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Family communication and genetics: developing a framework for effective interventions

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Family communication and genetics: developing a framework for effective interventions

by

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Thesis submitted to the Faculty of Graduate and Postdoctoral Studies
In partial fulfillment of the requirements for the M.Sc. degree in Epidemiology and Community Medicine

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ABSTRACT

Objective: To create a theory-based framework to guide the development of interventions for assisting genetics health services clients to communicate results of genetic testing to at-risk family members.

Methods: Systematic review methods were used to collect evidence on the barriers and facilitators of disclosure. After appraisal of several theories against key criteria, the Theory of Planned Behaviour (TPB) was chosen as the theory to base the framework on. The framework was developed through a qualitative process which incorporated the all the available evidence from the literature. A preliminary examination of the utility of the framework was done through a cross-sectional survey.

Results: The framework explains the act of disclosing a genetic test results to at-risk family members in terms of the TPB. Results from the cross-sectional survey, conducted in a population not previously studied, were generally consistent with the information presented in the framework and found in the literature.

Conclusions: The framework has potential to be used in developing interventions; however, results need to be replicated in prospective studies with larger samples.

EXECUTIVE SUMMARY

Context: Genetic risk information from one family member's genetic test provides relevant information about other family member's risk of developing that same disorder. Thus, communicating the results of a genetic test to at-risk family members can be very important. The responsibility for communicating this information lies with the client who underwent genetic testing; however, non-disclosure has been documented and raises several issues in both the familial and clinical setting. To date, there is a considerable amount of literature on the topic, yet there is no coherent understanding of the process, barriers and facilitators of this communication. Having this understanding is essential to developing and evaluating interventions for patients and families who have problems conveying this information to at-risk family members.

Objective: To create a theory-based framework to guide the development of interventions for assisting genetics health services clients to communicate results of genetic testing to at-risk family members. Specifically, this research aims to: a) select a theory to organize the barriers and facilitators of family communication of genetic risk information; b) update a previous systematic review on barriers and facilitators involved in disclosure of genetic test results to family members; c) use the evidence identified from the systematic review and the chosen theory to develop a framework for disclosure of genetic risk information between family members, as a way of organizing all the information such that it would aid in the development of interventions; and d) assess the initial applicability of the framework in a patient population using a cross-sectional survey.

Methods: Traditional systematic review methods were used to collect evidence on the barriers and facilitators of disclosure. Through consultation with experts and appraising

various theories against 5 key criteria, the Theory of Planned Behaviour (TPB) was chosen as the foundation of the framework. The framework was developed through a transparent, qualitative process where all the available evidence from the literature on the topic was organized according to, and framed in terms of the TPB. The framework was validated in a full-day workshop with a panel of experts. A preliminary examination of the utility of the framework was done through a cross-sectional survey of myotonic dystrophy patients from the Children's Hospital of Eastern Ontario genetics clinic. The cross-sectional survey consisted to two main sections: a) a TPB section which examined the applicability of the framework in this population; and b) an open-ended question section focused on attaining information on the experiences of this patient population with disclosure.

Results: The framework explains that the act of disclosing a genetic test result to at-risk family members is formed by 5 factors which contribute to one's attitude about disclosure, 3 factors which influence the pressure one feels to disclose, and 4 factors which form one's perceived control to disclose. Attitude is formed from the individual's 1) desire to protect oneself or family members from potentially harmful information, 2) perceptions of relevance of the information for family members, 3) perceptions of responsibility to disclose the information, 4) perception of rights to information, and 5) perception of the usefulness of communicating. Subjective norm (pressure to perform the disclosure) is influenced by pressure from genetics professionals, other family members and society. Perceived control over disclosure is determined by 1) the family relationships and dynamics, 2) the communication skills of the client, 3) the ability of the client and family members to understand the information, and 4) the coping skills of the client and family members.

Of 99 eligible participants, 28 responded to the survey (response rate of 28.3%). Results from the cross-sectional survey were generally consistent with the information presented in the framework and found in the literature to date. In this sample there were high rates of disclosure and first-degree relatives were more likely to have been informed than more distant relatives. The most important reasons in deciding to disclose to their family member identified were: a) having a close relationship with the family member, b) perceiving that the information was relevant for them , c) believing that they were capable of handling the information and d) feeling a responsibility to tell them about their genetic risk. This sample had moderate and positive attitudes towards disclosing the genetic risk information to family members, had low pressure from society or family members and moderate pressure from genetics professionals to communicate and felt they had moderate control over the disclosure in general, yet did not have control over certain aspects of disclosing the behaviour.

