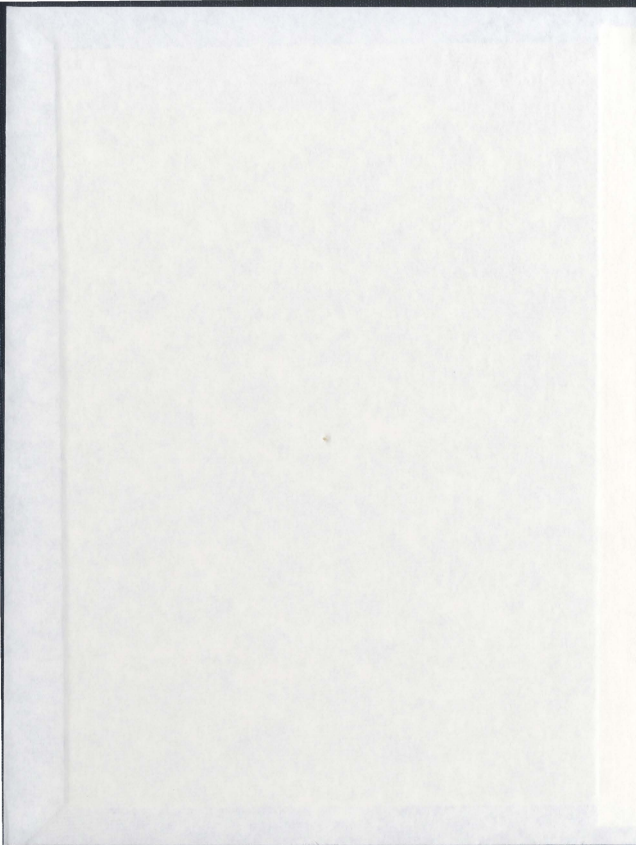


'I HAVE THE GENE, BUT I DON'T HAVE
HUNTINGTON DISEASE':
NEGOTIATING GENETIC RISK

HOLLY ETCHEGARY



Running Head: GENETIC RISK FOR HUNTINGTON DISEASE

'I have the gene, but I don't have Huntington disease': Negotiating genetic risk

by

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Abstract

In the emerging risk society (Beck, 1992), healthy bodies, rather than the sick or diseased, are the focus of medical attention. Nowhere is this more evident than in the field of predictive genetic testing. Few empirical studies of predictive testing have explored the everyday reality of living at risk for a fatal inherited disorder. Fewer still have focused on those already living with such a disorder and their caregivers. Drawing upon 24 semi-structured interviews with at-risk persons and their family members, this study examined the implications of living at risk for, or with, the adult-onset disorder, Huntington disease (HD). Qualitative data analysis revealed that genetic risk was not understood or retained as an objective numerical fact, much as it is constructed so by Mendelian genetics. Rather, genetic risk for HD was re-conceptualized as an index of threat to self and other family members. Discussion about genetic risk for HD was infused with emotions and moral undertones; the latter reflected a felt obligation to other family members. As such, decisions around genetic risk were sometimes constrained by perceived responsibility to others in the family. Living with risk for HD or with the illness itself had noticeable effects on self-identity and relationships with others. While the response of social others was often sympathy, perceived stigma did exist in relation to HD, affecting communication about the illness and sometimes restricting social behavior. Implications for research and clinical practice are discussed.

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CHAPTER 1

Middle of nowhere

These melancholy filled tears
 which encase all my animosities and fears,
 As that final defining sunset
 is descending my way

Will that last smidgen of sand
 from life's hourglass
 let me see another forsaken day?
 Will I ever again see the sun
 absorb the morning sky,
 or be enchanted by the sleepy morning dew?

As I lay my faith into destiny's sweet hands,
 As the winds of change storm mercilessly
 through my life,
 and into the epitome of my soul,

I'm in the middle of nowhere
 with no place to hide,
 As the angel of death descends upon me,
 do I have to go for the ride?

Middle of nowhere is a disturbing poem written by one of the remarkable individuals I have met during the course of this research. In the interest of my ethical obligations to him and his family, I will call him Jason. His words stand as testimony to a period of despair and hopelessness in Jason's life, a time when suicide seemed the only option. Genetic testing had confirmed his worst fears: He carries the altered gene for a fatal genetic disorder. His child lives, in blissful ignorance, at 50% risk for the same disorder. Jason chose life; he is currently a young man living with Huntington disease (HD). The current research endeavored to tell his story and the stories of others, affected in some way, by HD.

THE GENETICS REVOLUTION

We used to think our fate was in the stars. Now we know, in large measure, our fate is in our genes.

-James Watson, first director of the Human Genome Project
(cited in Holtzman, 1998)

We are on the brink of a genetic age. With distressing regularity, news media regale us with the latest genetic discoveries. The “gene for” not only disease, but also, behaviors and personality traits is revealed almost weekly. Due in part to the work of the Human Genome Project (HGP), a billion-dollar international research effort, a draft sequence of the entire human genome has been completed (e.g., Venter, Adams, et al., 2001). The genome had been described as the ‘Book of Life,’ and the quest to decipher it, the ‘Holy Grail’ of biology. Molecular biologist Walter Gilbert argued, “the possession of a genetic map and the DNA sequence of a human being will transform medicine” (1992, p. 94). The identification of more and more disease genes intimates a concomitant increase in the number of genetic tests that are available. The use of information derived from genetic testing raises important social, ethical and legal considerations. Underscoring these concerns, three to five percent of the U.S. Department of Energy’s and the National Institutes of Health’s annual HGP budgets have been earmarked for the empirical study of the ethical, legal and social issues (ELSI) arising from new genetic technologies (administered by the ELSI program).

Doubtless, the genetic age holds promise. Scientific discoveries underlying clinical genetics have the potential to provide new understandings of the genetic basis of disease with the hope of improving human health. A fairly immediate result of mapping

disease-causing genes is the development and availability of both diagnostic and predictive genetic tests. In fact, genetic testing for disease could soon become a *routine* component of medical care (Lerman, Croyle, Tercyak, & Hamann, 2002, emphasis added). Predictive genetic testing is distinct from either carrier or diagnostic testing in that it provides a currently healthy individual's *own risk* of developing a genetic disease in his/her lifetime. As the HGP continues to provide the tools for discovering genes predisposing to *common* diseases (e.g., cancer, diabetes or heart disease), predictive genetic testing will likely be the primary type of genetic test offered in the near future. Bell (1998) suggested, "The rapid advances in human molecular genetics seen over the last five years indicate that within the next decade, genetic testing will be used widely for predictive testing in healthy people and for diagnosis and management of patients" (p. 618).

Fundamentally, genetic risk information can have profound consequences for individuals, families and society at large. It could change how we think about life and death, normalcy and disability, kinship and the quality of life (Conrad, 1999). It affects how we see ourselves, potentially altering self-identities. It can influence how we label and categorize others and how we, ourselves, are labeled. For example, Carter (1995) suggested that people found to be carrying a genetic 'taint' could be relegated to a space of danger, that is, the place of the 'other.' In this way, boundaries of safety and danger are drawn. Behind the genetic age lurks the specter of eugenics, and with it, disquietude about discrimination, intolerance and stigma. Fears abound that society will become less sympathetic to disability of any kind since it will be assumed that the births of severely

handicapped people can be avoided. Genetic discrimination in employment and insurance contexts is a real possibility and therefore, a concern. Technology has outpaced our ability to anticipate and negotiate these myriad psychological, social and ethical consequences of providing genetic risk information (Sobel, 1997).

The benefit of disseminating genetic risk information can be further questioned in the context of disorders for which medicine offers no treatment or cure. It can be argued that, "In the absence of effective treatment or prevention for inheritable disorders, the provision of information about genetic risk is *itself* emerging as a new type of medical intervention" (Cox, 1999, p. 3, emphasis in original). While empirical research devoted to this new intervention is in its infancy, rapid advances in genetic medicine suggest such research will be increasingly important. Peters, Djurdjinovic, and Baker (1999) suggested that stories about illness have always figured prominently in family and individual life; however, it is *genetic* risk that will take centre stage in the illness narratives of the future.

Social scientists can contribute to the understanding of these genetic narratives. Richards (1993) argued that social scientists have been deterred from empirical research on the new genetics owing to the latter's highly technical language, coupled with a lasting distaste for biological reductionism. Much of the empirical work on genetic decision-making and response to genetic testing has been conducted from a clinical perspective. While valuable, this approach largely neglects the social and familial contexts within which genetic risk information is interpreted and acted upon (Cox, 1999).

Labeled 'at risk'

What are the experiences of those individuals who have chosen to ascertain their genetic risk of disease? Conversely, what becomes of those who resist the 'at risk' label, preferring uncertainty to definitive risk knowledge? How is risk information incorporated and negotiated in everyday life? How do people *make sense* of genetic risk information, give it meaning and significance in their lives? Alternatively, is it significant at all? And if so, when or under what circumstances? With whom is this information shared? From whom is it concealed? Does genetic risk information induce perceived, or actual, stigma? What is the effect of genetic risk information for other family members? These are pertinent questions for genetically at-risk families and for the healthcare professionals who work with them. They are also the focus of the current research.

This study sought to understand the meaning of genetic risk/illness for people living in the shadow of a fatal genetic disorder. The aim was not to determine participants' recall and comprehension of numerical risk information; although, some participants evinced a rather sophisticated comprehension of their genetic risk. Instead, the current research investigated how people talked about genetic risk in the course of informal conversation and how (or if) genetic risk was incorporated into everyday lived reality (cf. Parsons & Atkinson, 1992). I concur with Adelsward and Sachs (1998) who argued, "Lay assessment and evaluation of risk is a social process, not a scientific, technical one" (p. 203).

Patients before their time

Richards (1993) suggested that minimal empirical work had investigated the experiences of people whose lives have been touched by new genetic technologies. Since his admonition, a growing body of work has addressed this important area. However, there is still much we do not know. Broadly speaking, illness experiences have been extensively studied; it is more difficult to study the rather 'intangible' experience of health (Lawton, 2003). However, in the emerging 'risk society' (Beck, 1992), healthy bodies are the focus of medical attention, and currently healthy people are the usual targets of predictive genetic testing. Jonsen (1996) warned that in this climate, "Persons will become patients before their time: They will be described in disease terms but 'feel fine' and 'be fine' for years, perhaps always" (pp. 8-9).

'Risk,' its calculation, prediction and management is a typical feature of late modern society (Beck, 1992), and the advances in genetic medicine intimate that more and more people will be living at risk. Macintyre (1995) noted that the nature of the social identity of being at risk and the effect of this label are important issues for study. Kenea (1994), for example, warned that the at risk label will create a new social category of people, the 'possibly, potentially diseased' (PPD) or the 'diseased in waiting' (DIW). She argued we are all at risk of being labeled so, since we all carry within us some deleterious genes. A primary aim of the current research was to investigate the effect of an at risk label on everyday life.

Genetic testing is perhaps the pinnacle of what Armstrong called surveillance medicine. Armstrong (1995) suggested the 20th century witnessed the birth of

surveillance medicine, whose subject and object was the 'risky self.' The significance of surveillance medicine, "...lies in the way in which a surveillance machinery deployed throughout a population to monitor precarious normality delineates a new temporalised risk identity" (p. 403). Broadly speaking, 'risk identity' was the subject of the current research. Importantly, genetic advances anticipate that whole families, not solely individuals, can be subject to a risk identity. Yet little empirical work examines the implications of genetic-risk knowledge for the family (Tercyak, Streisand, Peshkin, & Lerman, 2000). The current study encompassed not only at-risk individuals, but also their family members, some of whom were not genetically at risk themselves. This in no way meant their lives were unaffected by genetic risk.

Huntington disease

Specifically, the current research comprised those with a family history of the genetic disorder, Huntington disease (HD) and, where possible, their partners or friends. HD has an autosomal dominant transmission. It affects both men and women and, at birth, each child of a HD parent has a 50:50 chance of inheriting the disease and is said to be 'at risk' for the disorder. Therefore, inheriting the altered HD gene is a random event that can be compared to the flip of a coin. *Not* inheriting the altered HD gene, also a random event, effectively eliminates HD in subsequent generations. The identification of the specific mutation for HD in 1993 (Huntington Disease Collaborative Research Group, 1993) allowed testing by direct mutation analysis. This meant that a simple blood test could now provide an individual's HD risk information; the cooperation of other family members, required in the older linkage analysis procedure, was no longer necessary.

Symptoms

HD is a progressive neuro-degenerative disease whose symptoms normally include a movement disorder, personality changes and intellectual decline (Huntington Society of Canada [HSC], 2002a). Therefore, the disease can encompass affect, behavior and cognition, leading to significant morbidity and early mortality in most cases. Uncontrollable jerking movements, typically called chorea, affect the trunk, limbs and face of a person with HD (SuttonBrown & Suchowersky, 2003), although as many as one fifth affected with HD suffer from muscle rigidity instead (Cox, 1999). Initial motor symptoms become worse as the disease progresses and can cause difficulty with walking, speaking and swallowing (HSC, 2002a). Persons with HD often have abnormal gait and slurred speech, for instance. Choking is a real possibility as the disease progresses and impairs normal swallowing. Cognitive impairments can include recall difficulty, attention deficits, difficulty in decision making and eventual dementia. Emotional impairment can include personality changes such as impulsiveness, disinhibition and aggression. Other emotional symptoms can include depression, irritability and obsessive-compulsive behavior. However, symptoms and their severity vary from individual to individual. For example, some people with HD suffer from severe involuntary movements, while others are more likely to suffer cognitive or emotional impairments.

Origin

HD was the first serious, autosomal dominant disorder for which predictive testing with DNA markers became available (Harper, Lim, & Craufurd, 2000). The genetic defect is confined to a small sequence of DNA on the short arm of chromosome

4. The gene is known as IT15, and its protein product is referred to as huntingtin.

Huntingtin is thought to play an important role in the normal functioning of the nervous system; however, it is currently unknown exactly how the defective gene leads to nerve cell damage in the brain. The gene is composed of three DNA bases – cytosine, adenine and guanine (CAG) - known as trinucleotide repeats. The number of repeats is significant since HD is caused by a CAG repeat expansion.

Notably, HD is one of the few genetic disorders that is 100% penetrant. This means that individuals who carry the altered HD gene *will* manifest the disease in his or her lifetime, excepting death from some other cause before the disease manifests. The normal allele contains 26 or fewer CAG repeats, with the most common length between 17 and 19 (Potter, Spector, & Prior, 2004). Conversely, HD appears fully penetrant when the CAG repeat expansion is equal to or greater than 40 (ACMG/ASHG, 1998). An allele size of 36-39 CAG repeats, however, can be equivocal. People bearing these repeat expansions may or may not develop symptoms of HD, and about one percent of people tested fall into this reduced penetrance category (HSC, 2002a).

While CAG repeats of 27 to 35 are rare, they can be associated with a HD phenotype, depending on the sex of the transmitting parent and allele size, among other factors (Potter et al., 2004). Therefore, some inherent uncertainty remains for individuals undergoing predictive genetic testing for HD, especially when CAG repeats fall in the 27-39 range. Further, while some alleles are 100% penetrant (e.g., 40 or more CAG repeats), even these individuals at risk for HD do not know when the illness will strike, which symptoms they are most likely to manifest or how severe those symptoms will be. Thus,

while some HD risk information may be fairly certain, much uncertainty remains in the life-world of a person at risk for HD.

Timing and prevalence

Age of onset for HD is usually between 30 and 45, although it can appear in young children or in adults as old as 70 (HSC, 2002a). Age of onset is associated with the length of the CAG repeat, such that the longer the repeat, the lower the age of onset (Potter et al., 2004). Regardless of age of onset, HD is a progressive disease. Complications from the disease (e.g., aspiration, infection or heart failure) typically cause death 10-30 years after disease onset (SuttonBrown & Suchowersky, 2003). The seriousness of this illness should not be underestimated: "The end result of the relentless progression of HD is a thin, bedridden, spastic, dysphagic, densely demented shadowy remnant of the former self" (O' Shea, 1997, p. 136). There is currently no cure for HD, and limited options for treatment.

The prevalence of HD is about five to eight per 100,000 in Europe and North America; it is less common in non-European ethnic groups (SuttonBrown & Suchowersky, 2003). The exact prevalence of HD is difficult to specify for a number of reasons (HSC, 2002b). For example, HD is often misdiagnosed. O'Shea (1997) noted that in psychiatric practice, HD has been differentially diagnosed as other dementing disorders, schizophrenia or affective and personality disorders. Mortality statistics for HD are not accurate either. Cause of death is often a secondary complication such as infection, aspiration or heart failure; HD might never be noted on a death certificate. Finally, some families are secretive about HD for fear of discrimination or social rejection. The best

estimate suggests that one in every 10,000 Canadians has HD, while approximately five in every 10,000 are at risk of developing the disease. However, it is estimated that one in every 1,000 Canadians is touched in some way by HD, whether as an affected individual, friend, family member or caregiver (HSC, 2002a), underscoring the fact that HD is a disease of *families*.

Reviewing the psychological consequences of genetic testing and counseling for HD, van't Spijker and ten Kroode (1997) ironically noted that in spite of "severe difficulties" for families, minimal research attention has been devoted to specific problems experienced by family members. Every participant in the current study strongly endorsed the notion that HD was a family illness; they were able to relate many specific experiences of family members in relation to the illness. In fact, talking about genetic risk for HD seemed synonymous with recounting numerous and varied stories about multiple family members, as well as oneself. Similarly, describing six different family cases of HD, Dudokdewit, Savenije, Zoeteweyj, Maat-Kievit, and Tibben (2002) showed the complex effects and implications of HD risk information for transitions in the family life cycle. Participants in the current research also referred to wide-spread familial effects and the relevancy of risk information during certain critical family-life moments (e.g., the transition of their children to teenagers or young adults or their own transition to later life).

Note, however, that the variability in onset and severity of symptoms precludes discussion of *the* HD individual or *the* HD family. Throughout this research, I have been struck repeatedly by individual and family differences in the meanings of HD, the

importance accorded to the illness, the response to testing, the effect(s) of testing and diagnosis and the negotiation and dynamic nature of the at risk status.

Evers-Kiebooms and Decruyenaere (1998) suggested that in order to understand motives for genetic testing and the effect of receiving a predictive test result, "It is of the utmost importance to realize what it means psychologically to be at 50% risk for Huntington's disease. This risk influences the entire life of the at risk individual" (p. 16). The current research was an attempt to address this understanding.

'At risk' in context

By definition, genetic risk is family risk. Arguably, the meaning of and importance accorded to the risk, communication about genetic risk and ways of coping with living at risk (and HD itself) will depend heavily on the family context. However, families also exist in a wider social context. Kenen, Ardern-Jones, and Eeles (2004), discussing communication about breast cancer risk, noted the changing landscape of societal views about cancer. Once dubbed the 'Big C,' cancer was often a family secret. Those times have changed. The media have placed breast cancer squarely on the public agenda and interventions and testing options for breast cancer are marketed widely. Compare the social context of breast cancer to the social context of HD: The latter is a relatively rare genetic disorder, largely invisible to society at large. In a promotional video, the Huntington Society of Canada noted that HD was largely unknown, even in the medical community. This context of 'ignorance' was often referred to by participants in the current research and affected response(s) to risk information, communication about genetic risk and perceptions of social stigma.

In keeping with the importance of context, Lawton (2003) has called for an examination of illness which pays attention not only to timing and context, but also, to individual biographies in order to understand “the complex and often variable ways in which people experience, and ‘live with’ illness” (p. 27). The current research used in-depth, semi-structured interviews to access the individual biographies of genetically at risk people, individuals clinically affected with HD and their family members. Importantly, both the timing and context of HD emerged as important considerations in understanding participants’ responses to risk information. For example, at-risk persons who have only recently learned of their at risk status often show a high degree of interest in taking a genetic test. Many do reconsider, however, once they have adjusted to the newly acquired at risk status (Tyler, Ball, & Caufurd, 1992). It has been suggested that such individuals wish to dispense quickly with a new source of anxiety (Cox, 1999). Cox (1999) noted, however, that these at-risk persons might be unfamiliar with HD, raising concerns about their ability to provide truly informed consent for genetic testing.

Genetic testing has been introduced into a social climate dominated by (at least) two themes: (1) risk and (2) individual responsibility for health, with associated notions of culpability and blame. These ideas informed and drove the current research. They are elaborated in the chapters to follow. Notably, the concepts are intimately related. It can be argued that knowledge of genetic risk, whether *suspected* because of family history, or *determined* through genetic testing, encourages individuals and families to reflect on their situation and act morally in accordance with this knowledge. Huniche (2001) noted that this “moral prompting” is not exclusive to genetic science or practice, but rather,

“...comes about through our understanding of the relation between knowledge and responsible decision making in our part of the world. Namely, the former as a solid foundation for the latter” (p. 35).

In the chapters to follow, I review and critically evaluate selected portions of the literature on risk, predictive testing for HD, living at risk for HD and social stigma. In so doing, I draw from the fields of health and social psychology, sociology, medical and clinical genetics and genetic counseling. In discussing the social and familial implications of living at risk for (or with) a genetic illness, I draw from medical sociology, and to a lesser extent, anthropology, since social psychology has been relatively silent on these issues. Drawing upon literature from numerous disciplines was deliberate. Test protocols stress a multi-disciplinary approach to the provision of genetic testing, with geneticists, neurologists, family physicians, nurses, social workers and other healthcare professionals playing a pivotal role. In the same vein, a multi-disciplinary approach to empirical research should inform the understanding of what it means to live at risk for, or with, a genetic illness such as HD.

CHAPTER 2

WHAT IS 'RISK'?

A constant companion

A depleting ozone, nuclear weaponry, terrorist attacks, rising levels of unemployment, escalating crime rates, the threat of serious illness and new genetic technologies. Those of us living in contemporary Western societies live in an age of risk. It is argued that 'risk society' (Beck, 1992) is one characterized by uncertainty, doubt and constant insecurity. Traditional social roles and institutions (e.g., gender, occupation, the family or education) can no longer be trusted to provide guidance for, or give definition to, our lives. Risk has become a pervasive concept in the 21st century and "...the noun 'risk' and the adjective 'risky' have become very commonly used in both popular and expert discourses" (Lupton, 1999a, p. 9). As an example of the latter, Skolbekken (1995) cautioned that the increasing frequency of the term 'risk' in medical journals over three decades resembled "an epidemic."

Changes in the meaning of risk

It is instructive to briefly review the history of the concept of 'risk' since it dominates the social context into which genetic testing and the dissemination of genetic risk information have been introduced. Lupton (1999a) reviewed changes in the meaning and use of the word 'risk.' Emergence of the risk concept was linked to the hazards which plagued naval voyages in the pre-modern period. During that time, risk was perceived as largely beyond human control: Violent storms or other dangers of sea travel

were imputed to an act of God or a force of nature. Notably, then, this early concept of risk largely *excluded* human responsibility for danger and misfortune.

Lupton (1999a) suggested that changes in the meaning of risk were linked to the emergence of modernity, that is, "...the 'industrialized world', incorporating capitalism, the institutions of surveillance and nuclear weaponry, as well as the process of industrialism" (p. 6). One of the assumptions of modernity was the existence of an objective reality that could be known through scientific thinking and investigation. As an outgrowth of this epistemology, the science of statistics and probability was developed.

Developments in statistics would become important in the modernist notion of risk: This notion of risk accepted that unintended outcomes or misfortunes could be the result of human action. Through science, however, outcomes could be predicted. Risk, then, was no longer seen as an act of God or nature, but rather *the consequence of human agency*. Skolbekken (1995) suggested that this shift from external to internal agency was the background against which the current 'risk epidemic' could be understood. One function of the 'risk epidemic' he argues, is the prediction of future disease and death, suggesting that risk identification, prediction and management are rational ways to gain control over illness. This accords with Beck who noted that managing risk is a way of "colonizing the future" (Giddens, 1991) since "events that do not exist (yet) strongly influence our present affairs and activities" (Beck, 1998, p. 11). Joffe (1999) suggested that predictive genetic testing represents the ultimate contemporary example of 'colonizing the future.'

Note that during earlier time periods, risks could be ‘good’ or ‘bad.’ For example, Ewald (1991) discussed risk as it developed in the insurance industry. In insurance, risk is associated with ideas of probability and loss, but also of gain. It is true that accidents *can* happen, and people insure themselves against them. From the perspective of insurance, however, risk is a fairly neutral concept – it denotes the probability of an event, with the anticipated losses *or* gains. This risk perspective was prominent until the beginning of the nineteenth century (Ewald, 1991), but the distinction between ‘good’ and ‘bad’ risks had become lost by the end of the twentieth century. Douglas suggested that “...the word *risk* now means danger, *high risk* means a lot of danger” (Douglas, 1992, p. 24, emphases in original). Lupton (1999a) concurred, suggesting that in the public’s everyday language, risk refers almost exclusively to danger, harm and threat. Lindbladh and Lyttkens (2003) suggest that risk and worry tend to be synonymous in everyday discourse. Risk has become a negatively-charged word, referring to something threatening on the personal or familial level. In this context, it is suggested that risk is a lived dimension of life, rather than an objective, neutral concept. While risk has been theorized on a grand scale, far less empirical work is devoted to how people experience risk as part of their everyday lives (Finkler, 2003; Lupton, 1999b). This lived dimension of risk was a primary focus of the current study.

Approaches to risk research and their application in genetics

The realist approach: Rational decision makers

Lupton (1999a) reviewed several approaches to risk research in the social sciences. She noted the most common is the ‘realist’ perspective, especially widespread

in technical and scientific approaches to risk. This “technico-scientific” approach to risk (e.g., in such fields as psychology, epidemiology and economics) is defined by a probabilistic approach to dangers or hazards. Risks are seen to be the product of both the probability and severity of a hazard or threat. In this perspective, risks are accepted as pre-existing in nature. In principle, scientific method and measurement have the potential to identify, calculate and potentially control risks. Psychometric studies of risk tend to focus on risk identification (e.g., the events, activities or things people perceive as risks or as risky) and on people’s responses to risk (Lupton, 1999b). For example, a majority of this type of risk research is concerned with identifying the ways people respond cognitively, affectively and behaviorally to risk. Lupton (1999a) noted that this approach to risk often assumes that lay people simply cannot, or do not, understand risk and so respond to risk information with inferior knowledge or ‘intuition.’

‘Biased’ risk perception

In this vein, a large body of decision-making research has identified the heuristics, or rules of thumb for judgment, people use when taking decisions based on probabilistic information (e.g., Tversky & Kahneman, 1974). Within the genetics context, there are few studies conducted explicitly in the framework of heuristic decision making. Nonetheless, it has been suggested that this could be a useful avenue for research on genetic-test decisions (Shiloh, 1996). The decision to take a predictive genetic test is a situation requiring the processing of large amounts of information, most of which is likely new and difficult. Kenen, Ardern-Jones, and Eeles (2003a) suggested these were precisely the circumstances conducive to heuristic decision making. In their research with

women genetically at risk for breast cancer, Kenen et al. (2003a) noted that heuristics did influence women's judgements about the cancer risk they faced. For example, vivid, unpleasant images of a relative's breast cancer were easily recalled (i.e., the availability heuristic) and tended to color risk perceptions and test decisions. Other research has revealed heuristic decision making in genetic counseling situations (e.g., Shiloh, 1994).

In addition to availability, Tversky and Kahneman (1974) also identified a representativeness heuristic. That is, we sometimes assign an instance to a category simply because it seems representative of the category (i.e., is similar to it). It causes bias in risk perception because individuals fail to consider the impact of probabilities or the effect of the sample size on the representativeness of an observation. For example, when estimating their child's risk of developing an adult-onset genetic disorder, such as HD, parents often refer to the degree of parental resemblance (Shiloh, 1994). Boutte (1990) interviewed individuals at risk for Machado-Joseph disease, an inherited neurological disorder similar to HD. Discussing their risk for the disease, participants in that study also referred to their similarity in appearance or temperament to the affected relative, despite knowledge of their objective risk (i.e., 50% chance of inheriting the defective gene). Similarly, some participants in the current research seemed to 'pre-select' (Evers-Kiebooms & Decruyenaere, 1998) which at-risk family member would be the one to develop HD based on similarity to an affected parent and/or grandparent. It has been suggested that pre-selection is a coping mechanism for living at risk.

'Rational' responses to risk

As noted, the reliance on heuristics in genetic decision-making situations has received minimal research attention. Instead, risk researchers from the realist tradition attempt to identify the factors which influence lay people's response(s) to risk information. This approach to risk tends to regard human beings largely as information processors: Rational thinkers who are risk avoidant. For example, socio-cognitive models of health behavior (e.g., Health Belief Model; Janz & Becker, 1984; Protection Motivation Theory; Rogers, 1983) suggest a linear relationship between health knowledge, attitudes and behavior (see Armitage & Conner, 2000, for a review). In the context of genetic testing, this approach to risk research is common. A growing body of research, for example, has attempted to delineate the factors which influence the uptake of predictive genetic testing (see Etchegary, 2004; Lerman et al., 2002, for reviews of this literature). Very broadly speaking, individuals with a positive attitude towards genetic testing, higher perceived risk, higher perceived personal control beliefs, lower perceived barriers to testing and higher perceived benefits of testing are more likely to intend to be tested or to engage in actual test behavior. Notably, however, the significance of these constructs varied depending on the disease. For example, higher levels of distress seem to motivate test intention and behavior for genetic testing for breast cancer, while disease-specific distress appears to deter testing interest for HD (Lerman et al., 2002). Genetic risk research from this 'realist' tradition, however, can offer little in the way of explanation for this finding. In addition, comparison studies of tested and untested people found that perceived severity of HD and perceived susceptibility to the disease were

similar in both groups (e.g., Evers-Kiebooms & Decruyenaere, 1998). Thus, social cognition models that rely heavily on these constructs will not be sufficient to explain why one person chooses to be tested and another declines. Rather, Evers-Kiebooms and Decruyenaere (1998) suggest that personality profile and individual coping style seem to be the primary factors in the decision to take a genetic test. Results of the current study provide some support for this suggestion. For example, when asked why they did or did not have the genetic test for HD, some participants began their answers with, “Well, I’m just the sort of person who...” or “I just had to know, that’s just who I am...”

Communicating risk

Within the genetics context, the psychometric approach to risk is also evident in the growing body of work which attempts to evaluate and improve risk communication in at risk individuals (Croyle & Lerman, 1999). In this work, risk perception is normally measured on a quantitative or qualitative scale. For the latter, a woman considering breast cancer testing might be asked, “Compared to other women your age, *your* risk is much lower, lower, the same, etc.” A quantitative scale, on the other hand, would ask people to rate their risk of genetic disease on a scale from 0% to 100%, or some other numerical index. A main goal of this research is to reveal the deviation from actual (i.e., objective) risk status (Robertson, 2003). Misperception of risk status could be important since accurate risk comprehension could be critical to genetic-test decisions and to preventative health behavior following test result (Croyle & Lerman, 1999). Note, however, the latter is less important for HD given there are no known preventative health behaviors which will impede or arrest disease progression. Nonetheless, risk perception and

communication have become active areas of inquiry in the genetics context. Two consistent findings have emerged from this body of work. First, many individuals overestimate their actual risk of genetic disease (Croyle & Lerman, 1999). Second, *perceived* risk, rather than actual risk, is a more consistent predictor of genetic-testing interest for a variety of disorders (Lerman et al., 2002). In general, those with a higher perceived risk are more likely to intend to obtain genetic testing. In light of this finding, research then proceeded to potentially modify overestimates of risk through education or counseling. Croyle and Lerman (1999), for example, reviewed several studies of breast cancer risk counseling and concluded that inflated perceptions of inherited cancer risk were only mildly influenced by standard educational approaches. A more recent review (Butow, Lobb, Meiser, Barratt, & Tucker, 2003) found that while improvements in risk perception were observed immediately after counseling, 22-50% of women continued to overestimate their risk.

There is an inherent assumption in this work that at risk individuals do not understand genetic risk information, an assumption Lupton (1999a) calls “ill masked contempt” (p. 9). Like Lupton, other authors are questioning the knowledge-deficit criticism of the lay public. They argue that subjective interpretation and qualification of risk figures might be perfectly valid in the daily lives of at risk individuals and their families (e.g., Smith, Michie, Stephenson, & Quarrell, 2002). Robertson (2003) noted that research which is concerned with measuring or improving the ‘accuracy’ of lay risk perception typically ignores how people understand risk information and create meaning about it in the context of daily life. The current study was expressly concerned with the

subjective meanings and interpretation of genetic risk. For example, Hallowell and Richards (1997), in their review of studies in risk recall, suggested that following genetic counseling, many individuals convert numerical risk estimates into binary outcomes: It will happen, or it won't (cf. Parsons & Atkinson, 1992). Some participants in the current study expressed their risk for HD in this way, even those whose HD allele was not fully penetrant. More generally, genetic consultants give their own meaning to the risks presented to them during counseling sessions (Biesecker-Bowles, 1998). In the current study, family experiences with HD and vivid memories of the affected parent, grandparent and/or sibling influenced the meaning participants gave to genetic risk and illness. In this way, risk was a lived and dynamic dimension of life, not a static, numerical probability. As Eiser (1998) noted: "Risk information is not 'perceived.' It is actively processed by individuals and families with problems to solve and decisions to take" (p. 790).

Psychological 'effect' of genetic risk information

Beyond examining the role of risk perception in genetic decision making, other psychometric risk research in the genetics context is concerned with the *effect* of risk information on psychological wellbeing. This body of literature is perhaps the largest of empirical research on HD, and in fact, my frustration and dissatisfaction with it was (in large part) the impetus for the current research. For example, the psychological impact of receiving a genetic-test result has been well studied, normally with clinical samples (see Broadstock, Michie, & Marteau, 2000; Meiser & Dunn, 2000, for reviews). The majority of these studies administer standard psychological instruments such as the Beck

Depression Inventory, The Beck Hopelessness Scale, the State-Trait Inventory, or the Centre for Epidemiological Studies (CES) Depression scale, among others. These are primarily used to assess the degree of anxiety and depression in individuals (and sometimes partners) prior to, and following, test disclosure. Most of this research is generally carried out as part of predictive testing protocols in specialized genetics centres.

Several reviews of the psychological effect of testing have been specific to HD. An early review (van't Spijker & ten Kroode, 1997) found that almost every person who tested positive for the altered HD gene experienced short-term emotional reactions such as numbness, sadness, anxiety or anger. However, most returned to normal levels of anxiety and depression one year following test disclosure. Increased feelings of hopelessness were observed in carriers of the altered HD gene, while reduced scores were recorded for those testing negative (Tibben, Duivenvoorden, Niermeijer, Vegter-van der Vlis, Roos, & Verhage, 1994). Within six months, however, unwanted intrusive thoughts about HD decreased for both groups. After three years, there were no differences in intrusive or avoidant thoughts about HD or in hopelessness between those testing positive and negative for the altered HD gene (Tibben, Timman, Bannink, & Duivenvoorden, 1997). In addition, those testing negative for the altered HD gene do not necessarily experience immediate, uncomplicated relief; there can be survivor guilt and difficulty adjusting to a new identity (Sobel & Cowan, 2000).

Dudokdewit et al. (2002) provided a more recent synopsis of research on the psychological implications of testing for HD. As is the case for genetic testing for other diseases, the actual uptake of testing for HD has been much lower than that indicated by

earlier studies of test intention. This is a finding which will be elaborated subsequently. For now, however, consider the implications of the finding that only 10-20% of at risk individuals choose to be tested. This means that upwards of 80% of the population at risk for HD is excluded from the medical and research arena. Binedell and Soldan (1997) noted that little is known about those who do not request HD testing, even though they are in the majority. A better understanding of why individuals choose not to be tested could help to anticipate future demand for genetic testing should therapeutic interventions become available. At that point, uptake for HD testing could increase. Currently, for example, it is not known to what extent lack of knowledge about HD testing affects test uptake (Binedell & Soldan, 1997). Nor do we know with any certainty whether there are differences in test uptake between urban and rural centres (Binedell & Soldan, 1997). Although, it is easy to imagine that testing might be easier or more convenient in urban centres.

Other observations of Dudokdewit et al. (2002) in their review of the psychological impact of testing for HD included the following: Most test candidates espouse relieving uncertainty, planning for the future and informing offspring as the main reasons for seeking testing. Fear of being unable to cope with a gene-positive result, on the other hand, is the primary reason given for refusing the genetic test. In general, short-term impact of the genetic test, whether a positive or negative result, is fairly good. Anticipated psychiatric problems (e.g., suicide) have rarely materialized. Catastrophic events were investigated in the largest cohort of HD test candidates ($N = 4527$) in a worldwide study (Almqvist, Bloch, Brinkman, Craufurd, & Hayden, 1999). In that study,

the percentage of suicide, suicide attempt and psychiatric hospitalization following predictive testing was estimated to be only .97%. However, a recent Canadian study of adverse events following testing (Almqvist, Brinkman, Wiggins, & Hayden, 2003) found that 6.9% of participants (14 of 202) experienced an adverse event within two years after testing. Notably, this finding was observed despite the fact that *both* carriers and non-carriers of the altered HD gene showed a *decrease* in psychological distress over the study's five-year follow-up period. It should be noted, however, that 'adverse events' were more broadly defined in this later study, including not only attempted and completed suicide, but also planned suicide, a diagnosis of clinical depression, an increase in alcohol consumption and/or the breakdown of married or common-law relationships.

Within relationships, partners of those testing positive for the altered HD gene tend to look, with worry, towards the future, while the at risk individual tends to live for the moment. Echoing Tibben et al. (1997), it is not the case that those testing negative for the altered HD gene are necessarily psychologically resolute. Rather, these once-at-risk individuals can feel intense guilt and numbed emotions. Unsurprisingly, HD and the genetic testing process itself, has profound effects on partners and other family members. Tibben et al. (1997), for example, observed that partners of those testing positive for the altered HD gene showed the same course of distress as test candidates themselves. In addition, partners of those testing positive who had children were more distressed than partners of those testing positive who didn't have children.

Another recent review of psychological studies in HD also produced several notable findings (Duisterhof, Trijsburg, Niermeijer, Roos, & Tibben, 2001). Mean scores of psychological wellbeing and HD specific distress were within normal range prior to test result in the majority of studies. Following test disclosure, carriers of the altered HD gene showed more depression, hopelessness and decreases in wellbeing, although mean scores were within a mild range. Within a month, levels of anxiety and depression returned to baseline levels; within six months, levels of hopelessness and general wellbeing returned to baseline level. In those testing negative, levels of optimism were higher one week post-test, but had dropped to baseline levels by six months. Anxiety, depression and general distress all decreased one year after test outcome. Compared to carriers of the altered HD gene, non-carriers reported less general distress, depression and hopelessness one week after the test outcome; however, this difference disappeared within the first year. In terms of Huntington specific distress, carriers of the altered HD gene evinced a mild increase of avoidance behavior in the first six months subsequent to test disclosure; however, mean scores returned to baseline at three-year follow up. Non-carriers, on the other hand, showed a decrease in avoidance at six months which returned to baseline at three year follow up. Notably, one attitudinal study found that at six-month follow up, some who tested positive suggested the test result had no influence on their lives and they rarely thought of the results. Duisterhof et al. (2001) suggested these results could indicate denial and identify a subgroup of at risk individuals who report they are functioning well, but could in fact be having difficulty integrating risk knowledge into their lives.

Why the minimal 'effect' on psychological well-being?

The lack of profound psychological sequelae following disclosure of a positive HD test result may be surprising. It has been suggested that the relative absence of psychological reaction to testing could be due to the lack of physical signs and symptoms of HD (Craufurd, Dodge, Kerzin-Storbarr, & Harris, 1989). Williams (1996) suggested that in the course of everyday life, our bodies are 'phenomenologically absent.' That is, the body is taken for granted, as is our health; it is only when things 'go wrong' with the body through illness, pain or disability that the body becomes the object of focused attention. Results of the current research lend some support to this suggestion. Notable differences emerged in the interviews of at risk, asymptomatic individuals and caregivers of persons with HD, the latter facing undeniable signs of the illness every day. It is precisely this kind of qualitative, contextual difference that health questionnaires typically fail to detect.

Longitudinal studies on the psychological effects of predictive testing for HD are rare. However, one recent study suggested that the long-term adverse effects of predictive testing for HD have been underestimated (Timman, Roos, Maat-Kievit, & Tibben, 2004). Seven to 10 years subsequent to receiving a positive test result, hopelessness scores were higher than at baseline, both for those testing positive for the altered HD gene and their partners. Timman et al. (2004) suggested that for these test candidates, increased hopelessness could be related to the onset of HD (i.e., the first visible symptoms of the illness). The current research lends some support to this suggestion. Those nearing the

age of disease onset did admit to feeling nervous and fearful as they neared the time when the illness would 'strike.'

It has also been hypothesized that a self-selected group, more capable of coping with a gene-positive result, is requesting genetic testing (e.g., Codori, Hanson, & Brandt, 1994); this could partially account for the lack of significant negative impact in those testing positive for the altered HD gene. The current research also lends some support to this suggestion. Non-tested individuals did suggest that a perceived lack of ability to cope with a positive test result was a primary reason for declining testing. However, research suggests that at-risk persons could forego psychological benefits conferred by genetic testing. In the first large-scale study of the psychological effects of testing for HD, Wiggins et al. (1992) concluded:

Knowing the result of the predictive test, even if it indicates an increased risk, reduces uncertainty and provides an opportunity for appropriate planning. Therefore, as our findings suggest, people who receive informative results, regardless of the content, may derive psychological benefits not experienced by those who remain uncertain (p. 1404-5).

Almqvist et al. (2003), however, cautioned that family and social contexts are important determinants of response to testing, and approaches to risk research outside the psychometric paradigm illustrate their point.

Critique of the psychometric approach

A significant portion of existing research on predictive testing for HD has focused on the individual psychological aspects of the clinical experience. Genetic test result (i.e., positive or negative) tends to be the main independent variable, and the focus is on elucidating causal relationships between test result and various clinical outcomes. This

approach to the psychological implications of genetic testing is valuable as it can help identify subgroups of the testing population who experience the most distress following test disclosure and who could need additional counseling or other related assistance. Generally, research in this area uses standardized psychological measures of anxiety and distress as the major dependant variables. These methods of data collection, however, provide minimal insight into the primary *focus* of anxiety (i.e., what are people anxious about and why?). Cox (1999) noted that while existing research is important for clinical evaluation, it says little about the meaning and lived experience of predictive testing for HD in at-risk persons and their families.

Denial?

In addition, while denial could be a response to HD risk information, it is not necessarily a negative, improper or immoral response. Yet this is often the assumption of health professionals, health researchers, social scientists and even other at risk individuals (Hunich, 2001; 2003). In the current research, for example, there was seeming evidence in some participant accounts of 'denial.' As I learned to really listen to their stories, however, it became apparent that such 'denial' was perhaps an inevitable response given the multiplicity of situations and interpersonal relationships which formed the backdrop of their everyday lives. For some, other struggles in everyday life relegated HD to the background, at least for the time being. For some participants, then, 'dismissal,' rather than 'denial' of their risk status was evident. This finding underscores the criticism that psychometric risk perception research generally provides only a 'snapshot' of risk judgements outside of the context of everyday realities (Wilkinson, 2001b). In addition,

for those at-risk persons who do go on to develop HD, symptoms are usually fairly mild initially. Bloch, Adam, et al. (1993) refer to the conscious awareness of disease presence as the “incipient stage.” During this stage, denial may be adaptive, providing the person with time to prepare (emotionally and practically) for the conscious acceptance of a diagnosis of HD.

What is ‘risk perception’ anyway?

Wilkinson (2001b) also noted that there exists no agreement on the meaning of ‘risk perception.’ His argument is conceivable when one considers the varied scales used to measure the construct in genetic-testing interest studies. As noted, both quantitative and qualitative scales are common, yet these may not represent a ‘standard’ measure of risk perception, given the subjective interpretation of risk. Croyle and Lerman (1999) have argued there is a need for better risk perception measures than currently exist in psychometric risk research. Some authors have argued for a distinction between ‘risk’ as a numeric probability and ‘uncertainty’ within the context of predictive genetic testing, suggesting that “risk prognostications are often uncertain rather than risky” (Bharadwaj, 2002, p. 230). In the case of HD, however, risk estimates are almost always certain: The risk of carrying the altered HD gene is 50:50 for every child of a parent affected with HD. Genetic testing then transforms that risk into 0% or 100% certainty of developing HD later in life. This is not to deny the importance of uncertainty in HD. As noted, even with definitive genetic testing outcomes, persons at risk for HD face uncertainty. They do not know when the disease will occur, which symptoms they will manifest, or how severe they will be. Nonetheless, this notion of uncertainty is distinguishable from the notion of

uncertain estimates of future disease inherent in many genetic testing situations, especially for multi-factorial diseases (e.g., breast cancer or diabetes).

Considering demographics

Lupton (1999a) argued that technico-scientific investigations of risk often underestimate the effect of the socio-demographic characteristics of their participants. Variables such as age, gender, occupation or geography, however, could have an important bearing on how people identify, negotiate and live with risks generally and with genetic risks in particular. Some of these variables did appear consequential in the current research. For example, participants in isolated areas of the province responded to genetic risk in a somewhat different way than those living in more urban areas. In part, response was determined by the availability of services such as counseling and other support (e.g., support groups). Age was also important in giving meaning to genetic risk, underscoring the temporal context of health and illness. For example, younger participants who had not yet married or had children assigned different meanings to genetic risk than those whose family was well established. And, while the current research did not set out to explore the financial burden of illness, economic considerations were paramount for some participants, notably some caregivers.

Beyond individual cognition

As noted, psychometric risk analyses are founded on the idea that humans are rational actors – individual information-processing units, taking in information about the world and using it rationally with minimal consideration of anyone or anything else. Douglas (1985; 1992) was particularly critical of the emphasis of this approach on

individual cognition. She contends, "The professional discussion of cognition and choice has no sustained theorizing about the social influences which select particular risks for attention. Yet it is hard to maintain seriously that perception of risk is private" (Douglas, 1985; p. 3). Douglas maintains that risk judgments and perceptions are culturally determined, rather than individualistic.

Alternatives to the psychometric approach - Socio-cultural theorizing about risk

Socio-cultural approaches to risk are very much concerned with the social and cultural contexts in which risk information is used and understood. Lupton (1999a,b) identified at least three socio-cultural risk perspectives: the 'cultural/symbolic' perspective, largely advanced by anthropologist Mary Douglas and colleagues; the 'risk society' perspective, led by sociologists Ulrich Beck and Anthony Giddens; and the 'governmentality' perspective, informed by the work of French philosopher, Michel Foucault.

Undoubtedly, these perspectives have salient differences; however, all share some common assumptions. All see risk as a central concept by which contemporary Western societies (individuals and institutions) are managed and monitored. Lupton (1999a) also suggested that all perspectives see risk as a pervasive, central aspect of human subjectivity and each perspective acknowledges that risk is related to ideas of responsibility and blame, a relationship which informed the current research.

In socio-cultural writings on risk, a range of social constructivist positions are represented, ranging from the fairly weak to the strongly held. Regardless of the strength of the position, constructivists espouse the *subjective* construction, interpretation and use

of risk discourse. For constructivists, a risk is not fully objective or knowable outside of the socio-cultural context in which it is identified (Lupton, 1999a). "A risk, therefore, is not a static, objective phenomenon, but is constantly constructed and negotiated as part of the network of social interaction and the formation of meaning" (Lupton, 1999a, p. 29). Beck's (1992) 'risk society' perspective can be thought of as a weak social constructivist position; Beck suggests that 'real' risks exist, but these are mediated by social and cultural processes (Beck, 1995).

Broadly speaking, this was the approach taken in the current work as well. Risk of carrying the altered HD gene does objectively exist, but interpretation of, and response to, genetic risk was subjective and varied depending on a number of social and familial processes. Discussing the conflict between social constructivism and positivism, Finkler (2001) noted:

In the end, whether cultural beliefs and practices are socially constructed or rooted in an objective reality may arguably be less significant than how the individual, as agent, experiences and negotiates them. To assess this, we cannot simply theorize but must meticulously observe and 'carefully listen' to the culture bearers (p. 260).

The current research was an attempt to 'carefully listen' to at risk individuals as they told the story of their genetic inheritance. Merely asserting that genetic risk was a social construction seemed inadequate given the goals of the current research. Rather, to talk in any meaningful way about risk, social scientists must, "...go beyond the conception of risk and technology as mere social constructs and grasp instead how specific technologies are lived as future-creating social praxis and in what way particular risks are experienced, perceived, defined, mediated, legitimated, and/or ignored" (Adam

& van Loon, 2000, p. 6). The current research was an attempt to 'grasp' how genetic testing technology influences current and future agency and to understand how genetic risk is experienced on a day-to-day basis.

'At risk' in Beck's risk society

Joffe (1999) noted that investigations of the subjective experience of risk in the context of risk society were limited. However, aspects of Beck's 'risk society' thesis seemed applicable to the current research. For example, Beck (1992; 1994) has argued that current industrial society is one in which discussion and debate about risk dominates all spheres – political, public and personal. This appears likely regarding developments in the new genetics (e.g., Bell, 1998; Kerr, Cunningham-Burley, & Amos, 1997; 1998; Lock, 2000). Those of us living in this time are forced to deal with risk on a daily basis. However, the risks in late modernity differ from those of earlier times: According to Beck, today's risks are global and of greater magnitude, sometimes threatening the destruction of all life on earth (e.g., nuclear disasters or toxic foodstuff; Beck, 1995). The global nature of these risks makes it more and more difficult to quantify and/or prevent them. Beck (1996a) argued that the risks of late modern society are not easily calculable because of their macro nature and long-lasting effects: "...to express it by reference to a single example: the injured of Chernobyl are today, years after the catastrophe, not even born yet" (Beck, 1996a, p. 31). This notion has parallels in new genetic developments (e.g., the use of genetic testing to identify mutated genes in fetuses, or the use of genetic engineering to enhance food, animals, or for that matter, humans).

Beck (1992) further contends that assessing exactly what is a 'risk' is also more difficult today – it is often impossible to perceive risks with the naked eye (e.g., chemical warfare or nuclear weaponry). This is also problematic for developments in the new genetics (e.g., risks associated with genetic engineering or genetic testing). Such risks exist largely in the scientific sphere and are open to contest and debate. Beck suggests that science is no longer regarded as an expert authority. Rather, individuals have become skeptical about science since science itself has created many of the risks they now face and cannot solve the problems it has created (Beck, 1994). In a recent article, Beck and colleagues noted, "The institutionalized answers of first modern society to its self-produced problems – for example, more and better technology, more economic growth, more scientific research, and more specialization – are less persuasive than they once were, although it is not at all clear what should take their place" (Beck, Bonss, & Lau, 2003, p. 6).

There is evidence that the lay public questions science in general (e.g., Hipkins, Stockwell, Bolstad, & Baker, 2002) and the new genetics specifically (e.g., Kerr et al., 1998). Some participants in the current research also evinced skepticism and mistrust about genetics research. Although, this was not uniformly true. For others, science was a source of hope, possibly salvation, if not for them, for the next generation. Prior studies of women genetically at risk for breast cancer (e.g., Kenen, Ardern-Jones, & Eeles, 2003b) also found great trust in science and medicine, both in terms of current cancer care and hope for future cures.

Risk society and individual responsibility

Notwithstanding the trust in science displayed by some participants, Beck's risk society is one in which the very features of modern society that once secured social progress (e.g., education, government, medicine or science) rebound on themselves in such a way as to undermine their dependability (Bradely & Morss, 2002). Society is beginning to reflect upon and critique itself, such that the central institutions of modernity – technology, science, government – are now marked as the primary risk producers. Beck calls this process reflexive modernization. "Reflexive signifies not an 'increase of mastery and consciousness, but a heightened awareness that mastery is impossible'" (Latour, 2003; cited in Beck et al., 2003).

As a result of this process, traditional social categories such as class, gender, the family, marriage or education no longer reign in risk society (Beck, 1992). There is, instead, a trend toward individualization. Risk society is characterized by the loss of traditions and social bonds "as a means of structuring the life-course and forming personal identity" (Lupton, 1999b, p. 4). It is now the task of each individual to invent his or her own self-identity. In risk society, an individual's biography will be a do-it-yourself project. Thus, risk society is replete with choices. In this society, risk is understood as human responsibility, rather than the outcome of God, nature or supernatural forces, as was the case in earlier times.

A key result of this "dynamic of individualization" argued Beck (1992), is anxiety and uncertainty. People in risk society live with an awareness of the myriad choices to be made in writing their biography, but also an awareness of "the different and contradictory

consequences and risk” associated with particular identities and choices. For example, an individual who chooses to smoke in risk society is aware of the consequences of this choice, namely, the frowned-upon identity of ‘smoker’ and the associated blame for any ill health caused by one’s smoking. Thus, the ‘dynamic of individualization’ is especially relevant in the context of health and illness, including genetic illness. Beck (1995) was particularly scornful of ‘choices’ emanating from genetic technologies, especially those ‘choices’ of would-be parents:

Once the state of the art determines the norm, abstention from choice becomes a luxury that no one possesses any longer. The dilemma of having to decide, and not being able to decide, between yes and no unfolds with inexorable rigour. The helpless parents find themselves once more burdened, one way or another, with the unconscionable responsibility of the godlike role of creation that accrues and is assigned to them through technology (p. 33).

Responsible ‘risky’ subjects

In risk society, as every person surely knows, there is a host of risk factors to be avoided in order to maintain good health: Poor diet, smoking, drinking, lack of exercise, and so on. Failure to avoid these risks is equated with blame for resulting sickness, a consequence known to those living in risk society. Giddens (1991), for example has noted that, “Risk profiles do not remain the special preserve of the experts. The general population is aware of them, even if it is often only in a rough and ready way” (p. 120).

Thus, the responsibility for avoiding and managing risk rests squarely on the shoulders of the individual. In the case of a genetic illness, such as HD, the responsibility for the affliction is placed not only on the individual, but also the family. “A person’s family medical history presents a formidable risk factor that people must negotiate, and is

one of the few in their lives over which they have no choice" (Finkler, 2003, p. 63). Note, however, that even though there is no 'choosing' one's genetic history and concomitant risk, developments in biotechnology make possible the 'choice' to have genetic testing, along with 'choices' regarding procreation, including the termination of affected fetuses. By attaching the label 'genetic' to illness, the affliction is no longer an individual matter. It is familial, a matter of family history and potential family future. In this way, genetic forms of thought introduce an additional burden to illness, that of 'genetic responsibility' (Novas & Rose, 2000) which affects critical life choices such as marriage, procreation, and career.

Kenen (1996) suggested that the at risk health status is accompanied by "a diagnostic invitation and the 'gift' of knowing" (p. 1546). Such an invitation, she argued, rests on the belief that knowledge is fundamentally good and will enable informed decision making. However, "The down side of this 'gift' is that knowledge is only empowering if it is beneficial. But this may not be so when diagnosis merely reaffirms risk, but offers no cure in the near future" (Kenen, 1996, p. 1546).

Such is the current context of genetic testing for HD. Cox (1999) argued that this 'gift of knowing,' supported by the 'discourse of potential benefits' (Boutte, 1988) surrounding genetic testing was part of a larger meta-narrative about the value of information in risk society. She noted it is this narrative which, "attaches a particular moral worth to information which facilitates rational planning and, at least the appearance of, choice" (p. 87). Some participants considering testing in this study were keenly aware that the 'gift of knowing' could not provide treatment or cure if they were

found to carry the altered HD gene. Regarding testing, they wondered, "There's nothing you can do, so why bother?" For them, there was little perception of 'choice.' But does this reasoning guarantee acquittal for genetic illness or for transmitting it to the next generation in the current cultural climate of individual and 'genetic' responsibility for health? Novas and Rose (2000) suggested that the focus on the genetic basis of disease "creates an *obligation* to act in the present in relation to the potential futures that now come into view" (p. 486, my emphasis).

Being healthy is a moral virtue

Moral theories of illness are not new. Whether illness was seen as the result of some punishment from God or supernatural beings or the result of 'behavioural culpability,' the notion of responsibility for health and illness has a long history. Galvin (2002) suggests, however, that "it is the transformation of the notion of behavioural culpability into our current obsession with health and fitness, and the accompanying belief that both are a matter of individual choice, which now predominates and results in a new culture of victim blaming" (p. 109).

For the chronically ill, such as those manifesting symptoms of HD, this context of responsibility and blame can have rather severe ramifications. In risk society, it is becoming less acceptable to be ill. Illness does not sit comfortably with the image of the 'good' citizen, someone who is self-reliant and autonomous - a rational decision-maker who is an active participant in civic, economic and social life. In this context, chronic illness or being at risk for a chronic illness could be perceived as some sort of moral failing.

The introduction of the Lalonde Report (1974) in Canada and the Surgeon General's report, *Healthy People* (1979) in the United States, marked the genesis of "health promotion." The early vision of health promotion was disease-, rather than health-oriented, defining health as the absence of disease and constructing disease as being associated with generally known and controllable risk factors such as poor diet or smoking (Minkler, 1999). Both reports did, in fact, include a strong focus on the social and environmental determinants of health. However, it was the 'healthy behavior/lifestyle' determinants which came to be cited most often by others; subsequently, the reports were used to defend and support the focus on individual responsibility for health (Minkler, 1999).

In the current cultural context, adhering to the lessons learned from health promotion has become a moral duty, and illness, a moral failing. To give credit where due, it would be foolhardy to ignore some of these lessons. There is no argument that lessons stemming from health-promotion programs can be beneficial to people's health. However, I concur with Galvin (2002) who argued, "What is in contention here is the attitude which is rooted in a twist of the logic of the responsibility thesis which includes the premise, sometimes hidden, often blatant, that, if a person *does* become ill, it is necessarily the result of faulty behaviour" (p. 112, emphasis in original). The seriousness of this contention is reflected in the experience of those suffering from chronic illness. If sufferers are blamed for their affliction, they are faced not only with the burden of the illness itself, but also with the added burden of 'moral reproach.'

Note that it is the concept of 'risk' which plays a major role in conferring responsibility for illness onto the individual. As noted, in risk society, we are faced with myriad choices in relation to health and illness for which we must bear responsibility. These choices are often fuelled by scientific knowledge and research (e.g., the choice to have a genetic test). Beck-Gernsheim (2000) has argued that health is a universal value in Western society, and as such, paved the way for genetic technologies. "By referring to health, obstacles are pushed aside, doubts are allayed, critics are silenced (or fall silent of their own accord)" (p. 126-27). Galvin (2002) noted that as a concept, risk derives its ability to define illness as a matter of personal responsibility in a multitude of ways:

Risks are conceivably everywhere and our growing knowledge of the statistical correlations between illness and various behaviours results in a seemingly endless chain of possibilities for intervention. What we eat, how we move, where we work, whether our relatives suffer from health problems to which we may also be predisposed and even how we think are sites of possible risk (p. 120).

As a concept, risk encourages us to seek expert advice, read self-help books, attend fitness classes, and arguably, have a predictive genetic test, among other 'healthy' choices. As a result, those who are ill and will not recover (e.g., people affected with HD) could be perceived in a very negative light. Note also that in this age of genetic testing and screening, future illness can be detected long before physical symptoms ever manifest. Genetic interventions, such as predictive genetic testing, could serve to reinforce the current mantra of individual responsibility for health, not only for oneself, but also one's offspring.

Attitude towards genetic testing

In light of the preceding discussion, it is useful to ask, 'What is the prevailing attitude toward new genetic technologies such as predictive testing?' Is there any evidence that at risk families will be held accountable for their genetic 'taint'? Are they held responsible for managing their risk and preventing it from infiltrating subsequent generations? Frankly, we do not know since empirical research in this area is in its infancy. Specifically, studies of attitude towards testing for late-onset neurogenetic illnesses such as HD are scarce (Evers-Kiebooms, Welkenhuysen, Claes, Decruyenaere, & Denayer, 2000). Nonetheless, some existing research provides a starting point for speculation about these issues.

It has been argued that the dominant discourse of individualism, "masks strong cultural pressures to make particular decisions" (Cunningham-Burley & Boulton, 2000, p. 180-181). For example, in one study of HD, 75% of partners of HD patients were in favor of prenatal diagnosis of the disease (Evers-Kiebooms, Swerts, & van den Berghe, 1991); however, only 29% of at risk individuals were (Bloch, Fahy, Fox, & Hayden, 1989). A later study found that among pregnant women at risk for HD, only 30% requested prenatal testing; some women withdrew before having the test, and only 18% (of 38 couples) actually performed it (Adams, Wiggins, et al., 1993). All but one of the increased risk pregnancies was terminated. Evers-Kiebooms, Nys, et al. (2002) examined the subsequent reproductive choices of 451 HD test candidates in the European Collaborative Study. The percentage of non-carriers with one or more pregnancies was higher (28%) than in carriers of the altered HD gene (14%). A more refined analysis of

the data revealed that of the 57 carriers who endorsed 'reproductive decision making' as a motive for testing, 18 had a subsequent pregnancy. The total number of pregnancies in the 18 carriers was 31. In these 31 pregnancies, 20 prenatal tests were performed, with 12 fetuses found to be carrying the altered HD gene. All 12 were terminated. It should be noted, however, that most people who eventually develop HD already have children of their own before the disease begins (O'Shea, 1997). Thus, for many persons with HD or at risk for the illness, reproductive decisions have already been made.

