 1980s, the onset of physical signs and symptoms of ARVC were particularly critical in constructing ideas about risk. Many women had taken comfort in the fact experts had advised them that they were at low risk for the disease in comparison to their male counterparts and did not require immediate testing. For these women the fact that they were at low risk seemed logical, as it supported their existing beliefs about risk, which were based on their mental representations of the *at-risk relative*, their experiential knowledge, and the cardiovascular literature of the time that framed heart disease as predominantly male (Davison et al., 1989, 1991, and 1992).

²⁷ See previous footnote 25.

It seems like it's more on the male side.... It affects males harsher [worse] than females. Females seem to be older when they contract it. The males have a sudden cardiac death.

I had the mind-set that women couldn't be affected by this; but I did think I could pass down a gene to my boys without having anything wrong with me.

Incidences such as these illustrate how beliefs about a health condition can shape one's decisions about genetic testing. The above quotations capture the impact that the opinions of experts have on risk perception.

Based on the fact that ARVC was presented by the experts and perceived by these women as something that did not readily affect women, women had taken a "developing" approach to genetic testing. This approach soon changed to that of a non-decision, as many women in the 1990s started to have physical symptoms deemed cardiac in nature and wanted genetic testing, as explained in this narrative:

At work one day I turned to my left, and everything went dark. I got light headed, and felt like my heart was doing flip flops in my body. I just put my hand on my head and said, 'please God, don't let me die.'

This is an example of how conditions such as gender may shape science. As one ARVC participant discussed, it was only through the many voices of women advocating to be tested after the onset of physical signs of heart disease that experts were forced to re-evaluate the criteria for risk of ARVC to include women.

Condition 5: Test Relevancy. For the participants, the meaning assigned to being at-risk is contingent on the relevancy that the contextual dimensions or conditions have for any given individual at a particular time. In keeping with research by Cox and McKellin (1999), and as described in the previous sections, the onset of physical symptoms, the continuous losses within the family, and the introduction of a more

conclusive test are relevant conditions that not only heightened one's sense of risk but also prompted participants to engage in genetic testing.

In regards to making the decision to have genetic testing, relevancy implies not only seeing the test as something important to undergo but also having a sense that this is the right time in one's life to have the test and feeling confident that one can cope with the outcome. As found in Taylor's (2005) research on HD, participants' narratives reflect an emotional, intellectual, and circumstantial readiness to know if they had ARVC or not. This collaborative sense of readiness is evident in various segments of several participants' narratives:

There are two different issues [with having testing]. One is personal and the other is for the public good.... I think that [genetic testing] is the only way we are going to find out if we can get rid of it [ARVC].... That is what went through my mind when I came in for testing.

There is no way that you can be prepared. It [genetic testing] is just something you got to do.

I never really took time out to come and get tested.... Now I had the time, and I said, 'I'll come and get it done now.'

This sense of readiness was seen as both exciting and scary at the same time as this narrative highlighted:

I wanted to know, but I was scared to know, because I know what Mom went through, especially the last year of her life; that was worrisome and scary and still is.

The concept of risk relevancy provides some insights as to why studies have found that children at risk for a genetic condition do not experience high levels of psychological distress (Cordi et al., 1996, 2003; Michie et al., 2001; Smets et al., 2008). That is, as noted in the literature, it may not be until later in life—when children reach adulthood and

are faced with critical life decisions such as marriage, reproduction, and employment—that the outcomes of genetic testing have meaning for or are relevant to their lives (Armstrong et al., 2000; Binedell et al., 1998; Decruyenaere et al., 1997; Duncan et al., 2007; Evers-Kiebooms & Decruyenaere, 1998; Evers-Kiebooms et al., 2002; Lerman et al., 1994). Therefore, it is worth noting that the three young participants in this study did not mention having gone through any psychological distress when making the decision to have genetic testing.

Condition 6: Relational responsibility. Included in parents' decision to have genetic testing was consideration for the impact that this decision would have on their children. Similar to other studies in genetics (Cox, 2003; d'Agincourt-Canning, 2001, 2006a, 2006b; Hallowell, 1999; Hallowell et al., 2006; Klitzman, Thorne, Williamson, & Marder, 2007) concern for the risk status of the children was threaded throughout parents' narratives. Added to this was concern for the children's future if a parent was ARVC positive. These sentiments are threaded throughout the following comments made by the parents:

That's my biggest fear of all: that my two kids are going to end up with what I have.

I chose to participate in genetic testing for my three boys. That was my reason: to help them.

I owe it to the children to get tested.

I said, 'Okay, I don't want to know [genetic status].' Then I thought, well, I have got two daughters, I have to know for them. Whatever about me, I have to know for them.

This strong sense of relational responsibility felt by participants to the children reinforces previous studies demonstrating that individual choices are socially embedded within the

nexus of our social relationships and the meanings that we assign to these interactions (Blumer, 1969; Glaser & Strauss, 1967; Mead, 1934; Sherwin, 1998). In this study it is the intersection of the contextual dimensions and conditions in which participants interact that shapes risk perception and the decision to have genetic testing.

This sense of relational responsibility described by participants in the second phase of the genetic testing process (that is, in the period when a decision about testing is being made) is not in the broader societal sense that Dean (1999b) and Wilkinson (2010) described in their works. That is, most participants did not conceptualize ideas about risk and the participation in genetic testing in terms of a broader societal moral duty; their sense of moral duty was in relation to their children.

Embedded within parent narratives was the turmoil of having to make the decision about whether to have their children tested.²⁸ It is quite obvious that making the decision for the children was not a non-decision but was something that developed. Parents constantly weighed the pros and cons of getting their children tested by drawing on multiple types of knowledge to make the decision, including conditions such as gender, age, and modifiable factors, as well as experiential knowledge such as the history of loss in the family.

Being a mother, giving birth to my children and knowing that something could happen to them ... I mean, I look at my mother; mom buried her two boys within ten days of each other.

²⁸ The average age of the children at the time of genetic testing for ARVC in this study was 14. Ethical issues related to genetic testing were not the focus of this study; however, there is a body of literature that does address the ethical implications of predictive genetic testing of children (e.g., Bailey, Skinner, Davis, Witmarsh, & Powell, 2008; Caga-anan, Smith, Sharp, & Lantos, 2011; Clarke, 2010; Emsenauer, Michels, & Reinke, 2005; Hogben & Boddington, 2005; Ross & Moon, 2000; Tozzo, Caenazzo, & Rodriguez, 2012; Valente, Ferrais, & Dallapiccola, 2008)

Every year as [Son] gets older; I know it's getting closer to the time to get him tested because he's more involved now with sports and things.... I don't know if [Son] fully realizes it.

I didn't know if it was best for them to get the blood work. Did I want to know? I didn't want to know it.... I couldn't make up my mind.

They [health care providers] are more watchful over my son than they are over my daughter. The [Genetic Counsellor] said, '[Daughter] is 14—but for whatever reason, the females fares a bit better than the males.' I'm not saying it takes [Daughter] off the hook. Obviously, it doesn't, but it may be a longer time before she has any problems. When my son was getting ready to have genetic testing, [Genetic Counsellor] asked me what I wanted to do. [Genetic Counsellor] gave me time to think about it, and I kept saying, 'No, I don't want to know.' [Genetic Counsellor] said to me, 'Okay, put it this way. We have lost some at the age of seventeen, so what if the same thing happens to [Son]. He didn't have a chance because you didn't get testing done. [Genetic Counsellor] said, 'I'm not telling you what to do, but think about it.' So I thought about it, and I said, 'I wouldn't be able to live with myself that way, either, so I might as well go on and get it done.'²⁹

Even when a decision was made to have their children tested, parents questioned the children's ability to cope with news of genetic testing and were concerned with how it would impact their lives. Knowing firsthand what the experience was like, parents try to protect their children from going through the same thing. The concern for the children is evident in these comments:

I am concerned for him [son].... It would be bad for me to be out of the picture, but I don't want him growing up [wondering whether he has ARVC].

I want [Son] tested, but I don't want him tested. I'd like to bury my head, but for his sake I just can't.

Well, I think about [Son], if he ever wanted a family, would it stop him.... It will be the happiest day of my life if I find out he doesn't have it [ARVC].

²⁹ Again, this is an example of the gendering of science and clinical care. See footnote 25.

Similar concerns regarding the wellbeing of children are found in other research looking at the psychosocial impact of genetics (e.g., Andersen et al., 2008; Armstrong et al., 2000; Binedell et al., 1998; Bruno et al., 2004; Cox 2003; Chivers-Seymour et al., 2010; d'Agincourt-Canning, 2006a, 2006b; Douma et al., 2008; Decruyenaere et al., 1997, 2003; Esplen et al., 2001; Evers-Kiebooms et al., 2002; Haddow, 2009; Meiser et al., 2007; Norris et al., 2009; Smart, 2010; Smith et al., 2002). Concerns for the wellbeing of the children are discussed in more detail in chapter 8.

Even though parents were confident that they had made the right choice to have their children tested, this decision was met, at times, with resistance by their young children (in their teens), as one mother described:

The middle one didn't want to know at all; it took a little while for him to be tested. The oldest had a bit of resistance. And the youngest one said I don't want to know. He was okay with getting the blood work done, but not with knowing.... He was upset at the time.

[Daughter] did not want any part of it. Her blood pressure went up...she had a panic attack.

In contrast, other children seemed to accept their fate at a level of unexpected maturity, as illustrated in this parent's narrative that described his eleven-year-old child's response to genetic testing: "The dice was thrown the day I was born, because either I got [ARVC] or I haven't got it.... Isn't it better if I know I got it or not?" In some respects it seems that living in a family with numerous losses had become a part of the children's everyday norm at an early age. Participating in genetic testing was a natural part of this norm. Thus, as with the older participants, genetic testing was a non-event.

For one young adult who had genetic testing in his childhood, living with ARVC was not something that he had envisioned for himself. In fact he expressed some

resentment to the fact that he was not included in the decision to have genetic testing as a youth but as an adult had to live with the consequences of the decision:

I was 16 when I figured out that I could have a problem with it [ARVC]. I didn't really want to be tested, but there was nothing I could do because I wasn't of age; and Mom wanted to know if I had it.... I don't even really remember going in and getting tested for the first time.... I didn't really want to know. You know what I mean? I'd rather just go through life and not know at all.... I was pretty much forced to do it by my mother at that time because I didn't really want to know, but she did, and I agreed to do it.

The long-term repercussion of this unilateral decision was also felt by a parent who questioned the viability of a good parent/child relationship:

My son is upset because he wants to go into this cooking course. To do the course you need to pass a medical. He is afraid that he is going to flunk the medical.... So, according to him, this [being ARVC positive] has got his life totally screwed up.... Our relationship is strained. He believes that he had this [ARVC] dumped on him and he can't do anything about it, so he's not a happy camper.

Although one's sense of relational responsibility to have genetic testing evolves out of concern for the wellbeing for the children, it is sustained by the support of one's family.

Condition 7: Family Support. A genetic linked condition can have a long-lasting effect on a family for many generations; thus, the decision to have genetic testing does not occur in isolation. Family support is imperative in making the decision to have genetic testing done. Participants' accounts of how they construct ideas about risk dispute aspects of Beck's idea of individualization of risk, and they support the work on relational decision-making (d'Agincourt-Canning, 2001; Etchegary, 2005) in that, although participants do take an active role in assigning meaning to risk, they are strongly influenced by familial relationships. For many participants the decision to have genetic testing was considered a family matter, as one participant explained:

It wasn't made plain to us that we were dealing with something genetic. We just talked it over with the family.... No doctor went up to [Relative] and said, 'Well, this is in your family, so you'd better get tested.'

For many families, the collaborative nature of the choice to be tested was apparent in this comment: "Eight of us went in. We had blood work."

The importance of family support in making a decision regarding testing is evident in this participant's recollection of refusing to have a child tested without the spouse present for support: "I said no, I can't let you test [Son]. Because my husband wasn't there, and I just couldn't deal with it at that time." Support of their family members was also significant in maintaining a positive attitude, as one individual recalled:

[Husband] has always seemed to me to be the strong person. If we ever went anywhere and there's bad news that had to come, as long as I had him to lean on, I felt very good.

Noteworthy is that the participants in this study did not discuss objective risk (that is, their scientifically based risk status) as being a key factor when making the decision to have genetic testing. This supports the findings of Cox (2003) and others who have argued that objective risk alone is not sufficient to assign meaning to risk and is not a primary factor in making the decision to have genetic testing. Similar to Rapp's (1999) work on the social impact of amniocentesis and studies examining decision-making around testing for other genetic conditions such as HNPCC (Bombard et al., 2008) and BRCA1/2 (Shiloh & Ilan, 2005), decisions to engage in genetic testing, for this study's participants, were contextualized in relation to the experiential knowledge formed out of their everyday lives and the contextual factors that impact that experiential reality. Decisions were not made based solely on Mendelian theories of inheritance. As noted by

several researchers (e.g., Lupton, 1995; Lupton & Tulloch, 2002; Johnson & Tversky, 1983; Krinsky & Goulding, 1992), the decision to have genetic testing is not a “rational” choice based on statistical analysis or objective measures but something that develops in response to heuristics, the subjective meanings assigned to risk that evolve out of individual interactions with the contextual dimensions (scientific knowledge and experiential knowledge), and conditions of one’s everyday life.

Category 2: Building One’s Risk Portfolio.

This category examines participants’ experiences and shifting ideas surrounding risk as they continue to decipher what it means to be at risk for ARVC. This category includes the time period up to and including waiting to receive test results and the immediate responses to having either a positive or negative test result. Included in this discussion are the diverse coping mechanisms that participants employed to deal with perceived psychosocial stressors. The two properties, (a) *waiting for predictive genetic test results* and (b) *receiving genetic test results*, capture how the participants assign meaning to being at-risk as they wait for and receive their test result, the conditions that influence this experience, and participants’ psychosocial and behavioural responses.

Property 1: Waiting for predictive genetic test results. This property highlights family members’ experiences as they wait for their predictive genetic test results. It describes the various psychological and behavioural coping responses employed as participants wait to receive their test result. The two conditions that influence these responses are the (1) *waiting period* and (2) *coping mechanisms*.

Condition 1: Waiting period. Participants reported that, although they had been fully aware that the discovery of the ARVC gene was something that would take a long

time, they had not been prepared for the psychological and behavioural stressors they would endure while waiting for their genetic test result. Impacting on this experience was the fact that the waiting period took longer for some, as the facilities to do the testing did not at first exist in the province, as this narrative stated:

[ARVC participant 1] had his test done, but it was two years before he got the results of his test; [ARVC participant 2] was a year. They [researchers] were switching where the testing was taking place. They used to have to send the blood to Germany, but now they were going to start to do genetic testing here, in Newfoundland.... So [ARVC participant 1's] blood got sent to Germany, then back to Newfoundland and had to be retested. That's why it took a bit longer.

For those who engaged in testing more recently, the waiting period was not as long: "A month, I think, to get the results back."

There is a dearth of literature that explores the waiting period immediately post-blood collection and prior to receiving one's test result. Most of the literature concentrates on the experience prior to genetic testing or the period after receiving the test results (e.g., Bleiker et al., 2003; Broadstock, Michie & Marteau, 2000; Croyle et al., 1997; Decruyenaere et al. 1999; Gargiulo et al., 2009; Hendriks et al., 2005; Lodder et al., 2001; Meiser & Dunn, 2000; Murakami et al, 2004; Reichelt et al., 2004; van Oostrom et al., 2003; van Roosmalen et al., 2004). There is a general consensus in that literature that those who experience psychological distress in the pre-genetic testing phase are more likely to experience higher levels of anxiety as they move through the genetic testing process. In keeping with that research, and supporting the research of Esplen et al. (2001) on HNPCC, the psychological distress experienced by ARVC participants in the pretesting phase continued to manifest itself in the waiting period. Threaded throughout participants' narratives, in their descriptions of waiting to receive their test results, were

expressions of feelings of a loss of control over one's life, denial, depression, anxiety, and uncertainty, as reflected in the following participants' accounts:

When you don't want to deal with something, you put it in the back of your head and try to forget about it; but you can't forget about it [ARVC] when you start having adverse reactions.... I'd wake up at night, my heart pounding.... I was a wreck.... I wouldn't say anything because I figured they're going to think I was nuts.... I was so afraid that something was going to happen.

Waiting for my test results was stressful and nagging. It did affect me every day, but it is one of those things that I needed to know.... It is there all the time in everything I did. We were working on our house for seven years, renovating, and in the back of my mind I was thinking, 'Am I ever really going to live in this place?'

I got in bed that night and—bang—just like that, I had a panic attack. I was thinking about it [test result]. I was in such a state my friend had to get up and actually get in the bed with me. I was so upset. I was distraught. When all of this [anxiety attack] is happening to you and you don't know if it is real [heart-related or not], that makes it even worse. If it were real, you'd have to deal with it, get medication, or do something. But when you don't know if it's real or not ... it was awful.

This overwhelming sense of worry and anxiety, described above, was not isolated to the individual at-risk, but was also experienced by the spouses: "I usually worry about ARVC all the time. I even went on antidepressants. I couldn't take it, because every time the phone rang I'd be shaking." Similar accounts of psychological distress in spouses living in at-risk families have been noted in genetic research on familial adenomatous polyposis (Douma et al., 2010), and in the cardiovascular literature (Dalteg, Benzein, Fridlund, & Malm, 2011; Hendriks, et al., 2008; Pihl, Fridlund, & Martensson, 2010).

Feelings of worry and concern were not as apparent in the narratives of those whose waiting period lasted less than six months. This suggests that individuals living in at-risk families for a prolonged period of time have a heightened sense of risk and

experience more psychological stressors in comparison to those who experience a shorter waiting period.

Condition 2: Coping Mechanisms. In explaining their experiences during the period of waiting to receive their test results, several participants described various coping mechanisms to deal with the stress throughout this period and to understand their risk. These included: avoidance, gathering information, constantly surveilling one's body, modifying lifestyle factors, and initiating a risk behaviour.

Avoidance. Some participants described trying to normalize the family history of cardiac disease as an effective coping strategy. These strategies of normalization as a coping strategy had also been employed in the pre-testing phase, as captured in Construct One: *Awakening to a New Meaning of Being At-Risk*. Normalization is identified as a strategy in the cardiovascular literature (Anderson et al., 2008). The successful use of normalization as a coping strategy is contingent upon one's ability to avoid communicating with others about their impending test result and ability to downplay the significance of having genetic testing as described in these narratives:

You try and hide it from your family. You don't want to talk about it.

The accusation [being ARVC positive] is there, but you are deflecting it all the time.

Sometimes I felt that the less I knew about it [ARVC], it probably wouldn't happen to us or our family. I could be one of the lucky ones that are not affected by it.

First, when I had the test done, I just put it out of sight, out of mind; but the more I lived with it, the harder it was to live with.

Gathering information. A second coping strategy involved gathering information. For some participants, becoming more knowledgeable about ARVC was therapeutic: not

only did it help them cope; it also helped them prepare for the test result and helped them make their own judgments about what it means to be at risk for ARVC, as evident in this comment:

Waiting for the results was stressful, but there is a relief because you will eventually know if you have it [ARVC] or not. I do have a tendency to read everything.... There are probably some people out there, if you told them about this [ARVC], they'd just take what the doctor told them. I'd be in my office; I'd get a minute and go on the internet. I'd look it up and read about it. You try to find out yourself whatever you can about the disease.

Surveillance of the body. In addition to the above psychological coping mechanisms, participants also employed behavioural coping mechanisms as they waited to receive their genetic test result. Similar to Kavanagh and Broom's (1998) study on embodiment of risk among women with abnormal pap smears, participants became increasingly conscious of their body's physical state as an indicator of one's risk status: "When you are aware of your body at all, and if you are worried about something, you are very conscious. You are conscious of your breathing and every heartbeat and every palpitation." This constant surveillance was both comforting and stressful, as a large amount of effort was put into trying to prevent a cardiac episode.

Modifying lifestyle factors. Avoiding a cardiac episode meant that participants once again revisited the criteria of the *at-risk relative* and the typical coronary candidate to identify and modify those conditions that put them at risk for a cardiac event. As in the Phase one awakening period, lifestyle factors such as diet and exercise received the most attention: "Okay, got to get yourself to a point where you don't smoke, don't drink. You eat well." For some of those who self-ascribe a high risk for ARVC, physical activity is assumed to heighten risk and is therefore restricted, as this participant noted:

I wouldn't exercise. I'd go for a walk here and there but wouldn't push myself. I was always afraid that I was going to trigger it [ARVC] and I would end up dead like my brother.

The same principle was applied to the children, as parents removed them from physical activities such as sports in fear of evoking a cardiac episode pending their test results. For others, adherence to this strict self-imposed lifestyle was at times lenient, as those who believed they did not have ARVC continued to engage in physical activity as a means of improving their heart health. Hence, responses to the proposed link between physical activity and heart disease fluctuate depending on one's beliefs about their risk for ARVC and the degree of risk assigned to physical activity.

Initiating an at-risk event. Another, less frequent, coping strategy was to attempt to initiate a cardiac event, based on the idea that if it was going to happen anyway, it would be better to provoke it to happen earlier to get it over with.³⁰ This was the experience of one participant who had stopped all physical activity pending his test result and found that period to be extremely stressful. Thus, at one point he purposively engaged in vigorous exercise in hopes of evoking a cardiac episode. The incident is captured in this narrative:

You get fed up with the whole thing.... Once you get challenged with something like this, it is like being bullied. You get pushed so far you're going to punch back. You feel like you are being pressured. I got to do something, and then you

³⁰ This is similar to descriptions of responses in other traumatic processes such as living through domestic violence, whereby a victim may take charge of the experience by initiating the feared event in order to get it over with (e.g., Walker, L. [(1979).], *The Battered Woman*. New York: Harper and Row).

go out and say to hell with this; I'm going out and do my thing.... You go out and work chopping wood from sun up to sun down.

It is behaviours such as this that foreshadow the potential psychological distress that participants may experience as they wait for their genetic test and supports the fact that psychological counselling is critical throughout all phases of the genetic testing process, not just pre-testing and post-receipt of one's test results.

Property 2: Receiving genetic test results. This property highlights how and from whom family members receive the news of their genetic status. It describes participants' immediate thoughts upon receipt of their genetic test results. The two conditions that describe participants' variations in this experience are as follows: (1) genetic test results and (2) mode of receiving test result.

Condition 1: Genetic test results (positive, negative, and inconclusive)

All of the participants in this study received their genetic test results either from the genetic counsellor or the cardiologist, in a private office. Participants' narratives, however, did not focus on this interaction but alluded to the fact that this was a positive encounter:

I found [Cardiologist] was very helpful.... I found it [interaction with cardiologist] very comforting. I guess it comes from being a professional. You are dealing with very serious issues day in and day out, and you know how to deal with people in a positive way.

Reactions to having received a positive or negative test result varied.

ARVC negative participants typically described an overwhelming feeling of relief ("It [negative test result] was a huge relief"). This sense of relief was somewhat bitter sweet for one ARVC negative participant who had lived for over a decade with an inconclusive test from the 1990s. "I cried for three days; I was ten years not knowing, so

it was like freedom to me.... Finally, I knew, one way or the other, whether I was high risk or low risk.” This participant’s narrative contradicts the literature, which reported individuals who had an inconclusive test perceived themselves as being low risk (Hallowell et al., 2002; Meiser, 2005). For this person, an inconclusive test in 1998 was experienced as if his condition were a high-risk status, which caused episodes of panic and distress.

For several participants, despite experiencing an overwhelming sense of happiness with receiving a negative test result, there were reservations about “publically” celebrating, due to an overpowering sense of guilt that others in the family may not be so lucky, as reflected in this comment: “You don’t celebrate because there are others that you’re waiting on, and knowing that other family member’s news wasn’t so great.” For some, celebrating took place in less overt ways, such as resuming physical activities. Similar accounts of “survivor guilt” have been cited in the literature (e.g., Codori & Brandt, 1994; Davies et al., 2007; Duncan et al., 2008; Hallowell et al., 2006; McAllister,; Mireskandari et al., 2009; Murakami et al., 2004, Sobel & Cowan, 2003; Tibben et al., 1993).

Immediate reactions to having a positive test result also varied. One participant reported that receiving a result of “positive” did not translate into a heightened sense of risk, because he had no physical symptoms and, therefore, there were no immediate actions required. In a similar manner, an ARVC positive woman described not being overly concerned about her newly assigned risk, given that her experiential knowledge of the at-risk relative and coronary candidate coincided with what the experts were saying: that women were not as affected by ARVC as men. “I don’t think I was overly concerned

about it [positive genetic test], to tell you the truth. I took a lot of confidence from the fact that I was female.” In other words, the positive test result had no bearing on her lifestyle; no immediate action was required.

For another participant, the news of having a positive test result was expected, given the extensive family history of loss and subtle signs of heart disease:

I was shocked and I wasn't shocked, given—because in our family we had all kinds of stuff.... There's a lot of it [ARVC] out in [rural area], where my father was from. I wasn't surprised because I had been having trouble [signs of heart disease], just every now and again, for a few years. Two months after [participant's brother] died, I was working, and I started getting dizzy spells and electric shocks in my arm, even though I wasn't touching anything. I kind of clued in then that something was up. So I went to the doctor to see what was wrong. When they did all their tests, I had what [brother] had, so they put me in the hospital. I was in there for three weeks. After about a week, they woke me up in the middle of the night and told me that my [participant's other brother] came into the hospital; he was just admitted.

Similar responses have been found in the cardiovascular literature that looked at how people understand and cope with the news that they are at risk for coronary heart disease (e.g., Farrimond et al., 2010). Comments such as these bring to light that at-risk individuals embody information about the conditions that influence their risk perception with more ease if they fit within existing frameworks about risk. It is only when one's existing framework is challenged, and one's existing risk perception is reshaped by a test result, that at-risk participants experience an elevated sense of being at-risk, causing psychological stress and disembodiment—a disconnect between what one believes to be the case and what is true according to existing scientific knowledge. This sense of disembodiment can cause distress, disengagement in the genetic testing process, or hinder adherence to preventative health behaviours in a manner similar to that described in other research (Heshka et al., 2008; McAllister, 2002; Sanders et al., 2007).

Reactions to the news of either a positive or negative test result were particularly striking for parents of children who had been tested. As noted in other studies (Meiser et al., 2002; van Roosmalen et al., 2004; Watson et al., 2004) for many ARVC positive participants, while they were concerned for their own wellbeing, it was the receipt of receiving the news that their child was ARVC positive that proved to be psychologically overwhelming, even for those who felt they had been emotionally and intellectually prepared: "I thought I would be able to handle it, but when I got the news [that participant's child was ARVC positive], it was devastating for me." It seems, regardless of how many times parents have prepared to receive their children's test results, they still experience anxiety, as one participant's reaction to their child being found ARVC positive illustrates: "I was all geared up for it [genetic test results].... But you were still thinking in the back of your mind, 'No, we can't go through this again.'" When receiving test results, especially in relation to children's risk status, participants appreciate the significance of having a support person present, as one parent who received the news alone notes:

I wish I had somebody there with me at the time, because it was hard to get that news. It came back that the two of them had ARVC, so that was the worst thing that could happen.

Condition 2: Mode of receiving results. Participants reported being informed about their test result in a face-to-face meeting with a genetic professional, but then subsequently receiving, by mail, the written notification of the test result. For some participants, this objective evidence was welcomed and reconfirmed their risk status. For others, the letter brought with it a familiar sense of ambiguity. While the letter confirmed that the experts were 95% confident that they were negative for ARVC, what stood out

more to some was that they were still 5% at risk for this disease. For those with a positive test result, the opposite was true: they were 95% sure they had the ARVC gene, but questioned whether the experts were wrong and whether they might be lucky enough to be in the 5% who did not have the mutation. The letter, then, was a catalyst that caused ideas about risk to be renegotiated again, with participants now appreciating that the genetic test results were not 100% definitive. “ I think [the letter] said I might be 95% or 90% [not at-risk].... So it’s still there. I still got that bit of doubt there.” Some had simultaneous feelings of joy and doubt as they received their genetic test results: “That was just marvelous—to know that I had gone from 50/50 down to five percent, but, still, it was there.” The only viable solution noted by participants, to resolve these doubts, would be to receive written notice that they were 100% positive or negative, which they never did receive. In any case, these letters were a critical link in facilitating the shift towards a new understanding of risk, one that they would revisit many times over the course of their lives as they moved towards constructing a new perception of risk.

Chapter Summary

Participants’ experiences in the second phase of the genetic testing process, *Deciphering the Meaning of Being At-Risk*, are captured in two categories. The first category, *taking the first steps of the genetic testing process*, describes how at-risk individuals construct ideas about their risk as they are offered a predictive genetic test (by health providers) and are faced with the decision to have predictive genetic testing for ARVC, or not. Each offering of a predictive genetic test required participants to juxtapose the contextual dimensions (scientific knowledge and experiential knowledge) against the meaning of the test being offered at the time of testing (that is, haplotype

testing in 1990s or a definitive test in 2007), and interactions with health care providers. For most, being offered a genetic test was a relief; not only did it provide a rationale for what was going on in their families, but it also provided hope of a viable treatment option. Interactions with health care providers were considered to be informative, and did provide emotional support but, for some, also caused a sense of uneasiness, as they remained ambivalent as to their *true* risk. All participants spent considerable time trying to understand and assimilate their objective and subjective knowledge in relation to existing beliefs about risk.

The decision to have genetic testing was considered a non-decision, an evolving decision, or some combination of these two. Participants' experiences with genetic testing as they made the decision to undergo it were influenced by the existing genetic test available, the numerous losses within the family, having physical signs of the disease, gender, relevancy of the test, sense of relational responsibility, and family support. As participants were offered a genetic test and made the decision to undergo testing, a common theme is that ideas and decisions based on one's understanding of risk are pragmatic, fluid, and transient.

Category two, *building one's risk portfolio*, examines how at-risk individuals construct ideas about risk as they wait for and receive their genetic test result. The time frame from having a genetic test to receiving the results can be stressful, requiring the use of coping mechanisms. Coping mechanism employed by participants were avoidance, information gathering, surveillance, modification of lifestyle factors, and initiation of an at-risk event. Participants' responses upon receipt of the genetic test results were varied and related to the mode of receiving the test result. Although most ARVC negative

participants were relieved, some did experience survivor guilt. Meanwhile, some ARVC positive participants were not overly concerned about their risk status, being that they had no visible signs of the condition, were female, and did not fit the profile of the coronary candidate. For others, being positive was something expected given their existing experiential knowledge. It is only when their experiential knowledge was challenged or when a child was found to be positive that they experienced increased psychological distress.

As individuals living in a family at risk for ARVC try to *Decipher the Meaning of Being At-Risk*, they juxtapose their scientific knowledge against their experiential knowledge and specific conditions. Figure 7.3 illustrates the interactions between these three factors.



Figure 7.3. Rubik's Cube Model: Summary of the Conditions that Influence the Psychosocial Process of Deciphering the Meaning of Being At-Risk. Rubik's Cube ® used by permission of Seven Towns Limited, www.rubiks.com.

CHAPTER 8

EMBODYING A NEW MEANING OF BEING AT-RISK

This chapter explores participants' experiences as they entered the final phase of the genetic testing process. The theoretical construct *Embodying a New Meaning of Being At-Risk* captures the experiences of participants as they began to adjust to the reality of living in a family at risk for ARVC and the news of their genetic test (see Figure 8.1). The process of embodiment of risk unfolded as participants juxtaposed the three contextual dimensions (scientific knowledge, experiential knowledge, and phase of the genetic testing process), against the conditions that influenced the manner in which the risk was experienced, understood, and evolved.

The three categories, (a) *adjusting to living with or without a genetic condition*, (b) *recognizing the reality of living in a family at risk for a genetic disease*, and (c) *looking towards the future*, describe the variations in participants' experiences as they embody a new meaning of being at-risk.

<i>Category 1: Adjusting to Living with or without a Genetic Condition</i>
<ul style="list-style-type: none"> • Property 1: Accepting and Assigning Meaning to Treatments Regimes • Property 2: Questioning the Accuracy of the Predictive Genetic Test
<i>Category 2: Recognizing the Reality of Living in a Family at Risk for a Genetic Disease</i>
<ul style="list-style-type: none"> • Property 1: Facing Everyday Worries and Challenges • Property 2: Coping with Barriers to Resources
<i>Category 3: Looking Towards the Future</i>
<ul style="list-style-type: none"> • Property 1: Trying to Maintain a Positive Outlook • Property 2: Living with Concern for At-risk Children

Figure 8.1: Construct 3: Embodying a New Meaning of Being At-Risk. Emerging properties and categories that describe the third phase of the psychosocial process Constructing the Meaning of Being At-Risk, post-predictive genetic testing.

