PARENTS' PERCEPTIONS OF THE INFLUENCE OF GENETIC COUNSELLING ON REPRODUCTIVE DECISION-MAKING FOLLOWING THE BIRTH OF A CHILD WITH NEURAL TUBE DEFECT

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ABSTRACT

Parents' Perceptions of the Influence of Genetic Counselling on Reproductive Decision-making Following the Birth of a Child with Neural Tube Defect.

Neural tube defect (NTD) is a common genetic (multifactorial) disorder in Newfoundland and Labrador. This descriptive study explored parents perceptions' of genetic counselling for this condition. The purpose of this study was to assess how parents of children with a neural tube defect perceive genetic counselling, as well as other related factors, and how these influences impact on their reproductive decisions.

Parents of children born in Newfoundland between May, 1984 and June, 1987 who also received genetic counselling regarding this condition were contacted by mail with a questionnaire designed by the researcher. The sample consisted of 55 subjects (31 mothers and 24 fathers) from 31 families.

Data were collected exploring the parents' recall of the recurrence risk of NTD; their interpretations of this risk; the impact of genetic counselling on subsequent procreative decisions; perceptions of prenatal diagnosis; the influence of decision-making factors other than recurrence risk; and, the actual reproductive behavior ensuing genetic counselling.

Although parents reported minimal influence of genetic counselling on reproductive decision-making, they appeared to be using the information obtained during genetic counselling

to facilitate their decisions. They also appeared to be using a myriad of other family oriented factors in making reproductive decisions including the wishes of their partners, fulfillment as parents and taking care of their other children. Other influences such as religion, relatives' influence, and career goals appeared to have less important roles.

This study concluded that parents not only used the information obtained from genetic counselling in making reproductive decisions, but many other factors interacted to arrive at the reproductive outcome.

Interpretation of the findings and implications of the research for nursing practice are presented. Suggestions for future pertinent research are discussed.

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CHAPTER 1

PROBLEMS AND PURPOSES

In North America, there has been a shift of disease patterns from nutritional deficiencies and acute infectious diseases to chronic illnesses, many of which originate from birth defects and genetic disorders. Gordon (1971) estimated that at the turn of the century, one death in approximately twenty-five was due to congenital malformations and in 1971 it was estimated to be one in five. Clow, Fraser, Laberge, and Scriver (1973) reported that 30% of all admissions to a pediatric hospital are associated with genetic disorders. Congenitally inherited disorders are the second leading cause of death in children and account for one third of all pediatric admissions to hospitals; about 50 - 60% of all spontaneous abortions are caused by gross chromosomal defects (Dineen, 1978).

Nurses make up the largest single group of health care professionals and are frequently exposed to individuals and families who have experienced a birth defect or genetic disorder. Concomitantly, in recognition of their broad health care expertise, nurses are often employed as genetic nurses, genetic associates and genetic counsellors in clinical genetics services in Canada and the USA. Nurses and other health care professionals who are involved with clients with

genetic disorders, are interested in how clients utilize the information given in genetic counselling sessions to make reproductive decisions. A study of the influence of genetic counselling on reproductive decision-making should provide nurses and other health professionals interested in genetics with information on the effectiveness of genetic counselling.

Problem Statement

Insight into the dilemma of parents who are required to make decisions under uncertainty would greatly facilitate improvement of the interventions utilized in counselling the families of children with genetic disorders. A better understanding of how clients use genetic counselling information would help put genetic counselling into perspective since this is one of several factors clients do consider in making reproductive choices. Formal and informal evaluation of genetic counselling services, focusing in particular on the influence of genetic counselling on reproductive decision-making, would help improve those services.

The use of a population that is confronted with a single disease entity has been recommended in conducting evaluations of the impact of genetic counselling (Evers-Kiebooms & van den Berghe, 1979). Because neural tube defect (NTD) is a relatively common genetic (multifactorial) disorder in

Newfoundland (Frecker & Fraser, 1987), families of children with this condition, constitute the focus of this study.

A neural tube defect is a malformation involving the neural tube in which failure to close during fetal development results in an open defect (spina bifida) or in severe cases, absence of the brain (anencephaly). The prevalence of NTD varies considerably with geographic location. Internationally, the highest frequency is found in the British Isles (4 to 5 per 1000 live births) with great regional variation within the Isles. For example, in Northern Ireland the frequency is 8 to 9 per 1000, while in London the rate is 3 per 1000 (Leck, 1984). Other countries, such as Japan and Israel, report a low prevalence rate, fewer than 0.5 per 1000 live births (Cohen, 1987).

In the United States, the frequency is 1 to 2 per 1000 births (Cowchuck et al., 1980) while different areas of Canada report varying prevalence rates. The prevalence rate reported in British Columbia is 1.55 per 1000 births (McBride, 1979), Nova Scotia is 2 per 1000 (Winsor & St. John Brown, 1986) and Newfoundland's reported prevalence is 3.2 per 1000 (Fraser, Frecker & Allderdice, 1986). The statistical recurrence risks for NTD also vary, and are based on the multifactorial model of inheritance as well as on what has been observed and reported in a particular geographical location. The recurrence rate for Newfoundland after the birth of one child with NTD

is generally given as 4%, based on the data on frequency and precurrence risk of Frecker & Fraser (1987).

As will be noted in the literature review, there are relatively few studies on genetic counselling published in Canada. Because there are no studies available on genetic counselling and reproductive decision-making in the province of Newfoundland, it is important to identify the impact of sociocultural influences on decision-making. Using Newfoundland and Labrador with its unique population as the context for this study provides such an opportunity.

Purpose of Study

The purpose of this study is to assess how parents of children with neural tube defects (NTD) perceive genetic counselling, as well as other related factors, and how these influences impact on their reproductive decisions.

Conceptual Framework

For the development of the conceptual framework of this study, appropriate models of decision-making were examined and the pertinent elements associated with reproductive decisions highlighted. One of the factors known to influence this decision-making process, genetic counselling, will be the focus. Specifically, the main concepts that will be included are related to 1) content of information offered during genetic counselling, especially recurrence risk, 2) factors

affecting decision making, including genetic counselling, and, 3) elements of models of the decision-making process.

Genetic Counselling

Important goals of genetic counselling are to convey understanding of birth defects and genetic mechanisms to parents so that they can make informed reproductive decisions (March of Dimes, 1980). The subject matter addressed in genetic counselling sessions for clients with a child affected with NTD involves: an explanation of the embryology of the defect of NTD; the multifactorial mode of inheritance; specific recurrence risks for individual families, usually 4% in Newfoundland (Frecker and Fraser, 1987); methods of prenatal diagnosis; and, information on environmental prevention strategies with emphasis on proper nutrition and/or preconceptual vitamin supplementation. This information is presented to the parent(s) in a format matching their level of understanding based on their educational level, interest, and in response to specific questions. The main counselling session usually takes place at approximately three months after the birth of the child. Follow-up sessions are arranged depending upon parental request, the need for additional information or the assessment of the geneticist and/or genetic counsellor.

The anticipated goal of genetic counselling is to offer information to clients that will help them make informed

decisions regarding future pregnancies. Lappe and Brody (1973) have suggested that there would be no rationale for genetic counselling if it were not to influence the behavior of the individuals obtaining the service.

The statistical recurrence risk is often considered to be one of the most important components of genetic counselling (Pearn, 1973) as well as an important factor in helping clients make decisions (d'Ydewalle & Evers-Kiebooms, 1987). Some studies have focused on recall of statistical risk figures as a criterion for evaluating the effectiveness of genetic counselling (Hare, Laurence, Payne & Rawnsley, 1966). However, as reported by Swerts (1987, p. 72) "while the recalled risk figures will influence a couple's decision about pregnancy planning to a certain extent, the subjective evaluation of this recurrence risk may affect the decision process to a larger extent".

The subjective interpretation of the recurrence risk for genetic disease can be viewed in two opposite extremes. Lippman-Hand and Fraser (1979a) found that subjects in their studies perceived the chance of recurrence in binary form - it either will or it will not happen. Lippman-Hand and Fraser (1979b) also found that some clients did not use the probabilities as a basis for action because " no matter the size of the recurrence risk, something can happen - a one in the numerator never disappears no matter the size of the

denominator, and this "one" could be the counselee's child" (p. 332).

Conversely, Pearn (1979) discusses the "risky-shift phenomenon" where the interpretation of risk is subject to shift. Studies by experimental psychologists have revealed that prior discussion of a risk situation leads to an increased willingness on the part of the counselee to take greater risks (Chandler & Rabow, 1969; Horne, 1970; Rettig, 1966). Pearn (1979) contends that the discussion of all aspects of a genetic problem during genetic counselling acts as a catalyst for future reflection in the privacy of the home. Such a view implies that a thorough discussion of the disorder between the genetic counsellor and the parents helps the latters' subjective interpretation to be more realistic and even more objective.

Other Factors Involved in Decision-making

Factors other than the statistical recurrence risk of a particular genetic disorder are important to consider in reproductive decision-making. Although these factors are probably unique for every couple who face a reproductive decision in the face of genetic threat, recurrent factors have been identified. Those that have been reported include the " procreative drive,..., past experience with the disorder and a couple's ethical and moral beliefs" (Thompson, 1986, p. 122).

One's personal past experience with a particular disorder allows one to make a subjective view of the burden of the disorder, which in turn influences significantly the perception of risk associated with that disorder (Black, 1979) and the use of genetic information for reproductive planning (McCollum & Silverburg, 1979). Carter (1966) suggested that previous experience with a disease facilitates the decision to abort another affected fetus and eases the psychological sequelae of abortion.

The severity of the disorder itself is also recognized as an operating factor in influencing decision-making (Pearn, 1973). In the case of NTD, one could say that the implications of the disease for the couple who has lost a baby affected with anencephaly, would differ from that of the couple who is coping with a child affected with a less severe neural tube involvement.

Cultural and personal variables also influence parental desire for a healthy child. Burton (1975) has identified social class, ethnic origin, religious affiliation and financial status as some of these variables.

A person's beliefs regarding birth, death, health care practices, attitude toward parenthood and self-image are some other influential factors in reproductive decision-making (Sorenson, 1974). As well, "the marital relationship, the presence and needs of other children, religious beliefs, attitudes of other family members, [and] career

aspirations,..." (Levine, 1979, p. 124) are among the myriad of influences that determine in one way or another the parents' decisions to procreate. Decision-making is in itself quite a complex process.

Elements of the Decision-making Process

Decision-making is a part of everyday life, but few decisions assume a greater importance for a couple than to have a child in the face of a genetic threat. LaRochelle (1983) stated that there are two distinct types of decision making that a couple wishing to conceive must face. The first is whether to conceive at all; the second occurs after conception and involves the decision to proceed with prenatal diagnosis if indicated, and then to decide whether or not to terminate the pregnancy. These are precisely the types of decisions facing the parents of a child affected with NTD or indeed any genetic disease.

There are a number of proposed models of decision-making; it is this researcher's opinion that none of these models describe accurately the decision-making process of genetic counselling. It was necessary to use elements from two models in order to attempt to concretely explain how parents who have a specific known genetic risk make reproductive decisions.

In the first model, Janis and Mann (1977) offer a five stage schema for decision-making. This is a classic model that

explains the concrete steps in making a rational decision. The five steps include:

1) Appraising the challenge;

2) Surveying alternatives;

3) Weighing alternatives;

4) Deliberating about commitment; and

5) Adhering despite negative feedback.

Stages 2,3 and 4 are particularly applicable to the genetic counselling situation for parents of children with NTD who are deciding whether to have another pregnancy. Stage 2, Surveying the alternatives, involves obtaining information about the statistical recurrence risk of the problem of NTD and other information (e.g. availability of prenatal diagnosis and pregnancy termination services) necessary to make decisions for or against the alternatives.

Weighing the alternatives (Stage 3) leads to a more thorough search and evaluation to confirm the gains and losses to be expected from each alternative. This would require that the decision maker weigh all of the factors that he/she feels are important; the gains and losses for these available alternatives are also reviewed by the decision maker at this stage.

Deliberating about commitment (Stage 4) requires one to ponder about the best alternative for oneself from the available options and, in relation to genetic counselling, to decide if one will proceed with another pregnancy and/or

utilize the course of prenatal diagnosis with its associated considerations.

In the second model, a cognitive perspective which allows for the particular uncertainty one would expect in making a reproductive decision is addressed. Vlek (1987) reported that "it is generally acknowledged among decision theorists that any single decision under uncertainty should not be evaluated by its own consequences" (p. 190). This view supports the contention that there are many variables in the decision-making process that interact to co-determine the final outcome.