The Human Genetics Commission reported on a comprehensive examination of the public's attitude towards genetic information using the People's Panel (i.e., in-depth, structured interviews; see www.hgc.gov.uk for a copy of the report). In that study, a total of 1038 members of the U.K. public were interviewed. Some findings may lend support to the argument that at-risk persons could face 'cultural pressures' to make certain decisions (Cunningham-Burley & Boulton, 2000). For example, 75% of respondents agreed that people *should* be encouraged to be tested in young adulthood for adult-onset disorders such as HD. Thus, there is some evidence that the public holds individuals accountable for determining their genetic status when such testing is available. Fifty-seven percent of the sample also agreed that parents could use genetic testing information to decide if children with disabling conditions are born, although 25% disagreed. Potentially indicative of intolerance, 55% of those respondents aged 55 years or older, agreed that couples at risk of giving birth to a child with a serious genetic disorder *should* be discouraged from starting a family. Notably, there was a marked difference in opinion by age: Only 5% of those under age 25 agreed with this statement, underscoring the

significance of demographic variables in perceptions of genetic testing and at-risk persons.

Respondents also had definite opinions on the uses of genetic information. In addition to that noted above, 67% and 72% agreed that genetic testing should be used to develop ways to correct defective genes in individuals and to develop ways to correct defective genes in future generations, respectively. However, 70% thought it was inappropriate for an employer to obtain the results of genetic tests for the purpose of knowing whether an employee would develop an adult-onset disorder. Similarly, over three-quarters of respondents agreed that insurance companies should not be able to obtain genetic test results to set insurance premiums.

Specific to HD, an early study found that 82% of Scottish general practitioners (GPs) were in favor of predictive testing for HD, while 16% were undecided and 2% were opposed (Mennie, Holloway, & Brock, 1990). Comparatively, 68% of Dutch GPs were in favor of genetic testing for HD, while 26% were unsure and 6% were opposed to such a test (Thomassen, Tibben, Niermeijer, van der Does, van de Kamp, & Verhage, 1993). Recently, Elger and Harding (2003) surveyed Swiss law and medical students (i.e., future lawyers and physicians) about their views on genetic testing for HD, sterilization and other measures to reduce the frequency of the altered gene. Almost all participants (94%) agreed that "Genetic screening of a fetus 'at risk' should be proposed systematically to women concerned in order to proceed, if the women desire, to the abortion of a fetus carrier of the HD mutation" (p. 338). Notwithstanding its poor wording, Elger and Harding (2003) correctly noted that this statement was stronger than

simply informing women such a test exists. Law and medical students differed in their views on diminishing the frequency of the altered HD gene: 73% of the law students versus 39% of the medical students agreed that society should do everything possible to diminish the frequency of the HD gene, including recommending sterilization for carriers of the altered HD gene.

Eugenic attitudes have been recorded in regard to prenatal diagnosis. For example, 91% of Chinese geneticists agreed that a woman at risk of having a child with a genetic illness should undertake prenatal diagnosis (Mao, 1998). In the United States, only 38% of genetics professionals agreed with prenatal diagnosis for at risk women (Wertz, 1998); however, over three-quarters of primary-care physicians agreed (p. 502). Wertz (1998) has distinguished between 'individual eugenics' (e.g., directive genetic counseling) and 'social eugenics,' the latter referring to social pressure about genetic testing in the idea of 'responsible parenthood.' For example, majorities of genetics professionals in 19 of 36 nations (predominantly from Asia, South America, and Europe, but also 26% of U.S. geneticists, 55% of U.S. family physicians, and 44% of U.S. patients) agreed with the statement, "...it is socially irresponsible knowingly to bring an infant with a serious genetic disorder into the world in an era of pre-natal diagnosis" (Wertz, 1998, p. 501). Majorities in 24 nations agreed that, "It is not fair to bring a child into the world with a serious genetic disorder" (p. 501). Only 40% of Canadian genetics professionals agreed with this statement; however, this remains a substantial percentage.

In light of findings such as those noted above, it is informative to ask whether at-risk persons felt coerced to take the genetic test for HD (e.g., from friends, family,

medical professionals or society at large). It is also worthwhile to discuss perceptions of responsibility and accountability regarding genetic testing, and the current research did include discussion of these issues with participants. Taylor (2004) has rightly noted that we do not yet have a complete picture of societal attitudes towards genetically at risk individuals. However, Binedell, Soldan, and Harper (1998) suggested, "As predictive genetic testing becomes more commonplace and receives greater public attention, public opinion and subjective norms may exert a growing influence on decisions about testing" (p. 496).

CHAPTER 3

LIVING 'AT RISK' AND BEYOND

In this chapter, I review the somewhat limited literature on living at risk for, or with, HD. It must be noted that living 'at risk' for HD is not the same as clinically manifesting symptoms of HD. As the title of this dissertation suggests, and as one participant adamantly asserted, "I have the gene, but I don't have HD." Temporally, there comes a shift from living at risk to the chronic state of being a person with HD (that is, for those who *do* go on to develop HD). Qualitative studies of living at risk for HD and/or testing positive for the altered HD gene are beginning to emerge, and this literature will be reviewed shortly. I am unaware of any research (qualitative or otherwise) that focused on HD as a chronic condition. However, this shift in identity is important. For example, it will be shown that the shift has implications for perceptions of stigma. It is also significant in understanding the meaning of genetic risk and illness for other family members, especially those who care for the person with HD.

Chronic Illness

In a now classic article, Bury (1982) suggested that chronic illness is a form of biographical disruption. That is, a disruptive event or 'critical situation' which upsets the routines and structures of everyday life, the store of knowledge which underlies them, and initiates a rethinking of a person's biography. Illness often necessitates a re-examining of the self ('why me?', 'why now?') and of interpersonal relationships. It can involve an upset of roles and norms, and the rethinking of future expectations and plans. "Chronic illness involves a recognition of the worlds of pain and suffering, possibly even

of death, which are normally only seen as distant possibilities or the plight of others” (Bury, 1982, p. 169). For some at risk for HD, and for all who have tested positive for the altered HD gene, these states of pain and suffering are no longer ‘distant possibilities.’ They will be definite realities, as they are *current* lived realities for persons clinically affected with HD and their caregivers. Echoing Bury’s (1982) respondents affected with arthritis, however, “There is rarely anything in the individuals’ biography which provides an immediate basis for recognition of the illness *as* illness” (p. 171).

Accordingly then, problems with the onset of symptoms and the recognition of a chronic illness, such as HD, are common. Participants in the current research spoke at length about the misdiagnosis of relatives, especially when there was no documented family history of the illness. Several visits to family doctors were often necessary before a diagnosis was confirmed, usually by a neurologist or geneticist. Bury (1982) suggested that seeking medical confirmation provides an opportunity to think about illness as separate from the self, the objectivity of medical science providing justification for the effects or symptoms of the illness. However, Bury (1982) noted an uneasy balance in his interview respondents as they tried to view the illness as an external force, but were forced to acknowledge its invasion in all aspects of life. They were ambivalent about seeing medical specialists – a specialist could confirm diagnosis, but could not provide much in the way of treatment or a cure. “Medical intervention was, therefore, regarded at the same time as both important and limited” (Bury, 1982, p. 173). Participants in the current research expressed similar sentiments.

Bury (1988) suggested that two types of meaning converged around chronic illness. The first meaning of chronic illness revolves around the practical *consequences* of the illness for the affected individual and other family members. In this meaning, the effects of disruptive symptoms on everyday life (including at work and at home), time devoted to management of symptoms and the financial burden of the illness (both now and in the future) are the priorities. Second, the meaning of chronic illness also resides in its symbolic significance (Williams, 2000). Bury (1988) suggested that illness carries with it different connotations and imagery, which will differ by condition. The symbolism attached to an illness has a profound effect on self-identity and how chronic illness sufferers think they are perceived by others (Williams, 2000). Scrambler and Hopkins (1988), for example, described how parents of children with epilepsy worked to conceal the family's "dark secret" for fear of social rejection and stigma. Some participants in the current study described how others thought their affected relative was drunk. This is a common misperception about persons exhibiting the choreic movements associated with HD. The Huntington Society of Canada (HSC) works to address this perception. A promotional pamphlet, for example, has the headline, "He's not drunk. He has Huntington disease."

While the bodily limitations and restricted activities due to chronic illness cannot be ignored, there is reason to suspect that not every chronic illness will be as 'disruptive' as others (Williams, 2000). For example, much of the chronic illness literature rests on adult-onset diseases which may be unanticipated, throwing normal life out of kilter. Alternatively, however, age can mediate the experience of, and response to, chronic

illness. Thus, when illness strikes at a later age, people could have a lifetime of experience and coping mechanisms with which to respond to chronic illness. In addition, illness may be expected with age or at least met with more acceptance. As such, "...chronic illness, particularly in the context of a lifetime's general hardship and adversity, may be a biographically *anticipated* rather than a disruptive (i.e., unanticipated) event" (Williams, 2000, p. 51, emphasis in original). If we apply this notion to HD, for some genetically at risk individuals, the onset of HD could be more anticipated than it is disruptive, especially for those who have sought testing earlier in life. Time is available to prepare for future chronic illness. This is a distinguishable feature of some genetic illnesses compared to chronic illness in general.

Chronic, but distinguishable, illness

Street and Soldan (1998) provided a conceptual framework of the range of psychosocial issues faced by families with genetic illnesses based on Rolland's research on chronic illness. Rolland (1994) identified five elements in relation to chronic illness that present a variety of psycho-social demands: Onset, course, outcome, incapacitation and uncertainty. The second of these elements, time phase of the illness (including the crisis, chronic, and terminal phases), is not sufficient to account for the *pre-illness phase* (Street & Soldan, 1998) of some genetic illnesses. This phase is especially relevant for persons at risk for HD and for those who have tested positive for the HD gene. During this phase, no physical manifestations of the disease are observable.

Following Street and Soldan's (1998) suggestion, Rolland (1999) later distinguished between the *pre-symptomatic* (1) pre-crisis, (2) crisis, and (3) chronic

phases in genetic illness. The former refers to life before a genetic test is available or even considered by family members. At this time, however, families could already know about the history of genetic illness in the family and members can have strong beliefs about their own vulnerability to the illness. The pre-symptomatic *crisis* phase begins for many families when a predictive test actually becomes available or when members actively consider taking the test. This phase extends to the entire decision making process and subsequent to the test. Finally, Rolland (1999) suggested that the pre-symptomatic *chronic* phase is similar in many respects to the chronic phase of living with chronic illness, and can extend over a person's lifetime.

While HD itself is a chronic illness, those at risk for HD and those testing positive for the altered HD gene can be thought of as in the pre-symptomatic pre-crisis or crisis phases, each with attendant psychosocial tasks and demands. This conceptualization is useful for understanding how at risk or tested positive persons give meaning to genetic risk and how that meaning can change over time. Other authors have distinguished chronic *illness* from chronic *risk*, which also helps distinguish genetic illness from illness in general.

Chronic illness versus chronic risk

Kenen et al. (2003b) have suggested a 'chronic risk' perspective for understanding how at-risk persons perceive and respond to genetic risk information. "The concept of chronic risk helps explain the lived experience of *risk* of illness, as opposed to illness itself." (Kenen et al., 2003b, p. 316, emphasis in original). This distinction is important since advances in genetic medicine intimate more and more of us will live 'at

risk.' While all residents of risk society feel they are at risk for something, "...the perception of chronic risk remains quiescent for many members of society" (p. 317). However, this is not always possible for genetically at risk individuals and families.

Kenen et al. (2003b) suggested that at-risk persons experience changes in identity, behavior and interpersonal relationships that are comparable, but not identical, to those observed in persons suffering from chronic illness. The primary difference between chronic illness and chronic risk, according to these authors, is perception. Those at chronic risk must cope with *thoughts* about the illness, including *possible* body deterioration, rather than actual deterioration. In their interviews with women genetically at risk for breast/ovarian cancer, for example, Kenen et al. (2003b) noted that most of the women perceived their risk as ongoing: "It was pervasive and chronic, though at times intermittent" (p. 321). Women were aware that their chronic risk was in the back of their minds most of the time, but they wanted to "just get on with it." Similar findings emerge from interviews with persons at risk for HD. The notion of living life, rather than passively waiting for symptoms of HD to emerge, is part of the negotiation of the at risk status. As one participant put it, "I can't give up my life because I have the gene for HD."

Negotiating the at risk identity

At risk individuals are caught in a state of limbo, somewhere between healthy and ill. While quantitative studies abound on the psychological implications of testing for HD, very few empirical studies have investigated the everyday lives of individuals at risk for, or living with, HD (Hunich, 2001; 2003; Taylor, 2004). As noted, the clinical (largely quantitative) body of work suggests that while emotional reactions of anger,

sadness and/or anxiety are common immediately following test results, levels quickly return to baseline. Qualitative studies, on the other hand, reveal a much more complex and dynamic picture.

Taylor (2004) argued for investigations of 'lay' understandings of risk, which account for the broader social and familial contexts within which test decisions are made and experiences of risk are located. While the current research was not specifically about the decision to take the genetic test for HD, the very availability of genetic testing makes genetic risk salient for many individuals. Participants in the current study often spoke spontaneously about their testing experiences, whether they were tested or not. Early studies of attitude toward, and interest in, genetic testing for HD revealed high levels of test intention (Evers-Kiebooms & Decruyenaere, 1998). As noted, however, test uptake has been substantially lower than initial indications of test interest. From a theoretical perspective, the decision to take a genetic test has been investigated most often in the framework of social cognition models. As a result, much of the psychometric risk research in the genetics context is underscored by individual cognition. Participants typically provide self-reports of attitudes, knowledge, risk beliefs, control beliefs, susceptibility beliefs, severity beliefs, and so on regarding a genetic illness. These are usually combined in an expectancy-value model to predict intention to take a genetic test. Percentage of variance in actual test behavior accounted for by these sorts of variables is typically low (Etchegary, 2004). As additional genes are identified, the 'clinical gaze' extends to the family, producing what Finkler (2001) calls a 'medicalization of kinship.' This intimates that more and more medical decisions are a family, as opposed to an

individual, matter. Risk research which is *not* underscored by individual cognition has revealed much broader social and familial contexts in which test decisions are made and genetic risk experienced.

To test or not to test

An immediately apparent difference between quantitative and qualitative analyses of living with genetic risk is the recognition by at risk individuals of the significance of the test decision. Qualitative studies of genetic testing for HD reveal that at risk individuals, whether or not they choose to be tested, are acutely aware of the import and impact of the availability of genetic testing (Taylor, 2004). This medical test is regarded as unique and serious when compared to other health and life decisions. Persons at risk for HD know the genetic test can provide them with critical life information, not only for themselves, but their entire family, including future generations (Chapman, 2002; Taylor, 2004). Qualitative investigations of decision making in this context, few though they are, also reveal the multiple contexts within which test decisions are taken.

Cox (2003), for example, provided a thoughtful analysis framed in narrative theory of *how* individuals take the decision to have the genetic test for HD. Participants in her study were clearly influenced by family history and interpersonal relationships in their decision to have the test. In clear contrast to the individualistic, rational-thinker approach to decision-making, Cox argued:

Choices are always hedged in by constraints; we are not free to decide upon just any course of action nor are we ever positioned in such a way that we can see what the full range of choices might consist of. As mothers and daughters, fathers and sons, sisters, brothers, aunts, uncles, cousins, spouses, life partners and friends, we exist in and through our social and familial ties with others. For those

at risk individuals who are in the process of deciding whether to request predictive testing for HD, such social and familial ties loom large (McKellin, 1997) (p. 262).

Cox's work is also notable in that beyond revealing that decision-making was not always a conscious and rational act, for some individuals, the perception of choice was not an important component of decision making at all. Psychometric risk research normally frames the experience of deciding to take a genetic test as a 'decision' – implying there is an opportunity for choice. Cox (2003), however, found evidence of 'having to know' in the stories of some people at risk for HD. For them, the decision to take the test was a 'self-evident act.' That is, for some, there was only one appropriate 'choice' which required minimal-to-no thought or discussion. Similarly, an in-depth interpretative phenomenological analysis of the decision to have the test for HD also found that the decision was described as 'automatic' or 'already made' before the at risk individual even had an initial clinic visit (Smith et al., 2002). Some participants in the current research revealed similar stories (see Chapter 8).

In contrast to those who 'had to know,' Cox (2003) also found evidence of 'evolving toward' the decision to take the genetic test for HD. In these narratives, at risk individuals had usually known for some time about their family history of HD. For these participants, the decision was a dynamic process where they moved from initially being either opposed to, or ambivalent towards, the idea of genetic testing, to a period of considering the ramifications of the test for themselves and others. Gradually, they felt ready to seek testing. These differing narratives about decision making help reveal the complexity, seriousness and fluid nature of genetic-test decisions.

The nature of these decisions is reflected in a recent publication of the Huntington Society of Canada (HSC; Cox, 2002). The booklet contains stories written by individuals throughout Canada about their own experiences with genetic testing. Some were still undecided about having the test, others were going through the testing process and others provided retrospective accounts after they had learned their test results.

The stories reveal a remarkable diversity in response to HD risk, underscoring the earlier argument that there is no such thing as *the* HD person or *the* HD family. For example, one individual who felt no need to know his genetic test results wrote, "I cannot think that either knowing I have or have not inherited the gene for Huntington's would change the way we live and are raising our children. Huntington's is not the only debilitating condition one can suffer with" (p. 8). Another wrote:

HD to me is cruel and unsympathetic to both the victim and the family...for me, predictive testing is something I really don't want. I really don't worry about getting HD. I also don't want to know if or when I could develop this disease. There's no cure for HD so why sit and wait for my hands to shake? (p. 11-12).

Another story would seem to fall within Cox's (2003) 'evolving toward it' narrative. A woman recounted how she originally had decided against testing as she was young, newly married and "HD was far away." However, many years later, that decision had changed:

I don't know if at 20 years I would have been tested, but now I find myself wanting to know the truth. I feel positive about getting tested; it will give me a sense of control over my life. It will help me to make some important decisions about my future and my family's future (p. 16).

This narrative reveals the dynamic nature of decision making about genetic risk. And, other narratives revealed the sometimes unexpected reaction to a favorable result

(i.e., testing negative for the altered HD gene). One man, for example, commenting on his negative result said, "I was relieved, but we honestly weren't expecting that result. I've been carrying this monkey on my back for about 24 years and it has been such a big part of me that I just can't throw it off my shoulder, walk out and do a few cartwheels" (p. 22). Just as notable, there is no single reaction to a positive result either. One woman commented on her reaction to learning she had inherited the altered HD gene. "I didn't have an emotional reaction, I didn't break down. And I still haven't" (p. 27). She went on to recount the follow-up phone call from a genetic counselor two weeks after the result:

They wanted to know how I was doing and if I was okay. And I'm thinking, 'What the hell's going on here? Is there something wrong with me?' Are they really looking realistically at what people are doing and dealing with and how it's affecting their lives? Before your test results, you have to fill out a package of questionnaires. I might understand the relevance of questionnaires if they were personalized but I don't like being forced to choose one or the other answer if it doesn't relate to me (p. 28).

For another woman, however, the positive test result was devastating:

I was devastated. Hearing my test results was like hitting rock bottom. I was pretty upset emotionally, and I do have days like that now. People don't usually get to know what's going on inside the heads of people that have it. The doctors are very clinical and they're into that kind of clinical arena. They never really touch the emotional impact or what's happening inside a person's head or heart (p. 44-47).

Two years later, however, this woman had found a new sense of direction. "For me the whole experience has been kind of like a gift. I've taken everyday and made that day special for myself" (p. 47).

These brief excerpts are revealing in that they clearly demonstrate there is no single response to genetic risk information. The usual blanket finding in most clinical risk

research (testing evokes short-term emotional reactions which return to baseline shortly after receiving test results) seems a rather inadequate and somewhat bland description of the ‘effect’ of risk information.

Like these stories, risk research which is not conducted in a psychometric paradigm reveals the importance of timing and context in deciding to have the genetic test for HD. Many individuals, for example, have no prior knowledge of HD in their family, and finding out about the illness often came at a later stage in life when reproductive and career decisions had already been made (Chapman, 2002). For these people, there is normally minimal knowledge about HD transmission and disease progression. Many already have children of their own, and the decision to test takes on a moral dimension as they come to realize the meaning of their own risk for that of their offspring (Chapman, 2002; Taylor, 2004). Participants in Chapman’s (2002) study, for example, had all tested positive for the altered HD gene. Some suggested they were tested primarily for their children, providing the next generation with accurate knowledge to be used in the planning of their own families. Similarly, Taylor (2004) observed many references by her participants as to what they “should” do or what was the “right thing to do.” Taylor suggested there was a moral sense of responsibility in some individuals at risk for HD. This is likely not only for those who have children, but for childless, unmarried persons as well; the latter often struggle with revealing their genetic risk to future life partners. These socio-cultural analyses of risk reveal an ethical dimension of decision making that is largely ignored or uncaptured in psychometric studies of genetic risk and decision making.

Beyond the test decision - What is 50:50 anyway?

Very few studies have explored the meaning of risk in persons at risk for HD. Smith et al. (2002) investigated risk perception in a small sample of people at risk for HD. Their participants revealed complex and contradictory interpretations of their genetic risk. In Mendelian terms, a 50:50 risk of carrying the altered HD gene appears rather straightforward. Smith et al. (2002), however, found the simple starkness of this figure difficult to negotiate; as a result, their participants suggested that an uneven risk (e.g., 70:30) might be more acceptable. Some of their participants sought information about their family that actually transformed the risk to 70:30. This was sometimes accomplished by mistaken lay theories of inheritance (e.g., HD skips a generation). Binedell et al. (1998) also found evidence of risk transformation in their participants at risk for HD. Some at risk individuals explained how they shifted their 50% risk towards greater certainty. One at risk individual, for example, suggested that while she knew her risk was 50%, she feared it was 75% since she was “like her mother.”

Similarly, Cox and McKellin (1999) found their participants interwove social and biographical information into their understanding of Mendelian risk in order to render their risk subjectively meaningful. For example, one female participant in their study was at 25% risk for HD. While she knew her risk of carrying the altered HD gene was one in four, family experiences of, and proximity to, the disease transformed her objective risk of 25% into a far more complex risk calculus: “Although when I speak of it as one in four, mentally it’s not...I couldn’t put a figure on it, one in twenty, whatever, quite a bit

less because of my mother's age and because I've never seen or been exposed to it" (p. 636).

Risk information is sometimes incorporated into daily life through processes which downplay the objective risk status. For example, a qualitative study of perceptions of familial hypercholesterolaemia (a dominantly inherited condition which is a precursor to heart disease) found that downward social comparison processes relegated the risk of heart disease to the background (Senior, Smith, Michie, & Marteau, 2002). Participants in that study compared their condition to other more serious genetic conditions, effectively downplaying their own risk status. Even in individuals at risk for HD, threat minimization can also occur with underestimates of HD risk reported (Codori & Brandt, 1997). These authors have suggested that individuals at risk for a serious, fatal genetic disorder such as HD, focus not on the disease itself, but on controllable outcomes, such as their ability to cope with the illness.

Other coping strategies include outright denial (although, recall the earlier suggestion that some participants could be dismissing their risk, rather than denying it), rationalization ("I could be hit by a truck"), and attempts to suppress all thoughts of being at risk (Bloch et al., 1993). These responses are not necessarily maladaptive; they provide at-risk persons with time to slowly assimilate disease information. The literature on the social and familial implications of HD also reveals that symptom watching and patient pre-selection are common, illustrating the degree to which family life becomes "saturated" by beliefs about HD (Cox, 1999). For example, Korner and Fitzsimmons (1985) observed that parents of at risk offspring were preoccupied with monitoring their

child's physical and emotional behavior. Participants in the current study also referred to "checking" themselves and other family members for symptoms of HD.

As noted earlier, at risk individuals often pre-select which members of the family are going to develop HD (Evers-Kiebooms & Decruyenaere, 1998), especially in families where a parent is beginning to manifest early signs of HD (Cox, 1999). Such pre-selection could also explain why at risk individuals interpret their risk status differently from that suggested by Mendelian genetics.

Effects of the at risk label

Beyond psychological well-being

As noted, the majority of at-risk persons choose not to take the genetic test for HD: These individuals are at risk for HD. Little empirical work has addressed the lived effect(s) of this label. In one of the first studies with HD families, Wexler (1979) interviewed 35 individuals at risk for HD. At the time, no genetic test was available to at-risk persons. Wexler (1979) discovered that HD was often perceived as a "time bomb," imposing a burden of apprehension and waiting. A second notable finding was the significance of early childhood experiences of HD. All of the at-risk persons had known their affected parent and had watched the parent change with the inexorable progression of HD. This was normally marked by bizarre movements, slurred speech and other uncontrollable behavior. Wexler (1979) noted that if changes in the affected parent occurred during the formative years of childhood, these at risk people later had especially sinister visions of HD as adults. For some of her participants, the inescapable progression of HD seemed to "strike at the core of their physical and psychological self esteem" such

that many envisioned “a vision of a Frankensteinian monster, one who approaches others with affection but from whom others recoil in horror” (p. 201). This finding would seem to underscore Bury’s (1988) contention that illness is symbol-laden.

Conversely, in families where the affected parent was able to remain even marginally functional, at-risk persons were better able to cope with their status as adults. Empirical research on the psychological impact of predictive testing often neglects such salient factors as whether the at-risk person has grown up with HD, witnessed its progression and how this, in turn, shapes the symbolic representation of HD. In the current research, such contextual factors proved important for participants’ interpretation of, and response to, genetic risk information

For all at-risk persons, however, the nature of HD symptoms evoked fear and dread. Wexler (1979) found that the threat of cognitive decline was the most frightening aspect of HD in at-risk persons. Her participants were also concerned about becoming incontinent and the extreme dependency involved in becoming chronically ill. Underscoring the latter fear, married at-risk persons felt that such a great responsibility should not be inflicted on someone they loved. Younger at-risk persons, on the other hand, despaired of finding a life partner. This latter finding was echoed by Taylor (2004) many years later who also found in her younger, at risk participants a moral sense of responsibility to take the genetic test for HD and share the results with potential partners.

Underscoring the familial nature of HD, at-risk persons frequently expressed their thoughts and feelings about HD in terms of responses to, and of, other family members (Wexler, 1979). This was particularly evident in at risk parents with children of their

own. At risk parents express horror and guilt as they realize they could have passed the altered HD gene to their own offspring. They also know that whatever emotions they themselves feel towards their affected parent – pity, anger, blame, or compassion – could be one day directed at them. “As they watch their parents, they watch themselves; all emotions rebound” (Wexler, 1979, p. 203). Binedell et al. (1998) also found that HD is regarded as a burden to the family, more so than a burden to oneself. Over one third of at-risk persons in their study suggested the burden of care was the primary feature of HD.

More generally, HD is often a part of family identity; in one study, family members who took the genetic test for HD and tested normal felt a loss of identity and membership in the family (Sobel & Cowan, 2000). Testing normal meant they no longer shared the common bond of the at risk status with other family members who remained at risk. Sobel and Cowan (2000) conducted interviews with families at risk for HD. One of their participants who received a normal test result, but whose siblings tested positive for the altered HD gene explained, “I don’t want to be left out of the family because I’m not sick. My sisters are special people now...I had been part of this very elite group of people who may be very ill, and it was like a claim to fame. You’re a special person because you may be dying” (p. 53). As this quote demonstrates, the at risk status can be incorporated as an integral component of one’s biography. When this identity is threatened (e.g., with a normal test result), uncomplicated relief or joy is not always the initial reaction to the test result (e.g., Cox, 2003).

An earlier study stressing the importance of recognizing the needs of at risk family members argued that the *time* of diagnosis is particularly crucial (Yale &

Martindale, 1984). Diagnosis of a family member will generally necessitate family discussion of genetic risk and for some family members, it represents the first time they become aware they themselves are at risk. Yale and Martindale (1984) suggested this discovery may invoke shock, disbelief and a profound sense of threat.

The threat to self-identity could help explain why studies employing psychological instruments to measure clinical outcomes following testing (e.g., stress or depression) do not observe notable decreases in such emotional reactions. Kavanagh and Broom (1998) suggest that risks located within the body, rather than the environment or those imposed by lifestyle choices, have been neglected by social scientists. These embodied or corporeal risks are located in the body and, "...define who a person *is* rather than what they *do* or what is *done* to them" (Kavanagh & Broom, 1998; p. 442, emphases in original). Even when previously at risk individuals are found not to carry the altered HD gene, they sometimes suffer from survivor guilt, especially when siblings test positive (Evers-Kiebooms & Decruyenaere, 1998; Sobel & Cowan, 2000). They also know that as the 'normal' sibling, they will be responsible for helping to care for family members who have tested positive (Sobel & Cowan, 2000).

The at risk status can translate into a sense of urgency – an emphasis on living for the moment, despite the fact that there is a 50% chance that the at-risk person does not carry the altered HD gene. This supports Beck's (1998) contention that events which have not yet happened, and could never happen, strongly influence agency in the present. Thus, the effects of living at risk are sometimes reflected in educational, career, marital and reproductive decision making: Some forgo career or family, and these choices have

consequences that ripple through the generations affecting the life chances and decisions of at risk offspring (Cox, 1999).

For the person at risk for HD, time becomes especially relevant (Cox & McKellin, 1999; Taylor, 2004; Wexler, 1979). In fact, the notion of time passing is often part of the 'at risk' definition (Taylor, 2004) for those at risk for HD. At-risk persons become more vigilant at symptom watching as the age of onset draws closer.

Binedell et al. (1998) have suggested it is the uncertainty associated with HD, rather than the objective risk of carrying the altered gene which is most relevant to the at-risk person. Some participants in their study expressly referred to the uncertainty of living at risk. For others, however, uncertainty was preferable to the certain knowledge afforded by the genetic test. In fact, uncertainty allowed hope.

Some participants in the current study expressed similar reasoning: Not only does being at risk maintain hope for individuals, but for their children as well. For those persons who have tested positive for the altered HD gene, hope is no longer for themselves, but for their children. There is hope that science will find a cure. For these individuals who know they cannot escape HD, the hope is for a good death, similar to Little and Sayers (2004) cancer survivors:

If 'cure' is not the outcome, and the participant accepts that he [*sic*] is now dying, his hope and his discourse change their objective to *good death*, a death which confirms meaning in the dwindling life, with symptoms controlled, dignity preserved, worth recognized and important relationships confirmed (Little & Sayers, 2004, p. 1336, emphasis in original).

Salience of genetic risk

Cox and McKellin (1999) have urged social scientists to resist assuming genetic risk information is of utmost importance in everyday life. In the accounts of their participants, genetic risk for HD became salient only at certain critical junctures (e.g., the diagnosis of HD in a relative). For much of the time, however, risk of carrying the altered HD gene existed as part of the diffuse, taken-for-granted information about family life. This finding accords with health research more generally. Health-related concerns are not prominent in everyday life and thinking, despite the fact that risk society is one which demands a future-oriented outlook (Lawton, 2002; Lupton, 1999a).

Lawton (2002) interviewed a sample of UK residents about current and future health, including morbidity and mortality concerns. She found that health was taken for granted; indeed, even a diagnosis of disease was not enough to redefine oneself as 'ill.' Rather, there had to be some tangible presence of the disease. This is particularly relevant to persons at risk for HD. These individuals could be asymptomatic for many years. Indeed, Chapman (2002) noted how some of her participants who had tested positive for the altered HD gene explained they could push HD to the background given they were not currently showing any signs of the illness. Similarly, Bharadwaj (2002) interviewed persons at risk for haemochromatosis and found that since they did not feel ill, they did not consider themselves patients nor perceive themselves 'at risk.' The lived experience of the condition was invisible, thus the presence of the gene was perceived as largely inconsequential for many of her participants. The lived experience of HD is important

since the presence or absence of physical manifestations of illness could affect perceptions of stigma, to which we turn next.

CHAPTER 4

STIGMA – A CASE OF SPOILED IDENTITY?

One aim of the current research was to explore perceptions of stigma associated with being a member of a family affected by HD. I am unaware of any empirical research that has attempted to relate living at risk or testing positive for HD with social stigma. As will be elaborated shortly, however, there are anecdotal accounts of social stigma against individuals affected with HD. This chapter reviews some pertinent literature on social stigma, drawn primarily from social psychology and to a lesser extent, sociology. The purpose is to define stigma and discuss some of its attributes and consequences that could be relevant to persons affected with, or at risk for, HD.

What is stigma?

The majority of research on prejudice has focused on those who are prejudiced against the stigmatized, rather than the stigmatized themselves (Swim & Stangor, 1998). Only recently has empirical work on stigma from the target's perspective been integrated more fully into the social psychological literature (e.g., see Crocker, Major, & Steele, 1998, for a review). The current research added to this growing literature by investigating stigma from one particular target's perspective. To my knowledge, it represents one of the few empirical studies that investigated, (1) whether persons at risk for (or with) HD, along with their partners and caregivers, perceive themselves to be the targets of stigma and, (2) with what consequences. Swim and Stangor (1998) outlined important social consequences of adopting a target's perspective in stigma research, including giving a

voice to target groups and validating their experiences. It is hoped these were consequences of the current work as well.

One cannot talk about social stigma without mention of sociologist Erving Goffman. In a classic text, Goffman (1963) defined stigma as a “deeply discrediting” attribute which can “disqualify one from full social acceptance” (preface). In that early text, Goffman distinguished three types of stigma. The first included ‘abominations of the body,’ referring to physical deformities (such as missing a limb). Another included blemishes of individual character typically linked to personality or behavior, such as being mentally ill or homosexual. Finally, there were ‘tribal’ stigmas such as race, nation or religion. Goffman cautioned, however, that not all discrediting attributes necessarily lead to stigmatization. Rather, only those which are incompatible with our stereotype of what a given person *should* be, in other words, his/her social identity.

Later researchers concurred, suggesting that a stigmatized person was flawed or devalued in the eyes of others owing to membership in some social category (Jones, Farina, et al., 1984). More recently, Crocker et al. (1998) noted there was no defining feature that signified a person or group would be stigmatized. “If forced to provide a single defining feature of social stigma, however, we would argue that stigmatized individuals possess (or are believed to possess) some attribute, or characteristic, that conveys a social identity that is devalued in a particular social context” (p. 505). Thus, stigma is considered to be a *situational* threat - it is context dependent.

The context into which genetic testing has been introduced revolves around both (1) ‘risk,’ including its measurement, management and avoidance; and (2) individual

responsibility for health, with associated notions of culpability and blame. Residents of the current risk society *should* be healthy, independent and personally responsible for avoiding and managing the risks in their lives. In this context, it is conceivable that persons at risk for a genetic illness could face societal intolerance or discrimination, hallmark features of social stigma (Lock, 2000). Sachs (1996), for example, argued that in the Western world, “the strong culturally created ambitions to prevent disease in its members also generate social stigma and a guilt complex” (p. 636).

Note that it is the concept of ‘normality’ from which a stigmatized person deviates. “The normalization practices of biomedicine define the normal in advance, and then, on the basis of that definition, proceed to isolate and deal with anomalies” (Sachs, 1996, p. 636). The mapping and sequencing of the human genome represents the ultimate contemporary example of defining the normal: We now know what the ‘normal’ genome should be. Genetic testing is *specifically designed* to expose deviations from the normal genome, even for severe illnesses such as HD where biomedicine offers no cure and only limited options for treatment. In this age of genetic medicine, persons at risk for (or with) HD could be seen to possess an attribute which conveys a negative social identity – that of being ill as opposed to healthy, genetically abnormal as opposed to normal.

More specifically, Taylor (2004) suggested that persons with a family history of HD suffer from “multiple social stigmas,” including those associated with both the physical and mental manifestations of the disease. This suggests it is not the disease itself that elicits stereotypes, prejudice or discrimination (all central to social stigma), but the physical and mental symptomatology of HD.

Crocker et al. (1998) suggested that for stigmatized social identities, there is widespread cultural agreement about the devaluation of those identities and the negative stereotypes associated with them. Children, for example, learn at an early age to devalue persons who have disabilities, who are overweight or who are non-white. Mass media portrayals depict stereotypical renditions of many stigmatized groups. The mentally ill, for example, are portrayed as incompetent, violent, less human and fundamentally different in such public media as advertising, movies and television (Wahl, 1995). As Taylor (2004) noted, the 'mental' (i.e., cognitive) manifestation of HD may be equated with being mentally ill by perceivers of persons with HD, even though the afflictions are quite different. Nonetheless, negative stereotypes about the mentally ill could be applied to the person with HD.

Whether stigma is real or perceived could also affect experiences of stigma for at-risk persons and their families. Scrambler (1998; Scrambler & Hopkins, 1988) distinguished between enacted and felt stigma. The former refers to *actual* discrimination or social rejection, while the latter refers to the fear of *future* discrimination. In his work with epileptics, Scrambler found that people with epilepsy normally generate acute felt stigma prior to any experience of enacted stigma. As a consequence of felt stigma, epileptics may conceal their illness and adopt a policy of nondisclosure to most people. According to Scrambler (1998), enacted stigma is rare in the case of epilepsy, but felt stigma is common and proves more disruptive to people's lives. Based on this distinction, it is possible that persons at risk for HD and those who have tested positive, both of whom are currently asymptomatic, could fear potential discrimination or social rejection

as the onset of HD approaches. Those manifesting symptoms of HD and their caregivers, on the other hand, could have experienced enacted stigma.

Dimensions of stigma

Rather than identifying *types* of stigma, recent researchers have tried to delineate the dimensions along which stigmatizing conditions differ. Jones et al. (1984), for example, identified six such dimensions: (1) concealability – the extent to which a stigmatizing attribute can be hidden from others; (2) course – the way the stigmatizing condition changes over time and its likely outcome; (3) disruptiveness – how much the condition hinders social interactions; (4) aesthetic qualities – how repellent or upsetting the condition is to others; (5) origin – how the condition was acquired and whether the person is perceived to bear responsibility for it; and (6) peril – the danger the condition poses to others, both real and symbolic. While all dimensions have implications for how stigma affects social interaction, Crocker et al. (1998) argued that concealability and controllability (origin) are critically important for understanding the subjective experience of being stigmatized. It is suggested these are also important dimensions for understanding the experience of persons at risk for, and currently affected with, HD.

Concealable stigma

Goffman (1963) was the first to provide a useful distinction between visible and concealable stigmas that could be relevant to persons at risk for HD. The distinction is important: Santuzzi and Ruscher (2002) suggested that stigma effects among targets of visible stigmas might not be relevant to those with concealable stigmas, as the latter could interact in social contexts without ever revealing the stigma. At-risk persons

generally do not manifest visible symptoms (e.g., chorea) of HD until disease onset, usually around mid-life. For much of their life, therefore, these at-risk persons have a concealable stigma. In Goffman's (1963) terms, they are 'discreditable.' Discreditable individuals can participate in social interactions without their negative social identity filtering others' perceptions of them. However, they know stigma *is* a possibility if their devalued attribute were discovered (Crocker et al., 1998). Goffman argued that for such discreditable stigmatized individuals, the issue is not ... "managing tension generated during social contacts, but rather that of managing information about his [*sic*] failing. To display or not to display; to tell or not to tell; to let on or not to let on; to lie or not to lie; and in each case, to whom, how, when, and where" (Goffman, 1963, p. 42). Goffman suggested that a widely employed technique of information control used by discreditable persons is a division of the world into two groups: A large group to whom the person reveals virtually nothing about the stigma and a smaller group to whom the person fully reveals the stigma and upon whose help the person relies.

If at-risk persons do become clinically affected with HD, they are no longer discreditable, but 'discredited' (Goffman, 1963). That is, signs and symptoms of HD become visible. In Goffman's terms, the stigma becomes 'perceptible.' Crocker et al. (1998) noted that visible stigmas can provide the chief schema through which everything about the stigmatized person is understood by others. Discredited persons face not only the burden of 'managing tension' during social interactions, but also the additional burden of trying to explain HD to others, most of whom have no prior knowledge of the disease.

An assumption sometimes made in the literature is that visible stigmas are more problematic than concealable stigmas (e.g., Jones et al., 1984). However, people with concealable stigmas can possess negative self-perceptions. Frable, Platt, and Hoey (1998), for example, found that students with concealable stigmas (e.g., eating disorders) had lower self esteem and were more anxious and depressed than students with *valued*, concealable stigmas (e.g., high socioeconomic status) or visible, stigmatized students (e.g., African-Americans). Thus, secrecy can be a response to stigma, and fear of social disapproval is a common reason for keeping significant life events secret (Lane & Wegner, 1995; Major & Gramzow, 1999).

Research on the cognitive effects of keeping secrets (e.g., Major & Gramzow, 1999; Smart & Wegner, 1999) has shown that the effort of keeping a stigma concealed is mentally taxing and can lead to psychological distress over time. The preoccupation model of secrecy (Lane & Wegner, 1995) proposes that secrecy prompts thought suppression as the secret bearer tries to stop thinking about the secret. Thought suppression, ironically, causes thought intrusion - the very act of trying to suppress the secret renders it salient. Intrusive thought then causes renewed effort at thought suppression, and these cognitive processes continue in cyclic repetition. Lane and Wegner (1995) noted that secrecy induces a preoccupation with the secret that becomes intrusive and troubling over time.

Smart and Wegner (1999) applied the preoccupation model of secrecy to persons with concealable stigmas (eating disorders) and found that participants did become preoccupied with the control of stigma-relevant thoughts. They noted that, "People with

concealable stigmas, then, may not have conscious thoughts of their stigmas all of the time but rather experience thoughts of their stigmas as periodic intrusions as they try not to think about them” (p. 474). Participants in the current research also remarked on their efforts to put HD ‘out of’ their minds. Smart and Wegner (2000) later noted that in addition to the taxing cognitive effort of stigma concealment, stigmatized persons suffer in other ways. For example, in an effort to hide the stigma, they avoid interactions with stigmatized others, depriving themselves of the benefits that are available when one is open about a stigma (e.g., social support, social services and social relationships).

The majority of research on the effects of keeping secrets suggests that concealment is a negative activity, leading to stress-related physical and psychological problems. However, there are important reasons to *keep* secrets (see Kelly & McKillop, 1996, for a review). For example, revealing could elicit negative feedback from the listener or lead to social isolation if listeners subsequently avoid the revealer. In addition, people could also choose to keep negative secrets to themselves because they are concerned they will worry or upset others if they are revealed (Kelly & McKillop, 1996). This seems especially likely in the context of illness when the ill person does not want to be a burden on other family members. Earlier, it was noted that persons at risk for HD fear becoming dependant and a burden to other family members as disease onset begins. Because of such negative consequences of revealing secrets, Kelly and McKillop (1996) outlined some instances when it could be wiser to keep secrets. For example, if the secret is not troubling (e.g., causing no intrusive thoughts, anxiety or depression), it might be more beneficial to keep the secret. However, Kelly and McKillop (1996) did recognize

that not all secrets were alike. Decisions to reveal secrets which involve others (such as being at risk for HD) will be more complex than decisions about secrets which do not involve others. Interviews in the current study included discussion of communication or secrecy about HD (within and outside the family) and the frequency and nature of thoughts about the illness.

Controllability – Whose fault?

Crocker et al. (1998) suggested that a stigma is controllable when a stigmatized person is responsible for the condition, or when the condition results from or could be eliminated by the stigmatized person's behavior. In the case of being at risk for HD, no behavior of the at-risk person will eliminate his/her personal risk, nor should the person be perceived responsible for the condition. Nonetheless, at-risk persons *can* be held responsible for passing on the genetic risk to future generations. Responsibility is an important dimension of stigma since it affects how others judge and treat the stigmatized person and how the stigmatized person responds to the stigma. Persons with stigmas thought to be controllable are liked less, treated more harshly and are less likely to be helped than those with uncontrollable stigmas. For example, DePalma, Madey, Tillman, and Wheeler (1999) provided participants with an actual opportunity to assist a target medical patient with a fictitious blood disorder. Participants were randomly assigned to one of three target responsibility conditions: Perceived responsible for the medical condition (caused by unprotected sex), perceived not responsible (caused by a genetic abnormality), or a control condition (no mention was made of how the disorder was acquired). DePalma et al. (1999) found that participants were less likely to help patients

who were perceived to be responsible for their condition and were particularly inclined to help those who were perceived not to be at fault for the illness.

According to Weiner's (1995) theory of responsibility, responsibility is linked to perceptions of the cause of an event and whether the event is perceived as controllable. Outcomes attributable to controllable causes lead to the assignment of responsibility (Weiner, Perry, & Magnusson, 1988). If an illness is perceived as resulting from uncontrollable factors (e.g., genetic abnormality), the sufferer is not held responsible. Why is responsibility such an important dimension of social stigma? Judgements of responsibility affect subsequent emotional and behavioral reactions of perceivers. Weiner et al. (1988), for example, showed that persons held responsible for cancer, AIDS or heart disease evoked anger in perceivers, which in turn, affected willingness to help the sufferer. Sufferers who are not held responsible for their condition, however, elicit sympathy and helping behavior. Weiner's (1995) theory of responsibility can be conceptualized as follows:

Controllable outcome→Responsibility→Anger→Neglect

Uncontrollable outcome→No Responsibility→Sympathy→Willingness to Help

Menec and Perry (1998) tested Weiner's (1995) model in the context of stigmas. Participants read vignettes of nine different stigmatizing conditions (e.g., cancer, blindness, unemployment or obesity), each described as controllable or uncontrollable. The latter was sometimes accomplished by suggesting the cause was hereditary. Participants rated the controllability of each stigma, as well as their anger, pity and

willingness to help the affected person. When stigmas were perceived as controllable, participants evinced more anger and less pity. Greater pity, however, was associated with greater willingness to help. Notably, these effects were observed for all nine stigmas, suggesting the effects were not limited to one particular stigma.

A recent study examined the effect of birth outcome on participants' reactions to genetic testing and the birth mother (Menec & Weiner, 2000). Along with constructs from the theory of responsibility, the effect of hindsight bias was also measured. Hindsight bias, or the I-knew-it-all-along effect, is the tendency to overestimate the probability of occurrence of an outcome once the actual outcome is known. It is thought to occur automatically and unconsciously, and has been found to be a robust phenomenon (Hoffrage & Pohl, 2003; Menec & Weiner, 2000). In the case of a genetic outcome, once the condition of a child is known (e.g., born healthy or with a genetic disorder), people could overestimate the likelihood of that outcome. This hindsight bias may then affect responsibility judgments that, according to Weiner (1995), determine emotional reactions and behavior towards the birth mother.

In three studies, Menec and Weiner (2000) gave participants a vignette in which a woman declined to take a genetic test and subsequently gave birth to a healthy child or a child with a genetic disorder (Tay Sachs disease or Cystic Fibrosis). Retrospective judgements of the likelihood that the child would have the genetic disorder were higher given the negative outcome (i.e., the child had a genetic disorder). Importantly, the more likely a negative outcome was perceived to be, the more responsible the mother was held for not taking the genetic test. When the mother was held responsible, she elicited more

displeasure and less sympathy. Sympathy, in turn, was linked to willingness to help. This study has important implications for how others view persons at risk for HD who choose to have children. When others feel the birth of a child with a genetic disorder was likely, they could hold the parents accountable for that birth. In turn, the parents could be less likely to receive assistance and sympathy from others. This suggestion is supported by findings from a study with mothers of children with disabilities (Green, 2003). Interviews with these mothers revealed that family members and members of the community at large did appear to blame them for their child's disability. A mother of a son with Down's syndrome said, "...people would say to me, 'Did you know he was going to be Downs before you had him?'" Like I would have changed my mind if I had known and I used to just want to cry right in public" (Green, 2003, p. 1367).

Mantler, Schellenberg, and Page (2003) caution that controllability, responsibility and blame are distinct psychological constructs. In their study, students read vignettes about a target male with AIDs or lung cancer and rated his controllability, responsibility and blame regarding the illness. Supporting decision-stage models of attribution for negative events (e.g., Weiner, 1995), students did distinguish between the constructs which followed the controllability→responsibility→blame sequence. Blame was the construct most associated with behavioral intentions (e.g., willingness to help the target) and emotions (e.g., anger or sympathy). Notably, the attribution sequence became biased by social attitudes and personal biases (e.g., authoritarianism, belief in a just world, or attitudes towards gay men). In this study, these biases and attitudes were correlated with

emotional reactions and intentions even after controlling for responsibility, controllability and blame judgments. Thus, subjective factors (e.g., values or attitudes) can exert independent effects on behavioral intentions, and on blame, which is most closely related to the response a stigmatized person is likely to receive.

Applying this notion to those at risk for HD, it is unlikely at-risk persons would be blamed for their status as an at-risk person. They would have no control over inheriting the altered HD gene. However, those same people *could* be perceived to have control over marriage, procreation or career decisions. As such, they could be held responsible for those choices and blamed for a negative outcome that might ensue (e.g., birth of an affected child).

Severity

In addition to concealability and controllability, the severity of illness has also emerged as an important component of stigma. For example, in two studies, Crandall and Moriarty (1995) showed that illnesses thought to be under personal control and those perceived to be severe consistently predicted social rejection. In Study 1, students read “medical case histories” of 66 illnesses and rated the diseases on dimensions of illness stigma (e.g., severity or contagion). They also completed a social distance scale, a measure of social rejection. Stepwise regression analyses revealed that illness severity and behavioral control of the illness accounted for 46% of the variance in social distance. In Study 2, students read one of four vignettes about a fictitious disease – it was either under behavioral control or not under control and the illness was severe (poor prognosis, minimal available treatment) or it was mild (good prognosis, treatment was available).

Importantly, the use of a fabricated illness minimized any preconceived notions about its nature or cause. Participants again rated the illness and completed a social distance scale. Once again, diseases that were severe and thought to be behaviorally caused had higher social distance scores, while the interaction was not significant.

Dijker and colleagues (e.g., Dijker & Raeijmaekers, 1999; Dijker & Koomen, 2003) also found seriousness of disease to be a reliable predictor of fear responses in others. Examining emotional reactions to disease-related stigma, Dijker and colleagues found that diseases which were life-threatening or debilitating, though not necessarily contagious, were more likely to evoke fear or anxiety in others.

Thus, illness severity could have implications for potential stigma experienced by families affected by HD. Crandall and Moriarty (1995) noted that in their severe condition, the illness was difficult to treat, had widespread effects in the life of the target and had a poor prognosis compared to the mild illness condition. Also, the symptoms of the illness were more pronounced and were more likely to interfere with everyday life. These dimensions of severity all correspond very well to HD as an illness – the disease is fatal, there are limited options for treatment, the symptoms are pronounced (e.g., chorea) and interfere with daily life and social interactions. Thus, while HD sufferers cannot be blamed for their illness, the severity of HD could be a significant factor in the stigmatizing responses of others.

Stigma as threat to social identity

As noted, stigmatized persons are devalued in the eyes of others due to membership in some social category. That is, their social identity is somehow flawed.

From the perspective of social identification, people are not regarded as unique individuals, but as members of social categories. Rather, they are viewed by others as representative of a group of like people, and they normally perceive themselves as sharing certain attributes and concerns with others who share their social identity (Deaux & Ethier, 1998). Deaux and Ethier (1998) proposed that social identification is a dynamic process: People actively adjust their self-perceptions, change their reference groups and modify behavior to accommodate these changes. For these authors, "Identity negotiation is an ongoing process, best conceived as continual efforts directed at maintaining existing identities as well as adapting to changing circumstances" (p. 301).

Living at risk for HD can conceivably involve identity negotiation. Persons at risk for HD must maintain their identity as a 'normal' person, while acknowledging the changing circumstances involved with the illness, both at present and in the future. Broadly speaking, contextual factors, such as illness in the family, are crucial to identity negotiation. Self-categorization theory (SCT; Turner, Hogg, Oakes, Reicher, & Wetherell, 1987), for example, explicitly emphasizes contextual shifts in identity salience. According to SCT, the accessibility of a category and its contextual fit to the immediate environment determine which identity is salient. Thus, SCT would suggest that the at risk identity may not always be salient for persons at risk for HD. Indeed, participants in the current study did identify themselves as similar to a range of generalized others when contexts or attributes not specific to HD were being described.

The social nature of identity negotiation also suggests that the views of others affect identity claims. For example, perceivers could categorize a person into a particular

category that the person him/herself does not claim. Deaux and Ethier (1998) noted this was particularly likely for categories which were visible to others, such as gender. One might also add this is a possibility for those manifesting visible symptomatology of HD (e.g., chorea). Contextual change is relevant to identity negotiation by targets of stigmatization, prejudice or discrimination as these persons can often be 'misabeled' by others. For example, participants in the current research cited examples of social others labeling them and/or loved ones as "drunk" or "retarded." As a consequence of mislabeling, stigmatized persons can seek out other environments where the same identity is valued and supported rather than stigmatized. The popularity of support groups for various illnesses attests to this agency on the part of stigmatized persons.

Identity negotiation is likely to take place when a person perceives the need to adjust or redefine a particular identity due to some psychological, social or contextual demand (Deaux & Ethier, 1998). It is reasonable to assume that prior to discovering they are at risk for a fatal genetic illness, at-risk persons will see themselves as 'normal' and 'healthy,' barring any other illness. Thus, discovering they are at risk for HD represents a contextual change that at-risk persons must negotiate. Similarly, when a person passes from the at risk state to testing positive or negative for the altered HD gene or from the genetic test to being clinically affected by HD, identity negotiation might also be likely.

Identity negotiation strategies

Deaux and Ethier (1998) outlined two broad negotiation strategies: (1) identity negation; and (2) identity enhancement. Identity negation tactics are aimed at disassociating oneself from a social identity that is aversive or non-satisfying. Identity

enhancement, on the other hand, includes those strategies which attempt to assert or extend an existing identity. Broadly speaking, if at-risk persons perceive the responses of others in their social environment to be negative, they could try to negate the at risk identity. However, if at-risk persons include their at risk status as an integral part of the self, they could alternatively enhance the at risk identity.

Identity negation

Identity negation strategies include eliminating the identity altogether, denying the identity or decreasing the importance of the identity. Eliminating the at risk status could be attempted by choosing to have the genetic test, effectively changing the risk of carrying the altered HD gene to 0% or 100%, and concomitantly, escaping HD or developing HD, respectively. On the other hand, at-risk persons or those who have tested positive could choose to deny those identities altogether. This seems a possibility for persons who are currently a-symptomatic or young, as the possibility of developing HD is in the distant future. Decreasing the importance of the identity is perhaps less extreme than denying or eliminating the identity and the most flexible. It allows a person to respond to immediate situational demands without abandoning a well-established identity altogether. Persons who are clinically affected with HD or those who have tested positive could decrease the importance of these identities. The latter by claiming the illness is not affecting them at the present time, and the former by asserting they are more than their disease.

Identity enhancement

Identity enhancement strategies, on the other hand, include reaffirmation, re-mooring, intensified group contact and social change. Reaffirmation is aimed at proclaiming or reasserting an identity that is already part of the self. Reaffirmation might be applicable to HD persons in several ways. Those at risk for HD, for example, could proclaim they are still 'normal' and 'healthy,' despite being at risk for the illness. On the other hand, at-risk persons, especially those who have known about the family history of HD for some while, could reaffirm their at risk status rather than their status as a healthy person.

Re-mooring strategies involve more active behavioral involvement. Thus, a person could seek out information about the group, associate with new group members or take part in group events. At-risk persons might engage in information gathering about HD, for example, or speak to others who are also at risk. Intensified group contact is related to re-mooring – in the latter, a contextual change necessitates a re-positioning of the self that might not be possible in current environments. A person clinically affected with HD, for example, eventually cannot work (excepting late-onset cases where retirement has begun before the onset of symptoms). Thus, re-mooring could occur in other contexts such as support groups. Intensified group contact, on the other hand, can often be carried out in one's current social environment. For stigmatized persons, contact with other group members has a number of benefits including social support, information, enhancing self-esteem and counteracting discrimination. Those at risk for HD, for example, may choose to join support groups for others with HD. Caregivers could also

choose this identity enhancement strategy. Finally, social change might also be employed when the person attempts to change the beliefs that others hold about his or her social category or to change the social structure in some way that will be positive for the identity. Green's (2003) interviews with mothers of children with disabilities revealed attempts by the mothers to educate others about their child's disability, rather than negate the identity. Similar evidence emerged in the current study.

Negate or enhance?

The question of whether stigmatization leads to identity negotiation and which strategies are employed depends on the perception and interpretation of the stigmatized person. Deaux and Ethier (1998) suggested that the perception of threat, and its magnitude, was integral to the identity negotiation process. If an identity is being evaluated negatively, as is the case for the identities of many stigmatized groups, this constitutes a threat to that identity. Deaux and Ethier (1998) suggest that a social identity is both a categorical membership and a set of meanings, behaviors and attributes associated with that category. These meanings, behaviors and characteristics are socially defined by society at large and the in-group with which one identifies. At a macro level, the meanings associated with a particular social identity derive from the social representations of the culture (Moscovici, 1988), the stereotypes, media representations and communications of a society. It is these shared aspects of meaning which define the similarity among members of a category, both for in-group members and outsiders. It is not the case that individuals will accept *every* component of the social definition of their group; however, outsiders can quickly, and sometimes unconsciously, apply the set of

attributes they associate with a membership category. Decades of stereotype research confirm how readily perceivers attribute characteristics to individuals on the basis of group membership and act accordingly (see Fiske, 1998, for a review).

As noted, the context into which the 'at risk' label has been introduced is one of personal responsibility for health. Citizens of risk society should be healthy, self-sufficient, active in social and civic life and responsible for avoiding and managing the risks in their lives. Sufferers of chronic illness often cannot claim this identity, yet they know it exists as the gold standard for behavior in Western societies (Galvin, 2002). Persons at risk for HD know they could face a future of chronic illness (Chapman, 2002; Wexler, 1979) along with the possibility of intolerance and stigma. Those who have tested positive for the altered HD gene *know* they will face this future. It is conceivable, then, that at-risk persons and those testing positive could perceive a threat to their identity as a currently healthy, normal person.

Identity threats are subjective

As noted, while discrimination and prejudice typically prompt identity negotiation by the target, the strategy chosen will depend on whether the responses of others are perceived as threatening to one's identity. Undoubtedly, threats to identity are subjective. What one person perceives to be stigmatizing, another could ignore or be oblivious to. One factor that seems important in determining reaction to stigmatization is the degree of identification with the stigmatized group (Deaux & Ethier, 1998). For example, those who are strongly identified with their group tend to engage in identity enhancement strategies - they will attempt to maintain their identity in the face of threat. On the other

hand, for those who do not identify strongly with their group, identity negation strategies could be chosen. These persons could try to dissociate from the group by negating the identity or reducing its importance.

In the case of a genetic disorder like HD, it is likely that identity threats manifest themselves in a multitude of ways, owing to the late-onset nature of the disease, the usual late discovery of HD in the family, its multi-generational effects and the variability in symptoms. For example, those who discovered they were at risk for HD at a young age could have incorporated the at risk status into the self. These people might be strongly identified with the at risk group, and would be more likely to choose identity enhancement strategies. They might decline the genetic test, for example, as this would destroy their at risk status. Others, however, who discovered their at risk status later in life could be less strongly identified with the at risk group. These individuals might decide to have the genetic test as it poses no threat to the at risk identity. Whatever the test outcome, these individuals could choose to negate the HD identity. They might refuse to acknowledge it altogether (e.g., since they are a-symptomatic) or reduce the importance of testing and the test result. For example, they might suggest we are all at risk for 'something' and HD is no different from any other risk that exists in life. They could also engage in identity enhancement by trying to maintain their identity as a normal, healthy person. Identity threats could also become evident when signs of HD are visible. In this case, the person is no longer at risk, but affected. Those who identify strongly with having HD could attempt to enhance their identity. For example, they might be a member of the Huntington's Society, attend support group meetings or try to educate

others about the nature of HD. Importantly, these strategies are also available to caregivers of persons with HD.

What are the consequences of social stigma?

Genetic discrimination

Crocker et al. (1998) reviewed several noteworthy aspects of the phenomenological experience of being stigmatized. All pose a threat to the self-worth of the stigmatized person, whether manifested as threats to personal or collective self-esteem. Like earlier authors (e.g., Goffman, 1963; Jones et al., 1984), Crocker et al. (1998) noted the hallmark feature of the stigmatized was the constant possibility of being the target of prejudice and discrimination. While personal experiences with prejudice and discrimination can be rare or common, the *possibility* of encountering them always exists. “This is the reality that shapes and defines the experience of stigma” (Crocker et al., 1998, p. 516). As a result, stigmatized persons could be ‘on guard’ during social interactions: They might feel the need to be mindful of the fact that social others could be prejudiced.