Category 1: Adjusting to Living with or without a Genetic Condition.

This category captures the experience of beginning to adjust to the meaning of having a positive or negative test. For the younger participants, the time from blood collection to receiving the genetic test results was short; however, for the older participants, this was not the case. Many lived for years, until 2007, with an inconclusive test or the knowledge that the genetic test for ARVC was not definitive. For the participants in this study, adjusting to living with the outcome of the genetic test required them to come to terms with prescribed treatment regimes. This was reportedly difficult, as many, regardless of the test result, still questioned its reliability. The two properties, (a) *accepting and assigning meaning to treatment regimes* and (b) *questioning the accuracy of the predictive genetic test*, describe this experience. Evident within these two properties are the conditions that participants continued to juxtapose against the two contextual dimensions (scientific knowledge and experiential knowledge) as they embodied and adjusted to a new meaning of risk.

Property 1: Accepting and Assigning Meaning to Treatment Regimes.

In light of the presence or absence of physical signs and symptoms of ARVC, the meaning assigned to having a positive or negative test significantly influenced the ease with which participants accepted prescribed treatment regimes. The confounding factor of participants' understanding of available treatments added to this. The two common treatments referred to in participants' narratives that caused the most discussion were pharmacological management and the ICD. For many, these interventions were welcomed; for others, this was not the case. It was only as participants recognized that in order to survive, they would need some type of medical intervention that they begin to

embody a new meaning of risk. For a number of participants, the process of embodiment was easy; yet, for others, it was something that would evolve as they continued to adjust to living in a family with a genetically linked condition. The property *accepting and assigning meaning to treatment regimes* captures participants' psychosocial and behavioural responses to being offered and living with treatments that are indicative of disease progression. The key conditions that influenced participants' experiences are as follows: (1) the genetic test result, (2) the presence or absence of physical symptoms of ARVC, and (3) the believed efficacy of available treatments (medications and ICD).

Conditions 1 and 2: Genetic test result and physical symptoms. Participants frequently spoke of their genetic test result and the existence of physical signs and symptoms of ARVC as being significant conditions that shaped their perceptions of risk. These two factors influenced their decision to engage in prescribed treatment regimes or not. Figure 8.2 provides an overview of the relationship between having a positive or negative test, being symptomatic or asymptomatic, and the decision to have treatment or not.

Figure 8.2 illustrates how, for some participants, having a positive genetic test with no physical symptoms of ARVC caused them to question the need for treatment and, in several cases, to initially refuse treatment. Those who had received a positive test and were symptomatic generally welcomed the news of some treatment. For the majority of participants the fact that they were ARVC positive was enough to warrant treatment, regardless of whether or not they had experienced any physical symptoms of ARVC. Of those who tested negative for ARVC, they either wanted to continue with some method of monitoring themselves or their children (because they questioned the accuracy of the test

results, something that will be discussed later), or were happy having no further follow-up or treatments.

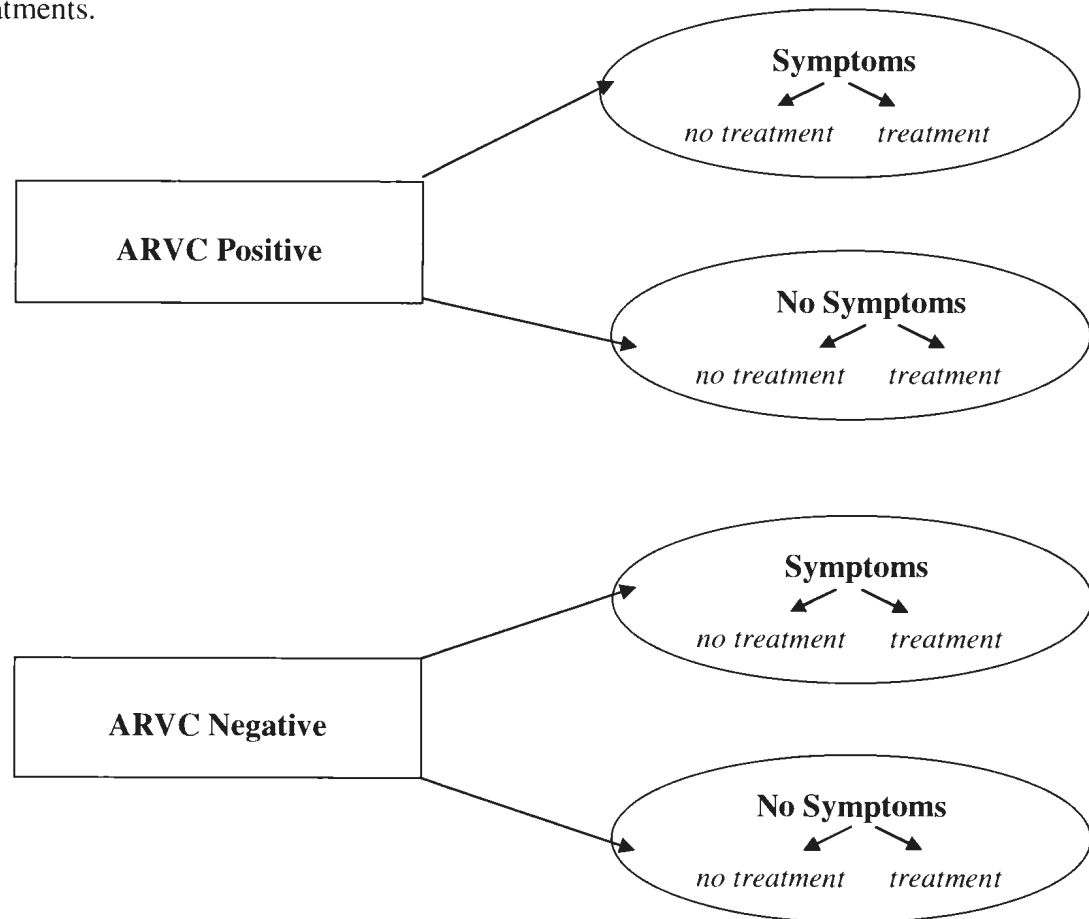


Figure 8.2 Relationships between Predictive Genetic Test Results, Disease Symptoms, and Treatments.

These responses can be explained by the fact that participants realized from an objective or clinical point of view that a positive test requires some medical intervention. However, this news was difficult to accept on an experiential or embodied level; being labeled as positive for ARVC did not immediately translate into a heightened sense of risk or anxiety, particularly for those who were asymptomatic, and therefore did not lead to the response that “being positive” could be relieved by some form of treatment. The same principles can be applied to those who are negative. That is, despite having a

negative test some participants still wanted to be monitored because it was not easy to ignore their experiential knowledge, and put trust in scientific knowledge. This may be because many participants had lived through the discovery of the causative gene for ARVC when the genetic test was not definitive and prescribed treatments were constantly changing. Other research has also shown that the meaning assigned to being at-risk may be more dependent upon subjective interpretations of risk and heuristics rather than objective estimates, and that subjective interpretations of what it means to be at-risk facilitate the ability to understand, cope, and make decisions about health in the context of everyday life (Cameron et al., 2009; Cox and McKellin, 1999; Sanders et al., 2007; Johnson & Tversky, 1983; Lupton & Tulloch, 2002; Sheldlosky-Shoemaker et al., 2010; Sivell et al., 2008; Weiner, 2009).

These two conditions (physical symptoms and genetic test results) in turn shape beliefs in the efficacy of pharmacological and ICD treatments, as the following discussion illustrates.

Condition 3: Believed efficacy of prescribed treatments: Pharmacological management. Participants' understanding and acceptance of the pharmacological management of ARVC was not something that took place in silo, but was understood in relation to the broader context of the state of available genetic testing at the time, and in relation to the individual's experience of having, or not having, physical symptoms of ARVC. These conditions intersect and shape each other in an individual's construction of risk and decisions about best treatment options.

In the early 1990s, the pharmacological management of ARVC consisted of a combination of medications such as antiarrhythmics, cholesterol reducing agents, and

antihypertensive drugs. It was common practice to be prescribed medications, as the ICD was not yet available: “It was 1991 when we were all diagnosed with some kind of heart problem and we were put on medications.” Medications were, at that time, and continue to be, individualized to a person’s physical symptoms, general health status and prior health history, and therapeutic response, as described by a spouse:

This is the second lot of medications that [Husband] has been on now. They [doctors] put him on one type of drug first but then increased it to one and a half. Then he had another attack [cardiac event] and the doctor put him on another pill. So now he is taking two kinds of pills.

Some participants who engaged in genetic testing in the early 1990s did not receive any treatment, despite undergoing clinical testing, as shown in the statement, “at the time [early 1990s] they were not doing defibrillators, and I wasn’t taking any medication.” This decision was based on the presence or absence of clinical signs of ARVC, or as in several cases, being assigned a low risk due to gender, as recalled in this narrative: “I remember the genetic counsellor said that maybe one woman had died from ARVC.” For some, then, it was the onset of symptoms of the evolving disease that determined prescribed treatments, as this participant explained:

Before I had an attack [cardiac event], I was not on medication. I had a defibrillator put in. It was an insurance policy the doctor told me. Since then, I have been taking Sotalol [antiarrhythmic drug]. I must say I have been pretty good after that.

The majority of participants accepted the introduction of medications regardless of when they completed the testing. They wanted to prevent symptoms of ARVC and avoid the onset of factors known to exacerbate heart disease, such as high blood pressure and

high cholesterol. This acceptance was credited to several factors. First, being put on medications for ARVC was perceived as pragmatic and logical; it supported existing beliefs and knowledge about the appropriate management of heart disease. Second, medication was considered to be acceptable because, despite altering lifestyle (e.g., diet), participants continued to have signs of ARVC, which they could not control. As was reported in previous research, participants started to align beliefs about ARVC and genetics with biomedical models of disease causation. That is, as the meaning of their illness was increasingly understood to be caused and controlled by their genes, it was assumed that the disease would be responsive only to medical interventions (Marteau et al., 2004; Senior et al., 1999; Senior, Marteau & Weinman, 2005; van Maarle, 2003). Third, recognizing that ARVC can be fatal and occur without any warning warranted preventative treatment. These three factors are reflected in the narratives of two ARVC positive participants who, despite modifications in their lifestyle, continued to exhibit signs of ARVC, causing them to turn to prescribed medications:

By the time I had my second appropriate shock [ICD firing] the cardiologist had talked about putting me on Sotalol [antiarrhythmic]. I said, 'No, I don't want to take Sotalol right now', so I didn't. When the ICD fired again I knew it was coming. I knew I was going to be put on Sotalol.

Once I settled down with medications I did not trigger it [ARVC] like I use to do before.

Most participants who received pharmacological treatment for ARVC stated that taking medications did not significantly impact their lives, "It [taking medications] does not affect me a lot because I can still do everything that I done before." On the other hand, several participants spoke of the adverse side effects of medications as being an

issue. This was captured in a participant's account of taking Amiodarone [antiarrhythmic] and going out in the sun:

I have been to the point that I was ready to toss them [medications] and not take any more. It gets to the point that you just get fed up with it. I told [Cardiologist] last time I was in to see him that I was going to stop, and he wants me to keep taking them until I get my heart catheterization done. Soon there has to be something done. I cannot do what I want to do the way that I am here now.... I don't know how to explain it but it is the simple things that affect you, such as being in the sun. I will go for a run on my skidoo but when I come back I have got a job to see anything. It is like someone put sand in my eyes. I can deal with the rest of it. I can deal with the attacks [cardiac events] but this is getting to me because it is continuous.

There were two ARVC positive participants who experienced extreme difficulty accepting that they needed to take medications to manage ARVC because they did not have any physical signs of the disease:

I am taking a whole Sotalol [antiarrhythmic] which I hate taking. I am taking Alsace [antihypertensive drug] which I don't need because my blood pressure is not up but it is suppose to have benefits for my cardiovascular system. I am taking Crestor [cholesterol reducing drug] but my cholesterol levels are always fine. Now I am taking drugs for what I have been all my life ... It changes the perception of who I am ... It's crazy ... If you had given me arsenic to take, it wouldn't have caused me so much stress and anxiety ... Every time I take that blue pill [Sotalol] I can't believe that my heart needs this bloody drug ... It [ARVC] is always there, and Sotalol is the constant reminder, twice a day ... I am not even symptomatic and my doctor had put me on Sotalol [antiarrhythmic].

For these participants, it is the meaning assigned to the absence of physical symptoms of ARVC that holds the most significance and not the positive genetic test. This finding supports the work of Cox (2003) and d'Agincourt-Canning (2005) that indicates that it is the personal meaning that one assigns to these conditions (e.g., physical symptoms, treatments) that influences risk perception. The construction of the meaning of risk is, however, conflicted by discrepancies between lay and expert understandings of what it means to be at-risk and the attributing factors that increase risk (as in d'Agincourt

–Canning, 2005; Allmark & Tod, 2005; Slovic et al., 1979) and, in the case of ARVC, prescribed treatments.

Although some individuals expressed feelings of dissatisfaction about having to take certain medications, they gradually resigned to the necessity of following their treatment regimes as captured in this comment:

I have got this condition. There is nothing I can do about it, so I am resigned to the fact that I probably have to take this bloody Sotalol [antiarrhythmic] and maybe Amiodarone [antiarrhythmic]. I don't want to take Amiodarone at all. To me that's nastier than Sotalol.

Another ARVC positive participant spoke of being treated for hypertension and hypercholesterolemia when he did not have either condition. In fact, he started to watch his dietary intake as if he had hypertension and hypercholesterolemia. This supports Nelkin's (1992) description of the pre-symptomatic ill: individuals who are assigned the label of being ill start to take on the illness role over time without being physically ill.

Condition 4: Believed efficacy of prescribed treatments: ICD management.

Participants described their experiences as they were introduced to the ICD as a viable treatment option for ARVC, as they assigned meanings to the ICD, and as they gradually began to accept the ICD as something they needed in order to survive.

For some, the initial reaction to being offered the ICD was one of anxiety, as the device represented an increasingly serious disease state that could not be managed solely with medications. Later, as they gained more experience and knowledge with ICD management, the ICD shifted from being something that symbolized an increase in risk state to a technology that prevented them from being at-risk. This shift is similar to that described in Lupton's (1999) work on the embodiment of pregnancy. She explains how

risk perceptions are constructed, shaped, reshaped, and regulated by technology and experts. In this study, participants' experiences were illuminated as they spoke of positives and negatives of having an ICD that resulted from the juxtaposition of experiential knowledge with scientific knowledge and the conditions (e.g., symptoms of disease) that influence risk perception.

For most participants, having an ICD gave them a sense of solace and comfort in that it could prevent the loss of family members in the event of a cardiac episode. Having this lifesaving device helped participants cope with having ARVC and restored a sense of confidence in their bodies. The negative aspects of having the ICD were that it were caused psychological distress as they tried to anticipate, prevent, and understand when and why it fired, framed with regards to psychological distress throughout participants' narratives. This distress was shown as they tried to accept that the ICD was something they needed to survive. They attempted to anticipate, prevent, and understand when and why it fired, as they struggled to anticipate, prevent, and understand when and why it fired as they, as they tried to cope with their body image, and as they worried about the impact of the ICD on spouses coping with the disease.

Positive aspects of having an ICD. Regardless of when participants entered into the genetic testing process (clinical testing in 1980s; haplotype genetic testing in 1990s; and definitive testing in 2007), being offered the ICD decreased psychological distress and restored a sense of control in most participants. The ICD was described as being a "lifesaver," an "insurance policy," or a "necessity" that instilled a sense of "security" assuring them that they were "safe" in the event of a cardiac episode. Similar descriptions of the ICD are found in the literature (Bose, Hamilton, Flanagan, Carroll, &

Fridlund, 2005; Dickerson, 2002; Kamphius, Verhoever, de Leeuw, Derksen, Hauer, & Winnubst, 2004; Kantor, Bullinger, & Gal, 2012; Morken, Severinsson, & Karlsen, 2009; Zeigler & Nelms, 2009). The meanings that participants assigned to their own risk status in relation to the ICD (that is, how they believed the ICD would affect their risk status) shaped the decision about whether to have one inserted.

As participants reflected on their experiential knowledge (e.g., family loss, family history) and scientific knowledge about ARVC and genetics (e.g., the sudden onset of symptoms), they accepted that they required some medical intervention if they wanted to survive. Most participants wanted the ICD implanted as soon as possible either for themselves or their children in order to manage and cope with their psychological and physical needs as explained by several participants:

Just imagine if the children never had the ICD. They would not have any chance whatsoever [if experienced a cardiac event]. Now you know there is something there that will defibrillate them if needed.

I could not wait for the time to come for the children to get an ICD because I felt it would make me more at ease.

I wanted to have it [ICD] because of the sudden death [in the family]. ...If there was a chance that the defibrillator was going to give me another shot at life I wanted it as soon as possible.

It seems like the ICD has proven itself.

It [ICD] is a safety net.

Faith in the ICD's capabilities was reinforced each time the ICD fired and saved a life.

As found in the literature (Kantor et al., 2012), these episodes provided evidence for the participants that they had made the right decision to have the ICD inserted.

I woke up in the morning and when I was out in the bathroom I heard a bang. I knew my defibrillator had fired, and that my heart was racing. I did not pass out this time. I did pass out one other time. Then the ICD did give me a shock and brought me back to life. It was only eight months after I had it implanted.

Adding to the sense of confidence in the ICD was the feeling that even if one were to engage in a “risky” behaviour such as physical exercise that evoked a cardiac event, the ICD would fire and shock one back into a normal heart rhythm, as this participant recalled:

Another time the ICD fired was when I was helping my husband bring in some wood. I was feeling tired that day, and I'd been lying around and I was getting over a cold. I thought maybe if I go out and do something, go outdoors and get a bit of fresh air, I would feel better. I was only out there five minutes, bending over picking up one chunk of wood at a time and throwing it in the wheel barrel, and the defibrillator fired.

This metaphor of the ICD as a “life saver” had a powerful impact on how individuals embodied their risk. This strong sense of faith in the technical capabilities of the ICD prompted some women who had received a positive haplotype test in late 1990s to approach health care providers to have an ICD implanted as soon as possible, as this woman stated:

I fought to get the defibrillator. If there was a chance that the defibrillator was going to give me another shot at life I wanted it.... So, I fought to get the defibrillator because I didn't really have any symptoms other than the inverted T waves.

These women were shocked by the reluctance of the medical community to treat women with ARVC the same as their male counterparts and prescribe an ICD. One participant posited that it was only after the media released a story of a woman who wanted an ICD because of her extensive family history of sudden cardiac death that the medical

community was challenged to reassess the treatment of women from families with

ARVC:

I was put on Sotalol [antiarrhythmic], and [Cardiologist] said I was not a candidate for a defibrillator because I was a female. Five years later, [cardiologist] had a woman who went to the newspaper and insisted that she have a defibrillator put in because she had a strong family history of ARVC. The lady had the defibrillator implanted and a couple of weeks later it fired and saved her life... Then all of a sudden I got a call, 'You need a defibrillator.' So I asked the cardiologist, 'What changed for me?' After so many years coming here and you telling me, 'No you are fine, except for the premature ventricular contractions, but it is not putting you at-risk.' I was on Sotalol which made me miserable and all of a sudden I am a candidate for and defibrillator?' He said, 'We have changed our mind-set on how it affects females.' So I had the ICD put in and gave up the Sotalol.³¹

As in the previous two phases of the genetic testing process (*Awakening to a New Meaning of Being At-Risk* and *Deciphering the Meaning of Being At-Risk*), the above example illustrates how individuals' beliefs about the conditions that influence risk shape the trajectory of scientific knowledge.

Negative aspects of ICD. Despite the many positives associated with living with an ICD, for some, having the device was not a pleasant experience. Living with an ICD has been noted in the literature as causing varying levels of psychological distress, including distress related to its management, which negatively impacts at-risk individuals' quality of life and that of their spouses (Bosle et al., 2005; Friedmann et al., 2006). Issues related to the management of the ICD that caused the most concern were: (a) accepting that the ICD was needed and anticipating its firing, (b) understanding why the device had fired, (c) trying to prevent it from firing, (c) body image, and (d) impact on spouses.

³¹ This is yet another example of the gendering of science and clinical care. See footnote 25.

Accepting the ICD and anticipating first shock. For many participants it was the realization that they needed the ICD to manage ARVC, coupled with the fear of it firing, which caused the most distress. Some felt that having the ICD took away their once “carefree” attitude and replaced it with a constant state of anxiety, as one participant described:

I had a cardio-version in the shower, so now every time I take my shower; it's pretty much a dribble. I don't want to have water going full force because I want to be able to get away from the water [if the ICD fires].

For some, who had engaged in clinical investigations in the early 1980s and genetic testing in the 1990s, being offered a defibrillator meant they that could no longer be managed with medications because they were getting sicker:

It [firing of the ICD] has changed the way I see my future because I think I am going to die young. I don't think that I am going to live to be in the 70s ... probably ten-years from now I will be gone, anything more than that is a bonus.

Regardless of the time that participants received an ICD they all shared a sense of dread in anticipation of the first ICD shock, and this caused significant distress. Both the children and the adults felt this sense of dread:

I [parent] have often heard the children say they were afraid that the defibrillator would cut in.

When I [ARVC positive adult] first had my ICD put in I went home and I was fine. After I got home I got nervous. I got nervous about having the ICD.

I [ARVC positive child] was frightened to death [to get a shock] at first!

For the nine participants whose ICD did fire, the first incident was the most memorable. It left them with feelings of anxiety, and made them fear the possible outcomes of subsequent shocks, when a shock would occur, and what it would be like:

I was sitting in a hotel room by myself [when he had the first shock]; there was nobody else there . . . So, if I never had the ICD the chances probably are that I would not have made it because I was alone [when cardiac event happened].

Nobody realizes what you go through when you have these shocks... I was scared to death.

That's what scares me: having another shock.

For one person, the anticipation of having a shock caused panic attacks: "I started to have panic attacks, at least one a night there for awhile." Similar feelings of anxiety prior to and post-shock have been documented in the ICD literature (Kamphius et al., 2004; Zeigler & Nelms, 2009). Two studies have reported that this post-shock anxiety was so severe that several participants considered removing it (Dickerson, 2002; Morken et al., 2009).

Gradually, as found in other studies, the apprehension associated with the ICD firing subsides, as participants become accustomed to its firing and recognize that this is to be expected (see also Bolse et al., 2005; Kamphius et al., 2004; Wheeler, Pretzer-Aboff, Hardie, DiSabatino, Saylor, & Lucey, 2009). For the participants in this study, having a defibrillator meant there was a high probability it would fire in response to a cardiac event. Many had the same opinion reported by participants in the study by Agard, Lofmark, Edvardsson and Ekman (2007); the ICD is an insurance policy and having a shock is just part of this policy. Living with an ICD eventually becomes "normalized" (as in Bourdieu, 1977), as described in this comment:

To me, after I had the first shock all the rest of them did not seem to be so bad, you do not find they hurt as much. It is probably because you are use to it after it happens. The last one was when I was sailing on the deck of the boat and I knelt down by a fellow. He told me all he heard was a grunt, and when he looked around I was down on my knees. He asked me, 'What happened?' I replied, 'The defibrillator just went off,' and I just got up and went on with my work.

From this discussion it is evident that individuals' understanding of technology plays a vital role in shaping ideas about risk and one's ability to cope with that risk. It solidifies the fact that an abnormal gene mutation (such as ARVC) can be conceived as something normal and the condition can be treated like other normal conditions (other heart conditions requiring an ICD).

Understanding why ICD fired. Over time, as participants adjust to living with a defibrillator and consciously make efforts to offset the negative connotations associated with the firing of the ICD, they begin to compile a kind of etiology of firing, with different styles of firings of the ICD being associated with different meanings of risk. For some the firing of the ICD more than twice warranted treatment: "If the ICD fires twice in a row you have to go to the doctor and get checked out, to see if there is anything wrong with it." For others the events surrounding the firing of the ICD were significant: "If you are not passed out it is probably a problem with the machine; whereas if you are passed out and it happens twice, it is probably something with your heart." Participants reported that as they became more familiar with the workings of the ICD, they soon started to assign their own meanings to the shock, the number of shocks requiring medical attention, and their risk.

The first shock we used to panic and go right on to town. Then it kind of mellowed... Now it's, we'll call the cardiologist in the morning and whenever he can get us in. So we've kind of eased off.

Revised interpretations of the ICD shocks come after many trips to the hospital post-ICD firing, which resulted in no medical intervention. Participants soon realize that the ICD fires because it is doing what it is designed to do; it fires to prevent an arrhythmia. In this

way, the experience of living with the constant threat of having the ICD fire at any time becomes normalized as part of everyday life. Despite this gradual normalization of the experience of living with an ICD, several spouses of individuals with ICDs expressed concern about the reliability of the ICD, saying, for example, “you still wonder if the ICD is going to work all the time.”

Preventing ICD from firing. Participant narratives emphasized the time and energy that goes into preventing firings of the ICD. Those who had an ICD seek to understand the function of the ICD and significance of each shock: was it inappropriate (the result of device malfunction), or was it appropriate (that is, fired in response to a cardiac event) and, if an appropriate firing, what were the precipitating factors? The rate of perceived success in managing the disease is measured by the effectiveness at identifying the modifiable factor(s) that triggered the ICD to fire and one’s ability to abstain from the “risky” activity. The need to engage in self-surveillance and in the self-regulation of lifestyle factors to prevent the ICD from firing is emotionally draining. It requires those with an ICD to resume their role as the lay epidemiologist (described in relation to Phase One: *Awakening to a New Meaning of Being At-Risk*), and to make a decision as to whether there is a correlation between the ICD firing and their behaviour.

Preventing the ICD from firing also has the effect of increasing anxiety about one’s escalating risk and sense of mortality (a point also made by Palacios-Cena et al., 2011). For some participants, despite modifying the factors identified as “risky,” the ICD continues to fire, which is very discouraging, as noted in this narrative:

Since I’ve had these appropriate [ICD firings], all I do is spend my time monitoring my heartbeat, and it’s driving me crazy ... You get negative reinforcement every time you get shocked.

Despite attempts to alter behaviours based on evidence gained from previous events, repeated firings of the ICD can trigger what has been described in other literature as a fatalistic attitude (Davison et al., 1989, 1991, 1992; Marteau & Lerman, 2001; Senior et al., 1999, 2000). Participants come to believe that it is futile to try and maintain a healthy lifestyle because no matter what is done, the ICD will still fire. Even though the participants in this study did get frustrated with the continuous firing of the ICD, they did not forego a healthy lifestyle. However, several participants noted that it would have been helpful to have some sort of formalized psychological support to help them adjust to the ICD firing: "What I think lacks, once you become symptomatic with this like me, is counselling."

Modifiable factors. For participants, successful identification of the factors that had caused the ICD to discharge was a relief, particularly when the precipitating factor was something that could be easily modified (for example, by altering sleep, diet, or medications). The finding that having a sense of control over the conditions that influence risk facilitates coping has been similarly reported in other studies (Hallas, Burke, White & Connelly, 2010; Howell et al., 2006; McAllister, Davies, et al., 2007; Senior et al., 2005; Sobel & Cowan, 2003; Vodermaier et al., 2010). On the downside, knowing that there are factors that could be controlled also led some participants to experience an enormous amount of distress, as they spent a lot of time and effort monitoring themselves or others for modifiable factors that might cause the ICD to discharge. Similar to Kavanagh and Broom's (1998) findings in women with cervical cancer, it is the continuous surveillance required in the ongoing management of ARVC

that contributes to participants' anxiety. The examinations of the factors thought to have triggered the ICD to fire are compared in order to create some frame of reference as to which activities to avoid, as captured in this participant's narrative:

The first time I triggered the ICD I was running with my dog. If I had not been running with my dog, it would never have happened. I know exercise can trigger the ICD to go off. The second time I set the ICD off was five months later. I triggered it through fatigue, and I think fatigue is well documented in the literature as causing heart problems. ARVC and fatigue, like a lot of heart conditions, are linked. I could have prevented the first shock; and if I had been more careful with sleep, I wouldn't have had the second one.

Similar accounts of the psychological stress that participants endured as they tried to predict when the ICD would fire have been reported in cardiovascular literature (Dickerson, 2002; Hallas et al., 2010; Morken et al., 2009; White, 2002). As explained in other research, a large part of the ICD related distress could be explained by participants' resistance to relinquish control over their lives to the ICD (Dickerson, 2002, Flemme, Hallberg, Johansson, & Stromberg, 2011). That is, they could not understand how their health could be so dependent on a machine and so resistant to modifiable lifestyle factors. Hence, the experience of being at-risk for ARVC is markedly different from other typical cardiac diseases given its resistance to modifiable factors.

ICD function. A big part of being able to determine an appropriate or inappropriate shock and adjust to the ICD was having a good understanding of how the ICD functions and an ability to troubleshoot technical problems. Participants spoke with ease about the day-to-day functioning of the ICD; they reported having a good understanding of the functioning of the ICD, as explained to them by health care providers. This included an understanding of when the ICD should appropriately discharge, and within which heart-rate parameters. Having a good knowledge of how the

ICD functions has been associated with better acceptance of the ICD (Wilson, Engelke, Sears, Swanson & Neil, 2012).

Your electrical system misfires and you go into ventricular fibrillation and the ICD will pick up on this and it will fire and knock you back into a normal heart rhythm... If your heart rate goes up past the setting, like 188 and stays there for so many seconds, the ICD is going to defibrillate you whether you are in ventricular fibrillation or not.

Participants also had a good understanding that technical difficulties with the ICD, such as displacement of wires or reprogramming, could cause an inappropriate shock, which is nicely summarized in this statement:

[Child] had his defibrillator put in September 1. On the 11th or 15th of September, he came out of school [high school] one day and his machine went off. So when he went to get checked out the doctor noticed that it was not his heart this time; it was a wire that had to be moved one centimeter in the heart. So he had to go back and get the wire moved.

Body image. Living with an ICD can be challenging in other ways. Several participants described difficulty with adjusting to the weight of the ICD, being nervous about having it in their body, having a sense of discomfort (see also Dickerson, 2002), and feeling weak and having problems sleeping (also reported by Bolse et al., 2005). Participants reported that over time, they came to realize that the ICD did not interfere with activities of daily living. One participant, however, argued that the ICD affected his body image in such a negative manner that he took action to have it removed. The following narrative captures this experience:

After I had the ICD in for six or seven months, I did not want it. I went through a lot to get the ICD taken out. I had meetings with anyone you could imagine; psychiatrists, nurses, genetic counsellors, doctors and members of the medical board. I had to go through all those steps just to prove I knew what I was doing and that I was actually sane. Where I am a small person it would grind against my ribs. Not only that, it was visible, about half an inch outside my body. Everyone

kept on saying, 'don't worry about it... nobody cares.' But people do care, and people will stare, and I didn't like it myself. So I did not want to have no part of it... Being a gay male, your visual appearance is very important. I could not go anywhere without any confidence... I would be constantly thinking are people looking at it? Are they staring at me? It was constantly on my mind. So my only option was to take it out, and if I died that was it... It was what I needed for me.

An alteration in one's body image, due to visibility of the ICD and incision scarring, is a factor identified in some literature that has caused ICD recipients psychological distress (Hallas et al., 2009; Sowell, Kohl, Sears, Klodell, & Conti, 2006). Alterations in body image impacting one's social relationships were also reported in individuals living with FAP, a genetically linked CRC wherein polyps develop in the intestine that require surgical intervention (Mireskandari et al., 2009).

Although the above participant experienced a tremendous amount of anxiety related to a disruption in his body image, he eventually reassigned his personal meaning to being at-risk post-cardiac arrest that rendered him critically ill for some time. Following this he had the ICD reinserted. Adjusting to the ICD for this participant was an ongoing process where he slowly learned to accept that he needed the ICD to live: "I have got no choice but to deal with it [having ICD]... I'd rather not have it, but I have to stay alive, so I got to deal with it." A significant part of his adjusting focused on trying to embody the ICD by incorporating it into his own body image:

The new ICD does not bother me as much, where it is less visible. I put on makeup to cover up the scars, so it makes it a bit easier. It's not too bad but if I touch it I can feel it ... I can actually cover it up a lot easier now. I put my arm in a certain position.

This participant's experience is very unique as it is the only documented case that illustrates an individual with ARVC having the ICD removed due to psychological distress because of an altered body image. On one hand, it represents resistance to

technology; however on the other, the reinsertion of the ICD following a cardiac episode reinforces beliefs about its efficacy and “life saving” capabilities.

Impact on Spouse. A systematic review by Van Den Broek, Habibovic, and Pederson (2010) examined the literature on the emotional distress of partners of individuals with an ICD. That review found that spouses experience a substantial amount of psychological distress linked to ICD management. This study supports the finding of this review. The psychological distress experienced by spouses is reflected in the following narrative of a spouse as she described what happened when her husband received a shock that rendered him unconscious and resulted in injury:

[Husband] came up to wash hands and take his pills, and I heard nothing before I heard that bang. I saw [husband] down by the steel corner [of the sink] and the blood pouring was pouring out of his head. All you could see was the white parts of his eyes. I said, ‘Oh my God! [Husband] is dead!