Vlek incorporates rational decision analysis, also called Bayesian decision analysis, into a model for genetic decision-making. The Bayesian hypothesis is a rule for revising probabilistic beliefs on the basis of new information. According to this hypothesis, if new evidence results in a change in a probability, the interpretation of the new probability will be affected by the direction of the change (Wertz, Sorenson, & Heeren, 1986). Vlek's model focuses on five steps:

1) Delineating and structuring the problem;

2) Uncertainty analysis and assessment of probabilities;

3) Analysis and assessment of goals, values, and/or utilities;

4) Optimization analysis; identification of the best course of action;

5) Sensitivity analysis: testing the nature of the final decision against variations in the results of steps 2 and 3.

Steps 2,3 and 4 are most applicable to the genetic counselling situation. Uncertainty analysis (Step 2) implies that one attempt to transform one's feeling of uncertainty into quantitative probability estimates assigned to the possible outcome of uncertain factors. This concept, applied to the genetic counselling situation, involves the statistical risk factor that is cited to parents who have had a child born with NTD. A second dimension of this step should lead one to collect additional information which would enhance the probability distribution in one's specific circumstances; this could be related to the additional information given in the genetic counselling sessions concerning embryology, prenatal diagnosis, environmental enhancers and so forth.

Step 3, Analysis of goals, values and/or utilities, involves the assessment of the relative attractiveness or seriousness or specific final consequences of the various options available. This step takes into account other factors such as procreative drive, past experience with the disorder, ethical and moral beliefs; attitudes toward abortion, birth, and parenthood.

Step 4, Optimization analysis, is aimed at the identification of the best course of action from among the options incorporated in one's decision-making. This refers to the decision to have or not to have a future pregnancy and,

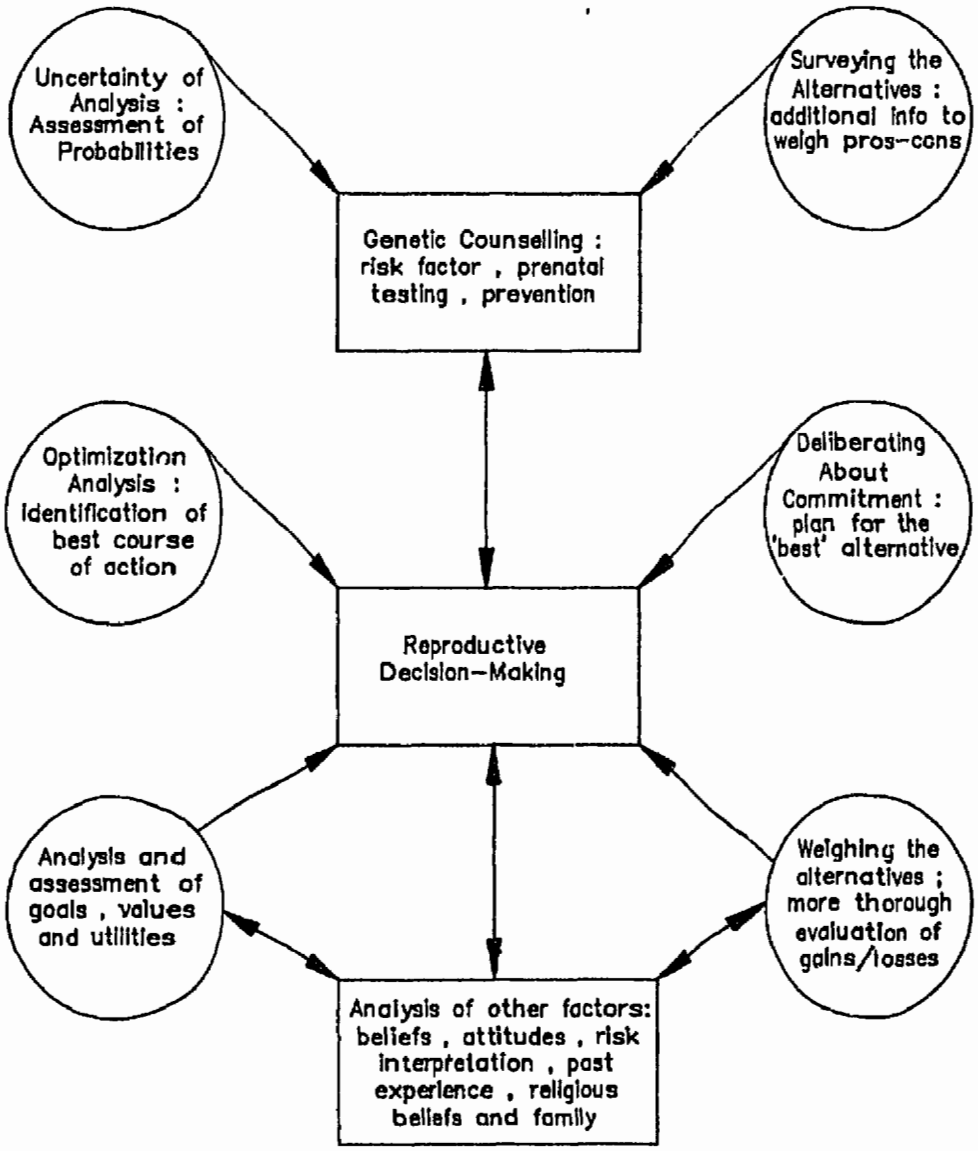
in the event of a pregnancy, the decision to use prenatal diagnosis for more information.

Some authors have difficulty with the rational decision-making model for genetic counselees and Black (1981, p. 15) refers to "the myth of rational decision making". Black believes that because rational models were derived from business and economics, these models are inadequate by themselves for studies of real life decision making. There is as yet no practical conceptual framework that could be utilized by genetic counselees for decision-making.

In this study, concepts from the above two theoretical approaches by Janis & Mann (1977) and Vlek (1987) have been combined into a model entitled "Model of Reproductive Decision-making of Genetic Counselees" (see Figure I). In this model, some of the phases involved in reproductive decision-making for the genetic counselee (as adapted from Vlek and Janis and Mann and depicted in the circled areas of the model) are incorporated. These phases demonstrate a dynamic interchange of the factors involved in decision-making. This dynamic process is not however a linear one, but reflects a continuous interchange of the information the genetic counselee may use in making a reproductive decision (note the double-headed arrows). The boxes in the middle column of the figure represent the conceptual framework of this study. This model notes some of the known variables and allows for the

FIGURE 1

MODEL OF REPRODUCTIVE DECISION—MAKING
OF GENETIC COUNSELEES



Adapted from Viek (1987) and Janis & Mann (1977).

unknown variables that would influence the reproductive decision-making process.

In summary, the information available in genetic counselling can be retained and utilized by the person in qualitatively distinct ways. According to Bringle and Antley (1980), at the primitive level, it is memorized (the risk factor), then it can be conceptualized (understanding) and finally it can be incorporated into decision-making (personalization). These authors propose that this process is a hierarchial one proceeding through these three stages in order to make a personal reproductive decision. In this study, assessment of how parents of children with NTD utilize genetic counselling, and the other outlined influences in making reproductive decisions, will be the focus. The following research questions will attempt to fulfill that purpose.

Specific Research Questions

1) How is the recurrence risk of NTD, discussed during genetic counselling, perceived by parents who already have a child with this condition?

2) How do parents of children with NTD perceive prenatal diagnosis in planning a subsequent pregnancy?

3) Does information given in genetic counselling sessions influence reproductive decision-making of the parents of children affected with NTD?

Definitions

1) Genetic counselling: at least one information session with content regarding embryology of NTD, the multifactorial nature of inheritance, recurrence risks, prenatal diagnosis and prevention strategies.

2) Reproductive decision: the decision whether or not to plan a pregnancy, and/or whether or not to terminate a pregnancy.

3) Parents: a biological mother and/or a father of a child born with a neural tube defect.

4) Perception: as defined by King (1981):" a process of organizing,interpreting,and transforming information from sense data and memory. It is a process of human transaction with environment. It gives meaning to one's experience, represents one's image of reality and influences one's behavior" (p. 24).

5) Risk factor: Following the birth of a child with NTD, provided that there is no significant family history, usually a 4% recurrence risk (Frecker & Fraser,1987) for the birth of another child with NTD.

6) Role: "A function performed by someone or something in a particular situation, process or operation" (Gove, 1986).

CHAPTER II

LITERATURE REVIEW

A large volume of literature on the subject of genetic counselling has been published in the last 30 years with both quantitative and qualitative approaches attempting to describe the efficacy of genetic counselling. Evaluations of the results of genetic counselling are usually influenced by what the investigators deem to be the goals of genetic counselling.

The Nursing literature concerning genetics has focused in general, on the basic principles of genetics with discussion on the implications for nursing (Cohen, 1984; Muir, 1983); the role of nurses in genetic counselling and patient support issues (Farnish, 1988; Fibison, 1983; Fitzsimmons, 1985); problems in the clinical speciality of genetics (LaRochelle, 1983); and, expectations and role development suggestions of genetic nurse clinicians (Tinley, 1987). However, despite the fact that many nurses assume the role of genetic counsellor, there is a paucity of nursing research literature concerning the process of genetic counselling and its effectiveness.

This literature review will focus on 1) a definition of genetic counselling; 2) studies analyzing how clients understand the particular genetic disorder and recurrence to which they are at risk, and, 3) studies related to how clients

utilize genetic counselling in terms of decision-making concerning future pregnancies.

Definition of Genetic Counselling

It is generally accepted among geneticists that the purpose of counselling is to provide families with medical information concerning a specific disease, and the main goal is to inform them of the risks they face in future childbearing. Once families have this information, it is assumed that they will make rational decisions regarding future children.

Leonard, Chase and Childs (1972) contend that the object of genetic counselling is not only to give information, but that it ought to be a form of preventive medicine. This preventive concept allows counsellors to assume that couples running high statistical risks will not reproduce while those having a low risk will be reassured about future pregnancies.

There are many definitions of genetic counselling available in the literature and although similar, they vary to a degree in how the process of counselling is defined and what are viewed as the goals. Shaw (1977) found after a review of 200 articles on genetic counselling, that the aims of genetic counselling could be divided into two broad categories: " to promote societal goals by enccuraging rational decision-making" (to prevent genetic disease) and

"to protect individual autonomy by encouraging counselees to make their own decisions, whether rational or not" (p. 35).

The March of Dimes (1980, p. 5) states that "genetic counselling provides and interprets medical information based on an expanding knowledge of human genetics, the branch of science concerned with heredity". However, the most widely accepted definition of genetic counselling has been formulated by the Committee on Genetic Counselling of the American Society of Human Genetics in 1973:

> Genetic counselling is a communication process which deals with the occurrence, or risk of occurrence, of a genetic disorder in a family. This process involves an attempt, by one or more appropriately trained persons, to help the individual or family:
>
> (a) to comprehend the medical facts, including the diagnosis, probable course of the disorder, and the availability of management;
>
> (b) to appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives;
>
> (c) to understand the alternatives for dealing with the risk of recurrence;
>
> (d) to choose the course of action which seems appropriate to them in view of their risk, their family goals, and their ethical and

religious standards, and to act in accordance with that decision.

(e) to make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder (Epstein, 1973, p. 2).

This definition encompasses both the ideal purpose and the whole scope of genetic counselling. In this descriptive study however, the researcher will be referring to item (d) of this definition as it focuses on one aspect of genetic counselling as a resource to the counselee, that is, as a decision-making facilitator.

Understanding of Disease and Recurrence Risk

A number of earlier studies have described counselees' knowledge about a particular genetic disease and the objective risks that they may have understood. Recurrence risks have been arbitrarily divided by geneticists into three categories: 1) high risk: greater than 1 in 10 (>10%); 2) low risk: less than 1 in 20 (<4%); and 3) medium risk: between these two extremes (Carter, Roberts, Evans & Buck, 1971; Emery, Watt & Clack, 1973).

Emery, Watt and Clack (1972) found that almost all mothers counselled about Duchenne Muscular Dystrophy, between 1965 - 1969, understood the genetics of the condition and remembered the recurrence risks. In a follow-up study, Emery

et al. (1973) found that general counselling cases at the same clinic showed a slightly poorer risk retention rate over time and that two-thirds of the errors were underestimates. Reynolds, Puck and Robinson (1974) found that 84% of 98 parents counselled at the University of Colorado Medical Centre after 1962 were judged to have adequate understanding. Twenty-three percent of these clients remembered the exact risk figures while 60% knew it approximately. The remaining 16% either had inaccurate recollections of the statistical risk figure cited them, did not believe this statistical figure, or were thought to have repressed the counselling experience. It was noted in a study of 27 families that the percentage of parents remembering statistical risks correctly varied with the genetics of the condition, with 82% for autosomal recessive conditions and only 25% for X-linked disorders (Clow, et al., 1973). Somer, Mustonen and Norio (1988) compared 791 families counselled for various conditions during a 10 year period, and found that 80% had adequate knowledge of mode of inheritance and 74% of recurrence risk.