Nelkin and Lindee (1995), ardent critics of genetic essentialism (i.e., the tendency to equate human beings with their genes, ignoring their myriad complexity), warned that the focus on genetics could enhance discriminatory attitudes towards at risk individuals. Guttmacher and Collins (2003) suggested the most commonly expressed fear about genetic information is that it will be used in ways which are detrimental to people – for example, to deny them access to health or life insurance, employment, education, and so on. At-risk persons do cite insurance concerns as an important reason to avoid taking a

genetic test (e.g., Barlow-Stewart & Keays, 2001; Hall & Rich, 2000). Hall and Rich (2000) noted that fear of potential discrimination was especially acute for persons at risk for late-onset disorders, such as HD. Their interviews with genetic counselors revealed that adults seeking testing for late-onset disorders such as HD had high levels of concern about potential discrimination, in sharp contrast to prenatal and pediatric counseling clients. The majority of genetic counselors (21 out of 25) indicated they routinely discussed the potential for insurance discrimination as a risk of genetic testing (Hall & Rich, 2000). Most counselors did suggest, however, that discrimination fears were not the primary reason for avoiding testing.

A later survey of genetic counselors (Bower, McCarthy-Veach, Bartels, & LeRoy, 2002), reported that 29% (N = 454) of either them or their clients frequently had concerns about genetic discrimination. Pfeffer, McCarthy Veach, and LeRoy (2003) interviewed 25 genetic counselors of cancer risk patients and reported that the vast majority (96%) almost always discussed genetic discrimination with their clients. Nine of the counselors recounted actual instances of discrimination against their clients, including denial of health/life insurance, social discrimination and employment discrimination.

Genetic discrimination has been defined as "discrimination against an individual or against members of that individual's family solely because of real or perceived differences from the "normal" genome of that individual" (Billings, Kohn, Cuevas, Beckwith, Alper, & Natowicz, 1992, p. 477; see also Natowicz, Alper, & Alper, 1992). Genetic discrimination excludes discrimination against individuals who are clinically affected with a genetic disorder (i.e., they are manifesting symptoms). Rather, the

'asymptomatic ill' are the usual targets of genetic discrimination. Thus, the denial of employment to an asymptomatic person who carried the altered gene for HD would constitute genetic discrimination, while the denial of employment to a person suffering pronounced chorea caused by the altered HD gene would *not* constitute genetic discrimination.

Billings et al. (1992) undertook one of the earliest studies on genetic discrimination. The authors aimed to discover whether incidents which reflected genetic discrimination were occurring in employment, insurance, in access to social services and in the delivery of health care. A variety of genetic illnesses were represented in the study, including HD. Billings et al. (1992) evaluated 29 narratives of genetic discrimination, representing 41 separate incidents of possible discrimination in the United States and Canada. All but two occurred in employment or insurance contexts. Specifically, thirty-two incidents were in the insurance context, while seven involved employment situations (e.g., hiring, firing, promotion and transfer). At-risk persons described difficulties in obtaining insurance (health, life, disability, mortgage and auto) and in trying to upgrade insurance. Difficulties in finding a job or being unable to change jobs were also noted. Two at-risk persons for HD were denied adoption, despite being asymptomatic at the time.

Since this early study, other instances of genetic discrimination have been documented (e.g., Barlow-Stewart & Keays, 2001; Hudson, Rothenberg, Andrews, Kahn, & Collins, 1995) with employers and health, life, and disability insurers using genetic information to deny or limit coverage and to raise rates. The first survey of genetic

discrimination in the United Kingdom found that approximately one-third of participants (drawn from support groups representing seven genetic disorders, including HD) encountered problems when applying for life insurance compared to only 5% of participants from the general population (Low, King, & Wilkie, 1998). Barlow-Stewart and Keays (2001) reported on 48 cases of alleged genetic discrimination in Australia. In each case, the individual was in good health at the time of the discrimination (i.e., the person was asymptomatic), but had received a positive genetic test result. Genetic discrimination was reported for a wide range of genetic disorders, including inherited predisposition to cancer (breast, ovarian, bowel and melanoma), heart disease, and HD. Incidents of discrimination were reported in the area of insurance, employment and health services (e.g., one woman was denied access to in-vitro fertilization (IVF), while another was denied a laparoscopy – tubes tied). It should be noted that most of this research relied on participants' perceptions of events. However, Billings et al. (1992) did comment on supporting documentation provided by some of their participants, providing some tangible evidence of instances of discrimination. A recent example of employment discrimination was the refusal of a permanent teaching position to a female teacher in Germany owing to her family history of HD (Burgermeister, 2003).

Notwithstanding these reported instances of discrimination, Reilly (2000) suggested that, "Despite little evidence to support it, a widespread public fear of genetic discrimination persists" (p. 494). Nowlan (2002) argued that the evidence for genetic discrimination, at least in the arena of health insurance, was largely anecdotal. He reported that a professional panel at the American Society of Human Genetics 1999

annual meeting could not identify any cases of health insurance discrimination. A recent law in Switzerland, however, allows insurance companies limited access to genetic test results, potentially creating a 'genetic underclass' of people who would have difficulty buying life insurance (Burgermeister, 2004).

Otlowski, Taylor, and Barlow-Stewart (2003) cautioned that there is very limited empirical research about the nature and extent of genetic discrimination. Further, much of the research to date relied on unverified and sometimes anonymous accounts of people's subjective impressions of discrimination. The current study included discussion of discrimination in insurance, employment and other social contexts. However, any perceived instances of discrimination were unverified. Participants who had a family history of HD, but had never been tested, were asked whether fears of discrimination entered their decision not to test. As part of a national survey, Moore-Orr and Longerich (2000) found no perceived discrimination in a sample of Newfoundland women genetically at risk for breast cancer. In that survey, 91% of women indicated that "My genetic test may influence my or my children's susceptibility to discrimination" was a 'not at all' or 'somewhat important' reason for declining a genetic test for breast cancer. These women did not appear to fear discrimination owing to their at risk status.

Beyond genetic discrimination: Other consequences of stigma

Beyond potential genetic discrimination, there are other important consequences for targets of social stigma: Social interaction between the stigmatized and non-stigmatized can be disruptive, awkward and embarrassing (Goffman, 1963), leading to anxiety, social isolation and depression in targets (Crandall, 2000). In addition, stigmas

have long been known to be barriers to intimate relationships and employment (Jones et al., 1984). Arguably then, there appear to be numerous cognitive, emotional and behavioral consequences of stigma. Crocker et al. (1998) argued that psychological well-being, including life satisfaction, self-esteem and depression and school achievement outcomes were critical to the experience of being stigmatized. The current discussion will focus on the consequences of stigma for psychological well-being, as that had more relevance for participants in the current study. School achievement outcomes had already occurred for the majority of participants.

Psychological well-being

An assumption sometimes made by 'normals' is that the stigmatized are unhappy and dissatisfied with their lives (Jones et al., 1984). After all, how could the stigmatized live with the knowledge that their social identity is devalued by others and confront the negative stereotypes, prejudice and discrimination about their group without feeling unhappy, bitter and angry? This assumption has also been shared by some psychologists, notably in early stigma research (e.g., see Crocker et al., 1998). However, empirical research generally does not support this assumption. Reviewing research on the self-esteem of many stigmatized groups, Crocker and Major (1989) concluded:

In short, this research, conducted over a time span of more than 20 years, leads to the surprising conclusion that prejudice against members of stigmatized or oppressed groups generally does not result in lowered self-esteem for members of those groups. These findings generalize across a variety of stigmatizing conditions, a variety of measures of global self-esteem, and a wide range of subject populations, from adolescents to college students to adults (p. 611).

In their interviews with women affected by chronic mental health problems, Camp, Finlay, and Lyons (2002) suggested that while women were aware of the negative views of mental illness held by others, they did not view them as valid. Nor had they internalized these negative attitudes or deemed them applicable to the self. As a result, they suffered no ill effects on their self-concepts. The women offered several explanations for society's negative attitudes including ignorance, fear and the media. For these women, then, "the stigma...was not presented as something they were responsible for, rather it was due to the flaws of those who stigmatize" (p. 830).

In addition to the relative lack of effect on self-esteem, Crocker et al. (1998) noted that stigmatized persons are not necessarily dissatisfied with their lives either. In fact, a large majority of stigmatized persons (e.g., the mentally ill and persons with numerous physical disabilities such as those who were blind or quadriplegic) reported positive levels of personal well-being (Diener & Diener, 1996). According to these authors, "although ethnic minority and disadvantaged groups sometimes report lower subjective well-being than broader samples, they nevertheless score in the positive range" (Diener & Diener, 1996, p. 7).

Crocker et al. (1998) noted, however, that depression seemed more prevalent in some members of stigmatized groups: Depression is usually higher in African-Americans than in European-Americans; women as a group experience more depression than men. Crocker et al. (1998) suggested, however, that it is less useful to examine differences between stigmatized groups and non-stigmatized groups than to examine within-group

differences in vulnerability to distress (i.e., which stigmatized individuals are vulnerable and why).

As noted earlier, a large body of work has suggested that those testing positive for the altered HD gene do score higher than those testing negative on measures of depression shortly after the test result. However, these levels are usually within normal range and return to baseline levels within a very short period of time. Longitudinal studies have shown that there is no significant increase in depression over time (up to five years) in those testing positive for the altered HD gene. I would suggest, however, that longer time periods of observation are needed, especially as the at-risk person moves closer to the age of onset. This suggestion is supported by Timman et al.'s (2004) recent observation of increased hopelessness in altered gene carriers and their partners seven to ten years following testing.

Are the genetically at risk really stigmatized?

Thus far, a variety of theoretical and empirical issues in stigma research have been reviewed, some of which could be relevant to HD families. However, is it the case that those at risk for (or with) HD *are* stigmatized? Do HD families perceive themselves to be the targets of stigma? In the context of genetic testing, very little empirical work (beyond the investigations of genetic discrimination reviewed earlier) has examined whether at risk individuals are actually stigmatized or perceive themselves to be stigmatized. There has been much theorizing and lamenting about the issue, however. Taylor (2004) suggested that persons with a family history of HD face multiple social stigmas, including "associations with physical as well as imputed psychiatric disability,

the latter for example through institutionalisation or suicide of affected or at-risk family members" (p. 2). A large body of research on mental illness confirms its status as stigmatizing (e.g., see Link & Phelan, 2001, for a review). It was noted earlier that HD is often misdiagnosed as other dementing disorders in psychiatric practice (O'Shea, 1997). Thus, sufferers of HD could be mislabeled as mentally ill due to HD symptomatology. As one participant put it, when others hear someone has HD, they think "you are a head case."

Cox (2002) edited a collection of personal accounts of predictive testing for HD. There was evidence in some of these accounts that felt stigma (Scrambler, 1998) was an important concern for some. As one contributor put it, "Families with HD hide behind a lot of shame and fear. This shame and fear seems to take over our life, till there 'ain't no more' life to look forward to" (Cox, 2002, p. 15). A man who had tested negative for the altered HD gene recalled:

I was told about the 'family secret,' Huntington disease. No one must know – people will be fired from jobs; the family name will become synonymous with "crazy," future prospects for the children will be non-existent (p. 52).

A woman who tested positive worried:

I just started a new job, so I worry about the right time to tell them. Do I wait until it is very noticeable, or do I tell them ahead of time and risk my job? For now, I think that I will just leave it for a later time (p. 57).

Felt stigma could be dependent on the type of genetic condition. For example, focus group discussion with persons at risk for colon cancer revealed that most participants felt there was nothing shameful about a positive genetic test result for colon cancer. Rather, they saw the test as an opportunity to inform others, especially other

relatives, about cancer risk and the benefits of screening (Ramsey, Wilson, Spencer, Geidzinska, & Newcomb, 2003). In contrast, qualitative interviews with persons who had tested normal for neuro-degenerative disorders (including HD) found that some individuals felt relief from perceptions of being stigmatized by their at risk status (Williams, Schutte, Evers, & Holkup, 2000). For example, some of these participants recalled experiences of discrimination in the workplace and from other family members prior to receiving their negative test result.

Interviews with families affected by HD in China did reveal social stigma. For example, one caregiver feared stigma if the person with HD was taken out in public (Leung & Leung, 2002). The authors also discussed a man with HD who was advised to avoid female passersby in the street as his choreic movements could be mistaken as indecent. Referring to persons with HD, Leung and Leung (2002) noted, "The subject can be regarded as bad or mad" (p. 308).

Beyond these cited examples, I am unaware of any other empirical research that has focused on being at risk for HD, or having HD, as a stigmatizing condition. Leung and Leung (2002) advised that although their research comprised a small sample, it did document instances of social stigma in HD, an observation that had never been recorded in the literature.

Despite the lack of empirical research, critics of the new genetics have been very vocal about the possibility of stigma and discrimination against at-risk persons (e.g., Lock, 2000; Nelkin & Lindee, 1995). Specifically, critics worry that the discourse of

genetics which permeates popular media today will enhance attitudes which are

biologically deterministic and socially discriminatory. The most active critics suggested:

...the images and narratives of the gene in popular culture reflect and convey a message we will call genetic essentialism. Genetic essentialism reduces the self to a molecular entity, equating human beings, in all their social, historical, and moral complexity, with their genes (Nelkin & Lindee, 1995, p. 2).

Nelkin and Lindee (1995) warned that genetic essentialism is deterministic and reductive, and it promotes discrimination: If one's genetic blueprint is read as 'causing' all disease and other human attributes, then one can be discriminated against simply because of one's blueprint (i.e., one's genes).

If an employer, or educator, or insurer can make the case that the 'predicted' future status of their client matters, then discrimination – denial of opportunity for medical care, work, or education – can occur with impunity. Indeed, predictive genetic typing may create an underclass of individuals whose genes seem to have marked them for the nowhere track (Nelkin & Lindee, 1995, p. 167).

Others concurred, suggesting that genetic differences underlying disabilities are inherently discriminatory. "The mind set behind genetic testing rests on societal views of disabilities that should not go unchallenged as tests that emphasize inborn genetic differences as the causes of potential disabilities are by their very nature discriminatory, because they sort people on the basis of factors that are beyond their control" (Hubbard & Wald, 1993, p. 135).

Others have warned of genetics' capacity to specify what is normal and abnormal (Lock, 2000). Taylor and Mykitiuk (2001), for example, discussed the implications of genetics for our understanding of normalcy and disability. These authors warned that genetic discourses of normalcy suggest that the ability to live a fully-functioning life is

related to, if not determined by, genetic makeup. Thus, genetic disability is thought to impair life opportunities, just as disability more generally is framed as limiting (Shakespeare, 1999). They argued that genetic knowledge changes our understanding of what normal bodies are: If we equate normalcy with the composition of one's genes, then genetically 'abnormal' bodies are signaled out for correction:

It predicates a movement away from (potentially) broadly based inquiries into what disabled people need to be full participants in society, and leads to the framing of questions about disability in terms of how disabled peoples' "abnormalities" can be corrected and improved upon. Indeed, it also raises questions about whether disabled people should exist at all (Taylor & Mykitiuk, 2001, p. 69).

Shakespeare (1999) argued that while overt eugenicisim in genetics discourse is rare, "a clear set of values does emerge from the literature, which is implicit and subtle, but undoubtedly reflects a consensus that disability is a major problem, which should be prevented by almost any means necessary" (p. 673). Shakespeare cautioned against the apparent ignorance about living with disability in genetics discourse. That is, ignorance exists about the experience of actually living with a particular condition since it is rare to hear the voices of people affected with disabilities in the genetics literature. According to Shakespeare, this represents a 'major absence in knowledge.' I would suggest a similar absence of knowledge exists in research on HD. Research abounds on the clinical outcomes following testing for HD (e.g., depression, stress or anxiety). However, it is rare to hear from HD sufferers themselves, their caregivers or those at risk for the illness. The current research addresses this gap in the literature.

Does a focus on genetics necessarily promote discrimination?

As noted earlier, it is possible persons at risk for, or suffering from, a genetic illness could elicit sympathy and willingness to help, rather than anger or neglect (e.g., Menec & Perry, 1998) since such persons cannot be held responsible for the condition. In the context of mental illness, for example, Phelan (2002) noted that (at least in the short term), the impact of genetic attributions should be to reduce the burden of blame, particularly suffered by parents. Condit (1999) argued that media portrayals of genetics are less deterministic and discriminatory than suggested by Nelkin and Lindee (1995). The latter authors have argued that the 'blueprint' metaphor (i.e., one who has a genetic illness has the illness as part of their hereditary blueprint) is the conduit by which discriminatory attitudes are enhanced. Condit (1999) had undergraduates read one of two hypothetical news articles representative of genetics discourse. Students were asked to interpret the articles and the blueprint metaphor specifically. Condit (1999) reported that only a minority of participants interpreted the blueprint metaphor as discriminatory. Further, participants clearly indicated that disability was not completely undesirable and disabled people could still lead happy, productive lives.

Thus, there is evidence that public attitudes towards persons with genetic illnesses are not wholly negative or stigmatizing. Kerr, Cunningham-Burley, and Amos (1998) conducted 20 focus groups with the lay public which covered a range of topics including genetic research and genetic testing, including responsibility and autonomy regarding the latter. Kerr et al. (1998) reported a sophisticated discussion of responsibility for taking genetic tests. When asked, participants clearly valued individual choice in genetic-test

decisions, but were aware of various cultural and social pressures that might constrain choice. Participants also discussed quality of life for various genetic conditions. Interestingly, these discussions included not just a consideration of the impairment caused by the condition itself, but also issues of stigma and discrimination which were considered equally debilitating. Across all focus group discussions, there was no evidence of overt discriminatory attitudes towards at-risk persons.

In addition to assessing the implications and interpretations of genetic risk and illness, the current study was expressly concerned to explore participants' views on HD as a stigmatizing condition. While critics of genetics have been quick to voice concern about the potential for stigma and discrimination toward at-risk persons, far less research has been devoted to investigations of stigma, including genetic discrimination, in at-risk persons themselves. A recent article noted that qualitative research was well suited to explore the nature of genetic discrimination according to at risk individuals' understanding and experience; although, to date, such methods have not been widely used (Treloar, Taylor, Otlowski, Barlow-Stewart, Stranger, & Chenoweth, 2004). The current study attempted to address this gap in the literature.

CHAPTER 5

**THE CONTEXT OF CONVERSATIONS ABOUT
GENETIC RISK AND ILLNESS**

It is one thing to read and think about Huntington disease (HD) in the sheltered world of academe, without ever having met a HD sufferer; it is quite another thing entirely to sit across a kitchen table with someone affected by the illness. Before I began interviewing people affected with, or at risk for, HD, I wondered (and worried about) how these interviews should be conducted. Was there a 'right' way to talk to people about genetic risk and fatal illness? And, if so, would I find it? Should I try to remain neutral or should I express my own emotions as stories were related to me? What would I feel anyway? Was it 'proper' for me to divulge information about myself? As interviews progressed, it was apparent participants expected some disclosure on my part. How many probing questions were appropriate? Would I even know how or when to probe? Did I ask the right questions at all? Would it be painful for participants to talk about a devastating illness? Would I recognize signs of distress? What would I do if a participant became upset?

Many of these questions did get answered as the research progressed, often by participants themselves, and some questions became (more or less) moot with the very first interview. With that interview, for example, I realized I would (or could) not be a completely 'neutral' participant in these conversations. I knew immediately that some aspects of these narratives would be painful for the participant and for myself. It was several interviews later, however, before I began to reflect on my own contribution to the

stories unfolding before me. Rapley (2001) suggested the analysis and reporting of research interviews should “include some degree of the interactional detail and *at the very least* interviewers’ talk should *always* be included” (p. 306, emphasis in original). The current research takes his suggestion seriously. This chapter and the next will attend to the ‘interactional detail’ of interviews during their consideration of context, methodology and research design. Instances of the researcher’s talk are included in the results chapters as data are presented.

This chapter outlines the context of conversations about genetic risk for HD (and the illness itself) as they occurred in the current research. It takes context seriously, suggesting that proper consideration of context is vital for interpreting the stories related herein. Serious reflection on the context in which at risk individuals and their families live their lives is paramount to understanding the meanings of, and reactions to, genetic risk for a fatal illness. Mainstream social psychology has often underestimated the impact of the social context in which individuals negotiate their lived reality. It has also failed to consider how research is an intervention into participants’ lives, not merely an investigation. Each conversation in the current research – both on and off the record – was not only an opportunity to *tell* a life story, but *was* an event in that life story (cf. Cox, 1999). Some participants explicitly acknowledged this reality. For example, a young woman, herself at risk for HD, reflected on the future when her parent would be affected with full-blown HD. “It’s going to be really hard. To know what my [parent] is like now...*I will remember doing this interview and saying to you, ‘I can’t even think about when they will be like that.’ It’s going to be so hard*” (my emphasis).

This chapter and the next are much more than typical ‘method’ sections; they are an attempt to describe the context in which interviews about genetic risk and HD occurred. This context includes not only the interpersonal, dynamic level of the interview itself, but also the wider social context of HD within which these conversations occurred. Accordingly, this chapter will discuss what is known about HD families in Newfoundland and Labrador (NL); in fact, remarkably little information is maintained by the province on this fatal illness. Genetic testing for HD in the province is discussed and statistics on the number of individuals who have been tested and/or received genetic counseling are provided. Participant recruitment is outlined and the challenges involved in recruiting a study sample are noted. Somewhat unusual in mainstream social psychology, this chapter and the next include reflexive commentary as I reflect on my own assumptions, beliefs and reactions over the course of this research. Reflexivity, “where researchers engage in explicit self-aware meta-analysis” is the defining feature of contemporary qualitative research (Finlay, 2002, p. 209). The reflexive commentary scattered throughout this paper should facilitate both comprehension and evaluation of the current research.

Ethical considerations are also outlined in this chapter. From the outset, I was acutely aware of specific ethical issues this sort of work would raise, and provisions were made for informed consent and confidentiality prior to ethical review. However, a number of issues were largely unanticipated, and it was through my own reflexive practices I came to realize and negotiate them. I chose to formally discuss reflexivity throughout this chapter, and elsewhere in this dissertation, with the sincere hope that my

own experiences will provide guidance for other investigators who undertake research in this relatively nascent field.

DNA testing program

Worldwide, international consultation and debate preceded the implementation of predictive DNA testing programs for HD. Guidelines were established by an ad hoc committee of the International Huntington Association and the World Federation of Neurology (1994). It was suggested that requests for predictive testing for HD ideally be approached by an interdisciplinary team consisting of a clinical geneticist, a neurologist, a psychologist and/or a social worker or genetic nurse. Generally speaking, test candidates receive at least two counseling sessions prior to the genetic test. These sessions normally follow a structured protocol according to international guidelines. For example, full information is provided on HD, inheritance pattern and the genetic test. Test candidates are encouraged to reflect on the meaning and effect of a positive test result, a negative test result or on not being tested at all. Inclusion criteria for testing include: Age 18 years or older, absence of serious mental illness or intent to commit suicide after a positive test result, family history of HD and the ability to give informed consent for testing. Test candidates are strongly encouraged to have a support person accompany them to all counseling sessions.

In Newfoundland and Labrador (NL), the provincial medical genetics program (located at the Health Sciences Centre in St. John's, NL) generally follows the international guidelines for testing, with some modifications if necessary. For example, while individuals must normally be 18 years of age to have genetic testing, exceptions are

possible (e.g., a pregnant, mature, at risk minor could be tested; M. Crowley, personal communication, 2004). Referrals are normally necessary to proceed with genetic testing, and they are typically provided by a family physician or neurologist. Referrals can include individuals with a family history of HD or individuals presenting with neurological symptoms indicative of HD, but with no known family history of the illness. In the latter case, these persons are normally referred by a neurologist for molecular confirmation of HD.

Generally speaking, at least three counseling sessions are available to test candidates. The first counseling session includes a review of the family history, education about HD, Mendelian inheritance pattern and predictive testing. This session also includes discussion about reasons for testing and any local support systems that are available in the province. Test candidates will also normally be given written material to review at home. It is possible to provide the DNA sample for testing at this first session, and many test candidates avail of that choice (M. Crowley, personal communication, 2004). DNA analysis is not conducted in Newfoundland; rather, test samples are sent to Alberta for analysis. Test candidates are advised of a two to three month wait for genetic test results, although it is sometimes possible to receive results sooner (M. Crowley, personal communication, 2004).

A second counseling session can be offered while candidates are waiting for test results. At this session, role playing about the results can take place. The test candidate is encouraged to plan the actual results session (e.g., arranging time off work or reflecting on with whom the results will be shared). During this session, a date can be arranged for

the results session. The third session discloses the results of the genetic test, and a support person must accompany the test candidate to this session. A geneticist and genetic counselor can be present; although, normally, the genetic counselor alone delivers the results (M. Crowley, personal communication, 2004). A psychologist is also available for these sessions should that support be required. Alternatively, it is possible for a family physician to deliver test results. This is sometimes preferable when a test candidate lives outside St. John's. Depending on the results, further referrals (e.g., to a neurologist, psychologist or social worker) or follow-up counseling sessions can be discussed.

At the provincial genetics clinic, it is rare to progress to the third counseling session; test candidates usually request their results as soon as possible (M. Crowley, personal communication, 2004). Subsequent to results, a phone call to the test candidate is desirable where other follow-up support can be discussed. At minimum, for those who test positive, an appointment should be offered within two weeks, and for those who test negative, within six months.

Uptake rate for predictive testing for HD

A recent review article reported on the uptake, utilization and outcome of predictive, pre-natal and diagnostic testing for HD in Canada from 1987 to 2000 (see Creighton et al., 2003 for a complete review). For current purposes, only predictive testing uptake will be discussed. In that review, the uptake for predictive HD testing in Canada was roughly 18% of the at risk HD population, ranging from 12.5% in the Maritimes to 20.7% in British Columbia. [Note: Test uptake was defined as the percentage of those who were actually tested, relative to the expected number of at-risk

persons in the population. The 'expected' number was calculated using data from Statistics Canada, factored by the proportion of those at risk for HD – the latter based on a prevalence of 8.4 per 100, 000 with an estimated five persons at risk for every person affected with HD; see Creighton et al., 2003.]

Creighton et al. (2003) found significantly more females than males sought predictive testing, and there were significantly more low-risk test results (i.e., the individual did *not* carry the altered HD gene). The mean age of test candidates in that review was about 39 years ($SD = 12.5$). Data from Newfoundland and Labrador (NL) revealed 38 individuals had undergone predictive testing for HD, translating into an uptake rate of approximately 16.8% for this province. Perhaps unsurprisingly, the uptake rate for predictive testing increased with the advent of direct mutation analysis in 1993 (i.e., since the discovery of the HD gene), but has since remained stable.

The Creighton et al. (2003) review is the sole article I have been able to locate which provides *somewhat* definitive, recent figures on predictive testing uptake for HD in Canada (e.g., Alberta and Quebec did not participate in the review owing to ethical constraints). I have been unable to locate any definitive figures on the number of persons affected with HD in Newfoundland and Labrador (NL). As noted, the Huntington Society of Canada (HSC) suggests the best estimate is one in every 10, 000 Canadians is affected with HD, while five in every 10, 000 are at risk for the illness. It was hoped to elucidate the context of HD in the province, particularly for sample size and recruitment considerations in the current research. Relevant questions included: How many individuals were affected by HD in NL? How many were at risk for the illness? How

many have undergone genetic testing? What have been the test outcomes? What is a typical test candidate profile (e.g., gender, age, marital status, etc.)? Notably, study participants were also very keen to have this information, and several of them were provided with the Creighton et al. (2003) article, which was well received.

Fortunately, the provincial medical genetics program did provide information about the number of persons tested, which will be presented shortly. However, I was unable to locate any definitive statistic on the total number of HD families or individuals in this province, despite searches in several sources. For example, the Huntington Society of Canada (HSC) maintains a mailing list of self-identified members across the country. The society's mailing list for Newfoundland and Labrador (NL) includes 62 self-identified client members (R. Silvestro, personal communication, 2004). However, it is unclear how many of these individuals are affected, at risk, or are caregivers and/or spouses. The only other 'numbers' maintained by the society are quarterly contact statistics provided by HD social workers in the provinces. These record the number of home visits, phone calls and other services provided to HD families in any given month. However, these records still do not accurately represent how many HD families have been contacted in any given quarter, since phone calls or visits are simply tallied. Thus, the same affected individual (or family) could have been called or visited any number of times (W. MacInnis, personal communication, 2004). And, while there is a provincial chapter of the HSC, it does not maintain a formal record of affected individuals or families in the province (M. Janes, personal communication, 2004).

Consultation with several provincial government departments revealed no diagnosis registry for individuals affected with HD in the province. Neither could any statistics be retrieved from the provincial drug program or MCP databases (G. Valvasori, personal communication, 2004). It was suggested that the most reliable data source was hospital separation information (G. Valvasori, personal communication, 2004) which was provided by the Newfoundland and Labrador Centre for Health Information (J. Knight, personal communication, 2004). Hospital separation refers to discharge from hospital after a stay. Table 1 presents the discharge information involving a diagnosis of HD for all the years in which this data was available (1995/96 – 2000/01).

Table 1

Hospital separations for Huntington disease in Newfoundland and Labrador, 1995/96-2000/01 by episode and unique individual

Selected demographics	Hospital separations with most responsible diagnosis or otherwise* of HD	Selected demographics	Unique individuals hospitalized with diagnosis of HD, most responsible diagnosis or otherwise
Total number	42	Total number	20
Mean age at discharge (SD)	59.95 (11.33)	Gender	
		Male	11
		Female	9
Mean length of stay in days (SD)**	17.35 (23.54)	Place of residence	
		Urban NL	6
		Rural NL	14

Source: Clinical Database Management System (1995/96-2000/01), Newfoundland and Labrador Centre for Health Information, St. John's, NL

* Most responsible diagnosis or otherwise indicates that even though separations had a diagnosis of HD, the reason for hospital admission could be HD or any other condition/reason.

**Excludes two separations with a length of stay greater than 365 days; total N = 40 for mean length of stay.

Table 1 requires some explanation. Hospital separations are normally recorded by distinct *episode* during each fiscal year. As shown in Table 1, there were 42 hospital separations from 1995/96 – 2000/01 involving a diagnosis of HD. However, it is possible the same person could have been discharged in multiple years. Therefore, the total number of *unique* individuals with a diagnosis of HD is less than the number of

separations, namely 20 (see last column of Table 1). Note that Table 1 reflects hospital stays due to HD or *any other condition*, as reflected in the 'most responsible diagnosis or otherwise' stipulation.

Patients with a diagnosis of HD were about 60 years of age at the time of hospital separation and the average length of stay for separations was approximately 17 days. Considering the 20 unique individuals hospitalized with a diagnosis of HD, males slightly outnumbered females. Note that most of these 20 individuals resided in rural Newfoundland and Labrador (NL). [Rural NL = any community with a population of 7000 or less, while Urban NL = communities with more than 7000 residents.]

These are the most definitive statistics I have been able to locate about the number of persons diagnosed with HD in NL. It is acknowledged, however, that even this number is incomplete. For example, the majority of participants in the current research had not been admitted to hospital since 1995; thus, they would not be included in the 20 unique individuals or 42 discharges represented in Table 1. Additionally, other demographic information could not be released from the Newfoundland and Labrador Centre for Health Information owing to ethical considerations of confidentiality and anonymity (e.g., mean age of unique individuals; J. Knight, personal communication, 2004). Nonetheless, hospital separation data begins to provide some information about the context of HD in the province.

The provincial medical genetics program provided more recent statistics (to May 2004) on HD clients in NL (M. Crowley, personal communication, 2004). Since the inception of the genetics program, a total of 139 clients were referred for HD. Of these,

HD was ruled out in seven cases, while six clients did not keep appointments. Of the remaining 126 clients, 11 were clinically affected with HD (Note: No genetic test was available at that time; rather, HD was clinically diagnosed). The remaining 115 clients were offered genetic testing. Table 2 displays the number of clients who chose to have either genetic counseling alone or counseling and the genetic test. The outcome of genetic tests for HD is also presented.

Table 2

Number of clients and results of genetic testing for Huntington disease in Newfoundland and Labrador

Genetic test result	Number of clients
Genetic counseling only - not tested	36
Tested, positive result	40 (13 were affected with HD at the time of the test)
Tested, negative result	27
Tested, intermediate allele	7
Tested, results pending	1
Tested, did not wish to receive results	4
Total	115

A full 36 at risk individuals received genetic counseling, but chose not to have the genetic test. Interestingly, four people had the genetic test, but chose not to receive their results. These findings are notable and underscore the complexity of living with the incontrovertible knowledge the genetic test for HD provides. Note also there were more positive than negative test results, in contrast to Creighton et al.'s (2003) review of predictive testing for HD in Canada. Of the 115 at-risk persons who were offered genetic testing, 50 were male, while 65 were female. The mean age at referral was 42.4 years (*Range* 18-90 years).

Mortality data for HD rounds out the available statistical information about the illness in the province (See Table 3).

Table 3

Deaths due to Huntington disease in Newfoundland and Labrador by gender and age

Year	Number of deaths due to Huntington disease	Gender		Mean Age	
		Male	Female	Male	Female
1983	0				
1984	1	Male		65	
1985	2	Male	Female	66	42
1986	1	Male		37	
1987	3	3 Males		53	
1988	1	Male		40	
1989	0				
1990	1		Female		83
1991	1		Female		72
1992	1		Female		60
1993	0				
1994	2	Male	Female	57	54
1995	1		Female		75
1996	5	4 Males	Female	58	72
1997	1	Male		82	
1998	1	Male		74	
1999	1		Female		45
2000	0				
Totals	22	14	8		

Source: Statistics Canada Mortality Datafiles (1993-2000), provided by the Newfoundland and Labrador Centre for Health Information, St. John's, NL

As shown in Table 3, there were 22 deaths in the province due to HD, 14 of which were male. Age at death is also provided, except in years with multiple deaths, where mean age is displayed. Readers are cautioned about the accuracy of mortality data, however. As noted, HD is often not recorded on death certificates; rather, a secondary complication such as infection or aspiration may be the official cause of death.

Interim summary: Provincial context of HD

Definitive information on the number of individuals affected with, or at risk for, HD in NL was seriously lacking. This deficit was frustrating, notably for sample size and participant recruitment considerations. It should be noted that almost every participant in the current study asked the researcher for this information. They too, are frustrated by the perceived ignorance which surrounds HD. Families want to know they are “not alone” with this illness, especially when care-giving becomes necessary and the unique issues of caring for a person with HD must be confronted.

Pragmatically, it is possible the social context of ignorance contributes to the lack of empirical research on HD in the province. The logistics and politics of acquiring access to a rare clinical population is likely a deterrent to social scientists interested in the new genetics. This is extremely unfortunate since almost every participant in the current research expressed his/her hope to raise awareness about this devastating illness. Some recounted struggles for government assistance, life insurance or social support which they believed would be mitigated (somewhat) if only someone ‘cared enough’ to tell their stories. I suspect this is partly why I was so well received by participants in the current research. I have become the one who ‘cared enough’ to give HD families a voice. This was both humbling and overwhelming.

Participant recruitment

No other empirical research has investigated living at risk for, or with, HD in Newfoundland and Labrador (NL). As a result, there was no established local protocol for recruiting participants in the current research. Nor could I anticipate the reception this

work would receive from the provincial genetics clinic, the local and national HD associations, genetic counselors, and most importantly, HD families in the province. As noted, little is known about the prevalence of HD in the province. Accordingly, there was concern to inform as many potential participants as possible about the study, and a variety of recruitment avenues were utilized. Random sampling was not appropriate (or even possible) for the current research. Rather, systematic, non-probabilistic sampling is often used in qualitative research where, "The purpose is not to establish a random or representative sample drawn from a population, but rather to identify specific groups of people who either possess characteristics or live in circumstances relevant to the social phenomenon being studied" (Mays & Pope, 1995, p. 110). Recruitment practices (and the research in general) received full ethical approval from Memorial University's Human Investigation Committee (HIC).

(1) Representatives from the provincial genetics clinic (all current or prior genetic counselors or geneticists) informed past and current HD clients about the study. Not all counselors agreed to assist with recruitment as there were concerns that contact with former clients would necessitate longer conversations than necessary for study recruitment. Recent clients were advised of the research during regular clinic appointments, while past clients were telephoned. In an effort to reduce the amount of time spent on recruitment, it was agreed that counselors would contact only one individual in HD families and ask him/her to inform other family members about the research. This practice, while useful for reducing time and effort spent on recruitment, could have attenuated participation, as there is no way of knowing if the contacted

individual actually informed other family members about the study. In total, 14 families were contacted by the provincial medical genetics program about the current research. Clinic representatives provided clients with the researcher's name and contact information and/or sent an information packet about the study (provided by the researcher; See Appendix A). In some cases, participants gave permission for the researcher to contact them directly to provide more information about the study. Decisions about which individuals should be informed about the research were taken by counselors. Participants who were deemed by genetic counselors as too vulnerable to participate (e.g., because of recent family death or cognitive impairment) were not invited to the study. This practice was consistent with the local ethics committee policy.

It must be noted that the use of recruiters, such as genetic counselors, could have introduced a sample bias into the current research. There is no way of ascertaining exactly how potential participants were identified. For example, perhaps counselors (or other recruiters, for that matter) only invited educated persons to participate. As we will see shortly, the study's sample was well-educated. Underscoring a possible sample bias, one counselor asked me if a certain potential participant had called me (a question, of course, I could not ethically answer). The counselor hoped the person had taken part since s/he thought, "it would be good for him/her to talk to you."

Unfortunately, acquiring access to this rare clinical population was very difficult. Without the cooperation of the provincial medical genetics clinic, there is no guarantee ethical approval would have been obtained for the current research. Additionally, without the support of some counselors at the clinic, I would not have acquired what little

information exists about HD families in the province. Thus, despite the potential bias, the current methods of recruitment were likely the most feasible.

(2) A social worker with the Huntington Society of Canada (HSC) and the chairperson of the provincial chapter of the HSC also informed HD families and individuals about the current research. Recruitment followed a similar method as outlined for genetic counselors. HD clients were either telephoned about the research or informed during face-to-face contact. Once again, participants who were deemed by the social worker and the provincial chairperson as too vulnerable to participate were not invited to the study. And again, some participants gave permission for the researcher to contact them directly about the study. Others were directly provided with information packets about the study from the social worker (See Appendix A). For example, members of HD support groups were recruited in this way. Recruitment outside the genetics clinic was thought to be important since upwards of 80% of the at risk population do not undertake genetic testing. This important group might have been missed had recruitment been solely through the genetics clinic. More broadly, Conrad (1990) has argued that clinic samples, "cause us to miss people with the same malady that manage it outside the medical gaze" (p. 1257). And, indeed, through the chairperson of the provincial chapter of the HSC, several untested individuals were recruited.

(3) A proposal for the current research was conferred to the HSC in Fall 2003. The director forwarded the proposal to the HSC's National Research Council for consideration prior to assisting with study recruitment (I. Horvath, personal communication, 2003). In January 2004, the HSC agreed to include an insert in its

newsletter, *Horizon*, about the current research (See Appendix B). The HSC has 227 members in total on its mailing list for NL who are identified as 'family member,' 'donor,' 'health professional,' and others (D. Wanzo, personal communication, 2003); of these, 62 are self-identified as family members. Three participants responded to the insert about the research and all three agreed to participate in the study.

(4) Family physicians were informed about the current research through an announcement in the Newfoundland and Labrador Medical Association's newsletter, *Nexus* (See Appendix C). Physicians were invited to contact the researcher for more information about the study and to inform any eligible patients; however, no inquiries were received from family physicians, and no participant was recruited through his/her physician.

(5) There are seven neurologists in the province; all were mailed a cover letter and information packet about the current research (See Appendix D). However, no inquiries were received from neurologists, and no participant was recruited through his/her neurologist.

(6) Advertisements about the study were posted in various community locations (e.g., genetics clinic, nursing homes, hospitals and shopping centres; See Appendix E).

(7) Finally, participants themselves informed other family members about the research. This method of recruitment proved most helpful to the current research. For example, one participant provided the names of several other family members who were interested in hearing more about the study; all eventually participated.

In total, 24 individuals participated in the current research; demographic details are presented in the next chapter. Unfortunately, response rate for the current study cannot be accurately provided, given the relative lack of data about HD in the province. As noted, 115 persons were offered genetic testing for HD, and 14 families were contacted by the genetics clinic about the current research (M. Crowley, personal communication, 2004). However, it is not known how many families are represented by the 115 individuals, making it difficult to estimate response rate. Further, there is no way of knowing whether contacted individuals informed other family members about the research. Nor do we know with any confidence the number of individuals who are at risk for HD, but manage it outside the 'medical gaze.' I would guess, however, this number is substantial compared to the 115 at-risk persons counseled by the provincial genetics clinic. On- and off-the-record conversations in the current research revealed numerous at-risk persons. It was common for participants to inform me of aunts, uncles and cousins who had not been tested and didn't want to 'deal with' HD. As a single example, one participant estimated over 20 cousins who are at risk for HD, many of whom have children of their own. To the best of this participant's knowledge, none of these persons has any association with any genetics clinic.

I am aware of only two outright refusals to participate in the current research. A genetic counselor informed me that two individuals declined to participate owing to deaths in their families at the time of recruitment.

Ethical concerns

As noted, the current research received ethical approval from Memorial University's Human Investigation Committee (HIC). Full provisions for informed consent, confidentiality, anonymity and data security were included in the application for ethical review. Additionally, all participants received a two-page information sheet about the study, the consent form and the interview topic guide well before the interview took place (See Appendix A). Consent was obtained during face-to-face interviews, while consent forms were mailed to the researcher for telephone interviews. This research has impressed upon me the necessity of *constant* attentiveness to ethical concerns, long after receiving ethical approval. Research ethics are not as straightforward as ethical applications and graduate ethics classes would have us believe. Here, I wish to highlight some specific ethical issues that arose during the current research.

Like Cox (1999), I found it difficult to obtain *meaningful* informed consent from many participants. They tended to sign the consent form very quickly, sometimes not reading it at all. Of course, it is possible the form had been read prior to the interview since participants received it well before our talk. Nonetheless, in these situations, I took particular care to paraphrase the salient points of the consent form, reminding participants of their right to privacy, confidentiality, anonymity, and refusal to answer any question or stop the interview at any time.

Issues of confidentiality did arise for some participants. For example, one participant was expressly concerned about others discovering her children were at risk. She suggested, "You could write my name all over it [the research report], but I don't

want anyone to know for my children's sake." She was concerned primarily for the employability of her children, fearful that employers would "hold it [their at risk status] against them." Other participants were very clear about who in their family I could (or could not) contact about the study. In these cases, participants were reassured no family member would be contacted about the research without their express permission. In some cases, a participant would tell me about other family members who were at risk, but had not been tested. It was sometimes suggested these individuals might talk to me, but with 'no guarantees' since they appeared to disavow the illness (e.g., they didn't discuss it with these family members and had claimed they would never be tested). During these situations, I normally declined the family member's name. This was a difficult choice since I was very interested in talking to at-risk persons. Theoretically, interviews with this population could help clarify our understanding of some of the issues surrounding the decision to have genetic tests. However, I was concerned about intruding into the lives of persons who seemed to have little interest in talking about HD (at least, according to other family members). In addition, I did not want to be the cause of rifts between family members. Some participants, for example, had already related to me problems that arose in their families when one branch acknowledged the reality of HD, while another didn't.

Others might challenge my decision to decline asking for these interviews; it is possible a somewhat larger sample could have been obtained for the current research. However, I felt it appropriate to place the well-being of participants ahead of the interests of the current research when I felt the two were in conflict (cf. Beeson, 1997). In some cases, I recognized (more clearly than participants) the potential impact of contacting

persons who did not want to discuss HD. And, I reasoned, there were several avenues of recruitment utilized in the current research. Others might hear about the research through one of these other sources and contact the researcher directly if they wanted to participate.

Issues of confidentiality were particularly germane to me during data collection and analysis. In some cases, several members of a family were interviewed for the current research. It became clear they were talking with each other about the study and their own interview experiences. At the start of interviews, for example, participants might refer to a sibling's interview with me. These conversations had to be very carefully negotiated, as I did not want to reveal anything told to me in confidence. And, there were times when participants asked me 'not to tell' another family member what they had said. Detailed notes and memos assisted in maintaining confidentiality as suggested by Cox (1999). I flagged any and all pieces of information which were in any way 'off the record' and kept scrupulous notes on how, and from whom, I received each piece of information about a family. This is one of the ethical issues which differentiates genetics research from other health research. By definition, genetics research involves families. Participants will know each other, and in many cases, they will know each other well. I had not anticipated how difficult it would be to refuse to answer participant questions about a family member's interview. It was common, for example, to be asked, "So, you spoke to my sister yesterday?" I normally responded by reminding participants about privacy and confidentiality and suggesting I could not comment on any interview specifically. Participants seemed to respect this position; however, at the same time, many would

laugh and suggest I was far more concerned about these issues than they were. However, I often knew more about what relatives thought about other members of the family and the choices they have made (e.g., having the genetic test or not), than some participants knew. I had to continually remind myself of the sensitivity of my situation. Constant vigilance was required in order to avoid inadvertently divulging information which might harm a participant.

Issues of confidentiality and anonymity also loomed large during data analysis and writing the results. Sample numbers were small for the current research since HD is a relatively rare single-gene disorder. A broad range of participants was included in the current study (e.g., tested positive, at risk, affected with HD, etc.); however, there were no large numbers in any of these groups. And, as noted, many participants knew each other well, whether because they were related or attended support groups together. As a result, I was acutely concerned with others being able to identify participants, despite the pseudonym issued to each of them. Anonymity was addressed in at least three ways. First, all participants received a copy of their interview transcript (excluding one participant who declined the offer). They were all encouraged to read the transcript and advise the researcher of any portion they might want removed. I reasoned this would give participants an opportunity to reflect on what was said during the interview and whether they wanted to chance someone attributing the remark(s) to them. No participant requested any removals.

Second, significant social details have been changed in order to protect anonymity (e.g., number of children or siblings, place names). Additionally, the plural "they/them"

is used in participant quotes, rather than he/she or his/her, when participants were talking about other family members or affected parents. Changing the pronouns did not alter the meaning of participants' stories in any way, and helped to conceal whether participants were talking about their mother or father, son or daughter - potentially identifiable information. The plural was chosen rather than 'he' or 'she' since it avoids the biases and stereotypes associated with gender. Finally, there are instances throughout this dissertation where absolutely no descriptive information is provided with participant quotes. The reader is not advised of the gender, age or at risk status of the speaker. In these cases, I felt it was the only way participant identity could be disguised and anonymity assured. In some respects, this is not ideal. For example, age emerged as an important variable in considering the meanings and salience accorded to genetic risk. When I thought noting the speaker's age would identify him/her, however, it is unspecified in the quote. While this might be frustrating, it is the only defensible position in light of ethical concerns about anonymity and confidentiality.

Finally, there were also instances of misunderstanding HD and/or the process of genetic testing which has been reported in other HD research (e.g., Cox, 1999; Smith et al., 2002). One participant, for example, thought HD skips a generation and asked me to confirm this. When I was asked for information, I reiterated I was not a geneticist, nor a genetic counselor, and I offered relevant sources of contact information (e.g., local and national HSC, provincial social worker or genetics clinic). However, I felt it would be unethical to withhold basic information, especially when it was asked for directly. Thus, I informed the participant that HD does not skip a generation. This particular incident is a

tangible example of how research can be an intervention into people's lives, not simply an investigation. This participant had thought she had potentially 'escaped' HD, since her parent tested positive and (she thought) the disease skipped a generation. The discovery that HD does not skip generations could now have implications for her thoughts about her own risk status and that of her children.

CHAPTER 6

METHODOLOGICAL APPROACH AND RESEARCH DESIGN

This chapter outlines the research design and methodology, justifying and explaining the design chosen. The current research is about the meaning of being at risk for, or affected with, HD. It is about how families and individuals negotiate genetic risk and illness in their daily lives, but also at more critical life junctures (e.g., marriage, childbearing or career decisions). It explores how, and with whom, families communicate about genetic risk and illness. To date, psychological research into HD has been conducted largely from a clinical perspective. In this body of work, test candidates are represented in the form of pre- and post-morbidity measures (e.g., anxiety), coping skills, or personality traits (e.g., optimism). Clinical research has relied heavily on well-validated survey instruments (e.g., Beck Depression Inventory or Impact of Events Scale) which have been instrumental in establishing individual psychological effects of predictive testing and the relationship between test outcome and individual short- and long-term psychological well-being. Existing studies are also useful in measuring attitudes toward and reasons for (or against) genetic testing, having important clinical and policy implications. However, extant research reveals little about the meaning and everyday lived experience of predictive testing for HD (Cox, 1999). Additionally, it neglects the experience of living at risk for HD (Hunniche, 2001), and I would argue, it also fails to explore the experience of *having* the illness or of caring for a person with HD. Despite widespread recognition within the medical genetics community that predictive testing has serious implications for families (e.g., Tercyak et al., 2000; van't

Spijker & ten Kroode, 1997), few studies have attempted to elucidate just what those implications might be. As such, Cox (1999) argued, "There remains a vast and as of yet unmet need to understand the experience of predictive testing from the perspectives of at risk individuals and their families" (p. 89). I would add the need to move beyond the test and the genetics clinic to explore the experience of living at risk for, or with, HD on families' everyday lives.

In light of the limitations of existing psychological research, an alternative research approach was needed – one that would allow at risk individuals, test candidates and their family members to express in their own words what HD means to them in the context of their own lived reality. Therefore, the methodological approach for the current research was shaped by the necessity of acquiring a rich, contextualized understanding of the everyday lives of families affected by HD. Conrad (1990) argued that the meaning and subjective experience of any illness must be grounded in the sufferer's world.

Therefore, relevant questions should explore:

...how people first notice 'something is wrong' and what it means to them, what kinds of theories and explanations they develop to make sense of these unusual events, what they do about their problem, how they come to seek medical care and with what concerns and expectations, what impact diagnosis has on them and how they cope with a medical label and managing regimes. It must examine the relationship with family members, friends and work associates...consider how people contend with formal and informal disenfranchisements based on a diagnosis, how people adapt to physical discomfort...how medical personnel and others appear to patients...and what strategies people use simply to 'get by' in their lives (p. 1260).

The nature of the research questions suggested a qualitative approach. With few exceptions (e.g., Cox & McKellin, 1999; Cox 1999; 2003; Huniche, 2003; Wexler,

1979), little empirical work has explored the meaning of being at risk for HD or the experience of living with the illness every day. The search for meaning is particularly well-suited to qualitative methodology. Beeson (1997) has argued that, "above all, qualitative research is about meaning" (p. 22). Huniche (2001) suggests that psychological research into HD should be about:

how people live when at risk for HD, how they conduct their lives and what issues are of concern to them. The focus is thus not one of disease in and of itself, but of how, where and when disease becomes an important issue and how it is sometimes not an issue at all (p. 39).

This was a particularly germane perspective for the current research. While critics of the new genetics lament the potential dire consequences of new genetic technologies, I was interested in the perceptions and experiences of those *actually* affected by these technologies. For example, I did not assume that genetic risk or illness would *always* be salient and/or grievous. Nor did I assume that at-risk persons would necessarily perceive themselves as stigmatized; although, I hypothesized that visible symptoms of HD would affect perceptions of stigma (cf. Goffman, 1963). And, contrary to some critics, I did not assume that at risk families would necessarily feel 'pressured' to have the genetic test for HD or to use genetic testing for procreation decisions (e.g., Beck, 1995). I preferred, instead, to elicit narratives from the very people about which critics speculate.

This approach is consistent with qualitative methodology: Common in all approaches to qualitative research is a commitment to studying the world from the perspective of the acting individual (Lincoln & Denzin, 1994, p. 575). While the current analysis was situated within a larger socio-psychological theoretical perspective on risk

and stigma, a grounded inductive method allowed exploration of the issues and stories most salient for participants, rather than imposing a framework *a priori* on their accounts. Beeson (1997) argues this is a critical feature of qualitative work. "Qualitative research is productive because it enables us to discover and document aspects of reality that we cannot necessarily anticipate, and thus to transcend the limitations of our own perspective" (p. 24). Pope and Mays (1995) argued this feature of qualitative methodology is particularly appropriate for studying the social consequences of new genetic technologies.

In-depth, semi-structured interviews and participant observation (e.g., attending a support group, meeting persons affected with HD) seemed the most promising methods for the current research. Open-ended interview questions were chosen over closed questions or survey instruments since it was thought that close-ended responses could not capture the variable responses to risk information for a fatal genetic illness. For example, Wolff and Walter (1992) found that participants gave very different motivations for choosing to have a genetic test when asked to give spontaneous reasons as opposed to a fixed choice questionnaire. In addition, there have been instances of question misinterpretation and ambiguity in studies of predictive testing for HD. Binedell and Soldan (1997) explained how participants in their interview study grappled with variable interpretations and 'subtle distinctions' in questions posed. Questions raised by their respondents included: "Do you mean my immediate response to a test result or how I would cope after a few weeks? By family, do you mean my extended or my nuclear family?" (p. 429). Personal interviews allowed refinement and clarification of questions,

resulting in more precise and hopefully valid information. Notably, participants in the current study posed similar questions. Perhaps the best justification for personal interviews, however, came from a participant in the current research. During our first phone conversation, I had been explaining the research to him - what I hoped to accomplish and why I chose to elicit people's stories about genetic testing and HD, rather than send a survey of some kind. He said, "Know what I would have done with a survey? Thrown it straight in the garbage."

Beyond questions and answers: Illness narratives

For every participant in the current research, questions about genetic risk and illness immediately elicited personal and family narratives about HD. In fact, answers to interview questions sometimes emerged naturally as participants recounted their family's history with HD. The structure of the interview likely contributed to this narrative. At the outset of interviews, participants were asked to "Tell me how it is you are affected by HD." This open question (or some variant thereof) usually elicited long accounts of the family's history with HD, from experiences of predecessors to the family members (including self) of present day.

Within psychology, there has been a growth of interest in the stories people tell about illness experiences (e.g., Crossley, 2003; Frank, 1995; 1998). Frank (1998) reminds us that stories reveal the meaning the ill have constructed around their illnesses. Given this research's primary goal of understanding the meaning of genetic risk and illness, recognition of, and explicit attention to, participant narratives was vital. As Mathieson (1999) argued, "...responses are never just answers to questions. In every interview about

health and illness, there is a narrative underway. What ultimately drives this narrative is the universal need to find meaning” (p. 130). Through illness narratives, then, people give meaning to their maladies and assert some control over the affliction.

Crossley (2003) argued that it is through traumatising events in particular (such as terminal diagnosis) that people frequently, “...experience a renewed need to rebuild and restructure their worlds and they tend to do this through the use of stories” (p. 295). Participant accounts were not analysed strictly within a narrative theory/analytic framework in the current research. However, close attention was paid to participants’ entire narrative. This focus revealed interesting contradictions in participant accounts. For example, some participants downplayed or denied feelings of guilt or blame in response to genetic illness; however, their narratives did contain discourse reflective of both. Similar findings have been reported in other genetics research (e.g., Beeson, 1997)

Initial contact

Initial contact with participants was through a telephone call, whether a potential participant called directly about the research, or when the name of a potential participant was provided through a recruiter. It is worth noting that many of these phone conversations lasted over an hour, underscoring participants’ seeming desire to tell their stories. Before asking for consent to participate, all participants were mailed (in a minority of cases, emailed) an information packet about the research (See Appendix A). The packet included an information sheet about the study, the consent form, and the topic guide for interviews. There is some debate as to whether participants should have access to the topic guide prior to research interviews. However, it was thought that making the

questions accessible to participants prior to the interview was appropriate in the current research for a variety of reasons. As noted, this is the first study in NL on living with, or being at risk for, HD. Families and individuals had not participated in empirical research prior to the current study and were understandably curious (and sometimes apprehensive) about the sorts of questions which would arise during conversation. The initial telephone call to participants often revealed these concerns, sometimes subtly, sometimes more directly. It was common, for example, for participants to wonder aloud about how much 'real help' they could be or if they would even be able to answer any of the interview questions. Provision of the topic guide seemed to help alleviate these concerns. In addition, I suspected that talking about a fatal illness would not be easy. I wanted to reassure participants from the very beginning that there would be no 'surprises' and allow them time to prepare themselves in whatever way necessary for the interview. This practice seemed to work well: Many participants expressed their gratitude for the way they were contacted and the time they received between initial contact and the actual interview. I also wanted participants to consider whether any important issues or topics had been neglected in the interview questions. Given this was my inaugural foray into the lifeworld of a HD family, I was acutely conscious and critical of the interview questions: Were they 'right?' What did I miss? Prior to conducting any interviews, feedback on interview questions was sought from the HSC, the chair of the provincial chapter, the HSC social worker, and genetic counselors, in addition (of course) to academic feedback. No particular suggestions were made regarding interview questions; all agreed the questions 'looked fine,' or had no comment at all. However, several participants raised

issues that had not occurred to me, but they deemed important. Clearly, having the questions beforehand gave participants time to consider other issues that were of import.

A second phone call was made to participants two to three weeks subsequent to the information mail-out (or email). The purpose of this call was to answer any questions and arrange a time for the interview. It is notable that, once again, some of these conversations lasted over an hour. It is likely that these initial conversations, and the transparency with which research was presented to participants, were vital in establishing some sense of trust between participants and the researcher. By the time of the actual interview, participants had already spoken with me twice and had received a mail-out. This method of contact has implications for the depth and breadth of the stories related to me: Many participants acknowledged my sincere interest in hearing their stories of living with HD, regardless of how painful those stories sometimes were. This, coupled with the transparency of the research process, allowed them to feel comfortable in recounting (usually in great detail) their family's experience with HD.

Participants

There were 24 participants in the current research. A range of test candidates/outcomes, at-risk persons, and caregivers/partners was represented (See Table 4).

Table 4

Risk status of interview participants

Genetic test result and number non-tested		Relationship of family member	
Tested positive	3	Spouse	2*
Tested negative	5	Parent	1**
Tested, intermediate range	2	Friend	1***
Tested, but did not receive results	2		
Tested, now affected with HD	2		
Family history, never tested	6		
Total	20	Total	4

* Includes a spouse of person who tested positive but is currently asymptomatic, and one spouse of person affected by HD. This person is also the primary caregiver for her child who is also affected by HD.

Child is currently affected by HD; *Friend is currently affected by HD; all family members are female.

In all, 14 participants had undergone genetic testing, resulting in a range of test outcomes (Table 4). Six participants had not been tested for the altered HD gene and are said to be at risk for the disease. Four family members participated in the interviews. One was the spouse of a currently a-symptomatic person, one was both spouse and parent to persons currently affected with HD, one was parent to a person affected with HD, while one was a friend of a person affected with HD. In the latter three cases, the family member was the primary caregiver for the person(s) affected by HD. These 24 participants represent ten different families affected by HD.

The mean age of all participants was 46.17 years ($SD = 11.26$; $Range\ 21 - 73$).

Other demographic information is summarized in Table 5.

Table 5

Participant demographic information (N = 24)

Gender		Employed	
Female	18	Yes	15
Male	6	No	9
Marital status		Residence	
Single	3	Urban Newfoundland	13
Married/common law	17	Rural Newfoundland	11
Separated/Divorced	2		
Widowed	2	Education	
		< High school	2
		High school graduate	3
		College diploma	8
Children		Some university	4
Yes	21	University, undergraduate	5
No	3	University, graduate	2

Three quarters of the participants were female, and most participants were married/common law at the time of the interview. Almost all participants had children. Over half were employed at the time of the interview. Of these, most were working full-time. Of the nine who were unemployed, three were caregivers of persons with HD, two were currently affected with HD and could not work, two were students, and two were largely seasonal workers. For current purposes, "Urban Newfoundland" was defined as any community with a population of 7,000 or greater, while "Rural Newfoundland" consisted of any community with less than 7,000 residents. Rural and Urban Newfoundland were roughly equally represented in the current research (Table 5). Participants resided in the Avalon, Central, Eastern and Western regions of the province.

One participant was visiting his/her home-town at the time of the interview, but currently lives out of province. He/she is included in the Urban count. Most participants were fairly well-educated; all but two had completed high school and most went on to complete college diplomas or university degrees, two at the graduate level.