It is incidents such as this that remind us that the health of family members impacts the entire family unit, not just the affected individual.

Property 2: Questioning the Accuracy of the Predictive Genetic Test. The history of the ARVC gene (see Figure 1.3) depicts the discovery of a definitive test specific for the ARVC gene in 2007. A definitive test is one that can determine with a high degree of certainty that one has the gene or not. Regardless of when participants received a particular genetic test result they all questioned its accuracy. The two conditions that account for the variations in participant experiences as they question the accuracy of their predictive genetic test are: (1) the news of a more definitive test and (2) discrepancies in ARVC management.

Condition 1: News of a more definitive test. Participants entered into investigations for what became known as ARVC at three critical points; (a) in the 1980s, when a diagnosis of heart disease was based on clinical investigations only; (b) in the 1990s, when haplotype genetic testing was offered but not a definitive test; and (c) in 2007, when a definitive genetic test for ARVC became available. Evident in participants' narratives, regardless of when they entered the genetic testing process, is that risk perception changes constantly and is contingent upon the meanings assigned to the three contextual dimensions (scientific knowledge, experiential knowledge, and phase of the genetic testing process) and the conditions at any particular point in the genetic testing process. With each step towards gene discovery, participants were presented with a more definitive genetic test. Each offering of another test caused participants to compare their objective risk with their subjective ideas and experiences about what it means to be at-risk. Added to this was the fact that many participants had received their genetic test results using haplotype analysis and thought that this was the most conclusive test available.

It was difficult for some participants upon finally receiving a negative test result to disregard segments of their experiential knowledge that conflicted with their assigned objective risk: "I guess you've lived with it so long you still think maybe you are positive." As was the case in the previous two phases of the genetic testing process, participants in this post-testing phase used their family stories to create personal theories about inheritance and to assign meaning to being at-risk:

I find it very hard to sleep. I am always wondering, 'Can this be true [I am ARVC negative]. Will my sons be alright?' The worry is always there. I don't think it is

ever going to go away. Since my teens it [the worry] has been there, and now that I am at this age, I don't think it is every going to go away. It is still there.

It [death] is so ingrained in this family that I don't ever think it's ever going to go away, the worry part of it.

This strong reliance on one's experiential knowledge explains why ARVC negative individuals still experienced psychological distress, and reflects what has been described in the literature on HD (Codori & Brandt, 1994; Decruyenaere et al., 1999, 2003; Gargiulo et al., 2009) and BRCA 1/2 (van Oostrom et al., 2003). This co-existence of competing beliefs about being at-risk ("I'm at risk"; "I'm not at risk"), manifests as a sense of disconnect between the embodied sense of being at-risk and the rational or objective knowledge of not being at-risk, as the following two narratives illustrate:

I have had to leave my classroom and run next door to the next classroom. It just overcomes me and it still does... I got to say, 'Hold on here! Hold it. It's not real'. But old habits die hard.

Panic attacks have happened to me four or five times since the genetic counsellor gave me the news [ARVC negative], especially when I talk to my [relative] who has a defibrillator.

In order to cope with this anxiety, participants used knowledge of their objective risk to put things into perspective. As noted in the following narrative of one ARVC negative person maintaining, this balance was not easy:

When I talk to [other relatives] that have this condition, I feel the panic welling in me. I feel the knot in my stomach. I can feel myself getting hot, my heart racing, and then I got to stop myself, 'Stop it! This is not you! You do not have this'.

Those with a negative test also questioned the accuracy of the test. After having been so many years without a diagnosis, some could not help but think that researchers may have missed something. In fact, many participants still spoke of doubt as to the accuracy of the genetic test:

There is still a nagging, because you think that maybe the gene is delayed [that is, the individual is in fact positive for the mutation but this has not yet been detected].

Even now I still got doubt in my mind. Even with what they have discovered ... I am still going to carry on worrying over it, over my son and daughter.

For the one participant who had gone from being 50% at-risk in 1998 to being negative in 2006, adjusting to this news was difficult. Living with a 50% risk for so many years, he found it difficult to embody this new risk status, as described in the following narrative:

They couldn't decipher my genes, they couldn't tell me. So I stood for the longest time at a 50/50 chance... And those ten-years were horrid... When they told me back in 2006 that they had unraveled my genes, that I was low risk... and that they may follow-up with me every two or three years. I came home and I think I cried for three days; finally, I knew one way or the other whether I was high risk or low risk. I was ten-years not knowing, so that was like freedom for me. That was just marvelous; to know that I had gone from 50/50 down to 5%; but, still, it was there. It was still there, but I was starting to believe what my husband kept telling me: "Somebody has to be like your father. [Participant's siblings] were like your mother, but somebody has to have your father's genes." He was trying to ease me and get me through these panic attacks and the misery. So I started believing that then. I am like Dad and I don't have this gene, and I don't have any symptoms; but then I thought a lot of times women's symptoms don't really show. Every now and then it would surface and I'd still have the panic attacks, but not nearly like I did.

The participants' comments in this property remind us that objective risk is not the most significant indicator of one's risk perception. As noted in some other research, it is how one assimilates knowledge of the genetic test result into one's existing realities, beliefs about genetics, and the relevancy of conditions (e.g., physical symptoms) that influence risk perception which are of utmost importance (Cox & McKellin, 1999; Parsons & Atkinson, 1992; Shiloh et al., 2006; Richards, 1992; Ozanne et al., 2010).

Condition 2: Discrepancies in ARVC management. Many participants, in the period following the receipt of their genetic test result, were concerned that there

appeared to be discrepancies, not only in the accuracy of the test result, but also in their medical management and follow-up. Participants had difficulty understanding why some ARVC negative relatives had follow-up appointments with the cardiologist, whereas others did not: “Why do I have to go back and see [cardiologist] again if I am okay [negative]?” It seemed that being negative had different connotations for different people. For some, being negative meant they did not require any follow-up; for others, haplotype analysis was viewed as not being absolute and being negative still meant there was a five percent chance that they might still have the gene, requiring follow-up care. So, when the cardiologist told them, “You do not have to come back anymore,” it was very confusing. They could not understand how the cardiologist could make such a statement when they only had haplotype testing, which was not considered to be “fool-proof.” They thought statements such as this should be reserved for those who had had genetic testing with a more definitive test. Incidences such as these reinforced how difficult it was to embody a new meaning of risk when communication was not clear. This sense of ambivalence is noted in the following narratives of those who were considered ARVC negative:

Until Jesus Christ himself comes down and says, “No you have not got it [ARVC]” I am not totally convinced that it is not going to happen. I mean there is nothing 100% proof.

I am more at ease than I was, but I am not 100% sure if there is not anything wrong.

I wanted them to say 100%, because even with that little bit of percentage, you are still wondering if you have it.

I try to relax with the negative a low risk verdict, but I do have my anxious moments with it.

Category 2: Recognizing the Reality of Living in a Family at Risk for a Genetic Disease

This category describes how individuals living in these at-risk families continue to live with the practical challenges of everyday life post-genetic testing, including restrictions on their life. It explores the variations in participant responses as they come to recognize and accept living in a family at risk for ARVC. A large part of dealing with what one participant described as the “constant cloud of being at-risk” is developing effective coping skills and having a good support network. The following three properties: (a) *facing everyday worries and challenges*, (b) *coping with barriers to resources*, and (c) *drawing on formal and informal supports* capture this experience.

Property 1: Facing everyday worries and challenges. This property illustrates participants’ everyday worries and challenges related to living in a family at risk for ARVC. The two conditions that describe participants’ varied experiences in this property are: (1) living with the restrictions on daily activities (e.g., driving), and (2) being a spouse of an at-risk person.

Condition 1: Living with the restrictions imposed on oneself and others.

Participants spoke of numerous worries in their everyday lives as they cope with the restrictions imposed upon them by themselves or by others. Despite knowing their genetic test result, many still continued to experience psychological distress and feelings of losing control over their lives and bodies, as they adjusted to living with ARVC and the restrictions of lifestyle. Their narratives expose the fact that technology does not always promote autonomy or empowerment but that it can be restrictive; treatments such as the ICD are not always perceived as desirable treatments that one would opt for, but

are used only because experts have imposed them, presenting them as the only option (as in Lupton, 1995). The restrictions that caused the most distress were related to modifiable lifestyle factors, including physical activity, sleep, diet, driving, education, employment opportunities, and social interactions.

Modifiable lifestyle factors. While participants reported placing restrictions on many modifiable lifestyle factors (specifically, physical activity, sleep, and diet), it was decisions about whether and how to restrict physical activity that caused the most anxiety. Restrictions on physical activity were self-imposed, usually on the advice of a health care provider. Over time, however, the decision was made to reintroduce physical activity in moderation.

For those who had restricted physical activity in the pre-test “awakening period” (described in construct one under the property, *the struggle to understand the meaning of being at-risk for oneself and others*), restricting physical activity to avoid firing of the ICD made sense.³² For one thing, most participants had either personally experienced, or heard stories of a relative who had experienced, a cardiac event that caused the ICD to fire which was precipitated by physical activity. Second, health care providers had advised participants of the strong correlation between physical activity and the triggering of a cardiac event. In keeping with the available scientific knowledge (upon the advice of health care providers), and experiential knowledge (witnessing or experiencing a cardiac episode while exercising), many elected to forego physical activity. This decision is

³² Restricting physical activity out of fear of causing the ICD to discharge has been noted in the literature to result in feelings of a poor quality of life (van Ittersum et al., 2003).

made by the at-risk person and reinforced by others (such as a health care provider, a parent, or a teacher). The following narratives captured this experience.

The cardiologist told [Son] he is not allowed to do the fitness test that they are doing in school anymore. The gym teacher will not let [Son] do the test either.

I could not go to the gym or do stuff outside in the summer time. I couldn't do any of it anymore, in my mind.

My other daughter who had an ICD is a jock, or was a jock. She was training for a marathon and was a scuba diver. All of her sports activity and her lifestyle have changed because of this condition. She was exercising and got her pulse rate up to 208. The defibrillator went off, knocking her back on her butt. Next thing, she was in the ambulance and at the hospital. She was fine but she scared everyone else half to death. She has given up the marathon idea. When she is scuba diving she cannot go below 50 feet, and she will not exercise without a pulse meter on.

For most, restrictions on physical activity resolved over time. As participants became more accustomed to living with an ICD, became increasingly familiar with its role in disease management, and gained trust in its capability to respond to a cardiac event, they cautiously began to introduce light to moderate physical activity back into their routines.

For many of the ARVC positive adult participants, the re-introduction of physical activity represented what Morken et al. (2009) referred to as "regaining control" over their lives. In this case, reintroducing physical activity is part of adapting to living with the ICD, having faith in its capabilities, and incorporating the uncertainty as to when the ICD will fire next into the new norm.

I can still be active, go out, socialize; dancing and pretty well do everything that a normal person can do. I go to the gym once a week. I walk about two to three miles a day.

It [ICD] never stopped me from doing whatever I wanted to do. If I wanted to go swimming, if I wanted to go skydiving, I was going to go.

Two individuals who had had a negative genetic test described an immediate overwhelming sense of relief and, not surprisingly, resumed physical activities more quickly than those who were ARVC positive. Having a negative result reinstated a sense of confidence in one's body as this participant explained, saying, "I am not afraid now. I don't hesitate to do something for fear of triggering something and when my heart rate goes up I say that's good. I'm burning calories." The second of these two participants who had tested negative, however, was not as confident and did take a bit longer to adjust to the fact that he did not have ARVC and that engaging in physical activity would not evoke a cardiac event:

I played basketball a couple of weeks ago. I played volleyball against the high school students twice last week. This was the first time in a long time. I am not back there, but I am more confident that I do not have this disease.

This study found that regardless of participants' carrier status they modified their behaviours in keeping with their existing scientific and experiential knowledge at any particular time. That is, on the issue of physical exercise, participants deferred to clinical knowledge; their experiential knowledge was completely shaped by science and was not reshaped by other conditions. For example, when physical activity was considered a "risky" behaviour based on their beliefs or upon recommendation of their health care provider, they restricted it. Upon receipt of their genetic test result and prescribed treatment (medications or ICD), and being told by health care providers that they could engage in some level of physical activity, they resumed it.

Driving. Participants reported that living with restrictions imposed by others, such as the removal of one's driver's license, made them feel as if they were being punished for something that is out of their control: "It's a classic example of negative

reinforcement... every time you get shocked, and then you get punished when they take your driver's license away." For some, not having a driver's license made them feel socially isolated, as they were no longer able to be independent in getting to work, or to do the practical activities of running a household, such as picking up groceries.

Participants understood the rationale behind having their license revoked; what was annoying was the lack of consistent protocols surrounding the criteria for removal, as this participant explained:

What I don't understand is they will take it from you for six months and you could be fine for that entire time but as soon as they give it back to you, two weeks later you could have an attack. Now, I am glad [Husband] gets his license back but it is still a danger.

Knowing that having this disease puts others at-risk, participants imposed driving restrictions on themselves. It was common practice to drive only when absolutely necessary, or to drive slowly and with extreme caution. The following commentary summarizes the stress linked to having one's drivers license removed, getting it back, and driving:

I'd lost my license for eleven months because of those two shocks [firing of ICD] in a row—I had my license back for nine days before I even drove. I wasn't afraid to drive, but I think you lose your confidence in your own body. You don't know what's going to happen. Even now, since I had another shock in October—and I'm still driving because motor registration hasn't asked for them yet—I was only driving back and forth to school and keeping on the inside lane, going slow and always cautious if I see a pedestrian, and making sure where can I pull off—you know, just crazy stuff.

These findings support other research indicating that individuals with an ICD who have had restrictions placed on their driver's license experience feelings of resentment, anger, loss of independence, worry that sudden incapacitation while driving may pose risk to

themselves or others, and in some cases, frustration with not being able to get daily taken-for-granted chores completed, such as grocery shopping (Shea, 2004).

Social restrictions. Participants' social lives had also been restricted to varying degrees. For some, social isolation has become a key issue of concern. One spouse spoke about the loss of her husband's friends once they became aware of his risk for ARVC and that he had a defibrillator which actively fires: "A few of his friends don't like being around him anymore." In contrast, for others this social isolation was self-imposed out of fear of the ICD firing: "I was afraid to go out in public. I didn't want to go on the bus; I didn't want to go to school; I didn't want to go to work because I was afraid it was going to happen." Similar feelings of social isolation are reported in the ICD literature (Dickerson, 2002). The impact of this self-imposed isolation was described by one participant as follows:

There is no spontaneity in my life anymore. I am always worried and concerned.... I find it embarrassing for people to know that I have had an incident [ICD firing]. I have gone into avoidance.... I spent most of July this year sleeping, I would sleep nine, and ten hours a night, and then I would spend the afternoons in bed. So I did not do anything. I did not do any gardening or mow the lawn. I took my dog for about six walks around the block. I walked once on the beach, that was it. The dog has not had a walk since October. All last winter I was afraid to take her out. We got a bit of land around our house, and I wouldn't even go out in the yard and play with her.

For the above participant, this self-imposed isolation was in a response to a "loss of confidence in his body" and the fear of "being embarrassed" that the ICD would fire causing a "scandal." Comparable accounts of anxiety, depression and avoidance have been noted in the ICD literature (Bilge et al., 2006; Bolse et al., 2005; Bostwick & Sola, 2011; Friedmann et al., 2006; Hallas et al., 2009; Lemon, Edelman, & Kirkness, 2004; Newall, Lever, Prasad, Hornabrook, & Larsen, 2006; Wheeler et al., 2009)

Employment and education opportunities. Participants described accounts of psychosocial distress related to the restrictions that having an ICD imposes on employment and educational opportunities, as reported by Probst et al., (2011) and Shea (2004). Acquiring gainful employment and having the education to secure employment was critical to those for whom it was important to be able to provide financially for the family. This concern was obvious in the case of an ARVC positive fisherman who fished alone and had to hire another laborer to handle the boat in case the ICD fired and he was rendered unconscious. Adding to the stress was that this position was difficult to fill as potential candidates were reluctant to take the job knowing that they would be expected to respond to a cardiac event: “I got a guy fishing with me and he is frightened to death all the time. That is one change I guess. I had always worked alone. I had to get a guy to go fishing with me.” Individuals living with chronic heart disease have also voiced similar life challenges with employment related to social interactions (Martensson, Karlsson, & Frilund, 1998; Nordgren, Asp, & Fagerberg, 2007).

A lack of opportunities to enter into a field of interest was noted by one participant who felt he would not pass a required medical examination: “I’d like to be a personal trainer but you have got to be able to teach other people to be fit and how can you when you are not healthy yourself?” This restriction was self-imposed and supports the findings of Rose (2007) who noted that it is the meanings that one assigns to being at risk for a genetic condition that influences one’s beliefs about how they should conduct one’s life and, as in this case, place restrictions upon oneself.

Condition 2: Being a spouse. Living in a family at-risk was also challenging for the spouses. The spouses frequently talked about being worried over the health of their

partners, as described in the cardiovascular literature (e.g., Martensson et al., 1998). They reported finding it difficult watching a family member endure the constant firing of the ICD unaware of the outcome (see also Hazelton, Sears, Kirian, Matchett & Shea, 2009). Some expressed fear that the ICD might not save their spouse every time; however, at the same time, knowing that one's spouse had the ICD (in keeping with perceptions of the ICD as "life-saving"), gave them some relief as this participant stated: "Since she got an ICD, as a husband I feel more relief." Participants noted that it was common practice to constantly observe their spouse for any signs of the disease. This constant observation on behalf of the spouses was a common topic of discussion in the three focus groups:

"[Participant A to Participant B]: Do you ever wake up in the middle of the night and [Husband] is there making sure you are still alive? Because that is what my husband does."

The concern for spouses did not include significant changes in roles, alterations in social life or changes in social relationships, as described in some cardiovascular literature (Dalteg et al., 2011; Hazelton et al., 2009; Hendriks, et al., 2008; Pihl et al., 2010). This may be because although ARVC is a life threatening condition and can impose restrictions, for the most part, participants were able to maintain employment and do activities involved with daily living. Although the spouses did acknowledge that there were times that they had to assume added responsibilities, such as driving, this was not overly concerning. Another factor that may account for the lack of emphasis on role changes is that the mean age of the ARVC-positive participants was only 41; thus, one might expect that this study did not capture any changes in familial roles and responsibilities with such young participants. This may be documented more fully in a

follow-up study with older participants. What was frustrating for spouses, however, was gaining access to resources (e.g., health care providers knowledgeable in ARVC, treatments, and technology). This is discussed in the next property, *coping with barriers to resources*.

Property 2: Coping with barriers to resources. This property describes the conditions that influence participants' experiences as they tried to access the information and resources they felt were necessary to manage and live with being at risk for ARVC. These conditions included barriers to (1) human resources, (2) physical resources, (3) financial resources, and (4) informal and formal supports.

Condition 1: Human resources. Formal supports are imperative throughout all phases of the genetic testing process. The support of the cardiologist, genetic counsellor, and nurse was noted to be instrumental in facilitating positive health outcomes. Participants did not focus on the role of health care providers in understanding their objective risk; however, many described them as playing a significant role in helping them cope with the outcome of the test. The role of genetic counsellors in helping individuals cope with their new risk-status is referred to in several systematic reviews (Braithwaite et al., 2006; Butow et al., 2003; Kaphingst & McBride, 2010; Meiser & Halliday, 2002; Sivell et al., 2008).

Participants emphasized that an important component of this formal support relationship is trust. For many, the excellent clinical and communication skills of a health care provider led to a sense of trust in their abilities. Although participants recognized that the cardiologist and the genetic counsellor did not have all the answers as to what the future holds for them and their families, they were satisfied, for the most part, with their

ability to communicate risk information appropriately. There was an overwhelming sense of respect and appreciation for the attending cardiologist who helped them understand and cope with the realities of living in an at-risk family, namely, providing them with a sense of being “safe” despite the potential outcomes of a fatal disease. Equally important were responses to the immeasurable support of the genetic counsellor: “If it wasn’t for [the genetic counsellor] I would be foolish.” Similar accounts of formal supports have been found in the literature (Bolse et al., 2005).

Although participants appreciate the ongoing expertise of the cardiologist and the genetic counsellor simultaneously, they also recognize the need for more experts in the field. The general consensus for those living in rural communities, as they reflect on the positives and negatives of the genetic testing process, is that they need someone who is familiar with family history and ARVC management to provide continuity of care. In fact, participants requested that health care providers familiar with ARVC visit the communities on a regular basis.

The most frequently cited barrier to care identified by participants was human resources and the lack of knowledge by health care providers for the diagnosis and treatment of ARVC. This finding is similar to that of McAllister, Payne et al. (2007), who reported that a lack of diagnosis and inappropriate care was a key factor that caused significant distress to at-risk families. This lack was particularly noted in rural and remote areas of the province. The lack of knowledge was both surprising and frustrating for participants. Not surprisingly, participants assumed health care providers would be knowledgeable about a disease prevalent in the population they service. This expectation is evident in these two comments: “I think that all physicians working in every area

affected by it [ARVC] should know about it... Newfoundland is such a melting pot of genetic conditions they need to be educated about these people,” and “They [health care providers] did not know the family history. They did not know what ARVC is all about. They can’t help you.” Accounts of health care providers in rural settings not knowing how to manage ARVC added to the lack of confidence in the abilities of health care providers:

[Son] went in, took his ICD card out at outpatients, and said, “I have a defibrillator implanted, and it went off.” The lady behind the desk said, “Does anybody know anything about one of these thingamajigs? He has one in!”

I carried mother in to the local hospital and got her checked out. I had to tell the doctor what was on the screen. The electronic device on the screen tells you how many irregular heartbeats you are having. The doctor said to mom, “You did not have any attacks.” I had to correct him on it and when he looked he said, “You are right.” This was the first trip, so that did not go over well with me.

It is in situations such as those described above that caused concerned families to forego local treatment and travel to the city.

I just get in the car and take my boys to [the city]. The bottom line is they [health care providers of smaller centers] are not educated in it, or they are just learning about it... I want the best there’s around.

Adding to the frustration over lack of access to care in rural areas is the high turnover rate of physicians and nurses, poor medical coverage for emergencies, and long wait times in local emergency departments in rural communities. In fact, many participants felt that they were often left to coordinate their own care:

The local doctor can be up on this [ARVC] but he is not in the clinic 24/7. I could take an attack at one o’clock in the morning and he could be away somewhere for the day. This could be his day off. Now what do I do?

If I have chest pains or anything, I know that I got to go right away. So I don’t want to go up there [hospital] and have to wait in the waiting room for 45 minutes to an hour before someone sees me.

The people who have worked in the cardiac department for a long time, when I go there they know me. They know all about the history, but a lot of those are retired and they are gone. So if I went down and had the monitor put on they would know what was going on and be able to say to the doctor, "This is [Participant] and they have this wrong with them."

Another barrier related to human resources identified by some participants was the lack of access to genetic counsellors and cardiologists. Although participants did appreciate those genetic counselling resources that were available, they also recognized that there is only so much that can be done given the resource limitations.

My biggest problem is, if I need anything I've got to phone [genetic counsellor] because there is no one else to phone. If [spouse's] defibrillator goes off I can call [the hospital] until I am blue in the face, and you won't reach anyone.

The genetic counsellor is not only dealing with me but a lot of other families as well.

Similar frustrations were noted concerning the limited accessibility to the cardiologist with the most expertise in ARVC:

They have a clinic once a month [at local hospital] but that's perfect for someone who is not having any episodes. Once you have an episode, automatically, the cardiologists want you in [city] to see you.

There's one thing that I mean to ask every time I go in, but usually when you see [cardiologist], you get in, get out. He's so busy and you don't get time to sit and talk.

Notwithstanding, the consensus was that participants were pleased with the cardiologist's bedside manner, support, and knowledge about and explanations of treatment regimes, as summarized in these three narratives:

[Cardiologist] keeps saying when the girls go in to get rechecked, 'You are fine, you are probably going to live to be 98 and die of something else.' [Cardiologist] gives you the feeling that, okay, you got this thing [ICD] in, and you can feel good and safe.

[Cardiologist] gives you a good feeling. It is kind of uplifting to go in and have your appointment, believe it or not, with the cardiologist.

The thing about [cardiologist] is that no matter how knowledgeable he is, he can always come to your level and talk to you right.

The participants' comments about the genetic counsellor shared similar sentiments as those above:

I had a lot of conversations with [genetics counsellor], he is always available if you need anything . . . I felt support was available for me at any time . . . when I was first tested I felt that the resources were there for me. But how much can one person do? How many times do you want to call somebody?

When you do meet somebody like [genetics counsellor], I felt I could tell him anything.

When I contacted [genetics counsellor] . . . the response was very quick through e-mail, and I talked to him on the phone. [Genetics counsellor] called me at work because I had given my information. I think that quick response sort of tells you [genetic counsellor] is trying to help.

They were fabulous out there at that clinic. The [genetics counsellor] and [cardiologist] and the crew [nurses] I can't thank them enough, and I can't praise them enough.

Aware of the limitations in human resources, many participants soon recognized that they had to be their own advocate in managing their disease. For example, individuals have advocated for appropriate blood work to monitor the therapeutic range of certain medications, for ARVC protocols to be placed on charts, and for better treatment.

I am supposed to be tested every three to six months for thyroid and liver and everything else when I am on this medication. When I went in and saw [cardiologist] we brought it up and told him I never got tested and yet I have been on this medication for the last two or three years. Also, I was taking this medication for two years before they even put the sticky on it that said I should be out of the sun.

We need an ARVC protocol posted up the local emergency room.

The first time [spouse] went up to the clinic with an attack, the doctor said, 'there is no good phoning into [city] now because we won't reach anyone in there this evening.' I said, 'Look there is a doctor in there on call all the time. This is what you do: you phone the main switchboard and tell them to page whatever cardiologist is on call.' This is what you deal with all the time.

Condition 2: Physical resources. In addition to human resource barriers, many narratives alluded to the fact that even if there were adequate human resources to meet the population's needs, there remained a dearth of physical resources. Smaller local facilities do not have the technology to download (nor health care personnel to interpret) information from the ICD. Therefore, individuals with ARVC who go to a local facility either find themselves being told to go home and to come back if the ICD fires again or are sent by ambulance to the city. Upon arrival in the city, unless there has been some technological issue with the ICD itself (such as loose wiring or reprogramming), individuals are typically sent home with few or no treatment changes. For many, these trips to local medical facilities become futile efforts, and individuals learn to draw on their own experiential knowledge about the ICD to decipher why it fired and to make decisions regarding their actual risk. This situation also solidifies the notion of the lay expert (Lupton, 1995), as participants begin to make more decisions about their care.

Condition 3: Financial resources. The financial implications of living in a family at risk for ARVC were also seen by many participants as a barrier to care and, subsequently, a barrier to the ability to understand and (re)assign meaning to the risks associated with living with ARVC. Regarding the findings of other research that examined quality of life issues of individuals living with an ICD (Shea, 2004), of particular concern was the cost of medications, the amount of work time lost due to

appointments, and travel expenses related to geographical restrictions on available resources.

For one participant, the cost of medications was unsustainable, and he found it necessary to accept social assistance in order to manage: "I am on social assistance because I cannot pay for my pills if I work. They are very costly, and they are always putting you on different ones." Another ARVC-positive participant spoke of the financial strains of treatment and follow-up appointments: "It costs money, gas, hotels, meals, and then there are work commitments and time off." Given that some participants required additional follow-up visits to the regular six-month clinic visits, or ambulance services that were a chargeable service, care could be particularly expensive. One participant described having made 18 trips to the city in one year. Added to these financial strains was the stress linked to poor driving conditions due to the weather; although, as many participants noted, this was often a moot point: "we just don't have what we need in this area." Hence, for many, treatment came with a huge financial commitment.

Being diagnosed with ARVC means that life and health care insurance may be unavailable or unaffordable. Several participants spoke of the challenges they faced when applying for insurance for their homes, cars, and disability or life insurance. Most were fortunate that they had insurance plans prior to being diagnosed, but for some this coverage was limited, as in the case of one participant: "I have life insurance. Luckily I had it before I got diagnosed. Other than that, I got nothing. They will [offer] life and disability insurance, and [when] I tell them what I have, forget it." For this participant, his inability to receive full insurance coverage caused him to worry, "If I get sick tomorrow, there is no one going to take care of me; what the hell am I suppose to do? I

got a mortgage, car payments, and kids going to school.” Limited access to insurance was also raised as a barrier to affordable care, a challenge that has also been discussed in relation to cardiovascular disease (Probst et al., 2011; Shea, 2004), and BRCA 1/2 (Lynch et al., 2006; Watson et al., 2004). Fear of health or life insurance discrimination was cited as a key concern for those at risk for BRCA 1/2 (Allan, Friedman, & Senter, 2012; Peterson, Milliron, Lewis, Goold, & Merajver, 2002) and HD (Bombard et al., 2012; Erwin et al., 2010).

Overcoming Barriers. Based on the previous discussion it is evident that individuals living in families at risk for a genetic condition do experience psychosocial distress throughout the genetic testing process and continue to experience it for many years after they receive their test results. This finding does not support the large body of literature that states that psychological distress subsides within a year (Almqvist et al., 2004; Aktan-Collan et al., 2001; Arver et al., 2004; Codori et al., 2004; Collins et al., 2007; Gritz et al., 2005; Heshka et al., 2008; Meiser & Dunn, 2000; Meiser et al., 2002, 2004; Shaw et al., 1999; Watson et al., 2004). It also suggests that concerns about one’s health do extend beyond five years post-testing, as noted in some other studies (Codori & Brandt, 1994; Decruyenaere et al., 2003; Gargiulo et al., 2009; Schwartz et al., 2002; Timman et al., 2004; Meiser et al., 2002; van Oostrom et al., 2003).

For the participants in this study, the level of psychological distress post-genetic testing is not constrained by time nor by carrier status as suggested in some studies (Almqvist et al., 2004; Aktan-Collan et al., 2001; Arver et al., 2004; Codori et al., 2004; Codori & Brandt, 1994; Collins et al., 2005, 2007; Decruyenaere et al., 2003; Gargiulo et al., 2009; Gritz et al., 2005; Heshka et al., 2008; Meiser & Dunn, 2000; Meiser et al.,

2002; Shaw et al., 1999; Schwartz et al., 2002; Timman et al., 2004; van Oostrom et al., 2003; van Roosmalen, 2004; Watson et al., 2004); rather, it is something that is ever-present and tends to resurface in response to risk perceptions. At times it is the juxtaposing of the conditions and contextual dimensions influencing risk that causes the distress; however, in other instances it is the strong presence of one of these factors (e.g., the onset of signs and symptoms, another loss, another ARVC positive relation, the presence of a more accurate genetic test), that causes a heightened sense of risk and feelings of distress. Similar accounts of how mitigating factors influence how one perceives and copes with their risk have been reported in other studies (McAllister, 2002; McAllister, Davies et al., 2007; van Oostrom et al., 2003). This finding explains why some researchers have found that negative carriers do not have any psychological distress pre- and post-testing (Croyle et al., 1997; Lerman & Croyle, 1996; Lodder et al., 2001; Reichelt et al., 2004; Schwartz et al., 2002).

Participants offered several suggestions that would help them overcome or deal with some of the identified barriers that create this ongoing distress. These suggestions included the following: the implementation of standards of practice, such as clinical practice guidelines that address ARVC management; an interactive ARVC website where they could ask questions; medical alert bracelets; a community support group; more devices that can download ICD activity in rural community hospitals and in the home; education for health care providers about ARVC; a stronger visible genetics program at the community level; and, more access to genetic counsellors, cardiologists, and researchers in times of need or to obtain progress reports on new discoveries. Many of these suggestions appear in the following narratives:

A webpage would help that has little bits of information...where you could input a question and somebody would answer you and put your mind at ease. It's probably just little things that pop up that you could ask. Things you might think it's important. . . More contact [with researchers] probably would be nice with little letters here and there, or progress to date.

Going to a support group you would meet people who are in the same situation you are.

I don't even care if it's an ID [medical alert] bracelet or whatever, but when we go to hospital that you get seen and [health care providers] know what we have.

Research has found that support groups are beneficial in helping at-risk individuals cope with their disease and prescribed treatments (Bolse et al., 2005; Dickerson, 2002). The experiential knowledge shared helped individuals gain insight into the practical every-day management of their disease that could not be acquired at the local doctor's office.

Finally, several narratives suggested that the need for health care resources post-genetic testing is critical, as many still have psychological challenges that persist long after testing and treatment.