While the previous studies demonstrated adequate retention rates, conversely, some studies revealed variability in the reports of information and recurrence risk retention. Leonard et al. (1972) studied 61 families of children with Cystic Fibrosis, Phenylketonuria (PKU), or Down Syndrome. All the parents in the study had received genetic

counselling whether they requested it or not and from different sources. Responses given by these families led the authors to believe that 1/2 of the families had good understanding, 1/4 had partial understanding and 1/4 had learned very little from the sessions. Hsia and Silverburg (1973) found that 70% of their counselees remembered that a recurrence risk was given but could not state the figure. Ives, Peterson and Cardwell (1973) found that 95% of 63 clients in the high risk groups for a given genetic condition remembered their statistical risk figure more accurately than 40% of the 120 patients belonging to the low-risk group. Pearn and Wilson (1973) have also reported on the nature of the disease and recurrence risk for Werdnig-Hoffman disease (acute spinal muscular atrophy). Although at least 70% of 42 families had received genetic counselling, only 26% knew the autosomal recessive recurrence risk of 1 in 4. Pearn and Wilson interpreted this as a subconscious rejection of more bad news when the parents were trying to cope with the stress of a dying child.

It is difficult to compare these studies and judge the efficacy of genetic counselling on the basis of retention of knowledge due to the fact that these studies vary in many respects including, the genetic disorders counselled, the different counselling techniques, the time elapsed since counselling and the lack of assessment of counselees' knowledge before counselling. In one study that examined

counselees' precounselling knowledge, Seidenfeld and Antley (1981) assessed 47 mothers of children with Down Syndrome who were evaluated on their knowledge of genetic facts pertaining to the condition. They found that more knowledge of the disorder was acquired by counselees before counselling than from actual counselling.

A study specific to parents of children with NTD found that of 47 parents of one year old children with spina bifida, 45 stated that they had received genetic counselling while 43 recalled the recurrence risk correctly (Freeston, 1971). In another study, Morris and Laurence (1976) found that couples who had a very high or a very low recurrence risk tended to remember and understand well the recurrence risk of NTD and that, before prenatal detection was available, counselees tended to accurately remember this risk. After antenatal testing was made available, the counselees apparently no longer made the effort to remember the recurrence risk. The researchers felt that the counselees probably thought recurrence risks were no longer relevant when prenatal diagnosis was available.

Utilization of Genetic Counselling for Decision-making

Any evaluation of the influence of genetic counselling is guided by what the investigators consider to be the goals of counselling. Shaw (1977) stated that "determination of

reproductive behavior is perhaps the most objective criterion that can be measured in follow-up studies" (p. 45).

The following studies review attempts to ascertain how counselling has influenced reproductive decisions. The review of this criterion will be achieved by firstly citing studies that report utilization of genetic counselling as having an influence on reproductive decision-making, followed by research that does not support this assumption.

It is anticipated in diseases that have a high recurrence risk such as cystic fibrosis (CF) that genetic counselling information will be important in preventing the birth of more children affected in a family (Dodge, Burton, Cull & McCrae, 1978; McCrae, Cull, Burton & Dodge, 1973). Dankert-Roelse, te Meerman, Knol and ten Kate (1987) studied 44 CF families to assess the influence of genetic counselling for cystic fibrosis on family planning, using neonatal screening, family size at time of diagnosis and maternal age as possible determinants of reproductive behavior. A 50.8% reduction in childbirth was found in the study group when compared to the control group births born to mothers of equal age and parity.

Other studies of the effects of genetic counselling for cystic fibrosis on reproductive decision-making showed that a considerable proportion of couples embarked on new pregnancies when a high recurrence risk existed (McCrae et al., 1973; Dodge et al., 1978; Leonard et al., 1972). These results may indicate that couples were not utilizing genetic

counselling information, or that they did not understand their statistical risks correctly, or that they were willing to reproduce despite the odds.

In a large study of clients seen in 47 genetic counselling clinics located in 25 states and the District of Columbia, Wertz and Sorenson (1986) found that of 628 clients completing questionnaires six months after genetic counselling, 43.5% reported that their reproductive plans had been influenced by the genetic counselling session. The investigators reported that clients who acknowledged genetic counselling influence did: come to counselling to get information for making a decision about whether to have a child; discuss this decision indepth with the counsellor; and, have a higher educational level than clients who said they were not influenced. These findings are difficult to interpret due to the fact that the subjects were counselled by different counsellors and that a variety of genetic conditions had been involved. The data permitted analysis of self-reports of influence but not of actual utilization of health related information provided in counselling. In interpreting clients' self-reports of influence, the investigators were unable to distinguish between influence resulting from utilization of knowledge and influence resulting directly from advice or direction given by the counsellor.

Reynolds et al. (1974) ascertained what a couple's reproductive plans were before and after genetic counselling.

Of the 97 respondents in their analysis, 41 couples reported having been influenced by genetic counselling. In six instances, couples who had decided not to have children before counselling changed their minds. Twenty-six of the couples came to genetic counselling undecided about attempting future pregnancies but felt reassured after counselling to proceed with another pregnancy. These results support the findings of Townes (1970) that the majority of individuals seeking genetic advice were found to have a relatively low recurrence risk and genetic counselling reassured them about future pregnancies Hsia and Silverburg (1973) found that 26 out of 36 clients wanting more children considered themselves as totally or partially influenced by genetic counselling. Sixty-two percent of 212 families followed-up by Abramovsky, Godmilow, Hirschhorn and Smith (1980) who reached a procreative decision indicated that genetic counselling had influenced their decision-making. Somer et al. (1988) found that 62% of the 791 clients questioned in their retrospective study felt that the counselling had a great or moderate impact on their reproductive plans.

Other research studies fail to show any relationship between genetic counselling and reproductive decision-making. Some studies examined genetic counselling and discovered that it made no significant difference in reproductive plans of some couples. In a controlled retrospective follow-up study of the impact of genetic counselling on parental reproduction following the birth of a Down Syndrome child (DSC), 23 couples

who had received genetic counselling after the birth of a DSC were closely paired by race, religion, maternal age, parental education and sex sibship of the DSC with 23 noncounselled couples who had also had a DSC. Evaluated at least one and one half years after the birth of the DSC or the counselling, the investigators found no significant differences between the counselled and the noncounselled groups in knowledge of general genetics, recurrence risks for Down Syndrome, initiation of subsequent pregnancies or utilization of prenatal diagnosis (Oetting & Steele, 1982).

Two clinical geneticists, Lippman-Hand and Fraser (1979c), utilized a qualitative design (grounded theory) to ascertain how decisions regarding reproduction after genetic counselling were made. They interviewed seven couples postcounselling using open-ended questionnaires and developed a model based on their findings to explain the information processing approach to postcounselling reproductive decision-making. These investigators contend that parents develop a perspective unique to their situation and that their many uncertainties are incorporated into a scenario that allows them to "try out the worst" consequences. This concept permits parents to explore ways of neutralizing the perceived consequences and limits their uncertainties. Apparently, postcounselling reproductive decision-making can most clearly be seen as parents' responses to uncertainty. "The parents' attention focuses not on recurrence rate or burden information

per se, but on how probable and manageable they feel the rate and prognosis are" (p. 337).

Swerts (1987) studied the impact of genetic counselling and prenatal diagnosis upon family planning for parents of a child with NTD. Ninety-four parents who had a child with NTD were divided into 3 groups; one group received genetic counselling, another group had already had an amniocentesis performed and a third group had neither counselling nor an amniocentesis. A significantly better recall of relevant objective risk figures was found in the counselled versus the uncounselled groups and 80% of the counselled group decided to plan a subsequent pregnancy. For more than half the families, the availability of prenatal diagnosis was of crucial importance in the decision to plan future pregnancies.

Another study determined that factors other than availability of prenatal diagnosis and recurrence risk affected reproductive decisions. Steele, Rosser, Rodman and Bryce (1986) compared the reproductive behavior of 132 white married couples of whom 44 had a child with CF, 44 had a child with either DS or NTD, and 44 with Cerebral Palsy (CP) to the reproductive behavior of the general USA, white, married population. The two groups were matched on race/religion, maternal age/paternal age and occupation, and sex of the affected child. Most of the study group had received genetic counselling and all were followed 3 years after the diagnosis of the affected child. These investigators found that,

regardless of the degree of recurrence risk and whether or not prenatal diagnosis was available, couples were likely to reproduce again when the affected child was the first born. They concluded that the three major determinants of reproductive behavior in couples after having a genetically handicapped child were the same as those of the general population: a) parental desire for more children; b) past reproductive experience (sibship size and outcome), and; c) maternal age.

Summary

It is evident throughout this literature review that conflicting results in the research literature indicate that there is no definitive answer as to whether or not reproductive decision-making is influenced by genetic counselling, or indeed how one should measure this influence. In particular, multiple conditions are being evaluated in many of the studies and data have been collected from many centers that may have different mandates for the provision of genetic counselling services. It might be expected that the recurrence risks and genetic burden of different conditions would influence reproductive decision-making in varying degrees.

Models of decision-making (Janis & Mann, 1977; Vlek, 1987) have been presented in the conceptual framework and it is this investigator's view that rational decision-making

cannot be used alone as a basis for reproductive decisions after the birth of a genetically handicapped child.

Lippman-Hand and Fraser's (1979c) results indicate that in order to fit into the rational model, genetic counselling would have to answer three major questions: "How likely am I to have an affected child?", "What would it be like if it happens?" and "How will others react to my choice?" (p. 330).

The present acceptable practice of genetic counselling allows the genetic counsellor to answer some but not all of these questions. The mandate of genetic counselling itself raises questions as to the need for such objectivity as non-directive counselling demands. One may also question the ethical and practical considerations of dissemination of information without analysis of personal meaning for the recipients. The process of genetic counselling however is not the direct focus of this study.

In the present study, the researcher will attempt to investigate the reproductive decision-making of genetic counselees affected by a single disease entity (NTD) and who have been counselled by a single genetics team (clinical geneticist and genetic counsellor). This study will incorporate a unique conceptual framework that attempts to measure genetic counselling as one of several identified variables influencing parental reproduction. It is anticipated that the results of this study may help to fill some of the

gaps in our present knowledge base so that more focused and complete genetic counselling can be provided to counselees.

CHAPTER III

METHODS

The Population and Sample Selection

The population under investigation included parents who 1) have had a child with NTD, and 2) have received related genetic counselling.

The sample consisted of parents who utilized the genetic counselling services of the Janeway Genetics Clinic, situated in St. John's, Newfoundland. In an effort to have a reasonable study sample, a sample consisting of all parents of children affected with NTD and born between May 1984 and June 1987 was selected. Thirty-eight families (two parents or single mothers) met the inclusion criteria and were contacted to participate in the study. Only 36 could be contacted (two families had moved and could not be located) and they all agreed to accept correspondence from the investigator.

Setting and Procedure

This study utilizes a descriptive, retrospective, uncontrolled research design. For the study, a questionnaire was designed by the researcher to be completed by the participants in their homes and returned to the researcher by mail (see Appendix A). Because the target population was distributed over a large geographical area and because of

limited resources for travelling, it was determined that the use of a mail questionnaire would be a convenient approach.

Prior to the beginning of the study, a letter was sent to the Director of the Janeway Genetics Clinic (see Appendix B) explaining the study and requesting permission to contact the families who met the criteria outlined in the methods section. Once permission was granted to proceed with the study, the parents were contacted by the Genetics Clinic secretary - as per the ethical review recommendation and were asked if they were willing to receive correspondence from the researcher.

The researcher then sent to those who were agreed to receive correspondence, a letter of introduction explaining the purpose of the study, and inviting them to participate. At the same time, they were also sent two questionnaires (one for the mother and one for the father, if he was available) with two consent forms which they were requested to complete and return within two weeks in the enclosed, stamped, self-addressed envelope.

In order to avoid the problem of decreased response rate, the potential participants were also informed that if the form was not returned within two to three weeks, they would be contacted by the researcher as a follow-up reminder. At that time, if the participants preferred, the researcher would fill out the questionnaire over the telephone.

The Instrument

A search of the literature revealed that there was no tested, validated instrument to measure the parents' perceptions of genetic counselling. Specific variables were identified by the researcher and selected based on the conceptual model derived from the literature review. These variables were used to design the questionnaire.