It is also useful to compare demographic information of test candidates and at-risk persons, excluding the four family members who participated in the current research. Of these 20 participants, 11 had inherited the altered HD gene (or were at risk for it) from the mother, while the remaining nine had inherited (or were at risk) from the father. Of the 14 participants who had undergone genetic testing, the mean number of years since having the test was 6.5 ($SD = 4.1$; *Range* 0 – 15 years). The mean age at the time of the genetic test was 40.1 years ($SD = 5.2$; *Range* 31 – 48).

Those who had genetic testing ($N = 14$) were older [$M = 46.6$; $SD = 6.04$] than those who chose not to have the genetic test ($N = 6$) [$M = 37.8$; $SD = 13.5$]. Sample numbers are too small to permit meaningful statistics; nonetheless, this finding is consistent with published studies on genetic testing for HD (e.g., van der Steenstraten et al., 1994). Eight predictive test candidates were female, while six were male. The greater number of females seeking genetic testing is consistent with prior research on genetic testing for HD. However, it is not particularly close to the 2:1 ratio typically found in testing studies (e.g., Cox, 1999). All six at risk participants were female.

Of the 14 predictive test candidates, 12 had children. In total, these participants had 23 offspring, some of whom also have children of their own. Five of the six at-risk persons also had children, eight in total. When these children and grandchildren are taken

into account, it is clear that predictive testing produces risk information with significant implications for other family members.

The interviews

The majority of interviews were conducted in participants' homes, with some in the researcher's office. One took place in a local hotel at the request of a participant visiting the study site. All participants were offered the choice of a face-to-face interview; however, one third of the interviews were conducted by telephone. Sturges and Hanrahan (2004) noted that telephone interviewing has typically been regarded as appropriate only for short, structured interviews or in very specific situations. They suggested, however, that the suitability of interview mode was more complex than these simple guidelines suggest. They argue, instead, that suitability can only be determined in light of the particular research endeavor. For example, participants could prefer the relative anonymity of telephone, versus face-to-face interaction with the researcher. In the current research, it is notable that three participants *requested* a telephone interview. This seemed to be the case for participants who were initially nervous about taking part in the study, while convenience was the motivation for one participant. The context of ignorance within which this research was conducted must also be considered in judging the suitability of telephone interviews. As noted, there were no definitive sample numbers from the outset of this research. Time constraints and a provincial public sector labor strike contributed to the delay in receiving the testing statistics from the provincial medical genetics program. These were not received until June 2004, quite some time after obtaining ethical approval for the study in late November 2003. There were days,

sometimes weeks, between discovering potential participants. Rather than risk losing the interview altogether, telephone interviews were conducted with some participants when this seemed expedient and convenient for both the participant and researcher, normally when participants resided more than four hours away from the study site. Sturges and Hanrahan (2004) argued that telephone interviews could be used productively in qualitative research. For example, they found no discernable differences in length or quality between their telephone and face-to-face interviews with visitors and correctional officers at a county jail. Similar findings are reported for the current research. For example, the mean single-spaced page length of typed transcripts for telephone interviews was 17; for face-to-face interviews, 18. Nor could I detect any obvious differences in the richness and depth of the stories related to me. Additionally, similar themes arose in all participant accounts, regardless of interview mode.

That said, however, it is acknowledged that telephone and face-to-face interviews are simply not identical interview modes. In particular, body language and other non-verbal cues are impossible to record in telephone interviews. Thus, particular attention was paid to sighs, pauses or other hesitations in speech during telephone interviews. I also remained alert to signs of distress (e.g., crying).

Interviews lasted from 50 minutes to three hours, with the average being about an hour and 15 minutes. Interviews were semi-structured; they attended to a core set of topics such as family history and experience with HD, meaning of genetic risk and HD, the effect of the illness or of being at risk on everyday life, perceived or actual stigma associated with the disease and healthcare concerns (See Appendix A). Questions were

chosen following a wide reading of the literature in diverse fields including health and social psychology, medical sociology and anthropology and medical and clinical genetics. Discussion with key informants, including representatives from the local and national HSC and genetic counselors, also contributed to the interview guide. Questions were focused in that they inquired about specific issues. However, their derivations represented a constant interplay between reading the literature, theoretical formulations, respondent feedback and key informant advice. In addition, field notes and memos also contributed to question revision as interviews progressed. For example, Beck's (1992) theoretical proposition that 'risk' seems to induce a future-oriented outlook was the impetus for the question, "Do you think that having genetic risk information makes you future-oriented?" After an interview with a participant who had tested positive for the altered HD gene, I had written under the heading *Post-interview reflections*:

I don't know about the future-oriented question. It seemed to be a difficult one for this participant – uncomfortable, painful even to answer. As indicated by the pauses and hesitancy in the response. In essence, this participant indicated there was no future at all. I need to reconsider using this question in upcoming interviews.

Following this interview, I was especially sensitive to participants' reactions to questions and the 'future' question was not asked faithfully in all interviews. Rather, thoughts about the future were probed when participants spontaneously spoke about it and when there appeared to be no apparent participant distress.

Questions were not confined to a specific order; although in general, almost all topics were covered in all interviews. Participants were actively encouraged to discuss any other issues *they* felt were important. Mishler (1991) suggested this approach

enhances the validity of the data since participants themselves pattern the timing, sequence, content and context of the topics discussed. Fortunately, several participants did raise important issues not included on the topic guide (e.g., quality of death and lack of support for children of families affected by HD).

Probing questions were used during interviews to elicit further information or to clarify information provided. These usually took the form of, "Could you tell me a little more about that?" I also regularly paraphrased salient aspects of participants' accounts (e.g., "So, if I am understanding you correctly, that was a really hard decision for you?"). These sorts of probes were vital for my interpretation of participants' stories and helped ensure the validity of the presented interpretation.

Asking a question sounds simple. In reality, however, I sometimes struggled with interview questions. On the one hand, I wanted to be responsive to each participant's conversation. That is, I wanted the interview to *be* an informal, comfortable conversation. This, of course, necessitates the give and take of speech, the dynamics of talking, rather than merely listening or recording answers on my part. It meant that questions could not be asked with *exactly* the same wording, or even in the same order from interview to interview. Nonetheless, I wanted to ensure that the essential *meaning* and content of questions were constant for every participant, despite a slightly different order of administration or deviations from original wording, and importantly, despite my dynamic conversations with participants.

Generally, this tension was managed well; all participants had received the same interview topic guide prior to our meeting. Despite some different wording that emerged

naturally during interviews, I believe each interview encompassed the same content, and I did not perceive any notable differential interpretations of interview questions.

Emotional context

An observation on the emotional context of these interviews is in order. As noted, prior to the interviews, I was worried about the emotional difficulty they might pose for participants. After all, HD is a progressive, fatal illness with limited options for treatment. I reasoned, correctly, that some participants would have watched a parent suffer and die and might even have children of their own who were now at risk for HD. How could anyone, I wondered, talk about these experiences without some degree of pain or upset? I wondered if the interviews would be too painful for participants; I wondered far less about my own emotional reaction. Though, in retrospect, I wondered why I assumed I would be relatively unaffected. (I later realized it was the objective, 'neutral' scientist in me.) There were times during interviews when participants became visibly or audibly upset (i.e., they were crying). I often found my own eyes filling with tears during these moments as well. Prior to the interviews and at their start, all participants had been informed that they could refuse to answer any question or stop the interview at any time. If participants became upset, I immediately asked if we should stop the interview. I also apologized for my own tear-filled reaction, believing this could not be helping the situation. However, no participant wanted to end our conversation. In fact, one participant captured the mood of many when she said:

It's hard to talk about Holly, but you have to, you know? It's good to get it off your chest. And I don't mind you crying, it shows you're human. That's a good thing when you're dealing with this disease.

Corbin and Morse (2003) have cogently argued that while qualitative interviews can cause some emotional distress, there is no evidence indicating this distress is any greater than in daily life. In addition, they suggest that researchers can be more interested in, and empathetic to, accounts of sensitive topics than family and friends. The latter may be embarrassed by the storyteller's emotional response.

With participants' permission, all interviews were tape-recorded and transcribed verbatim. At the beginning of each interview, participants were reminded that they were free to withdraw from the study at any time, for any reason, without penalty. I also assured them they could refuse to answer any question and the tape recorder could be turned off at any point. Participants were reminded their names would not appear in any report published as a result of the study, and their data would be immediately destroyed if they chose to withdraw. I transcribed sixteen interviews; however, due to illness, the final eight were completed by a transcriptionist. Portions of these were randomly checked for accuracy, as recommended by MacLean, Meyer, and Estable (2004) in their discussion of improving transcript accuracy. All participants, excluding one, were provided with a copy of their interview transcript; one participant indicated he wanted only a summary report of research findings. Participants were invited to check the transcript for accuracy and to contact the researcher with any corrections, removals or additions. One participant did request a minor alteration, while another requested additional thoughts be added. Both requests were accommodated. It is notable that in off-the-record conversations with participants, many indicated their gratitude in receiving these transcripts. I believe it gave

them a real sense of agency in this research. One commented, "I can't believe I had so much to say! I just hope it helps other families who are going through the same thing."

Participant observation and other sources of data

In-depth interviews with participants were the primary source of data for the current research. However, I also had the opportunity to attend a support-group meeting for caregivers of persons with HD. The meeting is facilitated by a social worker with the HSC and has been ongoing for about a year and a half. When I attended, the meeting included four caregivers and one at-risk person. It is worth noting that one of the regular meeting members would not attend while an outsider was present. (I was unaware of this until I arrived.) Other group members explained that HD was largely a secret in her family and she did not want to risk exposure to a researcher (or to anyone else for that matter). This is a noteworthy finding and underscores the secrecy and shame that are sometimes associated with HD.

I was apprehensive prior to the meeting and wondered what my reception would be. My concerns were alleviated when, outside the meeting room, one member introduced herself, hugged me, and said, "Thank you for choosing Huntington's." To this day, I am humbled by members' commitment to this research. The meeting lasted for over three hours, and another member informed me this was "a short one." The participant observation occurred early in the research and was instrumental in providing a backdrop for understanding many of the issues in the lives of HD families. For example, it was during this meeting I came to realize the uniqueness of caregiving issues.

My original goal at the meeting was to be as non-obtrusive as possible; this proved impossible, however, as group members encouraged my active participation in their discussion. The group was quite articulate and there were clearly issues of import they wanted me to hear. With the group's permission, the session was tape-recorded and transcribed verbatim. Portions of it are referred to in the results chapters. Four of the five group members also participated in the in-depth interviews. Their demographics are included with the total sample's information in Table 5. The other member (the primary caregiver for her spouse) did agree to participate in an interview; however, scheduling constraints ultimately precluded her participation (despite our arranging the interview on four different occasions). This is, in itself, a notable finding. For caregivers of persons with HD, time is always a premium, and her inability to find time to participate is a tangible reminder of the incredible devotion of caregivers to the person affected with HD.

Not only did participants in the group session provide invaluable insight into the experience of caring for a person with HD, their stories poignantly portrayed the stark reality of this illness on daily family life (even more so than the stories of test candidates and at-risk persons). In many respects, interviews with caregivers were the most difficult for me. There were often tears at some point during these interviews (mine and the participant's), when they described the incredible losses as the disease progressed in a loved one. It was through their stories in particular that I came to care deeply about participants and their families. I worried for them and about them, and became even more committed to giving them a voice. I felt this was the only real thing I could give them.

Their courage and resilience and the dignity with which they face each day never fail to move me, both as researcher and fellow human being.

Honorary participants

At the outset of this dissertation, I introduced the reader to “Jason,” a young man living with HD. During the course of this research, I also met another person with HD, who I will call “Francis.” While Jason and Francis did not participate in the current research in the usual sense (e.g., they could not provide informed consent and did not complete an interview), I refer to them as ‘honorary participants.’ I do so since they were instrumental in allowing me to observe firsthand many aspects of HD I might otherwise be unaware of. For example, through them in particular, I saw how difficult it was for some people with HD to engage in ‘normal’ conversation. I learned to anticipate this in some participant interviews and modify my own response in order to allow a meaningful exchange (e.g., I talked more slowly). I also observed how difficult even the simplest of tasks can be for a person affected with HD (e.g., sitting in a chair or walking across a room). In many respects, it was the people with HD (including Jason and Francis) who taught me most about what it means to live with this disease, how it feels to lose control over your own body and mind, and how it is possible to grieve the incredible losses and yet, remarkably, face each day with courage, strength and dignity.

Notes and memos

An extensive number of field notes and analytic memos were also maintained throughout the course of this research (over 70 single-spaced typed pages). A participant file was established early in the study that included demographic information, method of

recruitment, and my impressions of participant reactions to, and thoughts about, the research. These impressions were normally recorded immediately after any contact with participants (e.g., after the first and second telephone calls and after every interview). They were informative in many ways, but were particularly helpful in recording participants' reactions (including non-verbal cues and body language or, in the case of telephone interviews, pauses, sighs or tears) as the research progressed. This was especially useful for those participants who seemed initially wary about participating. After the very first call to one participant, for example, I had written:

This lady seems nervous about taking part. The family member who provided her name did warn me of this. However, we spoke for almost a full hour. She has already recounted her family experience with HD, her fears about the illness and her reasons for declining the genetic test. Yet, I sensed some reluctance in her too. I am not sure how well I can articulate this – it was more of a feeling I had while talking with her. She was very pleased with my offer to send information for her review. It seems to make me more 'legit' I think. I told her I will touch base again in about three weeks.

After the follow-up phone call to this participant, under the heading "Pre-interview thoughts" I wrote:

She had received the information packet in just a few days and had considered calling me herself to arrange a time for our interview. She seemed very much at ease talking with me today. She said she had been thinking about the interview questions, and in fact, began recounting a rather incredible (and sad) story of the institutional care received by her parent during the last years of life. This was a topic I had not originally thought to ask about, beyond the question about healthcare concerns. After this conversation, I feel much better about interviewing her. She has even provided me with a relative's name. I am especially pleased about this, as I am concerned to speak to people who have not been tested – they are, after all, the majority of the at risk population. We will do the interview [time/date]. I am looking forward to it.

These detailed notes were a valuable, additional source of information during data analysis as well, sometimes assisting with the interpretation of participant accounts. MacLean et al. (2004) have suggested that verbatim transcription, supplemented by detailed researcher notation of nonverbal behavior during interviews, is critical to the reliability, validity and trustworthiness of qualitative research. These notes also, of course, serve as tangible records of my own reflexive practices, and they are quoted when applicable.

Data Analysis

From the outset of this research, data analysis was a constant concern: How could answers to interview questions be analysed while maintaining the integrity of the entire narrative? As noted, the current research was situated in the social psychology of risk and stigma. Thus, some preliminary theories had been identified from the literature before data collection even began. However, other ideas emerged during fieldwork or later analysis. For example, the contention that risk is primarily a negatively-charged concept (e.g., Beck, 1992; Lupton, 1999a,b) led to a focus on the meaning of being 'at risk' during data collection and analysis. Other observations, however, were recorded in field-notes and memos or emerged during participant narratives. For example, the difficulty that many participants conveyed when asked directly to explain what 'at risk' meant to them became evident during data collection. Additionally, the uniqueness of caregiving issues became obvious to me only when I attended the support group meeting. And, as noted, themes arose from interviews which had not been directly queried (e.g., quality of death). In essence, the process of formulating hypotheses, searching the data and revising

hypotheses (e.g., living at risk is not an altogether negative experience) was dynamic and iterative, rather than linear. It began before the first interview was completed and continued throughout data analysis. This form of data analysis did not aim for, and does not claim to be, the 'truth.' Rather, the interpretation presented is one that was systematically warranted from the data analysed.

Interpretative Phenomenological Analysis

Admittedly, the above is a rather vague description of data analysis. As interviews progressed, I became more preoccupied with exactly *how* they would be analysed. Specifically, I was concerned to 'match' the data analysis to the research questions and methodology. Simple coding of data seemed rather bland and an injustice to the depth and richness of participant accounts. Early in the research, I stumbled upon a special issue of the *Journal of Health Psychology* devoted to Interpretative Phenomenological Analysis (IPA) and the new genetics (*Vol. 7, [2] 2002*). IPA (Smith, 1996; Smith, Flowers, & Osborn, 1997; Smith, Jarman, & Osborn, 1999) is a particular qualitative approach having its roots in phenomenology and symbolic interactionism. However, it has been developed in the last decade as a distinct approach to empirical research in psychology (Chapman & Smith, 2002). Of particular interest to the current work, the special issue highlighted the value of IPA to issues surrounding the new genetics. Perhaps more importantly, IPA seemed to suit the current study's goal of understanding the *meaning* of genetic risk and illness for families affected by HD.

Broadly speaking, the aim of IPA is a detailed exploration of how people make sense of their experiences. It is recognized, however, that the researcher's own

perceptions are needed in order to make sense of the personal world being studied. As

Smith (1996) introduced it:

The aim of interpretative phenomenological analysis (IPA) is to explore the participant's view of the world and to adopt, as far as is possible, an 'insider's perspective' (Conrad, 1987) of the topic under investigation. Thus, the approach is phenomenological in that it is concerned with an individual's personal perception or account of an object or event as opposed to an attempt to produce an objective statement of the object or event itself. At the same time, IPA also recognizes that the research exercise is a dynamic process. While one attempts to get close to the participant's personal world, one cannot do this directly or completely. Access is both dependant on, and complicated by, the researcher's own conceptions which are required in order to make sense of that other personal world through a process of interpretative activity. Hence the term interpretative phenomenological analysis is used to signal these dual facets of the approach. (p. 70).

IPA seemed compatible with the aims of the current research: Namely, to discover what being at risk for, or affected with, HD meant to participants, rather than eliciting objective facts about the disease. Smith has also suggested (Smith, 1996; Smith et al., 1997, Smith et al., 1999) that IPA and the social-cognitive approach to health psychology are compatible: Each shares a belief in, and concern with, the chain of connection between verbal account, cognition and physical state (or more generally, 'behavior' in social psychology). That is, much of health psychology is premised on the fact that people think about their bodies, and their talk about those bodies, including talk about illness, somehow relates to these thoughts (Smith et al., 1999). This feature of IPA was particularly notable to me since it accords with the social cognitive bent of mainstream social psychology (largely my own familiar, comfortable background). However, IPA can enrich the social cognitive paradigm in health research with its focus on personal meaning and interpretative activity. For example, a typical health

questionnaire derived from the Health Belief Model could reveal individual differences in perceived genetic risk of disease, despite *equivalent* objective risk. However, the Health Belief Model (and similar social-cognitive models) cannot say much about *why* these differences exist. IPA, on the other hand, can help reveal the nature of these differences. As Smith et al. (1997) noted, "A phenomenologist may choose, for example, to focus on the way two people may speak very differently about what is ostensibly, and medically categorised as, the equivalent illness precisely because of the light that may be shed on the subjective perceptual processes which are operating in the person's interpretation of their health status" (p. 71).

This feature of IPA proved important in the current research since differences emerged in meanings and interpretations of genetic risk (and illness) depending on risk status (e.g., tested vs. not tested or being currently affected by the illness). Chapman and Smith (2002) have argued that IPA is a useful approach for exploring psychological aspects of the new genetics since many of these issues are, "...complex, dynamic and dilemmatic and IPA allows the possibility of engaging with such issues" (p. 127).

Doing IPA

Data analysis in the current research generally followed the guidelines of Smith et al. (1999), which are outlined here. IPA methodology requires a detailed case-by-case analysis of individual transcripts. The aim of understanding and analyzing, in detail, how participants make sense of their experiences requires a flexible data collection method. Chapman and Smith (2002) argued the best way to collect data for an IPA study is with the semi-structured interview. This allows an exchange between participant and

interviewer whereby initial questions are modified according to participant responses.

This method also allows for follow-up or probing of new and interesting issues.

Additionally, the interview structure normally allows participants to raise issues or ideas that are of concern to them, but are not necessarily queried directly. And, as noted, this occurred in the current research.

IPA is flexible, and is appropriate for either an idiographic, case-study approach or a more exploratory approach which aims to theorize about themes at a group level (see Smith et al., 1999, for a detailed review). Either way, IPA follows an idiographic approach to analysis, beginning with an initial transcript and slowly working up to more general categorization or theory (Smith et al., 1999).

The first transcript is read and examined several times. With each reading, one side of the margin is annotated with initial comments and thoughts. These comments may be attempts at summarizing what the participant is saying, some could be connections or relationships that come to mind, some could even be initial interpretations. The next stage attempts to transform the initial comments into broader themes that capture the main features of the initial readings. The other side of the margin is used to document these emerging themes which need not be definitive at this early stage. Connections are then forged between themes until a coherent thematic account of the transcript is produced. To facilitate this process, emerging themes are listed on a separate sheet. Smith et al. (1999) noted that some themes could cluster together at this point or some could be potential super-ordinate themes. A master list or table of themes is constructed next which can also include any sub-themes. At this point, themes can be dropped if they do not fit well with

the overall thematic analysis or if there is little evidence within the transcript supporting it. It is possible at this point to find themes that were not anticipated. In the current research, for example, quality of death emerged as an important theme in some participant accounts.

Once this detailed examination of the first transcript is completed, the master-theme list can be used to begin the analysis of the second interview or the process can begin anew and a master list of themes produced for this second transcript. This process is repeated for all transcripts. Subsequently, master lists for all interviews are read together and a consolidated list of master themes for the group is produced. In the current study, the master list of themes from the first interview was used to inform the analysis of other interviews. It was helpful to remember what had already emerged before identifying what was novel or different in subsequent transcripts. This method also allowed me to pay attention to participants' entire narrative. The downside of this approach, however, is that one can be 'primed' to certain themes or ideas in the data from transcript to transcript. Vigilance and focused attention were required during every transcript reading.

Smith et al. (1999) noted that as each interview is analysed, a final master list of themes should emerge. Connections *across* participant accounts are then made until a set of super-ordinate themes for the group of participants is produced. Each super-ordinate theme is connected to the underlying themes, which of course, are connected to the original annotations and supported by participant extracts.

The above represents a detailed case-by-case analysis of transcripts. Smith et al. (1999) noted that for larger groups of participants (greater than ten), the process is similar; however, early coding is at a somewhat broader level. Rather than identifying 'higher-order' themes, meaningful 'groupings' are identified which collect together the emergent themes. Thus, 'clusters' of themes are generated for each transcript. Once again, clusters of themes are compared for each transcript until a final set of shared themes is identified. In the current research, for example, 'something is wrong,' 'we should have known,' and 'memories of affected relatives' all seemed to cluster together in a meaningful way to describe the 'initial discovery' of HD in a family.

Smith et al. (1999) suggested that diagrams are useful during analysis as they assist in capturing the relationships between emergent themes (e.g., the relationship between risk and stigma and a variety of other variables). The current research used this approach (see results chapters).

Doing IPA, therefore, requires a considerable investment in time and energy on the part of researchers, owing to its painstaking analysis of transcripts. Both approaches to IPA were utilized in the current research. Each transcript was first analysed in detail, as outlined for the case-study approach. This approach was especially useful for becoming "as intimate as possible with the account" (Smith et al., 1999, p. 220). However, I was also interested in theorizing themes at the group level; thus, the transcripts were compared with each other in an effort to capture the similarities and differences between accounts.

There were 431 typed, single-spaced pages of interview and group transcripts generated in the current research. However, computer packages (e.g., Ethnograph) were not used during data analysis. The nature of IPA demands considerable researcher immersion in the data. Therefore, it was felt that relying on computer-aided data analysis would not provide the detailed analysis required by IPA. As a result of this intensive involvement with the data, IPA studies typically employ small samples, raising questions about generalizability (Smith et al., 1997). However, the primary goal of IPA is to capture how particular individuals perceive and respond to their experiences, thereby highlighting the value of each particular case.

Reflections on rigor

Qualitative research is often criticized for lacking scientific rigor (Mays & Pope, 1995). Two common criticisms are that it is strongly subject to researcher bias and it lacks generalizability. That is, when one researcher is so thoroughly immersed in the data collection and analysis, with all of his/her biases, values and beliefs, how can the research be 'objective' and how can the interpretation offered be generalized to any other group or situation? Mays and Pope (1995) contend, however, that all research is selective and there is no way any researcher can discover the 'literal truth' of events (p. 109). And, as noted, generalizability is not the goal of research utilizing IPA. Although, the deliberate search to include a variety of at risk, tested, carer or affected perspectives does increase the representativeness of the current study's sample.

A variety of safeguards were employed to protect the qualitative reliability and validity of the analysis and the findings presented herein (see Denzin & Lincoln, 1994; Mays & Pope, 1995; Mays & Pope, 2000; and Silverman, 2000, for reviews).

(1). Meticulous records of interview transcripts, participant observation, pre- and post-interview researcher comments and analytic memos were maintained throughout the research. In addition, the process of analysis has been documented in detail. Thus, another trained researcher could (in principal anyway) repeat each stage of the analysis (Mays & Pope, 2000). Although, I have serious doubts about the feasibility (e.g., time, ethics, logistics) of this suggestion. In addition, IPA recognizes that researchers will bring their own backgrounds and biases to the analysis, contributing to the unique interpretation of the data gathered in a specific time and place.

(2). Triangulation is an approach to data collection which deliberately seeks evidence from more than one source and/or by different methods. The aim is to look for patterns of convergence to develop or to corroborate an overall interpretation (Mays & Pope, 2000). The current research relied on two methods of data collection: Participant interviews and participant observation. In addition, written accounts of living at risk for, or with, HD (e.g., Cox, 2002) were compared with the oral testimonies of participants in the current research. Additionally, a wide range of participants was sought for the current research (e.g., test candidates with a variety of test outcomes, at-risk persons and caregivers). Mays and Pope (2000) called this, "fair dealing." It is helps ensure that the viewpoint of one group is never presented as though it represents the sole truth of any situation (p. 51).

In addition, the constant comparative method was employed during data collection and analysis. This method requires a constant shifting back and forth between (and within) transcripts to continuously compare the perceptions and experiences of participants who had been selected purposively in an effort to illuminate somewhat subtle, but important differences in the meaning and salience accorded to HD. Barbour (2001) argued that without this constant comparison, samples may have been *selected* purposively, but are not *used* purposively to question the data collected.

(3). Respondent Validation. All transcripts, findings and interpretations were fed back to participants for their review. Participants were explicitly asked, first, to note any errors or discrepancies on their interview transcripts. They were then invited to comment on the validity of the interpretation of interviews as they were represented in a summary report of research findings. Specifically, they were asked to consider whether the presentation reflected a reasonable account of the experience of living at risk for, or with, HD.

Silverman (2000) suggested that both triangulation and respondent validation, while important, are insufficient to settle questions of validity. He advocates use of the constant comparative method, but also argues for the 'refutability principle' (p. 178) – a process whereby researchers actively seek to refute their initial assumptions about their data. Only if researchers cannot refute assumed relations between phenomena can they speak at all about valid knowledge. This process is facilitated by 'comprehensive data treatment' (p. 180) where *all* cases of data are used in the analysis. This suggestion fits well with IPA's detailed analysis of transcripts.

- (4). Analysis of negative cases. Attention was paid to those accounts of genetic risk and illness which seemed to contradict the emerging explanation or the literature in general (see results chapters). Deviant case analysis is a “long-established tactic” for improving the quality of explanation in qualitative research (Mays & Pope, 2000, p. 51). Silverman (2000) noted that it follows logically from comprehensive data treatment.
- (5). Reflexivity. As noted, reflexivity is crucial to evaluate how the researcher and the research process have shaped or influenced data collection and analysis. I have discussed reflexivity and will not reiterate that discussion here. Readers will note the reflexive commentary throughout the results and discussion chapters, however.
- (6). Verification strategies. Verification is the process of checking, confirming and being certain. “In qualitative research, verification refers to the mechanisms used during the process of research to incrementally contribute to ensuring reliability, validity, and thus, the rigor of a study” (Morse, Barrett, Mayan, Olson, & Spiers, 2002, p. 4).

Morse et al. (2002) argued that post-hoc evaluation of qualitative work is inadequate for ensuring reliability and validity. Rather, through verification at every stage of the work, errors can be identified and corrected before they are built into the developing theory. These strategies include methodological coherence, sampling sufficiency, a dynamic relationship between sampling, data collection and analysis, and thinking theoretically. The aim of methodological coherence is to achieve congruence between the research question(s) and the method. In turn, the method should match the data and the analysis procedures. As noted, the nature of the current research questions

seemed well-matched to a qualitative interview and narrative approach, and IPA seemed a good fit to the research methodology.

Morse et al. (2002) also recommend ensuring the sample is appropriate for the research question(s), which will, in turn, ensure saturation of themes or coding in the data. As noted, a wide range of participants was sought for the current study; although greater numbers were desired than achieved for some participant groups (e.g., spouses of test candidates and at-risk persons).

I have discovered what (I suspect) seasoned qualitative researchers have long known: There is a dynamic and iterative interaction between collecting and analysing data in qualitative research. During the course of this research, transcripts were read and reread in an effort to reveal “what is known and what one needs to know” (Morse et al., 2002). This practice encourages theoretical thought – ideas and themes emerging from data were reconfirmed in new data as interviews progressed. With additional interviews, new ideas arose which were reconfirmed (or disconfirmed) in data already collected. Morse et al. (2002) suggested, “Thinking theoretically requires macro-micro perspectives, inching forward without making cognitive leaps, constantly checking and rechecking, and building a solid foundation” (p. 6). I can think of no better way to describe the process of data collection and analysis in the current research. These verification strategies contributed to the qualitative reliability and validity of the current work, hopefully improving the study’s rigor.

CHAPTER 7

DISCOVERING THE FAMILY HISTORY OF HUNTINGTON DISEASE (HD)

This chapter describes participants' initial awareness of their family history of HD. The context of initial discovery extends to current day: It is from this moment onward that participants (knowingly) begin the journey of life with genetic risk, and eventually (for some), illness. Specifically, the chapter illustrates how participants recalled and narrated their initial discovery of the family history of HD and their realization that they themselves were at risk. It is notable that the initial discovery of HD and the awareness of implications for oneself and one's children generally did not coincide. Rather, it was when participants (or other family members) began actively seeking out information about HD that its hereditary nature became apparent. Even then, full awareness of the implications for self was normally gradual, rather than immediate.

It should be noted that many participants expressed uncertainty about where to begin their story. Recollections often extended far back in time to predeceased relatives in an attempt to explain the emergence of HD in the family. Behaviors or personality traits which were unexplained or had been attributed to some 'quirk' of the relative's personality were re-evaluated in light of the family history of HD. In hindsight, many suggested they "should have known" since 'something' was wrong with a parent or other family member. Conversely, other participants suggested they had "always known" about the illness. For them, the family history of HD was just one more element in the taken-for-granted backdrop of family life. Nonetheless, this did not mean they had always

understood the illness or its implications for themselves. Thus, for some participants, knowledge that HD was part of the family did not *instantly* translate into awareness of risk for self. In some cases, this discrepancy was partly due to misconceptions about HD's hereditary nature (e.g., women are not affected).

As noted, memories of unusual behaviors or personality 'quirks' of affected relatives were often invoked to recall the process through which a personal awareness of HD emerged. However, the official label of "Huntington disease" was not normally attached to the relative's illness at the time. It was only in retrospect that participants could classify the behaviors or personalities of relatives as symptomatic of HD. Notably, no participant recalled hearing about HD in any other context prior to discovering his/her own family history. Ignorance about HD was, by far, the most salient feature shaping participants' narratives of discovery in the current research.

Chronologically, participants can be roughly divided into two phases of 'discovery:' (1). Those who discovered the family history of HD in the last decade or so, and (2). Those who have been aware of their family history of HD for many years. These latter participants have known about HD since childhood, or at least since adolescence, and many are now in their forties and fifties. However, years since discovery is only a rough estimate and is meant as a heuristic device to provide a chronological context surrounding discovery for participants in the current research. As noted, many participants had difficulty pinpointing the exact time of emergent knowledge about HD.

Table 6 provides additional key elements of the family history of HD for at risk and tested individuals (N = 20) and for family members (N = 4).

Table 6

Family history of Huntington disease

Tested/at risk (N = 20)		Family member (N = 4)	
Origin of HD		Origin of HD	
Paternal	9	Paternal	2
Maternal	11	Maternal	1
		Unknown	1
Parent with HD		Parent with HD	
Living	4	Living	1
Deceased	16	Deceased	3
Has sibling(s) at risk		Has sibling(s) at risk	
Yes	18	Yes	4
No	2	No	0
Has sibling(s) diagnosed		Has sibling(s) diagnosed	
Yes	5	Yes	1
No	14	No	3
Unknown	1		

As indicated in Table 6, for 11 out of 20 tested or at-risk person, the family history of HD originated on the maternal side; nine traced the history on the paternal side. The affected parent of most test candidates, at-risk person and family members was deceased, some fairly recently. Almost all participants in the current study had a sibling at risk for or already diagnosed with HD; two participants were an only child.

Emergence of HD in the family

Alice Wexler, herself at risk for HD, offered a powerful description of the emergence and perpetuation of HD in families:

First there is the grandfather who has died of “nervous trouble” on the back ward of a state hospital, the uncle who attracts whispers and stares from the neighbors as he staggers down the street, the doctor who says, “Women do not get it.” Rumors of hereditary insanity linger about the family in question, along with a certain atmosphere of secrecy and suspicion. Divorce, arrests, abandonment, suicide punctuate the action. There is always a moment of discovery, when the protagonists finally learn the truth, usually after having several children. In the end, the characters all come to resemble one another, and the action winds down to a predictably gruesome close, with no resolution or release and always the promise of more performances to come. This is the drama of families with Huntington’s disease (formerly called Huntington’s chorea), played out with minor variations on stages around the world (Wexler, 1995, p. xi).

Her depiction is echoed, though only in part, by some participants in the current research. Many participants recalled relatives who were ‘diagnosed’ with ‘bad nerves,’ and all acknowledged that HD was a “never-ending” disease. Importantly, however, most narratives in the current study did not evince a singular ‘moment of discovery.’ Rather, there was often only gradual awareness of HD in the family, sometimes after months or years of odd behavior in relatives. Even for those who had grown up with HD, awareness of one’s own genetic risk was normally gradual, not instantaneous.

Initial discovery of HD in the current research can be categorized into four (sometimes interrelated) themes: (1) ‘Something’ is wrong; (2) Out of the blue; (3) Knowing, but dismissing; and (4) Growing up with HD. Each of these ‘moments’ of discovery is discussed in turn. It is not meant to suggest that these routes to discovery are mutually exclusive, nor exhaustive; rather, they were the central themes which seemed to

organize participants' narrative accounts of how they found out HD was part of their family.

'Something' is wrong

For nine participants in the current research, the family history of HD was unknown until a relative, usually a parent, began to manifest symptoms of the illness. The undocumented family history, however, usually meant that neither participants, nor general practitioners (GPs), initially suspected HD. Odd behavior in a family member, or a general sense of 'something' being wrong, motivates a search for answers, beginning with visits to GPs. While the 'odd' behavior was worrisome for the family, it was often initially attributed to some benign origin. For example, Michelle describes why she did not originally worry about her parent's twitching movements:

...ever since I can possibly remember, [parent] always had this twitching, and I had gone to the doctor with my [parent], and I asked him what it was, and he said that it was the twitching nerve syndrome...it wasn't anything too serious. (...) Huntington's didn't mean much more to me at that time. It was just, well, they told me they had a little virus and that it was going to get better and could be treated, that's all I bothered. It meant nothing more to me at that time. - Michelle, at risk

The GP's assurance that her parent's twitching was nothing 'too serious,' coupled with an unknown family history, obviated the need for Michelle to seek out information about HD. It is only when her parent's 'mysterious illness' progresses beyond a certain point that other medical investigations are initiated and the diagnosis of HD is eventually confirmed. Prior to her parent's diagnosis, Michelle had never heard of HD. "Actually,

we had to ask what it was because we had never heard of it, and I had never heard the title put on anything before.”

Similarly, Serena recalled how she first heard the words ‘Huntington disease:’

I only remember it as a nightmare, that part of it. My [parent] was to several doctors and we all knew something wasn’t right. We knew there was something wrong with them, but no one seemed to know what it was. So the doctor finally referred them to a neurologist – he diagnosed them right away. –Serena, at risk

Serena confirms that neither she, nor her siblings, had ever heard of HD prior to their parent’s diagnosis. Note also she describes the process of initial discovery as ‘a nightmare.’ The family begins to search for information about the illness; what they discover is both frightening and devastating:

We didn’t know what Huntington disease was. It was the first time we had ever heard of it. That was our first experience. Then, [partner] went to the library and when he came back, it was even more devastating. (...) We found out about us, and our kids, and heredity. –Serena, at risk

Note that knowledge of HD in the parent did not *immediately* translate into Serena’s understanding of her own risk and that of her children’s. It is only subsequent to researching HD that an awareness of the implications for herself and her family begins to emerge.

Kathleen also remembered her parent’s irrational behavior and her feeling that ‘something’ was wrong:

Years back, I noticed something with [parent]. (...) Just their behavior I guess. I knew there was something wrong with them, I just couldn’t put my finger on it.

I: So, you didn’t know what was wrong, but you suspected something?

Yes. I didn't know what it was, but I knew their behavior wasn't rational. Their temper also. [Parent] had a bad temper anyway, but sometimes, they would just mouth off – spew from their mouth, spittle and everything coming. It was like a temper that they couldn't control. –Kathleen, tested, intermediate gene

Like Serena, Kathleen suggests that she and her siblings had no prior knowledge of HD. Kathleen explained that a predeceased relative's death certificate noted another illness as the cause of death, underscoring the unreliability of death certificates in estimates of the prevalence of HD (HSC, 2002a).

When her parent was officially diagnosed with the illness, Kathleen confirms she knew nothing about HD, but in hindsight, suggests she can 'see it' now.

I: At this point, though, you didn't know anything about HD, did you?

No, and believe you me, that's come back to haunt us. That bad temper. We just let it go. And [parent] looked kind of spacey now that I look back on it. (...) We got the pictures out once after we knew about this Huntington disease, and you know, could see this vacant look. –Kathleen, tested, intermediate gene

Like Serena, Kathleen didn't realize the implications of her parent's illness for herself and her children until she began to search for information about HD:

Huntington disease, we didn't know what Huntington disease was either. What is Huntington disease? I went on the Internet and got all the information. I was kind of obsessed with it. I found out about it. It was frightening when I first read through. I said, 'Oh my God, what have I got? 'Oh my God.' It was total devastation. People don't know. Even the medical profession don't know.

For families like Serena's and Kathleen's, the undocumented family history of HD leaves family members in shock and disbelief once they begin to research the illness.

Lacking a known family history, HD is often misdiagnosed as other dementia disorders (O' Shea, 1997). One family member, for example, recounted the misdiagnosis of her relative and her suspicion the initial diagnosis was incorrect:

Well, to put a face on it and say it's Huntington's, that must have been a year. I knew there was something neurologically wrong for a long time, but I didn't know anything and they got misdiagnosed, which happens with Huntington's a lot. For a time, they were diagnosed as Alzheimer's. (...) When they were diagnosed, I said, 'No, that's not it.' But I didn't know.

I: You said that to the doctor?

Yeah. He wasn't listening. But I didn't really know, I didn't really have a face or a name. When they said Huntington's, and I began to educate myself about Huntington's, it was, 'yes, yes, yes, yes.'

I: So you could see the symptoms you mean?

Oh yes. I could go down the line. If there was 20 items, I could tick off 16 or 17. They had had it for a long time before I picked up on what it really was. I knew there was something amiss. At first, I didn't really clue in on the neurological part, but I just knew there was something amiss. – Shirley, caregiver

Summary: 'Something' is wrong

This trajectory of discovering the family history of HD was most common in the current research. There was usually a sense that 'something' was wrong with a family member, but this 'something' was generally unknown, often for several years. Lacking a known family history, HD was not normally suspected by family members and GPs alike.

Initial awareness about the family history of HD for participants in this theme normally encompassed several stages: (1) A family member begins to exhibit unusual behavior or personality traits; (2) The family suspects 'something' is wrong, but usually does not know what that something might be. Initially, the cause of odd behavior is

attributed to something relatively benign; (3) The family begins a search for answers to discover exactly 'what' is wrong. This usually involves several visits to GPs before the family member is referred to a specialist (e.g., a neurologist) who normally diagnoses HD; (4) Diagnosis of the family member is followed by personal searches in libraries and/or the Internet for information about HD. Often, a lone family member (usually a female) seeks information and educates the rest of the family. This finding is consistent with Richards' (1993) speculation that females are the 'genetic housekeepers' of the family; (5) Following personal information searches, family members begin to recognize the implications of a parent's diagnosis for themselves and their own children.

It is not meant to suggest that initial awareness followed the straightforward linear path these stages suggest. Nonetheless, discovering that 'something' was wrong with a family member normally involved some or all of these stages.

Out of the blue

Three participants in the current research narrated the initial discovery of their family history of HD as emerging 'out of the blue.' In each case, there was no known family history of HD (or at least the participant was unaware of the history). For these participants, the initial reaction to such news was normally (but not always) one of shock, confusion and disbelief. Note that this trajectory of discovery can contain elements of '*something*' is wrong. For example, an unknown family history of HD and 'odd' behavior(s) by a family member. However, it is distinguished by social and/or geographic distance from a family member affected with HD (cf. Cox, 1999).

Gerald described how a relative died with (what was later discovered to be) HD; however, he had no specific memories of his affected relative:

I don't remember anything about my [relative] who had the disease, other than the fact I went to their funeral when I was about [age]. And that's all I remember. No one talked about it then. As far as what we knew about it? Nothing. –Gerald, tested negative

It was many years subsequent to his relative's death before the family realized the cause of death and the implications for other family members. Gerald recounted a relative's description of the affected family member:

She had said that they were really strange for the last couple of years, they were very abusive and all that kind of stuff. (...) Then, there was all the twitching and jumping. She was told it was sort of an early old age. (...) It was [number] years after their death, or more, before some guy – he was a young doctor, out of practice a couple of years – said, 'Yes, we did a thing on that sometime in class, a paragraph or two.' He sent her on to a geneticist who started the whole ball rolling and here we are.

Gerald's reaction to the discovery of his family history of HD was somewhat different than the other two participants who became aware of HD *out of the blue*:

[Parent]'s onset, as they say, I think they were in their early [age] when we noticed it. At that stage, [neurologist] said it was not likely going to kill you, because it's so late onsetting. It might be a contributing factor, but it is not likely going to be the cause of anything. And it wasn't really. But, from there, we just learned to live with it, knew what to expect to come up because [parent] was so late. –Gerald, tested negative

Gerald's account of discovery is informative since it highlights the importance of the temporal context of HD in one's own reaction to testing and risk. Gerald suggests that because his parent's illness was very late onset, he could 'learn to live with it' and 'knew what to expect.' I asked if he remembered being particularly upset about having the

genetic test. There was no evidence this was a particularly stressful time for him. "As far as I was concerned, it was a done deal basically. If I had it and passed it on, the kids had it anyway, so there was nothing to be done there." He implied that earlier awareness of the family history of HD could have affected reproductive decision-making.

For Gerald then, the geographic and social distance from his affected family member results in discovery of HD 'out of the blue.' At the time, his parent was not showing any signs of illness. Additionally, his parent's very late onset translated into a seemingly less stressful and confusing time than other participants who discovered HD 'out of the blue.' It is also important to acknowledge that Gerald tested negative for the altered HD gene; thus, his reflections on discovering his genetic risk could be different, perhaps less emotionally charged, from those who are still at risk or those who have tested positive.

For example, Lori recently discovered her parent has HD. She does not live in the same community as her parent, and this geographic distance limited her proximity to signs and symptoms of her parent's illness. Adding to her shock and disbelief at discovery, Lori's unaffected parent had recently passed away. She describes her initial reaction to finding out the family history of HD:

I had no idea. I didn't even know what it was. I had just lost my [unaffected parent] with [names illness], and when I did find out what it was, I thought, 'My God, how can this happen to both my parents?' (...) When I found out the risk I was at with the Huntington's [pause]...I'm struggling. -Lori, tested, waiting for results

The context of Lori's life, then, contributes to the difficulty of discovering her family history of HD. She has just cared for a parent through a horrific illness and now must come to terms with her own risk for HD.

Similar to the theme of *'something' is wrong*, discovery initiates an information search and contact with healthcare professionals. As she put it, "That night, I got into it up to my elbows, and I've been into it up to my neck ever since." For Lori, discovering her own risk of HD did come 'out of the blue,' shortly after a parent's death.

Laura, currently caring for two family members affected with HD, recalled her disbelief and devastation at the discovery of HD in her family. An affected relative was long since deceased, and while she knew he/she wasn't 'well,' neither she nor her partner knew there was a family history of HD:

[Relative], we always thought they were an alcoholic and they had epilepsy. But apparently, they had Huntington disease. They were abusive, but this is what it was – Huntington disease. But no one knew. –Laura, caregiver

As in the trajectory of *'something' is wrong*, Laura recalls how she initially noticed 'something' in her immediate family member:

Well, I knew there was something wrong with them. They were losing the use of their legs. One day they came to me and said, 'There's something wrong with my legs. They are not working properly.' They were kind of off-balance.

This prompted visits to GPs and eventually the genetics clinic where testing confirmed a diagnosis of HD. Laura recounted her devastation at the discovery of HD in her family:

When I found out, I went to the church and I cried. I went into the confession box. The priest told me to come around and talk to him. I went around and I told him what happened, and I said, 'Why did it have to be [family member]?'

Laura's GP recommended she talk to another caregiver, presumably believing they could provide support or information. Unfortunately, this experience left Laura even more distressed:

When I went to see them, I must have caught them on a bad day. I was looking for some comfort or some information. They told me, 'Go home, leave your [family member], because you are not going to have a life.' And that's it. I left there, I parked my car by the park, and I must have sat there for two hours crying. I had nowhere to go. (...) It was the most devastating...[crying]...it's the most devastating thing to ever come into our lives. –Laura, caregiver

Laura's discovery of HD in her family member came only months before the confirmation of HD in one of her children. There was no sense in Laura's narrative of a temporal disjunction between her awareness of HD in the family and her awareness of the implications for her children and grandchildren. Rather, she described the entire 'moment' of discovery as coming 'out of the blue;' testing seemed to progress quickly during a time of stress, uncertainty, confusion and fear. Underscoring this difficult time in her life, Laura related a horrific nightmare she had shortly after discovering her family member had HD:

I remember once I woke up – I had had a really bad dream and I woke up screeching. I dreamt [family member] died and I was laying on their headstone. There was grass there and I was laying on their grave...I was trying to haul the sods up to get them out. That was my dream. I woke up screaming. It was so real. The Huntington disease is like this nightmare that I can't wake up from. –Laura, caregiver

The fact that Laura still vividly recalls this nightmare from over a decade ago is testimony to the frightening and stressful situation surrounding the discovery of HD in her family. It was only when her family member was tested, that the family begins to consider the implications for children and grandchildren:

They [children] didn't realize what it was. Myself and [family member] didn't know. We didn't know. It was like someone saying to you, 'Hey, there's a spaceship out there.' [laughs] We had never heard about it.

Summary: Out of the blue

For Gerald, Lori and Laura, the news that HD was part of their family histories came out of the blue. Although they had known (or suspected) a family member was 'unwell,' none had any reason to suspect there was a family history of HD. For all three, geographic and/or social distance precluded their knowing about HD in family members. Gerald's and Laura's relatives had both died without HD specified as the cause of death. Lori's geographic distance severely limited exposure to her relative's illness. Therefore, awareness of the family history of HD was sudden, rather than gradual, for these participants, despite having some vague sense of 'something' being wrong with a family member.

All perceived the news as troubling and began to actively seek out information about HD. Similar to some participants in Cox's (1999) research, all three immediately decided to have genetic testing (in Laura's case, her family members). It has been suggested that taking the genetic test is a method for dispensing quickly with unexpected, troubling information (Cox, 1999). Despite these similarities, however, there was much

variation in these three narratives. Gerald, for example, has known about his family history of HD the longest, and it has been some time since receiving his negative result. His narrative was, in many respects, matter-of-fact and unemotional. This is not to suggest that discovering his family history of HD and/or taking the genetic test were ‘easy’ events; however, testing negative allowed Gerald to relegate HD to the background of his life. Lori, on the other hand, had only recently become aware of her genetic risk. At the time of our interview, she had had very little time to assimilate this information and consider the implications for her life. Also at that point, she did not know if she carried the altered gene for HD. Laura had known about the family history of HD for over ten years; however, she cares for two affected family members, and as such, is faced with the illness every moment of every day.

It should also be noted that elements of *out of the blue* can be seen in ‘*something is wrong*’. Despite knowing ‘something’ was wrong with a relative, the unknown family history of HD contributed to the official diagnosis as coming out of the blue. Recall Serena’s recollection of that time in her life as a ‘nightmare.’

Knowing, but dismissing

For four participants in the current research, the initial discovery of HD can be described as *knowing, but dismissing*. That is, there had always been some vague knowledge that a family member had HD. [It is this element that distinguishes this route of discovery from *out of the blue*.] However, geographic and/or social distance precluded day-to-day exposure to the illness. Additionally, the immediate parent had never shown

any symptoms of HD. Therefore, while the family had knowledge that the illness was 'in' the family, they had no direct experience with HD in immediate family members.

This theme, therefore, contains elements of '*something*' is wrong since all family members knew a distant family member was 'not well,' but the illness was not originally diagnosed as HD. As Marjorie told me:

[Deceased family member], we thought they had bad nerves. We were *told* that was their problem. I remember they had all these movements, but we thought it was nerves. No one knew about Huntington disease. No doctors knew Huntington disease. – Marjorie, caregiver, emphasis in original

It was only when a second family member was diagnosed with HD that the family can, in hindsight, see the similarities in the two relatives. Jackie described her memories of finding out about HD in this way:

Really early on, when [sibling] and I were [age] or so, my [relative] was sick. We knew that there was something wrong with them and it was neurological or whatever. But there had never been a name put to it. Then we found out that my [relative] who lived away in [names place] was diagnosed with HD.
– Jackie, tested negative

For Jackie, there was a vague recognition that HD was part of the family; however, the first affected relative had passed away when she was fairly young and a second affected relative lived quite some distance away. Thus, social and geographic distance intervened to distance HD from her immediate family. And, as noted, her parent was not exhibiting any signs of HD. In this sense, then, HD was dismissed as relevant to the immediate family. "We thought we might have escaped."

It is not until an immediate family member begins showing signs of HD that the family begins to realize they have not escaped and there are implications for others. Julie remembers:

[Affected family member] was living away and we didn't see; there wasn't any sign of anything until they came home...but then we noticed the coordination things and [sibling] and I are on the Internet and the whole time you are thinking that you don't want it to be because you know what it is going to mean for you. -Julie, at risk

The affected family member returned home as the illness progressed. Marjorie remembers:

[Affected family member] came back from being away, and they were very sick. They were very thin. They looked really sick. I thought they had AIDs and had come home to die. That's how they looked. (...) After a while, we noticed a few movements. I noticed they were really moving their head. That's the first thing I noticed, and [family member] noticed too. We went to see [geneticist]. The fact that it was in the family... -Marjorie, caregiver

Testing confirmed that a family member did, in fact, carry the altered HD gene.

For some participants, then, even though the family history of HD had been known for some time, it had no immediate relevance. It is not meant to suggest that participants were in denial about the family history of HD in any derogatory sense. Rather, HD could be dismissed as not relevant to their immediate family since an affected parent was advanced in years and remained relatively asymptomatic.

Summary: Knowing, but dismissing

In *knowing, but dismissing*, participants had known for some time about their family history of HD; however, geographic and/or social distance from affected relatives, coupled with an asymptomatic parent, allowed them to dismiss the relevance of HD for

their own lives. Indeed, for quite some time, these participants thought they had 'escaped' HD. In this sense, this theme also contains elements of *out of the blue*, since the return of a visibly sick family member did occur out of the blue. It is distinguished from *out of the blue*, however, since participants were *aware* of their family history of HD. Despite this awareness, however, it was only with visible signs of the illness in an immediate family member that participants began to acknowledge implications for self.

Growing up with HD

Finally, eight participants in the current research grew up knowing about the family history of HD and many had vivid memories of their affected relative. Some had cared for an affected parent or sibling. In this theme, the family history of HD was generally not hidden from participants, at least not deliberately. However, in some cases, there was a period of time when 'something' was wrong with a family member, but this 'something' was not immediately attributed to HD. This was the case 30 or 40 years ago when HD was even more obscure than current day. [Although, most participants would argue HD remains a largely unknown and obscure illness.] Thus, the historical context of discovering the family history of HD is notable. Brenda, for example, recalled her early experience with HD in this way:

Well, my [parent] died from the disease. I was [age] when they died. In my family, it seems to have manifested in the late twenties – early thirties, whereas normally, from what I understand, people are in their sixties even before the disease starts to manifest. But in my family, it seems young. Actually, when I was growing up, we never really knew what my [parent] had. We didn't know what it was, but they used to have these rages. I thought my [parent] was foolish. I thought they were crazy because they used to take these fits. –Brenda, at risk

Brenda's narrative was filled with vivid memories of her parent; however, it was not until her parent's death that the illness was officially pronounced to be HD:

My [parent] died on [day], and we went to the hospital and this doctor had come from [place] and had experience with Huntington patients and he was the one who diagnosed it.

It is not until Brenda is in her mid-teens that she hears the words 'Huntington disease' attributed to her parent's 'crazy' behavior. She doesn't remember when she realized the implications for herself exactly, but admits certain life decisions (e.g., reproductive decisions) could have been affected by knowledge of her genetic risk:

Myself, I don't know if it's there sub-consciously, but I was [age] before I had [children]. And, I don't know if that's subconscious because the onset is always in the late twenties, early thirties. I feel today that I'm past that and I probably won't get it. —Brenda, at risk

Similar to Brenda, Dorothy had vivid memories of her affected parent's 'illness,' without immediately knowing it was HD. By the time she was entering her teenage years, she knew her parent was 'sick,' but it was not until her mid-teens that a label was attached to the illness:

I: Can you remember what your childhood experience was like with Huntington's? Do you remember understanding what it was?

No, I didn't, not for a long time. It was a nice while after. I guess I must have been probably [age] or [age], before I understood about it. And it wasn't called Huntington's then, it was called St. Vita's Dance. That's what they said they had, but it was years and years after that before they said it was Huntington's disease. —Dorothy, tested, intermediate gene

Brenda and Dorothy, both in their fifties, grew up with affected parents, but neither knew the illness was HD until their mid-late teens. It must be recalled that the

timing of their parent's illness would have been the mid-late 1960's. As both noted, there was very little communication about HD at the time (publicly or privately). There was no sense in either narrative, however, of the deliberate concealment of the family history of HD.

Like most of the participants in the current study, Dorothy did not immediately realize the implications of her parent's diagnosis for herself. In part, this was because Dorothy cared for her parent, and her siblings after her parent's death, which dominated her life. It was not until the birth of her grandchildren that Dorothy proceeded with genetic testing - as much for her own children, as for herself. "I decided I was going to get tested because I wanted to find out and [children] wanted to find out if I had the gene or if they might have the gene, and that way, they wouldn't have any more children." Genetic test decisions will be discussed in the next chapter. For now, note that events in the family (e.g., the birth of a child) are sometimes the catalyst for the salient awareness of one's own genetic risk, not the diagnosis of a parent and the concomitant discovery of the family history of HD.

Another participant also discovered the family history of HD in his late-teens. Unlike Brenda and Dorothy, however, 'Huntington disease' was suspected in his parent, not some mysterious illness or an unidentified 'something' wrong:

I remember something about my [relative] who said to my [parent] they have the best neurological institute in [names place] and should we have the test. They said 'while you're here, why don't you go get it?' So, they must have exhibited some sort of physical symptoms then for them to suggest that. -Tony, tested negative

As to whether the discovery of the family history of HD adversely affected him,

Tony noted:

No, I don't think I knew what was happening. Although, I was starting university at the time. I don't remember being overly upset. In the context of my [parent]: They were already disabled at the time because they had [names illness] and they couldn't work. (...) We were both going to university and I was young. So, the big disability kind of issue with the HD a few months later was sort of an add-on to [names illness]. -Tony, tested negative

Tony's experience underscores the importance of family context and age in explaining the effect of discovering HD in the family. As Tony suggests, his youth and the new experience of entering university combined to preoccupy him, not his parent's illness. Additionally, his parent was already unwell. In that sense, as he suggests, HD was an "add-on" to the preexisting disability.

Other participants grew up with HD in full recognition of the illness. These participants are currently in their twenties and thirties, and in general, there was no question that their relative was affected by HD. Nonetheless, this doesn't mean they fully understood the illness or the implications for self.

I: Can you tell me a little bit about your memory of when you first realized that HD was part of your family?

Well, I've known it always actually. My [relative], I was only [age] when they died, so it was long before that. I remember being terrified of them, so I couldn't have been more than [age] or [age] I guess. That was quite a few years before they died, at that time we called it Huntington's Chorea of course. I have always been aware of the name – you know, not from knowing so much about the disease. I have always known that it could be passed on and so on, but not knowing everything, like neurological and everything like that. -Stacey, tested negative

As Stacey suggests, she had always known the name of her relative's illness and had vivid memories of her fear of her relative. She also implied an awareness of the hereditary nature of HD when she said, 'it could be passed on.' However, at such a young age, she did not understand the neurological basis of the illness.

Stacey's narrative is suggestive of someone who understands the implications of a parent's genetic test result for herself:

...we found that they had an intermediate and a normal. Still not quite sure how this was working, but I was like, 'Intermediate, that's not a normal gene, right?' So, it was only a short time after that that I called [doctor] and asked them if I could get it done [genetic test], just to be certain. If [parent] had the intermediate, I just wanted to make sure that I didn't have either of them.

Like others who grew up with HD, Cheryl discovered her relative was affected with HD in her early teens. While geographic distance precluded everyday exposure to the affected relative, Cheryl recalls changes evident during family trips and visits:

So, grade [] we found out, but they had been sick. I can remember seeing a change since I was a little kid. (...) I think around that time – I never knew about it at that time. I was so young. [Parent] talked about it in bits and pieces. (...) Still there were changes, and I knew things weren't the same as they used to be.
–Cheryl, at risk

Cheryl's narrative suggests that awareness of HD in the family was a gradual process for her. As a child, she remembers being excited about her relative coming to visit, but as childhood progresses, 'things weren't the same.' Despite her youth at the time of initial awareness, Cheryl knew the implications of the family history for herself:

I: So when all this came about, grade [] is still pretty young, did you know what it was? Or that it had implications for you?

Well, my [parent] was really good about it. They really started to investigate it and get a lot of information. And they never kept it from me. As soon as there was a concern raised, me and [siblings] were informed. They talked to us. So we always knew about it. Pretty much, I knew what it was as soon as I was told. [Parent] explained it to us. They didn't try to go around the fact that my [parent] might have it. I understood what was going on. Nothing was uncertain for me. I knew exactly what was going on. –Cheryl, at risk

As this excerpt illustrates, Cheryl was adamant in her knowledge about HD and the implications of her parent's risk for herself. Note the importance of family communication about the illness: Cheryl's parent gathered and distributed information to her children, and as such, Cheryl "knew exactly what was going on."

Cheryl's narrative is distinguished from some of the other participants who 'grew up with HD' by her knowledge of the family history of HD and the illness itself at a relatively young age. Additionally, she implies that she always knew about her own at risk status. Her story is informative in underscoring the importance of the historical context of discovery. Cheryl is a young woman, and discovery occurred in the last decade. While HD remains a relatively unknown genetic illness, this is less so than during the 1960s and 1970s – the temporal and historical context in which Brenda's, Dorothy's and Tony's discovery of HD occurred.

Summary: Growing up with HD

Generally, those who grew up with HD can be classified into two groups: (1) Those who were told the relative's illness was HD; and (2) Those who knew 'something' was wrong with a relative, but could not necessarily label it as HD until later in adolescence. Historical context is important in this respect. Participants in the former

category are in the twenty- to thirty-year age range. These participants experienced more open discussion about the family history of HD than those participants who became aware of their family history in the 1960s and 1970s. This not to say there was active family concealment of the family history of HD. Rather, ancestors were deceased without a diagnosis of HD, leaving an undocumented family history.

The process of discovering the family history of HD in this route was normally gradual, rather than immediate, as in *out of the blue*. Generally, it was marked by living in close geographic and/or social proximity to a family member affected by HD. For those participants who discovered HD several decades ago, there was a time when the family member was unwell or otherwise 'odd' prior to an official diagnosis of HD. In Brenda's experience, official diagnosis came only after her relative's death. For younger participants, on the other hand, there was no protracted period of suspicion about a relative's 'mysterious illness.' Rather, they knew the family member had HD, even if they did not fully understand the neurological and hereditary nature of the illness upon initial discovery.