Yes, I think it [support] is very important because it wasn't my health that was at issue. It was my mental health.

It did not even come to me to go see a counsellor [for psychological help]. But I needed to see somebody. I needed two things: I needed someone to help me wrap my head around this avoidance that I was getting into, and the anxiety and depression. I also needed someone to assure me that it is okay for me to go out and walk, like I use to walk; to pick up a bag of groceries and walk up over the stairs. To do the normal everyday things that I have avoided doing that I don't have to be afraid of going out.

Condition 4: Informal supports. Family and friends were identified as having a significant role in balancing feelings of risk with the normalcy of everyday life. For many, knowing that other family members were readily accessible if needed was helpful, as this participant described: "It was six in the morning, so I phoned my sister and told

her she'd better get over here because my defibrillator just fired... So she got ready and came over." Similar accounts were provided by spouses: "[Husband] tried to get me through these panic attacks and the misery [of dealing with having a negative test result after ten years of not knowing genetic status]." For young adolescents, it tends to be parents, siblings, or cousins who have had similar experiences that are called upon for support and to gain a better understanding of prescribed treatments such as the ICD: "Before [Son] went in and had his defibrillator he use to talk to [male cousin] about it."

The knowledge and advice of mature adult family members were reported to be invaluable, as these individuals took on the role of the lay experts: "We got the knowledge. We lived through it. When we were diagnosed there were no defibrillators and half the medications they got now." The knowledge acquired by those who lived through the genetic testing process seemed to provide reassurance to others that they, too, could cope with this disease, that there was access to someone knowledgeable about ARVC, and that they were not alone. The importance of relying on informal supports from family members contributes to family cohesion and its importance in the management of ARVC.

Category 3: Looking Towards the Future Realities of Living in a Family at risk for a Genetic Disease

This category describes participants' experiences as they start to think about the future implications of living in a family at risk for ARVC and how they tried to focus on the positive aspects of genetic testing and their lives in general despite their concerns for the children living in these at-risk families. The two properties: (a) *trying to maintain a positive outlook* and (b) *living with concern for at-risk children*, describe participants'

experiences as they embody risk and look towards the future realities of living in a family at risk for ARVC.

Property 1: Trying to maintain a positive outlook. This property explains participants' efforts to maintain a positive outlook on life post-genetic testing. The two conditions that capture this experience are: (1) the positives of predictive genetic testing and (2) being able to deal with adverse events.

Condition 1: Predictive genetic testing: Positive aspects. Post-predictive genetic testing participants tried to maintain a positive outlook and focus on the good qualities of their lives. In doing so, participants highlighted the positive aspects of the predictive genetic testing process. Many recognized that they need to focus on preparing for their own future and the consequences of living with the disease. A large part of maintaining a positive attitude meant acknowledging the positives of knowing one's genetic status, of having the ICD, informing others about the benefits of genetic testing, and being able to identify the good qualities in one's life.

In order to sustain a positive outlook for the future participants focused considerable attention on the benefits of knowing their genetic status. In keeping with previous research on other inherited cardiovascular conditions (Aatre & Day, 2011; Andersen et al., 2008; Christiaans et al., 2009; Marteau et al., 2004) and BRCA 1/2 (Butow et al., 2003), knowing one's genetic status was viewed as something advantageous. This information was considered valuable because it could be used to make life decisions and alleviate the psychological distress of not knowing one's risk status, as summarized in this comment:

Now they [researchers] know what causes ARVC, I think it eases people's minds one way or the other. Because if you don't have it, they'll tell you that you don't have it; whereas, before we weren't really sure we had it or not.

Knowing one's risk status was also helpful in that it helped participants prepare for future management of ARVC, which instilled in them a sense of control over their lives: "It [genetic testing process] is positive because the more we know about ARVC, the more prepared we are." Successful disease management, as noted in the literature, requires individuals to be able to anticipate symptoms, prepare for future consequences of the disease, and engage in disease prevention and management strategies (Anderson et al., 2008; Decruyenaere et al., 2003; Dinc & Terzioglu, 2005; Esplen et al., 2001; Gritz et al., 2005; Heshka et al., 2008; Hodge, 2004; Shaw et al., 1999). Thus, many participants availed of the opportunity to know their risk status, to prevent the onset of ARVC and receive proper treatment (such as the ICD), prior to having physical signs of the disease. This was the case for the three younger participants who did not have any signs of the disease but had an ICD, "[Children] did not have any symptoms of the disease. They only knew because of the DNA testing."

Cases such as the one above emphasize the fact that most participants believed that the opportunity to prevent a sudden cardiac death outweighed any negative aspects of genetic testing, and made participants feel as if they were being taken care of: "I think it [genetic testing] was very positive. While it was being done, I sort of felt safe." Given the opportunity to relive their choice to be genetically tested, most participants agreed that they would do it again as this participant noted, "It [genetic test result] gave me piece of

mind which I haven't had....since 1983.”³³ In keeping with some other studies (Anderson et al., 2008; Hendriks et al., 2005; Smets et al., 2008), those who have made the decision to get their children tested did not regret it. Overall, participants were so confident in the benefits of having genetic testing they constantly reiterated to other family members the importance of having genetic testing done in order to plan for their future health:

I made them [children] aware of the history. I made them aware that they need to take responsibility. I think they're going to take responsibility when it comes to their children. They probably don't talk about it a lot, but I think they know deep down within them.

I was trying to tell my son, “go in to see the doctor and get tested.”

I am going to push them to do it [have grandchildren tested].... We know it is in our genes so we need to get it checked.

Being able to focus on the good quality of one's life. In order to maintain a positive mind-set, individuals aligned their perception of health with having a “good quality of life.” The meaning of “good quality of life” expressed in participant narratives emphasized daily life activities, accepting that one cannot control all aspects of the disease (and, in some cases, comparing one's situation to others less fortunate with their health), and accepting that living in a family with a genetic condition means not having 100% control over one's fate, as described by one participant:

I am not living life for ARVC. I am living life the way that I want to... If the ICD is going to go off, it's going to go off anyway. I am not going to sit at home

³³ For the most part all the participants in this study wanted genetic testing. I did not interview anyone who declined genetic testing. It is important that future research include this group to determine if there are alternative opinions that contradict this study's findings and findings of other research that show eager uptake of genetic testing. This point is discussed in the conclusion.

and grieve myself to death because I got this problem. I'm going to do what I can do until the day comes that I can't.

To successfully cope with ARVC, individuals took the position that they must appreciate what life has to offer and move on with their lives. This appreciation meant that participants had to reassign the meaning of the firing of the ICD from being something "risky" to something "normal" that saves their lives. As noted by Dickerson (2002) and Bolse et al. (2005) living with the ICD means getting on with one's life, accepting the restrictions imposed by the ICD, and appreciating life as it is lived.³⁴

Similar sentiments are reflected in these commentaries:

You have to live with it [having ARVC and ICD]. You have to live every day to the fullest. There is no good to grumble about it, or complain about it... You know the seven times that you passed out [from ICD firing] you got another ten or fifteen times yet.

You can go in now and you can get this test done; it won't stop the progression of the disease yet, but it'll give you a second chance if something were to happen, once you have the ICD in...so this kind of research got to continue.

For many, the meaning of what constitutes a "good" level of health was determined by comparing one's existing health with the potential negative health problems associated with ARVC. As the use of the ICD becomes normalized during the illness process, participants come to accept it as critical to maintaining this "good" health, a finding that is supported in the literature on the ICD (Flemme et al., 2005).

Participants also made downward comparisons with other individuals and families, similar to those described by Farrimond et al. (2010) in order to cope with living in an at-risk family and to confirm that they had a "good" level of health. A downward

³⁴ Being optimistic as to the ICD capabilities has been identified in the literature as an effective coping strategy to deal with the stress of having the device implanted (Flemme, Johansson, & Stromberg, 2011).

comparison is a social comparison where individuals compare themselves to others less fortunate in order to judge their own circumstances. In doing so, individuals are often able to see the positive aspects of their situation. In this study, participants drew upon stories of other families who experienced numerous losses to put the “good qualities” of their own lives into perspective. “[Family X] is a real horror story. They have lost seven generations of males from ages 17 upwards” and “[Cousin] lost three brothers. It is horrible!” Likewise, for one ARVC-positive participant, downward comparisons were used to confirm that he was coping well: “I just charged ahead and did what I had to do; [whereas sibling] quit work, lay around, and got depressed.” For several participants, discussions with non-family members gave them the opportunity to make comparisons amongst individual situations and to gain insight into how others had handled the news of being positive. For one participant, just knowing that other families were dealing with similar things, such as concern for the wellbeing of the children, gave him a sense of relief in knowing that what he was feeling was “normal,” and reconfirmed that he was indeed living a good quality life in comparison to others.

Condition 2: Being able to handle an adverse event. Efforts to be positive about one’s life and surrounding circumstances were also described by the younger participants (ages 15, 16, and 18). Although they understood the potential negative outcomes of a cardiac episode, they described a sense of confidence in their abilities to handle such a situation. Embedded within the narratives is a message to parents to stop worrying about them because they “have things under control.” For these individuals, this confidence is empowering, as noted in this narrative that described an incident where one participant’s defibrillator fired and the other two had to respond:

Our parents are worried about us going out alone. I remember one time when we were all in the pit hanging around and [Cousin] was there and his defibrillator went off and all of us were around. We knew what to do. We took care of him and called his parents, and waited for them to come. We knew he was going to wake up.

None of these young participants described any psychological distress with the genetic testing process other than the initial anticipation, anxiety, and fears related to the ICD firing and follow-up care. As described previously, these concerns gradually resolved as participants became more familiar with the ICD firing and more comfortable with management of the ICD. Similar to some earlier studies of adolescents with an ICD and their parents (Zeigler & Nelms, 2009), the younger participants in this study emphasized how they tried to live a normal regular life. A significant part of this “normality” is the firing of the ICD, as one participant summarized, “I was afraid at first [of the ICD firing], but now I let it slide by.”

Property 2: Living with concern for at-risk children. Threaded throughout the entire genetic testing process is concern for the health of the children. This property builds on the parents’ concern for their children specifically, their future wellbeing. Explicit within participants’ narratives, once they knew their own genetic status, was a shift in concern from oneself to one’s children, which was also reported by McAllister, Davies et al. (2007). This property describes the two conditions that influence participants’ concerns for the future wellbeing of the children. These two conditions are: (1) physical signs and symptoms of ARVC and (2) children’s knowledge of ARVC.

Condition 1: Physical signs and symptoms. Despite the appearance of coping well with their child’s potential risk, any subtle change in a child’s physical status caused parents to interpret the event as being “risky,” indicative of ARVC, requiring immediate

attention. The assignment of something as being “risky” forced the parents to momentarily reconstruct their ideas about risk and to respond to the event in a pragmatic manner; removing the child from school, seeking medical attention, monitoring the children, and try to maintain a cohesive family. Coinciding with parents reshaping of the meaning of risk were familiar feelings of anxiety and fear about their child’s future, as noted in this parent’s comment:

My son had some kind of flu-induced asthma. So I phoned the hospital and got the pediatrician on the phone and said, ‘Now this is one of my children, and I want his checked out right away’. He was okay . . . but I have warned the school if he complains of chest pain, call me. You are always afraid they will have chest pain.

Concern for the wellbeing of the children often led parents to remove them from school in order to see the family doctor or to simply to watch over them until they no longer were considered ill, as several parents noted, they were “not taking any chances.” One mother’s account of removing her child from school due to health concerns and receiving a note from the teacher inquiring about why he has missed so much time is captured this experience:

I received a letter from the school complaining because he has been so ill with chest congestion, and I am so afraid to take a chance that it is just chest congestion and not something else. He said he feels his heart pounding and he feels tightness in his chest. I’ll take him to the doctor instead of school. He has missed so much school that I got a letter from the school now because I have taken him out of school so often getting him checked out.

The perception that others (such as teachers), are criticizing a parenting decision to exercise caution with regard to risk of a cardiac episode adds to the distress experienced by caregivers in families with ARVC.

Concerns about the future wellbeing of the children were common amongst the parents whose children have not had genetic testing for ARVC, but exhibit physical signs

of the disease. This is evident in the narrative of one ARVC positive parent who placed restrictions on the physical activity of her 11 year old son.

My son was playing hockey up until last year. He is 11 now. He has not been tested and he passed out in the gym the other day.

The worry about the children's' future wellbeing in this phase of the genetic testing process is similar to that described in construct one: *Awakening to a New Meaning of Being At-Risk*. That is, the worry knowing that ARVC can cause a sudden cardiac death without any warning. In order to alleviate some of this anxiety many felt they needed to keep a watchful eye over the children. Research has also identified similar accounts of anxiety with having a child at-risk (Andrews et al., 2006; Dinc & Terzioglu, 2005; Duncan et al., 2007; Farnsworth et al., 2006; Hendriks et al., 2005). This anxiety has caused parents to maintain constant surveillance of the children's whereabouts and physical state (e.g., Featherstone, Bharadwaj & Clarke, 2006; Hendriks et al., 2005), regardless of age. These feelings are captured in the following narratives:

You just have to know where they [children] are. My daughter says, 'I'm 15 years old. Why can't I go [to a friend's house]?' She doesn't understand that you have to know wherever she is all times.

I know he is 29; but if he is going in the woods with his buddies overnight, he needs to phone and let me know.

If my son and his wife go out of town... I say to him, 'phone me when you get there.'

It seems like you are always looking for it [ARVC], not wanting anything wrong with them, but always looking at them. Anything that happened out of the ordinary, I would always think, could it be ARVC?

This concern for the children coupled with an awareness of the potential negative outcomes of this disease (that is, sudden death), has created a strong sense of family

cohesion. As a result family members do tend to spend a lot time on family outings and celebrating special occasions. These interactions serve two functions: (1) to provide a sense of security that they are not alone in case one of them has a cardiac event; and (2) to spend as much time together creating memories in case of an untimely death. These thoughts are threaded throughout several participants' narratives:

It is no good for me to go somewhere with him [ARVC positive husband] by myself, because I run [if husband has a cardiac event]. I just panic over it [cardiac episode]. They [brother and sister in-law] have a cabin just up the pond from us, just up the road. I will not go to our cabin unless they are at their cabin. I brought walkie-talkies so that we can be in touch. They keep their radio on and I keep mine on. So if anything happens I will call [sister-in-law] to come down.

We have always been close, the three boys, and us. My oldest son just turned 30 in August, and we still had to have all the family together to celebrate his birthday . . . Whether it's Christmas, New Years, Mother's Day, Father's Day, Easter we are together. I don't know if I should be doing it but I try to drill it into the boy, who knows where we are going to be next year.

Knowing the whereabouts of the children and creating a strong sense of family cohesion as methods to cope with the uncertainty of having a child at risk for a fatal disease has also been described in the cardiovascular literature (Fansworth et al., 2006; Hendriks et al., 2005) and in relation to HD (Sobel & Cowan, 2003).

Condition 2: Children's knowledge of ARVC. Concern for the children's' future wellbeing prompted many parents to educate them at an early age about ARVC, its symptoms and appropriate management strategies. For some parents, educating the children about ARVC was imperative to ensure that they were prepared for testing and aware of any signs of the disease. Parents felt that educating their children about ARVC was fulfilling their parental responsibility. This gave them a sense of solace in that that

they had prepared their children to deal with the disease in a proactive manner, as this parent explained:

I've tried to educate them on it. I don't know if I overstepped. I was trying to stress the importance of being aware of the disease, and keep a step ahead of it. You look after yourself your children and your grandchildren.

Educating the children about ARVC served another purpose: to relieve the parents of the guilt felt for passing the ARVC gene on to them. This narrative noted, "There is a lot of guilt attached to that [passing ARVC on], even though I didn't intentionally give it to them." Similar accounts of guilt have been found in the literature on genetics (Howell et al., 2006).

A large component of informing the children about ARVC is in anticipation of the difficult choices that they foresee their children having to make in their future, as they mature into adults, get married, and are faced with life decisions, such as whether to have children themselves. This is evident in one participant's account of educating a child about ARVC and then observing that same child make life decisions: "She [daughter] had chosen not to have kids until she found out she didn't have the gene."

Chapter Summary

Participants' experiences in the final phase of the genetic testing process, *Embodying a New Meaning of Being At-risk*, is captured in three categories. The first category, adjusting to living with or without a genetic condition, described how participants started to adjust to and assign meaning to prescribed treatments (pharmacological and ICD). The meaning assigned to treatments and one's willingness to adhere to them was based on the presence or absence of signs and symptoms of ARVC, one's genetic test results, and perceived efficacy of the treatment. A lot of effort went

into trying to understand the need for treatment, as they weighed the negatives and positives of those proposed. The challenge faced by participants as they tried to adjust to living with or without ARVC was that they were not totally convinced of the accuracy of the genetic test results. Adding to this were the discrepancies noted in the management of ARVC. As a result participants continuously juxtaposed one's experiential knowledge and scientific knowledge against each other in order to assimilate new information into existing beliefs about genetics and construct their perception of being at risk for ARVC.

Category two, *recognizing the realities of living in a family at risk for a genetic disease*, describes how participants live with the practical challenges of everyday life such as restriction related to lifestyle, driving, education, employment, and social interactions. A large part of coping with everyday life is identifying, coping, and overcoming the barriers related to human resources, physical resources, and financial resources. Informal and formal supports make coping with one's risk tolerable. Suggestions to dealing with identified barriers included providing more supports at the community level, more access to resources in rural settings, more practitioners familiar with ARVC management, financial planning, and help with coordinating care.

Category three, *looking towards the future*, highlights the realities of living in a family at risk for ARVC. It is here that participants focus on maintaining a positive outlook about the future for themselves and their children. To maintain a positive outlook in this context means that participants must accept and focus on the good aspects of their lives. That is, they must accept that they have this condition, accept that many facets of this disease are out of their control, accept that it can be fatal, and accept that it can be passed on to their children. In order to do so, attention is shifted to the benefits of

knowing one's genetic status, which contribute to what many have come to appreciate as a "good quality of life." These benefits include being able to prepare, manage, and get treatment for the disease prior to its onset. All participants hoped that their children could have a similar "good quality of life." Hence, as part of their parental responsibility, they constantly informed the children about the risks of ARVC, monitored them for the signs and symptoms of ARVC, and tried to help them prepare to make future decisions about their life, wellbeing, and genetic testing.

In this final phase of the genetic testing process, the continuous juxtaposing of the contextual dimensions and conditions and the outcomes of this juxtaposition determine responses to risk and account for the variations in participants' experiences. Figure 8.3 summarizes the conditions that influence the process of *Embodying a New Meaning of Being At-Risk*.



Figure 8.3. Rubik's Cube Model: Summary of the Conditions that Influence the Psychosocial Process of *Embodying a New Meaning of Being At-Risk*. Rubik's Cube ® used by permission of Seven Towns Limited, www.rubiks.com.

CHAPTER 9

CONCLUSION

This study examined the experiences of individuals living in a family at risk for Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) as they moved through the predictive genetic testing process. A grounded theory methodology, drawing on the tenets of symbolic interactionism and pragmatism, was used to discover the substantive theory, *Constructing the Meaning of Being At-Risk*. This theory describes how participants assign meaning to being at-risk and make subsequent health care decisions based on these meanings. Three theoretical constructs, (1) *Awakening to a New Meaning of Being At-Risk*, (2) *Deciphering the Meaning of Being At-Risk*, and (3) *Embodying a New Meaning of Being At-Risk*, capture the psychosocial process of constructing risk. In this final chapter, I provide an overview of the key findings. Threaded through this discussion are recommendations for future research, practice, education, and health policy. Limitations to the study are addressed.

Findings

Throughout each phase of the genetic testing process, participants' ideas about risk are shaped and reshaped as they juxtapose the three contextual dimensions (scientific knowledge, experiential knowledge, and phase of the genetic testing process) against each other and relevant conditions (e.g., age, gender, and so on). These contextual dimensions influence how individuals assign meanings about risk to the conditions. The reverse is also true; the meanings they assign to the conditions in turn (re)shape the ways in which the contextual dimensions influence risk assessment. Given that the conditions are not

the same for everyone, and that participants have varying lived experiences, the meanings assigned to being at-risk will not be the same for everyone.

This study demonstrates that one's sense of embodied risk is pragmatic, transient, and fluid. It is pragmatic in that participants draw on contextual factors in their everyday lives in order to understand risk. It is transient in that the meaning one assigns to risk fluctuates in relation to the contextual dimensions in one's life and the relevant conditions surrounding it. It is fluid in that risk perception is not static but something that continues to evolve and get reshaped with each new interaction or experience, with new scientific advancements, with each phase of the genetic testing process, and as one experiences each condition.

At a basic level, this study supports the work of other scholars who have argued that experiential knowledge, rather than an objective understanding of risk or biomedical models of disease causation, is the most significant factor in assigning meaning to being at-risk (Cameron et al., 2009; Cox, 2003; d'Agincourt-Canning, 2005; Etchegary, 2010; Lock & Ngyen, 2010; Lupton, 1999; McAllister, 2002, 2003; Norris et al., 2008; Shiloh & Saxe, 1989; Smith et al., 2002; van Oostrom et al., 2003).

A cross-cutting theme found in this study is that risk construction is a family affair. Ideas about risk are constructed in relation to interactions with other family members. This is not surprising. Within families many of us carry the same genetic material and have many reminders of this. Many participants in this study felt that having family support throughout the genetic testing experience helped them cope and make decisions. Moreover, the concern related to risk goes beyond the affected individual and to the children. Hence, genetic counselling services and psychological supports should be

offered to all members of the family. During these sessions specific attention should be paid to clients' sense of relational responsibility and concern for the children. Although this study did not specifically address the ethical considerations of genetic testing, health care providers need to be prepared to address questions surrounding this.

This study found that risk perception was a key factor in influencing how participants made the decision of when to undergo genetic testing. For many the decision to engage in genetic testing was a non-decision; for others, it was something that developed over time. By being more aware of the decision-making process, health care providers can better recognize and assess critical events or conditions that may change clients' decision to engage or disengage in genetic testing and can better support clients to make this transition. Finally, further studies that explore the experiences of those who refuse genetic testing are imperative if we are to gain a complete picture of the relationship between experiences of risk and decisions about genetic testing.

Previous scholars have demonstrated that individual representations of risk vary greatly and that it does not suffice to provide information about risk; rather, health care providers need to explore these representations and move towards a more collaborative understanding of risk in order to facilitate informed decision-making for those considering predictive genetic testing (Cox & McKellin, 1999; Ozanne et al., 2010; Parsons & Atkinson, 1992; Richards, 1992; Shiloh et al., 2006;). My findings support that work and point specifically to the need to create a relational space (e.g., common meeting place), wherein the users of genetic services (perhaps including those who choose not to avail of genetic services), clinicians, and scientists can "freely" discuss competing ideas about risk using a multidisciplinary approach. This relational space is

one where ideas about risk are discussed collaboratively amongst laypersons and experts; where the contextual nature of risk can be discussed; and where the implicit and explicit meanings of risk can be scrutinized and used, altered, or dismissed during care plan decision-making. This plan needs to be fluid; it needs to account for how individuals' ideas about risk shift and evolve. The failure to create such a relational space can result in disengagement in the genetic testing process.

Another finding of this study is that—even though science has proven that ARVC does affect women—laypersons as well as experts continue to associate ARVC as being a male disease. Further research into the gendering of genetically linked conditions would provide a fuller understanding of how the gendering of a disease influences risk construction.

A theoretical understanding of risk provides a framework that can guide fundamental questions about how individuals at risk conceptualize risk. This knowledge is critical in order to anticipate, identify, and provide interventions for those factors that may negatively influence health outcomes. From a theoretical standpoint studies such as this one provide a model that health care providers can use to become aware of how individuals at risk for other genetically linked conditions and chronic diseases assign meanings to their own risk and make subsequent decisions related to that risk. Specifically, this study provides further insights into the role of technology in constructing the sense of risk. In light of new genetic advancements (e.g., nanotechnology and genetic engineering) this knowledge will be beneficial in resource planning and implementation.

Further, my study highlights the importance of having psychological counselling services available to members of at-risk families throughout all phases of the genetic testing process, regardless of genetic status or length of time since receiving one's test results. Likewise, knowing that one's behavioural responses to being at-risk are constructed in a pragmatic manner—participation in healthy behaviours, genetic testing, or prescribed treatments depends on how one's beliefs about genetics and risk fits within the proposed model of care (as noted also by Condit, 2010)—access to psychological supports is imperative. Psychological support (informal and formal) in this study was noted to have facilitated coping as participants reshaped risk perceptions, made informed decisions about their health, and adjust to a new meaning of being at-risk. Finally, given that one's beliefs about inheritance are continuously being shaped, it is important that the psychological supports provided reflect the client's current existing reality of risk; otherwise, the probability of disengagement in the genetic testing process will be high.

At a deeper level, this study moved beyond previous studies on the psychological and social contexts of predictive genetic testing, illustrating the complexity of those contexts. Previous research has tended to be cross-sectional and thus not sensitive to how the meaning assigned to being at-risk is pragmatic, transient, and fluid. My findings illustrate how, as the contextual factors affecting one's risk perception changes, so does the meaning assigned to risk and one's responses to the risk. For example, as scientists moved closer and closer to discovery of the ARVC gene, participants were invited to engage in genetic testing at various stages throughout this process. Each time they were invited to testing, their decision to participate was reflective of the contextual nature of their lives and knowledge at the time. Hence, the decision to engage in genetic testing for

those in the 1980s, 1997, and 2008 were significantly variable, as was the meaning assigned to risk. Evident throughout this study is that risk is saliently embedded within participants' lives and contextual in nature.

Further, this study found that, although the relevancy of the conditions influencing risk perception does vary from person to person and is dependent on one's experiences, there are several conditions that significantly impact risk perception (e. g., physical signs of the disease). Figure 9.1 provides a synopsis of the key conditions that can shape and reshape how participants construct ideas about their risk. That is, as individuals move closer to assigning meaning to their risk, they move closer to completing the metaphorical Rubik's Cube puzzle. Being aware of these specific conditions, practitioners can assess for their presence and provide resources to help participants decipher their true relevancy and meaning in terms of the framing of risk. Also, knowing that the existence of certain conditions (e.g., approaching age of disease onset) can heighten one's sense of risk, providing access to psychological supports during these critical times is important. Knowledge of these conditions can be used in future research to both develop psychometric tools that assess for the presence of these conditions and provide appropriate resources to deal with any concerns. The metaphoric model of the Rubik's Cube can be used as an assessment tool to help practitioners understand the conditions that influence how users or non-users of genetic services construct ideas about risk and make subsequent health decisions. This model can be useful as a focal point during discussions with individuals in order to identify conditions that hold relevancy at any particular point. Future research is needed to test the applicability of this model to other genetic and chronic conditions.

One important incidental finding that emerged out of this attention to the specific conditions that shape risk perception was the variation between children's and adult's responses to being at-risk. We can draw on the findings of this study to shed light on why research has found that children living in at-risk families do not have a high level of psychological distress. For children, risk is not a relevant concept in relation to a genetic disease that will develop later. It is only as these at-risk children mature and are faced with decisions about their lives or start to have clinical manifestations of the disease that they revisit the meaning of being at-risk. Given that these findings emerged only incidentally in my study (when two children participated in focus groups at the request of their parents), and that the stories of the older participants were retrospective, further research is needed to confirm this assumption.

A second incidental finding, again related to the focus on how the specific conditions may shape risk perception for the individual, was that a treatment modality itself might significantly shape or reshape risk perception. A significant finding of this study is that participants spent a considerable amount of time worrying about the actual need for treatments (particularly in the case of those with no visible signs), the efficacy of treatments, and the management of treatments, particularly in terms of the ICD. A considerable amount of psychosocial distress was related to receiving an ICD, warranting further research on the impact of adjusting to living with a new and complex treatment technology. Further research will help to identify whether and how coinciding psychological resources should be available to help individuals become knowledgeable about the purpose, capabilities, and management of novel treatment devices.

This research also led to important information about how *absent* or *inconclusive* test results may shape the understanding of risk. A key finding in this study was the dissatisfaction among at-risk individuals when scientific knowledge over a span of many years remained unable to provide them with answers as to why family members were dying so young of heart conditions. In fact, even when there was a more definitive test, many still questioned its accuracy and experienced psychological distress, regardless of carrier status, for many years. This finding—that clients themselves may question the assumption that evidence produced through biomedical research is an objective and static truth, as it, too, evolves over time— is significant for our understanding of how risk is understood and reshaped in light of scientific advances. Added to this was the fact that existing treatment modalities were inconsistent, and many felt they had to be their own advocates for care. Based on these findings it is important that laypersons are included as part of the research team at all stages of new gene discoveries as “lay experts.” Only by understanding the contextual and historical factors of their everyday lives, captured through their stories, can health care services be reflective of the needs of at-risk individuals.

Findings specific to ARVC and its management within the NL context were also noted. First, the emphasis among participants on pragmatic but varied approaches to coping with the disease is significant for health services delivery. Individuals drew upon the coping mechanism that best fit with their existing perception of risk. For example, several participants tried to normalize the firing of the ICD as a routine occurrence given the nature of ARVC. Knowing that ARVC has a sudden onset, many spent considerable time surveying their bodies and that of their children for signs of ARVC. The diverse

coping mechanisms that have proven effective throughout the genetic testing process to help participants cope with living in a family at-risk can be offered as suggestions to other at-risk families.

Second, this study identified specific significant barriers to genetic testing related to human resources, finances, and physical resources that hold relevance for future health policy surrounding genetic services, particularly in rural communities. Currently there is a lack of dedicated genetic services in central or western NL. Access to health care practitioners with an expertise in genetics is needed in the rural areas. Technology may be the most efficient way to address this gap, such as telehealth services. Another option to deal with identified gaps in knowledge related to ARVC management is the further development of ARVC clinical practice guidelines. In addition, I recommend that continuing education about ARVC management be provided for health care providers, particularly locum physicians. Orientation programs could include the required information on this condition and associated treatment. These suggestions were noted by participants to be essential for safe, competent, consistent, and timely care.

In particular, geography was a barrier to effective care. Living with ARVC can be financially challenging. Given the geographical constraints related to access to health care in NL, policy makers need to consider making the costs incurred while accessing genetic services and obtaining medications an insurable benefit. The implementation of more physical resources within the community—specifically, access to the technology to download information from the ICD—would offset travel costs. A qualified health care practitioner could then review this information by remote access and give timely advice. Not only would this solution alleviate financial costs but also the psychological distress

that comes with not knowing why an ICD fired. Being at risk for ARVC also means that life and health insurance may not be available or that it may be too costly. Given that NL has become known for its myriad of genetic conditions, it is important that health policy makers spend time developing guidelines for insurable services that avoid genetic discrimination. This includes regulations around driving restrictions post-firing of the ICD.

This study, by examining how ideas about risk are constructed alongside new genetic discoveries, provides direction to best practices for introducing and applying novel genetic technologies. Evident throughout this study is that the concept of risk is best understood within the nexus of social, historical, and biological relationships. Unfortunately, these factors that motivate individuals to participate in genetic testing seem to be discussed in silo in the literature. In order to understand how risk is constructed, it is important to acknowledge the intersection of these three contextual factors. To discuss either of these factors in silo will fragment our understanding of how individuals construct their meanings of risk and make health care decisions. Individuals in this study juxtaposed scientific knowledge against experiential knowledge and specific conditions.

An important finding of this study that requires further research is that experiential knowledge shaped the trajectory of scientific knowledge to some extent. It is therefore important for health care providers and researchers to actively engage laypersons in the research process, the implementation of best practice guidelines, and the development of health care policy.

This study demonstrates that qualitative research is the most appropriate method to discover and understand the needs of at-risk families, to discover the psychosocial process of how ideas about risk are understood and constructed, and to offer suggestions about allocation of health care resources. Qualitative studies such as this also challenge the static construction of risk that emerges from quantitative studies. The majority of studies on the psychosocial impact of genetic testing are either quantitative or cross-sectional, and hence they do not capture the social process of genetic testing and can often fragment the experience. Cross-sectional studies are not sensitive to changing concepts over time such as relevancy, which was shown to have a significant impact on risk perception in this study. Qualitative longitudinal studies would be a more appropriate means to capture the transient nature of risk perception. Many of the tools used to study risk are one-dimensional in nature, have been adapted from other measurement tools, and borrowed from other disciplines such as psychology, which may not be sensitive to measurements of risk perception in genetic conditions. Also, many psychometric tools are used at inconsistent intervals, thus capturing different realities of risk, and may miss critical transient events that shape risk perception. Risk as shown in this study is a multidimensional concept that persists for a long time after receiving one's genetic test result, regardless of carrier status. More qualitative longitudinal studies are needed to gain a fuller understanding of how risk is shaped and reshaped in relation to scientific knowledge, experiential knowledge, and conditions that impact risk.