Because the questionnaire was new, content validity was ascertained by having three experts in the field of genetic counselling examine it. The instrument was also pretested by a pilot administration to three mothers of children with NTD who did not meet the inclusion criteria for the study (children were born before May, 1984). Based upon their suggestions as well as the suggestions of an expert from the Faculty of Education at Memorial University who reviewed the questionnaire for school grade level comprehension, the instrument was revised.

The questionnaire (see Appendix A) consisted of two sections: Section I was to be completed by the mother. It contained questions related to sociodemographic data such as ancestry, education, occupation and religion. As well, 31 questions were designed to determine 1) the recall of the recurrence risk of NTD; 2) the parent's interpretation of the magnitude of the risk; 3) the impact that genetic counselling had on subsequent procreative decisions; 4) perceptions of prenatal diagnosis; 5) the influence of decision-making

factors other than genetic counselling,and 6) the actual reproductive behavior ensuing genetic counselling.

Section II was to be completed by the fathers, and included the same 31 questions as mentioned before. Many of the questions were designed so that the participants circled the appropriate number corresponding to their selected answer, while some questions gave the participants an opportunity to make additional comments.

Ethical Considerations

The proposal for this study was presented to the Human Investigation Committees at both the Memorial University School of Nursing and the Janeway Child Health Centre. The research proposal was approved by these two committees.

The subjects were asked to sign two consent forms, one of which they could keep for their own records and one to be returned with the completed questionnaire (see Appendix C). Full explanations on the procedures for participating were given to the parents (see Appendix D). They were assured of confidentiality and that they could withdraw from the study at any time without negative consequences to their childrens' care. The questionnaires were coded so that the participants were not identified in any way; all completed questionnaires were destroyed when analysis was complete.

Limitations

This study used a convenience sample in the form of clients who received genetic counselling and who agreed to participate in this study. It is possible therefore that the study population presents unique characteristics that may limit the generalizability of the findings.

As is the case with any retrospective study, a limitation of this study is that the tool required the subjects to rely on their ability to recall some of the information obtained. The circumstances surrounding a birth defect are a traumatic experience. This, in itself, is a significant factor that could influence the accurate recall of information obtained during this period. Depending on individual subject's reactions to and/or acceptance of the birth of a child with NTD, the birth circumstances may have enhanced or diminished recall of genetic counselling information.

The content of this questionnaire examines a socially sensitive area and the participants' desire to provide socially acceptable answers may have biased the results. It would have been possible to control for this bias by using more than one question to examine each perception. However it was determined that this would make the questionnaire too lengthy and may have affected the response level. As well, the parents may have questioned why they were being asked the same question twice and this may have influenced their confidence in the questionnaire.

Statistical Analysis

This study describes the population being examined based on the questionnaire content and the sociodemographic variables. Although the researcher primarily used descriptive statistics, inferential statistics using correlations (Pearson Correlation Coefficient) were also used to determine statistical support for an observation at a significance level of $p \leq .05$. The reported correlations have not been corrected to allow for multiple analysis, resulting in a high Type II error. Conditional transformations, data transformations that construct or alter variables one way for one set of cases and other ways for other sets, were also used in the data analysis (SPSS-X User's Guide, 1988). The frequencies, correlations and conditional transformations were computed (VAX 8800) using the computer program: the Statistical Package for the Social Sciences (SPSS-X).

CHAPTER IV

RESULTS AND DISCUSSION

The results of this study will be presented in three sections. The first section will describe the characteristics of the population (pp. 39-45). The second section (pp. 46-68) will describe parents' perceptions of recurrence risk, prenatal diagnosis and genetic counselling. The third section will identify and discuss other reproductive decision-making influences (pp. 68-73).

A total of 55 subjects, 31 mothers and 24 fathers, participated in the study for an overall response rate of 84% of the mothers and 65% of the fathers eligible to participate. The mothers provided the sociodemographic data for the 31 couples so that these data are available on both parents. Therefore, the response rate for sociodemographic data was 84%.

The data presented in the tables to follow correspond to the responses of the available subjects. Because some subjects chose not to answer every question, the proportion calculations are therefore based on the numbers of responding subjects.

For purposes of comparison, correlational statistics are only carried out on couples (n=24) where both partners participated in the study. Because of the length of the

questionnaire and the multiple independent variables, only significant correlations are reported.

I: Population Characteristics

Parents' Descriptors

All of the parents were Caucasian and the mothers' ages ranged from 19 to 41 years, with the mean maternal age being 29 years (SD=± 5.5;n=28). The fathers' ages as reported by the mothers, ranged from 22 to 42 years with the mean paternal age reported at 32 years (SD=± 4;n=27).

The highest level of education obtained by the majority of parents in the sample was a high school education or higher with 20 of the mothers (67%) and 23 (79%) of fathers achieving this level of education. In addition, 12 (60%) of these mothers and 11 (48%) of these fathers reported that they had completed technical school or university courses (see Table 1).

The educational levels of the participants are slightly higher than those noted by Swerts (1987). She assessed the educational level of 63 parents of children with neural tube defects who received genetic counselling, and found that 54% of the mothers had a high school or greater education and 63% of fathers achieved this educational level.

A possible explanation for this discrepancy might be that the parents who chose to participate in the study were the more highly educated segment of the population under

Table 1

Sociodemographic Data

	Mothers	Fathers
Employment	n=30	n=28
unemployed	19 (63%)	6 (21%)
employed outside the home	11 (36%)	22 (78%)
Religion	n=30	n=30
Roman Catholic	12 (40%)	14 (47%)
Anglican/United	13 (43%)	12 (40%)
other	5 (17%)	4 (13%)
Ancestry	n=30	n=26
English/Irish	21 (70%)	16 (62%)
other	9 (30%)	10 (39%)
Occupation	n=28	n=26
service	1 (4%)	4 (15%)
construction	--	5 (19%)
transportation	--	6 (23%)
clerical	7 (25%)	1 (4%)
scientific/technical	5 (18%)	2 (8%)
homemaker	11 (39%)	--
other	4 (14%)	8 (31%)

Table 1 cont'd

	Mothers	Fathers
Education	n=30	n=29
some schooling	10 (33%)	6 (21%)
completed high school	5 (17%)	7 (24%)
some technical school	3 (10%)	5 (17%)
completed technical school	6 (20%)	6 (21%)
some university courses	5 (17%)	3 (10%)
completed university	1 (3%)	2 (7%)

study. Many of the subjects were relatively young and these findings may also be a reflection of a more highly educated population in general.

The occupations of the subjects were varied with 69% of fathers engaged in service, construction or transportation occupations. While many mothers (39%) reported their occupations as homemakers, 43% were engaged in clerical or scientific/technical occupations.

Forty percent of mothers and 47% of fathers reported their religion to be Roman Catholic while 43% and 40% respectively stated their denomination as Anglican/United.

Seventy percent of mothers and 62% of fathers reported their ancestry as English or Irish; this proportion is similar to the Newfoundland population as a whole (Statistics Canada, 1986). The frequency of NTD in Newfoundland is reported by Fraser et al. (1986) to be 3.2 per 1000 births. Similarly high frequencies are noted in the British Isles at 4 to 5 per 1000 live births (Cohen,1987). It is possible that Newfoundland's relatively high frequency of NTD is partially related to the high percentage of persons with English/Irish ancestry.

Twenty-two couples reported that they used a method of contraception and 64% of these had utilized sterilization.

Affected Child and Siblings Descriptors

Thirty-five percent (n=11) of the children affected with NTD were male and 65% (n=20) were female. This data correlates with the findings of Seller (1987) who reported a 2 to 1 female to male preponderance for NTD. The birth order of the child affected with NTD was determined. Fifteen (55%) of the affected children were the first born in the family, while fourteen (45%) of the affected children had older siblings.

The severity of the NTD of the affected children was determined from their clinic charts. Eleven (36%) of the children had lumbo/sacral involvement, 10 (32%) had thoracic involvement, and 10 (32%) of the children had been affected with anencephaly and had not survived the neonatal period.

Mothers' Reproductive Behaviors

Fourteen (46%) mothers proceeded with pregnancies following genetic counselling and 12 (86%) of these stated that they used prenatal diagnosis to determine if the fetuses they were carrying were affected with NTD. Fifteen children were delivered (one mother reported two pregnancies). None of the families had a second child affected with NTD. Ten (66%) of these post counselling pregnancies resulted in children who were male and the remaining 5 newborns (33%) were female. In addition, two

mothers reported that they were pregnant at the time of the study.

Abramovsky et al. (1980) noted similar results when 41% of couples with a statistical risk of <5% had another child after counselling but only 63% of these pregnancies were monitored by prenatal diagnosis. Somer et al. (1988) found that the number of families with postcounselling pregnancies was higher (57%) when the statistical recurrence risk was low (5% or less). These studies however evaluate counselling concerning a variety of genetic conditions and this may account for the noted differences in decisions to have subsequent pregnancies and utilization of prenatal diagnosis. In addition, as Hsia, Leung and Carter (1979) noted, the burden of the disorder itself is probably an influential factor in perception of risk and consequently reproductive decisions.

Another finding in the present study may also provide support for Hsia's et al. (1979) assumption on the implication of the burden of disorders concerning clients' reproductive behaviors after the birth of a child with NTD. Of the 14 mothers who chose to reproduce, 7 had previously delivered a severely affected child with NTD who had not survived. Although the death must have been a traumatic event at the time, these mothers have not experienced the burden of raising a handicapped child. Fourteen of the sixteen families who chose not to reproduce again had a

living child with spina bifida. This finding may lend further support to the assumption that caregiver burden may influence reproductive decisions. There was indeed a negative correlation noted between the severity of the defect and whether or not families had reproduced since genetic counselling ($p=.032$).

Hsia, et al. (1979) also reported that procreative intentions seemed to be influenced by the sex of the affected child. In the present study, it was noted that when the affected child was female ($n=14$), 71% of families embarked upon another pregnancy.

When the child affected with NTD was the firstborn ($n=15$), 60% of couples reproduced again. In contrast, when the child with NTD was not the firstborn, only 35% of couples had another child. Phelan (1983) found similar results with parents of children affected with cystic fibrosis ($n=207$); when the affected child was the firstborn, 69% of couples had another child and only 22% of couples with children older than the affected children had another pregnancy. Steele, et al. (1986) found that couples were much more likely to reproduce when the affected child was the firstborn rather than the later born, regardless of recurrence risk.

II: Parents' Perceptions

Perception of Recurrence Risk

Recall of Statistical Recurrence Risk

Twenty-nine (97%) mothers stated that the genetic counsellor had told them the numeric recurrence risk of NTD following the birth of their affected child (only one mother reported that the counsellor had not told her the risk). Of these, 25 (86%) reported that they knew or thought they knew the numerical risk. However, only 17 (59%) mothers correctly stated this statistical risk. (As stated before, this statistical risk figure was usually 4%).

Nineteen (79%) of the fathers reported that they were told the numerical recurrence risk (two fathers stated that they did not remember). Seventeen (89%) of these stated that they knew or thought they knew the statistical risk, while 12 (63%) of the fathers recalled it correctly (see Table 2).

Other studies have noted varying degrees of recall of risk figures given in genetic counselling. Swerts (1987) obtained a similar result in her assessment of 63 parents of whom 65% correctly reported the recurrence risk. Evers-Kiebooms and van den Berghe (1979) and Evers-Kiebooms, Vlietinch, Fryns and van den Berghe (1984) found that statistical recurrence risks in general were not well recalled by genetic counselees.

Table 2

Accuracy of Reported Recurrence Risks

Nature of Recall Total Responses	Mothers n=29	Fathers n=19
Correct	17 (59%)	12 (63%)
Underestimate	1 (3%)	1 (5%)
Overestimate	7 (24%)	3 (16%)
Did not recall	4 (14%)	3 (16%)

On the other hand, Abramovsky et al. (1980) and Somer et al. (1988) noted slightly higher recall figures with 72% and 74% respectively, of their study participants demonstrating adequate knowledge of statistical recurrence risks. Reynolds et al. (1974) actually demonstrated an 84% accurate recall figure. Emery, Raeburn, Skinner, Holloway and Lewis (1979) in their prospective study, found that while 77% of their clients remembered the recurrence immediately after the session, this recall figure fell to 53% after two years.

It is important to note that the counselees in this investigator's study had received genetic counselling up to four years prior to the study and this relatively long period for recall may have accounted for the slightly lower retention rates. In addition, Morris and Laurence (1976) noted in their study on neural tube defect counselees that subjects did not make as much effort to recall the numeric recurrence risk when the option of prenatal testing for NTD was available; it is possible that this hypothesis may also exist in the present study. Data were not obtained to verify or deny this assumption.