Discussion

The preceding narratives illustrate four inter-related (but distinguishable) experiences of discovering the family history of HD. Michelle's, Serena's, Kathleen's and Shirley's stories exemplify the type of discovery I have referred to as '*something*' is *wrong*. In this experience, the family history of HD was unknown. Following the protracted 'odd' behavior of a relative, or an uneasy sense that 'something' is wrong, a

search is initiated to discover the source of the 'odd' behavior. The family member is eventually diagnosed with HD. This was the most common route to discovery in the current research. Given the undocumented family history of HD in many families and the low public profile of the illness, this type of discovery is likely quite common. It should be reiterated that this trajectory of discovery also contains elements of *out of the blue*. Since there is no family history of the illness, the diagnosis of HD does appear to come out of the blue. It is distinguished from the latter since there is usually a period of 'something' being wrong with a family member.

For Gerald, Lori and Laura, initial discovery of the family history of HD was *out of the blue*. In each case, the family history of HD was unknown to the participant and initial discovery was often a time of upheaval, disbelief and shock. This trajectory of discovery can also contain elements of 'something' is wrong; however, it is distinguished by social and/or geographic distance from an affected relative.

In *knowing, but dismissing*, there is usually a vague knowledge that a relative has HD; however, social and/or geographic distance precluded day-to-day exposure to the illness. For members of these families, there is a sense of having 'escaped' HD, especially when a parent reaches an advanced stage in life without having shown symptoms. As in *out of the blue*, there is usually upheaval in the family when an immediate family member becomes symptomatic.

Finally, some participants' initial awareness of their family illness was the result of *growing up with HD*. This process of discovery was marked by living in close social or

geographic proximity to an affected relative. Awareness of the illness was normally gradual, and the family history of HD was not hidden from family members (at least not deliberately). This theme can contain elements of 'something' is wrong, in that the family member's illness was not immediately diagnosed as HD. This was normally the case during the 1960s. This route to discovery, in particular, highlights the importance of historical context in the initial awareness of the family illness.

It is reiterated that these 'moments' of discovery are not exhaustive; rather, they seemed to best organize participants' narratives of discovery in the current research. While they are clearly related, they can be contrasted according to several dimensions. These include: the temporal context of discovery (e.g., childhood or adulthood), the geographic context of discovery (e.g., close proximity or distance), the process by which awareness of the family history emerged (e.g., immediate or gradual), the process of recognizing the implications for self (e.g., immediate or gradual), and prior knowledge of HD (even if 'knowledge' means simply having heard of the illness, without necessarily understanding it). These dimensions are summarized in Table 7.

Table 7

Four trajectories of discovering the family history of Huntington disease

Trajectory of discovery	'Something' is wrong	Out of the blue	Knowing, but dismissing	Growing up with HD
Timing	Adulthood	Adulthood	Childhood and adolescence, but sometimes adulthood	Childhood and adolescence
Social or Geographic Distance	Close; but, no one is aware of family history of HD	Usually distant	Usually distant	Close; may or may not initially be aware illness is HD
Process of awareness of family history	Gradual	Abrupt	Abrupt; usually when an immediate family member becomes sick	Gradual
Process of awareness for self	Gradual	Gradual	Gradual	Gradual
Knowledge of HD prior to discovery	None	None	Some; but, usually in a distant family member	Some have full knowledge of HD; some can only label HD at later stage of the illness

While Table 7 is useful as a summary device, it belies how the initial discovery of the family history of HD is but one complex element in participants' personal biographies. Initial awareness of the family history of HD, and of implications for the self, are strongly interwoven with the issue of how participants recall and narrate the events which are integral elements of their life stories. The construction of these life

stories is not an individual enterprise, much as Beck's (1992) 'risk society' perspective might suggest. Rather, participants situated themselves and their memories squarely within the context of their families. As such, an overarching theme within all four trajectories of discoveries was the temporal context of genetic risk and illness. Families have a past and a future, and genetic narratives, in particular, vividly illustrate this simple (but often overlooked) point. Remembering the 'quirks' or 'odd' behavior of long-deceased relatives and speculating on the fate of children and grandchildren of the future, gave the sense that the family history of HD extends infinitely backward and forward in time.

It is important to note, therefore, that to talk of a singular 'moment' of discovery is misleading and fails to do justice to the intricate familial context within which such 'moments' occur. The complexity of family history and family future within which awareness is situated is underscored by the difficulty many participants had in choosing exactly *where* in their biographies to begin their narratives. Awareness of the family history of HD and that one is at risk for a fatal genetic illness are perhaps better described as on a continuum, rather than a binary aware/unaware state of knowledge.

Cox (1999) has cogently argued that the issue of 'when it started' has been overlooked in clinical studies of predictive testing for HD. Clinical research on the psychological effects of testing typically choose an arbitrary baseline (e.g., one month prior to taking the test) from which to measure morbidity and to determine the impact of genetic test results. However, this practice fails to understand or account for the

perceived significance of disease-related events that (normally) long precede such baseline measures. Thus, whether awareness of the family history was abrupt or gradual, known or unknown, could have implications for psychological well-being or decisions about genetic tests. These issues will be developed in the chapters to come.

A note on the act of recalling the process of initial awareness is in order: Memory is not exclusively cognitive. Readers will have noted the strong emotional component in the narratives of many participants. Emotions appear frequently throughout this dissertation since they were prominent in many issues relating to genetic risk. At the time of initial discovery, participants referred to it as a “frightening” time, a “nightmare,” a time of tears and pain. The emotional tone of the stories told by participants is revealing: It underscores a much deeper and far-reaching effect on psychological and familial well-being than clinical research reveals. Further, emotions are intricately implicated in genetic-test decisions, a point generally under-developed in much of the research on genetic decision making. I introduce their importance here in the discussion of initial discovery; however, they are featured prominently in the discussions to come.

Finally, it is hoped the discussion of the initial discovery of the family history of HD has destroyed the validity of talking about *the* HD family. Such a thing simply does not exist. Participants’ narratives revealed much variability in how they found out about the family history of HD and how they came to understand that they themselves were at risk. Similar findings were reported by Forrest, Simpson, et al. (2003) in their interviews with persons at risk for HD.

The tendency to homogenize the experience of living at risk for HD, prominent in much clinical research, fails to account for the temporal and historical contexts in which family history was discovered. Younger participants in the current research, for example, had 'always known' about their family history of HD. They have escaped much of the confusion, uncertainty and upheaval faced by their parents and grandparents. And surely, new genetic technologies and other medical advancements (e.g., medications) are changing the phenomenological experience of living at risk for a genetic disorder. As many of the parents in the current research suggested, their children and grandchildren now have access to information and knowledge that they themselves didn't (i.e., knowledge of the family history of HD). Parents in the current research implied or outright suggested that such information could or should be used in their children's decision-making (e.g., reproductive choices). It is to decisions about genetic risk to which we turn next.

CHAPTER 8

TO HAVE OR NOT TO HAVE: GENETIC TESTING FOR HD

...the crux of this is the impact of the genetic testing. It's a wonderful thing that it's available, but it is one of the biggest decisions you'll ever make in your life. -Julie, at risk

The previous chapter highlighted how participants recalled and narrated their initial discovery of the family history of HD. In that analysis, I emphasized the heterogeneity of routes to discovery and the notable gap between initial discovery and the realization of one's own risk status. Minimal research has investigated how (or if) the initial discovery of HD influences genetic-test decisions (Cox, 1999). While the current research was not expressly concerned with test decisions per se, talking about genetic risk inevitably summoned discussion about genetic testing.

This chapter presents the stories participants told about the experience of deciding upon the genetic test for HD. The chapter is chronologically situated given that the decision-making process follows from discovering the family history of HD.

Motives for testing

Extant literature is replete with studies and reviews that describe reasons for and against testing for HD (e.g., Binedell et al., 1998; Bloch et al., 1989; Evers-Kiebooms & Decruyenaere, 1998; Evers-Kiebooms et al., 2000; Meiser & Dunn, 2000). Findings from this literature suggest the most common reasons cited for testing include a desire for certainty, planning for the future and to inform children. Generally, women tend to be over-represented in testing programs (Cox, 1999; Craufurd et al., 1989) which could reflect their greater involvement in child-bearing decisions (Bloch et al., 1989).

Early research suggested child-bearing decisions were an important testing motive for one quarter of at-risk individuals (Bloch et al., 1989; Craufurd et al., 1989). Later studies confirmed a procreation motive: Evers-Kiebooms and colleagues (Evers-Kiebooms & Decruyenaere, 1998; Evers-Kiebooms et al., 2000) reported that one major motive for taking the test was 'family planning.' For example, post-test, longitudinal data collected in Belgium revealed a definite impact on reproductive decision-making: Approximately one third of those testing positive refrained from having children, one third chose to have prenatal diagnosis, and one third was undecided about future pregnancies (Decruyenaere, Evers-Kiebooms, et al., 1996). A ten-year review of genetic testing for HD in the U.K. revealed that many test candidates were 50 years of age and older (not typical reproductive age). Harper, Lim, and Craufurd (2000) suggested that these participants could be requesting testing for their adult offspring, many of whom were confronted with reproductive decisions of their own.

Findings from genetics centres world-wide confirm that people who have tested positive for the altered HD gene are confronted with reproductive dilemmas (e.g., having children, using prenatal testing). Generally, however, the uptake of prenatal testing is low, although this varies by country and genetics centre (Evers-Kiebooms & Decruyenaere, 1998). It has been suggested that optimism about future treatment for HD will further decrease the demand for prenatal testing (Hayden, Bloch, & Wiggins, 1995). In the current research, none of the at risk participants who had children had undergone

prenatal testing, and many of these participants did comment on their hope for a cure by the time their children were adults.

Reasons for declining testing

Existing research reveals much about motives underlying the genetic test for HD. Far less is known about those who do not request testing, despite their being in the majority (Binedell & Soldan, 1997). Generally, common reasons given for declining testing are related to the emotional and psychological consequences of coping with a positive test result (Binedell & Soldan, 1997; Meiser & Dunn, 2000). For example, those at risk for HD worry they will become hyper-vigilant about symptom watching. Also, a definitive test result could threaten whatever hope the uncertainty of being at risk provides. Little and Sayers (2004) noted, "Certainty is not an object of hope. Hope implies a degree of uncertainty" (p. 1330). Underscoring their suggestion, at risk narratives in the current research did reveal discourses of hope.

The absence of effective treatment is also a barrier to testing for HD (Marteau & Croyle, 1998) as is worry for children following a positive result (Evers-Kiebooms & Decruyenaere, 1998). Fear of employment or insurance discrimination can also be a barrier to testing (Binedell & Soldan, 1997; Guttmacher & Collins, 2003). Notably, one study found non-tested persons were more likely to have discovered their family history during adolescence, rather than adulthood (van der Steenstraten et al., 1994); it was suggested that being at risk for HD had become part of these participants' identities.

'How' is testing for HD actually taken up by at-risk individuals?

Despite the relatively large literature on testing uptake for HD, very few empirical studies have addressed *how* test decisions are taken. Cox (1999) has rightly questioned this curious gap: Clinical guidelines for offering the test stress autonomy in decision-making and the importance of adequate social support during the testing process. Understanding *how* test decisions are taken could help address these issues.

Therefore, the current research explored not only *why* participants requested or declined testing, but also *how* HD testing was actually taken up by at-risk person. It is notable that many participants had difficulty in defining exactly how they came to their test decision. For some, this was due to the perceived lack of choice regarding testing. That is, for some participants, there was no 'decision' to be made, no perceived opportunity for choice.

In what follows, I present the stories told by test candidates (N = 14) and at-risk person (N = 6) about deciding upon genetic testing. Stories can be arranged according to four themes. In the first, the narrator does not speak of the test as a decision per se; rather, there is *no decision to be made*. Stories of *re-evaluating the decision* generally acknowledge a decision to be made. However, decision-making is constructed as a dynamic process evincing shifts in the narrator's thinking about the test over time. Stories of *constrained decisions*, on the other hand, depict the decision to test in terms of obligations to other family members, usually offspring. Finally, some participants request the genetic test only when a specific event triggers their thinking about the family history

of HD and their own risk. These stories illustrate a *test trigger* that motivates agency on the part of the narrator.

It is not meant to suggest that these pathways to the test decision are mutually exclusive, nor exhaustive. They do, however, illustrate the diverse ways in which participants recalled their experience of deciding to request or decline the genetic test for HD. It should also be noted that the test had different salient meanings for participants. These themes appear throughout many of the pathways to the decision, including *test as moral phenomenon*, *test as emotional reflection* and *test as knowledge*. These themes are elaborated throughout the chapter.

It is also striking that virtually every participant constructed the test decision as if it were his or hers alone (cf. Cox, 1999). No participant suggested there was any coercion or pressure to have the genetic test from family, friends, biomedicine or society at large. However, as we shall see, very few stories upheld the ideal of autonomous decision-making (i.e., rational, self-involved, self-directed behavior). Rather, genetic-test decisions occurred in the context of one's family; they were shaped by social and familial ties, obligations and responsibilities to others.

No decision to be made

Three tested and four non-tested participants talked about the genetic test for HD as if there was no decision to be made – they either *had* to know whether they carried the altered HD gene or they simply did *not* want to know. In general, these narratives showed

no evidence of a dynamic decision-making process. Rather, all participants suggested they knew *immediately* that they either did or did not want to take the test.

I: Can you remember what went through your mind when you were trying to make that decision? Was that a tough decision for you to make?

No. For me, I'm the type, I'd just as soon know and deal with it. (...) I'm not the type who could live with not knowing. For me, it was, yes, I was going to get this done. –Victoria, tested positive

There was no evidence in Victoria's narrative of having to think about the test and its implications for any extended period of time. Rather, she seemed to know she wanted the test as soon as her family history of HD was discovered, around mid-life. Victoria did admit, however, that her decision to test was partly for her children. In this way, her narrative also contained elements of *constrained decisions*. "I wanted to find out for myself, but also for them too because it would have been so nice to hear that you didn't have it. Then, you wouldn't have to worry about your kids."

Patsy also discovered her risk for HD during mid-life, long after her children were born. Regarding the genetic test, she noted:

I *had to*. I would have died not knowing. I had to know. Whether it was good or bad, I had to know. Now, I don't know what I would have done if it had been positive. But I do know, and it's a terrible thing, but I was not going to live like [parent]. I would not live that way. –Patsy, tested negative, emphasis in original

Like Victoria, there was no evidence in Patsy's story of a prolonged period of thinking about the genetic test as a decision per se. As she says, she simply *had* to know. Having watched a parent suffer with HD, however, she knew she would not 'live that way' if she had tested positive for the altered HD gene. This issue will be elaborated

upon shortly. For now, note how disease-related events in the family color members' perception of genetic risk and illness.

Non-tested persons also spoke about the genetic test as if there was no decision to be made. In these narratives, however, at-risk individuals did *not* want to know whether they carried the altered HD gene. Participants cited many reasons for rejecting the genetic test, replicating previous research: Fear of being unable to cope with a positive result, fear of constant symptom-watching, anticipated guilt if other siblings tested positive, worry for at risk children and the fatal nature of HD. Despite these reasons, there was no evidence that participants engaged in a cost-benefit analysis of the test for any protracted period of time:

If I had the gene that carries it, I think I would dwell on it too much. (...) Like I had no, even from when they asked me if I wanted it done, I had no desire to have it done right from the start. –Sherri, at risk

I just said no, I don't want to know. If I'm going to get it, I'll get it, but I don't want to know because I felt that if I knew, I would be waiting for it to happen and every move I would make I would be wondering, 'Well, is this the start of it?' and I just didn't want to know. –Brenda, at risk

For non-tested persons in particular, emotions seemed to play an important role in the 'decision' to decline the genetic test. For these participants, the availability of the test motivated *emotional reflection*, both of current and anticipated emotions. Anticipated fear, guilt and worry permeated participant narratives:

Right now, I don't think I want to know. It's kind of scary. I think if you know, I'm afraid, I think it might come on quicker. (...) I'm afraid I would be thinking about it all the time. (...) Another thing I think about, if me and [siblings] did get tested, and I don't have it and they do, I think I would feel guilty for being the one who was lucky enough to get away with it. –Roxanne, at risk

I looked at it in a way in which, if I had cancer, I wouldn't want to know – there is no cure for it, so I would only worry about it. I am an anxious person...so I felt that it would make my life more miserable. –Michelle, at risk

It is worth noting that at risk participants in their twenties and thirties also cited their age as an important reason for declining the genetic test. The affected relatives of these participants did not develop HD until later in life (usually in their sixties). Participants noted they were still 'too young' to start worrying about HD at this point in the life course. These findings challenge Evers-Kiebooms and Decruyenaere (1998) who claimed, "It is obvious that demographic variables hardly play any part in the decision" (p. 23). Gender differences in test uptake also challenge this contention (Cox, 1999).

Summary: No decision to be made

Typical research on genetic decision-making constructs the rejection or acceptance of genetic testing as a 'choice.' As the preceding narratives illustrate, however, there is *no decision to be made* for some at-risk individuals. As Cox (1999) put it, the 'decision' is a 'self-evident act.' In these narratives, there was little ambivalence and minimal conscious reflection on the test as an opportunity for choice. Rather, for these participants, they simply knew they did (or did not) want to know whether they carried the altered HD gene. Nowhere in their narratives did these participants regret their choice or suggest they would do differently.

It is notable that five of the six at risk (i.e., non-tested) persons in the current study grew up with HD. Each had some experience with an affected relative - some had even cared for the relative. The relationship between initial discovery of HD in the family

and decisions about genetic testing has been relatively unexplored in the literature (Cox, 1999). Findings from the current study would seem to support the limited research which suggests that knowing about HD since adolescence (van der Steenstraten et al., 1994) or being in close proximity to someone with a genetic illness (Duster & Beeson, 1997) is associated with declining a genetic test. Participants who have cared for an affected relative had vivid, often frightening, memories. Perhaps it is these memories which underlie the emotional undertones which were quite evident in the narratives of at risk participants.

Constrained decisions

For six participants, the decision to have the genetic test was taken not for themselves, but for their children. I have categorized these narratives as examples of *constrained decisions*. There was a sense that participants had *no choice* but request the test. As in the narratives of 'no decision to be made,' the test was generally not conceived as an opportunity for choice, at least not *for self*. Rather, taking the test provides participants' children opportunities for choice, especially regarding reproduction. In these narratives, in particular, the genetic test is experienced as a *moral phenomenon*: Perceived moral duty to one's family constrained the perception of the genetic test as an opportunity for choice. The salient meanings of the test for these participants included: the fulfillment of obligations to others (especially offspring), an opportunity to rule out HD or a contribution to HD research.

For example, Dennis recalled how he didn't originally plan to be tested. "When [siblings] found out about it, I wasn't tested at the time – I didn't want to be tested." Later in the interview, however, I asked if it was acceptable for those at risk for HD to decline genetic testing:

To a certain point, yes, if they don't want to find out. But the part is, what about their children and grandchildren? Maybe I never would have gotten tested if my [children] hadn't asked me. And, they had the right to know. That's my point of view. I didn't want to know. It was for them. (...) It came to the point where I could have said no and walked away, and they could probably have gotten tested anyway. But, why put them through it if they didn't have to? I had no other choice but do it then for the sake of my family. –Dennis, tested positive

Dennis was clear that he didn't want to know if he carried the altered HD gene; however, when his children were considering starting their own family, Dennis suggested they 'had the right to know.' As he noted, Dennis perceived 'no other choice' but to take the test. At no time during the interview did Dennis appear to re-evaluate his initial thoughts about declining the test; rather, his perceived obligation to his children motivated him to take the test.

Like Dennis, Julie also noted, "Because my testing is not for me, it's for [children]." Julie has not yet received her genetic test result, preferring uncertainty to certain knowledge. Duty to her offspring, however, constrains her ability to sustain a 'wait and see' approach. As she said, "It's knowing that they're at risk and they'll be [age] in a couple of months and they need to know soon because I think it is an issue when you are deciding about children or not." –Julie, at risk

There was a sense in these narratives that genetic testing could provide *beneficial* knowledge for at risk children. Jerry, for example, noted how he could not ignore his risk:

The thing about it now, is knowing this, I sort of have to deal with it because I have four kids. I want them to know somehow, for them to know someday, and give them the fair choice of whether, ok, do I have kids when I get married, or whenever? – Jerry, tested positive

Jerry suggests he has to ‘deal with’ his risk and perceives the knowledge derived from genetic testing as beneficial to his offspring. Many tested participants in the current research spoke about genetic risk information in this way. When I asked him if it was acceptable for at-risk person to decline genetic testing, Jerry said:

From a scientific or health perspective, I think that’s wrong. I think they should know. I think it opens a lot of doors, a lot of options for people. That’s the way I look at it. Denying something never fixes the problem. (...) You have to realize there are choices in life one has to make with diseases. This is a cruel disease I’d call it. So, it [testing] sort of gives you options. With children, with family, I guess that’s the biggest thing. –Jerry, tested positive

For Jerry then, taking the genetic test is the only acceptable ‘choice’ given his belief that genetic test information can provide ‘a lot of options for people.’ Primarily, his test result can be used by his children in their own marriage and reproductive decisions.

Summary: Constrained decisions

Stories of constrained decisions poignantly depict the social and familial ties which limit the range of acceptable ‘choices’ open to persons at risk for HD. Perceived responsibility to offspring, in particular, often acted to constrain participants’ test agency.

Narratives of constrained decisions had distinct moral undertones. At times, there was a strong conviction that it was unacceptable to have children without first ensuring

they were free from HD. Clinical guidelines for offering the genetic test for HD all stress autonomy in genetic decision-making, and non-directive genetic counseling aims to promote this goal. Yet, narratives of constrained decisions make it difficult to maintain seriously that test decisions are purely rational – self-interested, self-directed and proceeding in a cost-benefit analysis. Test decisions are taken in the context of one's family, and it is vital that healthcare professionals and social scientists who work with at risk families recognize this simple, but oft overlooked, point. More broadly, Finkler (2001) argued that advances in the new genetics are medicalizing the family and kinship. She noted, "In contrast to the broader societal process, in which individualism and freedom of choice are emphasized, the medicalization of kinship creates a tension between individualism and choice and an orientation to family and kin" (p. 244). Stories of *constrained decisions* would seem to support her argument.

It should also be noted that the theme of *test as moral phenomenon*, while prominent in these stories of constrained decisions, was also evident for participants in other decision pathways. One tested participant who perceived *no decision to be made* spoke about at risk children: "Before they have kids, I hope they do get tested. I don't want this passed on to the next generation." Another participant expressed disappointment in the reproductive choices of offspring:

Now, [children] don't know if they have the gene or not. They went ahead and had [name]. But it's kind of disappointing to me. Like I told them, I went through all this for you guys. For myself and for you, so you could be aware if you had it or not. You make the decision to have children. I was hoping they would make the right one, but I guess they wanted children.

Negative case analysis revealed only a minority of participants who expressed their disapproval of using the genetic test to inform reproductive decisions. One at risk person commented, "What do we want – a society of all perfect people? I like to think that we will never become that cruel as a society that we would expect people who have genetic diseases to give up life." Another expressed anger at a relative who suggested their offspring should take the genetic test before having children:

...this was the one thing that bothered me, my [relative] said they had it and they were getting their [children] tested, and if they had it, they didn't ever want them to have children. That to me bothered me. (...) There's a possibility they might have children who might not have it. (...) What's the point in not having kids? What's the point in basing your whole life around a disease that's not going to affect you, more than likely, until after you're fifty?

It is notable that both participants who rejected the use of genetic testing to inform reproductive decisions each had children of their own and each had not been tested; both also grew up with HD.

Re-evaluating the decision

Narratives of *re-evaluating the decision* revealed the (sometimes) dynamic nature of genetic-test decisions. In these stories, the narrator generally *does* perceive the test as an opportunity for choice, both for self and offspring. Some participants initially wanted the test, while some initially did *not* want the test. Others decided to have the test, but were quite some time before requesting their test result.

Taken together, these narratives represent the kinds of stories I initially expected to hear – permeated with ambivalence, confusion, uncertainty and possibly fear about the test result. My expectation arose, in part, because I have never been able to decide

whether *I* would take the test if I had a family history of HD. I am grateful to Cox (1999) who recognized that *expecting* certain stories would risk 'not hearing' other types of narratives (e.g., no decision to be made).

In stories of *re-evaluating the decision*, participants exhibited changes in their thinking about the genetic test over time. Thus, they illustrate the shifting nature of thinking about genetic risk and help explain how variables such as disease-related events in the family, perceived coping ability and age influence test decisions. These narratives, in particular, evinced critical thought about the genetic test for HD, less pronounced in stories of *no decision to be made* and *constrained decisions*. It should be noted, however, that elements of re-evaluating the decision could be seen in at least one of the narratives of constrained decisions. For example, while Julie noted that her testing was primarily for her children, there was ample evidence in her narrative of critical thought about the implications of her test result for herself, her partner, her siblings, her work-life and her life in general.

One participant recounted how she was originally 'quite adamant' about not having the genetic test. Over time, however, a family member begins to show visible signs of HD and as such, "...it was in your face and then it was harder to ignore it." She went on to say:

And, I was in my late [age], so it was more immediate. And then there was, well, making choices about how you would live the rest of your life. (...) And there was for [partner] and I together. I mean, you think when I retire, I will travel. When I, you know, in a few years time, we'll build a summer house...-Female, tested negative

This narrative illustrates the many factors (and people) which are sometimes figured into genetic-test decisions. There is the additional burden of symptom-watching:

For about a year, every time I groped for a word, or couldn't find my keys, or dropped something on the floor, I would have to think if that was the start of it. (...) And then second-guessing and worrying and it did actually get to the stage where it would be less worrying to know than not to know.

Finally, this participant introduced a theme that I had not anticipated but which was broached by several other participants as well, that of rational suicide (Davis, 1999; recall Patsy's vow in the last chapter, "I would not live that way.").

When it came right down to it, it would change how I would live the rest of my life. There are decisions that you make and the things that you postpone and the 'after I retire' and the 'later on we will' things and I wanted to be able to do that. The other thing is, I never told this to the nice people down at the clinic, I also wanted to be able to control my own death.

I: In the event that you had carried the gene?

That before it got too bad that I could do anything about it, that I would probably be the kind of person who took that kind of control.

I: You are not the only one to say that. That has been an issue raised to me as well, which is interesting because it's one I didn't think of.

Oh yes, at the top of the list. I didn't think that I should tell [genetic counselor] that, but, they are all so sweet and all caring people, I didn't tell [genetic counselor] that one.

For this participant, then, a number of variables combine to influence their decision about the genetic test: An affected family member, age, thoughts about the future, concern for partner, and a desire to have some control over the end of life experience. It is notable that the latter motive was not discussed during genetic counseling sessions.

A second narrative of *re-evaluating the decision* highlights the opposite pattern of thinking about the genetic test: Initial thoughts are favorable, but with disease-related events in the family, the test is eventually seen as unacceptable:

Well, at the beginning, I was undecided. In fact, I was saying, 'Yes, I'm going to get tested.' Then I said, 'No, I'm not.' What made me *really* not get tested is when, of course, my [relatives] went for their testing. [Names relatives] tested positive. So, that was a real, real...that was another really big setback. Well, I just made up my mind then. Personally, I said, 'No, I don't want to know.' That's not saying that just because two out of their three....it could be none of us. Hopefully, there's none out of the four. But, we all know what the chances are. We decided it's better not to know. There's nothing that can be done. —Female, at risk, emphasis in original

For this participant, the test results of other extended family members, coupled with the fatal nature of HD, dissuade her from taking the test herself. Worry for her children also affect the decision:

To me, if my children know, they would worry about me, if they knew for sure. Whereas, now, they don't know. It's for your family that you have to think about. Even though they know I'm a 50:50 chance, the same thing goes for them, I think if they knew, I don't know how they would feel about it. I'm afraid that they would really worry about it. It's a lot to think about.

Regarding reconsidering taking the test she said:

I don't think so. There are insurance issues. If you know, there's no insurance. Sometimes I feel that even medical doctors, if they know you have the HD gene, then anything that goes wrong with you, they'll assume it is the HD. They don't check for anything else. There are different things I thought about and I said, 'No, I don't think I will ever get tested.'

Now, in the meantime, my kids, they know about HD. They know and they know that for them, it's up to them. We told them, we talked to them, and we told them if they wanted to go get the test...I know then if they have it, I have it. If I don't have it, they won't have it.

I: You're right. If they choose to have the testing, you will know then for yourself. Would you be ok with that?

Oh yes. I would. It's their decision.

This narrative, in particular, highlights a distinct ethical issue that surrounds genetic testing. Should her children decide to take the genetic test, she could be forced to know her own genetic test result. And while she indicates she would be 'ok with that,' it is an issue that deserves attention during counseling sessions. Notably, this participant has had no contact with the genetics clinic. Her narrative also vividly illustrates the myriad factors that sometimes contribute to genetic-test decisions.

Unlike those narratives of *no decision to be made* and *constrained decisions*, narratives of *re-evaluating the decision* were filled with shifts in the narrator's thinking. This fluidity of thought was also revealed in the narratives of those who decided to take the genetic test, but did not immediately request results. One participant, for example, recounted how concern for self and for siblings delayed the receipt of test results:

I just couldn't because I think I had went through a lot mentally. And I didn't think I could handle another. And, I didn't want to put it on my [siblings] either. (...) I thought it was too much for me and for them to handle.

Over time, however, there came a point where she simply *had to know* the test result:

I knew I had to get it done because every day it was on my mind. Do I have it? Do I not? I had to come to some sort of a resolution. I had to find out, I couldn't go on the way that I was. So, when I wanted my results, I knew I was ready.

I: So for you, it was something you just had to know?

Something I had to know, and in order for me to move on with my life, I *had* to know. I was caught in the limbo of everyday thinking about it, 'what if, what if.' So, I just couldn't do it anymore. –Female, tested, intermediate gene, emphasis in original

In this narrative, then, there are elements of *no decision to be made*. While initial thoughts about the test were favorable, disease-related events in the family and her own fear of coping with a positive test result combined to delay hearing her results. However, there came a point where she simply 'couldn't do it anymore' and like the narratives of *no decision to be made*, she 'had to know.'

A final narrative highlights age as a factor in genetic-test decisions. For younger at risk participants, there is a need to get career, marriage, and insurance issues settled before testing. As one participant referred to it, "real life things."

I haven't had the test. One of the interesting things is I did [think about it]. When I first found out, I was like, 'I don't know why anyone wouldn't find out if they had it or not.' (...) But at the time, I was too immature to know what the implications are. There are certain things: Before I get married, before I decide to have children, I will definitely test. –Female, at risk

For this participant then, her youth at the time of initial discovery of HD prevents her from considering the full implications of genetic testing. As she ages, she begins to think about marriage, career, insurance, her future – 'real life things.' Additionally, she recognizes the burden of coping with a positive test result and questions having that knowledge at a young age:

The other thing is, do I want to know right now? Why would I keep myself awake at night going, 'Oh no, I have Huntington's.' I keep coming back to cancer... I could die from cancer before I die from Huntington's, or anything else. I could get hit by a car tomorrow and sometimes not knowing is just as good as knowing. I'm only young. –Female, at risk

Summary: Re-evaluating the decision

Narratives of re-evaluating the decision revealed the changing nature of thinking about genetic testing for HD. In these stories, participants moved from initially wanting the test, to considering the implications of a positive result for self and others, to eventually rejecting the test. Others shifted from initially rejecting the test, to coping with the burden of uncertainty, to eventually taking the test and/or receiving results.

In these stories, participants normally perceived the test as an opportunity for choice. In fact, some participants specifically cited planning for the future as a motive for having the test. This was the case for participants in mid-life, but also for younger participants who wanted to have future affairs settled (career, marriage, insurance) before proceeding with the test. These narratives are illustrative as they highlight the importance of age, perceived coping ability and disease-related family events in the decision to take the test.

Test triggers

In these narratives, an event or behavior normally triggered participants' thinking about their family history of HD and their own risk. Three participants described their motive for taking the genetic test in this way. David, for example, recounted how he could see changes in himself, primarily at work:

Well, I was seeing a counselor about a year before, through an employee assistance program. I was sensing, finding my way with them on this, because I could see problems occurring at work. (...) I wanted to protect myself, so I wanted to get things documented and decided to go for a particular test. I didn't disclose that I could be having problems with HD. I wasn't sure, and when I went to get the test, I didn't expect a positive result. –David, affected with HD

I: Is that right? Okay.

No. But, I knew that the problems could be HD, but I didn't expect a positive result.

I: Ok. So then you noticed problems yourself, or things happening?

I was in a stressful workplace...It was a competitive workplace and short-staffed and they wanted me to work extra hours. I experienced a lot of paranoia, obsession, isolation. I was snowballing really fast and really big. I ended up and got the predictive test done.

David grew up with HD and had known about his own risk since adolescence.

However, it is only when he noticed changes in himself – primarily at his place of employment – that he was motivated to have the genetic test. As he later told me, “I felt it was the thing I needed to do at the time to protect myself. I didn't want to end up getting fired.” David noted he did not expect a positive test result, despite his knowledge that his a priori risk was 50:50. Perhaps his expectation that he did *not* carry the altered HD gene helps explain why he did not have the test until some specific behavior(s) triggers his thinking about his risk.

As in stories of *no decision to be made*, Tony recounted how:

There came a day when I had to know. I was going through...I recognized it as something more going on with me than normal. I recognized it, as there was a psychiatric side to HD. I was well aware that I wasn't physically showing symptoms or signs. But I felt that what was going on with me at the time was certainly more of a - how shall I put it? I need to know if I'm just being a jerk now or whether this is part of a disease process. That's the way I can put it. That it got to a point where I had to know. –Tony, tested negative

While Tony had known about his risk for some time, he ‘never bothered’ with testing until he perceived changes in himself. Perceived changes in his behavior trigger

his thinking about the family history of HD; he realizes that genetic testing can eliminate HD as the cause of that behavior:

For me, [year of testing], well I would have been [age], all the major life decisions were made. But obviously, at that time, I was under a lot of stress or something was going on with me - whether it was depression, whether it was whatever I had, I hadn't spoken to a doctor about it per se, but I needed to know. I needed to eliminate that as a possibility. That's why I ended up going.

Finally, Steven recalled how he was already manifesting signs of HD at the time of his genetic test:

I was showing signs of the disease then.

I: Really?

Yes. [geneticist] suggested I be tested right away. My head was moving. My head was moving when I was reading newspapers. That's what Dr. [geneticist] saw.

I: Ok, so [geneticist] suggested you go have the DNA testing?

Yeah. -Steven, affected with HD

Unlike David and Tony, Steven did not grow up in close proximity to an affected relative. He was aware, however, of the family history of HD since a distant relative was affected. It is this knowledge which motivates Steven to see a geneticist when he notices changes in his body (e.g., movement).

Summary: Test triggers

In these narratives, participants recalled a specific incident that triggered their thinking about their risk for HD and the availability of the genetic test. All three had

known about their family history of HD for some time; two had lived in close proximity to an affected relative.

There was only minimal evidence in these narratives of re-evaluating the test decision; as Tony said, there came a point where he 'had to know.' Note, however, that when other members of his family were originally tested, Tony did not proceed with testing at that time. In this sense, then, he did re-evaluate that decision when something triggered his thought about his risk.

It could be argued that these stories also contain elements of *no decision to be made*, given that something (usually non-normal behavior) triggers the test agency of participants. With that trigger, there was little evidence of sustained, critical thought about the genetic test (unlike the stories of *re-evaluating the decision*).

Discussion

This chapter highlighted the myriad factors that can influence genetic-test 'decisions.' Importantly, it also attempted to describe *how* HD testing was actually taken up by participants. For some at-risk individuals, there was *no decision to be made*. Victoria, Patsy, Sherri and Brenda all described their decision in this way. Two requested, while two declined, the genetic test for HD. All, however, confirmed they simply *had* or *did not want* to know whether they carried the altered HD gene. Their narratives are revealing as they challenge much of the dominant research on genetic decision-making.

Dennis's and Jerry's narratives, on the other hand, showed evidence of *constrained decision making*. In these stories, the genetic test was generally not perceived as an opportunity for choice, at least not *for self*. Rather, test results were seen as a way of alleviating children's worry about their own risk or as providing beneficial information for children to be used in their own decision-making. These narratives, in particular, constructed the genetic test as a moral phenomenon: Perceived responsibility to offspring often acted to constrain participants' test agency. It is acknowledged, however, that no participant seemed in any way disturbed or distressed by his/her 'decision,' even those who had tested positive. That is, *participants* didn't necessarily perceive their decision as constrained. This could be partly due to the belief that the test can provide their offspring with *beneficial* knowledge. And as noted, all participants suggested they had never been pressured in any way, by anyone, to take the genetic test.

The (sometimes) dynamic nature of genetic decision-making was most clearly revealed in narratives of *re-evaluating the decision*. In these stories, the test was normally perceived as an opportunity for choice, both for self and offspring. These narratives unveiled critical thought about the genetic test and illustrated shifts in participants' thinking over time. Some originally wanted the test, but moved towards rejecting it, while others initially rejected the test, but moved towards requesting it.

Finally, David, Tony and Steven recalled how unusual behaviors in themselves motivated them to proceed with testing. Narratives of *test triggers* revealed how

knowledge of one's own risk became salient and relevant only when the narrator noticed some kind of change in him or herself.

It is reiterated that these processes of decision-making are not mutually exclusive, nor exhaustive. They are illustrative, however, of the many variables that can influence test decisions and in their depiction of how testing is actually taken up by at-risk individuals. The latter issue has been under-represented in the literature.

It should also be noted that all participants except one indicated they were comfortable with their test decision, whether they had accepted or declined. That is, no one expressed regret over having taken the test, regardless of test outcome. Nor did any at-risk participants regret declining the test. Laura, however, expressed regret that her affected relative was tested:

I wish they hadn't gotten tested. I find in that [number] years, that would have given them [number] more years and they would still be ok today if they had not gotten tested. They know they have it and that [number] years put a toll on their life. [number] years already. If they hadn't been tested, they would still be alright, but they would not have known for sure. –Laura, caregiver

Laura suggests that the certain knowledge provided by the test has negatively affected her relative. Knowing they carry the altered HD gene has 'put a toll' on their life, despite being asymptomatic for some time after the test.

What distinguishes tested and non-tested persons?

Pathways to the genetic-test decision represented in the preceding four themes described *how* test decisions can sometimes be taken. I was also interested in motives underlying genetic testing. Evers-Kiebooms et al. (2000) have rightly highlighted the

difficulty in trying to “unravel” why only a minority of at-risk person choose to be tested, while the majority reject testing.

It is my contention that this difficulty is a notable finding in and of itself. It suggests that the phenomenological experience of genetic decision-making is a complex process, sometimes lasting for years. Conversely, for others, there is no decision to be made, and there is minimal conscious reflection about the test as an opportunity for choice. It also warns against any simplistic discussion about motives for testing during genetic counseling sessions and highlights how some concerns of test candidates are not discussed at all (e.g., rational suicide/quality of death).

Justifying decisions

Before reviewing the variables that seemed to influence genetic-test decisions in the current research, a word on justifying decisions is in order. The narratives of decision-making of at risk participants seemed, in some ways, richer and more detailed than those of tested participants. This could reflect a need to *justify* their decision to decline the test. In all cases, I was the first person from the ‘public’ genetics arena to whom participants told the story of their genetic risk and test decision. Given the public discourses which depict the ‘power and promise’ of the new genetics (Smith, Michie, Allanson, & Elwy, 2000), participants could have felt the need to justify their non-participation in testing. Tested participants, on the other hand, need not have felt the same pressure to justify their behavior since it could be perceived as congruent with current cultural norms about responsibility for health and management of one’s personal risk. Despite this, the stories

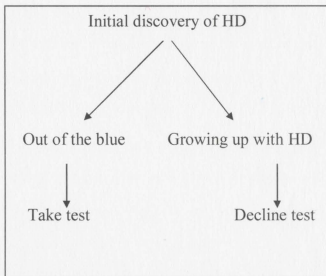
of at risk participants were revealing as they highlighted the multiplicity of factors that were considered in taking the decision to decline the genetic test for HD.

Initial discovery and testing

Virtually no literature exists on the relationship between initial discovery of HD and genetic-test decisions. Cox (1999) interviewed predictive HD test candidates and found those who 'had to know' generally had little direct contact with family members affected with HD and had known of the family history for only a short time. In contrast, predictive test candidates who had grown up with HD generally expressed some degree of ambivalence about the test; there was usually a period of weighing the implications for self and others.

The relationship between initial discovery of HD and uptake of genetic testing in the current research is depicted in Figure 1.

Figure 1. *Discovering HD and taking the genetic test*



In the current research, the narratives of the three tested participants in *no decision to be made*, would seem to support Cox (1999). For one participant, initial discovery of HD did come *out of the blue*; and, while there was a period of '*something*' being wrong with a family member for the other two participants, their undocumented family history did lead to awareness of the family history out of the blue. In all three narratives, participants had minimal direct experience with an affected family member at the time of their test, and all three were tested fairly quickly following initial discovery as depicted in Figure 1.

In contrast, narratives of *growing up with HD* did reveal a degree of ambivalence about the test decision and/or receiving results. In general, those who had grown up with HD were more likely to decline the genetic test (see Figure 1).

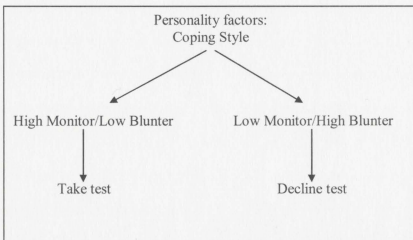
Other motives for testing

Decruyenaere et al. (1996) suggested that personality profile and individual coping style seem to be the chief variables in the decision to be tested. The current research provides some support for these suggestions. For example, recall both Victoria and Patsy who suggested they just *had to know*, suggesting it was the 'sort of person' she was. Similarly, non-tested participants for whom there was *no decision to be made* spoke about declining the test as if it were a logical reflection of their personality. And, others who were not tested suggested they were worried about how they would cope with a positive test result, feared excessive symptom-watching, and/or anticipated survivor guilt and worry for their offspring.

Personality differences between those who *had* to know and those who simply did *not* want to know could reflect a different style of coping with health risk information more generally (Miller, 1996). “Monitors” attend to and are more likely to process threatening information, while “blunters” avoid health threat information and cues. Blunters are less likely to manage any malady inside the medical gaze (e.g., see a physician). Importantly, Miller has demonstrated that people fare better when health information is tailored to their personal coping style – monitors will generally do better when given more information, while blunters generally fare better with minimal information. I did not formally measure coping style; however, participants’ discourse about coping with genetic risk for HD was reminiscent of Miller’s (1996) monitoring/blunting distinction.

Figure 2 depicts the relationship between individual coping style and genetic test decisions in the current research.

Figure 2. *Coping style and taking the genetic test for HD*



As depicted in Figure 2, monitors were more likely to take the genetic test for HD, while blunters were more likely to decline. The distinction between monitoring and blunting coping styles is an area worthy of future research in the genetics context. This difference may have implications for the type and amount of information presented during counseling sessions. Importantly, however, the difference may have implications for those who make *constrained decisions*. Consider, for example, people with a blunting coping style who are not normally inclined to process health risk information. Assume further they take the genetic test for their children. How will they cope with knowledge of their own genetic risk? And, how well will they cope with the large amount of information presented during counseling sessions?

I am aware of only one empirical study that investigated the contribution of a monitoring coping style on genetic-testing interest (Shiloh, Ben-Sinai, & Keinan, 1999). Shiloh et al. (1999) randomly assigned participants to one of four hypothetical testing situations differing by the degree of control and certainty each provided. As expected, tests for conditions that offered control over the illness were preferred to tests that did not by monitors and blunters alike. Additionally, both groups were least likely to prefer tests that provided no certainty or control. Differences emerged, however, for tests which offered high certainty and low control: For these tests, monitors showed significantly more interest in testing than blunters. It is notable that this empirical scenario reflects the current context of genetic testing for HD. The test is 100% penetrant (i.e., certain), but

offers no control over the illness. Shiloh et al. (1999) argued that under conditions of health threat that offer no possibility of control, monitors still strive for uncertainty reduction, a form of emotion-focused coping.

Nonparticipation in genetic testing

The monitoring/blunting distinction also stresses the importance of the right *not* to know, an important ethical principle. Virtually every participant in the current research staunchly defended the right not to know one's genetic risk, and all stressed the highly personal nature of the test decision. In the literature, however, at-risk persons are often characterized as pessimistic (van der Steenstraten et al., 1994) or as possessing negative coping appraisals (Wolff & Walter, 1992) about living with a positive result. More broadly, they are regarded as 'passive' in their response to genetic risk (Hunnicke, 2003). Binedell and Soldan (1997) have warned against this negative portrayal of non-tested persons in much of the testing literature. Their concern is that the negative characterization of at-risk persons could influence healthcare professionals in the genetics context and bias them against the perception of nonparticipation as a valid decision. Binedell and Soldan (1997) suggest, however, that non-tested persons may in fact be insightful, rather than pessimistic.

Despite the focus on risk avoidance, management, and control, risk society (Beck, 1992) is one in which risk *intolerance* may not be the best course of action for those living with genetic illness. Forde (1998) reminds us that uncertainty, unpredictability and risk will always be an inherent part of human life. Those at risk for HD are perfect

exemplars of such lives: They do not know whether they will eventually manifest the illness or, if they do, which symptoms will strike them, when, and with what severity. Forde noted that risk aversion and intolerance might not be the best basis for coping and self-realization in the uncertain environment of life. Non-tested participants in the current research were well aware of the uncertainty and risk with which they live. Some seemed almost unconcerned about their genetic risk, but this could well reflect a tolerance for, rather than a denial of (or intolerance for) risk.

Considering age

It must be reiterated that age seemed an important influence on genetic-test decisions in the current research, especially for non-tested participants. Five of the six at risk participants specifically cited their age as a motive underlying their test decision. For example, three participants were in their twenties and thirties, and all claimed they were too young to begin worrying about a disease that may (or may not) manifest until much later in their life. And, recall Brenda, who suggested that since typical onset was early in her family, she feels she is past the age at which she needs to worry about HD.

Age has been under-examined as a mediator of test decisions. Psychometric risk research sometimes underestimates the impact of socio-demographic variables which can influence how people identify and respond to risk (Lupton 1999a, b). Narratives of at risk participants, however, clearly highlighted the importance of age in thinking about and responding to genetic risk for HD. Tested participants, as well, cited age as an important factor in their decision and in coping with risk.

The benefits of knowledge

Striking differences emerged in the narratives of tested and non-tested participants regarding perceptions of the *beneficial* knowledge provided by genetic testing. Almost all participants who had been tested (and those who claimed they would be tested at some point in the future) were unanimous in their endorsement of knowledge – knowledge about the test, HD itself, and the beneficial knowledge the test can provide. Recall Jerry's suggestion that the knowledge provided by testing gives people 'some options.' Similarly, Jackie commented, "I firmly believe that the more you know, the better choices you can make. And, you should have as much knowledge as possible." Lori said, "I've been educating myself. I figure the more I learn, the better it is for everybody." This discourse is reflective of the public health discourses that construct the new genetics as allowing prediction and control of future health (Petersen & Bunton, 2002). For many tested participants, then, genetic testing can be seen as a way of 'colonizing the future' (Giddens, 1991).

Non-tested participants, on the other hand, (excepting Cheryl who plans to be tested in the future) were far less likely to speak about the benefits of knowing genetic risk information or genetic testing *per se*. Roxanne commented, "There's no one who can even tell you once you does the testing - they can tell you if you got HD, but there's no one going to tell you it's going to come on when, or how bad it's going to be." Non-tested participants recognized that many questions still remain following a positive test result (although, several tested participants noted this as well). Thus, at-risk individuals

sometimes resist the 'power and the promise' of genetic testing (Smith et al., 2000). As such, the predictive aspect of genetic testing is rarely interpreted as enhancing one's power or providing a sense of control for some at-risk person (Duster & Beeson, 1997).

In general, at risk participants in the current research seemed less interested in predicting and controlling their futures than tested individuals. In part, this was due to pre-existing views about future health, frequently couched in the language of luck or fate (e.g., 'if I get it, I get it,' 'whatever will happen, will happen'). For these participants, if the future is perceived as uncontrollable or unmanageable, genetic testing is not seen as a useful form of risk management. Giddens (1991) has argued that entrusting one's life to fate, ... "relieves the individual of the burden of engagement with an existential situation which might otherwise be chronically disturbing" (p. 133).

The 'right' thing to do – test as moral phenomenon

Notable differences also emerged in the narratives of tested and at-risk individuals regarding the moral duty to be tested. Tested participants (and those who plan to be tested in the future) were far more likely to talk about the test as the 'right' thing to do, and as noted, this was especially evident for *constrained decisions*. It is unclear as to why some at-risk individuals perceive a moral duty to be tested and others do not. The explanation is not as simple as having children. Recall both Roxanne and Brenda - each has children, each is at risk. In neither narrative was there any evidence to suggest the decision to test was perceived as a moral duty. Tested participants, especially those with children, were far more likely to speak of the test as something they 'should' do.

A possible interpretation for this difference is a belief in the meaning of the test as an instrument for planning the future. In the current research, non-tested participants were far less likely to speak of the genetic test as an instrument that would allow them to assert some control over their futures or the futures of their offspring. These participants were also more likely to note the therapeutic gap that exists between having the test and treating the illness. They were usually aware that even if they tested positive, no one could tell them when the disease would affect them, which symptoms they were most likely to manifest or how severe those symptoms would be. I am not suggesting that participants who do not perceive a moral duty to be tested are in any way wrong, nor do I wish to disparage them in any way. Hunniche (2001) has cogently argued that with the rapid and changing proliferation of genetic knowledge and technology, how can we speak of well-informed or responsible choices in any universal sense? As she put it, "How can we be well informed, or pass on solid information if the information and associated technologies are constantly revised? And how then, can decisions be uniformly ethical?" (p. 44). I wish to highlight, however, that the responsibility ensued by genetic testing can be seen to reduce some at-risk individuals' choices (e.g., see *constrained decisions*).

Concluding comments

Narratives of genetic decision-making in the current research revealed the complexity of such decisions. Myriad factors interacted to influence the test decision, including disease-related events in the family, age, responsibility to others and the nature of both the test, and of HD, itself. Additionally, there was no single pathway to the test

decision. These findings have both theoretical and clinical implications. Theoretically, it may be inadequate to define health decision-making as a cognitive, rational, static process. Obligations to others and anticipated emotions also influence health decisions. Yet, social cognition models such as the Health Belief Model do not explicitly account for such variables.

For some at-risk individuals, it may be better to characterize decision-making as on a continuum, rather than a single, static choice. Narratives of *re-evaluating the decision* clearly revealed the dynamic nature of decision-making. As in life more generally, we often change our minds, come to a different decision or do differently than intended in accordance with the complexity that is life (Hunich, 2001).

Participants' stories in the current study also challenge the conventional construction of a decision as an opportunity for choice. Their narratives raise questions about what it is that allows some at-risk individuals to see that there is a decision to be made. Conversely, what is it about some participants' experiences that seems to point - without hesitation - to one, and only one, course of action? These life experiences deserve attention during counseling sessions if we want to uphold the gold standards of autonomy and informed consent in genetic decision making.

Some narratives in the current research also revealed issues that were not discussed during counseling sessions (e.g., quality of death or rational suicide). Davis (1999) noted that as a general rule, when suicide is mentioned as a response to HD or Alzheimer's disease (AD) in the medical literature, it is negatively characterized (e.g., an

adverse reaction or a risk, rather than an option). Davis contends, however, that persons at risk for genetic diseases such as HD have unique reasons for the serious consideration of suicide. In the current research, at least three tested participants spoke about the importance of quality of death, one of whom had tested positive.

The topic of quality of death was not included on the interview guide; rather, participants spontaneously discussed their concerns in this area, suggesting it is important for some at risk and tested individuals. I concur with Davis (1999) who noted, "Although probably only a minority of those facing HD or AD will choose this option, that minority does exist, and it is unhelpful and less than frank to ignore it" (p. 317). Reports of genetic counseling in practice confirm that counselors are frequently unaware of the issues their clients want to discuss; as such, the issues are not addressed (Smith et al., 2000). Yet, these issues are important. In the words of a participant who had tested positive:

You want quality of life, but I think people should have quality of death too. (...) I think for myself, I don't want to suffer like my [parent] did. I'd like to have a choice whether I want to go through with it till the end or if I want not to. I think that's the thing I'd like to see dealt with.

CHAPTER 9

**RISK AS LIVED REALITY: BROADENING THE MEANING
OF GENETIC RISK**

The current research explored meanings: What does it mean to live at risk for a fatal genetic illness? What does it mean to test positive and carry the knowledge that future illness is inevitable? Conversely, does testing negative mean 'freedom' from a fatal genetic heritage? How do people talk about genetic risk, and what does it mean to them in the context of their daily lives?

The previous chapter highlighted the experience of genetic testing for HD, including a discussion of why and how such 'decisions' were taken. In that analysis, I argued that not all test decisions were perceived as 'choices,' especially for constrained decision-makers. Rather, perceived responsibility to others acted to constrain test agency. Responsibility to others reappears in the discussion to follow.

This chapter, and the next, present the stories told by participants about living with genetic risk and/or illness in the context of their own lived reality. The current chapter highlights the meanings that converged around living 'at risk' for HD and discusses when genetic risk is or is not salient. The next chapter continues the discussion of living at risk by exploring perceptions of stigma associated with HD and by investigating how participants cope with genetic risk information. Taken together, these chapters paint a picture of living with genetic risk for HD.

Numerous factors combined and interacted to influence what people thought about risk, how they responded to it and when (or if) it was an issue in their lives. Common in all participant narratives, however, was a discussion of risk and illness as a *lived reality*, not a numerical probability. Participants' stories in the current research suggested that genetic risk for HD was not perceived as an objective probability. Rather, it was actively processed, discussed, reflected and acted upon by at-risk individuals and their families, albeit with much variability.

Several themes clustered around the meanings and saliency of genetic risk for HD. These are broadly subsumed under: (1) Risk as emotion; (2) Risk as responsibility; and (3) Chronic Risk, including risk as biographical disruption and risk saliency.

Risk as emotion

Genetic risk as unique threat

For many participants, the meaning of risk lay in its propensity to evoke emotions, particularly negative emotions. Genetic risk was simply not interpreted as a neutral probability, much as it is constructed so by Mendelian genetics. Rather, its meaning derived from its representation as an *index of threat*, to self and other family members. Narratives in the current study were replete with discourse reflective of the worry, confusion, anxiety and fear invoked by genetic risk for HD. In this way, risk was very much a lived dimension of reality. Julie, for example, spoke about the meaning of being at risk in this way:

I think that people keep saying 50:50. But it's not - it's one out of two, you know what I mean? When people keep talking about 50% risk and 25% risk and

whatever. When you say to someone 50:50, they think you have got 50 good chances out of 100 and 50 bad chances, but it is not. You have got one good chance and one bad chance. And that's what changes things. -Julie, at risk

Note that genetic risk was not maintained as a mathematical figure; rather, it was transformed into a qualitative statement with a binary outcome (cf. Parsons & Atkinson, 1992). The idea of 'one good chance and one bad chance' also gives meaning to Julie's children's risk, and it is fear that she could have passed on the altered HD gene that permeates her talk about living at risk:

It's the fact that you have passed it on that's the issue. That would be hard for me. And, they talk about [names children] would be 25% risk because I don't know my results yet. I am 50%, they're 25% - well, the chances of them being positive are just the same as the chances of me being positive because if I have it, they either got the good one or the bad one. So, the numbers don't mean anything. And that's why when you read these studies and you see 25%, you think, 'Oh gee, that's not a big deal, 25% at risk,' but when you look at the reality of it, it is a big deal. -Julie, at risk

For Julie, then, the 'reality' of living at risk means far more than her numerical risk can convey. Similarly, Victoria suggested that risk means worry, regardless of whether one is tested or not:

Well, if your [parent] has it, you think you have a very good chance of getting it - it's 50:50. To me, once you know it's in the family, there's no peace of mind anyway. I took the chance of having peace of mind, and what a good feeling for you and your kids if you didn't carry the gene. For me, it didn't work out. For my [siblings] I think in some ways they're still tortured because they feel guilty about not having it. But I think that when you're at risk, you worry anyway, whether you get tested or not. -Victoria, tested positive

Victoria's narrative was illustrative of the emotional meanings converging around genetic risk. For her, living at risk means 'no peace of mind,' 'worry,' and 'tortured' guilty siblings: Genetic risk is a threat to her and her family.

Kathleen's understanding of her genetic risk was also given meaning in her concern for her children. Her risk is a threat to them, and it is a threat over which she has no control. Despite this, she expressed guilt at the possibility of passing on the altered HD gene:

I do worry about my children a lot. I think that's the thing I'm most obsessive about. Them having the gene. I can take it for myself, but for my children who I love more than anything, who I would give my life for, I have no control over them, and that's the most painful aspect of this for me.

I: That's an important issue you are raising. For people who have children, that is something to be considered, isn't it?

Oh yes. I think you have a lot of guilt too. I know it isn't my fault, it's beyond my control, but sometimes I say to myself, 'Oh my God, what have I done to my children?' I think that's a natural process, because my [parent] feels the same way about us. —Kathleen, tested, intermediate gene

In addition to the worry about her own uncertain future, Kathleen carries the emotional burden of worry and guilt about her children's risk.

Rejecting the threat of genetic risk

While some participants noted that being labeled 'at risk' did invoke feelings of dread and anxiety, they rejected this label. Roxanne commented:

At risk almost makes me think it's a deadly disease, something that's going to kill you. I think at risk makes it scarier sounding than what it really is. It is a bad thing to have, you lose your memory, you forget the best part of your life, and who everyone around you is. But I don't really think that throughout your life it really affects the quality of life that you have. Putting that term at risk on it makes it sound a lot scarier to me. —Roxanne, at risk

Roxanne's understanding of her genetic risk highlights the importance of personal experience with HD in the meaning accorded to risk. Her affected relative did not

manifest symptoms of HD until later in life (sixties). Roxanne is able to distance herself from her scary 'at risk' status since she is young and HD is not an immediate threat.

Similarly, Michelle noted her difficulty in assigning meaning to living 'at risk' at this point in her life course:

I: Does anything come to mind when you hear the term 'at risk' for HD?

Not really, to be honest with you. Now, maybe if I was older. But, at my age, I am only [age], and to me that is unnecessary worry. I may not even have it, so why am I worrying? – Michelle, at risk

Michelle is in her thirties, and her youth allows her to distance herself from the state of being at risk. And, like Roxanne, the onset of HD was late in her affected relative.

While many participants constructed genetic risk as a unique index of threat to their families, negative case analysis revealed some participants who viewed genetic risk as just one more risk in the myriad risks that exist in life. Notably, this construction of risk was most common in the narratives of participants who had tested negative and no longer had to worry about themselves or their children developing HD. One said, "I think that you are at risk for everything in life (...). So, in a weird kind of a way, it was kind of one thing that I didn't...I could check it off my list of things I worry about." Gerald also questioned the uniqueness of being 'at risk' for a genetic illness:

As far as being classified 'at risk,' it's kind of a strange term. We're all at risk for heart attacks, but no one says that. This society in Canada alone, Newfoundland alone, heart attacks are almost mandatory. But no one says Newfoundlanders are 'at risk' in a derogatory term. It's just that the stats show we are this way.
-Gerald, tested negative

For participants who had tested negative, the threat of HD was eliminated for themselves and their children, and concomitantly, worry and fear about it. As such, their construction of being at risk for HD was less emotionally charged than some at risk participants and those who had tested positive. This is not to suggest, however, that participants who had tested negative were not worried for other affected and at risk family members (e.g., siblings).

Beyond threat: Does genetic risk invoke anger?

Readers may wonder if at risk participants expressed anger toward their parent for having passed on the altered HD gene or anger towards being at risk more generally. No participant talked about risk in this way, nor was any anger expressed by participants toward their affected parent. Early research reported similar results that, at the time, were unexpected and surprising (e.g., Wexler, 1979). Since Wexler's seminal work, however, other studies have found little or no expressed anger in at risk participants toward their affected parent (e.g., Chapman, 2002). It has been suggested that it is in poor taste to be angry at a person who is ill and dying (Cox, 1999). In the current research, participants explained that their parents simply did not know about the family history of HD and, as such, could not be blamed. As Kathleen told her parent:

You didn't know. You were an innocent victim. No one blames you. I don't blame you. One of these days, I'm going to get sick, but I don't blame you.
-Kathleen, tested, intermediate gene

The current sample could have limited the possibility that participants would express anger or resentment towards affected parents. An average of six years had passed

since participants had their genetic test. It is possible that sufficient time had passed for most participants in the current research to work through any anger or resentment they could have felt toward their parent or toward being at risk more generally. Only one participant had recently discovered the family history of HD and had undergone testing. At the time of our interview, she had not yet received the test result; perhaps insufficient time had passed to work through thoughts and feelings at all. Finally, the interview guide did not expressly ask participants whether they were angry with their parent(s) or angry about being at risk for HD more generally.