Conclusions: These preliminary results indicate that a TPB-based framework may be a useful way to examine the barriers and facilitators of communication of genetic risk information in general. This study has established preliminary evidence of the face validity of the framework and that the framework has the potential to be used in developing interventions; however this must be interpreted cautiously because of the small sample available for analysis and the low overall response rate. The framework requires further evaluation and possible refinement in a wider range of study populations.

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CHAPTER I

INTRODUCTION

Overview

The rapidly expanding area of human genomics has lead to an increase in the amount of information available about the contributions of genetics to our health. Accompanying this expansion of knowledge has been a myriad of psychosocial, ethical and legal issues.[1] Genetic testing has altered the meaning of personal health information because it provides the individual not only with disease risk information about himself or herself, but also about family members. As such, genetic information is often referred to as family information and genetic testing a family affair.[2] Because genetic information has implications for individuals and their family members, communication of genetic test results within families has become and important issue in clinical genetics health services.

Communication between family members about genetic risk information provides potentially important information and knowledge about risks for diseases, and in some instances provides information which might save the lives of family members, as is the case for Long QT Syndrome (LQTS). LQTS is a genetically inherited cardiac arrhythmia which can cause syncope, seizures and sudden death. [3-5] Lifestyle modification to avoid triggers for cardiac arrhythmia, and use of beta-blockers, pace makers and implantable defibrillators can help treat the disorder and reduce mortality.[5] LQTS presents a clear example of how family communication of genetic test result information would be of great importance to other family members; it is a potentially fatal disease with treatment and prevention options available if mutation carriers can be determined through genetic testing.

Genetic testing is also available for disorders where test results do not provide definite answers regarding risk and disease, such as for hereditary breast and ovarian cancer (HBOC), or where treatment methods are not available or less effective (e.g. Huntington's disease). However, knowledge of genetic risk information in these instances can still provide useful information which provides relief from uncertainty, information relevant to children, and more information for planning for the future, marriage or reproduction decisions.[2] Communication of genetic risk within families in these situations can be complicated and raises ethical issues pertaining to patients' right to confidentiality, a family member's right to know (or not know) their genetic risk, and a genetic counsellor's duty to maintain patient confidentiality and their 'duty to warn' other family members of possible risk status.[1]

Familial non-disclosure of genetic test results has emerged as an important area for clinical genetics with potential implications for genetic counselling. Guidelines typically suggest that the genetic counsellor discuss with the client the importance of communicating the risk information to their family members, but advises against breaching patient confidentiality except in extraordinary cases.[1;6] However, there are documented instances where genetic counsellors would like to intervene and inform family members directly,[7;8] and in fact, such acts have led to legal cases.[9;10] Genetic counselling aims to be non-directive where the client is given all the information about the disorder, genetic risk, options for managing that risk, and is offered support in adjusting to their risk of a genetic disease.[11] Yet, a review of guidelines and position papers on communication of genetic information in families[12] found that three common principles were articulated in the guidelines identified.

1) Individuals have a moral obligation to communicate genetic information to their family members;
2) genetic health professionals should encourage individuals to communicate this information to their family members; and
3) genetic health professionals should support individuals through the communication process (Forrest 2007, p. 612).

Thus, the roles of a genetic counsellor include 1) not causing harm to the patient (by respecting privacy and not causing distress) 2) assisting the patient (by helping to alleviate distress and provide information and support) and 3) respecting patient autonomy (by remaining non-directive and accepting their right to make their own choices). However, a fourth role may include a duty protect the patient's family and a fifth role to 'strive for justice' – a balance between good and harms. Therefore, at the clinical level, the counsellor is placed in a difficult position where several different professional ethical principles may conflict.

At the family level, disclosing such sensitive information to different family members can also raise a variety of different issues. For example, suggesting that the ideal position for counsellors is to encourage the communication of genetic risk information ignores the possibility that this might produce more distress and anxiety than benefits. For example, a patient undergoing genetic counselling may feel pressured to discuss the topic with others, which may be anxiety-provoking and undermines autonomous decision making of the patient. In addition, the act or process of telling might open up difficult or distressing issues which already exist within the family.