Subjective Interpretation of Recurrence Risk

The respondents were also asked to give their own views about the subjective magnitude of the risk, that is, the subjective interpretation of the risk of NTD (see Table 3).

Table 3

Subjective Interpretation of Risk

Magnitude of risk	Mothers	Fathers
Total responses	n=31	n=21
Moderate to very high	21 (68%)	11 (52%)
Low to very low	10 (32%)	10 (48%)

Twenty-one (68%) of the mothers perceived their risk of having another child with NTD as moderate to high while 11 (52%) fathers felt the same way. Wertz et al. (1986) reported that the interpretation of risk was a better predictor of client reproductive intentions than was the numeric risk. In the present study, it was found that there was no significant correlation between the magnitude of risk, as subjectively perceived by the parents, and the decision to have another child among either the mothers ($p=.345$) or the fathers ($p=.406$). Wertz et al. also found that clients who were pessimistic about their perceived risk were less likely to plan future pregnancies than were clients who were optimistic.

Other studies have also noted similar results; Swerts (1987) found that 66% of families with NTD estimated that their 3-5% risk figure was high to very high. Hsia et al. (1979) noted that a large proportion of the parents that they questioned (41%) felt that their recurrence risk was "big" and 43% were undecided as to whether they perceived their risk as high or low. These authors felt that the severity or burden of the disorder was a more potent factor than quantitative predictions in a family's perception of risk. In contrast, Somer et al. (1988) reported that 38% of 102 clients whose risk figure was 1-5% perceived that this risk was high while 60% perceived it as low.

One study has found that the presentation of risks as either proportions or percentages may indeed affect the subjective interpretation of risk. Kessler and Levine (1987) asked subjects to compare numerical risks as proportions and as percentages. When presented as unrelated to genetic risk, they found that percentages tended to be chosen as having greater magnitude than their equivalent proportions. However, when these risks were framed as genetic risks, these authors provide empirical evidence that the "linguistic framing of probabilities affects its cognitive processing" (p. 362); they suggest that clients may be using "person-reasoning" (p. 369) in which proportions have a greater magnitude than their equivalent percentages.

In the present study, the genetic risk was presented to the participants as a percentage. In contrast to Kessler and Levine's (1987) findings, these participants do not appear to have used "person-reasoning" (p. 369) to further minimize their risks.

Role of Recurrence Risk in Reproductive Decision-making

When parents were asked to interpret the risk of having another child with NTD in terms of planning future children, 89% of mothers and 91% of fathers felt that the risk had a very important role in their family planning (see Table 4).

Table 4

Role of Recurrence Risk When Planning More Children

Role of recurrence risk	Mothers n=26	Fathers n=21
Very important or important	23 (89%)	19 (91%)
Unimportant or no	3 (12%)	2 (10%)

There was no correlation noted between the role of the risk of NTD when thinking of having more children and the subjective interpretation of risk for either the mothers ($p=.119$) or the fathers ($p=.444$). This finding is in contrast to Evers-Kiebooms (1987) who, in another context, noted a positive correlation between a higher subjective evaluation of risk and an important effect on family planning for cystic fibrosis families.

Analysis of Recurrence Risk

This researcher attempted to examine recurrence risk perceptions using another statistical approach. Using conditional transformations, responses from three appropriate questions from the questionnaire addressing the parents' perception of recurrence risk were combined. Hypothetically, genetic counselling rationale anticipates that parents of children with NTD who have received genetic counselling would 1) know the statistical recurrence risk for this problem, 2) subjectively evaluate this risk as low, and 3) believe that this risk should not have an important role in a decision to plan another child.

When the three corresponding variables in the available data on this question were combined however, it was found that the hypothetical genetic model did not evolve: only one (4%) mother and one (4%) father satisfied these three interdependent factors. Six (25%) mothers and five (21%)

fathers knew the statistical risk, but they perceived it as a moderate to high risk and felt that it had an important role in their reproductive decisions. Interestingly enough, six (25%) mothers and four (17%) fathers who had not recalled the recurrence risk subjectively evaluated this risk as low and stated that it had an unimportant role in a decision to plan another child.

A discussion of these findings will be further examined later.

Perceptions of Prenatal Diagnosis

Availability of Prenatal Diagnosis

Mothers' reproductive outcomes and use of prenatal diagnosis have already been discussed in the first section. Four mothers (13%) in the present study planned another child in the future and 9 (30%) remained uncertain as to their future plans regarding reproduction.

Regardless of whether they had used it or not, the respondents were asked whether prenatal testing should be available to parents who have had a child affected with NTD and whether they would use this testing in a subsequent pregnancy. The opinions of the parents about prenatal testing were very clear; thirty mothers (98%) and 22 fathers (92%) stated that prenatal testing by fetal ultrasound should be available for parents who have had a child with NTD. Twenty-nine mothers (94%) and 20 fathers (84%) agreed

that prenatal testing by amniocentesis should also be available to parents in this situation. When the parents intentions toward the future use of prenatal diagnosis were explored, 90% of mothers and 79% of fathers stated that they would use prenatal testing in a subsequent pregnancy. In contrast, Hsia et al. (1979) found that 78% of 167 families who had a child with spina bifida thought that amniocentesis should be offered to all couples at risk, while only 56% stated they would use the test in a subsequent pregnancy.

Use of Prenatal Diagnosis

Parents have many reasons for wishing to avail of available prenatal diagnosis. The subjects were asked to select their most important reason to use prenatal testing (see Table 5).

While most mothers (96%) and fathers (74%) would use prenatal testing to determine if the baby was affected with NTD, or, to reassure them that the child was unaffected with this condition, a few parents would use the testing so that they could terminate a fetus affected with NTD.

These findings indicate that most parents would use prenatal testing to find out and/or be reassured that their children were unaffected with NTD. There was however a significant difference in couple agreement in respect to

Table 5

Most Important Reason to use Prenatal Testing

	Mothers n=25	Fathers n=19
To find out if child affected with NTD.	11 (44%)	7 (37%)
To be reassured that the child is unaffected with NTD.	13 (52%)	7 (37%)
To terminate pregnancy if child affected with NTD.	1 (4%)	5 (26%)

termination of pregnancy. While only one mother felt that she could terminate a fetus with NTD, 5 fathers (26%) felt the same way.

Evers-Kiebooms, Swerts and van den Berghe (1988) noted that not all mothers were reassured by a prenatal test result that was negative for NTD. In an assessment of the psychological aspects of amniocentesis, they found that women who had given birth to a child with NTD more frequently reported concerns about an unfavorable result than did mothers who were having the amniocentesis for other problems (advanced maternal age or a previous child with Down syndrome). A high percentage of the mothers who had given birth to a child with NTD (50%, n=42) were also not reassured by the results of the test as opposed to 25% of the other mothers. The authors hypothesized that the results of the amniocentesis (usually a chromosome analysis) were more conclusive for the mothers whose child was not affected by NTD; the alpha feta protein test performed on the amniotic fluid for NTD can only detect open lesions and therefore the possibility of a false negative test may be a concern to these mothers.

Opinions About Pregnancy Termination

Subjects were also asked their opinions about termination of pregnancy in general and had opportunity to give their comments about this subject (see Table 6).

Table 6

Opinions About Termination of Pregnancy

Total Responses	Mothers n=28	Fathers n=23
No termination	11 (39%)	6 (26%)
Termination if mother wishes	5 (18%)	4 (17%)
Termination if fetus has a genetic defect	7 (25%)	8 (35%)
Other comments	5 (18%)	4 (17%)

While 39% of mothers and 26% of fathers did not agree with termination of a pregnancy, 18% of mothers and 17% of fathers agreed with termination if the mother wished and 25% and 38% of these respective groups agreed with termination of pregnancy if the fetus had a genetic defect.

The couples appear to be more in agreement in their opinions about termination of pregnancy in general than when asked about termination of pregnancy if expecting a child affected with NTD (see Table 5). It is possible that mothers had a problem agreeing with the termination of a fetus with the specific defect of NTD because they already have had a child with that problem. Asking about termination of pregnancy in general may allow them to impart a more impersonal view on pregnancy termination when referring to genetic defects in general.

Other parental comments about termination of pregnancy were: would need to be in the situation to be able to make a decision about termination; if the mother's life was at stake, would agree to termination; the issue is more complex; mixed feelings; opinion is related to a person's beliefs; and depends on the seriousness of the defect.

Davies (1983) states that parents who already have a child with a genetic defect may experience a conflict when they consider the abortion of a fetus with a similar problem. They may be concerned that it will affect their relationship with the first child particularly if and when

the child is able to understand the implications, that he/she might have not have been a wanted child because of a disability. For some of the families in the study this scenario would not be applicable because their affected children were severely affected with NTD (anencephaly) and did not survive the neonatal period. The severity of the affected child's condition did indeed appear to have affected the opinions about termination of pregnancy. Over 50% of parents who agreed to termination of a subsequent pregnancy if the fetus had a genetic defect had previously had a child affected with anencephaly. While it was not determined if these parents had the option to terminate the original affected pregnancy, these findings may suggest that these parents would not wish to proceed with a pregnancy when it was known that the outcome would be a child who might not survive.

Blumberg (1984) does propose that upon the discovery of a fetal defect, "the wish to avoid the anticipated burden of bearing a defective child usually supersedes the desire for a child and may lead the couple to interrupt pregnancy even in the face of preexistent morals or religious objections to abortion" (p. 211).

This researcher speculated that in this province where a large percentage of the population is Catholic, one might expect a relationship between religious affiliation and attitudes towards termination of pregnancy. However, there

was no correlation noted between religious affiliation and termination of pregnancy for either the mothers ($p=.155$) or the fathers ($p=.324$).

Analysis of Perceptions of Prenatal Diagnosis

To further analyze the perceptions of prenatal diagnosis, three questions addressing this topic were combined to produce a new variable. This new variable combined questions that ascertained whether parents thought prenatal diagnosis should be available, if they would use it and the reason they would utilize this procedure.

Out of 22 couples, 18 (75%) mothers and 13 (54%) fathers reported that prenatal diagnosis by ultrasound and amniocentesis should be available to parents who have had a child with NTD, and they would use it in a subsequent pregnancy to find out if their child had NTD or to reassure themselves that the child was unaffected.

Only one (4%) mother felt that the prenatal techniques should be available so that she could terminate the pregnancy if the child was affected with NTD while five (21%) fathers felt that way.

Perceptions of Genetic Counselling

Genetic Counselling Influence on Family Plans

Parents were also asked their perceptions about the genetic counselling they had received and its influence on

their reproductive decisions. Subjects were asked if their family plans had been changed by genetic counselling. Fifty percent of both mothers and fathers reported that their family plans had not been changed while the remainder reported that they were influenced or were uncertain about the influence of genetic counselling upon their family plans (Table 7).

Although only 27% of mothers and 29% of fathers reported changes in family plans due to genetic counselling, only about half of the subject reported no influence from genetic counselling, while the remainder reported uncertainty. These results support the conceptual framework which notes genetic counselling as just one of many factors that clients use in decision-making.

Some studies have reported a higher degree of influence of genetic counselling. Abramovsky et al. (1980) reported that 62% of 212 families indicated that genetic counselling had influenced their decision making. Morris and Laurence (1976) found that only 22% of couples counselled for neural tube defects thought that the counsellor was the strongest influence in reaching their decisions.

Reproductive Decisions Following Genetic Counselling

In order to explore reproductive intentions and the influence of genetic counselling on these plans, parents were specifically questioned about reproductive decisions

Table 7

Family Plans Changed by Genetic Counselling Sessions?

	Mothers n=30	Fathers n=24
Yes	8 (27%)	7 (29%)
No	17 (57%)	12 (50%)
Uncertain	5 (17%)	5 (21%)

after genetic counselling and if genetic counselling information/guidance had affected these decisions. Table 8 summarizes the responses obtained.

Again, these results indicate that genetic counselling is but one influence that genetic counselees utilize in making reproductive decisions. Other important reproductive decision-making influences will be explained in the next section. Other studies have reported higher degrees of influence of genetic counselling. Somer et al. (1988) found that 62% of their respondents felt that counselling had a great or moderate influence on their reproductive plans and within this group 62% had postcounselling pregnancies. Swerts (1987) reported that 80% of parents who had a child with NTD decided to plan another pregnancy after genetic counselling.

Family Size Limitations

In this study, of the 14 mothers who planned to have more children after genetic counselling, 9 planned the same size family as before counselling, 4 planned fewer children and one mother reported that she planned more children. Of thirteen fathers who reported plans to have more children after genetic counselling, 7 planned the same size family and 6 planned fewer children than before counselling.