Summary: Risk as emotion

For many participants, the meaning of genetic risk did not reside in an objective probability, despite that fact that a majority of participants knew their objective risk was 50:50. Rather, genetic risk was re-contextualized as a *threat*, both to self and other family members; it was especially pronounced as a threat to one's children.

The construction of risk as a negatively-charged concept, annotated with images of dread and anxiety, accords with some recent writings on risk (e.g., Beck, 1992; Lupton, 1999a, b). In the current research, the threatening meaning of risk could be influenced, in part, by participant views of HD itself. Almost without exception, participants saw HD as an uncertain, unpredictable 'horrible' illness, a 'never-ending' illness, a 'disease of loss.' To speak of being at risk for this illness (or testing positive for the altered HD gene) necessarily invoked memories of affected relatives. Generally speaking, these memories were vivid and disturbing, or as David said, "pretty traumatic."

Participants' stories clearly demonstrated the strong emotional meanings associated with genetic risk – fear, anxiety, dread and guilt; anger was not expressed by any participant. Testing negative appeared to mitigate these negative emotions (at least partly); although, survivor guilt and/or worry for other affected family members remained.

These findings highlight the emotional pre-test state in which some test candidates can find themselves and raise questions about the ability to absorb and integrate complex information imparted during counseling sessions. According to Janis and Mann's (1977) decisional conflict theory, stress interferes with the ability to consider the salient features of a situation and to deliberate carefully about the pros and cons of alternative options. Lerman, Lustbader, et al. (1995), for example, found an inverse relationship between risk comprehension (for breast cancer) and levels of distress: They suggested that distress interfered with information processing.

Risk as responsibility

Socio-cultural writings on risk (e.g., Beck, 1992; Lupton, 1999a,b), argue for a consideration of the social context in which risk is experienced and lived. Genetic testing has been introduced into 'risk society' – an era of surveillance medicine (Armstrong, 1995) and personal responsibility for health. It is perhaps unsurprising, therefore, that many participants spoke about their 'genetic responsibility' (Novas & Rose, 2000).

The language of genetic risk increasingly provides a grid of perception which informs decisions on how to conduct one's life, have children, get married or pursue a career. With the emergence of the genetically at risk person, genes themselves have been constituted as what Foucault (1982) might term an 'ethical substance' that one works upon in relation to the self (genetic identity,

reproduction, health) and in relation to others (siblings, kin, marriage, children) (Novas & Rose, 2000, p. 502).

Responsibility to future generations

Many participants spoke about their obligations to others, especially children, and this was especially pronounced for tested participants. Thus, genetic forms of thought have become entwined with the ethical dilemma of how to live one's life, what goals to set and how to plan for the future in relation to genetic risk. These ethical dilemmas are all occurring in risk society, (supposedly) populated by responsible, well-informed citizens, expected to manage and control their genetic risk.

It is not only test candidates themselves, however, who perceive a personal responsibility to ascertain their genetic risk; some social others (even other at-risk individuals) also expect those at risk for HD to determine their genetic risk and act accordingly. A participant who tested negative, for example, said:

My [relatives], they continue to have children. To me, who in their right mind would do this if you knew you were going to inflict such terrible things on your children? If you don't know, I suppose that's different. If you do know it's in the family, I just can't see it.

For this participant, then, choosing to have children despite the family history of HD was incomprehensible. Early in this interview, the right of individuals *not* to know their genetic history was defended. However, this passage would seem to suggest a belief in genetic responsibility. This contradiction could be partly explained by personal experience with HD. The participant had experience caring for an affected relative, and those memories were vivid and painful. As such, s/he 'just can't see' inflicting such

'terrible things' on one's children. Another participant who had personal experience with HD similarly commented, "If I had known about this disease before I had [children], as much as I love my children, I would never have had them. I wouldn't want to inflict this on my children."

These examples highlight the responsibility felt by at-risk individuals to future generations. However, genetic responsibility revolved not only around reproduction, but also, marriage, career and future care-giving obligations - for self and for other affected family members. In this chapter, I wish to highlight these other aspects of the general theme, *risk as responsibility*.

Responsibility to partners

Participants with current partners expressed their unease with their partners having to provide extended care for them if (or when) they develop HD. Responsibility to partner was especially evident in the narratives of tested participants: Their partners should not be responsible for providing the comprehensive, long-term care often needed by a person affected with HD. Kathleen, for example, said:

I made the decision years ago, a couple of years ago, that when I get sick, I'm not going to make my [partner] have the total responsibility of my health. I don't want him to. I know it's for better or worse, but I don't think he should have to give up his life for me. (...) So, when I start to get sick and can no longer look after myself, I want to go in a home. -Kathleen, tested, intermediate gene

Similarly, Lori also spoke about giving her partner the option of 'getting out' before, or if, she developed HD:

Because I had already gone through a lot of health problems...And I said, you know, he's just had enough. [Parent] just died, he helped me with them. And I

said to him when I found out, I said, 'You know, if you want to go, I totally understand.' I said, 'I totally get it. You've done more than enough.' –Lori, tested, waiting for results

Neither Kathleen's nor Lori's partner indicated he would leave and both have suggested they will provide future care for their partner should that be necessary. However, these passages are illustrative of the perceived *personal* responsibility for one's care and a desire to avoid burdening one's partner in the future.

There was also a recognition of responsibility to *future*, hypothetical partners in the narratives of some participants. For example, while Stacey has tested negative, she admits that if her children were still at risk, future in-laws might hold her children accountable for knowing their genetic risk. "I would think that if my [children] met somebody and they were going to get married and it was still up in the air about us, I would think that their family would certainly want them to go and get tested and at least be accountable for that. I know I would."

Cheryl also expressed concern for her future partner:

I don't think it's fair knowing that you might have something like...your husband will be completely – well, if they love you, they have to take care of you right up until you die. And that's 15 years, or about that. That's a long time to watch someone you love die. I think anyone that's going to spend the rest of their life with you has the right to know. –Cheryl, at risk

For participants with partners, and even for participants without, there was a perceived responsibility to inform them of the potential future of HD. As Serena put it, "So that leaves no one getting into this and not knowing what's coming." Similar results

were reported by Chapman (2002) and Taylor (2004) in their interviews with persons at risk, or testing positive, for HD.

When the discovery of HD comes out of the blue, *unaffected* partners of at-risk person are faced with the knowledge of impending disease in their partner, and possibly their children. These narratives too, reveal a sense of responsibility to the at risk partner and their children, although the uncertain future is terrifying:

That's very hard. Thinking about that. I suppose for me, I'm not sure what's going to come. I don't know if I'm going to be looking out to [partner]. I'm afraid that my children may come down with something and I'm going to be the one who's going to be looking out to everybody. That's a very scary thing for me.
—Daphne, partner tested positive

Partners of those at risk or having tested positive for HD can perceive the future as uncertain and frightening. Changing roles in the relationship (e.g., from spouse to potential caregiver) can lead to marital distress (Decruyenaere, Evers-Kiebooms, et al., 2004), both before and after HD symptomatology begins. Daphne, for example, noted how she sometimes watches her asymptomatic partner and children for signs of HD.

Decruyenaere et al. (2004) also reported on marriage breakdowns following both positive and negative genetic test results for HD. In the current research, at least two participants noted marriage breakdowns, both of whom tested positive (in one case, the participant's family member). Both also suspected the partners left because they did not want 'to deal with HD.' Nowhere in Daphne's narrative did she suggest she ever thought of leaving her relationship. "It didn't even cross my mind. No. Actually, even when I found out [partner] had it, I said, if I could have taken it and put it in my own self, I

would have.” Nonetheless, perceived responsibility for her partner’s potential long-term care is scary and confusing.

It should also be noted that while I have focused on responsibility to partners, there was also evidence in participant narratives of responsibility to other family members, especially affected members. Stacey, for example, commented on the perpetuation of HD in families and recognized that many of her relatives remain at risk – as she put it, “we are only just beginning.” She perceived a responsibility to help those who were not lucky enough to escape the family legacy. “The time is going to come when I am going to have to go down and deal with this and help them, and they are going to need a major load of help in the next few years coming up.” –Stacey, tested negative

Beyond responsibility to, and felt by, unaffected partners, perceived responsibility for future care was most poignantly expressed by caregivers, especially those caring for their children. Caregivers feared their increasing age and ability to continue to provide care for their affected family member(s). Every caregiver in the current research expressed responsibility for the care of a family member, in two cases, an adult child. Their narratives were often emotional and illustrative of the pain and fear invoked by acknowledging they will not be able to provide care indefinitely.

Laura, for example, does not know what the future will bring, but she was adamant that her affected family members would always be with her:

You know, I don’t know where I’m going. I try not to think about where I’m going. I just wait for tomorrow to come. I only know that right now, they’ll always be with me. For as long as I can do it. And when I can’t do it, I’ll have somebody here to do it. But they’ll still be with me. At home.

I: Rather than put them in a home, you mean?

I will not. Unless it gets to the point where they don't know me. And that might never happen. But as long as they know me, know who I am, they'll always be with me. I know they are cared for when they're with me. -Laura, caregiver

For Laura, then, caring for her affected family members at home is important, and her entire narrative was reflective of her obligation to them. She prefers not to think about the future as the prospect of her not being able to care for her family members is painful. Similarly, Marjorie suggested that she feels *she* is the one who needs to care for her child, despite the assurances of her other, unaffected children:

That's the worst I find – they're young, and I'm old.

I: So, your concern is for later on down the road?

Yes. Now, the [children] tell me not to be concerned. No matter what happens, they'll be taken care of. I know that. But, of course, I think *I* need to do it.
-Marjorie, caregiver, emphasis in original

For parents of affected children, there was a strong sense of responsibility to be the caregiver. This responsibility can be painful as caregivers face increasing age and sometimes poor health of their own.

At risk children can also feel responsible for providing future care when their parent becomes affected with HD. The idea of caring for a parent like a child is distressing and frightening. One at risk participant said:

They're my [parent], and no matter what happens, I should be there to take care of them. But, I just don't think I could change them, and bathe them and change their diaper, to give them the proper care they would need. I don't think I'd be able to do it. The whole bathing them and taking care of them like a baby, I can't picture ever seeing them like that. -Female, at risk

Responsibility to plan future care

Perceived responsibility to plan for the future was salient in the narratives of tested participants. Jackie said, "If you did have it, you want to manage the rest of your life - whether it's a living will or do not resuscitate order or anything else. I didn't want to leave that to somebody else to have to decide." –Jackie, tested negative

Similarly, Lori suggested:

The first thing we did was, the day after I found out, I made my will. You know, power of attorney and what I wanted if things turn bad. (...) So for me, the future is having my affairs in order, having my directive, you know, all that stuff done. Then I don't have to worry about it. –Lori, tested, waiting for results

For some tested participants, then, there was a perceived responsibility to have future affairs settled before the manifestation of HD impeded their ability to do so. David suggested that many decisions that might have been left to the future were made earlier in light of his having HD, particularly those about finances:

My wife is only going to [work] for 20 years and then she is going to take care of me, so we know she's not going to have enough insurance with her [work]. So now we have to plan to put money away for her retirement because she was going to need 30 years of retirement after that. So we have to plan for that. –David, affected with HD

For some participants, genetic risk creates a responsibility to act in the present in response to future events that have not yet happened (Novas & Rose, 2000).

Responsibility to communicate genetic risk

It has already been noted that many participants expressed a responsibility to communicate their risk for HD to current and future partners. Some participants also expressed a felt responsibility to inform other, extended family members of their

potential risk. One participant recalled how, following confirmation of the family history of HD, they provided a copy of the confirmation letter from the genetics clinic to the extended family:

So we sort of picked one person in each family and said that, here is a copy for everybody, and not so much you decide what to do with it, but we kind of left it at that. But, I sort of felt that each person needed to have their own copy of the letter, and I felt that that was our responsibility. Some of them hated us for that because it changes your life. –Female, at risk

This account is notable since it highlights how rifts can occur in families when some branches do not want to acknowledge or talk about the family history of HD.

Other participants spoke about their responsibility to inform children of their risk, replicating research with other HD study populations and women at risk for breast cancer (Forrest et al., 2003). Most participants with children in the current research endorsed their children's right to know about their genetic risk and had spoken with their children about the family history of HD. There were only two exceptions to this: One exception involved parents of young children. These participants suggested their children were still too young to handle such news. All confirmed, however, they would talk to their children as soon as they were old enough to understand the family history of HD. There was an overriding theme of responsibility to protect young children from such devastating news. At the same time, parents felt their children should know about the risk before critical life junctures (e.g., marriage or reproductive decisions).

In a second exception, a participant waited to inform an adult child of the family history of HD since the child was already coping with a painful life event. Concealing the

family history was stressful, and even though they knew the news would be devastating, they perceived a responsibility to inform the child of his/her own risk:

I was living with this and keeping it so close to my chest. I was so overwrought and overwhelmed (...). They had the right to know. I felt horrible having to tell them, but I knew I couldn't not tell them.

Note how this narrative highlights the discourse reflective of clinical genetics – the language of ‘rights.’ This meaning of genetic risk often converges around ethical questions of the right to know and just as important, the right not to know.

While it is beyond the scope of this study, questions of rights as they pertain to genetic information are significant and complex. Who owns genetic information, and who should have access to it? If a person's right *not* to know his or her genetic risk conflicts with a family member's right to know, how is this conflict resolved? In the case of HD, a ‘non-disclosing’ prenatal test (Wexler, 2001) is available that can tell with a high degree of certainty whether a foetus is not going to have HD. However, it cannot tell with certainty whether the child carries the altered HD gene. In this way, parents who do not want to know their own risk status are protected from disclosure. Nancy Wexler (2001), herself at risk for HD, cautioned that each individuals' position must be considered, and her discussion of the availability of the non-disclosing test for HD in Venezuela revealed the complex ethical choices that at-risk person and their families must negotiate.

Choices such as using prenatal testing highlight the distinct ethical dilemmas surrounding genetic testing, and a growing literature is addressing these issues (e.g., see

Everett, 2003; Gaivoronskaia & Solem, 2004; Juengst, 1999; and the Nuffield Council on Bioethics, 1993, for reviews).

Summary: Risk as responsibility

In the previous chapter, I highlighted how some at-risk person were constrained in their test agency by perceived responsibility to others, especially offspring. I also noted how test candidates were more likely than at-risk person to experience the test as a moral phenomenon – something they felt they ‘should’ do. In this chapter, I aimed to expand upon the theme of risk as responsibility by presenting some of the myriad responsibilities expressed by study participants.

Responsibility to current and future partners emerged as an important lived dimension of genetic risk. For younger at risk participants who had not yet married, there was a belief that future partners have the ‘right to know’ what ‘they’re getting into.’ Participants with current partners, on the other hand, worried about their own future care and felt their partners should not have to ‘give up’ their own life to care for them, if or when they become sick. Some participants, notably those who had been tested, felt a responsibility to plan for their futures, including financial considerations, retirement, future healthcare needs and medical directives. This was most likely for those participants who perceived the genetic test for HD as an instrument for planning the future. Perhaps unsurprisingly, then, the majority of at risk participants did *not* express responsibility to plan for a future that might include HD.

Finally, there was also a perceived responsibility to communicate one's genetic risk to others, notably children and, in some cases, extended family members. In the latter case, tensions can arise when some members of the family do not want to acknowledge or discuss the family history of HD. In general, communication of genetic risk in the current research presented a dilemma of *when* and *how* to tell, rather than *whom* to tell, and this was especially pronounced for participants with children.

When risk is experienced as a responsibility, it can create tension and anxiety for at-risk individuals. 'Responsible genetic subjects' (Novas & Rose, 2000) are those who take an active role in the management of their genetic risk – they gain as much knowledge about HD as possible and apply it to self or to a person for whom one cares. They govern their lives with a consideration of others, often future generations. Thus, when the meaning of genetic risk converges around responsibility, it is infused with ethical considerations that must be negotiated by at-risk person, and more broadly, at risk families (Juengst, 1999). Should they have children? Should they get married? When do they tell future partners about their family history? When should they tell siblings and children they are having the genetic test?

In this context, where genetic information is thought of as containing the potential to transform one's life, the disclosure of genetic risk information gets framed in terms of the language of rights – the right to know – a right of one's kin, a right of one's children – the withholding of this knowledge is seen as an incursion upon the right to choose. Yet, the right to know comes into tension with another right, the right not to know, the right not to be known, the fear of the consequences that that knowledge may bring for one's conduct of one's own life and for one's treatment by others – friends, employers, teachers or insurers. (Novas & Rose, 2000, p. 505)

In this light, genetic risk is not a 'neutral' probability, but a lived, often anxiety-provoking, dimension of reality.

Chronic risk

Kenen et al. (2003b) advocated use of a 'chronic risk' perspective for studying how individuals live with a heightened awareness of their genetic risk. The concept of chronic risk helps explain the lived experience of the *risk* of illness, as opposed to illness itself (p. 316). Kenen et al. argued that individuals at risk for genetic illness exhibit changes in behavior, social relationships and self-identity that are similar, though not identical, to those observed in people suffering from chronic illness.

The chronic risk perspective is heavily influenced by Bury's work on chronic illness (1982; 1991) and Parsons and Atkinson's (1992) work on genetic risk. Two conceptualizations from these works are relevant for the chronic risk perspective and for the current research:

(1). An analysis of chronic risk in terms of biographical disruption. Bury (1982, 1991) contends that chronic illness disrupts daily life such that its taken-for-granted features are no longer stable: Uncertainty is a key feature of the disruptive experience. It involves existential questions such as 'why me' 'why now?' Further, it necessitates a re-thinking of oneself as healthy, capable and in control. This biographical (re)examination is often linked to the growing dependency so often a feature of chronic illness, including HD.

Chronic illness also affects social relationships with others and can cause disruption in interpersonal relations. Disruptions occur because of functional limitations, but also because of the embarrassment such disabilities can create (Bury, 1982). Bury suggests that sufferers attempt to *normalize* in the face of disruption in an effort to cope with the illness – that is, the psychological ‘bracketing off’ of the impact of the illness such that its effects on identity are minimal (Bury, 1991, p. 460). In the current research, several themes that arose in relation to genetic risk clustered under *implications for self-identity*.

(2). The idea of zones of relevance (Parsons & Atkinson, 1992) – the conditions under which genetic risk is or is not salient. People at risk for HD do not necessarily spend all of their time thinking about it (Cox, 1999; Cox & McKellin, 1999). Rather, genetic risk becomes relevant at specific times and/or events throughout the life course and lessens at other times.

The remainder of this chapter will report on how people live with a heightened awareness of their genetic risk for HD in terms of biographical disruption and risk saliency.

Genetic risk for HD – when is it salient?

In the earliest stages of this research, I (rather naively) assumed that genetic risk for a fatal disorder such as HD must *always* be salient. How could you *not* think about it all the time, I wondered? As I quickly learned, for the majority of individuals at risk for HD in the current research, and even for those who had tested positive, genetic risk was

not always salient. Rather, risk becomes salient under several zones of relevance. If it is necessary to talk about risk perception, it is suggested that risk not be represented as a binary salient or not salient dichotomy or a low or high categorization, prominent in much clinical research. Rather, risk saliency interacts with numerous zones of relevance. The relationship can be represented as a matrix with various connections and interactions (Figure 3).

Several zones of relevance emerged in the current research, encompassing many factors that seemed to influence the saliency of genetic risk for HD (Figure 3). These included: Stage in the life course; family history of HD (e.g., geographic/social distance from affected relatives and parent's age of onset); disease-related events in the family (e.g., own test results, others' test results or the death of an affected family member); personal beliefs about whether one carries the altered gene; personality factors (including monitoring coping style); and other important life events that can be specific to an individual (e.g., a divorce, starting university, or noticing 'odd' behaviors in oneself such as twitching). It is worth noting that stage in the life course and family history of HD were most influential on risk saliency for participants in the current research. Their influence is developed in the discussion to follow.

It is not meant to suggest that the zones of relevance in Figure 3 are mutually exclusive nor exhaustive. Rather, these are the zones of relevance that were warranted from a systematic analysis of interview transcripts in the current research. For most participants, several of the subsequent factors interacted to influence risk saliency. For

example, young participant age, combined with a late age of onset in the affected parent interacted to diminish the salience of genetic risk (Figure 3).

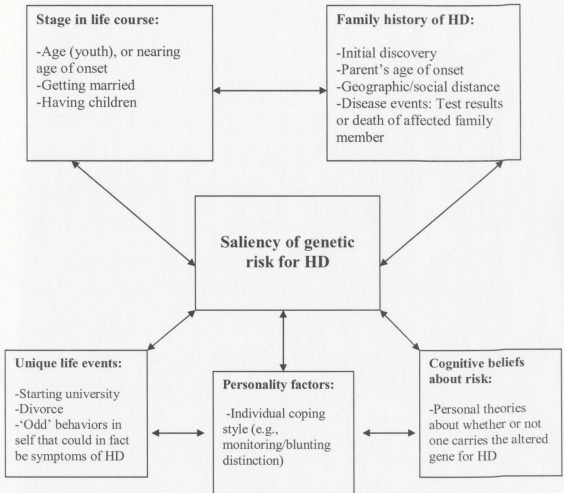
Figure 3. *Zones of relevance for genetic risk for HD***Primary zones of relevance****Potential moderators**

Figure 3 depicts the zones of relevance that emerged in the current study and displays their interaction with risk saliency. Zones of relevance interact with each other

and with risk saliency to affect the heightened or diminished saliency of risk for HD as at-risk person progress through the life course. Stage in the life course and family history of HD appeared to be the most significant zones of relevance in the current research, having a direct influence on risk saliency. They are noted as the primary zones of relevance in Figure 3.

For example, for younger participants who had not yet married or had children, their risk becomes salient when they consider starting a family or committing to a partner. If marriage and reproduction decisions are not prominent, however, at risk participants in their twenties and thirties suggested their youth allowed them to distance themselves from the possibility of HD since it was 'very far away.'

For older at risk participants (forties and fifties), age affected the saliency of genetic risk in at least two ways. For some, it *diminished* the saliency of risk since the threat of developing HD becomes smaller with increasing age. However, for those at risk participants who were nearing the age of onset of an affected parent, risk was likely to be salient. Serena told me, "I know my age and I'm at the age of developing symptoms right now."

Participants' family history of HD also influenced risk saliency in the current research (see Figure 3). Namely, geographic and/or social distance from affected relatives, parent's age of onset and disease-related events in the family (e.g., own test results, others' test results or the death of an affected family member).

For instance, when participants were in close proximity to an affected relative, their own risk became salient. Sherri explained:

So, when [sibling] came to live with me, there was always a reminder there. Even when I lived with Mom and Dad, there was always a reminder there.

I: So it seems like, if it's something you can see, like visible signs, it kind of makes you think about your own risk more?

Yes. I mean, you do think about it, but you don't think about it as much.

I: So it's there, but it's not necessarily in the front of your mind everyday?

That's right. –Sherri, at risk

Sherri's comments were echoed by many participants in the current research, regardless of whether they were at risk or had been tested. Understandably, for participants who had tested negative, the risk of their siblings or cousins was salient when they were in close proximity to an affected relative (rather than their own risk).

Paradoxically, while proximity to an affected relative can heighten risk awareness, it can also work to *diminish* anxiety about one's own risk. When there is a living relative with disease (e.g., several forms of cancer; Sanders, Campbell, Sharp, & Donovan, 2003), a person's *own* risk status is not the priority. Rather, the relative's illness and helping him or her cope becomes the main priority of at-risk person. The current research provides some support for this suggestion. Apparent throughout participants' entire narratives, was worry for affected family members.

This finding has implications for how we might speak about denial of genetic risk. It is not necessarily the case that at-risk person are denying their own risk of developing

illness: Recognizing the implications of their own risk status simply does not translate into worry when there is a living relative close to the age of onset or currently showing signs of the disease. In this context, risk saliency is based on a wider set of social and personal circumstances than one's own risk status – the emotional preoccupation of worry about (or caring for) an affected relative pre-empted thinking about one's own risk.

Conversely, for participants who were separated (geographically or socially) from at risk or affected relatives, genetic risk was much less salient, particularly for participants who had tested negative. Two such participants in the current research, for example, suggested they did not know whether siblings had been tested and there was no indication in either narrative that they worried excessively about their siblings' risk. For both participants, testing negative relegated HD to the background of their lives. For another participant who had tested negative, however, genetic risk was very salient since siblings had tested positive and the family was close (geographically and socially). "Somewhere down the road, I'm going to face a lot of devastation again, as one by one, they are all targeted with HD. I hate the thought of it, but it's coming." Test results of other siblings served to make risk salient and worrisome. And, despite testing negative, this participant admitted to occasional symptom-watching in self.

Participants' own test results, of course, also render risk salient, especially when the test result is positive or intermediate. Kathleen, for example, recalled, "First, when I was diagnosed, I was really obsessive about it. First, I would cry and cry and cry everyday." Similarly, Jerry suggested that receiving a positive test result made his risk

salient. Notably, the death of an affected parent was a particularly difficult time (understandably) which also served to heighten awareness of his own risk:

I don't dwell on it, although it is in the back of my mind all the time. Especially when [parent] was sick and dying was probably one of the strongest points. And, probably even when I first found out, even. Those were the two strongest times ever – first when I found out and when [parent] died with it. –Jerry, tested positive

Unlike stage in the life course and family history of HD, variables such as personality factors, personal theories about risk and unique life events can be thought of as potential moderating zones of relevance (Figure 3). For example, while coping style can have a direct influence on risk salience, stage in the life course (particularly age) can override the influence of monitoring or blunting coping style on saliency of risk for HD.

Summary – Saliency of genetic risk for HD

Individuals at risk for HD do not spend all of their time thinking about their genetic risk. Risk is chronic – it is 'always there,' but whether or not it is salient depends on several zones of relevance. Figure 3 depicts the interconnections of risk saliency and the zones of relevance that emerged in the current research.

Notably, numerous factors combined and interacted to influence the saliency of risk for HD, especially stage in the life course and family history of HD. For example, as at-risk person neared their parent's age of onset, risk saliency was high and symptom watching was likely.

It is not meant to suggest that at-risk person undergo a step-by-step progression from low- to high-risk salience over the course of their lives. Rather, depending on the

zones of relevance, the salience of genetic risk will wax and wane many times over the life-course. These findings challenge much of the clinical research on HD which treats 'test result' as a sole independent variable with a range of measurable effects, some positive (e.g., relief from uncertainty) and some negative (e.g., depression or heightened anxiety). Participant narratives clearly revealed that genetic test result is only one of many factors that affected thinking about and responding to genetic risk.

These zones of relevance also have implications for genetic counseling and follow-up support. While the bulk of clinical research on genetic testing for HD suggests minimal post-test psychological distress, distress can occur some time later (e.g., with the death of an affected relative or as the age of onset approaches), and support could be required at that time. Almost every participant in the current research commented on the lack of follow-up psychological support for families affected by HD. In the only longitudinal study of the effects of predictive testing for HD greater than five years, Timman et al. (2004) suggested that research to date could have underestimated the real impact of a positive test result. They found increased levels of hopelessness in participants who tested positive for the altered HD gene over the study's 7-10 year follow-up. Timman et al. (2004) suggested, "Testing for fatal inherited diseases creates a long-term, lifelong stress reflected by gradually increasing levels of hopelessness as the onset of disease approaches. This pattern may have implications for follow-up of cases" (p. 196). Findings from the current study lend support to their argument.

Chronic risk as biographical disruption

Changes to self-identity

Most participants in the current research acknowledged their risk for HD as being ongoing: It was chronic, though, as noted, not always salient. Participants were aware of changes in their self-identity due to their risk for HD, but attempted to normalize their risk in order to get on with their lives. In fact, an underlying theme in many participant narratives was *let's get on with it*. For example, I had asked if being at risk for HD was something that affected daily life; Serena explained it this way:

Well, yes. Yes and no really. Sometimes, you just block it out. That's the way you have to deal with it sometimes. But, yes, I'd be wrong if I said no. Everything that you see in yourself, you're thinking, 'Oh, this might be just the beginning.' So, there are things like that that you ...yes, so, in every day, yes, I can say you do think about it. Then, you sort of say to yourself, 'No, block it from your mind.' But every little thing you associate with HD. –Serena, at risk

Serena's account is illustrative of the cognitive work at-risk individuals sometimes perform in an effort to 'bracket off' their risk and minimize its impact on identity and daily life (Bury, 1991).

Sherri attempted to normalize being at risk for HD by comparing it to any other risk that exists in life. "You are at risk really for everything. So, if you think about it, I would say it could drive you crazy." Despite her attempt to bracket off the risk, however, she acknowledged it does affect her identity as a healthy person; although, its intrusion is often contingent on a specific incident, such as a body movement:

Sometimes, I will lie down on the bed or on the chesterfield or just sitting there by myself and maybe sometimes my finger will just move or something, twitch or...that's when I think about it. –Sherri, at risk

Rejecting the 'at risk' identity

Other participants who were at risk for HD seemed to resist the at risk label since it threatened their identity as a normal, healthy, independent person. Roxanne was most adamant about this:

Now for me, I say if I got it, I'm not letting it affect me anyway. I'm one of those people who's a fighter and I always tell myself that whatever happens, nothing is going to affect me enough that someone has to take care of me full time. No matter what, I'll struggle through on my own. –Roxanne, at risk

For some at-risk individuals, on the other hand, there was no evidence that they perceived themselves as someone at risk for HD. That is, there was no sense this identity was ever *consciously* acknowledged as part of their self-concept. Brenda explained, “You want to forget that this is even part of you. You want to get on with a normal life.” I asked if she had ever thought of herself as a person at risk for HD:

No, I don't. No, I don't. Like I said, I don't know if subconsciously I made decisions along the way – like I said I was [age] before I had [children]. Now, whether subconsciously these things were in the back of my mind...I feel, today, I feel confident that I am over the hump. I am over the age in my family because they were all so young. –Brenda, at risk

Note that it is Brenda's age which seems to allow her to bracket off the impact of being at risk in her life. Similarly, Michelle also explained that her age permits her to do the same. I had asked if being at risk for HD was something that affected her every day. She said, “No. I have to honestly say, no. I can honestly say it does not affect anything that I do, and especially not at my age.”

Testing and self identity

Tested participants also displayed biographical disruptions in terms of self-identity. They too, attempted to bracket off the impact of risk information in their lives. Kathleen, for example, explained how her risk intrudes on her daily life and her attempts to keep the intrusion to a minimum.

I: Do you find that you are checking yourself for signs or symptoms?

Yes I am. I think when you are diagnosed with the gene, you are always checking yourself. I think that's just part of your life everyday. For example, a couple of times, I just flopped down for no reason, and I thought, 'My God.' I didn't trip. I just went down, my legs just gave out under me. I wondered about that. Twice that happened. So, that kind of scared me, and I thought, 'Oh my God.'

I: So, you do kind of think about it everyday. That's something I'm curious about. Is this something that impacts your daily life?

No, I don't let it. First, when I was diagnosed, I was really obsessive about it. First, I would cry and cry and cry everyday. I would have a spurt of crying and it would be on my mind 24/7 more or less. But, I learned not to do that now. Now, if something happens, I do think, 'Oh my God, is it the HD?' but, I don't let myself dwell on it in that sense anymore. I can't change it. It's something I have to live with. I have to realize that I have to make the best of what I've got left. I can't be...give up my life because I have the gene for HD. -Kathleen, tested, intermediate gene

Kathleen's courageous attitude was typical of participants who had tested positive or intermediate for the altered HD gene in the current study. Participant narratives revealed their risk was chronic in that it was 'always there,' but it could be mitigated somewhat by trying to 'get on with it.' As Jerry put it:

For me, I just sort of put it on the back burner most days. It's with me. I know I have it. I don't show any signs. I know someday I'm 9 chances out of 10, if there's no cure, 10 chances out of 10 with me, I'm going to have it. Of course, the

other way I look at it, I could be dead with something else before then. So, you can't dwell on it. —Jerry, tested positive

Some participants who had tested positive for the altered HD gene or had received an intermediate result also spoke of the ways HD had made them a stronger, or somehow better, person. Kathleen, for example, explained it this way:

I think finding out I have HD has made me a better person. I think it's given me a knowledge of what other people go through in their life. I think it makes me more empathetic to other people's illness. I think it made me a kinder person. It's made me really listen to people, instead of brushing them off. —Kathleen, tested, intermediate gene

Jerry also commented on changes in his identity and outlook since his testing:

I suppose you probably look at life sometimes somewhat different. I'd say that's probably the brightest part of it. (...) For me, it's brought me more spiritual satisfaction I guess. (...) I know I'm a better person. I think I'm a better person. My heart is as big as what the world is. I just feel good sometimes. I see the spiritual part of it, the religious part of it, or whatever. A lot of people don't believe in God, but through this stuff, I think I've seen...I feel a lot better about things than I did when I found out. —Jerry, tested positive

These attitudes stand in stark contrast to those reported by Charmaz (1983) in her study of living with chronic illness, including heart disease, multiple sclerosis and cancer:

The language of suffering these severely debilitated people spoke was a language of loss. They seldom talked of gaining a heightened consciousness of the world, revelations about self or insight into human nature from their experiences (1983, p. 191).

This contrast is notable, however, since it highlights how being *at risk for*, rather than being *affected by*, HD are two different phenomenological experiences. The

narratives of those affected with HD or those who have tested positive and are nearing the age of onset were illustrative of this difference. These narratives did echo Charmaz (1983). Victoria, for example, spoke of her fear of 'losing herself' to HD:

The most frightening part of it is losing yourself. I think you lose your personality, you lose your energy. You become a different person. (...) Once it strikes, I'm a very strong willed person and very capable. But, it takes you out in so many ways...I think, 'What is going to happen to me?' When you lose your spirit, you lose a lot I think. –Victoria, tested positive

David noted how social others view him differently now which threatened his identity as a normal, capable person:

People kind of, they don't expect you to be able to do much. They don't expect you to take on any responsibility or to do anything. Even my own [relatives], I think even the first time they saw me when I had to pick something up they said 'I'll take care of that for you.'

I: So once they know that you carry the gene, you mean?

Yes. So they think that you are weaker physically and mentally. –David, affected with HD

David also commented on the bitterness he experiences as a consequence of testing positive for the altered HD gene and as HD symptoms progress:

I am envious of other people's success and people making long-term planning for retirement and grandchildren and so on. I am bitter over that. That is the aspect of living with knowledge.

Chronically ill people, and those nearing the age of onset for a chronic illness such as HD, can perceive developing limitations, loss of control and constraints on their futures as 'losses of self,' losing their former identities as healthy, capable people without developing new, equally respected ones (Charmaz, 1983).

Summary: Chronic risk as biographical disruption - changes to self-identity

At-risk individuals were aware of the impact of their genetic risk for HD on their self-identities and on their life in general. Many of these participants went back and forth in their minds between a heightened awareness of their risk and active efforts to bracket off (Bury, 1991) the at risk label and get on with their lives. This was also observed in participants who had tested positive or intermediate for the altered HD gene. In these narratives, participants attempted to minimize biographical disruption and maintain a normal life in the face of chronic risk and future chronic illness.

Some individuals at risk for HD appeared threatened by the at risk label, perceiving it as an affront to the view of themselves as a healthy, independent person. Roxanne's narrative was illustrative of such rejection. In this narrative, there was little evidence that chronic risk intrudes on daily life and, as such, little mention of efforts to normalize.

Others showed no evidence of having accepted the at risk label as part of the self; age often allowed this minimization of the biographical disruption such acceptance might invoke.

Concepts of suffering and loss in relation to chronic illness were prominent themes in early studies of living with chronic illness (Thorne & Paterson, 1998); recent research adopts a more optimistic perspective by uncovering those aspects of chronic illness that are healthy, transforming and positive. Kathleen's and Jerry's narratives were, in some ways, reflective of this shift. However, participants who had tested positive (and

were currently asymptomatic) often made reference to the future when they would be manifesting symptoms of HD. They acknowledged that while they were currently normal, healthy, asymptomatic people, that would change in the future when HD symptomatology began. At that point, they expect different reactions than they currently receive from social others. Therefore, it is suggested that chronic risk and chronic illness not be dichotomized into 'loss and suffering' on the one hand, with 'transforming and positive' on the other. Narratives in the current research suggested that the lived experience of chronic risk and chronic illness was multi-faceted, containing elements of current and future loss and suffering, but also positive aspects such as greater empathy and a positive attitude.

Disruptions to social relationships

Numerous social relationships participants had with others were affected by their chronic risk. This biographical disruption intruded on interpersonal relationships, notably communicative interaction. Many participants, for example, did not want to worry other family members. In this sense, then, communication (or lack thereof) was used as a form of protection. For example, Julie recounted how, regardless of test result, she and her siblings decided to tell her parent the test was negative:

We decided then that if any of the rest of us were positive, we weren't going to tell [parent] because it probably wouldn't be an issue in their lifetime. They didn't need to know because there was absolutely nothing they could do about it. –Julie, at risk

Similarly, Lori explained how she had not told her affected parent about her own genetic testing, fearing their reaction if her test result was positive:

They know nothing. Like I have tremors and things that I've been hiding from them. Or when I bump into walls and trip or something, I make a joke out of it. I don't want them to know. My [parent] (long pause in speech) – they would kill themselves if they knew that.

I: Really?

There's no doubt. If they knew that I was somehow damaged from something that they had no control over? Oh yes. –Lori, tested, waiting for results

For some at risk participants, then, their communicative interactions were disrupted when they felt they must protect a family member. This was particularly difficult when an at risk family member lived out of town. As Julie explained it, "I don't know if I'm supposed to talk about the things that I hear or not talk about them because it's not all positive." And, I have already noted how parents of at risk children actively protect them until such time they are mature enough to hear about the family history of HD.

Other types of communication disruptions occurred as a result of genetic risk and illness. Numerous participants recounted stories of other family members who did not want to acknowledge the family history of HD or their own risk. Brenda, for example, recounted how a sibling distanced him/herself from her for fear of having to acknowledge symptoms of HD:

And for a period of four years they never spoke to me..[pause]..over this incident. And I really believe at that time they were starting to show the signs. And that they had to keep me away because if I started to see the signs there, I would bring them up. –Brenda, at risk

Brenda suggested that her sibling wasn't ready to face the illness, and as a result, became distant towards her.

Caregivers also commented on the distancing of family and friends as a result of HD.

I find that with family – here's the way my family is anyway, my nieces and nephews and everybody else – they don't come around, they pretend it's not there. They just left us there. (...) I find that with the family, it's like they won't come to see you because they don't want to see what's there. It's like if they don't see it, it's not happening. –Laura, caregiver, excerpt from support group meeting

They [relatives of person she cares for] are angry with me because I suggest that maybe they could look into getting the testing done. But they're not talking to me anymore. –Shirley, caregiver, excerpt from support group meeting

For both Shirley and Laura, there is a sense of isolation from other family members who do not want to 'see what's there.'

Summary: Disruptions to social relationships

Biographical disruptions in terms of relationships with others were observed in the current research, and these were most evident in communicative relationships. These disruptions were sometimes anxiety provoking and upsetting for at-risk individuals, contributing to rifts in families affected by HD.

This section has focused on disruptions to communicative relations specifically, since relatively little is known about how families communicate about genetic risk and with what consequences. It should be noted, however, that relationships with others were affected in additional ways as well. One at risk participant, for example, recounted how her in-laws did not approve of her relationship with her husband owing to her family history of HD. Others who had cared for an affected relative in the past commented on the impact of this experience on their spouse and their marriage (e.g., lack of privacy,

minimal time to spend with spouse). And recall how some family members did not approve of other members' decision to have children despite knowledge of the risk for HD. What all of this suggests is that while genetic risk is not always salient, there are many tacit social processes occurring in the lives of at-risk individuals and their families (e.g., social disapproval of reproductive or marriage decisions, protective communication patterns, and social or geographic distancing of family members). These findings highlight the broad impact genetic risk can have on whole families, not just the at risk individual – frequently acknowledged by participants in the current study.

Discussion

This chapter highlighted the variable meanings and lived experience of being at risk for HD. Broadly speaking, living at risk or proceeding with genetic testing had significant implications for self and others, notably other family members. Each and every participant in the current research situated and negotiated his/her risk in the moral and material contexts of their families.

As such, participants' narratives challenge Beck's (1992) contention that risk society is marked by a trend towards individualization. In 'risk society,' the core features of modern society that once worked to secure social progress (e.g., government, science and medicine) are now marked as the primary risk producers as a result of reflexive modernization. Since (supposedly) these entities can no longer be trusted, it is now the sole responsibility of individuals to invent their own identity. In so doing, risk society is replete with choices we all must negotiate. The 'responsible' citizen of risk society

chooses wisely, managing and controlling (as far as is possible) the myriad risks of such a world. In this context, risk is understood as human responsibility.

In general, narratives of genetic risk were permeated with the discourse of responsibility in the current research – responsibility to future generations, to current and future partners, to be tested, to plan future care and to communicate genetic risk. This observation accords with research on other autosomal dominant disorders (e.g., Boutte, 1990). However, the perception of risk as responsibility was *not* an individual enterprise, much as Beck's (1992) risk society suggests. Rather, *risk as responsibility* often meant duty to others, not self, as the narratives in this chapter have demonstrated. Thinking about risk was squarely situated within the family history of HD – past, present and future. Risk was a family matter, not an individual one, and this was especially pronounced for those participants who had had genetic testing. Similar findings were reported with women at risk for breast cancer (Hallowell, Foster, Eccles, Ardern-Jones, & Watson, 2004). The majority of women in that study viewed their participation in genetic testing as an 'altruistic act' that would provide other family members with valuable risk information.

Even for participants who had not been tested, there was a moral duty to care for, or in some way assist, other affected or at risk family members (current and future). This was pronounced in participants who were close, geographically and/or socially, to their families. It was most pronounced in the narratives of current caregivers.

When the meaning of genetic risk resides in responsibility to others and to self, anxiety and tension can permeate life choices. This was evident in the narratives of participants in the current research. Younger participants who had not yet married worried about how or when to tell a future partner about their family risk. Married participants worried about being a burden to their spouse. Partners of at-risk person and those affected with HD worried about their ability to provide extended, long-term care.

Partners have received little research attention in predictive testing programs (Quaid & Wesson, 1995; Tibben, Timman, Bannink, & Duivenvoorden, 1997). Quaid and Wesson (1995) found that spouses were more depressed than their at risk partners at baseline, while Tibben, Frets, et al. (1993) observed that spouses had the most difficulty coping with the impending threat of HD, and this was especially likely for partners with children (Tibben et al., 1997). Findings from the current research were in line with these earlier findings. Recall Daphne who noted the future was 'a very scary thing' for her. Laura also preferred not to think about the future since it was perceived as uncertain and frightening.

The perception of risk as responsibility also has implications for how social others view at-risk individuals and families. If others expect at risk families to be responsible for controlling and managing their genetic risk, what 'choices' are open to persons at risk for HD? Should they have children? Should they get married? And, if family members disagree on how best to manage genetic risk, are family rifts inevitable? This chapter has revealed how some family members disapproved of other members' reproductive

choices. And recall the participant who remarked that some members of the family 'hated' her and her siblings when they informed the extended family of HD. In this light, genetic risk is not a 'neutral' probability, but a lived, often anxiety-provoking, reality.

Beyond responsibility, the meaning of risk also resided in its representation as an index of threat to self and other family members. Risk was equated with worry, anxiety and fear, especially for at risk offspring. This conceptualization of risk accords with current constructions of 'risk' as a danger or hazard (e.g., Douglas, 1985; Lupton, 1999a, b).

The threatening meaning of genetic risk in the current research could be related to participant views of HD itself. Almost without exception, participants spoke of HD as a 'devastating,' 'horrible,' 'never-ending' illness, a 'nightmare.' Living at risk for HD (and testing positive for the gene) invoked memories of affected relatives and comparisons to self, or at least, to future (potentially affected) self.

Notably, however, some at risk participants were able to manage the threat of genetic risk by minimizing the danger. This was sometimes accomplished with fatalistic beliefs about future healthcare (e.g., 'if I get it, I get it') or by regarding genetic risk as just one more risk in the many that already exist in life. Thus, despite the focus on *risk as emotion*, not every participant spoke about genetic risk as an index of threat, at least not to self. This was likely for those participants who had tested negative and no longer had to worry for themselves or their children. Nonetheless, they often recognized the remaining threat to their at risk or affected family members (usually siblings or cousins).

For many participants, their risk was chronic (e.g., 'it's always there'), but not always salient or anxiety provoking. Rather, several zones of relevance emerged in participant narratives, highlighting when genetic risk for HD might be salient. The discussion of zones of relevance illustrates how participants in the current research attempted to 'localize' their risk for HD (Sarangi, Bennert, Howell, & Clarke, 2003). "Localisation strategies contextualise the scope of reference from the comprehension of risk in the probabilistic sense...to a fuller appreciation of the significance of the risk for the whole life of the individual and their family" (Sarangi et al., 2003, p. 161). While many participants knew their risk was 50:50, it was re-contextualised along a temporal dimension by drawing upon memories of affected relatives as a way of localising potential onset for self. Note that the process of localisation was contingent on family history of disease, not population risk figures. Thus, when at risk participants in their fifties have lived through most of their risk of developing HD without doing so, they correctly interpret their current risk as small.

Finally, the narratives of some participants challenge the pessimistic view of at-risk person and individuals who have tested positive in the literature (e.g., they are unable to cope with a positive test result or they are 'in denial;' Duisterhof et al., 2001). Such individuals might not be in denial at all. Results from the current study suggest they *dismissed* their genetic risk for HD as an *immediate* priority. An overriding theme of 'let's get on with it' pervaded participant narratives, despite the biographical disruptions that often accompanied discovering the family history of HD.

CHAPTER 10

STIGMA, RISK AND COPING STRATEGIES

The previous chapter highlighted some of the salient meanings converging around genetic risk for HD and outlined some of the implications of living with risk information in daily life. In that analysis, I highlighted the distinct emotional and moral meanings with which genetic risk for HD was infused. The notion of chronic risk was employed to demonstrate the biographical disruptions that arose as a consequence of living at risk. Notably, however, I argued that genetic risk was salient only under certain zones of relevance. This chapter continues the discussion of living with genetic risk and illness by exploring (1) participants' narratives about stigma and (2) their stories about coping with genetic risk (and/or illness) in everyday life.

Is living at risk for HD stigmatizing?

The current research aimed to explore participants' perceptions of (and/or experiences with) the stigma associated with being a member of a family affected by HD. Beyond anecdotal accounts (e.g., Cox, 2002), I am unaware of any empirical research that investigated the relationship between living at risk or testing positive for HD with social stigma. Similarly, minimal research documented social stigma towards persons *affected* with HD (but see Leung & Leung, 2002, for an exception). To address this gap, participants were asked if they thought there was any stigma associated with living at risk for HD or with the illness more generally. Crocker et al. (1998) noted that discrimination is a hallmark feature of social stigma. Accordingly, participants were also asked if they,

or their affected relative(s), had ever experienced any form of stigma, including genetic discrimination in employment and insurance contexts.

Participants had very similar views on whether they felt HD was a stigmatizing condition. They also adopted similar coping strategies (e.g., educating others) for addressing responses they perceived to be stigmatizing. In response to a direct question about whether stigma existed in relation to HD, participants suggested there was no real stigma since (1) the illness was out of their control, and (2) ignorance surrounds the illness. Despite the overt disavowal, however, analysis revealed subtle responses to perceived stigma, such as not informing an employer about a positive test result or advising children to start a career or obtain life/health insurance prior to testing.

Participants' perceptions of stigma diverged, however, with caregiver narratives. Caregivers (current and former) recounted very specific instances of social stigma in a variety of social contexts. This difference was notable, and it highlighted the distinction between visible and concealable stigmas.

Crocker et al. (1998) argued that two dimensions of stigmatizing conditions were critically important for understanding the subjective experience of being stigmatized: Concealability and controllability. Participant narratives in the current research can be interpreted within these dimensions. For example, asymptomatic participants suggested that stigma would not be likely until disease onset when HD symptomatology was clearly visible. And, even when specific examples of stigma were recounted, there was little

evidence that participants suffered negative consequences. The illness is beyond their control; as such, negative responses of others could be dismissed as not valid to the self.

The next section discusses perceptions of, and experiences with, stigma under four interrelated themes: (1) Sympathy, not stigma; (2) There's not really a stigma, but...; (3) Enacted stigma; and (4) Dismissing stigma – ignorant others. Subsequent to the presentation of stigma narratives, the second half of this chapter will discuss participant coping strategies for living with genetic risk information.

Sympathy, not stigma

Excepting some current caregivers, nearly every participant suggested that most people were sympathetic to them and/or their affected relative. Michelle's comment was typical:

I don't think so. No, not around here. I find where we live, I find that people are more supportive and sympathetic and like, 'My God, it's terrible that that happened.' But I think that they are more sympathetic than anything else.
–Michelle, at risk

Participants who had tested positive for the altered HD gene also commented on the sympathy they received from others:

I think most people are pretty sympathetic. (...) I think everyone felt sorry for us and about our [parent]. I don't think people that I know ever seem to feel any differently because I've come down with this. –Victoria, tested positive

Everybody is good with it. They'll ask questions like, 'How do you know?' and 'What is it?' I told them I had gone and got testing for it, and tested positive. I look at them and laugh and say, 'I'm going to be normal yet for a while.' I think people feel bad for you. I've never had any bad experiences with it. –Jerry, tested positive

Jerry's comment about remaining 'normal' was notable since it underscores the distinction between a concealable and a visible stigma. Asymptomatic individuals have a concealable stigma: Others only know about future illness if at-risk individuals inform them, as Jerry did. When illness begins, however, HD is often marked by visible symptoms (e.g., movements). Participants were aware that stigma was a *future* possibility with disease onset; in Scrambler's (1998) distinction, a felt, rather than enacted, stigma.

Kathleen, for example, speculated:

Right now, I'm a normal person. I look normal. I don't think there's a stigma attached right now. But, down the road, when I start to look retarded and I start to shake and I can't walk, and get that vacant look, I think it's going to be different. I think there will be a stigma attached then. –Kathleen, tested, intermediate gene

Thus, some participants who could be facing a future with HD were well aware that future stigma remained a possibility. While participants were asymptomatic, however, negative responses from others were rare. Rather, the dominant response to participants in the current research was sympathy.

Interestingly, participants cited place as a determining factor in the responses they received from others. Specifically, Newfoundland was perceived as a friendly, welcoming place with good-hearted people. Jerry said:

People here are sympathetic. We are good-natured people. (...) Like I said, in New York, if you fell down, they'd probably walk right over you. Whereas in Newfoundland, they would ask what's wrong. –Jerry, tested positive

Similarly, Gerald noted:

Smaller towns in Newfoundland do bring out the best in people. They don't have the anxiety about people who are different mentally or physically. –Gerald, tested negative

While other participants agreed the response to their affected relative was usually sympathy, rather than stigma, some suggested it was a result of ignorance about HD more generally, rather than place. When I asked if she thought there was any stigma associated with HD, Roxanne noted:

Not really, because most people in my area don't understand what HD is. I remember when [relative] found out they had it, I told a couple of my friends and they were, 'What is HD?' They didn't even know that it existed. They had never heard of HD before. –Roxanne, at risk

For Roxanne, then, the ignorance that surrounds HD precluded the perception of HD as a stigmatizing condition since most people 'don't understand what HD is.' Perceived ignorance about HD seemed to allow participants to dismiss any sort of stigmatizing response they received from others, as we will see shortly.

Summary: Sympathy, not stigma

The majority of participants in the current research recalled sympathetic, or at least non-negative, responses from others in relation to HD. Some participants thought these responses could change with disease onset; although, there was no sense that they were particularly distressed by that possibility at the time of our interview. Participants cited both place and ignorance for the relative lack of stigma associated with HD.

It is not meant to suggest that *no* perceived stigma existed in relation to HD. While the day-to-day experience of living at risk was generally perceived as non-stigmatizing, stigma certainly was perceived and experienced in specific social contexts.

There's not really a stigma, but...

Despite the generally sympathetic response from social others, participants did recount specific instances of what they perceived to be stigma, notably in insurance contexts. For example, eight participants (in one case, a participant's family member) had difficulties obtaining life and/or extended health insurance. In nearly all cases, the participant was at risk at the time of the application (i.e., asymptomatic). Some were outright rejected, while others could not afford the exorbitant cost of insurance for a person at risk for HD. If the participant had subsequently tested negative for the altered HD gene, obtaining insurance was no longer problematic, as was the case for two participants. When the test result was positive, however, obtaining life or extended health insurance was far more difficult, if not impossible. Laura, for example, noted:

We had it done [genetic testing] before we had insurance or anything. We had nothing. If I could look back now over time, I would have had insurance first before we did this. –Laura, caregiver

Other participants noted how potential insurance discrimination was a motive in their delaying or declining genetic testing. One at risk participant commented:

There's also insurance purposes. I'm sorry, but I don't want to have to put down something and get jipped out of insurance that anybody should be allowed to have. You could get cancer at the same time as I could get sick. And I don't get insurance? I don't think that's fair. I really don't. So I'm not going to get tested until I have some kind of insurance. –Female, at risk

Others had difficulty changing the amount of their insurance policy:

I went to increase my insurance – to put it up – and one of the questions at the time was, 'Do you or any of your family have a hereditary disease?' which of course I had to mark 'yes.' I got turned down because of it. –Sherri, at risk

Sherri perceived this refusal as discrimination since she could be healthy for many years before, or if, HD manifested:

Here I was [age]-something years old and it was almost like I was going to die tomorrow. I still have 20 to 25 years even if I did get it. (...) It's discrimination. That's what I told them. –Sherri, at risk

For some at risk participants, then, obtaining or increasing life and extended health insurance was problematic, if not impossible.

When the family history of HD was discovered at mid life (as it was for many participants in the current research), participants could already have insurance policies that were unaffected by the discovery of HD. This is only possible if at-risk person do not disclose the discovery of HD in the family – and some don't. For example, one participant noted, "We would never, ever tell the insurance company or anything like that. We would probably be taken right off the list."

Fear of employment discrimination was also evident in some participant narratives, if not for self, then for at risk offspring. It should be noted that most participants had disclosed the family history of HD to their employer and many of their work colleagues; no participant perceived any stigma at their place of work. However, participants recognized that discrimination could be possible in specific work environments. As such, risk for HD was not disclosed to every employer. One participant commented on a tested relative:

Obviously, they're not going to tell an employer that they've been tested or anything. We wouldn't put that on their resume or anything like that. We keep it as much a secret as possible because we don't want anybody to look at them like, I guess put a stigma on them.

Other participants also noted to need to 'be quiet' about the family history of HD in insurance and employment contexts:

I realize in the business world, there's some people with a crueller streak I suppose. That's why the kids won't get tested I guess. Some people might hold that against them.

Like I said to [names children], don't get tested for insurance purposes. If they ever ask you if you've got a disease, you can say no and you're being honest. No one can discriminate against them then.

The preceding narratives revealed a tacit recognition of potential discrimination in employment and insurance contexts, despite the suggestion of most participants that sympathy, not stigma, was the usual response to their family history of HD.

Finally, at least two at risk participants feared that healthcare professionals could be 'biased' if they knew about the family history of HD. One commented:

I don't think there's a lot of stigma attached. I think people are more sympathetic with you. But, from our experience, I find the medical professionals might be a little more biased. I think once they know you have this disease, no matter what happens, they are, 'Why should we try to save them, let them go?' Everything is Huntington disease. —Female, at risk

The participant felt an affected relative had not received proper medical care owing to the fatal nature of HD. Similarly, another feared any health problems that might surface would be attributed to her family history of HD and not investigated further:

I guess if I am the gene carrier for Huntington's, if I am to go to the doctor and I was tested, I think that a lot of the problems would come out, like I said, once again, 'That could be your Huntington's.' And I don't want anyone to know it in the professional field because a lot of the things wouldn't even be tested to get further results. It's just blamed. —Female, at risk

For these participants, then, while sympathy was the dominant response to their affected relatives, they did perceive a potential stigma attached to HD in the healthcare arena. That is, both were concerned that once their family history of HD was known, health problems that arose would not be properly investigated. Rather, healthcare professionals would simply attribute the problem to HD.

Summary: There's not really a stigma, but...

Most participants perceived *sympathy*, not *stigma* in response to their family history of HD. However, the preceding narratives demonstrate a tacit awareness of potential discrimination, notably in employment and insurance contexts, but sometimes also in healthcare contexts.

These stories are illustrative of the effect perceived (or felt; Scrambler, 1998) stigma can have on communication of genetic risk and on genetic-test decisions. Several participants did not disclose their (or their relatives') risk for HD to employers, insurers and healthcare professionals. Others also advised their children to postpone testing until career and insurance were obtained. And, at least two at risk participants noted that potential insurance discrimination was a motive in their declining or postponing the genetic test for HD.

It should be noted that several participants recounted specific instances of social stigma in contexts other than insurance and employment. For example, some commented on the stares and pointing their affected relative received from social others when they were in public contexts such as parks or restaurants. These recollections were almost

exclusively from participants who are current (or were former) caregivers. It is not meant to suggest that enacted (i.e., actual) stigma was experienced solely by caregivers. In the current research, however, experiences of enacted stigma were most often narrated by participants with caregiving experience.

Enacted stigma

Participants who were quicker to agree there was a stigma associated with HD were usually caregivers. They could recount specific examples of what they perceived to be stigmatizing responses from others. Shirley, for example, recalled other families affected by HD and their experiences with stigma:

One person – you’ve probably heard of it, I don’t know – one person, a lady who has Huntington’s, they fired her, immediately. Another person - Huntington’s sometimes can seem like, or have the same sort of actions as a person who is drinking? (...) I know of one person who didn’t drink and was fired for drinking on the job. So that’s a stigma – Shirley, caregiver

These sorts of incidents make Shirley angry, and her *own* experiences caution her to be selective about public outings:

It makes me angry because I have to be very careful. When [names person being cared for]’s having a good day, I’d like to, you know, take them out to dinner or out to lunch for a bowl of soup or a coffee or something. And I have to be very careful of how their behavior is that day.

I: Ok, before you choose to go outside in public, you mean?

Yes. Yes. And whether I stay inside or not. Because people are staring. And they’re a very proud person. They’ve always been a very well dressed, very well groomed, very well spoken person. And for them to realize that someone is staring at them, they’re very hurt by that. So I’m not going to subject them to something like that if I can possibly prevent it. – Shirley, caregiver

Shirley, like all caregivers in the current research, is protective of her affected relative and tries to avoid situations that have the potential for stigmatizing responses from others. However, this constrains her behavior and social relationships with others, just as it limits the outings for her affected relative.

Laura recounted several instances of stigma in response to her affected relatives. One of her relatives enjoys walking, but as Laura told me, "They always do it in the evening because they don't want people staring at them." The same relative, "went out once and got beat up because they thought they were drunk." She recalled a public outing when another family member (unaffected) was visiting:

People stare. My [unaffected relative] came down last year and we went to the park. They stared so bad. My [unaffected relative] was so upset. If you know there's something wrong with a person, you don't stare them down. You have the courtesy and respect they deserve. –Laura, caregiver

Marjorie has also experienced stares of others while out with her affected relative. When I asked if she thought there was any stigma associated with HD, she said:

We haven't found it. Not from friends and family. Definitely not. The thing that bothers me is – I guess it's because people don't know – they stare at [relative]. Like in the mall. People will actually turn around. Now I don't mean kids – you can overlook that. But grown adults. I mean [relative]'s walking with a cane. (...) If they weren't walking with a cane, they might think [relative] was drinking. –Marjorie, caregiver

The unsteady gait typically associated with the choreic movements of HD can mimic a drunken stagger. One participant who had cared for an affected relative in the past, for example, remembered, "Now once when we were walking down the road,

somebody sang out from a truck and said, 'Pretty early in the day to be dragging a drunk around.'" Similarly, Sherri recalled:

I think the worst time I had when I was younger was people thought [relative] was drunk. We would hear people snickering. We used to have a [event] every summer and [relative] went to them. I was off with my friends but I would see [relative] walking and people would look and stare and point and laugh at them because they were drunk. –Sherri, at risk

Nearly every participant perceived widespread ignorance surrounding HD.

Participants suggested that others simply do not know what HD is or how it manifests. As a result, affected individuals can be perceived as drunk, contagious or somehow mentally challenged. Sherri, for example, commented on others' perception of HD as 'contagious' and the shame this can cause in people at risk for, or affected with, the illness:

There is not a lot of awareness, and I think people are ashamed.

I: Do you think?

Yes I do.

I: So, people maybe don't talk about it?

They don't talk about it and they are probably ashamed to say that, 'Well, my mother or my grandmother had Huntington disease.'

I: So do you think that's because of the way other people react?