I believe that the concepts discussed in this study (three contextual dimensions and conditions) have conceptual generalizability (as discussed by Green & Thorogood, 2009) to other at-risk populations or those living with a chronic condition. That is, how

an individual constructs ideas about their risk is dependent on the interactions between the three contextual dimensions and the conditions that have been experienced in everyday life. Although the components of the contextual dimensions and conditions may change, the process remains the same and can be used as a framework for understanding risk. The Rubik's Cube model can be used as a way to understanding how individuals at risk for a genetically linked condition or another chronic condition move towards puzzle completion, or, in other words, a greater understanding of their risk.

Limitations

As with every study there were limitations to my research, many related to constraints of time and geography.

Sample

There were six limitations related to the sample. First, the majority of participants were of English or Irish descent (in keeping with this founder population), middle class, employed, and had completed high school education. Thus, the perspectives of individuals living under lower social economic conditions may not have been fully captured in this study. Second, the fact that the majority of participants lived in small rural communities may account for the numerous references related to barriers to health care services. These barriers may not hold true for those closer to tertiary care centers. Third, there were no participants between the ages of 21 and 30; hence the voices of this population were based on retrospective accounts of the older participants, who may not remember relevant details of their experience with predictive genetic testing. This may be significant given that many momentous life choices (e.g., career choices, marriage) often happen in that age group and may not be adequately represented in this study.

Fourth, there was only one ARVC negative male, which may predispose the study to some gender bias. Fifth, there were only two younger participants (ages 14 and 15); hence, this study did not fully examine the experience of this population, which warrants further research. Finally, although I did have one individual who refused the ICD, I did not have any participants who refused genetic testing. Further research with this population of resisters would add to the understanding of the relationship between risk construction and uptake of genetic services.

Method

Another critique of this study is that I used focus groups as a data collection method. It has been argued that focus groups are not as useful as individual interviews in grounded theory, as they do not usually provide a rich source of data (Morse, 2001). In contrast Carey and Asbury (2012) argue that through the use of focus groups researchers are enabled to get the most of the interactions between participants and as a consequence "enhance the collection of deep, strongly held beliefs and perspectives" (pp. 17). Therefore, I argue that the use of focus groups was more appropriate for this particular topic given that genetics has to do with the shared nature of genes and has implications for all family members. By using focus groups I was able to capture how ideas about risk were made in relation to others. More importantly, I was able to observe the fluid nature of risk first-hand by listening to at-risk family members tell their stories to other members of the group, as risk for many was reshaped during this very encounter. Finally, there has been a dearth of literature looking at the experiences of spouses and other family members living in an at-risk family; by using focus groups I was able to fill this gap.

Regarding the actual data analysis and collection, I faced two challenges. The first was in relation to research bias. Given my clinical experience as a nurse, I found that I had to consciously bracket my own personal thoughts from those of the participants. As discussed in chapter 5: Methodology, I created diagrams and memos and labelled them “ME.” This helped me to situate myself within the research process and be cognizant of my own preconceived ideas about predictive genetic testing in order to allowed transparency of the emerging theory, *Constructing the Meaning of Being At-Risk*.

The second challenge related to method was in regards to the practical aspects of recruitment for the focus groups. Two of the focus groups occurred within one day. This limited my ability to reflect on the interviews and data analysis prior to the subsequent interview. Also, although I knew for the most part who would be participating in each group, I was not aware that the two young participants would be coming; thus, I did not follow “true” theoretical sampling for focus groups. Ethical approval was given to include the two young participants; however, in order to fully capture the experiences of children, more research using this population is needed.

Final Reflection

This study provided invaluable insights into how ideas about risk are constructed throughout the genetic testing process. This study is unique in that it examines the historical and contextual evolution of risk for a new genetic condition. The Rubik’s Cube is proposed as a framework that health care providers can employ to gain a fuller understanding of how the key contextual dimensions and conditions fit together and influence one’s sense of being at-risk. It is only by gaining a fuller understanding of how at-risk individuals juxtapose their experiential knowledge against their scientific

knowledge and specific conditions in their everyday lives that adequate resources to meet the needs of this population can be developed.

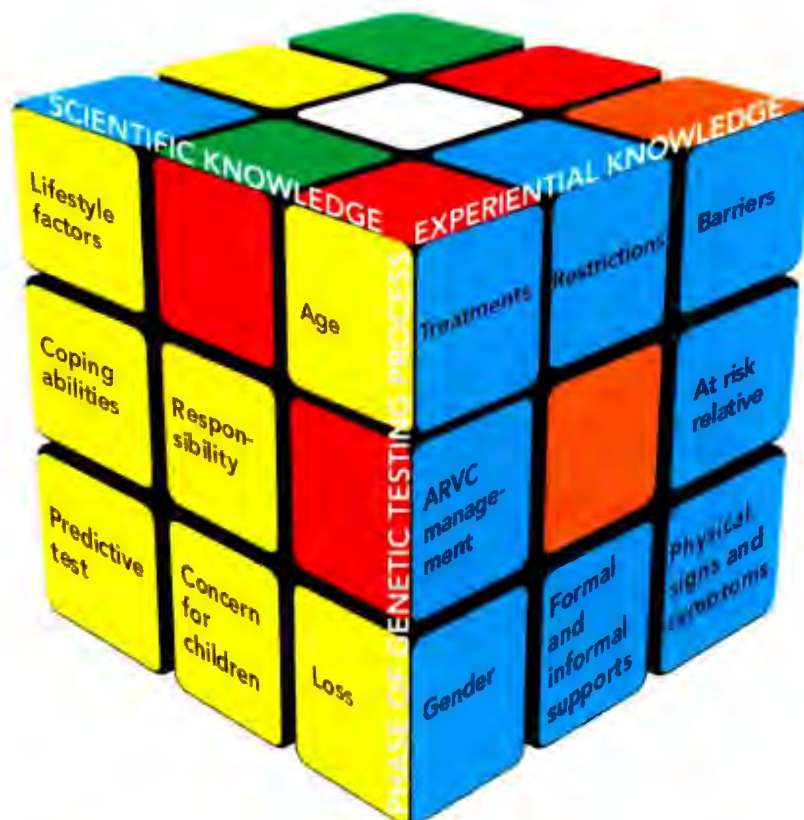


Figure 9.1. Rubik's Cube Model: Summary of the Conditions that Influence the Psychosocial Process of *Constructing the Meaning of Being At-Risk*. Rubik's Cube ® used by permission of Seven Towns Limited, www.rubiks.com

References

- Aatre, R. D., & Day, S. M. (2011). Psychological issues in genetic testing for inherited cardiovascular diseases. *Circulation: Cardiovascular Genetics*, 4(1), 81-90. doi: 10.1161/CIRCGENETICS.110.957365
- Agard, A., Lofmark, R., Edvardsson, N., & Ekman, I. (2007). Views of patients with heart failure about their role in the decision to start implantable cardioverter-defibrillator treatment: prescription rather than participation. *Journal of Medical Ethics*, 33, 514-518. doi: 10.1136/jme.2006.017723
- Ahmad, F., Li, D., Karibe, A., Gonzales, O., Tapscott, T., Hill, R., ... Roberts, R. (1998). Localization of a gene responsible for arrhythmogenic right ventricular dysplasia to chromosome 3p23. *Circulation*, 98, 2791-2795. doi:10.1161/01.CIR.98.25.2791
- Aktan-Collan, K., Haukkala, A., Mecklin, J. P., Uutela, A., & Kaarianen, H. (2001). Psychological consequences of predictive genetic testing for hereditary non-polyposis colorectal cancer (HNPCC). A prospective follow-up study. *International Journal of Cancer*, 94, 608-611. doi: 10.1002/ijc.1372
- Allain, D. C., Friedman, S., & Senter, L. (2012). Consumer awareness and attitudes about insurance discrimination post enactment of the genetic information non-discrimination act. *Familial Cancer*, 11, 637-644. doi: 10.1007/s10689-012-9564-0
- Allmark, P., & Tod, A. (2005). How should public health professionals engage with lay epidemiology? *Journal of Medical Ethics*, 32, 460-463. doi: 10.1136/jme.2005.014035

- Almqvist, E., Brinkman, R., Wiggins, S., & Hayden, R. (2003). Psychological consequences and predictors of adverse events in the first 5 years after predictive testing for Huntington's disease. *Clinical Genetics*, 64, 300-309. doi:10.1034/j.1399-0004.2003.00157.x
- Anderson, E (2006). Arrhythmogenic right ventricular dysplasia. *American Family Physician*, 73, 1391-1398. Retrieved from <http://www.aafp.org/afp/2006/0415/p1391.html>
- Andersen, J., Oyen, N., Bjorvatn, C., & Gjengedal, E. (2008). Living with long QT syndrome: A qualitative study of coping with increased risk of sudden cardiac death. *Journal of Genetic Counseling*, 17, 489-498. doi:10.1007/s10897-008-9167-y
- Anderson, S.L. (2004). The human genome project. In S. S. Sanbar. (Ed.), *Legal Medicine* (6th ed., pp. 256-263). Philadelphia: Mosby.
- Andrews, L., Mireskandari, S., Jessen, J., Thewes, B., Solomon, M., Macrae, F., & Meiser, B. (2006). Impact of familial adenomatous polyposis on young adults: Attitudes toward genetic testing, support, and information needs. *Genetics in Medicine*, 8, 697-703. doi:10.1097/01.gim.0000245574.75877.b9
- Angus, J., Evans, S., Lapum, J., Rukholm, E., St. Onge, R., Nolan, R., & Michel, I. (2005). "Sneaky disease": The body and health knowledge for people at risk for coronary heart disease in Ontario, Canada. *Social Science & Medicine*, 60, 2117-2128. doi: 10.1016/j.socscimed.2004.08.069
- Annels, M. (1996). Grounded theory method: Philosophical perspectives, paradigm of inquiry, and postmodernism. *Qualitative Health Research*, 6, 379-393. doi: 10.1177/10497323960060030

Armstrong, K., Calzone, K., Stopfer, J., Fitzgerald, G., Coyne, J., & Weber, B. (2000).

Factors associated with decisions about clinical BRCA1/2 testing. *Cancer*

Epidemiology, Biomarkers, 9, 1251-1254. Retrieved from

<http://cebp.aacrjournals.org/content/9/11/1251.full.pdf+html>

Arver, B., Haegermark, A., Platten, U., Lindblom, A., & Brandberg, Y. (2004). Evaluation

of psychosocial effects of pre-symptomatic testing for breast/ovarian and colon

cancer pre-disposing genes: A 12-month follow-up. *Familial Cancer, 3*, 109-16.

doi: 10.1023/B:FAME.0000039863.89137.f9

Bailey, D., Skinner, D., Davis, A.M., Whitmarsh, I., & Powell, C. (2008). Ethical, legal,

and social concerns about expanded newborn screening: Fragile X syndrome as a

prototype for merging issues. *Pediatrics, 121*, e693-e704. doi: 10.1542/peds.2007-

0820

Baker, C., Wuest, J., & Stern, P.N. (1992). Method slurring: the grounded

theory/phenomenology example. *Journal of Advanced Nursing, 17*, 1355-1360. doi:

10.1111/j.1365-2648.1992.tb01859.x

Bandura, A. (2004). Health promotion by social cognitive means. *Health Education &*

Behaviour, 31, 143-164. doi: 10.1177/1090198104263660

Basso, C., Corrado, D., Marcus, F.I., Nava, A., M., & Thiene, G. (2009). Arrhythmogenic

right ventricular cardiomyopathy. *Lancet, 373*, 1289-1300. doi: 10.1016/S0140-

673(09)60256-7

Beck, U. (1992) *Risk society: Towards a new modernity*. London: Sage.

- Beck, U. (1996). World risk society as cosmopolitan society? Ecological questions in a framework of manufactured uncertainties. *Theory, Culture & Society*, 13, 1-32. doi: 10.1177/0263276496013004001
- Beck, U. (2006). Living in the world risk society. *Economy and Society*, 35, 329-345. doi: 10.1080/03085140600844902
- Beck, U., & Beck-Gernsheim, E. (2002). *Individualization*. California: SAGE Publications.
- Beck, U., Bonss, W., & Lau, C. (2003). The theory of reflexive modernization: Problematic, hypotheses and research programme. *Theory, Culture & Society*, 20, 1. doi: 10.1177/0263276403020002001
- Beery, T. A., & Williams, J. K. (2007). Risk reduction and health promotion behaviours following genetic testing for adult-onset disorders. *Genetic Testing*, 11, 111-123. doi: 10.1089/gte.2006.0527
- Biesecker, B. B. (1998). Future directions in genetic counseling: Practical and ethical considerations. *Kennedy Institute of Ethics Journal*, 8, 145-160. Retrieved from http://muse.jhu.edu/journals/kennedy_institute_of_ethics_journal/v008/8.2biesecker.html
- Bilge, A. K., Ozben, B., Demircan, S., Cinar, M., Yilmaz, E., & Adalet, K. (2006). Depression and anxiety status of patients with implantable cardioverter defibrillator and precipitating factors. *Pacing and Clinical Electrophysiology*, 29, 619-626. doi: 10.1111/j.1540-8159.2006.00409

- Binedell, J., Soldan, J.R., & Harper, P.S. (1998). Predictive testing for Huntington's disease: Predictors of uptake in South Wales. *Clinical Genetics*, 54, 477-88. doi: 10.1111/j.1399-0004.1998.tb03768.x
- Bleiker, E. M., Hahn, D. E., & Aaronson, N. K. (2003). Psychosocial issues in cancer genetics--current status and future directions. *Acta Oncologica (Stockholm, Sweden)*, 42, 276-286. doi: 10.1080/02841860310004391
- Bleiker, E. M., Menko, F. H., Taal, B. G., Kluijdt, I., Wever, L. D., Gerritsma, M. A., . . . Aaronson, N. K. (2005). Screening behaviour of individuals at high risk for colorectal cancer. *Gastroenterology*, 128, 280-287. doi: 10.1053/j.gastro.2004.11.002
- Blumer, H. (1969). *Symbolic interactionism. Perspective and method*. New Jersey: Prentice-Hall Inc.
- Boenink, M., & van der Burg, S. (2010). Informed decision making about predictive DNA tests: Arguments for more public visibility of personal deliberations about the good life. *Medicine, Health Care, and Philosophy*, 13, 127-138. doi: 10.1007/s11019-009-9227-6
- Bolse, K., Hamilton, G., Flanagan, J., Carroll, D.L., & Frilund, B. (2005). Ways of experiencing the life situation among United States patients with an implantable cardioverter-defibrillator: A qualitative study. *Progress in Cardiovascular Nursing*, 20, 4-10. Retrieved from <http://www.ncbi.nlm.nih.gov/pubmed/15785164>
- Bombard, Y., Palin, J., Friedman, J. M., Veenstra, G., Creighton, S., Bottorff, J. L., . . . & Canadian Respond-HD Collaborative Research Group. (2012). Beyond the patient: The broader impact of genetic discrimination among individuals at risk of

- Huntington disease. *American Journal of Medical Genetics. Part B, Neuropsychiatric Genetics*, 159B, 217-226. doi: 10.1002/ajmg.b.32016
- Bombard, Y., Penziner, E., Suchowersky, O., Guttman, M., Jane, S. P., & Joan, L. B. (2008). Engagement with genetic discrimination: Concerns and experiences in the context of Huntington disease. *European Journal of Human Genetics*, 16, 279-89. doi: 10.1038/sj.ejhg.5201937
- Bourdieu, P. (1977). *Outline of a theory of practice*. Cambridge: Cambridge University Press.
- Bostwick, J. M., & Sola, C. L. (2011). An updated review of implantable Cardioverter/Defibrillators, induced anxiety, and quality of life. *Heart Failure Clinics*, 7(1), 101-108. doi: 10.1016/j.hfc.2010.10.003
- Boyce, W. (2002). Influence of health promotion bureaucracy on community participation: a Canadian case study. *Health Promotion International*, 17(1), 61-68. doi: 10.1093/heapro/17.1.61
- Braithwaite, D., Emery, J., Walter, F., Prevost, A. T., & Sutton, S. (2004). Psychological impact of genetic counseling for familial cancer: A systematic review and meta-analysis. *Journal of the National Cancer Institute*, 96, 122-133. doi: 10.1093/jnci/djh017
- Braithwaite, D., Emery, J., Walter, F., Prevost, A. T., & Sutton, S. (2006). Psychological impact of genetic counseling for familial cancer: A systematic review and meta-analysis. *Familial Cancer*, 5(1), 61-75. doi: 10.1007/s10689-005-2577-1

- Braithwaite, D., Sutton, S., Mackay, J., Stein, J., & Emery, J. (2005). Development of a risk assessment tool for women with a family history of breast cancer. *Cancer Detection and Prevention*, 29, 433-439. doi: 10.1016/j.cdp.2005.06.001
- Bratich, J. Z., Packer, J., & McCarthy, C. (2003). Governing the Present. In J. Z. Bratich, J. Packer & C. McCarthy (Eds.), *Foucault, Cultural Studies, and Governmentality* (pp. 3-21.). Albany NY: State University of New York Press.
- Broadstock, M., Michie, S., Gray, J., Mackay, J., & Marteau, T. M. (2000). The psychological consequences of offering mutation searching in the family for those at risk of hereditary breast and ovarian cancer-a pilot study. *Psycho-Oncology*, 9, 537-548. doi: 10.1002/1099-1611(200011/12)
- Broadstock, M., Michie, S., & Marteau, T. (2000). Psychological consequences of predictive genetic testing: A systematic review. *European Journal of Human Genetics*, 8, 731-738. doi: 10.1038/sj.ejhg.5200532
- Brorsson, A., Troein, M., Lindbladh, E., Selander, S., Widlund, M., & Rastam, L. (1995). My family dies from heart attacks. How hypercholesterolaemic men refer to their family history. *Family Practice*, 12, 433-437. doi: 10.1093/fampra/12.4.433
- Brunger, F., & Bassett, K. (1998). Culture, ethnicity and genetic testing. In B.M. Knoppers (Ed.), *Socio-ethical issues in human genetics* (pp. 7-42). Quebec: Les Éditions Yvon Blais.
- Bruno, M., Tommasi, S., Stea, B., Quaranta, M., Schittulli, F., & Mastropasqua, A. (2004). Awareness of breast cancer genetics and interest in predictive genetic testing: A survey of a southern Italian population. *Annals of Oncology*, 15(1), I48-I54. doi: 10.1093/annonc/mdh658

- Butow, P. N., Lobb, E. A., Meiser, B., Barratt, A., & Tucker, K. M. (2003). Psychological outcomes and risk perception after genetic testing and counselling in breast cancer: A systematic review. *Medical Journal of Australia*, 178(2), 77-81. Retrieved from <https://www.mja.com.au/journal/2003/178/2/psychological-outcomes-and-risk-perception-after-genetic-testing-and-counselling>.
- Caga-ana, E.C.F., Smith, L., Sharp, R.R., & Lantos, J.D. (2012). Testing children for adult-onset genetic diseases. *Pediatrics*, 129(1), 163-167. doi:10.1542/peds.2010-3743
- Cameron, L. D., Sherman, S., Marteau, T., & Brown, P. (2009). Impact of genetic risk information and type of disease on perceived risk, anticipated affect, and expected consequences of genetic tests. *Health Psychology*, 28, 307-16. doi:10.1037/a0013947
- Carey, M. A. & Asbury, J. (2012). *Focus group research*. Walnut Creek, CA: Left Coast Press.
- Castel, R. (1991). From dangerousness to risk. In G. Burchell, C. Gordon., & P. Miller (Eds.), *The Foucault effect: Studies in governmentality*. (pp. 281-298). Chicago: University of Chicago Press.
- Chan, C. W., Lopez, V., & Chung, J. W. (2011). A qualitative study of the perceptions of coronary heart disease among Hong Kong Chinese people. *Journal of Clinical Nursing*, 20(7-8), 1151-1159. doi: 10.1111/j.1365-2702.2010.03526.x
- Chenitz, W., & Swanson, J. (1986). *From practice to grounded theory: Qualitative research in nursing*. Menlo Park, CA: Addison-Wesley.
- Chivers Seymour, K., Addington-Hall, J., Lucassen, A.M., & Foster, C.L. (2010). What facilitates or impedes family communication following genetic testing for cancer

- risk? A systematic review and meta-synthesis of primary qualitative research. *Journal of Genetic Counseling*, 19, 330-342. doi: 10.1007/s10897-010-9296-y
- Christiaans, I., van Langen, I.M., Birnie, E., Bonsel, G.J., Wilde, A.M., & Smets, E.M.A. (2009). Quality of life and psychological distress in hypertrophic cardiomyopathy mutation carriers: A cross-sectional cohort study. *American Journal of Medical Genetics. Part A*, 149, 602-612. doi: 10.1002/ajmg.a.32710
- Claes, E., Evers-Kiebooms, G., Boogaerts, A., Decruyenaere, M., Denayer, L., & Legius, E. (2004). Diagnostic genetic testing for hereditary breast and ovarian cancer in cancer patients: Women's looking back on the pre-test period and a psychological evaluation. *Genetic Testing*, 8(1), 13-21. doi: 10.1089/109065704323015996
- Clarke, A. (2010). What is at stake in the predictive testing of children? *Familial Cancer*, 9, 19-22. doi: 10.1007/s10689-009-9260-x
- Clarke, A.M. (1998). The qualitative-quantitative debate: moving from positivism and confrontation to post-positivism and reconciliation. *Journal of Advanced Nursing*, 27, 1242-1249. doi: 10.1046/j.1365-2648.1998.00651.x
- Codori, A. M., & Brandt, J. (1994). Psychological costs and benefits of predictive testing for Huntington's disease. *American Journal of Medical Genetics*, 54, 174-184. doi: 10.1002/ajmg.1320540304
- Codori, A. M., Petersen, G. M., Boyd, P. A., Brandt, J., & Giardiello, F. M. (1996). Genetic testing for cancer in children. short-term psychological effect. *Archives of Pediatrics & Adolescent Medicine*, 150, 1131-1138.

- Codori, A. M., Petersen, G. M., Miglioretti, D. L., & Boyd, P. (2001). Health beliefs and endoscopic screening for colorectal cancer: Potential for cancer prevention. *Preventive Medicine, 33*(2), 128-136. doi: 10.1006/pmed.2001.0862
- Codori, A. M., Slavney, P. R., Rosenblatt, A., & Brandt, J. (2004). Prevalence of major depression one year after predictive testing for Huntington's disease. *Genetic Testing, 8*, 114-119. doi: 10.1089/1090657041797275
- Codori, A. M., Slavney, P. R., Young, C., Miglioretti, D. L., & Brandt, J. (1997). Predictors of psychological adjustment to genetic testing for Huntington's disease. *Health Psychology, 16*(1), 36-50. doi: 10.1037/0278-6133.16.1.36
- Codori, A. M., Waldeck, T., Petersen, G. M., Miglioretti, D., Trim bath, J. D., & Tillery, M. A. (2005). Genetic counseling outcomes: Perceived risk and distress after counseling for hereditary colorectal cancer. *Journal of Genetic Counseling, 14*, 119-132. doi: 10.1007/s10897-005-4062-2
- Codori, A. M., Zawacki, K. L., Petersen, G. M., Miglioretti, D. L., Bacon, J. A., Trim bath, J. D., . . . Giardiello, F. M. (2003). Genetic testing for hereditary colorectal cancer in children: Long-term psychological effects. *American Journal of Medical Genetics. Part A, 116A* (2), 117-128. doi: 10.1002/ajmg.a.10926
- Collins, V., Meiser, B., Gaff, C., John, D. J. B. S., & Halliday, J. (2005). Screening and preventive behaviours one year after predictive genetic testing for hereditary nonpolyposis colorectal carcinoma. *Cancer, 104*, 273-281. doi: 10.1002/cncr.21183
- Collins, V. R., Meiser, B., Ukoumunne, O. C., Gaff, C., St John, D. J., & Halliday, J. L. (2007). The impact of predictive genetic testing for hereditary nonpolyposis

- colorectal cancer: Three years after testing. *Genetics in Medicine*, 9, 290-297. doi: 10.1097/GIM.0b013e31804b45db
- Condit, C. M. (2010). Public understandings of genetics and health. *Clinical Genetics*, 77(1), 1-9. doi: 10.1111/j.1399-0004.2009.01316.x
- Corrado, D., Fonatine, G., Marcus, F.I., McKenna, W.J., Nava, A., Thiene, G., & Wichter, T. (2000). Arrhythmogenic right ventricular dysplasia/cardiomyopathy: Need for an international registry. *Circulation*, 101, e101-e106. doi: 10.1161/01.CIR.101.11.e101
- Corbin, J. (1986). Qualitative data analysis for grounded theory. In W.C. Chenitz, & J.M. Swanson (Eds.), *From practice to grounded theory: Qualitative research in nursing* (pp. 91-101). Menlo-Park, CA: Addison Wesley.
- Covello, V. T., & Mumpower, J. (1985). Risk analysis and risk management: An historical perspective. *Risk Analysis*, 5(2), 103-120. doi: 10.1111/j.1539-6924.1985.tb00159.x
- Cowan, J., Morales, A., Dagua, J., & Ray, E. H. (2008). Genetic testing and genetic counseling in cardiovascular genetic medicine: Overview and preliminary recommendations. *Congestive Heart Failure (Greenwich, Conn.)*, 14(2), 97-105. doi: 10.1111/j.1751-7133.2008.08217.x
- Cox, M.G.P.J., van der Smagt, J.J., Noorman, M., Wiesfeld, A.C., Volders, P.G.A., van Langen, I. M., ...Hauer, R.N.W (2010). Arrhythmogenic right ventricular dysplasia/cardiomyopathy diagnostic task force criteria. Impact of new task force criteria. *Circulation Arrhythmia and Electrophysiology*, 3(2), 126-133. doi: 10.1161/CIRCEP.109.927202

- Cox, S. M. (2003). Stories in decisions: How at-risk individuals decide to request predictive testing for Huntington disease. *Qualitative Sociology*, 26(2), 257-280. Retrieved from <http://web.ebscohost.com/ehost/pdfviewer/pdfviewer?sid=d7522ca7-f10e-4de9-8eed-9f67e4fea451%40sessionmgr113&vid=2&hid=112>
- Cox, S. M., & McKellin, W. (1999). 'There's this thing in our family': Predictive testing and the construction of risk for Huntington disease. *Sociology of Health & Illness*, 21, 622-646. doi: 10.1111/1467-9566.00176
- Cox, S. M., & Starzomski, R. C. (2004). Genes and geneticization? The social construction of autosomal dominant polycystic kidney disease. *New Genetics and Society*, 23(2), 137-166. doi: 10.1080/1463677042000237071
- Croyle, R. T., & Lerman, C. (1999). Risk communication in genetic testing for cancer susceptibility. *Journal of the National Cancer Institute. Monographs*, (25), 59-66. Retrieved from <http://jncimono.oxfordjournals.org/content/1999/25/59.long>
- Croyle, R. T., Smith, K. R., Botkin, J. R., Baty, B., & Nash, J. (1997). Psychological responses to BRCA1 mutation testing: Preliminary findings. *Health Psychology*, 16(1), 63-72. doi: 10.1037/0278-6133.16.1.63
- Cutcliffe, J. R., & Harder, H. G. (2009). The perpetual search for parsimony: Enhancing the epistemological and practical utility of qualitative research findings. *International Journal of Nursing Studies*, 46, 1401-1410. doi: 10.1016/j.ijnurstu.2009.05.005
- d'Agincourt-Canning, L. (2001). Experiences of genetic risk: Disclosure and the gendering of responsibility. *Bioethics*, 15, 231-247. doi: 10.1111/1467-8519.00234

- d'Agincourt-Canning, L. (2005). The effect of experiential knowledge on construction of risk perception in hereditary breast/ovarian cancer. *Journal of Genetic Counseling*, 14(1), 55-69. doi: 10.1007/s10897-005-1500-0
- d'Agincourt-Canning, L. (2006a). A gift or a yoke? women's and men's responses to genetic risk information from BRCA1 and BRCA2 testing. *Clinical Genetics*, 70, 462-472. doi: 10.1111/j.1399-0004.2006.00720.x
- d'Agincourt-Canning, L. (2006b). Genetic testing for hereditary breast and ovarian cancer: Responsibility and choice. *Qualitative Health Research*, 16(1), 97-118. doi: 10.1177/1049732305284002
- Dalla-Volta, S., Battaglia, G., & Zerbini, E. (1961). "Auricularization" of right ventricular pressure curve. *American Heart Journal*, 61(1), 25-33. doi: 10.1016/0002-8703(61)90513-0
- Dalteg, T., Benzein, E., Fridlund, B., & Malm, D. (2011). Cardiac disease and its consequences on the partner relationship: A systematic review. *European Journal of Cardiovascular Nursing*, *In Press, Corrected Proof*. doi: 10.1016/j.ejcnurse.2011.01.006
- Davison, C. (1996). Predictive genetics: the cultural implications of supplying probable futures. In T. Marteau, & M. Richards. (Eds.), *The troubled helix* (pp. 317-330). Cambridge: University Press.
- Davison, C., Frankel, S., & Smith, G. D. (1989). Inheriting heart trouble: the relevance of common-sense ideas to preventive measures. *Health Education Research*, 4, 329-340. doi: 10.1093/her/4.3.329

- Davison, C., Frankel, S., & Smith, G. D. (1992). The limits of lifestyle: Re-assessing 'fatalism' in the popular culture of illness prevention. *Social Science*, 34, 675-85.
doi: 10.1016/0277-9536(92)90195-V
- Davison, C., Smith, G. D., & Frankel, S. (1991). Lay epidemiology and the prevention paradox: the implications of coronary candidacy for health education. *Sociology of Health & Illness*, 13, 1-19. doi: 10.1111/j.1467-9566.1991.tb00085.x
- Dean (1999a). Risk, calculable and incalculable. In D. Lupton (Ed.), *Risk and sociocultural theory: New directions and perspectives* (pp. 131- 159) New York: Cambridge University Press.
- Dean (1999b). *Governmentality. Power and rule in modern society*. London: Sage Publications.
- Decruyenaere, M., G. E., Boogaerts, A., J. J. C., Cloostermans, T., & Demyttenaere, K. (1999). Psychological functioning before predictive testing for Huntington's disease: The role of the parental disease, risk perception, and subjective proximity of the disease. *Journal of Medical Genetics*, 36, 897-905. doi: 10.1136/jmg.36.12.897
- Decruyenaere, M., Evers-Kiebooms, G., Boogaerts, A., Cassiman, J. J., Cloostermans, T., Demyttenaere, K., . . . Van den Berghe, H. (1995). Predictive testing for Huntington's disease: Risk perception, reasons for testing and psychological profile of test applicants. *Genetic Counseling*, 6(1), 1-13. Retrieved from: <http://hdl.handle.net/10822/527315>
- Decruyenaere, M., Evers-Kiebooms, G., Boogaerts, A., Cloostermans, T., Cassiman, J. J., Demyttenaere, K., . . . Van den Berghe, H. (1997). Non-participation in predictive testing for Huntington's disease: Individual decision-making, personality and

- avoidant behaviour in the family. *European Journal of Human Genetics*, 5, 351-363.
Retrieved from <http://www.ncbi.nlm.nih.gov/pubmed/9450179>
- Decruyenaere, M., Evers-Kiebooms, G., Cloostermans, T., Boogaerts, A., Demyttenaere, K., & Dom, R. (2003). Psychological distress in the 5-year period after predictive testing for Huntington's disease. *European Journal of Human Genetics*, 11(1), 30-38. doi: 10.1038/sj.ejhg.5200913
- Dewey, J. (1922). *Human nature and conduct*. New York: Holt.
- Dickerson, S. S. S. (2002). Redefining life while forestalling death: Living with an implantable cardioverter defibrillator after a sudden cardiac death experience. *Qualitative Health Research*, 12, 360-372. doi: 10.1177/104973202129119946
- Dinc, L., & Terzioglu, F. (2005). The psychological impact of genetic testing on parents. *Journal of Clinical Nursing*, 15(1), 45-51. doi: 10.1111/j.1365-2702.2005.01228.x
- Dorval, M., Geneviève Gauthier, Maunsell, E., Michel, J. D., Rouleau, I., & Chiquette, J. (2005). No evidence of false reassurance among women with an inconclusive BRCA1/2 genetic test result. *Cancer Epidemiology, Biomarkers*, 14, 2862-2867. doi: 10.1158/1055-9965.EPI-05-0512
- Douglas, M. (1966). *Purity and danger: An analysis of concepts of taboo and pollution*. London: Routledge.
- Douglas, M. (1970). *Natural symbols*. London: Barrie and Rockcliffe
- Douglas, M. (1978) *Cultural bias*. London: Royal Anthropological Institute.
- Douglas, M. (1985). *Risk acceptability according to the social sciences*. New York: Russell Sage.