Table 8

Plans to Have Children After Genetic Counselling and Genetic Counselling Influence

After genetic counselling	Mothers n=29	Fathers n=23
Planning more children	14 (48%)	13 (57%)
Planning no more children	8 (28%)	6 (26%)
Uncertain	7 (24%)	4 (17%)
Influence of genetic counselling	n=28	n=22
Influenced	6 (21%)	11 (50%)
Not influenced	17 (61%)	9 (41%)
Don't know	5 (18%)	2 (9%)

As shown in Table 9, 48% of mothers and 44% of fathers thought that the condition of NTD had influenced their family planning.

Although one of the aims of genetic counselling is to reassure parents about future pregnancies regarding NTD (Townes, 1970), these findings may indicate why many of the couples did not plan to have children after genetic counselling. It is possible that parents were not reassured by genetic counselling and that they still had many reservations or concerns about the recurrence risk for this problem. These findings also support those reported by Hsia et al. (1979) regarding the reproductive influence of the burden of the genetic condition. These results are also consistent with Lubs (1979) who found that 43% of 199 families who had children with hemophilia or muscular dystrophy thought that these conditions had influenced their family planning.

Analysis of Genetic Counselling

Again, three questions were combined to create a new variable addressing the parents' perceptions of genetic counselling. This variable combined questions related to parents' reports of influence regarding 1) family plans changed by genetic counselling, 2) decision to have more children influenced by genetic counselling, and 3) role of

Table 9

Feeling About Limiting Family Size due to NTD in the Family

	Mothers n=29	Fathers n=23
No influence on number of children	14 (48%)	10 (44%)
Limit family size.	14 (48%)	10 (44%)
Do not know	1 (3%)	3 (13%)

the risk of NTD in future reproductive decisions.

Eight mothers (33%) and 8 (33%) fathers who 1) felt the decision to have more children was affected by, and 2) perceived that family plans had been changed by (or were uncertain about the effect of) genetic counselling, also felt that the role of the risk of NTD was an important factor in a decision to have another pregnancy. Nine (38%) mothers and 5 (21%) fathers who reported that the decision to have more children was not affected by, and family plans were not affected by genetic counselling, also felt that the role of the risk of NTD had an important influence on reproductive behavior. These results point out the consistency of the influence of the recurrence risk of NTD for these families in planning future children.

III: Decision Making Influences

There could be many hypotheses generated as to how parents make reproductive decisions. Although the findings of this study indicate that the majority of parents do not perceive genetic counselling as being influential in decision-making, it is evident that they utilize the information that they obtain in genetic counselling in making these decisions. The majority of parents perceived the recurrence risk factor as having an important role in planning future children. As well, the parents were very

positive about the use of prenatal testing for subsequent pregnancies. These two topics are a major component of the information given to parents who receive genetic counselling for NTD.

In an attempt to ascertain the role of other factors in family planning, the subjects were asked to rank the importance of certain factors in reproductive decision-making. The results that were rated as "somewhat" or "very important" are combined in a histogram presented in Figure 2. For this comparative analysis, only the responses of the couples (n=24) who participated in the study were used.

This list of reproductive considerations attempted to cover a broad spectrum of topics. Some considerations such as "fulfillment as a parent" and "number of children you should have" were oriented towards personal parenting goals. Some considerations speculated about the family as a social unit, such as "part of being married" and "carrying on the family line". Other items reflected more practical considerations, such as "taking care of other children", "career goals" and "impact on social life". It is clear that parents consider a variety of factors in planning pregnancy and some are more likely than others to be viewed as more important.

With a few exceptions, parents revealed that similar factors influenced their reproductive decisions. The data shown in Figure 2 demonstrate that "the wishes of their

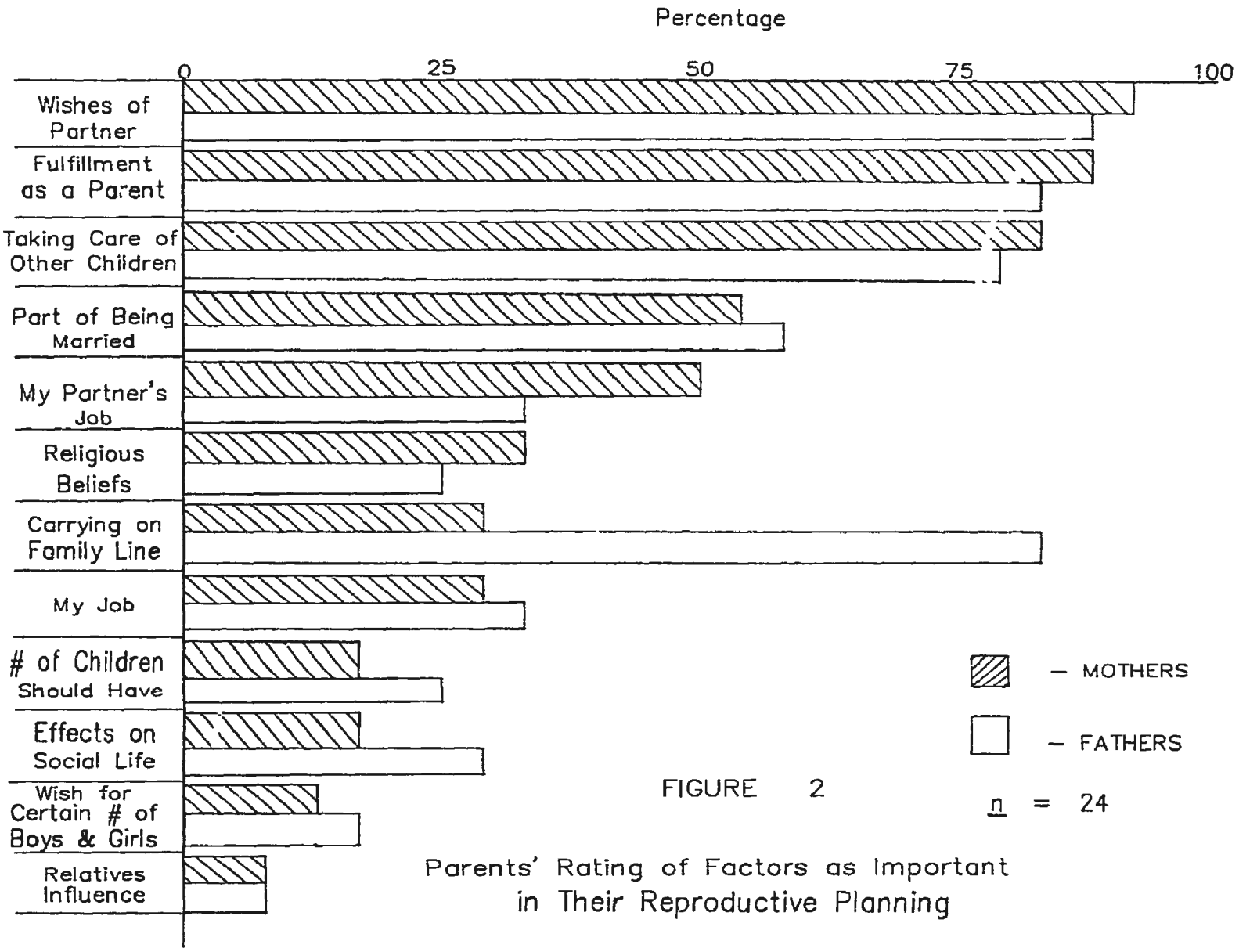


FIGURE 2

Parents' Rating of Factors as Important in Their Reproductive Planning

partner" was the most frequently cited factor that was rated important or very important by the parents. "Fulfillment as a parent", a personal parenting goal, was cited as important by almost as many parents. A more practical consideration, "taking care of other children", was of similar importance to the preceding factors. The fathers ranked "carrying on the family line" almost as important as these other considerations.

Over half the parents cited "part of being married", and half of the mothers reported the "partner's job" as important factors in reproductive plans.

When examining the most frequently cited considerations, it is evident that most parents cited a mix of personal, practical and interpersonal considerations in making reproductive plans.

Seventeen to 33% of parents reported "religious beliefs", and "my job" (mothers reported "carrying on the family line") as important. Finally, there were four factors, including "number of children you should have", "effects on social life", "wish to have a certain number of boys and girls" and "influence from relatives", that were rated as less important.

Clearly, parents consider many factors in their reproductive planning and these factors interact to influence the final outcome or reproductive decision.

A similar assessment by Sorenson, Swazey and Scotch (1981) was carried out on a large sample comprised of clients with assorted genetic problems from many clinics. With over 1000 mothers responding to this question, Sorenson et al. found that "fulfillment as a parent" was rated most often as important in reproductive decision-making, followed by "taking care of other children", and "wishes of partner". Twenty-one to 40% of these mothers cited "completing one's marriage", "financial concerns", "spouses career goals" and "religious or ethical concerns" as important. There were six considerations in Sorenson's et al. study that were less likely to be rated as important, including "carrying on ones family line", and "achieving an ideal family size" and "sex ratio".

Although the same three factors were rated as the most important in both studies, the ranking of these three factors by the participants varied. The high priority given to "the wishes of one's partner" as a decision making influence may be a reflection of the desire of parents to preserve the cohesiveness of the family unit. This finding may be reflective of a cultural expectation in this province where the nuclear family is still essentially the norm.

Although the respect for "the wishes of ones partner" takes precedence over the personal parenting goal of "fulfillment as a parent", this variable is also related to the family as an important concern for parents.

A high percentage of the fathers ranked "carrying on the family line" as very important as well. This too may indicate a cultural, or possibly a male expectation, that one should procreate and again, preserve the family.

Parental Comments

Because opportunity was given for comment, some additional information was obtained which could potentially expand clinical insights and possibly provide some direction for future research.

Parents were asked if the genetics clinic could be of more assistance to them and were asked to comment on how the genetic counselling sessions could respond to this need. Although some parents answered "no" to the offer of more assistance, other comments were explicit and included: requests for additional information to be sent on the subject; requests for additional counselling when planning future pregnancies; and, requests for counselling for other family members and offspring.

Some of the suggestions from parents included: review of counselling after one year; more public education about the problem; further research to determine the cause of NTD; group counselling sessions; provide written summary of counselling session(s); explain in simpler terms; and, more frequent sessions.

Other positive comments included: "clinic has already helped", and, "sessions were very adequate".

A few parents were more negative in their comments stating: "clinic could not give us the exact cause of this problem"; "be more specific about the problem"; and "could be more informative".

General Discussion of Research Findings

In summary, 46% of the mothers in the study had another child after genetic counselling and two were pregnant at the time of the study. These decisions to proceed with more pregnancies were probably influenced by several factors including the sex of the affected child, whether or not the affected child was the first born, and the severity of the defect of the affected child.

The three research questions posed at the beginning of this study were addressed.

How is the recurrence risk of NTD, discussed during genetic counselling, perceived by parents who already have a child with this condition?

Although most parents reported that they were told the statistical recurrence risk for NTD by the geneticist, only about 60% of parents could recall the exact risk figure. Most of the errors were overestimates. The recall of risk

figures may have been influenced by the time elapsed since counselling and/or the availability of prenatal diagnosis for this condition.

Although the objective risk of recurrence of NTD is generally thought to be low at 4%, (Carter et al. 1971; Emery et al. 1973) many mothers (68%) perceived their risk as moderate to high, while slightly fewer fathers shared these views. It was speculated that the perceived burden of this disorder has a significant effect on the perception of recurrence risk for this condition.

Many of the parents also stated that this perceived recurrence risk was very important in planning future pregnancies. It was not determined in what way this information was important in reproductive planning.

How do parents of children with NTD perceive prenatal diagnosis in planning a subsequent pregnancy?

Almost 100% of parents agreed that methods of prenatal testing (amniocentesis and fetal ultrasound) should be available to parents who have had a child with NTD. Most of these parents also reported that they would use prenatal testing in a subsequent pregnancy. The findings indicate that 86% of parents who had a subsequent pregnancy stated that they actually used prenatal diagnosis.

A large proportion of parents stated that the reason to

use prenatal testing in a subsequent pregnancy would be to determine if the fetus was affected with NTD, or, to reassure them that the fetus was unaffected with this condition. Only one mother stated that she would use prenatal testing to find out if the child was affected with NTD so that she could terminate the pregnancy, while 26% of the fathers felt this way.

However, while many parents were definite or ambivalent in their opinions about not terminating a pregnancy, 43% of mothers and 52% of fathers could justify termination of pregnancy if the mother wished or if the fetus had a genetic defect.