I think so, yes. It is almost like they think that it is contagious. –Sherri, at risk

Shirley agreed that HD is viewed by some as something that can be caught and must be avoided. "So, stay away from them, you know? It's almost like it's a contagious thing."

Some participants questioned whether the stigmatizing reaction of others was due to fear of the physical or mental manifestation of HD. Lori, for example, commented:

Is it the cognitive thing that makes people standoffish or afraid of it? Or, is it the physical stuff? I've found that with the Alzheimer's stuff, anything that was related to the mind, the brain, people were more afraid of than they were physical things. –Lori, tested, waiting for results

Taylor (2004) noted that the both the physical and mental manifestations of HD could evoke stigmatizing responses from social others, and the narratives presented herein would seem to support her claim.

However, it is interesting that both participants who were currently affected with HD did *not* perceive HD to be a stigmatizing condition, or at least, they were not distressed by the responses of others in their social environments. I asked Steven, for example, if he noticed people staring when he was in public contexts:

Occasionally, yeah.

I: And does that bother you?

No, not really. –Steven, affected with HD

David suggested that current medications are changing the experience of living with HD and also changing the reactions of others:

I think that for older people, they still remember Huntington disease being the cases like, I can remember my [relative], you know restraining and so on. But, for the younger people now, the people that they see with Huntington disease I imagine, including myself, they are not seeing the same thing.

I: Okay. So times have changed almost?

Yes. And the thing with medication is, and people don't have the movements. So, it's not such a shock to meet anyone with HD. –David, affected with HD

David's comment was illustrative of the changing phenomenological experience of living with HD resulting from new genetic knowledge and technologies. Medication can sometimes assist in keeping the chorea under control; although, this is not uniformly true since HD symptomatology is quite variable. His own experience, however, is quite different from his affected relative who had to be restrained.

Summary: Enacted stigma

In the previous section, *there's not really a stigma, but...*, I presented narratives of felt stigma experienced by some participants. That is, some at-risk individuals speculated about stigmatizing responses that *could* happen in the future. Conversely, some participants (usually caregivers) had experienced *enacted* (i.e., actual) stigma, normally in the form of stares or whispers of 'what's wrong with them?' in relation to their affected relative(s). These responses from social others were sometimes distressing and infuriating for caregivers who work to protect the affected person. Interestingly, affected persons themselves did not appear distressed by the reactions of others in their social environments.

It is reiterated that enacted stigma was not exclusively a caregiver phenomenon. At risk and tested persons also recalled instances of enacted stigma (e.g., see Sherri's comment above). However, they were always in relation to their affected relative, not self. It is unsurprising, therefore, that caregivers of persons affected with HD were most likely to suggest stigma exists in relation to the illness.

Participants mused about why others react as though a person affected with HD is somehow ‘contagious’ or ‘mental,’ and nearly every participant suggested that ignorance surrounds HD. The reader could wonder how participants cope with the stigmatizing responses of others, or how it is that some perceive stigma, and some do not. One interpretation is the perception of ignorance surrounding HD that allows participants to dismiss that stigma exists or to dismiss the perceived stigmatizing responses of others.

Dismissing stigma – Ignorant others

I have already noted how some participants suggested there could be no stigma associated with HD since no one knows enough about the illness to stigmatize at all. However, the previous sections revealed that some participants were aware of the potentially negative views of HD held by others, and caregivers recounted numerous examples of what they perceived to be stigma associated with HD. Notably, however, there was no evidence to suggest that participants viewed these attitudes as valid. Nor did they appear to internalize these negative attitudes or deem them applicable to the self. As a result, they appeared to suffer no ill effects on their self-concepts. In general, the stigma associated with being a member of a family affected by HD was not presented as something for which participants were responsible. Rather, it was due to the ignorance of those who stigmatize.

In fact, the most salient aspect of participants’ entire narratives in the current research was the perceived ignorance surrounding HD. Cheryl commented:

It’s like this exotic disease that no one has and no one knows about. It’s so uncommon, it’s almost like one of those diseases you see on TV. You know, a

child is born and they look fifty or something – they age really quickly. It's like that. It's so unknown, no one knows about it. It's so uncommon, but it should not be so uncommon because so many people are affected by it. –Cheryl, at risk

Participants suggested that social others simply did not recognize or understand (1) HD symptomatology and (2) that having the illness is beyond one's control. At-risk person did not blame themselves for having inherited the gene; however, they recognized that others who do not understand HD could blame the affected person for the illness or their behavior. Kathleen, for example, noted:

People aren't educated. A lot of people don't understand about diseases, especially Huntington disease. They don't realize that it's beyond your control. It's a fluke of nature that you've got this. –Kathleen, tested, intermediate gene

Similarly, Roxanne commented:

It's not their choice, it's not like they want to be making a fool of themselves and having trouble with communication and movement and all that out in public. It's something that they can't control. –Roxanne, at risk

Generalized others also do not know how HD manifests itself; as a result, HD symptomatology can be perceived as drunken behavior or mental illness. For example, Gerald recalled the reaction to his affected relative:

We used to take [relative] out and go to a store sometimes, and they're standing up and they're twitching back and forth, and doing what they call the 'dance.' You could look around and see people looking at them and wondering, looking them up and down and wondering, 'what the hell is wrong with them?' You take them by the arm and try to lead them in and they say, 'Well, what's wrong with them? Are they out of the Waterford?' –Gerald, tested negative [Author note: The Waterford is a local, primarily psychiatric hospital.]

Some participants suggested that these sorts of stares and whispers were irritating, but understandable, since such behavior was 'normal' when confronted with something we don't understand and haven't seen before. Dorothy, for example, explained:

I've been in the mall somewhere and I see somebody with Huntington's and I've seen other people staring at them. And it irritated me, but it's not like something that, if I saw somebody with their legs chopped off, I would look and see. You know, it's only normal [to stare]. I didn't think anything of it when they looked at [affected relative], they were just trying to figure out what was the matter with them.

I: That's one of the things that I am interested in, you know. Like, do you think that there's any sort of stigma associated with HD?

Yes at times. But not when my [relative] had it, because everybody around here knew that they were sick. So the only people that made any comments were strangers that were around. But apart from that, no. —Dorothy, tested, intermediate gene

Participants suggested the ignorance surrounding HD contributed to the stigmatizing responses of others. By acknowledging that others were ignorant about HD, participants seemed able to dismiss responses that were potentially stigmatizing and avoid internalizing these negative attitudes.

The most common response to the ignorance surrounding HD was an attempt by participants to educate others about the illness. Nearly every participant suggested they try to explain HD by comparing it to diseases with which others are familiar (e.g., Parkinson's disease). Cheryl's explanation was typical of many participants:

I say it's like if you took Parkinson's and maybe a little bit of Alzheimer's. That's what people understand. People know about Alzheimer's and they know about Parkinson's. They can relate to it better. —Cheryl, at risk

While this sort of explanation often helps end whispers and stares, as Dorothy suggested, it cannot begin to convey the devastating impact of HD on families. Laura explained:

Huntington disease is not just one person. It's everybody. Everybody is affected by it. That's one thing about Huntington disease. If you had cancer, one person has it. This is what people don't know about HD. When people approach me and say, 'What is that?' I try to explain it to them. And they just go, 'Oh yeah.' But they still don't know. They don't know how devastating it is. You got to walk in my shoes to understand. –Laura, caregiver

While participants could forgive the ignorance surrounding HD in generalized social others, they were far less willing to tolerate ignorance in GPs and other professionals (e.g., insurance companies). Many participants in the current research not only lamented the ignorance of GPs about HD, but were also frustrated and angered by it. Cheryl recalled:

How frustrating is it to go...we found it frustrating when we found out [relative] had it and no one knew what it was. My [relative] had to go and do their own research to get a full idea of what it was. Doctors, who you put your life in their hands all the time, can't tell you what it is. –Cheryl, at risk

Shirley similarly commented:

It makes you feel angry and frustrated, you know? I have no medical training whatsoever. I know nothing, you know, in your field at all, and yet I need to know more than they [GPs] do. Try living with it 24/7. –Shirley, caregiver

Summary: Dismissing stigma – ignorant others

Early research on social stigma assumed the stigmatized were victims, suffering any number of ill effects such as low self-esteem or psychological distress (see Crocker et al., 1998, for a review). Later researchers, however, remarked on the resiliency and intact

psychological well-being of a wide number of stigmatized groups (Crocker & Major, 1989). Participant narratives in the current research, whether at risk, tested or affected, also evinced strength and there was little evidence of psychological distress in response to stigma.

Participants were able to dismiss the (sometimes) perceived stigmatizing responses of social others by appeals to the ignorance surrounding HD. In fact, the most common explanation for potentially stigmatizing responses of others was ignorance about HD and the fear this could arouse in others. Participants who had formerly cared for an affected relative acknowledged that reactions such as staring and pointing were perhaps normal when others were confronted with a person affected with HD. Current caregivers, on the other hand, were more often angered by these responses. This is unsurprising since they *currently* face the illness (and others' reactions) every day.

There was no evidence in participants' narratives that they had internalized others' negative attitudes or deemed them applicable to the self. As a result, they appeared to suffer no ill effects on their self-concepts. Rather, any stigmatizing responses they or their relatives received were attributed to the flaws (e.g., ignorance and fear) of those who stigmatize. By far, the most common response to perceived ignorance was an attempt to educate others about HD. All participants, however, thought it was unacceptable for GPs to lack knowledge about the illness.

Beyond coping with stigma, the current research was also interested in coping with genetic risk more generally.

Coping with genetic risk

Minimal research has applied a stress and coping perspective to investigate living with genetic risk. Lazarus and Folkman's (1984) seminal work on stress suggested a stressor is an event in which environmental or internal demands somehow strain or exceed a person's adaptive resources. They suggested that stress occurs only when demands placed upon an individual strain or exceed the individual's coping resources; stress will therefore involve cognitive appraisals about the threat and about the resources the individual has to cope with that threat. The previous chapter demonstrated that genetic risk for HD was experienced as a threat for some participants; although, the salience of risk varied with numerous zones of relevance. Therefore, coping with genetic risk can be thought of as a dynamic process that changes in response to the shifting salience of risk.

The most widely used dimensions of coping are problem- versus emotion-focused coping, primary versus secondary control coping, and engagement (approach) versus disengagement (avoidance) coping. A comprehensive model of stress and coping that organizes most of these dimensions has been proposed by Compas, Connor-Smith, Saltzman, Thomsen, and Wadsworth (2001).

According to Compas et al. (2001), the most basic distinction between stress responses is those which are voluntary or involuntary. This distinction underscores the suggestion that not every response to a stressor constitutes coping. According to Compas et al. (2001), coping refers to "conscious volitional efforts to regulate emotion, thought,

behavior, physiology, and the environment in response to stressful events or circumstances" (p. 89).

The current study did not investigate involuntary, largely physiological, responses to a stressor. Not only would this have required an entirely different research design, it did not fit with the study's primary goal of exploring the subjective meanings of living with genetic risk. Accordingly, the subsequent discussion presents participants' voluntary coping responses to living with genetic risk and/or illness.

Both voluntary and involuntary stress responses can involve engagement or disengagement with the stressor; thus, engagement does not necessarily imply volitional, conscious cognitive processing. Voluntary efforts to engage with a stressor can be further distinguished by the goal of achieving either primary or secondary control. In the former, coping efforts are directed at influencing events or conditions in order to increase a sense of personal control over the stressful situation and one's own reactions to it (Compas et al., 2001). For example, seeking information, generating possible solutions and attempts to regulate emotions (e.g., regulate anger or anxiety about the stressor). Secondary control coping efforts, on the other hand, are aimed at adapting to the situation. In general, one tries to change how one feels about the negative situation. These coping strategies include distraction, acceptance, positive thinking and seeking support from others.

Miller and Kaiser (2001) suggested that secondary control coping is often adaptive; this is especially true for those stressors that are not controllable (e.g., being at

risk for, or having, HD). While research on the adaptive effects of acceptance has been mixed (Miller & Kaiser, 2001), acceptance could be beneficial for those testing positive for the altered HD gene, those who are clinically affected with HD and their caregivers. In these situations, there is nothing the person or caregiver can do to alter the progression of the disease.

Disengagement with the stressor, on the other hand, includes voluntary avoidance, denial or wishful thinking (Compas et al., 2001).

Some of these coping strategies emerged in the current research since living at risk for (or with) HD was a stressor, at least some of the time. Clinical studies of being at risk for HD show increased post-test anxiety and depression in those testing positive for the altered HD gene. Even those testing negative have been found to suffer from numbed emotions and survivor guilt. The nature of HD also limits any efforts of the affected person to alter or impede the progression of the illness. Thus, being at risk for HD can be thought of as a stressful situation. Participant narratives in the last chapter, in particular, provide support for this suggestion.

A recent study argued for a stress and coping perspective to explain adaptation to HD risk (Pakenham, Goodwin, & MacMillian, 2004). In that study, tested and non-tested at-risk person completed a battery of questionnaires measuring numerous stress and coping variables: (1). Adjustment, including depression, global distress and health anxiety; (2). Genetic testing context, including knowledge, contact and experience with HD; (3). Appraisal, including perceived control, threat and self-efficacy; and (4). Coping

strategies, including avoidance, self-blame, wishful thinking, seeking support and problem solving.

Differences emerged between the two groups on some of these measures. For example, non-testees reported lower self-efficacy and control appraisals and higher threat and passive avoidant coping strategies (e.g., avoidance, wishful thinking and self-blame) than testees. In addition, stress and coping variables were related to adjustment to HD risk. In both groups of at-risk person, for example, poorer adjustment was related to (1), higher levels of contact with HD, threat appraisals and passive avoidant coping and (2), lower levels of social support, self-efficacy and control appraisals, and problem solving. Pakenham et al. (2004) argued that a stress and coping model could be usefully applied to study adjustment to HD risk since individual coping style has been identified as a key factor in the decision to take the genetic test for HD (Evers-Kiebooms & Decruyenaere, 1998; and see Chapter 8 of the current research).

The next section presents participants' narratives about coping with genetic risk for HD. Coping strategies are discussed under three themes: (1) Primary control coping, (2) Secondary control coping, and (3) Social comparisons. It is not meant to suggest that these are the only coping strategies available to persons at risk for HD, those testing positive for the altered gene or their caregivers. For example, in the preceding chapter, I argued that as a consequence of the biographical disruption caused by living at risk for HD, participants tried to 'bracket off' (Bury, 1991) their risk in an attempt to maintain

normality. This process, is itself, a coping strategy for living at risk for HD. Victoria's comment was illustrative of the attempt to normalize:

I think I'm pretty much a normal person and am quite active, the same as anyone else. Until that time when it does start, I don't see any need for anything. I think I deal with it fairly well. –Victoria, tested positive

Lazarus and Folkman (1984) suggested there was no single response to a stressor – people will usually try several alternatives, and outcomes will feedback to alter which other coping responses might be made. Participant narratives in the current research were reflective of this variability. No participant relied on one, and only one, coping response to their genetic risk or their family member's risk. Rather, several coping strategies were employed as the salience of genetic risk waxed and waned over the life course.

Primary control coping

As noted, voluntary primary control coping aims to increase a sense of personal control over the stressor and/or one's own reactions to it. In the current research, strategies included (1) seeking information (including having the genetic test), (2) generating possible solutions (including lifestyle practices such as proper diet, exercise, and a positive attitude, and for those affected with HD and their caregivers, finding the proper balance of medications and/or the right physicians) and (3) attempts to regulate emotions (e.g., anxiety).

Attempts to seek out as much information about HD as possible, including having the genetic test, can be viewed as attempts to exert control over being at risk for HD. While at-risk person know there is nothing that can alter their risk or impede disease

progression, seeking information about HD allowed them to be prepared for a potential future of illness. As Cheryl put it:

I do want to know everything. I want to know to prepare myself. If you were going to have surgery, I don't think you would go blind into it. You should know what they're going to do to you. I should know what's going to happen to me and when I should expect it and who it's going to affect and how it's going to affect me, all that kind of stuff. —Cheryl, at risk

Cheryl intends to be tested; as such, her comments were typical of many participants in the current study who had already been tested. Kathleen, for example, who has tested with the intermediate gene said, “I know what’s coming my way. It makes me more prepared for it.” (See Chapter 8 for a detailed discussion of the benefits of knowledge and the use of genetic testing as a way of colonizing the future.)

Readers might wonder just what possible solutions could be generated for the stressor of living at risk for, or with, HD. Even though there is no cure for HD and only limited options for treatment, participant narratives were indicative of attempts at generating possible solutions. [Participants’ thoughts on a cure will be fully addressed later in this chapter.]

For example, participants who had tested positive or intermediate for the altered HD gene suggested that lifestyle practices such as proper diet, exercise, avoiding smoking, limiting or avoiding alcohol and maintaining a positive attitude, including the use of humor, could not hurt their situation. Thus, while these individuals knew there was no ultimate *solution* to their risk of HD (beyond a cure of course), these lifestyle practices

were perceived as at least not being harmful. It is suggested they are an attempt to regain some control over an illness that offers virtually none. Jerry, for example, said:

There's things you can do though, the same for Huntington disease. Like smoking, drinking and stuff like that. I think cutting down on that stuff helps it – you're killing brain cells. So all those things are a step in the positive way, they can't hurt. –Jerry, tested positive

Similarly, Victoria noted:

I really look after myself. I exercise a lot, and I eat well. I take a lot of vitamins. (...) They have found studies – I get the Huntington disease issue of the newsletter quarterly – and, well, they don't have any suitable treatment yet as such, but they are trying out that co-enzyme ten and things like that. They're finding exercise has been just as good as some of these treatments. Of course, those treatments aren't very effective. They think, and I do too, that if you look after yourself, you do have a better chance. –Victoria, tested positive

Thus, while participants acknowledged there was no suitable treatment for HD, lifestyle choices, such as exercising and avoiding alcohol and smoking, were perceived as affording at-risk person a 'better chance.'

Participants also referred to their attempts to regulate how they felt about being at risk for HD - an attempt to exert control over their own reactions to the illness. This coping strategy was understandable: Participants could do nothing to change their risk for HD or alter disease progression. They could, however, attempt to control their own reaction to it. Kathleen, for example, noted how she was initially quite distressed by her intermediate test result, but has learned to regulate her emotions:

First, I would cry and cry and cry everyday. I would have a spurt of crying and it would be on my mind 24/7 more or less. But I learned not to do that now. I don't let myself dwell on it in that sense anymore. –Kathleen, tested, intermediate gene

Participants suggested that having a sense of humor about their situation also helped them cope with their genetic risk:

We can even have a good chuckle about it at times. You have to try to look at it with a sense of humor if you can. Every now and then, something funny will come up. –Victoria, tested positive

This strategy is also available to caregivers and to persons affected with HD.

Marjorie told me:

In the meantime, you have to be funny sometimes. When I go to a meeting, I end up with a joke that sort of lightens things up a bit. –Marjorie, caregiver

Steven recounted a family member's joke that he found funny:

You know what [relative] said when we were diagnosed? They said we needed new 'jeans' for Christmas (laughing). –Steven, affected with HD

Caregivers also cope with HD by trying to ensure their loved one is getting the best possible care, including the best GPs, specialists and medications. This strategy is also employed by persons affected with HD, and it underscores the primary control coping strategies of seeking information and generating possible solutions. Shirley, for example, explained:

If I can keep them comfortable, you know, and do the best I can, get the best medication I can for them. (...) I want one thing, one priority. I'm not asking for a whole bunch of things. I want the meds that they need. These are practical matters. They can be solved easily with a doctor who understands. –Shirley, caregiver

Persons affected with HD also cope with the illness by keeping regular appointments with specialists and by constantly finding the right balance with medication. Steven, for example, told me he has everything he needs right now: He sees

several specialists regularly, although it is a struggle finding the right balance with medications:

I haven't spent a night's sleep in months.

I: Is that because of the Huntington disease?

Yeah. Well, it could be because of the medication. The medication can help ease the chorea. My chorea's gotten worse. I've been on [names drug] for the last couple of months. That's been keeping me awake. I'm on [names drug] now.
—Steven, affected with HD

David similarly suggested:

It is a fine line with the medication because I can up my meds and I can be here zonked out and I am not going to be freaking out about nothing. But then again, you want to participate too. —David, affected with HD

For David, participating in his family life is a priority:

The most important thing for me in my life, on a day to day basis, is how I function with my family. If I can't do that, my quality of life is squat.

David also noted he has found the professional support of specialists (e.g., neuro-psychologists, psychiatrists) helpful in coping with HD and in maintaining a balance in his family life:

I place a real emphasis on making sure people get to see specialists as soon as they can because it was a big help for me. (...) If somebody comes into a GP who has a neurological disorder, it should be automatic that they get a referral to a psychologist. —David, affected with HD

Even though there is currently no ultimate solution to being affected with HD, both David and Steven attempted to exert some control over this stressful situation. For example, by adjusting their medications and keeping regular appointments with healthcare specialists.

Summary: Primary control coping

Primary control coping strategies were employed in the current research as participants attempted to exert some control over the stressful situations of testing positive for the altered or intermediate gene, of being currently affected with HD or of being a caregiver of a person affected with HD. In general, coping efforts were directed at giving oneself (or one's affected relative) the best possible chance in relation to when HD would manifest or in how disrupting the disease currently is to one's life. Thus, some asymptomatic individuals attempted to eat right, exercise and limit alcohol and smoking, while currently affected persons and their caregivers strove to find the right balance with HD medications. Those currently affected with HD also try to make and keep regular appointments with healthcare specialists.

Notably, fewer at risk participants employed primary coping strategies. Rather, they were more likely to engage in secondary coping strategies.

Secondary control coping

Secondary control coping is aimed at adapting to a stressful situation. Generally speaking, one tries to change how one feels about the negative situation. In the current research, the secondary control coping strategies which emerged included: (1) Appeals to luck or fate and/or rationalization, (2) Trust in science, (3) Seeking social support from others, and (4) Acceptance.

Analysis of transcripts revealed that some at risk participants in the current research did not view genetic testing as an instrument for planning their futures (see

Chapter 8), and some referred to their futures as being fated with regard to developing HD, regardless of their actions (see Chapter 9). For some participants, appeals to luck or fate can be seen as a coping mechanism for living at risk. Giddens (1991) has argued that entrusting one's life to fate, ... "relieves the individual of the burden of engagement with an existential situation which might otherwise be chronically disturbing" (p. 133).

Roxanne, for example, noted:

If it's going to happen, it's going to happen, no matter what. I believe that everybody's life is pretty much mapped out. You can make a few changes to it, but you can't take it off track. –Roxanne, at risk

Similarly, Sherri said:

Right now, I feel comfortable not knowing. If I am going to get it, well, I'll get it. –Sherri, at risk

At risk participants were not the only ones to appeal to fate in an effort to cope with living at risk. At least one tested participant also couched the future in the language of fate:

So I was looking at it this way – if I got it, I got it. What are you going to do about it? There's nothing I can do. So I just went on with my day to day things. If it happens, it happens. What are you going to do? (...) I'm a strong believer in... I don't think anything is going to happen to any person not until their number comes. I don't know if that's the right way of believing. I believe in, well, when you get up in the morning, Peter says, 'Well, who's going to be here today at twelve o'clock?' And he says, '[names self].' So, if your number pops up, it doesn't matter what you're doing.

For some participants, then, perceiving the future as a matter of fate allowed them to manage the stress that comes with living at risk for HD. If the future is perceived as

unmanageable and uncontrollable, it makes little sense to participants to worry about what *could* happen.

Participants, both at risk and tested, also engaged in rationalization in an effort to cope with living at risk for HD. In this way, participants tried to adapt to the stressor by minimizing the risk of HD and placing it in the context of all the other risks that exist in life.

Jerry, for example, explained:

You can't dwell on it. I could get sick tomorrow and be diagnosed with cancer, or I could fall down with a heart attack. I could leave here after talking to you and go to the store and be hit by a tractor trailer. That's life. —Jerry, tested positive

Dennis concurred, suggesting:

I might die with cancer before that [being affected by HD]. I could get killed in a car accident before that. I could drown before that. —Dennis, tested positive

At risk participants also appeared to use rationalization to cope with living at risk:

I keep coming back to cancer. I could die from cancer before I die from Huntington's, or anything else. I could get hit by a car tomorrow. —Cheryl, at risk

When the risk of developing HD could be rationalized by comparing it to any risk, participants seemed able to adapt to living at risk, or testing positive for, HD.

Another secondary control coping strategy employed by participants in the current research was trust in science to find a cure. The vast majority of participants did trust that science would find a cure for HD, if not in time for them, then certainly for their children. This trust allowed participants to change how they thought about HD: With the perception of a cure just around the corner, HD could no longer be a fatal disease. Even if

a cure could not be found, there was hope that science would at least close the therapeutic gap that currently exists between testing positive and manifestation of symptoms.

Dorothy commented:

There's been so much research done since [relative] had it, you know? So, yes, I still have hope that there is going to be a cure or at least something that will slow down the process. And now with stem cell research and all this stuff. There's always that little bit of hope. –Dorothy, tested, intermediate gene

Similarly, Dennis suggested:

I think someday they are going to hit it. Well, they found the gene. They're getting closer to a cure. –Dennis, tested positive

At risk participants also showed evidence of trusting science to find a cure that served as a coping mechanism for living at risk. Michelle, for example, said:

I always keep telling myself that by the time I am diagnosed and the chances of my kids being diagnosed with it, there is going to be a cure. I am confident they will have a cure or something to really slow that process down by years by the time I am affected. –Michelle, at risk

Younger at risk participants, while trusting science to find a cure in their lifetime, were skeptical of a cure in their affected relative's lifetime. One said:

They have made so many breakthroughs, so many different things. I'm hopeful for myself, but I can't exactly say I'm hopeful for my [relative]. There's not enough hope and confidence and time, I don't think for my [relative]. –Female, at risk

Thus, while trust in science enabled some participants to engage in positive thinking (a secondary control coping strategy) regarding their own risk for HD, trust did not extend to their relative's situation. This was the case when a relative was nearing the age of onset or was already affected with HD.

While the majority of participants in the current research did trust that science would find a cure for HD, or at least some better treatment, a minority of participants noted they did not have faith in science. Kathleen explained:

No I don't. No, I'm sorry, but I don't. I guess basically, I'm too afraid to hope. I can accept it for myself, but I'm too afraid to hope for my children. I don't want to get my hopes up and say, 'Oh yes, they will find a cure for the children if they do have it,' and then, down the road, there isn't. I don't want to build up my hopes, so I just go with the flow and say maybe there will never be a cure.
—Kathleen, tested, intermediate gene

At least two participants at risk for HD also dismissed science's ability to find a cure. Notably, both of these participants engaged in fatalism as way of coping with genetic risk (see beginning of Secondary Control Coping section).

Roxanne, for example, said:

I don't know. It seems like things have a way of changing themselves. If they find a cure, it changes itself and mutates enough that...like, they came up with a cure for the common cold, they give everybody flu shots. Then, that doesn't work because the flu mutates so much, there's no way to keep track of it all. By the time they defeat one strain, they pretty much changes it and you end up with something new.

I: That the flu shot doesn't work for then? So, do you think that might happen with Huntington disease?

Yeah, it's the same with cancer. It would be great if they could come up with a cure. If they did...it's God nature that everything has to die sooner or later and there got to be a way for it to die. I think that's just God's way of saying, 'Look, no matter what you do, it's always going to be there.' —Roxanne, at risk

Thus, while the majority of participants had hope in science and research into HD, a minority of participants were either too afraid to hope for a cure for their children or skeptical that a cure could be found at all.

A third secondary control coping strategy evident in participant narratives was seeking social support. This was most evident in the narratives of those who attended support groups for those affected with HD and caregivers of persons affected with HD. And, at least one tested participant was a member of an online support group. Their narratives were similar in that they all suggested it was important to know how others were coping with the same issues. The sample only included two individuals who were currently affected with HD, and one of those did attend support group meetings with fellow HD sufferers. He told me the meetings were "wonderful" since others knew exactly what he was "dealing with."

In general, participants wanted to know they were not alone and were curious about how other families found out about HD, how they were experiencing the disease, and importantly, how they were coping. Kathleen's comment was typical:

It was really nice sitting down and to know that these people feel the same way I do. They are hurting the same as I am. At the time, you think you are the only one who's hurting. (...) It was so good to get together and know that there were more people like you out there. —Kathleen, tested, intermediate gene

Some at risk participants lamented the lack of social support for families affected by HD. When I asked if she had any suggestions for healthcare professionals, Serena, for example, commented:

I guess support is the main thing. I think that is the main thing that you need when you're going through something like that. You have people out there in the same situation that you're in. You know that they know what you're going through. That's one thing that really helps. They understand. You can talk to them and they understand what you're going through. —Serena, at risk

Of course, seeking social support is not a panacea for everyone at risk for HD. Other at risk participants did not currently feel the need for social support, fearing it could make their risk salient. They did, however, suggest that such support would have been beneficial when they were caring for an affected relative. One commented:

I think if I was attending support groups and stuff, it would play on my mind more because I would be more actively involved in things. (...) It would have been [beneficial] earlier in life when I was looking out to [affected relative] and ran into a lot of problems of different things happening. –Female, at risk

Finally, at least two participants spontaneously suggested that professional social support is essential, but seriously lacking, for *children* of families affected by HD. This finding was notable since it was not directly queried during the interview. David, for example, suggested:

I think that people don't know that kids need counseling too. They are living in a verbally abusive environment. Children need it too. –David, affected with HD

Brenda also suggested there was a need for such support. She was concerned for her at risk nieces and nephews:

There's not enough of the social workers dealing with these cases to be able to come and see somebody. Every six months, or once a year? That's not much. (...) I think there needs to be more support for the children. Like I said, growing up in a home with a parent with Huntington's, you know, we didn't know what it was. Now these children know what it is. But things go on and there has to be stuff going on in their minds and wondering, 'Do I have it?' 'Don't I have it?' And do they really have the support to talk about it at that age? –Brenda, at risk

For some individuals affected by HD, social support was perceived as an important coping mechanism for children of families affected by the illness.

The final secondary control coping strategy that emerged in participant narratives was acceptance; although, this was the least frequent strategy in the current research and was normally used in conjunction with some of the other strategies outlined in previous sections. Kathleen, for example, suggested:

I don't let myself dwell on it anymore. I can't change it. It's something I have to live with. I have to realize that I have to make the best of what I've got left. I can't be...give up my life because I have the gene for HD. –Kathleen, tested, intermediate gene

Shirley suggested that as a caregiver, HD simply becomes part of your life:

At first, it's a very frightening thing, but after a while, it sort of, you know, it doesn't sound very nice, but it's a lifestyle. –Shirley, caregiver

While initial discovery of HD in the family is often a scary, confusing experience, with time often comes acceptance.

Summary: Secondary control coping

A number of secondary control coping strategies were employed by participants in the current research as they coped with living at risk for, or with, HD. Some at risk for HD used a fatalistic discourse when talking about their futures (e.g., 'if I get it, I'll get it'; 'there's nothing you can do'). If the future was perceived as fated, it allowed at-risk person to manage the stress of being at risk for HD since the outcome was out of their control. Additionally, both tested and at risk participants engaged in rationalization – an attempt to minimize the risk of HD by comparing it to any of the other risks that exist in life.

The majority of participants trusted science to find a cure for HD, or at the very least, some treatment that could slow down disease progression. This trust allowed participants to change the way they viewed HD. If there is a cure, or better treatment, HD is no longer fatal, and as such, less frightening. Those who did not evince great trust in science were more likely to be those who engaged in fatalistic thoughts about their futures. Some participants also coped with HD by seeking support from others; this was especially pronounced for caregivers of persons affected with HD. Finally, a minority of participants demonstrated acceptance of their risk for HD. These secondary control coping strategies were not mutually exclusive; as with primary control coping, several secondary strategies were employed by participants in the current research.

Social comparisons

Festinger's (1954) social comparison theory holds that we learn about our own abilities and opinions by comparing ourselves to others. That is, we use others in our social environments for self-evaluation. Since Festinger's original 'information value' conceptualization, research has detected a clear motivational function behind social comparison. Social comparisons do provide valuable information about the self; however, they are also key determinants of affect and self-esteem. While Festinger suggested that comparison choice would likely be oriented toward superior others, a robust body of work suggests that positive affect and self-esteem can be *impaired* by unfavorable comparisons of valued attitudes or outcomes with others (upward comparisons) and enhanced by favorable comparisons with others (downward comparisons) (Crocker et al.,

1998). Comparison of the self to others who are thought to be doing better can be threatening. This is especially likely for attributes over which one has little or no personal control (Crocker et al., 1998), such as being at risk for, or having, HD.

In his influential downward social comparison theory, Wills (1981) argued that in contexts that produce a decrease in well-being (e.g., illness), people will compare themselves to others perceived to be worse off in an effort to increase their well-being, especially when instrumental action is not possible – the notable context of HD. In pioneering research with women with breast cancer, Taylor, Wood, and Lichtman (1983) observed that women always commented on others who were worse off, despite the severity of their own illness. Further, if no social comparison other was available, the women imagined or fabricated one.

The past two decades have confirmed the prevalence and beneficial effects of downward comparisons for a variety of illnesses including chronic pain, arthritis and cancer (see Tenne, McKee-Iberhardt, & Affleck, 2000, for a review).

It was thought that social comparisons would be observed in the current study. Recent research confirms that stress and uncertainty do tend to promote social comparisons with similar others, and underscoring this, it is in the area of health psychology that social comparison research has seen a revival in recent years (Buunk, Gibbens, & Visser, 2002). Many participants did not discover the family history of HD until later in life. It represented a new and often unexpected situation (see Chapter 7) that seemed conducive to social comparison processes. A recent study found anxiety,

frustration and lack of control over illness were all associated with a need for social comparison information (e.g., information regarding how similar others were doing, feeling and coping; Buunk, 1995). These attributes can characterize the situation of being at risk for, or clinically affected with, HD (at least for some individuals).

As noted, a majority of social comparison research within health psychology has focused on downward social comparisons, particularly to in-group members. A recent study of people with schizophrenia, however, observed upward, downward and lateral comparisons (the latter two being more frequent; Finlay, Dinos, & Lyons, 2001). In addition, comparison targets were not always in-group members (e.g., other schizophrenics). Frequently, comparison targets were an unspecified, general target (e.g., everybody, other people). Finlay et al. (2001) found that people chose a wide range of attributes (not solely having schizophrenia) and target others when making comparisons; in addition, positive representations of the self were more frequent than negative. In most instances of lateral comparison, their participants saw themselves as similar to a range of other people, on a range of other attributes. Finlay et al. (2001) cautioned social researchers that the stigmatized belong to a multitude of identities and have multiple comparison targets available to them.

The multiple identities of participants with schizophrenia in their study prompted Finlay et al. (2001) to conclude, "Although the identity [having schizophrenia] was found to be salient in some contexts and to have negative consequences, the findings reported here should temper stigma theories which often imply that such identities are chronically

salient, are overwhelmingly negative, and are best defined in opposition to the non-stigmatized majority” (p. 590). This admonition proved quite relevant to the current study. The current research did not attempt to probe for social comparisons *per se*; however, participants spontaneously engaged in social comparisons when asked about how they were dealing with the illness (or being at risk).

Victoria’s comment earlier in this chapter, for example, was a typical example of lateral comparisons of the self to generalized others. “I think I’m pretty much a normal person and am quite active, the same as anyone else.” Like Finlay et al.’s (2001) participants, participants in the current research compared themselves to generalized others on a wide range of attributes, not just being healthy or ill. Additionally, positive representations of the self were much more common than negative portrayals. Most participants in the current research believed they were coping well with their risk for HD.

While there were many such comparisons observed in the current study, here I wish to highlight the type of comparison most pronounced in participant narratives – comparisons to affected relatives.

If I can be like Mom...

The majority of participants in the current research seemed to actively compare themselves to their affected relative (who was often deceased), specifically remarking on their relative’s age of onset. These comparisons seemed to be an effective coping mechanism for both at risk and tested participants. Jerry, for example, remarked:

If I look at my odds and if I look at my [parent], if I could live as long as they did, with a normal life – before the sickness I mean – and do the things that they did, it wouldn't be all bad I suppose. –Jerry, tested positive

Similarly, at risk participants compared themselves to their affected relatives.

Julie, for example, explained how her parent's very late age of onset helped her cope with her own risk:

If I can say in my mind I am going to be like [parent] and I am not going to show obvious symptoms until I am [age], well, that gets me through the next period relatively easy. –Julie, at risk

Roxanne also actively compared her risk to her affected relative's experience; the comparison allowed her to cope with the possibility of HD in the distant future:

I think about the fact that [relative] was old. They were in their [age] before they even found out they had it. So, I mean, in that respect, I think to myself, I can still live a good, healthy normal life. –Roxanne, at risk

For tested and at risk participants, comparisons to affected relatives were actively undertaken in an effort to cope with personal genetic risk, underscoring (once again) the influence of the familial context on the experience of living with genetic risk/illness.

Summary: Social Comparisons

Participant narratives in the current research were replete with social comparisons. However, these comparisons were not exclusively downward to 'worse off' individuals or families affected by HD. Although, this did occur. For example, some caregivers noted how much more difficult their situation would be if they had to care for two or more individuals affected with HD.

Lateral comparisons were common in the current research. At risk and tested participants compared themselves to a range of generalized others, on a range of attributes – not just being at risk for (or having) HD. Participants saw themselves as ‘normal’ and ‘just like anybody else.’ In this section, however, I highlighted the type of comparison that seemed most effective for participants in coping with their risk of HD – comparisons to affected relatives. For most participants in the current research, age of onset in an affected relative was later in life (the fifties, sixties, or even the seventies). Participants noted that if they did not manifest symptoms of HD until that age, they could still live a normal, healthy life, and in Jerry’s words, it “wouldn’t be all bad.”

A final note on coping with genetic risk

The preceding sections highlighted some of the coping strategies employed by participants in the current research. Most of these were engaged, voluntary coping responses (Compas et al., 2001). However, a note on disengagement coping strategies is in order. Compas et al. (2001) suggested that voluntary avoidance, denial and wishful thinking were all suggestive of disengagement with a stressor. In the current research, there was no evidence of wishful thinking in any participant narrative. And, as I will argue shortly, little evidence of denial of genetic risk. At least two participants, however, evinced voluntary avoidance of genetic risk for HD. Serena, for example, suggested she actively tries to suppress thoughts about her risk:

Yes. Everyday, yes, I can say you do think about it. Then, you sort of say to yourself, ‘No, block it from your mind.’ –Serena, at risk

She also remarked that, as a family, she and her siblings sometimes do not discuss the family history of HD since it is depressing:

We don't talk about it sometimes. When you do, it is depressing. It is depressing and I guess that's why you keep it and don't discuss it.

Dorothy suggested she no longer worried about personal risk, but does worry for her other relatives who remain at risk. Despite trying to 'block' the worry, there are times it simply cannot be avoided:

I do worry about it. I try not to think about it most of the time. I block it out. But there are times when it comes out strong in your mind that you have no other choice but to think about it. —Dorothy, tested, intermediate gene

Discussion

This chapter completes the story of living with genetic risk that began in Chapter 7 with the discussion of discovering the family history of HD. Here, I presented participants' perceptions of, and experiences with, the stigma associated with HD. I also highlighted participant coping strategies, not only for coping with stigma, but with genetic risk/illness more generally.

The majority of participants suggested that sympathy, not stigma, was the dominant response to them and/or their affected relative, supporting research suggesting that those *not* held responsible for their illness elicit sympathy and willingness to help (e.g., Menec & Perry, 1998). Sympathetic responses were likely when others understood HD or *after* participants had explained HD to others. As several participants suggested, it was normally when others understood that HD was a genetic disease over which their affected relative had no control that others' responses were sympathetic.

Despite sympathy, perceived stigma did exist in relation to HD. Scrambler's (1998) distinction between felt (i.e., future) and enacted (i.e., actual) stigma proved relevant to participant experiences. Some at risk and tested participants generated felt stigma in acknowledging the possibility of *future* stigma. Participants who tested positive or intermediate, for example, recognized that while they were currently 'normal,' the perception of normality could be threatened when HD symptoms began. Some at risk participants also acknowledged the potential for stigma, notably in insurance and employment contexts.

Scrambler's work with epileptics (1998; Scrambler & Hopkins, 1988) revealed that felt stigma could be more disruptive to people's lives than actual stigma. For example, as a result of acute felt stigma, epileptics concealed the illness and adopted a policy of nondisclosure to most people. Some participants in the current research adopted a similar policy; although, this was normally in very specific contexts (e.g., employment or insurance).

Contrary to Scrambler (1998), there was little evidence that felt stigma was overly disruptive or worrisome for at risk and tested participants in the current research. Participant narratives also challenge other research suggesting that concealable stigmas generate anxiety and depression (e.g., Frable et al., 1998). All participants in the current research indicated that being at risk for HD was completely out of their control. It was this dimension of a stigmatizing condition that seemed to mitigate any potential negative effects of stigma on participants in the current research. This finding supports Crocker et

al.'s (1998) suggestion that controllability is a critically important stigma dimension for understanding the subjective experience of stigma. Since the illness is beyond their control, participants could dismiss any negative responses from others as not valid to the self.

Caregivers were more likely to recount instances of actual (i.e., enacted) stigma. This is perhaps unsurprising since illness severity has been shown to be an important component of stigma (Crandall & Moriarty, 1995). HD is a progressive, severe condition having pronounced symptoms that interfere with daily life and social interactions. Crandall and Moriarty (1995) found that illness severity consistently predicted social rejection. Accordingly, caregivers of persons with a severe illness such as HD did recount instances of social rejection, particularly from strangers. They were also more likely than any other participant to be angry and/or frustrated by these negative responses.

Their response arose in part because of their need (and perceived obligation) to protect their affected relative. However, social identity theory can also help explain their reaction.

From the perspective of social identification, people are not viewed as individuals, but as members of social categories. All caregivers in the current research strongly identified with being a caregiver of a person affected with HD. Each narrative was replete with discourse reflective of this identity. Deaux and Ethier (1998) suggested that in the face of a threat to one's identity, identity negation or enhancement strategies

were likely. Further, these strategies are most likely for those who identify strongly with their group (as caregivers clearly did in the current research).

No caregiver seemed to engage in identity negation strategies, aimed at dissociating oneself from a social identity that is aversive or non-satisfying. All did, however, engage in identity enhancement strategies. That is, they attempted to assert the already existing identity of caregiver of a person affected with HD. In fact, during the support group meeting I attended, caregivers were quick to inform me that caring for a person affected with HD was not the same thing as being a caregiver *per se*.

Deaux and Ethier (1998) outlined several enhancement strategies, of which two were observed in the current research, re-mooring and social change. Re-mooring strategies involve active behavioral involvement. Caregivers sought information about HD and HD support groups, associated with new group members, and actively took part in a monthly support group meeting. They also showed evidence of social change when they attempted to change the beliefs that others held about persons affected with HD. Every caregiver in the current research actively tried to explain the illness to others; one had even taken part in a media interview to help raise public awareness of HD and to counteract the mistaken impression that HD sufferers are somehow 'mental,' 'contagious' or at fault for the illness.

Despite the experiences of felt and enacted stigma in the current research, there was little evidence that participants suffered pronounced ill effects on their self esteem or life satisfaction. As noted, participants suggested they could not be blamed for their risk

(or illness), and if others' reactions were negative, it was because they simply didn't understand HD. Thus, while participants were aware of the negative views of HD that could be held by others (e.g., associations with mental illness or alcoholism), there was no evidence they viewed them as valid. Nor did they seem to internalize these attitudes or deem them applicable to the self. Like the women with mental health problems in Camp et al.'s (2002) research, they appeared to suffer no ill effects on their self-concepts.

Nonetheless, caregivers could be seen to have constraints on their behavior as a result of the negative reactions of others to their affected relative. For example, they were selective in their communication about their relative's condition and in public outings.

All participants in the current research coped with perceived stigmatizing reactions from others by attempting to educate the other about genetic risk for HD and/or the illness itself. Response to stigma was fairly homogenous across study participants. Broadly speaking, coping with genetic risk for HD, however, was more variable.

Coping appeared to be a dynamic process – no participant relied on a single coping strategy. Instead, numerous coping responses were employed by participants, and nearly all suggested they were coping well with their risk or illness. The study's sample could have contributed to the positive perceptions of coping ability. Most participants had known about their risk for some time, and an average of six years had passed since taking the genetic test (*Range*: 0-15 years). Only one participant had known about his/her risk for less than a year; this narrative showed the least evidence of perceived positive coping

ability. Understandably, coping takes time, and I suspect this participant had not had enough time to fully process the news of his/her risk at the time of our interview.

Both primary and secondary control coping strategies, in addition to social comparisons, were observed in participant narratives. Primary control coping included seeking information about genetic risk and HD (e.g., having the genetic test), generating possible solutions (e.g., proper diet, limiting alcohol) and attempts to regulate emotions (e.g., anxiety).

Wilkinson (2001a) described two constructions of risk that he suggested were related differentially to anxiety. In the first, 'risk' is an outcome that is largely unwanted and incalculable. The future is perceived as containing any number of unknown dangers or hazards. This is the construction of risk that permeates Beck's (1992) 'risk society.' According to this view, uncertainty about these future dangers (i.e., risk awareness) has become a major source of anxiety in modern Western society.

The second and contrasting usage of 'risk,' however, is related to certainty. It is this usage that underlies genetic testing as a primary control coping strategy. In this construction, knowledge of risk is seen as permitting one to predict the likelihood of future events, and by implication, suggests a degree of control over the future (Hallowell et al., 2004). In this sense, risk information is viewed as beneficial since it can inform life choices. Wilkinson (2001a) argued that risk calculation can therefore, "...allay anxiety by making clear the proper dimensions of an anticipated danger so that it can be faced as manageable fear" (p. 44). Thus, risk awareness can be seen as a coping mechanism for

living with genetic risk, and the narratives of tested participants revealed this coping strategy.

At risk participants, on the other hand, were less likely to talk about the benefits of knowing the future. Rather, they preferred to 'take it one day at a time.' As Roxanne put it, "I try not to worry about what's going to happen in the future. I try to take care of what I can today and worry about everything else when it comes." This view was also held by participants affected with HD and caregivers, and it supports recent research in Australia that comprised those affected with HD and their caregivers (Dawson, Kristjanson, Toye, & Flett, 2004). In that study, participants reported they 'take each day as it comes' (p. 126).

Tested participants, affected persons and caregivers also coped by trying to generate possible solutions. Several participants spoke about lifestyle choices that were in their control such as exercise, proper diet, stress reduction and maintaining a sense of humor. Even though all participants readily acknowledged these practices could not change their risk/illness, they believed they were at least worth doing, or as Victoria put it, gave them a 'better chance.' Affected participants and their caregivers also worked to ensure the former have the best possible care (e.g., medications and specialists).

Primary control coping strategies such as problem solving have been associated with better adjustment in a wide range of chronic health problems (e.g., Pakenham, 1999), including HD (Pakenham et al., 2004). Poorer adjustment, on the other hand, has been related to passive avoidant strategies such as wishful thinking, self-blame and

avoidance. Few participants spoke about self-blame; although several did comment on blaming themselves for having possibly passed on the altered HD gene to their children. Even these participants, however, acknowledged the fault did not lie with them since they had no knowledge of their family history of HD at the time of reproduction.

Additionally, there was no evidence of wishful thinking in participant narratives. Some at risk narratives did reveal voluntary avoidance as participants actively tried to stop thinking about their risk. At least one participant suggested she and her siblings sometimes did not discuss the family history of HD since it was 'depressing.' In the context of their interviews with women at risk for breast cancer, Kenen et al. (2004) suggested that restricted communication about cancer in the family provides a means of distancing at-risk individuals from their own increased risk.

Despite some voluntary avoidance, however, no apparent differences emerged in participant perceptions of how well they were coping with their risk. This corresponds to Pakenham et al.'s (2004) lack of differences between those at risk and those tested for HD on a variety of adjustment measures (e.g., global distress, depression, health anxiety and social role functioning). Similarities between at risk and tested persons with respect to perceived adjustment are consistent with the suggestion that some people at risk for HD cope well with their increased genetic risk and do not need a predictive test (Evers-Kiebooms et al., 2000). Additionally, study participants who revealed avoidant coping also relied on other coping strategies; these could have mitigated any negative effects of avoidant coping.

For example, secondary control coping strategies included appeals to luck or fate, rationalization, seeking social support, trusting science and acceptance of genetic risk/illness. It has been suggested that secondary coping strategies are often adaptive, particularly for stressors that are beyond one's control – the notable context of living at risk for HD.

At risk participants were more likely than tested participants to construct their futures as determined by the vagaries of luck and fate, rather than genetics. Similar findings were reported with women at risk for breast cancer (Hallowell et al., 2004). In the current research, comments such as 'if I get it, I'll get it' were typical of this coping strategy. Giddens (1991) suggested that people turn to fate as a means of explanation when they face risks over which they have little or no control. Some at risk participants in the current study perceived their futures as uncontrollable. By suggesting they would face whatever the future might bring, they were able to manage their fears about a future they could not change, but that might include HD.

It could be argued that appeals to fate represented a form of denial of genetic risk (Hallowell et al., 2004). This seemed unlikely in the current research. Every participant acknowledged the implications of his/her own risk status for children and/or other family members (e.g., siblings). And, some non-tested participants could see the utility of genetic testing for *other* at-risk individuals, suggesting they dismissed, rather than denied, their personal risk.

Rationalization was also common in participant narratives, whether tested or at risk. It is suggested that rationalizing genetic risk for HD is an effective coping strategy that allows people to change how they feel or think about risk/illness. Specifically, rationalization minimizes risk for HD in comparison to all the other risks that exist in life.

Trusting science to find a cure for HD can also be an adaptive coping strategy. If a cure is found, HD is no longer a fatal disease and is perhaps less anxiety-provoking. Kenen et al. (2003b) also observed great trust in science and medicine in the narratives of women at risk for breast cancer. Trust in science represented a secondary control coping response for many participants in the current study, whether at risk, tested, affected or caregivers. This finding challenges Beck's (1992) contention that risk society is marked by *mistrust* of science.

According to Beck (1992; 1995), increased reflexivity has led to increasing individualisation, de-traditionalisation and a heightened risk awareness. Risk society is accompanied by a corresponding erosion of trust in risk 'experts' (e.g., scientists, medical professionals, government, etc.). Horlick-Jones (2004) has argued that Beck's (1992) treatise does not capture the complexity of lay assessment of risk and trust in risk sources. For example, he argued that parents with a health decision to make are concerned only about the effect on *their* child, not a population of children. Therefore, even if risk projections or judgements are associated with objectivity and authority, expertise about risk tends to be viewed as incomplete: Risk assessments fail to capture the quality of risk (Horlick-Jones, 2004). That is, people want to know about *their or their children's risk*,

rather than global risks per se. Horlick-Jones (2004) contended that, "In the same way as there are severe doubts about the extent to which a 'golden age' existed when lay people had implicit trust in governments, so the authority of traditional expertise has always, it seems, been tempered by the specificity of situations" (p. 111).

The situation in which individuals at risk for, or affected with, HD find themselves is conducive to trust. In the current research, many participants perceived little choice but trust in science since their situation affords them little control. There is nothing at-risk individuals can do to change their risk for HD; science and medicine were perceived as the only hope.

Secondary control coping was also observed in the current research as participants actively sought social support from others affected with HD. This strategy was particularly likely for those affected with HD and their caregivers. Although, some tested participants also confirmed that contact with similar social others would be beneficial, if not now, than in the future when HD symptomatology began.

Very few studies have examined social support in persons at risk for HD; although, higher levels of social support have been shown to be related to better adjustment for a wide range of health threats (Pakenham, 1999). Specific to HD, Tibben et al.(1993) found social support to be related to better adjustment following receipt of the test result. Pakenham et al. (2004) observed social support was related to better adjustment across several domains (e.g., global distress, depression and health anxiety) for both at risk and tested persons. Genetic testing protocols imply the importance of

social support in the encouragement given to test candidates to have someone accompany them throughout the testing process.

In the current research, participants wanted to know that they were not alone and were curious about how other families found out about HD and how they were coping. This finding corresponds with a study of (ex-) cancer patients who indicated the need for social comparison information and contact (Bennenbroek, Buunk, van der Zee, & Grol, 2002). Several variables were found to be associated with the need for social comparison information: The more patients evaluated their own health negatively and the higher the level of anxiety, uncertainty and depressive symptoms the patients experienced, the greater the need for social comparison information. The best predictor of the need for social comparison information was patients' evaluation of their own health. These findings, and those of the current study, have practical implications. A readily apparent implication lies in patient education and information. Patients and their family members want information about their diseases, especially serious ones like HD. It would appear they also want social comparison information, especially about those who are coping well with the illness. In light of their findings, Bennenbroek et al. (2002) recommended that physicians provide patients with information about support groups in their area. That suggestion seems applicable to the current research as well. Some participants did indicate the need for support, especially from others who were "going through the same thing." Some research has shown that patients can be unaware of such support groups (e.g., Eakin & Strycker, 2001), and participants in the current study confirmed this for

HD support as well. A recent study found that support groups for people affected with HD and their caregivers were perceived as beneficial, both for exchanging practical information about the disease and for bolstering the spirit (Dawson et al., 2004).

These findings confirm the importance of social comparisons in adjusting to genetic risk for HD and the illness itself. In the current research, comparisons to other families affected by HD seemed most adaptive for caregivers. In this way, they could exchange practical advice and encouragement. Interestingly, participants who had *formerly* cared for a person affected with HD spontaneously suggested that such social support and social comparison information would have been very beneficial at the time.

The most common type of comparison in the current research, however, was comparisons to affected relatives. Both tested and at risk participants employed this coping strategy to live with their increased risk. Those who had tested positive or with the intermediate gene, for example, compared themselves to their deceased relative. Since the age of onset was late, participants suggested they could still live a fairly long, normal, healthy life before (or if) HD manifested. At risk participants also employed this coping strategy. If the age of onset was early in an affected relative and at risk participants had passed that age, this comparison also seemed to serve as an effective coping strategy for living at risk.

CHAPTER 11

SUMMING UP – CONCLUSIONS AND IMPLICATIONS

The current research explored the stories of individuals affected in some way by HD. It represents an alternative to mainstream clinical research on HD in its phenomenological approach. As such, the central research questions that comprised this work revolved around meanings: What does it mean to live at risk for HD? What does it mean to be affected with the illness? What does it mean to take the genetic test for HD? In this final chapter, I summarize the findings of the current research, emphasizing the most central conclusions. In so doing, I discuss the theoretical and clinical implications of the main findings. I also describe some of the limitations of the research design and the study sample. Finally, I offer directions for future research.

Situating this research

When biomedicine offers no cure and only limited options for treatment, predictive genetic testing is about risk and the management of uncertainty (Cox, 1999). Both concepts have garnered considerable attention, not only in how we talk about health and illness, but also in modernity more generally (e.g., Beck, 1992; Giddens, 1991; Lupton, 1999a,b). As Wexler (1995) noted, however, predictive genetic testing creates a 'third space' that exists outside the boundaries of conventional talk about health and illness. Participants such as Cheryl, Michelle and Serena currently inhabit this space, and this dissertation has been an attempt to understand what that might mean. The proliferation of new genetic knowledge and technology (Bell, 1998) suggests that

increasing numbers could inhabit this 'third space.' It is within the context of efforts to chart this new terrain that I situate the findings of the current research.

Study findings and implications

Presently, there exists a rather large literature on HD and predictive genetic testing for a variety of adult-onset disorders, notably hereditary cancers. With few exceptions (e.g., Chapman, 2002; Cox, 1999; 2003; Taylor, 2004), much of this literature presents quantitative findings of studies undertaken by service providers in clinical settings (Lerman, 1997). This work serves an important evaluative function for genetic testing protocols and helps identify subgroups of the testing population that could benefit from psychological support. However, this work largely ignores the vast array of factors that both shape and differentiate the experience of predictive testing for HD (Cox, 1999) and the experience of living at risk for the illness. Following Cox (1999), the current investigation sought to break from mainstream research on HD in order to explore the meaning and salience of genetic risk for HD within the context of everyday life.

Such an approach is needed if we are to account for the perspectives of at-risk individuals in policies and procedures related to genetic testing (Cox, 1999; Kerr et al., 1998). Further, decreasing economic resources for health care will likely constrain existing levels of counseling and support available to at-risk person. Therefore, it is important to investigate how a variety of non-clinical factors (e.g., initial discovery or personal experience with HD) shape the experience of living with genetic risk.

Living genetic risk every day

Conrad (1990) argued that understanding the subjective meaning and experience of illness could only be attempted if the inquiry was grounded in the sufferer's world. One of the strengths of this research was its focus on the everyday lived reality of participants, rather than the clinical setting in which genetic risk for HD is salient (cf. Cox, 1999). People at risk for HD do not think about and discuss risk solely at the genetics clinic; indeed, sometimes they do not think about it at all. Further, some at-risk individuals who decline testing *never* enter the genetics clinic, as the stories of Serena, Roxanne and Michelle illustrated. These findings suggest that researchers must move beyond the clinical setting in which the majority of genetics research is located in order to explore how hereditary risk is understood and lived when at-risk person are at home, at work, or doing whatever they do when they are not 'patients' (Conrad, 1990; Cox, 1999).

Study participants drew from a wide range of life experiences, events and memories that differed from those typically told in a clinical setting. In particular, participants' stories were a testimony to the fact that there is, at this particular moment in history, no prototypical HD family or predictive testing experience (Cox, 1999). This finding has both clinical and research implications. It suggests that healthcare professionals who work with HD families cannot assume that a single medical protocol (e.g., for genetic testing, counseling or other healthcare referrals) will be appropriate for *every* individual at risk for HD. Rather, tailored, individualized healthcare could be necessary to address the wide array of factors that shape the experience of being at risk

for HD, the experience of undergoing predictive testing and the experience of actually having the illness.

This suggestion is supported by findings from an interview study with persons currently affected with HD and their caregivers (Dawson et al., 2004). In that study, participants suggested that both practical support and care services should be flexible and individualized for HD families, including in-home support, constant respite workers and varying levels of short- and long-term residential care. It is notable that several at risk participants in the current research also lamented the lack of appropriate practical and psychological support available to them while they were caring for a (now deceased) relative. Current caregivers, such as Laura and Shirley, expressed similar concerns.

Other participants suggested the need for psychological support for children of HD families which could have implications for genetic counseling protocols. World guidelines for HD testing (International Huntington Association and World Federation of Neurology, 1994) suggest that at-risk person be 18 years of age before proceeding with testing. While this seems a reasonable and protective conclusion, it fails to address the needs of at risk children under the age of 18. In the current research, for example, both David and Brenda suggested that their children and/or nieces and nephews could benefit from psychological support provided by counselors or social workers.

The variability in participants' experiences also has research implications. It suggests that isolated variables (e.g., test outcome) are not independent constructs with some single effect (e.g., morbidity) on the experience of living at risk. As this dissertation

illustrated, a host of variables such as stage in the life course, experience with HD and unique life events all shaped the experience of living at risk for HD (see Figure 3, Chapter 9). These variables were not independent of each other; rather, they interacted to influence risk saliency, and the importance of each will wax and wane over the at risk experience. Therefore, test outcome is not the only, nor even the most important, independent variable with a range of measurable effects in the life-world of a person at risk for HD.

The hermeneutical approach of this dissertation has broadened our understanding of what it means to be at risk for HD and what it means to undergo predictive testing for a fatal illness. The central data chapters explored how participants described and narrated their experiences of: 1) discovering the family history of HD, 2) deciding to request or decline the genetic test, 3) living at risk for, or with, HD, and 4) social stigma. Some of these research questions have been relatively neglected in the literature (e.g., initial discovery, social stigma). Table 8 summarizes the primary themes and subthemes arising from analysis of participant interviews. These were the findings from which the main conclusions and implications of the study were drawn.

Table 8. *Summary of interview themes and subthemes*

<p>Discovering HD in the family</p> <ul style="list-style-type: none"> ▪ 'Something' is wrong ▪ Out of the blue ▪ Knowing, but dismissing ▪ Growing up with HD 	<p>What does living at risk mean?</p> <ul style="list-style-type: none"> ▪ Risk as threat ▪ Risk as responsibility ▪ Risk as biographical disruption ▪ Risk saliency
<p>Genetic test decisions – How are they taken?</p> <ul style="list-style-type: none"> ▪ No decision to be made ▪ Constrained decisions ▪ Re-evaluating the decision ▪ Test triggers 	<p>Stigma about HD</p> <ul style="list-style-type: none"> ▪ Sympathy, not stigma ▪ There's not really a stigma, but... ▪ Enacted stigma ▪ Dismissing stigma – ignorant others
<p>Genetic test decisions – Why are they taken?</p> <ul style="list-style-type: none"> ▪ Stage in the life course ▪ Emotions ▪ Personality (e.g., monitors/blunters) ▪ Responsibility to others ▪ Belief in the benefits of knowledge 	<p>Coping with genetic risk/illness</p> <ul style="list-style-type: none"> ▪ Normalizing ▪ Primary control coping ▪ Secondary control coping ▪ Social comparisons to affected relatives

Finding out about HD

Participants' accounts of discovering their family history of HD were varied.