- Douglas, M. (1990). Risk as a forensic science. *Daedalus*, 119(4), 1–16. Retrieved from <http://www.jstor.org/stable/2002533>
- Douglas, M. (1992). *Risk and blame*. London: Routledge.
- Douglas, M., & Wildavsky, A. (1982a). *Risk and culture: An essay on the selection of technological and environmental dangers*. Berkeley: University of California Press.
- Douglas, M., & Wildavsky, A. (1982b). How can we know the risks we face? why risk selection is a social Process. *Risk Analysis*, 2(2), 49-58. doi: 10.1111/j.1539-6924.1982.tb01365
- Douma, K. F., Aaronson, N. K., Vasen, H. F., & Bleiker, E. M. (2008). Psychosocial issues in genetic testing for familial adenomatous polyposis: A review of the literature. *Psycho-Oncology*, 17, 737-745. doi: 10.1002/pon.1357
- Douma, K. F., Bleiker, E. M., Vasen, H. F., Gundy, C. M., Gerritsma, M. A., & Aaronson, N. K. (2010). Psychological distress and quality of life of partners of individuals with familial adenomatous polyposis. *Psycho-Oncology*, doi: 10.1002/pon.1716
- Duncan, R. E., Gillam, L., Savulescu, J., Williamson, R., John, G. R., & Martin, B. D. (2007). "Holding your breath": Interviews with young people who have undergone predictive genetic testing for Huntington disease. *American Journal of Medical Genetics. Part A*, 143A, 1984-1989. doi: 10.1002/ajmg.a.31720
- Duncan, R. E., Gillam, L., Savulescu, J., Williamson, R., John, G. R., & Martin, B. D. (2008). "You're one of us now": Young people describe their experiences of predictive genetic testing for Huntington disease (HD) and familial adenomatous polyposis (FAP). *American Journal of Medical Genetics. Part C, Seminars in Medical Genetics*, 148C (1), 47-55. doi: 10.1002/ajmg.c.30158

- Duster, T. (2003). *Backdoor to eugenics* (2nd ed.). London: Routledge.
- Einsiedel, E.F. (2000). Understanding “publics” in the public understanding of science. In M. Dierkers & C. vonGote. (Eds.), *Between understanding trust: The public, science, and technology* (pp. 144-150). Amsterdam: The Gordon and Breach Publishing Company.
- Emsenauer, R.E., Michels, V.V., & Reinke, S.S. (2005). Genetic testing: Practical, ethical, and counselling considerations. *Mayo Clinic Proceedings*, 80(1), 63-73. doi: 10.4065/80.1.63
- Emslie, C., Hunt, K., & Watt, G. (2001). Invisible women? The importance of gender in lay beliefs about heart problems. *Sociology of Health & Illness*, 23, 203–233. doi: 10.1111/1467-9566.00248
- Emslie, C., Hunt, K., & Watt, G. (2003). A chip off the old block? Lay understandings of inheritance among men and women in mid-life. *Public Understanding of Science*, 12(1), 47-65. doi: 10.1177/0963662503012001248
- Erwin, C., Williams, J.K., Juhl, A.R., Mengeling, M., Mills, J.A., Bombard,... & the IRESPOND-HD Investigators of the Huntington Study Group (2010). Perception, Experience, Response to Genetic Discrimination in Huntington Disease: The International RESPOND-HD Study. *American Journal of Medical Genetics. Part B* 153B, 1081–1093. doi: 10.1002/ajmg.b.31079
- Esplen, M. J., Madlensky, L., Aronson, M., Rothenmund, H., Gallinger, S., Butler, K., ... & McLaughlin, J. (2007). Colorectal cancer survivors undergoing genetic testing for hereditary non - polyposis colorectal cancer: motivational factors and psychosocial

- functioning. *Clinical Genetics*, 72, 394-401. doi: 10.1111/j.1399-0004.2007.00893.x
- Esplen, M. J., Madlensky, L., Butler, K., McKinnon, W., Bapat, B., Wong, J., . . . Gallinger, S. (2001). Motivations and psychosocial impact of genetic testing for HNPCC. *American Journal of Medical Genetics Part A*, 103, 9-15. doi: 10.1002/ajmg.1493
- Esplen, M. J., Urquhart, C., Butler, K., Gallinger, S., Aronson, M., & Wong, J. (2003). The experience of loss and anticipation of distress in colorectal cancer patients undergoing genetic testing. *Journal of Psychosomatic Research*, 55, 427-435. doi: 10.1016/S0022-3999(03)00511-7
- Etchegary, H. (2005). *I have the gene, but I don't have Huntington disease: Negotiating genetic risk*. (Doctoral dissertation). Retrieved from ProQuest.
- Etchegary, H. (2006a). Genetic testing for Huntington's disease: How is the decision taken? *Genetic Testing*, 10(1), 60-67. doi: 10.1089/gte.2006.10.60
- Etchegary, H. H. (2006b). Discovering the family history of Huntington disease (HD). *Journal of Genetic Counseling*, 15(2), 105-117. doi: 10.1007/s10897-006-9018-7
- Etchegary, H. (2009). Coping with genetic risk: Living with Huntington disease (HD). *Current Psychology*, 28, 284-301. doi: 10.1007/s12144-009-9061-2
- Etchegary, H. (2010). 'I put it on the back burner most days': Living with chronic risk. *Health*, 15, 633-649. doi: 10.1177/1363459310364162
- Etchegary, H., & Fowler, K. (2008). 'They had the right to know.' Genetic risk and perceptions of responsibility. *Psychology & Health*, 23, 707-727. doi: 10.1080/14768320701235249

Etchegary, H., Miller, F., deLaat, S., Wilson, B., Carroll, J., & Cappelli, M. (2009).

Decision-making about inherited cancer risk: Exploring dimensions of genetic responsibility. *Journal of Genetic Counseling*, 18, 252-264. doi: 10.1007/s10897-009-9218-z

Etchegary, H., & Perrier, C. (2007). Information processing in the context of genetic risk:

Implications for genetic-risk communication. *Journal of Genetic Counseling*, 16, 419-432. doi: 10.1007/s10897-006-9082-z

Evers-Kiebooms, G., & Decruyenaere, M. (1998). Predictive testing for Huntington's

disease: A challenge for persons at risk and for professionals. *Patient Education and Counseling*, 35(1), 15-26. doi: 10.1016/S0738-3991(98)00086-X

Evers-Kiebooms, G., Nys, K., Harper, P., Zoetewij, M., Durr, A., Jacopini, G., . . .

Simpson, S. (2002). Predictive DNA-testing for Huntington's disease and reproductive decision making: A European collaborative study. *European Journal of Human Genetics*, 10, 167-176. doi: 10.1038/sj.ejhg.5200781

Ewald, F. (1991). Insurance and risk. In G. Burchell, C. Gordon & P. Miller (Eds.), *The*

Foucault Effect: Studies in governmentality (pp. 197-210), London: Harvester Wheatsheaf.

Farmer, P. (1992). *AIDS and accusation. Hati and the geography of blame*. London:

University of California Press.

Farnsworth, M. M., Fosyth, D., Haglund, C., & Michael, J. A. (2006). When I go in to

wake them ... I wonder: Parental perceptions about congenital long QT syndrome.

Journal of the American Academy of Nurse Practitioners, 18, 284-90. doi:

10.1111/j.1745-7599.2006.00132.x

- Farrimond, H., Paula, M. S., Qureshi, N., & Philip, H. E. (2010). Making sense of being at 'high risk' of coronary heart disease within primary prevention. *Psychology*, 25, 289-304. doi: 10.1080/08870440802499382
- Featherstone, K., Bharadwaj, A., & Clarke, A. (2005). *Risky relations : Family, kinship and the new genetics*. Oxford, GBR: Berg Publishers.
- Fenwick, A. (2010). Are guidelines for genetic testing of children necessary? *Familial Cancer*, 9, 23-25. doi: 10.1007/s10689-009-9278-0
- Finkler, K. (2001). The kin of the gene: The medicalization of family and kinship in American society. *Current Anthropology*, 4, 235-263. Retrieved from <http://www.jstor.org/stable/10.1086/320004>
- Finkler K. (2005). Family, kinship, memory and temporality in the age of the new genetics. *Social Science & Medicine*, 61:1059–1071. doi: 10.1016/j.socscimed.2005.01.002
- Flemme, I., Edvardsson, N., Hinic, H., Jinhage, B. M., Dalman, M., & Fridlund, B. (2005). Long-term quality of life and uncertainty in patients living with an implantable cardioverter defibrillator. *Heart & Lung: The Journal of Critical Care*, 34, 386-392. doi: 10.1016/j.hrtlng.2005.05.003
- Flemme, I., Hallberg, U., Johansson, I., & Stromberg, A. (2011). Uncertainty is a major concern for patients with implantable cardioverter defibrillators. *Heart & Lung: The Journal of Critical Care*, 40, 420-428. doi: 10.1016/j.hrtlng.2011.02.003
- Fontaine, G., Fontaliran, F., & Frank, R. (1998). Arrhythmogenic right ventricular cardiomyopathies. Clinical forms and main differential diagnoses. *Circulation*, 97, 1532-1535. doi: 10.1161/01.CIR.97.16.1532

- Fontaine, G., Guiraudon, G., Frank, R., Vedel, J., Grosogeat, Y., Cabrol, C., & Facquet, J. (1977). Stimulation studies and epicardial mapping in ventricular tachycardia: Study of mechanisms and selection for surgery. In H.E. Kulbertus (Ed.), *Reentrant arrhythmias* (pp. 334–350). Lancaster: MTP Publishers
- Foster, C., Eccles, R., Ardern-Jones, A., Moynihan, C., & Watson, M. (2004). Juggling roles and expectations: Dilemmas faced by women talking to relatives about cancer and genetic testing. *Psychology & Health*, 19, 439-455. doi: 10.1080/08870440410001684168
- Foster, C., Watson, M., Eccles, R., Eccles, D., Ashley, S., Davidson, R., . . . Evans, D. (2007). Predictive genetic testing for BRCA1/2 in a UK clinical cohort: Three-year follow-up. *British Journal of Cancer*, 96, 718-724. doi: 10.1038/sj.bjc.6603610
- Foucault, M. (1977). *Discipline and punishment: The birth of the prison*. Great Britain: Penguin Books Ltd.
- Foucault, M. (1991). Governmentality. In G. Burchell, C. Gordon, & P. Miller (Eds.), *The Foucault effect: Studies in governmentality* (pp. 87-104). Chicago: The University of Chicago Press.
- Fox, N.J. (1999). Post modern reflections on 'risk', 'hazards' and life choices. In D. Lupton (Ed.), *Risk and soiciocultural theory* (pp. 12-33). New York: Cambridge University Press.
- French, D., Senior, V., Weinman, J., & Marteau, T. (2007) Causal attributions for heart disease: A systematic review. *Psychology and Health*, 11(1), 77-98. doi: 10.1080/08870440108405491

- Frich, J. C., Ose, L., Malterud, K., & Fugelli, P. (2006). Perceived vulnerability to heart disease in patients with familial hypercholesterolemia: A qualitative interview study. *Annals of Family Medicine*, 4(3), 198-204. doi: 10.1370/afm.529
- Friedmann, E. E., Thomas, S.A., Inguito, P., Kao, C.W., Metcalf, M., Kelley, F.J., & Gottlieb, S. S. (2006). Quality of life and psychological status of patients with implantable cardioverter defibrillators. *Journal of Interventional Cardiac Electrophysiology*, 17(1), 65-72. doi: 10.1007/s10840-006-9053-1
- Frost, C. J, Venne V, Cunningham, D., & Gerritsen-McKane, R. (2004). Decision making with uncertain information: Learning from women in a high risk breast cancer clinic. *Journal of Genetic Counselling*, 13, 221-236. Retrieved from: <http://www.ncbi.nlm.nih.gov/pubmed/15604633>
- Gaff, C. L., Clarke, A. J., Atkinson, P., Sivell, S., Elwyn, G., Iredale, R., . . . Edwards, A. (2007). Process and outcome in communication of genetic information within families: A systematic review. *European Journal of Human Genetics*, 15, 999-1011. doi: 10.1038/sj.ejhg.5201883
- Gargiulo, M., Lejeune, S., Tanguy M-L., Khadija, L-L., Faudet, A., Cohen, D., ... Durr, A. (2009). Long-term outcome of presymptomatic testing in Huntington disease. *European Journal of Human Genetics*, 17, 165-71. doi: 10.1038/ejhg.2008.146
- Genome Canada. (2009). From lab to clinic: Implementing discoveries of single-gene disorders in Atlantic Canada. Impact. *Genomics & Society*, No 2. Retrieved from <http://www.genomecanada.ca/en/ge3ls/newsletters/fall-2009/feature-stories.aspx#story-1>

- Gerull, B., Heuser, A., Wichter, T., Paul, M., Basson, C.T., McDermott, D.A., Lerman, B.B., Markowitz, S.M., ... Thierfelder, L. (2004). Mutations in the desmosomal protein plakophilin-2 are common in arrhythmogenic right ventricular cardiomyopathy. *Nature Genetics*, 36, 1162–1164. doi: 10.1038/ng1461
- Giddens, A. (1991) *Modernity and self-identity. Self and society in the late modern Age*. Cambridge, UK: Polity Press.
- Gifford, S.M. (1986). The meaning of lumps: A case study of the ambiguities of risk. In C.R. Janes, R. Stall, & S.M Gifford (Eds.), *Anthropology and epidemiology: Interdisciplinary approaches to the study of health and disease* (pp. 213-246). New York: D. Reidel Publishing Co.
- Glaser, B. G. (1978). *Theoretical sensitivity*. CA: Sociology Press.
- Glaser, B.G. (1992). *Basics of grounded theory analysis*. Mill Valley: Sociological Press.
- Glaser, B. G. (2004). Naturalist inquiry and grounded theory. *Forum: Qualitative Social Research*, 5(1). Retrieved from <http://www.qualitative-research.net/index.php/fqs/article/view/652/1413>
- Glaser, B. G., & Strauss, A. L. (1967). *The discovery of grounded theory: Strategies for qualitative research*. New York: Aldine.
- Goldman, R. E., Parker, D., Eaton, C., Borkan, J. M., Gramling, R., Coverm, R.T., & Ahem, D.K. (2006). Patients' perceptions of cholesterol, cardiovascular disease risk, and risk communication strategies. *Annals of Family Medicine*, 4, 205-212. doi: 10.1370/afm.534
- Gollob, M. H., Blier, L., Brugada, R., Champagne, J., Chauhan, V., Connors, S., . . . Woo, A. (2011). Recommendations for the use of genetic testing in the clinical evaluation

- of inherited cardiac arrhythmias associated with sudden cardiac death: Canadian cardiovascular Society/Canadian heart rhythm society joint position paper. *Canadian Journal of Cardiology*, 27, 232-245. doi: 10.1016/j.cjca.2010.12.078
- Green, J., & Thorogood, N. (2009). *Qualitative methods for health research*, (2nd ed.). London: SAGE Publications.
- Gritz, E. R., Peterson, S. K., Vernon, S. W., Marani, S. K., Baile, W. F., Watts, B. G., . . . Lynch, P. M. (2005). Psychological impact of genetic testing for hereditary nonpolyposis colorectal cancer. *Journal of Clinical Oncology*, 23, 1902-1910. doi: 10.1200/JCO.2005.07.102
- Grover, S., Stoffel, E. M., Mercado, R. C., Ford, B. M., Kohlman, W. K., Conrade, S. P. G., . . . Syngal, S. (2009). Colorectal cancer risk perception on the basis of genetic test results in individuals at risk for lynch syndrome. *Journal of Clinical Oncology : Official Journal of the American Society of Clinical Oncology*, 27, 3981-3986. doi: 10.1200/JCO.2008.18.6940
- Guiraudon, G. M., Klein, G. J., Gulamhusein, S. S., Painvin, G. A., Campo, C. D., Gonzales, J. C., & Ko, P. K. (1983). Total dissection of the right ventricular free wall: Surgical treatment of right ventricular tachycardia associated with right ventricular dysplasia. *Circulation*, 67, 463-470. doi: 10.1161/01.CIR.67.2.463
- Haddow, G. (2009). "We only did it because he asked us": Gendered accounts of participation in a population genetic data collection. *Social Science*, 69, 1010-1017. doi: 10.1016/j.socscimed.2009.07.028
- Hadley, D. W., Ashida, S., Jenkins, J. F., Martin, J. C., Calzone, K. A., Kuhn, N. R., . . . Koehly, L. M. (2010). Generation after generation: Exploring the psychological

- impact of providing genetic services through a cascading approach. *Genetics in Medicine*, 12, 808-815. doi: 10.1097/GIM.0b013e3181f69dbb
- Halbert, C. H., Lynch, H., Lynch, J., Main, D., Kucharski, S., & Anil, K. R. (2004). Colon cancer screening practices following genetic testing for hereditary nonpolyposis colon cancer (HNPCC) mutations. *Archives of Internal Medicine*, 164, 1881-1887. doi: 10.1001/archinte.164.17.1881.
- Hall, R., Saukko, P. M., Evans, P. H., Qureshi, N., & Humphries, S. E. (2007). Assessing family history of heart disease in primary care consultations: a qualitative study. *Family Practice*, 24, 435-442. doi: 10.1093/fampra/cmm037
- Hallas, C. N., Burke, J. L., White, D. G., & Connelly, D. T. (2010). Pre-ICD illness beliefs affect postimplant perceptions of control and patient quality of life. *Pacing and Clinical Electrophysiology : PACE*, 33, 256-265. doi: 10.1111/j.1540-8159.2009.02641.x
- Hallowell, N. (1999). Doing the right thing: Genetic risk and responsibility. *Sociology of Health*, 21, 597-621. doi:10.1111/1467-9566.00175
- Hallowell, N., Arden-Jones, A., Eeles, R., Foster, C., Lucassen, A., & Moynihan, C. (2006). Guilt, blame and responsibility: Men's understanding of their role in the transmission of BRCA1/2 mutations within their family. *Sociology of Health*, 28, 969-988. doi: 10.1111/j.1467-9566.2006.00515.x
- Hallowell, N., Foster, C., Arden-Jones, A., Eeles, R., Murday, V., & Watson, M. (2002). Genetic testing for women previously diagnosed with breast/ovarian cancer: Examining the impact of BRCA1 and BRCA2 mutation searching. *Genetic Testing*, 6(2), 79-87. doi: 10.1089/10906570260199320

- Hansen, J., Holm, L., Frewer, L., Robinson, P., & Sandoe, P. (2003). Beyond the knowledge deficit: recent research into lay and expert attitudes to food risks. *Appetite*, 41(2), 111-121. doi: 10.1016/S0195-6663(03)00079-5
- Hazelton, A. G., Sears, S. F., Kirian, K., Matchett, M., & Shea, J. (2009). Cardiology patient page. coping with my partner's ICD and cardiac disease. *Circulation*, 120(10), e73-e76. doi: 10.1161/CIRCULATIONAHA.109.877613
- Hendriks, K. S. W. H., Grosfeld, F. J. M., van Tintelen, J. P., van Langen, I. M., Wilde, A. A. M., van den Bout, J., & ten Kroode, H. F. J. (2005). Can parents adjust to the idea that their child is at risk for a sudden death? Psychological impact of risk for long QT syndrome. *American Journal of Medical Genetics. Part A*, 138A(2), 107-112. doi: 10.1002/ajmg.a.30861
- Hendriks, K. S., Hendriks, M. M., Birnie, E., Grosfeld, F. J., Wilde, A. A., van den Bout, J., . . . van Langen, I. M. (2008). Familial disease with a risk of sudden death: A longitudinal study of the psychological consequences of predictive testing for long QT syndrome. *Heart Rhythm*, 5, 719-724. doi: 10.1016/j.hrthm.2008.01.032
- Heshka, J. T., Palleschi, C., Howley, H., Wilson, B., & Wells, P. S. (2008). A systematic review of perceived risks, psychological and behavioural impacts of genetic testing. *Genetics in Medicine*, 10(1), 19-32. doi: 10.1097/GIM.0b013e31815f524f
- Himmelstein, M. (2010). Genetic testing and risk interpretation: How do women understand lifetime risk results? *Judgment and Decision Making*, 5, 116-123. Retrieved from <http://journal.sjdm.org/10/91110/jdm91110.html>
- Ho, S. M., Ho, J. W., Chan, C. L., Kwan, K., & Tsui, Y. K. (2003). Decisional consideration of hereditary colon cancer genetic test results among Hong Kong

- Chinese adults. *Cancer Epidemiology Biomarkers & Prevention*, 12, 426-432.
- Retrieved from <http://cebp.aacrjournals.org/content/12/5/426.full.pdf+html>
- Hogben, S., Boddington, P. (2005) Policy recommendations for carrier testing and predictive testing in childhood: A distinction that makes a real difference. *Journal of Genetic Counseling*, 14, 271-281. doi: 10.1007/s10897-005-4840-x
- Hodge Jr., J. G. (2004). Ethical issues concerning genetic testing and screening in public health. Paper presented at the *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*, 125(1), 66-70. doi: 10.1002/ajmg.c.30005
- Hodgkinson, K.A. (2007). [ARVC Statistics in Newfoundland and Labrador]. Unpublished raw data.
- Hodgkinson, K., Connors, S. P., Merner, N., Haywood, A., Young, T-L., McKenna, W.J., ... Parfrey, P.S. (2012). The natural history of a genetic subtype of arrhythmogenic right ventricular cardiomyopathy caused by a p.S358L mutation in TMEM43. *Clinical Genetics*, 1-11. doi:10.1111/j.1399-0004.2012.01919.x
- Hodgkinson, K., Dicks, E., Connors, S., Young, T-L., Parfrey, P., & Pullman, D. (2009). Translation of research discoveries to clinical care in arrhythmogenic right ventricular cardiomyopathy in Newfoundland and Labrador: Lessons for health policy in genetic disease. *Genetics in Medicine*, 11, 859-865. doi: 10.1097/GIM.0b013e3181c20bb3
- Hodgkinson, K., Parfrey, P. S., Bassett, A. S., Kupprion, C., Drenckhahn, J., Norman, M. W., ... Connors, S. P. (2005). The impact of implantable cardioverter-defibrillator therapy on survival in autosomal-dominant arrhythmogenic right ventricular

- cardiomyopathy (ARVD5). *Journal of the American College of Cardiology*, 45, 400-408. doi: 10.1016/j.jacc.2004.08.068
- Holt, K. (2006). What do we tell the children? Contrasting the disclosure choices of two HD families regarding risk status and predictive genetic testing. *Journal of Genetic Counseling*, 15, 253-265. doi: 10.1007/s10897-006-9021-z
- Hunt, K., Davison, C., Emslie, C., & Ford, G. (2000). Are perceptions of a family history of heart disease related to health-related attitudes and behaviour? *Health Education Research*, 15, 131-143. doi: 10.1093/her/15.2.131
- Hunt, K., Emslie, C., & Watt, G. (2001). Lay constructions of a family history of heart disease: Potential for misunderstandings in the clinical encounter? *Lancet*, 357(9263), 1168-1171. doi: org/10.1016/S0140-6736(00)04334-8
- Hutchinson, S. (1986). Creating meaning. A grounded theory of NICU nurses. In W.C. Chenitz & J.M. Swanson (Eds.), *From practice to grounded theory: Qualitative research in nursing* (pp. 191-204). Menlo-Park, CA: Addison Wesley.
- Jallinoja, P., Hakonen, A., Aro, A., Niemelä, P., Hietala, M., Lönnqvist, J., . . . Aula, P. (1998). Attitudes towards genetic testing: Analysis of contradictions. *Social Science & Medicine*, 46, 1367-1374. doi: 10.1016/S0277-9536(98)00017-3
- Jeon, Y. H. (2004). The application of grounded theory and symbolic interactionism. *Scandinavian Journal of Caring Sciences*, 18, 249-256. doi 10.1111/j.1471-6712.2004.00287.x
- Joffe, H. (2003). Risk: From perception to social representation. *British Journal of Social Psychology*, 42, 55-73. doi: 10.1348/014466603763276126

- Johnson, P. D. (1981). *Sociological theory: Classical founders and contemporary perspectives*. New York: Wiley.
- Johnson, E. J., & Tversky, A. (1983). Affect, generalization, and the perception of risk. *Journal of Personality and Social Psychology*, 45(1), 20-31. doi: 10.1037/0022-3514.45.1.20
- Johnson, K. A., Trimbath, J. D., Petersen, G. M., Griffin, C. A., & Giardiello, F. M. (2002). Impact of genetic counseling and testing on colorectal cancer screening behaviour. *Genetic Testing*, 6, 303-306. doi: 10.1089/10906570260471831
- Kamphius, H. C., Verhoever, N. W., Leeuw, R., Derksen, R., Hauer, R. N., & Winnubst, J. A. (2004). ICD: A qualitative study of patient experience the first year after implantation. *Journal of Clinical Nursing*, 13, 1000-1016. doi: 10.1111/j.1365-2702.2004.01021.x
- Kantor, P. B., Bullinger, J. M., & Gal, C. S. (2012). Patient decision-making modes and causes: A preliminary investigation. *Journal of the American Society for Information Science and Technology*. 63, 1332-1349. doi: 10.1002/asi.22667
- Kaphingst, K. A., & McBride, C. M. (2010). Patient responses to genetic information: Studies of patients with hereditary cancer syndromes identify issues for use of genetic testing in nephrology practice. *Seminars in Nephrology*, 30, 203-14. doi: 10.1016/j.semnephrol.2010.01.011
- Katapodi, M. C., Lee, K. A., Facione, N. C., & Dodd, M. J. (2004). Predictors of perceived breast cancer risk and the relation between perceived risk and breast cancer screening: A meta-analytic review. *Preventive Medicine*, 38, 388-402. doi: 10.1016/j.ypmed.2003.11.012

- Kavanagh, A. M., & Broom, D. H. (1998). Embodied risk: My body, myself? *Social Science & Medicine*, 46, 437-444. doi: 10.1016/S0277-9536(97)00188-3
- Kelle, U. (2007). "Emergence" vs. "Forcing" of Empirical Data? A Crucial Problem of "Grounded Theory" Reconsidered. *Historical Social Research/Historische Sozialforschung. Supplement*, 19, 133-156. Retrieved from <http://www.jstor.org/stable/40981074>
- Kenen, R., Arden-Jones, A. & Eeles, R. (2003), Family stories and the use of heuristics: women from suspected hereditary breast and ovarian cancer (HBOC) families. *Sociology of Health & Illness*, 25, 838-865. doi: 10.1046/j.1467-9566.2003.00372.x
- Kinney, A. Y., Lindsey, E. B., Mandal, D., Sara, E. S., Bonnie, J. B., & Holubkov, R. (2005). The impact of receiving genetic test results on general and cancer-specific psychological distress among members of an African-American kindred with a BRCA1 mutation. *Cancer*, 104, 2508-2516. doi: 10.1002/cncr.21479
- Klitzman, R., Thorne, D., Williamson, J., Chung, W., & Marder, K. (2007). Disclosures of Huntington disease risk within families: Patterns of decision-making and implications. *American Journal of Medical Genetics. Part A*, 143A, 1835-1849. doi: 10.1002/ajmg.a.31864
- Klitzman, R., Thorne, D., Williamson, J., & Marder, K. (2007). The roles of family members, health care workers, and others in decision-making processes about genetic testing among individuals at risk for Huntington disease. *Genetics in Medicine*, 9, 358-371. doi: 10.1097/GIM.0b013e3180653c5a
- Krimsky, S., & Golding, D. (1992). *Social theories of risk*. Westport, CT: Praeger Publishers.

Landsbergen, K. M., Prins, J., Brummer, H.G., Kraaimaat, F.W., & Hoogerbrugge, N.

(2009). Genetic testing for Lynch syndrome in the first year of colorectal cancer: A review of the psychological impact. *Familial Cancer*, 8, 325-337. doi:

10.1007/s10689-009-9239-7

Landsbergen, K. M., Prins, J., Brummer, H. G., Kraaimaat, F. W., & Hoogerbrugge, N.

(2011). Shortened time interval between colorectal cancer diagnosis and risk testing for hereditary colorectal cancer is not related to higher psychological distress.

Familial Cancer, 10(1), 51-57. doi: 10.1007/s10689-010-9387-9

Landsbergen, K., Verhaak, C., Kraaimaat, F., & Hoogerbrugge, N. (2005). Genetic uptake in BRCA-mutation families is related to emotional and behavioural communication characteristics of index patients. *Familial Cancer*, 4(2), 115-119. doi:

10.1007/s10689-004-7991-2

Lanie, A. D., Jayaratne, T. E., Sheldon, J. P., Kardia, S. L. R., Anderson, E. S., Feldbaum, M., & Petty, E. M. (2004). Exploring the public understanding of basic genetic concepts. *Journal of Genetic Counseling*, 13, 305-320. doi:

10.1023/B:JOGC.0000035524.66944.6d

Lemon, J., Edelman, S., & Kirkness, A. (2004). Avoidance behaviours in patients with implantable cardioverter defibrillators. *Heart & Lung: The Journal of Acute and Critical Care*, 33, 176-182. doi: org/10.1016/j.hrtlng.2004.02.005.

Lempert, L.B. (2007). Asking questions of the data: Memo writing in the

grounded theory tradition. In A. Bryant & K. Charmaz (Eds.), *The SAGE*

Handbook of Grounded Theory (pp. 245-264). London: Sage Publications.

- Lerman, C., & Croyle, R. T. (1996). Emotional and behavioural responses to genetic testing for susceptibility to cancer. *Oncology-Huntington*, 10, 191-199. Retrieved from:
<https://member.ubmmedica.com/index.php?referrer=http://member.ubmmedica.com/cga.php?assetID=425&referrer=http://www.psychiatrictimes.com/display/article/10165/74358>
- Lerman, C., Croyle, R. T., Tercyak, K. P., & Hamann, H. (2002). Genetic testing: Psychological aspects and implications. *Journal of Consulting and Clinical Psychology*, 70, 784-797. doi: 10.1037//0022-006X.70.3.784
- Lerman, C., Daly, M., Masny, A., & Balshem, A. (1994). Attitudes about genetic testing for breast-ovarian cancer susceptibility. *Journal of Clinical Oncology*, 12, 843-850. Retrieved from <http://jco.ascopubs.org/content/12/4/843.short>
- Lerman, C., Hughes, C., Croyle, R.T., Main, D., Durham, C., Synder, C., . . . & Lynch, H.T. (2000). Prophylactic surgery decisions and surveillance practices one year following BRCA1/2 testing. *Preventive Medicine*, 31(1), 75-80. doi:.org/10.1006/pmed.2000.0684
- Lerman, C., Hughes, C., Lemon, S., Main, D., Snyder, C., Durham, C., . . . Lynch, H. (1998). What you don't know can hurt you: Adverse psychological effects in members of BRCA1-linked and BRCA2-linked families who decline genetic testing. *Journal of Clinical Oncology*, 16, 1650-1654. Retrieved from <http://jco.ascopubs.org/content/16/5/1650.short>
- Lerman, C., Lustbader, E., Rimer, B., Daly, M., Miller, S., Sands, C., & Balshem, A. (1995). Effects of individualized breast cancer risk counseling: A randomized trial.

- JNCI Journal of the National Cancer Institute*, 87, 286-292. doi:
10.1093/jnci/87.4.286
- Lerman, C., & Shields, A. E. (2004). Genetic testing for cancer susceptibility: The promise and the pitfalls. *Nature Reviews Cancer*, 4, 235-241. doi: 10.1038/nrc1301
- Lincoln, Y.L., & Guba, E.G. (2000). Paradigmatic controversies, contradictions, and emerging confluences. In N. K. Denzin, & Y.S. Lincoln, (Eds.), *Handbook of qualitative research* (2nd ed.), (pp 163- 188). Sage Publications: California.
- Lippman, A. (1994). The genetic construction of prenatal testing. In K. Rothenberg, & E. Thompson. (Eds.), *Women and prenatal testing: Facing the challenges of technology* (pp. 9-12). Ohio: Ohio State University Press.
- Lock, M., & Nguyen, V-K. (2010). *An anthropology of biomedicine*. UK: Wiley-Blackwell.
- Lodder, L., Frets, P. G., Trijsburg, R. W., Meijers-Heijboer, E. J., Klijn, J. G.M., Duivenvoorden, ... Niermeijer, M. F. (2001). Psychological impact of receiving a *BRCA1/BRCA2* test result. *American Journal of Medical Genetics*, 98, 15-24. doi: 10.1002/1096-8628(20010101)98:1<15::AID-AJMG1014>3.0.CO;2-0
- Lupton, D. (1993). Risk as moral danger: The social and political functions of risk discourse in public health. *International Journal of Health Services*, 23, 425-435. doi: 10.2190/16AY-E2GC-DFLD-51X2
- Lupton, D. (1995). *The imperative of health: Public health and the regulated body*. London: Sage Publications Ltd.
- Lupton, D. (1999). *Risk and sociocultural theory: New directions and perspectives*. New York: Cambridge University Press.