Does information given in genetic counselling sessions influence reproductive decision-making of parents of children affected with NTD?

Parents reports of the perceived influence of genetic counselling indicate that they generally feel that genetic counselling has not changed their family plans. Over 50% of parents stated that they planned to have more children after genetic counselling and most of these couples did have a subsequent pregnancy. Most parents disclosed that genetic counselling had not influenced their reproductive decisions, while 50% of fathers stated that they were influenced. These findings may indicate that more fathers than mothers felt

reassured by genetic counselling while mothers remained concerned about their reproductive outcomes.

Actually, almost half of th.. parents felt that they would limit the size of their families because of this condition in their families.

Despite these parental reports of minimal influence of genetic counselling in reproductive decision-making, there is evidence that parents are using the information obtained in genetic counselling sessions to facilitate their reproductive decision-making.

The data in this study indicates that the recurrence risk of this problem, one of the most important aspects of genetic counselling, is reported to be important to parents in planning more children. Another component of genetic counselling sessions, the education of clients concerning prenatal diagnosis, appears to be well received by parents. It is possible that if these parents had not received genetic counselling, they might not have obtained accurate information from other sources.

Influence of Other Decision-making Factors

The results of this study have provided nurses and other health care professionals with information on additional factors that influence reproductive decision-making of genetic counselees. The family unit and how it functions seem to be the paramount concern of the parents in

this study. In respect to this concern for the family unit, parents stated that the wishes of their partners is the first most influential factor. Fulfillment as a parent, the second most important factor rated in reproductive decision-making, is both a personal and familial goal. The third most important factor, taking care of their other children, would also appear to reflect the desire of these parents to preserve the family unit.

It should be noted that although correlations have been referred to in the findings, these are pair-wise correlations and cannot be seen as independent of each other. In essence, the reported probabilities would have to be disregarded if they were corrected for their lack of independence and would need to be many times less than $p=.05$ to achieve true significance. The reported correlations can only be considered in acknowledgement of a high Type II error.

CHAPTER V

SUMMARY, IMPLICATIONS AND RECOMMENDATIONS

Genetic counselling is a service offered to individuals with a family history of genetic disorders and/or birth defects. The purpose of this investigation was to assess how parents of children with neural tube defects perceive genetic counselling, as well as other related factors, and how these influences impact on their reproductive decisions.

Insight into how parents make reproductive decisions under uncertainty is a critical objective in the provision of genetic counselling services. Factors believed instrumental in determining the efficacy of genetic counselling were chosen from the literature to highlight important decision-making factors for health professionals interested in genetic counselling.

Information from such a study has implications for the provision of appropriate genetic services by providing a broader understanding of clients' perceptions of genetic counselling and other reproductive decision-making influences.

A descriptive design was selected for this investigation. Parents of children affected with NTD, and who received genetic counselling for this condition comprised the target population, and the target sample consisted of all those parents who had received genetic counselling for NTD within a prescribed time period. The study sample consisted of 36

families who had a child with NTD and who had received genetic counselling for this study. Fifty-five subjects (31 mothers and 24 fathers) from 31 families participated. The subjects completed a mailed questionnaire which was designed by the researcher to elicit knowledge relating to the recurrence risk of NTD, perceptions of prenatal diagnosis, the impact of genetic counselling on subsequent procreative decisions and the influence of other decision-making influences.

Findings of the study indicate that although parents reported minimal influence of genetic counselling on reproductive decision-making, they do appear to be using the information obtained during genetic counselling to facilitate their decisions. They also appear to be using a myriad of other family oriented factors in making reproductive decisions including the wishes of their partners, fulfillment as parents and taking care of their other children. Other influences such as religion, relatives' influence, and career goals appear to have less important roles.

In conclusion, this investigation was designed to explore parental perceptions of the influence of genetic counselling on reproductive decision-making. The parents were surveyed to obtain information pertaining to certain aspects of genetic counselling. The data obtained provided support for the conceptual framework as illustrated in Figure 1. Parents not only use genetic counselling in making decisions about

reproduction, but many other factors interact in their reproductive decision-making.

Implications

The findings of this study have implications for nursing practice, theory and research.

Nursing Practice

Nurses who are employed in genetic counsellor roles need to be more aware of factors that contribute to the reproductive decision-making of their clients. Examination of these influences with the client may help to further facilitate informed decision-making and should be a part of genetic counselling sessions.

The reported perception of a majority of parents of children with NTD that prenatal diagnosis should be available and would be used, suggests that nurses working in any health related field need to be aware of the availability of such services for their clients. Nurses should pay particular attention to clients who are affected by genetic problems who have not received genetic counselling services. These clients may not be aware of prenatal testing options to facilitate their reproductive decisions. Referral of clients to genetic counselling services, so that they can make informed reproductive decisions should be an important aspect of the care of clients in all health care settings.

Nurses consider the family to be the primary unit of health care. This study found that the family unit and family interactions appear to be important factors for parents who are about to make reproductive decisions. To foster health care at the family level, nurses need to be cognizant of identified factors that impact on reproductive decision-making for the family. In particular, since the family, especially the wishes of one's partner, appears to play such an important role in reproductive decision-making, nurses could help both parents to be aware of this factor during genetic counselling sessions. Nurses could encourage genetic counselees to discuss the information obtained during genetic counselling and to share the reproductive decision.

Nursing Theory and Research

Nursing theory will gradually be developed through the testing of theoretical relationships. This researcher has made an attempt to establish conceptual relationships through the adaptation of various models for reproductive decision-making. The conceptual framework which guided the investigator was supported by the results of the study. The conceptual framework outlined how genetic counselling information, especially recurrence risk and prenatal diagnosis, as well as other decision-making factors influenced reproductive decision-making for genetic counselees (see Figure 1). The study provides support that although a majority of parents

with NTD do not report that genetic counselling influenced their reproductive plans, they do appear to be using the information obtained from genetic counselling in reproductive decision-making. The risk of recurrence was identified by most parents as an important factor in making these decisions. The availability of prenatal testing was also valued by parents and most of them would use it in a subsequent pregnancy.

The analysis of other factors that influence reproductive decision-making is noted in the third box of the model (Figure 1). The parents revealed that they consider the wishes of their partners, fulfillment as parents and taking care of their other children as important reproductive decision-making influences. It would be helpful to apply this model to clients who have other genetic conditions to identify if these particular decision-making influences are considered important for all genetic counselees, and not only for parents who have a child with NTD. The conceptual framework identifies the dynamic interchange of these factors as they interact to allow parents to arrive at a reproductive decision.

A replication of this study would help further determine the scientific validity of the model for reproductive decision-making for genetic counselees.

Because nursing is a science that formulates "the diagnosis and treatment of human responses to actual or potential health problems" (American Nurses Association, 1980), studies such as this one could provide the practitioner

with some insight into client responses to reproductive dilemmas.

Recommendations

Modification of Present Study

Some suggestions for revisions to the present study are:

- A larger sample of parents.
- Changes to the present instrument so that specific questions related to content areas of genetic counselling for NTD could help determine the usefulness of these sessions and how the presentation of this material could be enhanced.
- Additional questions so as to: clarify the direction (positive or negative) to which information obtained from genetic counselling influenced reproductive decisions.
- Identification of parents' perceptions of the helpfulness of genetic counselling sessions.

Suggestions for Future Research

It is necessary that future studies orient themselves to the effects of counselling on other social and psychological factors which may significantly affect the manner in which genetic counselees interpret, accept and act upon the information and advice they receive in the genetic counselling sessions.

A study is needed that will specifically focus on parental recall of recurrence risk figures when prenatal

diagnosis is known to be available for a particular condition.

Other studies could be done to measure if genetic counselling provides relief of psychological distress and/or how this could be achieved by health care professionals.

While it is evident that a number of psychological and practical factors are influencing the retention and personal evaluation of genetic counselling, research is needed to further delineate psychological mechanisms that affect reproductive decision-making.

Studies that utilize a qualitative methodology may be an alternative to identify the personal meaning of giving birth to a child with a defect, living with such a child, and self esteem considerations. Nuances may emerge from research of this level which are difficult to obtain from questionnaires. Topics such as quality of feelings, emotional overtones and better insights into the effect of NTD on the marital relationship, may be better addressed by qualitative methodologies.

These potential studies could provide for a more comprehensive picture of the process of decision-making and add to the nursing knowledge base on how individuals, families and couples perceive and cope with the burden of genetic problems.

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Appendix A

Dear Parent(s);

Thank you for agreeing to take part in this study,

This questionnaire is divided into two sections, I and II. Most of the questions require you to circle an answer and a few questions provide you with an opportunity to comment.

SECTION I: FOR MOTHER ONLY: This section includes general background information, but does not reveal any identifying information about you. It also contains questions related to your family planning, genetic counselling and your beliefs about prenatal diagnosis.

SECTION II: FOR FATHER ONLY (if possible): This section contains questions related to your family planning, genetic counselling and your beliefs about prenatal diagnosis.

I would appreciate it if you would both fill out the questionnaires separately.

Your participation is appreciated.

SECTION I:(FOR MOTHERS ONLY)

Please circle the number that corresponds to your answer(s) or fill in the blank spaces.

1) Date_____ 2) Location: _____
(Name of city or town)

3) From where did your ancestors come?

England.....1
France.....2
Ireland.....3
Canadian Innu.....4
Canadian Innuit.....5
Other (specify).....6
Don't know.....7

4) From where did the father's (of your child with NTD) ancestors come?

England.....1
France.....2
Ireland.....3
Canadian Innu.....4
Canadian Innuit.....5
Other (specify).....6
Don't know.....7

5) Your Religion:
Anglican.....1
Baptist.....2
Jewish.....3
Pentecost.....4
Roman Catholic.....5
Salvation Army.....6
Seventh Day Adventist.....7
United Church.....8
Other.....9

6) The religion of the father of your child with NTD:
Anglican.....1
Baptist.....2
Jewish.....3
Pentecost.....4
Roman Catholic.....5
Salvation Army.....6
Seventh Day Adventist.....7
United Church.....8
Other.....9

7) Your Education Level:
Some Schooling.....1
Completed High School.....2
Some Technical School Courses.....3
Completed Technical School.....4
Completed University Courses.....5
Completed University Degree.....6

8) The father's Education Level:
Some Schooling.....1
Completed High School.....2
Some Technical School Courses.....3
Completed Technical School.....4
Completed University Courses.....5
Completed University Degree.....6

9) Mother's Occupation _____ _____

10) Are you presently employed?
Yes 1
No.....2

11) Father's Occupation _____ _____

12) Is he presently employed?
Yes.....1
No.....2

13) Your Children:	Age	Sex
Oldest:		Male..1 Female..2
Next:		Male..1 Female..2
Next:		Male..1 Female..2
Next:		Male..1 Female..2
Next:		Male..1 Female..2
Next:		Male..1 Female..2

* Questions #14 to 38 are about the history in your family of NEURAL TUBE DEFECT , better known as spina bifida or anencephaly. In this questionnaire, we are using the abbreviation NTD to cover this condition.

14) How many brothers are OLDER than the child who has/had NTD?__
How many sisters are OLDER than the child who has/had NTD?___

15) How many brothers are YOUNGER than the child who has/had NTD?____
How many sisters are YOUNGER than the child who has/had NTD? ____

16) What is the sex of your child affected with NTD?
Male.....1
Female.....2

17) How many children did you plan to have BEFORE your child was born with NTD? _____

18) When you came for genetic counselling, did the counsellor tell you what your chances were of having another child born with an NTD birth defect?
Yes.....1
No.....2
Do not remember.....3

19) If your answer to Question 18 is Yes, what are your chances of having another child with NTD?
1) I know the chances are _____percent.
2) I think the chances are _____percent.
3) I do not remember.....3

20) After genetic counselling, what do you think is your chance of having another child with NTD?
Very high.....1
High.....2
Moderate.....3
Low.....4
Very low.....5

21) Have you looked for information about the risk of NTD other than from genetic counselling?
Yes.....1
No.....2
Do not remember.....3

22) If YES to Question #21, circle the source from which you found more information?
Family.....1
Friends.....2
Nurses.....3
Family doctor.....4
Other doctor.....5
Newspaper article.....6
Other readings.....7
Spina Bifida Association.....8
Other (specify).....9
_____

23) Have you had another child since you received genetic counselling?

Yes.....1
No.....2
(If NO, skip to Question 26.)

24) If YES to Question # 23,how many children have you had since?

One.....1
Two.....2
Three 3
Four.....4
More than Four (specify).....5

25) If YES to Question #24,did you use prenatal testing to find out if the child you were carrying was affected with NTD)?