Awareness of the family history was often gradual, yet at other times, marked. It occurred

when a mysterious 'something' was finally diagnosed in a relative, as in Michelle's and Serena's stories of '*something*' is wrong. Alternatively, for participants like Gerald and Lori, awareness can come *out of the blue* when a socially or geographically-distant relative is suddenly found to have HD. For others, a vague awareness that HD was 'in' the family existed, but it was not until an immediate family member was diagnosed that the transition into initial awareness began, as in Julie's and Steven's stories of *knowing, but dismissing*. Finally, awareness of the family history could be just one more taken-for-granted element in the complex backdrop of family life, as in Brenda's and Tony's narratives of *growing up with HD*.

How at-risk individuals initially discovered the family history of HD has been relatively unexplored in the literature (for a notable exception, see Cox, 1999). However, an appreciation for the transition into initial discovery is significant for several reasons.

No participant recalled hearing about HD in any other context prior to discovering his/her own family history. Ignorance about HD was, *by far*, the most salient feature shaping narratives of discovery in the current research. Misdiagnosis and/or an undocumented family history were prominent in many participant stories and appeared to be significant factors shaping families' awareness of HD. It is notable that when asked if they had any suggestions for healthcare professionals regarding care provision to HD families, the most frequent and passionate suggestion revolved around destroying the perceived ignorance about HD. The perceived ignorance surrounding HD raises numerous research and clinical considerations.

From a research perspective, this perception underscores the importance of investigating whether (or how) recent advances in genetic knowledge and technology have translated into a corresponding increase in GPs', as well as, families' awareness of genetic disease. If, as Cheryl, Lori and Shirley suggested, there is minimal awareness on the part of GPs and other healthcare professionals about HD, why is this? What is the current attitude of healthcare professionals towards genetic testing for fatal inherited diseases? Very little is known about the attitudes of healthcare providers towards genetic testing (but see Wertz, 1998, for an exception). The current study did not investigate knowledge or attitude about HD in healthcare professionals. However, findings suggest it is an area worthy of future research. In the current study, for example, the perceived ignorance of GPs was unacceptable, frustrating and anger-provoking for at least two-thirds of participants, contributing an additional burden to families already facing a progressive, fatal illness.

Pragmatically, it was very difficult and frustrating to recruit a sample for the current study. It is not known exactly how many families are affected with HD, nor do we know the number of individuals at risk for the illness. A largely unknown at risk population could deter researchers who are interested in HD. Additionally, this knowledge deficit is a hindrance to policy makers. How can lobbying for services begin if the number of people affected with HD is not known? It should be reiterated that participants in the current study expressed great interest in these numbers. They want to

know how many others are affected by this illness, not least for social support and information-sharing needs.

The issue of when awareness of risk begins has been overlooked in clinical studies of predictive testing for HD. Clinical research on the psychological effects of testing typically choose an arbitrary baseline (e.g., one month prior to the test) from which to measure morbidity and to determine the impact of genetic test results. However, this practice fails to understand the perceived significance of disease-related events that normally long precede such baseline measures. Thus, whether awareness of the family history was abrupt or gradual, known or unknown, could have implications for psychological well-being or decisions about genetic tests. While some research has explored the effect of age and duration of awareness on genetic-test decisions (e.g., Quaid & Morris, 1993; van der Steenstraten et al., 1994), little attention is paid to the meaning of initial awareness. The current research has shown, for example, that being aware of the family history of HD does *not* translate into immediate awareness of implications for self or for one's children. Sherri's, Jackie's and Julie's narratives were exemplars of this gap in awareness. Thus, researchers cannot simply treat 'awareness of family history' as if it were some dichotomous variable, nor can we assume that discovering the family history of HD unfolds in a linear fashion. Participants' stories in the current research suggested that it does not.

Finally, the temporal and historical contexts in which discovery took place were also highlighted in the current research. Younger participants, for example, had 'always

known' about their family history of HD. They had escaped much of the confusion, uncertainty and upheaval faced by their parents and grandparents; Cheryl's narrative was most illustrative of this. And surely, new genetic technologies and other medical advancements (e.g., new medications) are changing the phenomenological experience of living at risk for a genetic disorder, as David suggested.

As many of the parents in the current research suggested, their children and grandchildren have access to information and knowledge that they themselves didn't (i.e., knowledge of the family history of HD). Parents in the current research implied or outright suggested that such information could or should be used in their children's decision-making (e.g., reproductive choices). Thus, clinicians and researchers alike should take seriously the temporal and historical contexts within which an awareness of genetic risk for HD emerges. This context had demonstrable effects on the experience of living at risk and on genetic-test decisions for participants in the current study.

Clinically, perceived ignorance surrounding HD has implications for (1) the provision, amount and type of information imparted during genetic counseling sessions, and (2) the provision of healthcare to families affected by HD. The complex and highly technical language of Mendelian genetics could be misunderstood by at-risk individuals who are already distressed when they arrive for counseling. Most participants who had attended genetic counseling sessions were very satisfied with the experience, perceiving counselors as 'very good' and 'caring people.' Despite this perception, however, difficulties in risk comprehension were evident. One participant, for example, suggested

the genetic counselors were 'very good;' however, the technical information imparted during counseling was confusing and hard to understand:

They [genetic counselors] were very good. It was useful, but not knowing what it was about and trying to soak it all in. Still, it was like you were stupid, and I didn't know what they were saying. I was trying to take it in. Sometimes, doctors have a way of putting words in there...I'm not a smart person.

Further, for at-risk individuals who choose not to be tested, they normally have no involvement with the public genetics arena and are left to founder about on their own in the quest for answers. Many such participants in the current research suggested the need for 'somewhere to go' following the initial discovery of HD in their family. As noted, this need was especially pronounced for those individuals who did not want genetic testing. This finding should caution clinicians and researchers against the assumption that declining the genetic test for HD equates with a disavowal of genetic risk. Findings from the current study suggest that at least some at-risk individuals *do* want the opportunity to discuss their risk with a knowledgeable professional (e.g., genetic counselor, social worker or psychologist) and have their questions answered, even if they never take the genetic test.

Psychological support would, therefore, be beneficial for some at-risk individuals. Importantly, healthcare professionals *must* acknowledge non-participation in testing as a valid choice. Some at risk participants perceived genetic counseling as a precursor to taking the genetic test, believing that attending counseling sessions would mean they *had* to take the test. For some of those who do not want to be tested, this deterred their

participation in counseling, leaving them alone with their search for information about HD.

Ignorance surrounding HD could also negatively impact healthcare delivery to affected families. If, as participants suggested, 'no one' knows about HD, there exists the potential for inferior care of individuals affected with HD. HD is a complex illness, displaying a wide and variable range of physical and psychiatric symptoms, affecting not only the person with the diagnosis, but also his/her family members. Both persons affected with HD and their caregivers noted the need for experienced healthcare professionals with knowledge and expertise in a wide range of services needed by HD families (e.g., Dawson et al., 2004). For example, expertise in the areas of nutrition, behavioral therapy, physiotherapy, and speech pathology were perceived as essential, to name a few. Due to the large numbers of people involved in the care of people affected with HD, there is the potential for lack of co-ordination and duplication or omission of services. Participants in the current study, for example, suggested the need for better information sharing between care professionals and co-ordination of services for HD families.

A recent project initiated by the Milton Keynes Primary Trust (located in Milton Keynes, UK) is attempting to respond to these sorts of concerns (see www.mkpct.org.uk/pdfs/huntingdons-disease.pdf for a copy of the report). It has established a single local service for persons with HD in the area. The project's aim is to provide 'seamless service,' accessed by a single point of contact and linked with a core

multidisciplinary team of professionals (e.g., a neuropsychiatrist, physical disability specialist, speech pathologist and a dietician). Program review is ongoing and may indicate the effectiveness of such an approach.

Questioning what it means to take a decision

Beyond exploring *why* participants chose to request or decline the genetic test for HD, the current research also investigated *how* such decisions were taken (Table 8). These stories, in particular, proved interesting since some of them challenged the conventional construction of a genetic-test decision as an opportunity for choice. Narratives in the current research suggested that this construction is inadequate, at least for some at-risk individuals.

Stories of *no decision to be made* were most challenging to the conventional framing of genetic testing as a 'decision.' In these narratives, participants knew they either did or did not want to know if they carried the altered HD gene; Victoria's, Patsy's and Sherri's stories were reflective of this theme. There was little ambivalence and minimal conscious reflection on the test as an opportunity for choice. Similarly, stories of *constrained decisions* also showed minimal evidence of perceiving the test as an opportunity for choice, at least not for self. Rather, taking the test provided participants' children with choices, especially regarding reproduction. In narratives of *constrained decisions*, participants (e.g., Dennis and Julie) suggested they had the genetic test for their children, not for themselves. In this way, perceived moral duty to one's family constrained the perception of the genetic test as an opportunity for choice.

In contrast, narratives of *re-evaluating the decision*, revealed a dynamic decision-making process. In these stories, the narrator generally did perceive the test as an opportunity for choice, both for self and children. Some initially wanted the test, but moved toward rejecting it, while others were initially opposed to the test, but moved toward requesting it. These narratives confirm the existing literature on genetic-test decisions which suggests at-risk individuals can experience anxiety, confusion and uncertainty as they consider taking the test; ending the uncertainty is, in fact, a major motive often endorsed for taking the test (e.g., Meiser & Dunn, 2000).

Finally, narratives of *test triggers* revealed how an odd behavior in self (e.g., twitching or inappropriate emotional responses) triggered at-risk individuals' thinking about their family history of HD. Knowing HD was 'in' their families, these participants recognized that the odd behavior could be indicative of HD and each sought genetic testing; Tony's, David's and Steven's narratives were reflective of this experience.

The question of *how* genetic-test decisions are taken has been relatively ignored in the literature on genetic decision-making. This omission is curious (Cox, 1999). From a clinical perspective, genetic-test 'decisions' should be autonomous and voluntary. No other person or group should unduly influence this decision. Tested participants in the current study often remarked on this admonition given to them during genetic counseling sessions. It is interesting that while participants in the current research upheld the clinical stance that the decision to test was theirs alone, many tested participants spoke about their felt obligation to others as an integral part of their test decision. Narratives of

constrained decisions were most illustrative of this. The obligation to others and its role in genetic-test decisions challenges the conventional meaning of autonomy in medical decision-making and questions the practice of constructing decision-making as a rational, self-directed activity (Cox, 1999).

Pathways to the test decision revealed in this study, therefore, have implications for some research in health decision-making. They challenge the construction of a 'decision' as an opportunity for choice, at least for potentially life-threatening decisions. Some participants' narratives raise questions about what it is that allows some at-risk individuals to see that there is a 'decision' to be made. Conversely, what is it about some participants' experiences that seems to point – without apparent hesitation – to one, and only one, course of action? These life experiences are worthy of future research and also deserve attention during genetic counseling sessions if we want to uphold the gold standards of autonomy and informed consent in genetic testing protocols.

Stories participants told about *why* they chose to take or decline the genetic test (Table 8) were also revealing and have implications for health decision-making research. Narratives contained numerous factors that seemed to influence genetic-test decisions, including age, diseased-related events in the family, responsibility to others, and current and anticipated emotions. Theoretically, this suggests the process of genetic decision-making may not be a rational, static process. Obligations to others and emotions also influenced these decisions. Yet, social cognition models such as the Health Belief Model

(often used in genetic decision-making research) normally do not explicitly account for such variables.

Stories of non-tested participants also have implications for how we might speak about denial of genetic risk. In the literature, at-risk person are sometimes characterized in a negative light (e.g., they are regarded as pessimistic, passive and/or unable to cope with a positive result). Binedell and Soldan (1997) have warned against this negative view, fearing it could bias healthcare professionals against the perception of non-participation as a valid choice.

Narratives of participants in the current study, however, revealed a 'dismissal' of their genetic risk, rather than denial. Some, for example, cited their age as a determining factor in their declining the genetic test at this time. There was no sense, however, that they were denying their family history of HD or its possible implications for future self. Psychometric risk research tends to underestimate the effect of socio-demographic variables, such as age, that can influence how people identify and respond to risk (Lupton 1999a,b). In the current research, however, stage in the life course had noticeable effects on the saliency of genetic risk and the decision to undergo genetic testing for HD.

Such findings challenge the 'reflexive' agent of Beck's (1992) risk society – a calculating, critically reflexive person who logically assesses risk(s) and contests expert risk discourses. As Lupton (1997) argued, depictions such as these, "...tend to portray a subject that is non-differentiated...there is little discussion of how gender, sexual

identity, age, ethnicity, social class and personal biography or life experiences can affect the taking up of “consumerist” or “reflexive” positions” (p. 374).

Expanding the meaning of genetic risk

If there is one overarching conclusion to be drawn from participant narratives, it is this: Genetic risk for HD was not perceived as a numerical probability. Instead, it was a lived reality, an “embodied risk” (Kavanagh & Broom, 1998) that existed inside persons at risk for HD.

For many participants, genetic risk was re-contextualized as a threat, both to self and other family members (Table 8). It was especially pronounced as a threat to one’s children. Narratives in the current research (e.g., Roxanne’s, Julie’s and Kathleen’s) clearly revealed strong emotional meanings associated with genetic risk – fear, anxiety, dread, blame and guilt. These emotional meanings of genetic risk for HD have both clinical and research implications.

Findings of the current research highlight the emotional pre-test state in which some test candidates can find themselves and raise questions about the ability to absorb and integrate complex information imparted during counseling sessions. Janis and Mann’s (1977) decisional conflict theory argued that stress interferes with the ability to consider the salient features of a situation and to deliberate carefully about the pros and cons of alternative options. Such deliberation is stressed during genetic counseling: Protocols for genetic testing for HD do encourage test candidates to consider the implications of a positive test result, a negative test result or of not being tested at all.

Some participants in the current study, however, commented on their difficulty in understanding and absorbing the information provided during counseling sessions. They recommended that counseling sessions be extended over a longer period of time and include less information, allowing at risk families more time to reflect upon and absorb complex risk information. Recall, however, that for some at-risk individuals there is *no decision to be made*. These people will likely not need additional time to understand or reflect upon complex risk information. Thus, counseling must be tailored to meet the diverse needs of persons at risk for HD, underscoring the earlier suggestion that a single counseling protocol may not be appropriate for every at risk person.

The emotional meanings of genetic risk also imply a need for researchers to measure the influence of emotions on genetic-test decisions. Participant narratives suggested that affect can play a crucial and sometimes immediate role in decision-making under risk, despite the assumption of traditional research in this area that risky decision making is essentially a cognitive activity. Researchers interested in genetic-test decisions should include some exploration and/or measure of affect.

Beyond emotion, genetic risk for HD was permeated with meanings of responsibility and moral obligations to others (Table 8). Tested participants, in particular, experienced their risk as a moral phenomenon: Taking the genetic test was perceived as something they *should* do. Responsibility to future generations and to current and future partners emerged as important lived dimensions of genetic risk. Study participants also noted their responsibility to plan for their futures and to communicate their genetic risk to

children, other family members, and in some cases, employers and friends. When the meaning of genetic risk resides in responsibility - as it did in the narratives of participants such as Julie, Lori, Jackie and Kathleen - there are significant clinical and research implications.

When genetic risk is experienced as responsibility, it can create tension and anxiety for at-risk individuals. Novas and Rose (2000) suggested:

...the new genetics also links up with contemporary practices of identity. It operates in a political and ethical field in which individuals are increasingly obligated to formulate life strategies, to seek to maximize their life chances, to take actions or refrain from actions in order to increase the quality of their lives, and to act prudently in relation to themselves and to others (p. 487).

Perceptions of what constitutes acting 'prudently' in relation to others raises questions about whether genetic-test decisions are purely autonomous and voluntary. If, as some study participants suggested, they took the genetic test out of perceived moral duty to others, what effect did this have on their psychological well-being, especially when the test result was positive? How do such at-risk person cope with knowledge of their genetic risk when it is knowledge they might never have wanted for themselves? Is a different kind or amount of psychological support needed for these individuals? Theoretically, how much of the variance in genetic-test decisions is accounted for by perceived obligation to others, and do differing perceptions of responsibility help differentiate tested from non-tested persons? These are all interesting questions for future research.

Theoretically, these findings also refute aspects of Beck's (1992) risk society thesis. Beck has argued that today's risks are global and democratic. Participants in the current study, however, were concerned about 'local' rather than 'global' risk. That is, they were concerned for themselves, their children, and other members of their family, rather than the global idea of genetic risk per se. As well, there existed a class bias to the management of risk, despite Beck's (1992) assertion that today's risks cross all class boundaries. In the current research, however, lower socio-economic, lower education participants were far less likely to understand the genetics of HD and to cope financially with the extended illness. As a result, they faced not only the physical, psychological, and emotional aspects of living with HD, but also the crippling economic burden associated with the disease.

While the economic burden of HD was not a focus of the current study, it did emerge as an anxiety-provoking reality for some caregivers, replicating research with persons affected with HD and their carers (Dawson et al., 2004). At several points throughout my interview with one caregiver, for example, she spontaneously talked about the devastating economic impact of HD:

It is so devastating. It's bad enough to have the disease, but when you haven't got food to go along with it. It's so hard. I've never gone to a food bank in my life until they got sick. –Female, caregiver

When a wage-earner becomes afflicted with HD, there comes a point when s/he can no longer work. This economic burden is heightened when the partner is the primary caregiver, precluding his or her opportunity to work. As caregivers stressed, caring for a

person affected with HD is a 'full-time job.' This finding raises questions about the type and amount of support provided by government to HD families. The caregiver quoted above became very emotional when she said:

The social assistance, the government, if only they would realize. If I wasn't around and they had to take care of [relative], how much money would it cost the government to do that? I'm asking them to give [relative] a life. If they don't want to give it to me, don't give it to me, but give it to them. [Relative] worked all their life, and I did too. Alright, now is the time. It's not their fault. (...) It's not fair. HD doesn't mean you have to lay down and die. They are real people. They don't just lay down and die. -Female, caregiver

Putting genetic risk on the back burner

Findings from the current study suggest that genetic risk for HD is chronic (Kenen et al., 2003b): It is 'always there,' but whether or not it is salient depends on several zones of relevance. These include, but are not limited to, stage in the life course, family history of HD, personal beliefs about carrying the altered gene, individual coping style and unique life events. These variables are not independent, but rather interact, to influence risk salience over the life course. This finding also raises important theoretical and clinical considerations.

Theoretically, the findings imply the need for a much broader consideration of the variables that affect genetic decision-making or other responses to genetic risk. Social cognition models typically measure the effect of perceived risk, severity, control and attitude on test decisions. However, findings from the current study suggest the importance of additional variables in some health decision-making contexts. The findings also imply the need for longitudinal data that would allow researchers to explore

fluctuations in zones of relevance over time. Longitudinal designs could also identify additional zones of relevance that might emerge as at-risk individuals progress through the life course.

Future research should also study zones of relevance and risk saliency in other regions, ethnic groups or genetic disorders. It is possible that cultural, religious or regional factors could account for observed differences in risk saliency. For instance, the interview guide did not ask participants about religiosity; however, if at-risk person believe their lives are fated (e.g., dictated by God), does this translate into less interest in genetic testing? Narratives in the current research, for example, suggested that at risk participants were more likely to speak of their futures as fated than tested participants. Further, what role does religiosity play in coping with genetic risk and/or illness? These are interesting questions for future research.

Zones of relevance in the current research also have implications for genetic counseling and follow-up support. While the bulk of clinical research on genetic testing for HD suggests minimal post-test psychological distress, distress can occur some time later (e.g., with the death of an affected relative or as the age of onset approaches), and support could be required at that time. Almost every participant in the current research commented on the lack of follow-up psychological support for families affected by HD. In the only longitudinal study of the effects of predictive testing for HD greater than five years, Timman et al. (2004) suggested that research to date could have underestimated the real impact of a positive test result. They found increased levels of hopelessness in

participants who tested positive for the altered HD gene over the study's 7-10 year follow-up. Timman et al. (2004) suggested, "Testing for fatal inherited diseases creates a long-term, lifelong stress reflected by gradually increasing levels of hopelessness as the onset of disease approaches. This pattern may have implications for follow-up of cases" (p. 196).

This suggests that follow-up support is needed for much longer time periods than current protocols suggest. One participant in the current research suggested that counselors 'touch base' with HD families every six months. The participant thought a simple phone call could help maintain contact with the genetics community, reduce feelings of isolation and provide the opportunity to address any problems or concerns that could have arisen in the ensuing time since last contact. It is not suggested that every individual at risk for HD would appreciate this kind of regular follow-up support. Findings from the current study suggest, however, that some at-risk person would find this a valuable service.

Coping with chronic risk and illness

At risk participants were aware of the impact of their genetic risk for HD on their self-identities and on their lives in general. Many participants went back and forth in their minds between a heightened awareness of their risk (notably during particular zones of relevance) and active efforts to bracket off (Bury, 1991) the at risk label and get on with their lives (e.g., Julie and Cheryl). This was also observed in participants who had tested positive or intermediate for the altered HD gene (e.g., Jerry and Kathleen, respectively).

In these narratives, participants attempted to minimize biographical disruption and maintain a normal life in the face of chronic risk and/or future chronic illness (Table 8).

Researchers studying genetic risk, therefore, should not assume that risk is always salient and anxiety provoking. Rather, future research could attempt to delineate when risk is salient and with what effects. Such research could inform genetic counseling and suggest the need for, and timing of, additional follow-up support. As noted, for example, findings from the current study suggest the need for support as the age of onset approaches and when caring for an affected relative.

It is also notable that some tested participants had concerns about their quality of death that were not discussed during counseling sessions. This finding raises questions about the process and content of genetic counseling. For example, how much control over these sessions is given to the at risk person? Process studies of counseling could help identify barriers to at-risk person's willingness to discuss important issues such as quality of death.

Both research and clinical practice have documented that psychosocial issues, rather than physical pain or symptom management, have a critical impact on a dying person's quality of life and end-of-life decision making (see Werth, Gordon, & Johnson, 2002, for a review). Werth et al. (2002) identified a host of psychosocial issues that are typically critical near the end of life, some of which were raised as concerns by participants in the current research: Autonomy or control over the end of life, maintaining dignity in death and not being a burden to loved ones. Psychosocial factors such as these

should be addressed if we are to provide comprehensive, quality end-of-life care to persons affected with HD. Future research must address end-of-life issues in persons affected with HD, their carers and in at-risk person nearing the age of onset. Palliative care service providers must also recognize the critical importance of psychosocial issues near the end of life. Werth et al. (2002) cautioned:

To the extent that alleviating suffering and improving quality of living and dying are the goals of improving end-of-life care, an emphasis on physical pain and symptom management that ignores psychosocial aspects may be short-sighted or seriously limited in effectiveness. Further, such an emphasis neglects the factors that appear to be the primary reasons why people make many of the end-of-life decisions that they do (p. 403).

This warning is all the more relevant to a person affected with HD since symptom management will likely be prominent near the end of life. For often, it is during this time HD symptoms are most pronounced (e.g., chorea, difficulty swallowing or various other forms of incapacitation). End-of-life issues should be addressed – in those who desire it – long before this stage in the illness. Findings of the current research suggest that at least some people at risk for HD had concerns about their quality of death, and these concerns were not addressed with genetic counselors.

The discussion of participant coping strategies for living at risk for (or with) HD highlights additional theoretical and clinical implications. Theoretically, findings support the utility of a stress and coping model for studying adjustment to stress associated with living at risk for HD or with proceeding with genetic testing (cf. Pakenham et al., 2004). Notable differences emerged between tested and non-tested participants in their reliance

on primary or secondary control coping: At risk participants were more likely to employ secondary strategies.

The current study is limited in its ability to explore differences in adaptation to genetic risk between these coping strategies. For example, is primary or secondary control coping more adaptive for people at risk for, or affected with, HD? Pakenham et al. (2004) suggested that reliance on passive avoidant coping, higher threat appraisals and lower self-efficacy beliefs were related to poorer adjustment in those at risk for HD. Future research could explore which coping strategies are most effective for persons at risk for HD, for those testing positive for the altered gene and for caregivers of persons affected with the illness. Findings of the current research, for example, suggested that secondary control coping seemed adaptive for caregivers and at-risk person, as assessed by participants' perceptions of their own coping.

Regarding clinical practice implications, findings suggest that a stress and coping framework could be useful in providing genetic counselors or other practitioners in the clinical genetics field with a framework within which to understand individual differences in coping with genetic risk and testing. The framework could identify stress and coping factors, such as perceptions of threat and self-efficacy, social support and coping strategies that genetic counselors should take into account when assessing the potential for adjustment difficulties in the context of living at risk or undergoing genetic testing for HD. Pakenham et al. (2004) rightly suggested that some of these stress and coping factors could be modified in clinical interventions in an effort to enhance

adjustment to living at risk. For example, reducing perceptions of threat or enhancing perceived self-efficacy.

Study participants also confirmed the benefits of social support for coping with genetic risk for HD, supporting genetic testing protocols that suggest test candidates be accompanied by a support person. Many lamented the lack of available social support in their hometowns, suggesting the need for organized support. Notably, even at risk participants who did not perceive support groups as beneficial while they were currently asymptomatic suggested the need for such support when they were caring for a (now deceased) affected relative. Some also suggested they could need such support in the future with disease onset. Study participants wanted to know they were not alone and were curious about how other families found out about HD and how they were experiencing the illness. Thus, the narratives of at-risk individuals, those affected with HD and their caregivers all implied the need for social comparison information.

These findings have implications for patient education and information. Since patients and their family members want information about their illness, GPs, neurologists, genetic counselors and social workers should provide at-risk individuals with information about support groups or other support (e.g., online illness groups or community groups such as the provincial chapter of the Huntington Society of Canada, HSC) in their area. A recent study found that support groups for people affected with HD and their caregivers were perceived as beneficial, both for practical information exchange and for bolstering the spirit (Dawson et al., 2004). Findings such as these and those of the current study

confirm the importance of social comparisons in adjusting to genetic risk for HD and the illness itself and suggest a useful theoretical framework for studying coping with genetic risk.

It should also be noted that several participants spontaneously mentioned the value of national conferences held by the Huntington Society of Canada (HSC). Over the past ten years, several study participants had had the opportunity to attend at least one of these conferences. Participants suggested the conference was a valuable experience that allowed them to acquire not only practical information, but also social comparison information about other families affected by HD.

Theoretically, findings from the current study suggest that social comparisons will not always be made to worse-off others, nor are negative representations of the at risk self the most likely. Rather, study participants engaged in lateral comparisons to a range of generalized others on a range of attributes, not just being at risk for, or affected with, HD. Additionally, representations of the self were overwhelmingly positive, not negative, regardless of whether participants were at risk, had been tested or were currently affected with HD. These findings should caution researchers that risk identities are not chronically salient and negative, nor are they best defined in opposition to a healthy, as opposed to an ill, majority. Individuals at risk for HD have multiple identities and social comparison groups from which to choose.

A seemingly adaptive coping strategy employed in the current research was social comparisons to affected relatives (Table 8). Both tested and at risk participants employed

this strategy to live with their increased risk. Those who had tested positive or with the intermediate gene, for example, compared themselves to their deceased relative. Since the age of onset was late, participants suggested they could still live a long, normal, healthy life before (or if) HD manifested. At risk participants also employed this coping strategy. If the age of onset was early in an affected relative and at risk participants had passed that age, this comparison seemed to serve as an effective coping strategy for living with genetic risk. Thus, the narratives of Jerry, Roxanne, Julie and Brenda underscore the importance of discussing the family history of HD with test candidates. They also caution researchers to take family history into account when studying adaptation to genetic risk.

Finally, the finding that participants employed the secondary control coping strategies of trust in science and appeals to fate also has theoretical and clinical implications. Theoretically, the trust in science displayed by most study participants challenges Beck's (1992) contention that risk society is marked by mistrust of science.

According to Beck (1992; 1995), increased reflexivity has led to increasing individualisation, de-traditionalisation and a heightened risk awareness. Risk society is supposedly accompanied by a corresponding erosion of trust in risk 'experts' (e.g., scientists, medical professionals, government, etc.). However, the context in which individuals at risk for, or affected with, HD find themselves is conducive to trust. In the current research, many participants perceived little choice but to trust in science since their situation affords them little control. There is nothing at-risk individuals can do to alter their risk for HD; science and medicine were perceived as the only hope. Thus, risk

research conducted within the framework of Beck's (1992) risk society treatise should be tempered by the context in which risk is lived and experienced. In the case of genetic risk for a fatal illness, trusting science is almost required in order to maintain hope.

Appeals to fate as a way of coping with a future that could contain HD also have clinical implications. Findings suggest that some individuals at risk for HD do not look ahead to a possible future of illness; rather, they prefer to leave the future to chance and 'deal with' whatever might come, when it comes. Findings of the current research and other research with HD populations (e.g., Dawson et al., 2004), therefore, suggest a reluctance to plan ahead for a possible future of HD, despite the normally long window of opportunity to do so. These findings contrast with a central tenet of good care at the end of life: Patients and physicians should plan in advance for possible future illness (Lynn, 1997).

Future planning could be important, not only to identify structural service needs, but also to assist with timely interventions to support informal caregivers and to avoid premature institutionalization (Dawson et al., 2004). I concur with Dawson et al. (2004), who suggested, "Introducing psychological support, including the provision of accurate information about care options and ensuring that financial needs are met, may mean that individuals and families facing Huntington's disease are willing to participate in long-term care planning" (p. 129). They suggested, as did participants in the current study, that a case management approach was the most useful way to cater to individual differences in the experience of living at risk for, or being affected with, HD.

Stigma and genetic risk for HD

The majority of participants suggested that sympathy, not stigma, was the dominant response to them and/or their affected relative (Table 8). Sympathetic responses were likely once others understood that HD was a genetic disease over which affected persons had no control.

Despite sympathy, however, perceived stigma did exist in relation to HD. Some at risk and tested participants generated felt stigma (Scrambler, 1998) when they acknowledged the possibility of future stigma with disease onset. Others noted the potential for discrimination in employment and insurance contexts. There was little evidence, however, that felt stigma was particularly distressing for study participants. All participants asserted that HD was completely out of their control. This dimension of a stigmatizing condition seemed to mitigate any potential negative effects of stigma on participants in the current research. Since the illness is beyond their control, participants could dismiss any negative responses from others as not valid to the self. As such, there was little evidence that participants suffered pronounced ill effects on their self esteem or life satisfaction.

Caregivers were more likely to recount instances of actual stigma, underscoring the importance of illness severity on experiences of stigma (Crandall & Moriarty, 1995; Dijker & Raeijmaekers, 1999). Caregivers (past and current) recounted numerous examples of social rejection which they experienced as frustrating and anger-provoking. Some noted how the stigmatizing responses of social others constrained their and/or their

affected relative's social behavior (e.g., limiting public outings, selective communication).

These findings have implications for both research and clinical practice. The relative lack of psychological sequelae in study participants supports recent stigma research (see Crocker et al., 1998, for a review) that suggests stigmatized persons are generally happy and satisfied with their lives. Therefore, it is perhaps fruitful for stigma researchers to explore *within* group differences in psychological well-being (or other stigma-related outcomes), rather than undertake comparative studies of the stigmatized and non-stigmatized. For example, the current study suggests that at-risk person closer to the age of disease onset have more concerns about stigma than younger at-risk individuals.

From a clinical perspective, a discussion of potential stigma, including discrimination in insurance and employment contexts, should be included in genetic counseling sessions. Follow-up of clients should also consider these issues since approaching the age of onset and visible HD symptomatology could mean enacted stigma for individuals affected with HD, a potential acknowledged by some tested participants in the current research (e.g., Kathleen).

The current study suggests that felt stigma affects communication about genetic risk for HD. Theoretically, we know very little about how families communicate about genetic risk and with what effects. Communication is significant in the context of clinical genetics, however, for a number of reasons: It can determine the accuracy of information

brought to counseling sessions; issues raised (or not raised) in communication between family members can lead to emotional distress; and, non-disclosure of risk information can undermine the decision-making autonomy of other at risk family members (Wilson, Forrest, et al., 2004).

In the current research, concern about potential stigma emerged as one variable that constrained communication about risk for HD in employment, insurance, clinical and social contexts. Future research could investigate the relative impact of felt stigma on risk communication and on genetic-test decisions, not only for single gene disorders such as HD, but also for multi-factorial diseases such as hereditary cancers.

More generally, we know relatively little about how social others view at-risk individuals and families. While the current study suggests that sympathy was the main response to participants and/or their affected relative, some narratives highlighted several examples of social stigma. These experiences underscore the perceived ignorance surrounding HD and highlight the need to dispel this unfamiliarity in social others and healthcare professionals alike. Future research should explore prevailing attitude towards, and knowledge of, hereditary disorders such as HD and persons at risk for the disorders. This latter component of research is necessary: Extant research addresses public attitude towards genetic testing more generally, such as use of genetic technology and information, access to genetic information, and the benefits/harms of new genetic technology (e.g., Human Genetics Commission, 2001). However, there are very few

studies that measure attitude towards at-risk person or towards adult onset diseases more generally (Evers-Kiebooms et al., 2000).

Lessons learned

It is my hope that the findings of this research, as I have summarized them above, will facilitate new understanding and prompt reflection and debate about the experience of living at risk and genetic testing for HD. At the end of this journey, however, I find myself reflecting on what I have not been able to accomplish in carrying out this research. Here, I wish to highlight some of the limitations of the current research and provide a succinct list of suggestions for future research.

Study limitations

Prospective, longitudinal designs are needed for research on genetic risk. A wide range of individuals, each affected in some way by HD, was included in the current study. However, I (formally) spoke to each of them only once. The cross-sectional nature of the research precluded an examination of how living with genetic risk changes over time. Yet, findings from the current research highlighted the significance of age and stage in the life course in negotiating genetic risk for HD.

Prospective designs, in particular, offer the opportunity to follow at-risk individuals prior to, during and after genetic testing. Recent longitudinal work (e.g., Timman et al., 2004) revealed changes in hopelessness at ten-year follow-up in those testing positive for the altered HD gene. Longitudinal work would permit researchers to track changes not only in emotional response such as hopelessness, anger, regret or

blame, but also in risk saliency and coping strategies over time. Such a design would also be useful for studying at risk populations as it would allow an investigation of decision-making over time.

The current study comprised a rich and diverse set of narratives about the experience of living at risk for HD. Study participants actively took part in research interviews, drawing on a wide array of life experiences and memories as they told their stories. In all, there were over 430 pages of interview data, coupled with countless notes and analytic memos. While this was fortunate, it was also a source of complexity. It meant I was often overwhelmed and had difficult choices to make about what stories to emphasize and what stories to merely mention, without any sustained critical analysis. Such choices are inevitable when faced with the rich data set of the current research. However, it is important to acknowledge their implications for the findings of this research.

As readers will know, this dissertation focused primarily on (1) individuals at risk for HD and (2) predictive test candidates. While family members were given a presence, the current study would be quite different were it to have emphasized the perspectives of spouses/partners or caregivers as central. Regarding the caregiver perspective, I was concerned that the experience of caring for a person affected with HD would become lost in this dissertation amidst the central focus of living at risk or undergoing genetic testing for HD.

As such, the experiences of caregivers of persons with HD will be addressed in a separate, forthcoming paper, hopefully rectifying their lack of central focus in the current discussion. The research was limited, however, by its inability to recruit spouses/partners of at risk and tested persons. The lone partner of an at risk person in the current study confirmed that partners have a different perspective on genetic risk than the person undergoing testing or living at risk. I referred only briefly to this interview throughout the dissertation as I was concerned about confidentiality and anonymity. I had hoped to recruit far more partners than I did, and I was quite disappointed by this deficit in the study sample. The perspectives of spouses/partners have been neglected in the literature, and unfortunately, in the current research. In hindsight, I am not sure how I could have increased the participation of partners/spouses. Each participant in the current research was invited to inform his or her spouse/partner about the study. Additionally, all advertisements about the research, including recruiters, invited spouses/partners to join the study.

The study's sample is also limited in that it was highly educated, largely excluding the voices of lower educated at-risk person. Recruitment of study participants was (partly) conducted in the clinical setting, and it is likely that the use of recruiters, such as genetic counselors, could have introduced a sample bias into the current research. There is no way of ascertaining exactly how potential participants were identified. For example, perhaps counselors (or other recruiters, for that matter) only invited educated persons to participate.

In an effort to reduce the amount of time spent on recruitment, it was agreed that counselors would contact only one individual in HD families and ask him/her to inform other family members about the research. This practice, while useful for reducing time and effort spent on recruitment, may have attenuated participation, as there is no way of knowing if the contacted individual actually informed other family members about the study.

Sample limitations also precluded examination of gender differences in response to genetic risk for HD. Females were over-represented in the current study, leaving little opportunity to explore gender differences in test uptake or communication about genetic risk. Similarly, the mean age of the study's sample was 46 years, and reproductive decisions had already been made for most participants. This left little opportunity to explore important issues about reproductive choice and parenthood.

The current research was fairly broad in scope, investigating core topics such as family history of HD, 'decisions' about genetic testing, meanings and implications of living at risk, communication about genetic risk, and experiences of stigma in at risk, tested and affected individuals, along with their caregivers. And, as noted, it was favored with a rich data set. Nonetheless, in the process of analysing interview transcripts and writing the data chapters, certain deficits were notable. For example, the interview guide did not expressly ask participants about whether or not they regretted having the genetic test (or not having the test) or whether they felt anger towards their affected parent for

passing on the altered HD gene. As such, the current research can say little about whether or how anger or regret affects test decisions or communication about genetic risk.

Additionally, interviews in the current research did not explore participants' lay beliefs about genetics and inheritance (e.g., what causes HD?) However, some beliefs were spontaneously mentioned by participants (e.g., HD skips a generation; a person who shares more resemblance in personality or appearance with relatives who have HD is perceived as more likely to carry the gene). In Chapter 9, I noted how personal beliefs about whether or not one carried the altered HD gene influenced risk saliency and genetic testing. Participants who had always felt they carried the gene were more likely to undergo testing and were more likely to have a heightened awareness of their genetic risk. The current research did not explore, however, the origin of these 'hunches' about gene carrier status. In retrospect, however, it is possible they were influenced by lay beliefs about heredity. For example, recall the participant who suggested she was the oldest of the grandchildren, and if any family member was going to 'get it,' it would be her (see Chapter 9).

A better understanding of lay beliefs regarding heredity could have implications for genetic counseling protocols. Richards (1996) argued that lay beliefs about inheritance did not develop in tandem with the coming of the new genetics; rather, "they have long been a part of family culture" (p. 249). He suggested that when a family is faced with the possibility that it carries a genetic disorder, members will try to make sense of the observable disease pattern in the family in terms of previously held

knowledge about inheritance. This could require further ideas being introduced to account for an "intermittent appearance of the disorder." For example, the disorder could be thought to skip a generation or appear only in first-born children. Richards (1996) suggested that such beliefs could serve a psychological defense function for some at risk family members. He indicated that difficulties in genetic counseling sessions could arise because lay beliefs conflict with the Mendelian account of disease typically provided by genetic counselors. Henderson and Maguire (1998) have suggested that counseling sessions begin with an exploration of each counselee's unique beliefs regarding genetics and inheritance; this seems a worthwhile suggestion.

Finally, I wish to comment on the rigor of the current research. The study is limited in its generalizability. It employed a relatively small sample of people at risk for, or currently affected with, a fatal genetic disease. Additionally, sample numbers were small for each individual group, for example, at risk, positive test result, negative test result, caregiver or affected. While the size of the sample is in line with many qualitative studies, findings might not generalize to persons living at risk for other genetic disorders or even to others at risk for HD. However, the deliberate search to include a variety of at risk, tested, caregiver and affected participants does increase the representativeness of the study's sample (Mays & Pope, 2000).

Concerns about generalizability are mitigated somewhat, however, since generalizability is not the goal of research utilizing interpretative phenomenological

analysis (IPA). Rather, the goal is to capture how particular people perceive and respond to their experiences, highlighting the value of each particular case.

Chapter 6 outlined, in detail, the many safeguards employed in the current research to contribute to the study's rigor. That discussion will not be reiterated here. It is worth repeating, however, that researcher reflexivity is particularly crucial in qualitative research. Throughout this research, I have constantly engaged in critical reflection on my own assumptions, beliefs and reactions, and my reflexive commentary is scattered throughout these pages. This practice is essential for readers who want to evaluate how the researcher and the research process could have shaped or influenced the data collection and analysis. Cox (1999) reminds us that:

...the integration of reflexive commentary about the nature of the relationship between personal experience and the research helps to establish trust: Without such reflexive commentary, the reader is in no position to assess the degree to which the researcher distinguishes between her own experiences and the experiences of those she studies (p. 401).

It is hoped the transparency of this research (both to participants and to readers), along with the use of triangulation, respondent validation, the constant comparative method and the refutability principle (Silverman, 2000) has contributed to the study's rigor.

Regarding respondent validation, I wish to inform the reader of some participant feedback that could have implications for the validity of the study findings. As noted, all participants received a summary report of findings and were encouraged to contact me with any concerns or suggestions. I specifically asked participants to consider whether

my interpretation of their comments was reasonably valid. Over half of the study participants contacted me after reading the summary report. Each participant strongly endorsed the research, and no one suggested the findings were in any way incorrect or their stories misinterpreted. The following comments were typical:

Just wanted to let you know I thought the summary was fantastic. I finished it in two days and was impressed. –Female, tested negative

I read the summary Holly. It is a great piece of work...-Female, at risk

Thanks very much for sending me your summary report of findings. I read every page and thought you did a great job. –Female, tested negative

It is acknowledged that respondent validation alone is not sufficient to ensure the validity of qualitative research; however, participant comments such as these were encouraging.

Directions for future research

There are numerous lacunae for future research on the experience of living with genetic risk. A brief list of topics would include: 1) Prospective, longitudinal designs that follow test candidates before, during and after testing, including comparisons with at-risk person who choose not to be tested. These studies should include a much broader array of outcomes than depression or anxiety. For example, other outcomes could include feelings of anger, blame or regret, relationship with partner/spouse or family communication patterns; 2) Qualitative investigations of *how* genetic-test decisions for HD and other genetic diseases are taken; 3) Qualitative and quantitative investigations of *why* at-risk person accept or decline testing. Such research must include a much broader array of

factors that affect test decisions, notably emotions, felt obligation to others, and personality factors such as monitoring/blunting coping style; 4) Process and content studies of genetic counseling sessions with a particular focus on (a) how much autonomy is given to test candidates during the session and (b) identifying barriers to candidates' willingness to discuss serious concerns such as quality of death; 5) Longitudinal investigations of the zones of relevance that affect salience of genetic risk for HD. These studies should be replicated in different ethnic groups, regions and genetic disorders; 6) Studies using a stress and coping framework to investigate adaptation to risk for HD; 7) Qualitative investigations of the experience of caring for an individual affected with HD or of being the spouse/partner of a person at risk for HD; and 8) Investigations of perceived stigma associated with HD and its effect on communication about genetic risk.

Within the larger community context, it would be valuable to investigate:

9) The attitude of GPs and other healthcare professionals towards genetic testing for fatal genetic diseases; 10) More broadly, the attitude of generalized others towards persons affected with HD; 11) The economic burden faced by families affected by HD and its possible effect on planning for future care; and (12) Current healthcare services available to families affected by HD. Evaluation studies would be useful in this area to identify limitations of current healthcare delivery.

Concluding comments

This research has broadened our understanding of what it means to live at risk for a fatal genetic disorder and what it means to face the prospect of genetic testing. While

genetic risk for HD is notably different than genetic risk for multi-factorial diseases, we have much to learn from families affected by HD. Predictive testing for HD, in particular, has raised numerous clinical, social and ethical issues. As genetic tests become available for a variety of other adult-onset disorders (e.g., Alzheimer's Disease, breast, ovarian and colon cancer), it will become increasingly important to know how those most closely affected by the new genetics comprehend, manage and adapt to genetic risk information.

It is my hope that the current research has given a voice to families affected by HD and will help raise awareness about this devastating illness. I also hope I have caught the attention of other social scientists and perhaps sparked their interest in the lives of people touched by the new genetics.

Afterwords

It is with regret that I inform the reader of the passing of a study participant. This participant was particularly inspiring to me: Despite the ravages of HD, they were able to maintain a sense of humor and a hope for a cure that I found remarkable. They trusted science and supported HD research, including my own. I am humbled by the trust they placed in me, and I hope I have done justice to their story. In some small way, I hope this research has given them a voice and called attention to the experiences of those families affected by HD. I dedicate this research to them.

Finally, and on a much happier note, I wish to update the reader on Lori. Readers may recall Lori, who was waiting for her test result at the time of our interview. I am delighted to announce that Lori tested negative for the altered HD gene.

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Appendix A

Information packet provided to all participants. It included:

- Cover letter
- Information sheet about the research
- Topic guide for interviews
- Consent form

Date
Participant Name
Address

Dear _____:

Enclosed, please find the information about my research. I've included an information sheet about the study, the consent form for participants, and the topic guide for the interviews.

The topic guide is simply an outline of the kinds of issues I'd like to discuss. However, there are no right or wrong answers, and participants can feel free to raise any other issues they think are important.

For your information, there are several people who are spreading the word about this research. It is possible you might receive a call from someone else about the study.

Thank you so much for your interest in this research. I would also be happy to send information to anyone else you may know of who might be interested. Please don't hesitate to call me at work or at home, or to e-mail me should you have any questions or concerns. My contact information is included on the information sheet.

I will phone you shortly to make sure you've received the information and answer any questions you might have. At that time, we can arrange a time for our talk if you are still interested in participating. Once again, my sincere thanks for your interest.

Sincerely,

Holly Etchegary
PhD candidate, Psychology
Memorial University

HUNTINGTON DISEASE STUDY

You are invited to participate in a research study about Huntington disease. We are interested in talking to people who are 19 years of age or older and fall into one of the following categories:

- ☐ You are clinically affected with Huntington disease **OR**
- ☐ You have a family history of Huntington disease, had genetic testing, and received either a positive *or* negative test result, or you have not received your results **OR**
- ☐ You have a family history of Huntington disease, but have *not* had genetic testing **OR**
- ☐ You are a member of a family in which someone has Huntington disease, even if *you* are not personally at risk for the disease. You may be an immediate (e.g., child, sibling, spouse) or extended (e.g., cousin or aunt) family member

Background to the study

Genetic testing has led to the labeling of individuals, and whole families, as “at risk” for a genetic illness. One such illness is Huntington disease. This study will explore what it is like to have this illness (or *possibly* be at risk for it) or be a family member of someone who has Huntington disease. We want to know how Huntington disease affects your daily life and the lives of family members. We are also interested in how you think others (such as friends, co-workers, or society at large) feel about you and your family.

Knowing what the “at risk” label means to you is important. It may help provide information for genetic counselors about the best way to offer counseling sessions. It may also help to identify unmet healthcare or information needs, perhaps improving the delivery of healthcare to altered gene carriers and their family members.

Description of the study procedure and length of time

If you decide to take part, a researcher (Holly Etchegary) will phone you to provide more information about the study and answer your questions. You will be invited to participate in either a group discussion or a personal interview.

Focus group

You will be invited to one group discussion with other people who are living with the risk of Huntington disease (HD). Talks will be held at a time and place that is good for everyone. The group will talk about what it means to have Huntington disease or be at

risk for it (or be a family member of someone who has HD), how this affects your daily life, your family, and how you think other people see you. Group talks will be tape-recorded to make sure your comments are not lost. At the end of the study, however, all tapes will be destroyed.

The focus-group discussion will be about one to two hours in length. However, if you would like to discuss other issues, there will be no set time to end the session. We will provide you with a list of possible topics for the talk before arriving at the group discussion. However, you can raise any other issues you feel are important to talk about.

Please note that taking part in the group discussion will mean that your status as a person with HD (or a family history of HD) or the status of your family member as a person with HD, will be known to others in the group. However, we will not reveal your full name or any family member's name to the group; only first names will be used during the group talk.

Personal interview

If you do not want to take part in a *group* discussion, you can still participate in this research. If you would rather, the investigator will invite you to a personal, one-on-one interview at a location of your choosing. The interview will cover the same topics as the group discussion, and you will be provided with the list of topics before the interview. Interviews will also be tape-recorded to make sure your comments are not lost. At the end of the study, however, all tapes will be destroyed.

The information you provide will be used to produce research reports and possibly academic papers, and we will ask for your signed consent at the time of the group discussion or interview. However, your name will not be attached to any written material and your contact information will be destroyed once the research is completed.

If you want to participate

If you have any questions or would like more information before deciding to participate, feel free to contact the principal investigator, Holly Etchegary. She would be happy to answer your questions. Thank you for your interest!

INVESTIGATOR: Holly Etchegary
 Psychology Department, Memorial University
 St. John's, NL, A1B 3X9
 Office phone #: 737-8496 or 722-7971 (home)
 Email: hetch@play.psych.mun.ca

Focus group or interview topic guide

Focus-group discussion or interviews will be somewhat unstructured as a main goal is to allow you to explain, in your own words, what it means to be affected by Huntington disease, even if you have received a negative genetic-test result, or have never had genetic testing. The following topics will be used as a guide for our talk in all focus groups and in all interviews. However, you can feel free to bring up other issues you feel are important to talk about. There are no right or wrong answers here. Your opinions and feelings are important.

1. History of personal experience with Huntington disease (HD)

- How/when did you first learn of your family history of HD?
- What was your childhood experience of HD?
- Did you have the genetic test?
- Demographic information such as age, education level, gender, marital status, age at diagnosis (if applicable), had genetic test/no test, children/no children. This information will be collected with a very short survey instrument during the interview or group discussion.

2. Perspectives on the disease and medical profession

- What is the most/least frightening aspect of the disease?
- What has been good/bad about the disease?
- Does the medical profession (such as your family doctor or genetic counselor) understand what it means for a person and their family to be affected by HD?
- Was, or can, the science of genetics be of any help to you and your family?

3. Risk awareness

- When you hear the term '**at risk**' for HD, what comes to mind?
- Is or was the label 'at risk' a part of your self identity?
- Does this label impact daily life? If so, how?
- Did you NOT want to know your genetic risk? Is it ok for someone to NOT want to have the genetic test?

4. Stigma

- Do you feel any stigma within your family as someone who does (or might) have this disease?
- What does society think of those who have (or might have) HD?

-What do you perceive the response of others to be once they know you carry (or might have carried) the HD gene?

-Do you feel responsible for your own health?

-Do you think society holds you accountable for your own health? For example, do you think society would expect those with a family history of HD to have a genetic test?

-Have you ever encountered any form of discrimination or prejudice because of HD? Or, if you received a negative test result, did you ever encounter any form of discrimination before you were declared gene-negative?

5. Communication about the disease

-Do you talk about it within the family (immediate and extended)?

-Do you talk about it with others outside the family (e.g., friends, co-workers)?

-If so, under what circumstances? If not, why not?

6. Healthcare needs

-What are your concerns, if any, regarding your own healthcare?

-Do you have adequate insurance coverage?

-Is there anything you need (e.g., health information or support) which is currently unavailable to you?

-Do you have any suggestions for medical professionals who work with HD families?

-Is there anything else you think we should talk about?

Your comments are truly helpful. Thank you so much for your time and for sharing your story with me.

**Department of Psychology
Memorial University
St. John's, Newfoundland, A1B 3X9**

Consent to Take Part in Health Research

TITLE: **Labeled with genetic risk: Meanings and implications of living with Huntington disease**

INVESTIGATOR(S): Holly Etchegary
Office phone #: 737-8496 (Home: 722-7971)
Email: hetch@play.psych.mun.ca

You have been asked 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 healthcare or the healthcare of your affected relative(s).

1. Introduction/Background:

We would like to speak to you about being at risk for a genetic illness or being a family member in a family affected by a genetic illness. We want to know how the disease affects your daily life and how you feel other people see you and your family. We feel it is important to speak to people who live with a genetic illness every day. At-risk individuals and their family members will be invited to a group discussion or a personal interview about living with Huntington disease.

If we know how people live with the “at risk” label, it could provide information about the best way to offer genetic counseling. It might also help pinpoint unmet healthcare needs, perhaps improving the healthcare of those living with a genetic illness.

2. Purpose of study:

We will explore what it means to have a family history of Huntington disease for those who carry the altered gene, those who do not carry the altered gene, and their family members.

3. Description of the study procedures and tests:

If you agree to take part, a researcher will call you to provide more information about the study and answer your questions. If you still want to take part, you will be invited to one group talk with other people who are living with Huntington disease. Or, if you would prefer, you could participate in a one-on-one personal interview with the researcher. Talks or interviews will be held at a time and place that is good for everyone. The group (or interview) will talk about what it means to have Huntington disease (or be a family member of an altered gene carrier), how this affects your daily life, your family, and how you think other people see you. Studies like this have found that groups of patients and family members enjoy talking about their lives. Group discussions and interviews will be tape-recorded to make sure your comments are not lost. At the end of the study, however, all tapes and typed transcriptions will be destroyed.

4. Length of time:

There will be one focus-group session (or interview), about one to two hours in length. However, if you would like to talk about other issues, there will be no set time to end the talk (or interview).

5. Possible risks and discomforts:

It is possible that discussion about living with Huntington disease may be upsetting or hard to talk about. You can refuse to answer any question. Also, you can leave the group talk (or stop the interview) at any time, and all data about you will be destroyed. However, most people enjoy telling their stories. We will reduce any inconvenience to you by trying to have a suitable time and place for our discussion.

6. Benefits:

It is not known whether this study will benefit you personally.

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:

Your name will not appear in any report or article published as a result of this study. No real names will be used when reporting any responses. Your contact information will be destroyed once the research is completed. You may withdraw from the study at any time without consequence and all data about you will be destroyed.

9. Questions:

You have been given a copy of this consent form. If you have any questions about taking part in this study, you can meet with the investigator who is in charge of the study. This person is: Holly Etchegary, 737-8496 (or 722-7971).

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: Labeled with genetic risk: Meanings and implications of living with Huntington disease

Name of investigators: Holly Etchegary

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>I have received enough information about the study.</i>	Yes { }	No { }

I have spoken to Holly Etchegary or a qualified member of the study team.	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 have focus-group discussions (or interviews) tape-recorded. Yes { } No { }

I agree to take part in this study. Yes { } No { }

Signature of participant

Date

Signature of witness

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 B

Advertisement in the Huntington Society of Canada's (HSC) newsletter, *Horizon*:

March 8, 2004

Dear Friend of the Huntington Society of Canada:

I'm writing to you because you receive our newsletter, *Horizon*, and may be interested to know of research that is happening in the province of Newfoundland.

Holly Etchegary, a Ph.D. student in Psychology at Memorial University, is working on a dissertation entitled "Labeled with genetic risk: Meanings and Implications of living with Huntington disease." This research is concerned with the subjective meanings and implications of being labeled 'at risk' for HD. A primary goal of the research is to investigate the effect of risk information on everyday life.

Ms. Etchegary is interested in speaking with any HD family members who may be interested in participating in this research. Discussion will comprise broad themes, such as views of genetic testing and genetic illness, the meaning of 'at risk', the perception of stigma, and other themes. The research is a qualitative interview study. Ms. Etchegary is hoping that a better understanding of how HD family members define and live with risk will assist genetic counselors as they help people within the HD family.

Although the Huntington Society of Canada is not affiliated with this study, we did want to make our membership aware of it.

If you are interested in learning more, or would like an information packet about the research, please contact Holly at hetch@whinge.psych.mun.ca, or call her at (709) 737-8496.

Sincerely,

Isla Horvath
Executive Director and CEO

Appendix C

Advertisement in the Newfoundland and Labrador Medical Association's newsletter,
Nexus

Huntington's disease research

A study is currently underway at Memorial University. The research is concerned with the subjective meanings and implications of being labeled 'at risk' for a genetic disorder, specifically Huntington's disease, for both at-risk individuals and their family members. The research will investigate how the 'risk' label is interpreted and coped with, how the label affects daily life, and how at-risk person perceive the responses of others in their social environments.

To be eligible for the study, participants must be 19 years of age or older and fall into one of the following categories:

- Be clinically affected with Huntington's disease, OR
- Had a family history of Huntington's disease, had genetic testing, and received either a positive *or* negative test result, OR
- Have a family history of Huntington's disease, but have *not* had genetic testing, OR
- Be a member of a family in which someone has Huntington's disease, regardless of personal risk status. We invite both immediate (e.g., spouse, sibling) and extended (e.g., aunt, cousin) family members to take part.

Recruitment for this research is currently underway, and physicians may receive patient requests for information about the study. Further, physicians around the province are invited to identify patients who might be eligible for this research and notify them about the study.

The investigation is part of the doctoral research of Holly Etchegary, and it has received ethical approval from the Human Investigation Committee, Memorial University. Please feel free to contact Holly for full details about this study.

Holly Etchegary, MASP
PhD candidate, Department of Psychology
Memorial University
St. John's, NL, Canada
A1B 3X9
(709) 737-8496 or
(709) 722-7971
Email: hetch@play.psych.mun.ca

Appendix D

Cover letter sent to all neurologists in the province. Note: Neurologists' mailout also included the information packet as outlined in Appendix A, including the information sheet about the study, interview topic guide and consent form.

December 17, 2003

Name

Address

Dear Dr. ____:

I am PhD student in the Psychology department, Memorial University. I am writing neurologists to request their assistance with participant recruitment for a study on Huntington disease (HD). According to ethical guidelines offered by the Human Investigation Committee (HIC), Memorial University, initial contact with patient participants in research should be made by someone knowledgeable of their medical history. Therefore, I am hoping to secure the assistance of neurologists in recruiting potential participants for a research study I will be carrying out as part of my doctoral dissertation.

The proposed research is concerned with the subjective meanings and implications of being labeled 'at risk' for a genetic disorder, specifically HD. The study includes both individuals at risk of HD and their family members, some of whom may not be at risk themselves. I am interested in how the 'risk' label is interpreted and coped with, how the label affects daily life, and how 'at risk' individuals perceive the responses of others in their social environments.

To be eligible for this study, participants must be **19 years of age** or older, be cognitively competent in order to provide free and informed written consent for the research, and fall into **at least one** of the following categories:

- Be clinically affected with HD
- Had a family history of HD, had genetic testing, and received either a positive **or** negative test result
- Have a family history of HD, but have not had genetic testing
- Be a member of a family in which someone has HD, regardless of personal risk status. We invite both immediate (e.g., spouse or sibling) and extended family members (e.g., aunt or cousin) to take part.

Recruitment for this research is currently underway, and neurologists may receive patient requests for information about the study. Further, neurologists around the province are invited to identify patients who might be eligible for this research and notify them about the study.

Potential participants can be provided with an information sheet about the research (see enclosed).

The study protocol, including the above-noted methods of recruitment, has received full ethical approval from the HIC and is currently under review by the Research Proposal Approval Committee, HealthCare Corporation of St. John's. If you would like to see the complete dissertation proposal, including theoretical background, please let me know. E-mail would likely be the quickest way to reach me. Otherwise, please do not hesitate to contact me should you require any additional information. Thank you for considering my request; I look forward to hearing from you.

Sincerely,

Holly Etchegary, MASP
PhD candidate, Social Psychology
Memorial University

hetch@play.psych.mun.ca

Phone: 737-8496 (Psychology general office) or 722-7971 (home)

Fax: 737-2430

Appendix E

Poster about the research.

Huntington disease research

You are invited to participate in a research study about Huntington disease. We would like to speak to you about your experience of having a family history of Huntington disease. We are interested in how the disease affects your daily life and how you feel others respond to you and your family. You will be invited to participate in one focus-group discussion *or* one personal interview.

In order to participate, you:

- ☐ Must be 19 years of age or older and fall into one of the following categories:
- ☐ You are clinically affected with Huntington disease **OR**
- ☐ You have a family history of Huntington disease, had genetic testing, and received either a positive *or* negative test result **OR**
- ☐ You have a family history of Huntington disease, but have *not* had genetic testing **OR**
- ☐ You are a member of a family (immediate or extended) in which someone has Huntington disease, even if *you* are not personally at risk for the disease

If we know how people live with the 'at risk' label, it could provide information about the best way to offer genetic counseling. It might also help to identify unmet healthcare needs, perhaps improving the healthcare of those living with a genetic illness and their families.

If you are interested in participating, or would like additional information, please call or email the principal investigator, Holly Etchegary.

Holly Etchegary
 Department of Psychology, Memorial University
 Office # 737-8496
 Email: hetch@play.psych.mun.ca