A previous systematic review on family communication of genetic risk[13] identified the general themes that have emerged as being important in the communication process. There is general consensus in the literature that communication of genetic test results within second- and third-degree relatives is less frequent and more selective than within first-degree family members.[13-22]

However, while communication tends to be more open in the nuclear family, there are documented instances of poor communication of genetic risks even within the immediate family unit, for hereditary breast/ovarian cancer, [17] cystic fibrosis,[17;23-26] hemophilia,[22] and fragile X syndrome.[27] Within families, women are commonly identified as the 'messengers' or 'communicators' of genetic risk information. Often women are told more frequently about family members genetic test results than men, or paternal relatives,[20;22] and have the implicit responsibility of communicating results of family members to other family members.[14;15;28]

Barriers to communication which tend to be frequently encountered often relate to the degree of closeness of family members and emotional issues associated with disclosing such sensitive information. Poor social contact, geographical distance, [14;15;17;18;26] and emotionally distant or tense relationships[19;26] have consistently been found to impede communication between relatives. Parental feelings of guilt, upset and shame, e.g. in recessively inherited conditions such as hemophilia and cystic fibrosis, often make communication of genetic risk information to children and other family members more difficult.[26;29] Another frequently cited reason for not disclosing includes not wanting to upset or alarm family members or cause too much emotional upset.[14;17]

A variety of other problems which have been documented in inhibiting communication of genetic risk information include: misconceptions that the family members have already been notified;[14;17] a negative experience at a person's first attempt to disclose the information;[21] taboos surrounding hereditary disorders;[24;29] concerns about insurance or employment discrimination;[21] and the age of the recipient of the information.[19;20;26;30] Some individuals have reported feeling that information was not relevant to some family members who were not having children at the moment and therefore, there was no reason to tell them.[26]

Factors which facilitate communication within families about genetic risk information have been examined to a lesser degree; nonetheless, some factors still emerge as being important contributors. One commonly identified motivation for telling family members about their risk is a sense of duty or moral obligation to disclose the information.[15;18;21] A patient's feeling that the information is important to family members or that relatives could benefit from being tested can also influence whether communication is likely to occur.[14;18] Furthermore, being in need of social support after receiving a genetic test result,[20] and external cues such as professionals suggesting communication, are also important in facilitating communication.[19]

Communication about genetic risks and disease status can be a stressful event, as it can potentially lead to negative experiences and consequences for the informant.[27] A wide variety of negative events arising from disclosure of genetic risks have been documented in the research, including: family members' denying they having any personal risk,[26;27] being reluctant to accept the diagnosis, refusing to speak to the informant after disclosure, and feeling angry that they were informed.[27] More extreme situations also occur, such as rivalry

between siblings where one is a carrier of a genetic disorder and the other is not, and survival guilt in siblings who do not carry a disease-associated genetic mutation.[27]

Interventions

Communication of genetic risk information amongst family members is not a simple process. Given the often complicated and diverse inter-relational nature of modern families and the complexity of genetic information itself, this type of communication can be very difficult or perceived as impossible in some circumstances to some patients.[31] Considering the potential for patient- and family-specific issues related to family communication of genetic information, facilitating family communication will likely require a variety of different types of interventions. Ideally we could have interventions which remove the ethical burden for counselors who feel a duty to both their patients and their patients' family members, while also providing options for patients to minimize their distress as well as distress to other family members. Thus, as well as the entire issue of family communication being complicated, it is highly likely that at least some of the necessary interventions will themselves also be complex. They could range from a very simple educational pamphlet to more involved tools such as the Genogram[32] or six-step skill building exercise[33].