Yes.....1
No.....2

26) When or if you were thinking about having more children , your risk of having another child with NTD had (has):

A very important role in your decision.....1
An important role in your decision.....2
An unimportant role in your decision.....3
No role in your decision.....4

27) Do you intend to have a child in the near future?

Yes.....1
No.....2
Undecided.....3

28) Do you think that your family plans have been changed by your genetic counselling session(s)?

Yes.....1
No.....2
Do not know.....3

29) What is your feeling about limiting your family size because of the NTD in your family? (please circle one statement only).

I feel that the condition has not influenced the number of children I have 1
I feel that the condition has not influenced the number of children I am planning to have.....2
I feel that because of the condition, I have decided to limit my family size.....3
I do not know.....4

30) Are you or your partner using any of the following methods of birth control at this time to limit your family size?

The pill.....1
An IUD (coil).....2
Condoms.....3
Diaphragm.....4
Foam.....5
Tubal ligation.....6
Hysterectomy.....7
Vasectomy.....8

31) Are you pregnant at this time?

Yes.....1
No.....2
Don't know.....3

32) Should prenatal testing by fetal ultrasound be available to parents who have had a child with NTD?

Yes.....1
No.....2
Don't know.....3

33) Should prenatal testing by amniocentesis be available to parents who have had a child with NTD?

Yes.....1
No.....2
Don't know.....3

34) If you were planning another pregnancy, would you plan to use prenatal testing to find out if the child you were carrying had NTD?

Yes.....1
No.....2
Don't know.....3

35) If YES to Question #34, check the MOST IMPORTANT reason for you to use the test (one answer only):

1) to find out if the child was affected? []
2) to reassure you that the child was unaffected? []
3) so that you could terminate the pregnancy if the child was affected? []

36) Check the ONE statement which agrees with your opinion about terminating a pregnancy?

1) I do not agree with terminating a pregnancy for any reason. []
2) I agree with termination of pregnancy if the mother wishes. []
3) I agree with termination of a pregnancy when it is known that the child one is carrying has a genetic defect. []
4) Other comments_____

37 a) After genetic counselling, did you plan to have more children?

Yes.....1
No.....2
Undecided.....3

b) If YES to 37a, did this decision mean having:
The same size family you had planned BEFORE counselling?.....1
Fewer children than you were planning BEFORE counselling?.....2
More children than you were planning BEFORE counselling?.....3

c) If NO to 37a, did this decision mean having:
The same size family you had planned BEFORE counselling?.....1
Fewer children than you were planning BEFORE counselling?....2

d) Was the decision in 37a about having more children affected by the information /guidance you received at the genetics clinic?

Yes.....1
No.....2
Don't know.....3

41) When you think about having children, how important were/are the following factors to you in your planning?

	VERY IMPORTANT	SOMEWHAT IMPORTANT	OF LITTLE IMPORTANCE	NOT IMPORTANT	DON'T KNOW
1) My wish to have a certain number of boys and girls.					
2) Fulfillment as a parent.					
3) Part of being married.					
4) Carrying on my family line.					
5) The number of children I think a person should have.					
6) The wishes of my partner.					
7) Taking care of my other children.					
8) Effects on my social life.					
9) My job.					
10) The father's job.					
11) Influence from relatives.					
12) Religious beliefs.					

13) Other(specify)_____

42) Could the genetics clinic be of more assistance to you or your family?

43) How could the genetic counselling sessions be more useful to you and other parents?

44) Do you have any other comments?

Thank you for your valuable help.

SECTION II (FOR FATHERS ONLY):

Please circle the appropriate answers or fill in the blank spaces.

*These questions are about the history in your family of NEURAL TUBE DEFECT, better known as spina bifida or anencephaly. In this questionnaire, we are using the abbreviation NTD to cover this condition.

1) How many children did you plan to have BEFORE your child was born with NTD? _____

2) When you came for genetic counselling, did the counsellor tell you what your chances were of having another child born with an NTD birth defect?

Yes.....1
No.....2
Don't Know.....3

3) If your answer to Question #2 is YES, what are your chances of having another child with NTD?

1) I know the chances are _____percent.
2) I think the chances are _____percent.
3) I do not remember.....3

4) After genetic counselling, what do you think is your chance of having another child with NTD?

Very high.....1
High.....2
Moderate.....3
Low.....4
Very low.....5

5) Have you looked for information about the risk of NTD other than from genetic counselling?

Yes.....1
No.....2
Do not remember.....3

6) If YES to Question #5, circle the source from which you found more information?

Family.....1
Friends.....2
Nurses.....3
Family doctor.....4
Other doctor.....5
Newspaper article.....6
Other readings.....7
Spina Bifida Association.....8
Other (specify).....9

7) Have you had another child since you received genetic counselling?

Yes.....1
No.....2
(If NO, skip to question #9)

8) If YES to Question #7,how many children did you have?

One.....1
Two.....2
Three.....3
Four.....4
More than Four (specify).....5

9) If YES to Question #7, did your partner use prenatal testing to find out if the child she was carrying was affected with NTD)?

Yes.....1
No.....2
Don't Know.....3

10) When or if you were thinking about having more children , your risk of having another child with NTD had (has):

A very important role in your decision.....1
An important role in your decision.....2
An unimportant role in your decision.....3
No role in your decision.....4

11) Do you intend to have a child in the near future?

Yes.....1
No.....2
Undecided.....3

12) Do you think that your family plans have been changed by your genetic counselling session(s)?

Yes.....1
No.....2
Don't Know.....3

13) What is your feeling about limiting your family size because of the NTD in your family? (please circle one statement only).

I feel that the condition has not influenced the number of children I have.....1
I feel that the condition has not influenced the number of children I am planning to have.....2
I feel that because of the condition, I have decided to limit my family size.....3
I do not know.....4

14) Are you or your partner using any of the following methods of birth control at this time to limit your family size?

The pill.....1
An IUD (coil).....2
Condoms.....3
Diaphragm.....4
Foam.....5
Tubal ligation.....6
Hysterectomy.....7
Vasectomy.....8

15) Is your partner pregnant at this time?

Yes.....1
No.....2
Don't know.....3

16) Should prenatal testing by fetal ultrasound be available to parents who have had a child with NTD?

Yes.....1
No.....2
Don't know.....3

17) Should prenatal diagnosis by amniocentesis be available to parents who have had a child with NTD?

Yes.....1
No.....2
Don't know.....3

18) If you were planning to have another child, would you like your partner to use prenatal testing to find out if the child she was carrying had NTD?

Yes.....1
No.....2
Don't know.....3

19) If YES to Question #18, check the MOST IMPORTANT reason for you to use the test (one answer only):

1) to find out if the child was affected ? []
2) to reassure you that the child was unaffected ? []
3) so that you could terminate the pregnancy if the child was affected? []

20) Check the ONE statement which agrees with your opinion about terminating a pregnancy?

1) I do not agree with terminating a pregnancy for any reason. []
2) I agree with termination of a pregnancy if the mother wishes. []
3) I agree with termination of a pregnancy when it is known that the child one is carrying has a genetic defect. []
4) Other comments_____

21) a) After genetic counselling, did you plan to have more children?

Yes.....1
No.....2
Undecided.....3

b) If YES to 21a, did this decision mean having:
The same size family you had planned BEFORE counselling?.....1
Fewer children than you were planning BEFORE counselling?....2
More children than you were planning BEFORE counselling?.....3

c) If NO to 21a, did this decision mean having:
The same size family you had planned BEFORE counselling?....1
Fewer children than you were planning BEFORE counselling?...2

d) Was the decision in #21a about having more children affected by the information /guidance you received at the genetics clinic?

Yes.....1
No.....2
Don't know.....3

22) When you think about having children, how important were/are the following factors to you in your planning?

	VERY IMPORTANT	SOMEWHAT IMPORTANT	OF LITTLE IMPORTANCE	NOT IMPORTANT	DON'T KNOW
1) My wish to have a certain number of boys and girls.					
2) Fullfillment as a parent.					
3) Part of being married.					
4) Carrying on my family line.					
5)The number of children I think a person should have.					
6) The wishes of my partner.					
7) Taking care of my other children.					
8) Effects on my social life.					
9) My job.					
10) My partner's job.					
11) Influence from relatives.					
12) Religious beliefs.					
13) Other(specify)_____					

25) Could the genetics clinic be of more assistance to you or your family?

26) How could the genetic counselling sessions be more useful to you and other parents?

27) Do you have any other comments?

Thank you for your valuable help.

Appendix B
Letter to Janeway Genetics Clinic

August 3,1988

Dr. E.J. Ives,
Director,Provincial Genetics Program,
Janeway Child Health Centre,
St. John's, Nfld.
A1A 1R8

Dear Dr. Ives,

My name is Marian Crowley and I am a registered nurse and a graduate student in the School of Nursing at Memorial University. I am interested in conducting a research study related to genetic counselling.

The purpose of the study is to determine if genetic counselling is useful to parents of children born with spina bifida or anencephaly (neural tube defects) in helping them make decisions about future pregnancies.

I would like to request access to your population of counselees who meet the following criteria: 1) they have had a child born with NTD between May 1984 to June 1987, and, 2) they have received at least one genetic counselling session (including their risk for another child for the same problem). Prior to sending these counselees any correspondence, I will arrange for them to be contacted by the clinic staff to obtain their permission to be contacted by the researcher. I would appreciate it if your secretary could record this contact with the name of the client, date of contact, client response (agree/disagree) and secretary's signature.

Participation for the counselees involves filling out a mailed out questionnaire. A letter of introduction will accompany the questionnaire, as well as a consent form for the parents to sign. Anonymity and confidentiality of the participants will be maintained at all times and only the researcher will have access to the data.

Although the study will provide no direct benefits or risks to the participants, it will provide nurses and other health care workers with information concerning the efficacy of genetic counselling for this population. Ultimately, it may help to improve genetic services.

A copy of the results of the study will be available at the Memorial University Library as well as at the Genetics Clinic. The research proposal will be submitted for approval to the Human Subjects Review Committees at both the Memorial School of Nursing and the Janeway Child Health Centre.

A copy of the research proposal is enclosed for your perusal. Thank you for kind consideration of this proposal.

Yours sincerely,

Marian Crowley, R.N., B.N.
Masters Candidate
School of Nursing,
Memorial University

Appendix C

CONSENT

- I have read the explanation of the proposed study.

- I agree to take part in the study as outlined in the explanation.

- If I take part in the study, my name and any personal information will be kept confidential and not available to anyone other than Marian Crowley.

- I do not have to answer any questions unless I want to.

- I may withdraw from the study at any time.

Father_____Date_____

Nurse_____Date_____

* please send this copy back with the questionnaire.

---

please cut along this line

CONSENT

- I have read the explanation of the proposed study.

- I agree to take part in the study as outlined in the explanation.

- If I take part in the study my name and any personal information will be kept confidential and will not be available to anyone other than Marian Crowley.

- I do not have to answer any questions unless I want to.

- I may withdraw from the study at any time.

Father_____ Date_____

Nurse_____ Date_____

* you may keep this copy for your records.

Appendix D
Letter of Introduction to Parents.

Dear Parent(s),

My name is Marian Crowley and I am a registered nurse and a graduate student in the School of Nursing at Memorial University. I am conducting a research study related to nursing and genetic counselling.

You are invited to participate in this study and if you decide not to participate, this decision will in no way affect the services available to you or your children at the Janeway Genetics Clinic. If you agree to participate, you are free to withhold any information during the study and you are free to withdraw from the study at any time.

The purpose of the study is to determine if genetic counselling is useful to parents of children born with spina bifida or anencephaly (neural tube defects or NTD)in helping them make decisions about future pregnancies. Participation involves filling out a questionnaire (which takes about twenty minutes of your time). Although there will be no direct benefit or risk to you from the study, it will provide nurses and other health care workers with information about the usefulness of genetic counselling. Ultimately, your input may help to improve genetic services.

The information that you provide will be kept confidential and will be destroyed after the study. You will be identified only by a number. I have included two consent forms for you to sign, one to return with the questionnaire and one for you to keep for your own records.

If I do not hear from you within two to three weeks from the mailing of the questionnaire, I will telephone you to see if you are still interested in participating. If you are interested, and I do not receive the questionnaire within an additional two weeks, I will telephone you again and with your permission, ask you the questions over the telephone.

A copy of the results of the study will be available at the Memorial University Library and at the Janeway Genetics Clinic.

Thank you for taking the time to consider participating in this study and if you have any questions about the study, you may call me collect at 437-6180 after 6:00pm.

Yours sincerely,

Marian Crowley, R.N.
P.O. Box 388,
Torbay, Nfld, A0A 3Z0


MAR 16 1988