- Lupton, D. (2006). Sociology and risk. In G. Mythen & S. Walklate (Eds.), *Beyond the risk society* (pp. 11-24). England: Open University Press.
- Lupton, D., & Tulloch, J. (2002). 'Life would be pretty dull without risk': Voluntary risk-taking and its pleasures. *Health, Risk & Society*, 4, 113-124. doi: 10.1080/13698570220137015
- Lynch, H. T., Snyder, C., Lynch, J. F., Karatoprakli, P., Trowonou, A., Metcalfe, K., . . . Gong, G. (2006). Patient responses to the disclosure of BRCA mutation tests in hereditary breast-ovarian cancer families. *Cancer Genetics and Cytogenetics*, 165, 91-97. doi: 10.1016/j.cancergencyto.2005.07.011
- MacDonald, M (2001). Finding a critical perspective in grounded theory. In R.S. Schreiber & P.N. Stern (Eds.), *Using grounded theory in nursing* (pp. 113-157). New York: Spring Publishing Company.
- Macdonald, K. G., Doan, B., Kelner, M., & Taylor. (1996). A sociobehavioural perspective on genetic testing and counselling for heritable breast, ovarian and colon cancer. *CMAJ : Canadian Medical Association Journal*, 154, 457-64. doi: <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1487608/pdf/cmaj00088-0027.pdf>
- Madlensky, L., Espen, M. J., Gallinger, S., McLaughlin, J. R., & Goel, V. (2003). Relatives of colorectal cancer patients: Factors associated with screening behaviour. *American Journal of Preventive Medicine*, 25, 187-194. doi: 10.1016/S0749-3797(03)00202-2
- Manuel, A., Brunger, F., & Hodgkinson, K. (2010). Psychological implications of genetic investigations. In R. Brugada (Ed.), *Clinical Approaches to Sudden Cardiac Death Syndromes* (pp. 311-316). New York: Springer.

- Marcus, F. I., Fontaine, G. H., Guiraudon, G., Frank, R., Laurenceau, J. L., Malergue, C., & Grosogeat, Y. (1982). Right ventricular dysplasia: A report of 24 cases. *Circulation*, 65, 384-398. doi: 10.1161/01.CIR.65.2.384
- Marcus, F. F. I., McKenna, W. J., Sherill, D., Basso, C., Bauce, M., Bluemke, ..., Zareba, W. (2010). Diagnosis of arrhythmogenic right ventricular cardiomyopathy/dysplasia: Proposed modification of the task force criteria. *European Heart Journal*, 31, 806-814. doi: 10.1093/eurheartj/ehq025
- Mårtensson, J., Karlsson, J. E., & Fridlund, B. (1998). Female patients with congestive heart failure: how they conceive their life situation. *Journal of Advanced Nursing*, 28, 1216-1224. doi: 10.1046/j.1365-2648.1998.00827.x
- Marshall, W.H., Furey, M., Larsen, B., Rose, J., Sharratt, G.P., Sussex, B., & Virmani, S. (1988). Right ventricular cardiomyopathy and sudden death in young people. *The New England Journal of Medicine*, 319, 174-175. doi: 10.1056/NEJM198807213190312
- Marteau, T. M., & Croyle, R. T. (1998). The new genetics: Psychological responses to genetic testing. *British Medical Journal*, 316, 693. Retrieved from <http://www.jstor.org/stable/25178420>
- Marteau, T. M., & Lerman, C. (2001). Genetic risk and behavioural change. *British Medical Journal*, 322, 1056-1059. Retrieved from <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1120191/pdf/1056.pdf>
- Marteau, T., Kinmonth, A., Pyke, S., & Thompson, S. (1995). Readiness for lifestyle advice: Self-assessments of coronary risk prior to screening in the British family heart study. Family heart study group. *The British Journal of General Practice*, 45,

5-8. Retrieved from

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1239105/pdf/brjgenprac00024-0009.pdf>

- Marteau, T., Senior, V., Humphries, S. E., Bobrow, M., Cranston, T., Crook, M. A., Day, L., ...Wray, R. (2004), Psychological impact of genetic testing for familial hypercholesterolemia within a previously aware population: A randomized controlled trial. *American Journal of Medical Genetics*, 128A: 285–293. doi: 10.1002/ajmg.a.30102
- McAllister, M. (2002). Predictive genetic testing and beyond: A theory of engagement. *Journal of Health Psychology*, 7, 491-505. doi: 10.1177/1359105302007005628
- McAllister, M. (2003). Personal theories of inheritance, coping strategies, risk perception and engagement in hereditary non-polyposis colon cancer families offered genetic testing. *Clinical Genetics*, 64, 179-189. doi: 10.1034/j.1399-0004.2003.00133.x
- McAllister, M., Davies, L., Payne, K., Nicholls, S., Donnai, D., & MacLeod, R. (2007). The emotional effects of genetic diseases: Implications for clinical genetics. *American Journal of Medical Genetics. Part A*, 143A, 2651-2661. doi: 10.1002/ajmg.a.32013
- McAllister, M., Payne, K., Nicholls, S., MacLeod, R., Donnai, D., & Davies, L. M. (2007). Improving service evaluation in clinical genetics: Identifying effects of genetic diseases on individuals and families. *Journal of Genetic Counseling*, 16(1), 71-83. doi: 10.1007/s10897-006-9046-3
- Mead, G.H. (1934). *Mind, Self, and Society*. Chicago: University of Chicago Press.

- Meiser, B. (2005). Psychological impact of genetic testing for cancer susceptibility: An update of the literature. *Psycho-Oncology*, 14, 1060-1074. doi: 10.1002/pon.933
- Meiser, B., Butow, P., Friedlander, M., Barratt, A., Schnieden, V., Watson, M., . . . Tucker, K. (2002). Psychological impact of genetic testing in women from high-risk breast cancer families. *European Journal of Cancer*, 38, 2025-2031. doi: 10.1016/S0959-8049(02)00264-2
- Meiser, B., Collins, V., Warren, R., Gaff, C., St John, D. J. B., Young, M. A., . . . Halliday, J. (2004). Psychological impact of genetic testing for hereditary nonpolyposis colorectal cancer (HNPCC). *Clinical Genetics*, 66, 502-511. doi: 10.1111/j.1399-0004.2004.00339.x
- Meiser, B., & Dunn, S. (2000). Psychological impact of genetic testing for Huntington's disease: An update of the literature. *Journal of Neurology, Neurosurgery & Psychiatry*, 69, 574. doi: 10.1136/jnnp.69.5.574-578.
- Meiser, B., Gaff, C., Julian-Reynier, C., Biesecker, B. B., Esplen, M. J., Vodermaier, A., & Tibben, A. (2007). International perspectives on genetic counseling and testing for breast cancer risk. *Breast Disease*, 27(1), 109-125. Retrieved from <http://iospress.metapress.com/content/y5465m7627n585x6/>
- Meiser, B., & Halliday, J. L. (2002). What is the impact of genetic counselling in women at increased risk of developing hereditary breast cancer? A meta-analytic review. *Social Science & Medicine*, 54, 1463-1470. doi: 10.1016/S0277-9536(01)00133-2
- Merner, N. D., Hodgkinson, K. A., Haywood, A. F. M., Connors, S., French, V. M., Drenckhahn, J. D., . . . Young, T.L (2008). Arrhythmogenic right ventricular cardiomyopathy type 5 is a fully penetrant, lethal arrhythmic disorder caused by a

missense mutation in the TMEM43 gene. *The American Journal of Human Genetics*, 82, 809-821. doi: 10.1016/j.ajhg.2008.01.010

Michie, S., Bobrow, M., & Marteau, T. M. (2001). Predictive genetic testing in children and adults: A study of emotional impact. *Journal of Medical Genetics*, 38, 519-526.

Retrieved from

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1734924/pdf/v038p00519.pdf>

Michie, S., French, D. P., & Marteau, T. M. (2002). Predictive genetic testing: Mediators and moderators of anxiety. *International Journal of Behavioural Medicine*, 9, 309-

321. Retrieved from [http://web.ebscohost.com/ehost/detail?sid=c8d607d3-1acf-](http://web.ebscohost.com/ehost/detail?sid=c8d607d3-1acf-4921-b747-75fd3d41eac0%40sessionmgr115&vid=1&hid=104&bdata=JnNpdGU9ZWwhvc3QtbGl2ZSZzY29wZT1zaXRl#db=s3h&AN=8554580)

4921-b747-

75fd3d41eac0%40sessionmgr115&vid=1&hid=104&bdata=JnNpdGU9ZWwhvc3Qtb

Gl2ZSZzY29wZT1zaXRl#db=s3h&AN=8554580

Michie, S., MacDonald, V., & Marteau, T.M. (1996). Understanding responses to predictive genetic testing: A grounded theory approach. *Psychology and Health*, 11, 455-470.

doi: 10.1080/08870449608401982

Michie, S., Weinman, J., Miller, J., Collins, V., Halliday, J., & Theresa, M. M. (2002).

Predictive genetic testing: High risk expectations in the face of low risk information.

Journal of Behavioural Medicine, 25(1), 33-50. Retrieved from

<http://link.springer.com/article/10.1023%2FA%3A1013537701374?LI=true#page-1>

Mireskandari, S., Sangster, J., Meiser, B., Thewes, B., Groombridge, C., & Spigelman, A.

(2009). Psychosocial impact of familial adenomatous polyposis on young adults: A qualitative study. *Journal of Genetic Counseling*, 18, 409-417. doi: 10.1007/s10897-

009-9231-2

- Modell, S. M., & Lehman, M. H. (2006). The long QT syndrome family of cardiac ion channelopathies: a HuGE review. *Genetics in Medicine*, 8:143-155. Retrieved from <http://www.ncbi.nlm.nih.gov/pubmed/16540748>
- Morken, I. M., Severinsson, E., & Karlsen, B. (2009). Reconstructing unpredictability: Experiences of living with an implantable cardioverter defibrillator over time. *Journal of Clinical Nursing*, 19, 537-546. doi: 10.1111/j.1365-2702.2009.02898
- Morse, J. M. (1994). Designing funded qualitative research. In N. K. Denzin & Y. S. Lincoln (Eds.), *Handbook of qualitative research* (pp. 220-235). Thousand Oaks, CA: Sage.
- Morse, J. M. (2001). Situating grounded theory within qualitative inquiry. In R. S. Schreiber & P. N. Stern (Eds.), *Using Grounded Theory in Nursing* (pp. 1-15). New York: Spring Publishing Company.
- Morse, J. M., & Field P. A. (1995) *Qualitative research methods for health professionals* (2nd ed.). California: Sage.
- Murakami, Y., Okamura, H., Sugano, K., Yoshida, T., Kazuma, K., Akechi, T., & Uchitomi, Y. (2004). Psychologic distress after disclosure of genetic test results regarding hereditary nonpolyposis colorectal carcinoma. *Cancer*, 101, 395-403. doi: 10.1002/cncr.20363
- Murray, S. A., Manktelow, K., & Clifford, C. (2000). The interplay between social and cultural context and perceptions of cardiovascular disease. *Journal of Advanced Nursing*, 32, 1224-1233. doi: 10.1046/j.1365-2648.2000.01593.x
- Molak, V. (1997). *Fundamentals of risk analysis and risk management*. New York: Lewis Publishers.

Nadesan, M. H. (2008). *Governmentality, biopower, and everyday life*. New York:

Routledge.

National Library of Medicine. (2012). *Where is the TMEM43 gene located?* Retrieved from

<http://ghr.nlm.nih.gov/gene/TMEM43>

Nava, A., Baucé, B., Basso, C., Muriago, M., Rampazzo, A., Villanova, C., ...Thiene, G.

(2000). Clinical profile and long-term follow up of 37 families with Arrhythmogenic right ventricular cardiomyopathy. *American College of Cardiology*, 36, 2226-2233.

doi: 10.1016/S0735-1097(00)00997-9

Nava, A., Thiene, G., Cancian, B., Scognamiglio, R., Daliento, L Buja, G., ...Fasoli, G.

(1988). Familial occurrence of right ventricle dysplasia: a study involving nine families. *The Journal of the American College of Cardiology*, 12, 1222-1228. doi:

10.1016/0735-1097(88)92603-4

Nelkin, D. (1992). The rhetoric of scientific revolution. *The Hastings Center Report*, 22(4),

38-39.

Newall, E. G., Lever, N. A., Prasad, S., Hornabroo, C., & Larsen, P. D. (2006).

Psychological implications of ICD implantation in a New Zealand population.

Europace, 9(1), 20-24. doi: 10.1093/europace/eul142

Nordgren, L., Asp, M., & Fagerberg, I. (2007). Living with moderate-severe chronic heart

failure as a middle-aged person. *Qualitative Health Research*, 17(1), 4-13. doi:

10.1177/1049732306296387

Norris, J., Spelic, S., Snyder, C., & Tinley, S. (2009). Five families living with hereditary

breast and ovarian cancer risk. *Clinical Journal of Oncology Nursing*, 13(1), 73-80.

doi: 10.1188/09.CJON.73-80

- O'Malley, P. (2004). *Risk, uncertainty and government*. London: Glashouse Press.
- Ozanne, E. M., Wittenberg, E., Garber, J.E., & Weeks, J.C. (2010). Breast cancer prevention: Patient decision making and risk communication in the high risk setting. *The Breast Journal*, 16(1), 38-47. doi: 10.1111/j.1524-4741.2009.00857.x
- Palacios-Cena, D., Losa-Iglesias, M. E., Alvarez-Lopez, C., Cachon-Perez, M., Reyes, R. A., Salvadores-Fuentes, P., & Fernandez-de-Las-Penas, C. (2011). Patients, intimate partners and family experiences of implantable cardioverter defibrillators: Qualitative systematic review. *Journal of Advanced Nursing*, 67, 2537-2550. doi: 10.1111/j.1365-2648.2011.05694.x
- Parsons, E. & Atkinson, P. (1992) Lay constructions of genetic risk, *Sociology of Health and Illness*, 14, 437-55. doi: 10.1111/j.1467-9566.1992.tb00131.x
- Peters, K. F., Kong, F., Hanslo, M., & B, B. B. (2002). Living with marfan syndrome III. quality of life and reproductive planning. *Clinical Genetics*, 62(2), 110-120. doi: 10.1034/j.1399-0004.2002.620203.x
- Peterson, E. A., Milliron, K. J., Lewis, K. E., Goold, S. D., & Merajver, S. D. (2002). Health insurance and discrimination concerns and BRCA1/2 testing in a clinic population. *Cancer Epidemiology, Biomarkers & Prevention: A Publication of the American Association for Cancer Research, Cosponsored by the American Society of Preventive Oncology*, 11(1), 79-87. Retrieved from <http://cebp.aacrjournals.org/content/11/1/79.short>
- Pierce, C.S. (1878). How to Make Our Ideas Clear. *Popular Science Monthly*, 12, 286-302. Retrieved from <http://www.peirce.org/writings/pl19.html>

- Pihl, E., Fridlund, B., & Jan Mårtensson. (2010). Spouses' experiences of impact on daily life regarding physical limitations in the loved one with heart failure: A phenomenographic analysis. *Canadian Journal of Cardiovascular Nursing*, 20(3), 9-17. Retrieved from <http://www.ncbi.nlm.nih.gov/pubmed/20718235>
- Pilarski, R. (2009). Risk perception among women at risk for hereditary breast and ovarian cancer. *Journal of Genetic Counseling*, 18, 303-312. doi: 10.1007/s10897-009-9227-y
- Ponder, M., Lee, J., Green, J., & Richards, M. (1996). Family history and perceived vulnerability to some common diseases: A study of young people and their parents. *Journal of Medical Genetics*, 33, 485-92. doi: 10.1136/jmg.33.6.485
- Ponz, d. L., Benatti, P., Gregorio, C. D., Pedroni, M., Losi, L., & Genuardi, M. (2004). Genetic testing among high-risk individuals in families with hereditary nonpolyposis colorectal cancer. *British Journal of Cancer*, 90, 882-827. doi: 10.1038/sj.bjc.6601529
- Popper, K. (1968). *Conjectures and Refutations: The growth of scientific knowledge*. Haper & Row: New York.
- Probst, V., Plassard-Kerdoncuf, D., Mansourati, J., Mabo, P., Sacher, F., Fruchet, C., . . . Le Marec, H. (2011). The psychological impact of implantable cardioverter defibrillator implantation on brugada syndrome patients. *Europace: European Pacing, Arrhythmias, and Cardiac Electrophysiology: Journal of the Working Groups on Cardiac Pacing, Arrhythmias, and Cardiac Cellular Electrophysiology of the European Society of Cardiology*, 13, 1034-1039. doi: 10.1093/europace/eur060

- Quaid, K. A., & Morris, M. (1993). Reluctance to undergo predictive testing: The case of Huntington disease. *American Journal of Medical Genetics*, 45, 41-45. doi: 10.1002/ajmg.1320450112
- Rapp, R. (1999). *Testing women, testing the fetus. The social impact of amniocentesis in America*. New York: Routledge.
- Rapp, R. (1988) Chromosomes and communication: the discourse of genetic counselling, *Medical Anthropology Quarterly*, 2, 143-157. doi: 10.1525/maq.1988.2.2.02a00040
- Rayner, S. (1992). Cultural Theory and Risk Analysis. In S. Krimsky & D. Golding (Eds.), *Social theories of risk* (p. 83-116). Westport, CT: Praeger Publishers.
- Rees, G., Fry, A., & Cull, A. (2001). A family history of breast cancer: Women's experiences from a theoretical perspective. *Social Science*, 52(9), 1433-1440. doi: 10.1016/S0277-9536(00)00248-3
- Rees, G., Martin, P. R., & Macrae, F. A. (2007). Screening participation in individuals with a family history of colorectal cancer: a review. *European Journal of Cancer Care*, 17(3), 221-232. doi: 10.1111/j.1365-2354.2007.00834.x
- Reeve, J., Owens, R. G., & Winship, I. M. (2000). Psychological impact of predictive testing for colonic cancer. *Journal of Health Psychology*, 5(1), 99-108. doi: 10.1177/135910530000500102
- Reichelt, J. G., Heimdal, K., Møller, P., & Dahl, A. A. (2004). BRCA1 testing with definitive results: A prospective study of psychological distress in a large clinic-based sample. *Familial Cancer*, 3(1), 21-28. doi: 10.1023/B:FAME.0000026820.32469.4a
- Richards, M. (1993) The new genetics: some issues for social scientists,

- Sociology of Health and Illness*, 15, 567–86. Retrieved from <http://onlinelibrary.wiley.com/doi/10.1111/j.1467-9566.1993.tb00363.x/pdf>
- Richards, M. (1996) Families, kinship and genetics. In T. Marteau & M. Richards (Eds.), *The troubled helix: Social and psychological implications of the new human genetics*. (pp. 249-273). Cambridge: Cambridge University Press. `
- Riedijk, S.R., de Snoo, F.A., van Dijk, S., Bergman, W., van Haeringen, A., Silberg, S., ... Tibben, A. (2005), Hereditary melanoma and predictive genetic testing: why not?. *Psycho-Oncology*, 14: 738–745. doi: 10.1002/pon.901
- Rose, N. S. (1993). Government, authority and expertise in advanced liberalism. *Economy and Society*, 22, 283-299. doi: 10.1080/03085149300000019
- Rose, N. S. (2007). *Politics of life itself: Biomedicine, power, and subjectivity in the twenty-first century*. Princeton: Princeton University Press.
- Ross, L. F., & Moon, M. R. (2000). Ethical issues in genetic testing of children. *Archives of Pediatrics & Adolescent Medicine*, 154, 873-879. doi: 10.1001/archpedi.154.9.873
- Sandelowski, M. (1986). The problem of rigor in qualitative research. *Advances in Nursing Science*, 8(3), 27-37. Retrieved from <http://psycnet.apa.org/psycinfo/1987-12268-001>
- Sanders, T., Campbell, R., Donovan, J., & Sharp, D. (2007). Narrative accounts of hereditary risk: Knowledge about family history, lay theories of disease, and "internal" and "external" causation. *Qualitative Health Research*, 17, 510-50. doi: 10.1177/1049732306297882

- Scheiber, R.S. (2001). The "how to" of grounded theory: Avoiding the pitfalls. In R.S. Schreiber & P.N. Stern (Eds.), *Using grounded theory in nursing* (pp. 55-83). New York: Spring Publishing Company.
- Schneider, R., Schneider, C., Kloor, M., Furst, A., & Moslein. (2012). Lynch syndrome: clinical, pathological, and genetic insights. *Langenbecks Archives of Surgery*, 397, 513-525. doi: 10.1007/s00423-012-0918-8
- Schwartz, M. D., Beth, N. P., Hughes, C., Main, D., Isaacs, C., & Lerman, C. (2002). Impact of BRCA1/BRCA2 mutation testing on psychologic distress in a clinic-based sample. *Journal of Clinical Oncology*, 20, 514-50. doi: 10.1200/JCO.20.2.514
- Scully, J. L., Porz, R., & Rehmann-Sutter, C. (2007). 'You don't make genetic test decisions from one day to the next'--using time to preserve moral space. *Bioethics*, 21, 208-17. doi: 10.1111/j.1467-8519.2007.00546.x
- Senior, V., & Marteau, T. M. (2007). Causal attributions for raised cholesterol and perceptions of effective risk-reduction: Self-regulation strategies for an increased risk of coronary heart disease. *Psychology and Health*, 22, 699-717. doi: 10.1080/14768320601020253
- Senior, V., Marteau, T., & Peters, T. (1999). Will genetic testing for predisposition for disease result in fatalism? A qualitative study of parents' responses to neonatal screening for familial hypercholesterolaemia. *Social Science*, 48, 1857-1860. doi: 10.1016/S0277-9536(99)00099-4
- Senior, V. Marteau, T. M., & Weinman, J. (2000). Impact of genetic testing on causal models of heart disease and arthritis: An analogue study. *Psychology*, 14, 1077.-1088. doi: 10.1080/08870440008407368

- Senior, V., Marteau, T. M., Weinman, J. (2005) Perceptions of control over heart disease in people with an inherited predisposition to raised cholesterol. *Psychology, Health & Medicine*; 10(1), 16-30. doi: 10.1080/13548500512331315334
- Senior, V., Smith, J. A., Michie, S., & Marteau, T. M. (2002). Making sense of risk: An interpretative phenomenological analysis of vulnerability to heart disease. *Journal of Health Psychology*, 7(2), 157. doi: 10.1177/1359105302007002455
- Shaw, C., Abrams, K., & Marteau, T. M. (1999). Psychological impact of predicting individuals' risks of illness: A systematic review. *Social Science & Medicine*, 49(12), 1571-1598. doi: 10.1016/S0277-9536(99)00244-0
- Shea, J. B. (2004). Quality of life issues in patients with implantable cardioverter defibrillators: Driving, occupation, and recreation. *AACN Clinical Issues*, 15(3), 478-489. Retrieved from http://journals.lww.com/aacnadvancedcriticalcare/Abstract/2004/07000/Quality_of_Life_Issues_in_Patients_With.13.aspx
- Shedlosky-Shoemaker, R., Tho, L. N., Amy, K. F., Porter, K., Leventhal, H., & Kimberly, M. K. (2010). Exploring perceptions of genetic testing: An examination of perceived accuracy over time. *Patient Education and Counseling*, 78(1), 34-39. doi: 10.1016/j.pec.2009.05.010
- Sheinfeld Gorin, S., & Steven, M. A. (2003). The meaning of risk to first degree relatives of women with breast cancer. *Women*, 37(3), 97-117. doi: 10.1300/J013v37n03_07
- Sherwin. S. (1998). A relational approach to autonomy in health care. In S. Sherwin (Ed). *The politics of women's health* (pp. 19-47). Philadelphia. Temple University Press.

- Shiloh, S., Gerad, L., & Goldman, B. (2006). Patients' information needs and decision-making processes: What can be learned from genetic counselees. *Health Psychology, 25*(2), 211-219. doi: 10.1037/0278-6133.25.2.211
- Shiloh, S., & Ilan, S. (2005). To test or not to test? moderators of the relationship between risk perceptions and interest in predictive genetic testing. *Journal of Behavioural Medicine, 28*(5), 467-479. doi: 10.1007/s10865-005-9017-4
- Shiloh, S., Petel, Y., Papa, M., & Goldman, B. (1998). Motivations, perceptions and interpersonal differences associated with interest in genetic testing for breast cancer susceptibility among women at high and average risk. *Psychology & Health, 13*(6), 1071-1086. doi: 10.1080/08870449808407451
- Shiloh, S., & Saxe, L. (1989). Perception of risk in genetic counseling. *Psychology & Health, 3*(1), 45-61. doi: 10.1080/08870448908400365
- Sivell, S., Elwyn, G., Clara, L. G., Angus, J. C., Iredale, R., & Shaw, C. (2008). How risk is perceived, constructed and interpreted by clients in clinical genetics, and the effects on decision making: Systematic review. *Journal of Genetic Counseling, 17*(1), 30-63. doi: 10.1007/s10897-007-9132-1
- Sjöberg, L., Moen, B. E., & Rundmo, T. (2004). Explaining risk perception. An evaluation of the psychometric paradigm in risk perception research. *Norwegian University of Science and Technology, 1*-33. Retrieved from http://paul-hadrien.info/backup/LSE/IS%20490/utile/Sjoberg%20Psychometric_paradigm.pdf
- Skolbekken, J. (1995). The risk epidemic in medical journals. *Social Science & Medicine, 40*, 291-305. doi: 10.1016/0277-9536(94)00262-R

- Slovic, P. (1987). Perception of risk. *Science*, 236 (4799), 280-285. doi: 10.1126/science.3563507
- Slovic, P. (1992). Perception of risk: reflections on the psychometric paradigm. In S. Krimsky, & D. Golding (Eds.), *Social theories of risk* (pp. 117-152). Westport, CT: Praeger Publishers.
- Slovic, P. (2000). *Perception of risk*. London: Earthscan Publications
- Slovic, P., Finucane, M. L., Peters, E., & MacGregor, D. G. (2004). Risk as analysis and risk as feelings: Some thoughts about affect, reason, risk, and rationality. *Risk Analysis: An Official Publication of the Society for Risk Analysis*, 24, 311-322. doi: 10.1111/j.0272-4332.2004.00433.x
- Slovic, P., Fischhoff, B., & Lichtenstein, S. (1979). Rating the risks. *Environment*, 21(3), 14-20, 36-39.
- Slovic, P., Fischhoff, B., & Lichtenstein, S. (1980). Facts and fears: Understanding perceived risk. In R. Schwing, & W. A. Albers, Jr. (Eds.), *Societal risk assessment: How safe is safe enough* (pp. 181-216). Plenum: New York.
- Smart, A. (2010). Impediments to DNA testing and cascade screening for hypertrophic cardiomyopathy and long QT syndrome: A qualitative study of patient experiences. *Journal of Genetic Counseling*, 19, 630-639. doi: 10.1007/s10897-010-9314-0
- Smerecnik, C. M. R., Mesters, I., Verweij, E., Nanne, K. d. V., & Hein, d. V. (2009). A systematic review of the impact of genetic counseling on risk perception accuracy. *Journal of Genetic Counseling*, 18, 217-28. doi: 10.1007/s10897-008-9210-z
- Smets, E. M. A., Stam. M. M. H., Meulenkamp, T. M., van Langen, I. M., Wilde, A. A. M., Wiegman, A., & Tibben, A. (2008). Health-related quality of life of children with a

- positive carrier status for inherited cardiovascular diseases. *American Journal of Medical Genetics. Part A*, 146A (6), 700-707. doi: 10.1002/ajmg.a.32218
- Smith, J. A., Michie, S., Stephenson, M., & Quarrell, O. (2002). Risk perception and decision-making processes in candidates for genetic testing for Huntington's disease: An interpretative phenomenological analysis. *Journal of Health Psychology*, 7(2), 131-144. doi: 10.1177/1359105302007002398
- Sobel, S., & Cowen, B. C. (2003). Ambiguous loss and disenfranchised grief: The impact of DNA predictive testing on the family as a system. *Family Process*, 42(1), 47-57. doi: 10.1111/j.1545-5300.2003.00047
- Sowell, L. V., Kuhl, E. A., Sears, S. F., Klodell, C. T., & Conti, J. B. (2006). Device implant technique and consideration of body image: Specific procedures for implantable cardioverter defibrillators in female patients. *Journal of Women's Health (2002)*, 15, 830-835. doi: 10.1089/jwh.2006.15.830
- Speziale, H. S., & Carpenter, D. R. (2007). *Qualitative research in nursing: Advancing the humanistic imperative* (4th ed.). Philadelphia: Lippincott Williams & Wilkins.
- Starr, C. (1969). Social benefit versus technological risk. *Science*, 165, 1232-1238. Retrieved from <http://www.jstor.org/stable/1727970>
- Strauss A.L. & Corbin, J. (1998). *Basics of qualitative research: Grounded theory procedures and techniques* (2nd ed.). Thousand Oaks, CA: Sage.
- Strauss, C.L. (1968). *The savage mind*. London: Weindenfeld & Nicolson.
- Stern, P. N., & Porr, C. J. (2011). *Essentials of accessible grounded theory*. Walnut Creek, CA: Left Coast Press.

- Tansey, J., & O'Riordan, T. (1999). Cultural theory and risk: A review. *Health, Risk & Society, 1*(1), 71-90. doi: org/10.1080/13698579908407008
- Taouqi, M., Ingrand, I., Beauchant, M., Migeot, V., & Ingrand, P. (2010). Determinants of participation in colonoscopic screening by siblings of colorectal cancer patients in France. *BMC Cancer, 10*, 355-364. doi: 10.1186/1471-2407-10-355
- Taylor, C. A., & Myers, R. H. (1997). Long-term impact of Huntington disease linkage testing. *American Journal of Medical Genetics, 70*, 365-370. doi: 10.1002/(SICI)1096-8628(19970627)70:4<365: AID-AJMG7>3.0.CO;2-S
- Taylor, S. D. (2005). Predictive genetic test decisions for Huntington's disease: Elucidating the test/no-test dichotomy. *Journal of Health Psychology, 10*, 597-612. doi: 10.1177/1359105305053442
- Thiene, G., Corrado, D., & Basso, C. (2007). Arrhythmogenic right ventricular cardiomyopathy/dysplasia. *Orphanet Journal of Rare Diseases, 2*(45), 1-16. doi: 10.1186/1750-1172-2-45
- Thiene, G., Nava, A., Corrado, D., Rossi, L., & Pennelli, N. (1988). Right ventricular cardiomyopathy and sudden death in young people. *New England Journal of Medicine, 318*, 129-133. doi: 10.1056/NEJM198801213180301
- Tibben, A., Duivenvoorden, H. J., Vegtervandervlis, M., Niermeijer, M.F., Frets, P.G., Vanderkamp, J.P., ... Verhage, F. (1993) Presymptomatic DNA testing for Huntington's disease: identifying the need for psychological intervention. *American Journal of Medical Genetics, 48*, 137-144. doi: 10.1002/ajmg.1320480305

- Timman, R., Roos, R., Maat-Kievit, A., & Tibben, A. (2004). Adverse effects of predictive testing for Huntington disease underestimated: Long-term effects 7-10 years after the test. *Health Psychology, 23*(2), 189-197. doi: 10.1037/0278-6133.23.2.189
- Tozzo, P., Caenazzo, L., & Rodriquez, D. (2012). Genetic testing in minors: Comparison between Italian and British guidelines. *Genetics Research International, 2012*, 1-4. doi: 10.1155/2012/786930
- Tversky, A., & Kahneman, D. (1973). Availability: A heuristic for judging frequency and probability. *Cognitive psychology, 5*, 207-232. doi: 10.1016/0010-0285(73)90033-9
- Tversky, A., & Kahneman, D. (1974). Judgment under uncertainty: Heuristics and biases. *Science, New Series, 185*, 1124-1131. Retrieved from <http://links.jstor.org/sici?sici=00368075%2819740927%293%3A185%3A4157%3C1124%3AJUUHAB%3E2.0.CO%3B2-M>
- U.S. Department of Energy Genome Programs. (2012). *Human genome project information*. Retrieved from http://www.ornl.gov/sci/techresources/Human_Genome/home.shtml
- Valente, E., Ferraris, A., & Dallapiccola, B. (2008). Genetic testing for paediatric neurological disorders. *The Lancet, 7*, 113-126. doi: 10.1016/S1474-4422(08)70257-6
- Van Den Broek, K. C., Habibovic, M., & Pedersen, S. S. (2010). Emotional distress in partners of patients with an implantable cardioverter defibrillator: A systematic review and recommendations for future research. *Pacing and Clinical Electrophysiology : PACE, 33*, 1442-1450. doi:10.1111/j.1540-8159.2010.02885.x; 10.1111/j.1540-8159.2010.02885.x

- van Dooren, S., Rjnsburger, A., Seynaeve, C., Hugo, J. D., Essink-Bot, M. J., Tilanus-Linthorst, M., ... Tibbenm, A. (2004). Psychological distress in women at increased risk for breast cancer: The role of risk perception. *European Journal of Cancer*, 40, 2056-63. doi: 10.1016/j.ejca.2004.05.004
- van Ittersum, M., de Greef, M., van Gelder, I., Coster, J., Brügemann, J., & van der Schans C. (2003). Fear of exercise and health-related quality of life in patients with an implantable cardioverter defibrillator. *International Journal of Rehabilitation Research*, 26(2), 117-122. Retrieved from <http://www.ncbi.nlm.nih.gov/pubmed/12799605>
- van Maarle, M. C., Southard, M. E., & Boonsel, G. J. (2003). Risk perception of participants in a family-based genetic screening program on familial hypercholesterolemia. *American Journal of Medical Genetics. Part A*, 116A(2), 136-143. doi: 10.1002/ajmg.a.10061
- van Manen, M. (1997). *Researching lived experience: Human science for an action sensitive pedagogy*. London, Ontario: The Althouse Press.
- van Oostrom, I., Meijers-Heijboer, H., Lodder, L. N., Duivenvoorden, H. J., van Gool, A. R., Seynaeve, C., & Tibben, A. (2003). Long-term psychological impact of carrying a BRCA1/2 mutation and prophylactic surgery: A 5-year follow-up study. *Journal of Clinical Oncology*, 21, 3867-3874. doi: 10.1200/JCO.2003.10.100
- van Rosmalen, M. S., Stalmeier, P., Verhoef, L., Hoekstra-Weebers, J., Oosterwijk, J., Hoogerbrugge, N., & van Daal, W. (2004). Impact of BRCA1/2 testing and disclosure of a positive test result on women affected and unaffected with breast or

- ovarian cancer. *American Journal of Medical Genetics*, 124, 346-355. doi: 10.1002/ajmg.a.20374
- Venter, J.C., Adams, M.D., Myer, E.W., Li, P.W., Mural, R.J., Sutton, G., & Zhu, X. (2001). The sequence of the human genome. *Science*, 291, 1304-1351. doi: 10.1126/science.1058040
- Vodermaier, A., Esplen, M. J., & Maheu, C. (2010). Can self-esteem, mastery and perceived stigma predict long-term adjustment in women carrying a BRCA1/2-mutation? evidence from a multi-center study. *Familial Cancer*, 3, 305-311. doi: 10.1007/s10689-010-9325-x
- Vos, J., Jansen, A., Menko, F., van Asperen, C., Stiggelbout, A., & Tibben, A. (2011). Family communication matters: The impact of telling relatives about unclassified variants and uninformative DNA-test results. *Genetics in Medicine : Official Journal of the American College of Medical Genetics*, 3, 305-311. doi: 10.1097/GIM.0b013e318204cfed
- Wainberg, S., & Husted, J. (2004). Utilization of screening and preventive surgery among unaffected carriers of a BRCA1 or BRCA2 gene mutation. *Cancer Epidemiology Biomarkers Prevention*, 13, 1989-1995. Retrieved from <http://cebp.aacrjournals.org/>
- Walker, L. (1979). *The battered woman*. New York: Harper and Row.
- Walter, F. M., & Emery, J. (2005). 'Coming down the line'--patients' understanding of their family history of common chronic disease. *Annals of Family Medicine*, 3, 405-414. doi:10.1370/afm.368

- Walter, F. M., Emery, J., Braithwaite, D., & Marteau, T. M. (2004). Lay understanding of familial risk of common chronic diseases: A systematic review and synthesis of qualitative research. *Annals of Family Medicine*, 2, 583-595. doi: 10.1370/afm.242.
- Watson, M., Foster, C., Eeles, R., Eccles, D., Ashley, S., Davidson, R., . . . Evans, D. (2004). Psychosocial impact of breast/ovarian (BRCA 1/2) cancer-predictive genetic testing in a UK multi-centre clinical cohort. *British Journal of Cancer*, 91, 1787-1794. doi: 10.1038/sj.bjc.6602207
- Weiner, K. (2009). The tenacity of the coronary candidate: How people with familial hypercholesterolaemia construct raised cholesterol and coronary heart disease. *Health*, 13, 407-427. doi: 10.1177/1363459309103915
- Weiner, K., & Durrington, P. (2008). Patients' understandings and experiences of familial hypercholesterolemia. *Community Genetics*, 11, 273-282. doi:10.1159/000121398
- Weiner, K., & Martin, P. (2008). A genetic future for coronary heart disease? *Sociology of Health*, 30, 380-395. doi: 10.1111/j.1467-9566.2007.01058.x
- Wheeler, E. C., Pretzer-Aboff, I., Hardie, T., Disabatino, A., Saylor, J., & Lucey, R. (2009). Psychological impact of implantable cardioverter defibrillator on their recipients. *Dimensions of Critical Care Nursing : DCCN*, 28, 176-181. doi: 10.1097/DCC.0b013e3181a4743b
- White, M. M. (2002). Psychosocial impact of the implantable cardioverter defibrillator: Nursing implications. *The Journal of Cardiovascular Nursing*, 16(3), 53-61.
Retrieved from <http://www.journals.lww.com/jcnjournal/pages/default.aspx>
- Wildavsky, A. & Dake, K. (1990). Theories of risk perception: Who fears what and why? *Daedalus*, 119, 41-60. Retrieved from <http://www.jstor.org/stable/20025337>

- Wilkinson, I. (2010). *Risk, vulnerability and everyday life*. London: Routledge.
- Wilson, M. H., Engelke, M. K., Sears, S. F., Swanson, M., & Neil, J. A. (2012). Disease-specific quality of life: Patient acceptance: Racial and gender differences in patients with implantable cardioverter defibrillators. *The Journal of Cardiovascular Nursing*, Advance online publication. doi: 10.1097/JCN.0b013e31824e072e
- Wiseman, M., Dancyger, C., & Michie, S. (2010). Communicating genetic risk information within families: A review. *Familial Cancer*, 9, 691-703. doi: 10.1007/s10689-010-9380-3
- Zajonc, R. B. (1980). Feeling and thinking: Preferences need no inferences. *American psychologist*, 35(2), 151. doi: 10.1037/0003-066X.35.2.151
- Zeigler, V. L., & Nelms, T. (2009). Almost normal: Experiences of adolescents with implantable cardioverter defibrillators. *Journal for Specialists in Pediatric Nursing : JSPN*, 14, 142-151. doi:10.1111/j.1744-6155.2009.00186.x
- Zinn, J., (2008). Heading into the unknown: Everyday strategies for managing risk and uncertainty. *Health, Risk and Society*, 10, 439-450. doi: 10.1080/136985708023808

Appendix A

ARVC Diagnostic Criteria

Criteria for diagnosis of right ventricular dysplasia

<p>I Global and/or regional dysfunction and structural alterations⁽¹⁻³⁾ •</p> <p>MAJOR Severe dilatation and reduction of right ventricular ejection fraction with no (or only mild) LV impairment Localised right ventricular aneurysms (akinetic or dyskinetic areas with diastolic bulging) Severe segmental dilatation of the right ventricle</p> <p>MINOR Mild global right ventricular dilatation and/or ejection fraction reduction with normal left ventricle Mild segmental dilatation of the right ventricle Regional right ventricular hypokinesia</p> <p>II Tissue characterisation of walls</p> <p>MAJOR Fibrofatty replacement of myocardium on endomyocardial biopsy</p> <p>III Repolarisation abnormalities</p> <p>MINOR Inverted T waves in right precordial leads (V2 and V3) (people aged more than 12 yr; in absence of right bundle branch block)</p>	<p>IV Depolarisation/conduction abnormalities</p> <p>MAJOR Epsilon waves or localised prolongation (>110 ms) of the QRS complex in right precordial leads (V1-V3)</p> <p>MINOR Late potentials (signal averaged ECG)</p> <p>V Arrhythmias</p> <p>MINOR Left bundle branch block type ventricular tachycardia (sustained and non-sustained) (ECG, Holter, exercise testing). Frequent ventricular extrasystoles (more than 1000/24 h) (Holter)</p> <p>VI Family history</p> <p>MAJOR Familial disease confirmed at necropsy or surgery</p> <p>MINOR Familial history of premature sudden death (<35 yr) due to suspected right ventricular dysplasia. Familial history (clinical diagnosis based on present criteria)</p>
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*Detected by echocardiography, angiography, magnetic resonance imaging, or radionuclide scintigraphy. ECG, electrocardiogram; LV, left ventricle.

From: McKenna, W.J., Theine, G., Nava, A., Fontaliran, F., Blomstrom-Lundqvist, C., Fontaine, G., et al. (1994). Diagnosis of arrhythmogenic right ventricular dysplasia/cardiomyopathy. *British Heart Journal*, 71(3), 215-218. This is the diagnostic criteria used to diagnose ARVC in this study's participants. This diagnostic criterion was revised in 2012.

Appendix B

The Theory: Constructing the Meaning of Being At-Risk

Construct 1: Awakening to a New Meaning of Being Risk

<i>Category 1: Making Sense Out of Numerous Losses</i>
<ul style="list-style-type: none"> • Property 1: Living in a Family Familiar with Loss. • Property 2: The Struggle to Understand the Meaning of Being At-Risk for Oneself and Others.
<i>Category 2: Struggling to Break the Cycle of Uncertainty</i>
<ul style="list-style-type: none"> • Property 1: Making Sense of and Living Through Early Clinical Investigations and Prescribed Treatments. • Property 2: Acknowledging a Possible Genetic Origin to Risk.

Construct 2: Deciphering the Meaning of Being At-Risk

<i>Category 1: Taking the First Steps of the Genetic Testing Process</i>
<ul style="list-style-type: none"> • Property 1: Being Offered a Predictive Genetic Test. • Property 2: Making the Decision to Participate in Predictive genetic testing.
<i>Category 2: Building One's Risk Portfolio</i>
<ul style="list-style-type: none"> • Property 1: Waiting for Predictive Genetic Test Results. • Property 2: Receiving Genetic Test Results.

Construct 3: Embodying a New Meaning of Being At-Risk

<i>Category 1: Adjusting to Living with or without a Genetic Condition</i>
<ul style="list-style-type: none"> • Property 1: Accepting and Assigning Meaning to Treatments Regimes • Property 2: Questioning the Accuracy of the Predictive Genetic Test
<i>Category 2: Recognizing the Reality of Living in a Family at risk for a Genetic Disease</i>
<ul style="list-style-type: none"> • Property 1: Facing Everyday Worries and Challenges • Property 2: Coping with Barriers to Resources
<i>Category 3: Looking Towards the Future Realities of Living in a Family at risk for a Genetic Disease</i>
<ul style="list-style-type: none"> • Property 1: Trying to Maintain a Positive Attitude • Property 2: Living with Concern for Others at-risk

Appendix C

Amendment for Ethical Approval

January 2012

Health Research
Ethics Authority

RECEIVED 11/15/2012

07.145

APPROVED 1/15/2012

Request For Ethics Renewal / Study Closure

- The Tri-Council Policy Statement- Ethical Conduct for Research Involving Humans (TCPS2, 2010) (article 6.14) requires ongoing review by the approving REB at least on an annual basis. The information provided in this form must be current to the time of submission and submitted to the HREA not less than 30 days nor more than 45 days before the anniversary of your approval date.
- Ethics approval is required if there is ongoing subject contact or data collection/transfer is active.
- Ethics approval is not required and the file may be closed if the project is in analysis or the writing stage.
- Please forward a summary of findings or published abstract to the HREA Office once the study is complete.
- Incomplete forms will not be accepted and may result in delay in the review and approval process.
- (For clinical trials only) if the project is complete - please submit the applicable Study Closure form.

HREA Ref Number 07 145	Expiry Date of Current Approval 01/15/12
Principal Investigator: April Manuel	
Title of study (with Protocol Number if applicable): Experiences of Individuals with Genetic-Linked Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) in the province of Newfoundland and Labrador: A Grounded Theory Study	
Email of PI: admanuel@mun.ca	Email of Key Contact:

Please choose one	
<input checked="" type="checkbox"/>	I am requesting renewal of ethics approval for this file.
<input type="checkbox"/>	I am requesting to close this file.
	yes

April Manuel
Name typed or printed

April Manuel
Signature of PI

08/09/2012
Date (MM/DD/YYYY)

This project was reviewed on _____	
Date	By Full Board Review <input type="checkbox"/> By Expedited Review <input checked="" type="checkbox"/>
Ethics approval for this project has been granted for a period of 12 months effective from _____ to _____	
<p>This research ethics board (the HREA) has reviewed and approved the study which is to be conducted by you as the qualified investigator/principal investigator named above. This approval and the views of this Research Ethics Board have been documented in writing. The Health Research Ethics Board operates according to Tri-Council Policy Statement, Ethical Conduct for Research Involving Humans, ICH Guidance E6 Good Clinical Practice Consolidated guideline and applicable laws and regulations. The membership of this research ethics board is constituted in compliance with the membership requirements for research ethics boards as defined by Health Canada Food and Drug Regulations Division 5 Part C.</p>	
This file has been closed as requested <input checked="" type="checkbox"/>	

January 2012

All Other Studies: Since Last Approval		Yes <input checked="" type="checkbox"/>	No <input type="checkbox"/>
1. Have there been unexpected events or problems related to participant risk since original approval or last ethics renewal?			
2. Has there been amendments submitted for this project?		Yes <input type="checkbox"/>	No <input checked="" type="checkbox"/>
<p><i>If yes, please describe the events/problems/amendments (Add an addendum to this form if necessary)</i></p> <p><i>A focus group was completed wherein three youths between the ages of 14-16 requested to participate as part of the family. The parents of all three youths were present and gave informed consent. At this focus group one of my research supervisors was present and provided consultation as to the participation of the youths. No psychosocial repercussions were reported upon follow up.</i></p>			

All Studies - Status At Local Site (check all that apply)	YES	NO	N/A
1. Intervention/data collection active		<input checked="" type="checkbox"/>	
2. Closed to recruitment/accrual	<input checked="" type="checkbox"/>		
3. Participants in follow up		<input checked="" type="checkbox"/>	
4. Site closed (clinical trials only)			<input checked="" type="checkbox"/>
5. For secondary use of data only is Data Transfer Complete			<input checked="" type="checkbox"/>

Knowledge Transfer	YES	NO	N/A
1. Have participants been informed of study findings?		<input checked="" type="checkbox"/>	
2. Have findings been presented/published?		<input checked="" type="checkbox"/>	
<i>Please indicate where: (Add an addendum to this form if necessary)</i>			

Additional Information :

Participants will be informed of study findings but not until my dissertation is complete; similarly, presentations and publications will follow after the dissertation has been completed.

Appendix D

Recruitment Script

I just wanted to let you know that there is a new study starting that is looking at the experiences of individuals in families with a history of ARVC. This study is funded by a group called Genome Atlantic. It's being conducted by a PhD student at Memorial who's working on the experiences of individuals in families that have ARVC.

If you want to be part of this study there would be two face to face interviews. The interviews would take place at a time and location of your convenience. If you give permission the interviews will be audiotaped and later transcribed to written form. Your identity will be kept confidential throughout the study and when the results are published. You are in no way obligated or expected to participate in this study. But if you do want to be part of it, then I will pass on your name and phone number to the researcher, April Manuel.

Are you interested in participating in this study? If so I'll pass on your contact information to April, and she will contact you and explain the study and answer any questions that you may have. And then when you're talking to her and have found out more about the study, you can decide whether you want to participate.

Appendix E

Research Study Cover Letter

Experiences of Individuals with Arrhythmogenic Right Ventricular Cardiomyopathy
(ARVC) in the Province of Newfoundland and Labrador: A Grounded
Theory Study.

March 18, 2008

Dear Sir/Madam,

Thank you for taking the time to look at the attached materials that explain this study. The goal of this study is to hear your ideas on what life is about being in a family with a history of ARVC. The purpose is to help health care providers know what services people with ARVC need.

If you choose to participate you will be interviewed two times about your experiences of being in a family with ARVC. The first interview will take about one hour and the second will take about 30 minutes. The interviews will be done at a time and place convenient for you.

In the attached package you will find a description of the study and a consent form. Once you have read them please feel free to contact me, April Manuel, the main researcher, at 1-709-777-6319, or at the toll free number 1-877-222-6319 with any questions you may have about the research study or your participation.

I will be contacting you by telephone about two weeks after you get the research package to confirm your interest in participating in this research study.

Thank you for your interest.

Respectfully,

April Manuel BNMN
Primary Investigator
Doctorate of Community Health and Humanities Student
Newfoundland and Labrador ARVC Study

Appendix F

Summary of the Research Study

Title: Experiences of Individuals with Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) in the Province of Newfoundland and Labrador: A Grounded Theory Study.

Investigator: April Manuel

Purpose of the study:

The main purpose of this study is to understand your ideas about what it is like to be in a family with a history of ARVC. The second purpose of the study is to see if your experience is similar to other people who belong to families with hereditary disorders.

Rationale for the study:

Health care workers need to know what the experience is like to have ARVC in the family, in order to provide the best service. It is thought that because of the seriousness of ARVC people deciding to have genetic testing may find it stressful. This study will look at how you learned that there was a possibility that you might have ARVC and how it has changed your life.

Description of study:

This study will look at people's experiences with genetic testing for ARVC in Newfoundland and Labrador. You will be asked to talk about how you learned that there was a possibility that you might have ARVC and how it has changed your life. The study will take place at a time and location of your choice. There will be two interviews in this study.

At the first interview will last about one hour. At the start of the first interview I will explain the purpose of the study, answer any questions that you have about the study and your participation. I will then ask you to sign a consent form.

The second interview will take place at a later agreed upon date. You will be asked to look at a summary of interview one. You will be given time to ask questions about the study or to add ideas that may help me to understand your experience with genetic testing.

Proposed starting date: December, 2008

Appendix G Consent Form

Consent to Take Part in Health Research

TITLE: Experiences of Individuals with Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) in the Province of Newfoundland and Labrador: A Grounded Theory Study.

INVESTIGATOR(S): April Manuel BNMN

SPONSOR: Atlantic Medical Genetics and Genomics Initiative, Genome Atlantic

You have been invited to take part in a research study. It is up to you to decide whether to be in the study or not. Before you decide, you need to understand what the study is for, what-risks you might take and what benefits you might receive. This consent form explains the study.

The researchers will:

- **discuss the study with you**
- **answer your questions**
- **keep confidential any information which could identify you personally**
- **be available during the study to deal with problems and answer questions**

If you decide not to take part or to leave the study this will not affect your usual health care or normal treatment.

1. Introduction/Background:

Many families in Newfoundland and Labrador have been affected with ARVC. Genetic testing is routinely offered in these families. You have been invited to join this study because you were offered genetic testing for ARVC.

Health care workers need to gain a better understanding of the experiences of people living in families with a history of ARVC, how the decision was made to have genetic testing (or not) and the impact of this decision on everyday life in order to meet the health needs of these families. This study will look at how you learned that there was a possibility that you might have ARVC and how it has changed your life.

2. Purpose of study:

The purpose of this study is to gain a better idea of how living in a family with a history of ARVC affects your everyday life. I am interested in hearing how you learned that you might have ARVC, how you decided if you should have genetic testing, and how you feel that decision affected your life. Another purpose of this study is to compare your

experience of genetic testing with other individuals who have been offered genetic testing for other hereditary disorders.

3. Description of the study procedures and tests:

You will be asked to participate in two face to face interviews. Both interviews will take place at a time and location that is best for you. In the first interview you will be asked to talk about what it is like to be in a family with a history of ARVC. You may be asked about how you learned that there was a possibility that you might have ARVC, your decision about whether to have genetic testing, and how that decision has affected your life.

In the second interview you may be asked to explain or talk more about what you said in the first interview. This is to be sure that I have understood your experience.

4. Length of time:

The first interview will take about one hour. The second interview will take about 30 minutes.

5. Possible risks and discomforts:

You may find talking about your experience stressful. At any time you may decide to stop the interview and leave the study. Then the audio-files and notes will be destroyed immediately. You have the choice to take a break at any time during the interview or postpone it for another time. I will be prepared with the telephone number of the Director of the Provincial Medical Genetics Program if at any time you would like to talk about concerns regarding your experience with genetic testing.

6. Benefits:

It is not known whether this study will benefit you.

7. Liability statement:

Signing this form gives us your consent to be in this study. It tells us that you understand the information about the research study. When you sign this form, you do not give up your legal rights. Researchers or agencies involved in this research study still have their legal and professional responsibilities.

8. Confidentiality:

Confidentiality and privacy of your personal information and interview will be maintained at all times. The only people who will see the interview are: the person who types the interviews to paper, and my supervisory committee. The interview and consent form will be locked in separate cabinets in my locked office where I will be the only person with a key to the cabinets. Any computer files will be password protected. Your personal information will be removed from your interviews by me. Each interview will be given a code so that you will not be identified.

There is a possibility that you may have others in the room during your interview. In this case confidentiality will be discussed with everyone present prior to the start of the interview.

9. Questions:

If you have any questions about taking part in this study, you can meet with the investigator who is in charge of the study at this institution. That person is:

April Manuel 1-709-777-6319 or Toll Free Number :1-877-222-6319

Or you can talk to someone who is not involved with the study at all, but can advise you on your rights as a participant in a research study. This person can be reached through: Office of the Human Investigation Committee (HIC) at 709-777-6974

Email: hic@mun.ca

Signature Page

Study title: Experiences of Individuals with Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) in the Province of Newfoundland and Labrador: A Grounded Theory Study.

Name of principal investigator: April Manuel

To be filled out and signed by the participant:

Please check as appropriate:

I have read the consent [and information sheet].	Yes { }	No { }
I have had the opportunity to ask questions/to discuss this study.	Yes { }	No { }
I have received satisfactory answers to all of my questions.	Yes { }	No { }
I have received enough information about the study.	Yes { }	No { }
I have spoken to April Manuel and she has answered my questions	Yes { }	No { }
I agree to have my interviews audio-taped	Yes { }	No { }
I understand that I am free to withdraw from the study	Yes { }	No { }
• at any time		
• without having to give a reason		
• without affecting my future care		

I understand that it is my choice to be in the study and that I may not benefit.

Yes { } No { }

I agree to take part in this study.

Yes { } No { }

Signature of participant

Date

To be signed by the investigator:

I have explained this study to the best of my ability. I invited questions and gave answers. I believe that the participant fully understands what is involved in being in the study, any potential risks of the study and that he or she has freely chosen to be in the study.

Signature of investigator

Date

Telephone number: _____

Appendix H

Sample Interview Schedule/Questions

I was wondering if you could please share your experience of what it was like to have had genetic testing for ARVC? You may start wherever you want and talk about any experiences, thoughts and feelings that you feel is important for us to gain a better understanding of the process of genetic testing.

Examples of Probes/Questions to Facilitate the Interview and Enrich Data

Could you please tell me about growing up in your family, did you know anybody sick?

(Probes: Did you have any relatives die at an early age? Where there any unexplained deaths in your family?)

Perhaps you could reflect on how you became aware of the possibility of some genetic condition in your family? How did you become aware of your potential for having ARVC?

(Probes: Have you been healthy? How did it make you feel when you learned that there was a high probability that you may have ARVC?)

Did you have any thoughts at that time on what it meant to have ARVC? How did you become aware of what it meant to have the gene for ARVC? What was your understanding of ARVC? What was your understanding at this time of treatment protocols and recommended screening for ARVC?

(Probes: What was your perception of being at risk for ARVC? Do you feel you had a good grasp on what it meant to be at risk for ARVC? Could you please guide me through your thoughts at this time? When did you become aware that ARVC was a genetic linked disorder? What was your perception of having a genetic linked disorder? What were your thoughts when you realized that if you have the gene for ARVC it could be passed to your offspring?)

Who was the first person who alerted you that you might have a hereditary disease? Who provided you with the information on ARVC?

(Probes: Do you feel that you had a good understanding of what it meant to be at risk for ARVC after discussion(s) with this person? After this discussion how did your perception of being at risk for ARVC change. What were your immediate concerns/thoughts when you learned that you might have a hereditary disorder? Did this knowledge change your perception of everyday life or how you carried out your activities of daily living? Where there any particular incidents during your discussions with this individual(s) that comes to mind that impacted your understanding of ARVC? Why was this incident so significant?)

How did the probability that you and your family may have the gene for ARVC affect the family and everyday life? Who were included in your support network?

(Probes: Was there a disruption in the family everyday functioning? Where there times that were particularly stressful on the family when you suspected that you or members of your family may have the gene for ARVC? Was there anyone outside your immediate family that was a support person(s) for the family? How did members of the family support each other? Are there any incidents that stand out in your mind you feel happened because of the uncertainty surrounding the risk of having ARVC?

Could you describe what it was like to have to wait for genetic testing? Could you tell me about your decision to participate (or not) in testing for ARVC? When/How did you learn about the availability of testing for ARVC? What were your initial thoughts when you became aware that testing was available for ARVC?

Who do you think made up your support network during this decision? Did you feel ready to participate in genetic testing?

(Probes: What thoughts went through your mind while you were waiting to be tested for ARVC? What factors impacted your decision to participate (or not) in genetic testing? Did you experience any thoughts associated with reluctance to genetic testing? Where all the family tested at the same time? Where there any discussion in the family surrounding not getting tested? Did you feel you had adequate knowledge regarding the procedure for genetic testing?)

How did you prepare for testing? At this time what meaning did genetic testing have for you and your family? Did you have any thoughts on the consequences of testing positive or negative? Could you please tell me about what thoughts went through your mind when you were waiting for the test results? What was your understanding of your risk of having ARVC and passing it on to your offspring? Did you discuss with your children any information on ARVC? Who provided you with support during this time?

(Probes: Did you have any thoughts on how you would deal with the test results? What did you understand were the treatment options? Who did you share your thoughts and ideas with thorough out this experience? Where there any immediate or longterm concerns that came to your mind?)

Where there any specific moments or images that comes to mind that you might recall as significant though out the testing process?

(Probes: Where there any times that you felt particularly sad or relieved through out the testing process?)

After you received your tests results what were your thoughts? How do you perceive your tests results? How did you cope with your test results? How do you feel your life has changed? Do you feel satisfied with the decision to participate (or not) in genetic testing?

How do you feel the test result has impacted your family? Do you have any suggestions for others who may be facing a similar situation? How supportive is your family and friends?

(Probes: How do you feel that your family responded to your tests results? (Probes: How did you reveal the results of your test to the rest of your family? How did this make you feel? What factors affected your decision to disclose your tests results to your family? How did your family members respond? Were there any immediate concerns that impacted the family?)

Has this experienced changed your life or the way you look at life? Do you perceive these changes in your life as positive or negative? What do you do to stay positive?

(Probes: How has genetic testing for ARVC changed the way you for see your future? Has this experience impacted your livelihood? Has this experience impacted your relationship with your children?)

What is your understanding of treatment for ARVC? What is your understanding of recommended screening protocols?

Do you think that this experience has altered your feelings towards genetic testing? Do you think your offspring should have genetic testing?

(Probes: Was your experience and positive or negative experience? Could you please give me an example of some positive or negative experiences?)

How helpful are health care providers involved in your care? Do you have any thoughts on how health care professional support could be improved?

Do you have any concerns regarding barriers to accessing genetic information, testing and screening that your might have experienced? Could you give me an example of a concern that you have had?

(Probes: Do you feel you were given enough education about ARVC prior to making the decision to participate (or not) in genetic testing, and post-test results? Did you have any concerns regarding access to necessary resources for your care? How has geography (where you live) impacted your experience? What are your thoughts on health care professional's communication skills when discussing genetic screening and treatment options?

Overall, how would you describe your experience with genetic testing? Are there any particular areas that you would like to see changed, improved or implemented?

Do you see any benefits from becoming involved in supportive groups? Having access to up-to-date information such as news letters or web sites? Having ongoing contact with individuals involved in ARVC testing, research and counselling?

Is there any area of the genetic testing experience that you would like to make a comment on or discuss?

Appendix I

Ethics Approval



Faculty of Medicine

SHIPPED OCT 12 2007

October 1, 2007

Reference #07.145

Ms. April Manuel
Memorial University of Newfoundland
School of Nursing

FILE COPY

ENTERED OCT 13 2007

Dear Ms. Manuel,

This will acknowledge your correspondence dated, September 16, 2007, wherein you clarify issues and provide a script for your research study entitled **"Experiences of Individuals with Genetic-linked Arrhythmogenic right ventricular cardiomyopathy (ARVC) in the province of Newfoundland and Labrador: A Grounded Theory Study"**.

At the meeting held on August 16, 2007, the initial review date of this study, the Human Investigation Committee (HIC) agreed that the response could be reviewed by the Co-Chairs and, if found acceptable, full approval of the study be granted.

The Co-Chairs of the HIC reviewed your correspondence, approved the script and, under the direction of the Committee, granted **full approval** of your research study. This will be reported to the full Human Investigation Committee, for their information at the meeting scheduled for October 11, 2007.

Full approval has been granted for one year. You will be contacted to complete the annual form update approximately 8 weeks before the approval will lapse on **August 16, 2008**. It is your responsibility to ensure that the renewal form is forwarded to the HIC office not less than 30 days prior to the renewal date for review and approval to continue the study. The annual renewal form can be downloaded from the HIC website: <http://www.mun.ca/hic/downloads/Annual%20update%20form.doc>.

The Human Investigation Committee advises THAT IF YOU DO NOT return the completed annual update form prior to or on the aforementioned date of renewal:

- Your ethics approval will lapse
- You will be required to stop research activity
- You will not be permitted to restart the study until you re-apply for and receive approval to undertake the study again

In addition, the Human Investigation Committee will inform the appropriate authorities. To ensure proper action is taken, the appropriate officials will be notified to terminate funding.

Modifications of the protocol consent are not permitted without prior approval from the Human Investigation Committee. Implementing changes in the protocol consent without HIC approval may result in the approval of your research study being revoked, necessitating cessation of all related research activity. Request for modification to the protocol consent must be outlined on an amendment form (available on the HIC website) and submitted to the HIC for review.

For a hospital-based study, it is **your responsibility to seek the necessary approval from Eastern Health and/or other hospital boards as appropriate**


This Research Ethics Board (the HIC) has reviewed and approved the application and consent form for the study which is to be conducted by you as the qualified investigator named above at the specified study site. This approval and the views of this Research Ethics Board have been documented in writing. In addition, please be advised that the Human Investigation Committee currently operates according to the Tri-Council Policy Statement and applicable laws and regulations. The membership of this research ethics board complies with the membership requirements for research ethics boards defined in Division 5 of the Food and Drug Regulations.

Notwithstanding the approval of the HIC, the primary responsibility for the ethical conduct of the investigation remains with you.

We wish you every success with your study

Sincerely,

John D. Hamett, MD, FRCPC
Co-Chair
Human Investigation Committee


Richard S. Neuman, PhD
Co-Chair
Human Investigation Committee

JDH:RSN jed

Dr. C. Loomis, Vice-President (Research), MUN
Mr. W. Miller, Senior Director, Corporate Strategy & Research, Eastern Health

Appendix J

Research Approval: Eastern Health



St. John's Mercy Hospital
St. John's, NL
Canada A1C 5A8
T: (709) 777-5233
F: (709) 777-5271
www.easternhealth.ca

December 4, 2007

Ms. April Manual
MUN School of Nursing
HSC

Dear Ms. Manual:

Your research proposal "HIC # 07.145 Experiences of individuals with genetic-linked Arrhythmogenic right ventricular cardiomyopathy in the province of Newfoundland and Labrador: a grounded theory study" was reviewed by the Research Proposals Approval Committee (RPAC) of Eastern Health at its meeting on December 3, 2007 and we are pleased to inform you that the proposal has been approved.

The approval of this project is subject to the following conditions:

- The project is conducted as outlined in the HIC approved protocol;
- Adequate funding is secured to support the project;
- In the case of Health Records, efforts will be made to accommodate requests based upon available resources. If you require access to records that cannot be accommodated, then additional fees may be levied to cover the cost;
- A progress report being provided upon request

If you have any questions or comments, please contact Donna Bruce, Manager of the Patient Research Centre at 777-7283

Sincerely,

Mr. Wayne Miller
Senior Director Corporate Strategy & Research
Chair, RPAC
Eastern Health

cc: Ms. Donna Bruce, Manager Patient Research Centre
Dr. Sandra LeFort, MUN School of Nursing