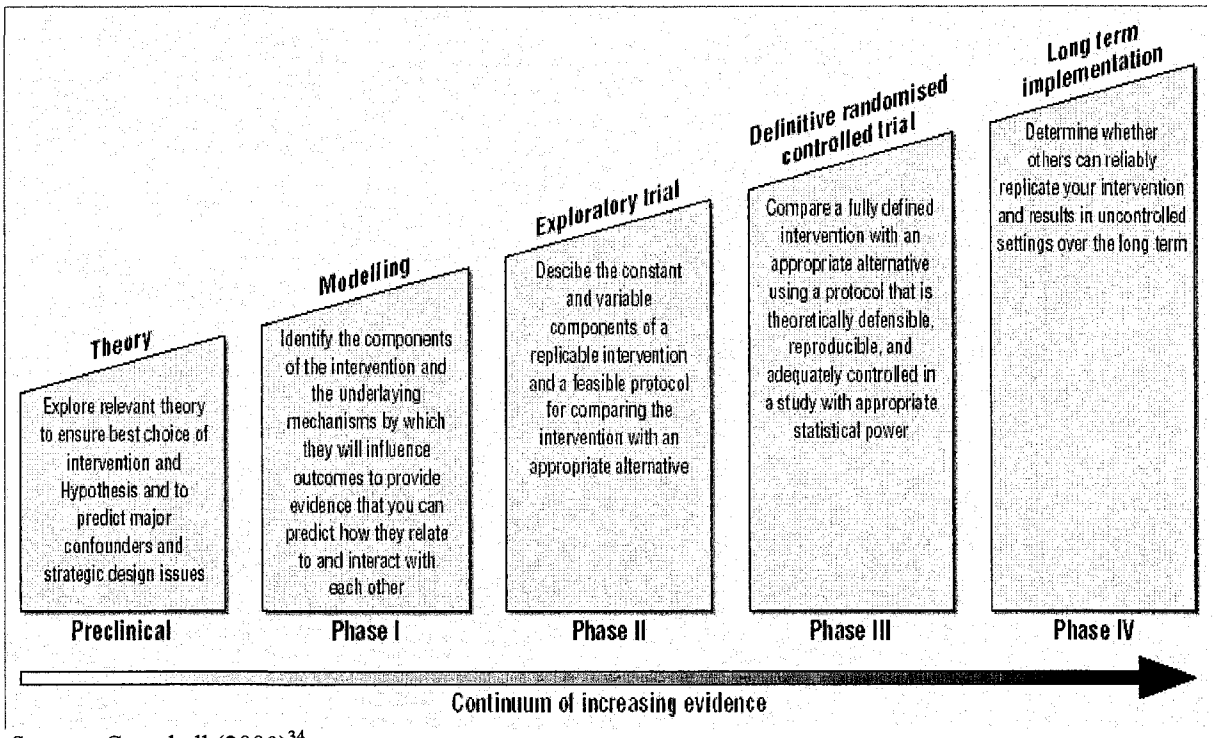
Interventions used in healthcare, including the type of interventions which might be needed to facilitate family communication of genetic risk, are also often very complex. A complex health intervention is composed of several different components which together appear to be important to the effectiveness of the overall intervention; however, it is usually difficult to discern any single element which acts as the 'active ingredient'.[34-36] Thus, the evaluation of complex interventions is difficult because of problems developing, identifying, documenting and reproducing the intervention which often arise because researchers have

not fully defined the intervention, or have developed it on a pragmatic basis without any type of conceptual underpinning.[35]

The United Kingdom Medical Research Council (MRC) has proposed a phased approach for the development and evaluation of complex interventions which emphasizes the importance of a coherent theory-based framework in the early phases of intervention development.[36] The framework consists of phases of investigation in the evaluation of complex interventions, which parallel the sequence of steps usually taken in the evaluation of drugs from pre-clinical research to post-marketing surveillance.[36] The first phase, within which the current study primarily falls, is the establishment of the theoretical basis which suggests the kind of intervention that would be effective. This phase is useful in identifying, in preliminary form, the kind of intervention needed and the study design needed later to evaluate it.[35] At the very least, incorporating this phase forces investigators to consider the underlying assumptions they are making about their intervention, the context for its use, and highlights issues which can result in modification and revisions of the complex intervention, as well as help to identify which kinds of interventions are implausible.[36]

The theoretical basis of an intervention can be any body of evidence ranging from a formal theory to accumulated wisdom from empirical evidence. For example, an intervention designed to modify health behaviour may benefit from consulting the health-behaviour theories, whereas a trial to change doctors' prescribing behaviour may be refined by considering research on factors influencing prescribing decisions.[36]

The other phases in evaluation include modeling, an explanatory trial, a definitive randomized control trial and long term implementation as detailed in Figure 1. The process is an iterative one and progression from one phase to another may not be linear.[35]



Source: Campbell (2000)[34]

Figure 1: Framework for development and evaluation of complex interventions

Therefore, regardless of the complexity of the interventions to be developed, an explicit theory-based framework would be useful in: mapping out the different types of barriers and facilitators in family communication, mapping out the different types of possible interventions, and identifying important methodological issues for an evaluation (e.g. outcomes).

Rationale

The issues surrounding familial disclosure of genetic results are complex. Methods for helping professionals facilitate the process in families are necessary, not only to resolve the ethical issues genetic counselors face, but to ensure that patient and their families interests are seen too. A framework which highlights the barriers to communication of genetic risk information, has a strong theoretical underpinning, and can be used to outline interventions which would be predicted to be effective in specific instances will be developed. This framework will enable researchers and professionals to understand what will inhibit and facilitate communication for their clients, and develop interventions to suit their needs. The evidence-based, theory-driven framework will serve as a resource for future creation of interventions to deal with these types of communication issues.

Research Plan

The main objective of this project is to complete the 'preclinical' phase in the development and evaluation of complex interventions by creating a theory-based framework which will be useful in the development, and later evaluation, of interventions for the issues associated with communication of genetic risk information in families. Thus, the research project occurs in three major parts:

1) choosing an appropriate theory to base the framework of interventions;

2) developing the framework based on the chosen theory which explains the barriers and facilitators of family communication of genetic risk; and

3) preliminary testing of the face validity of the framework with a cross-sectional survey in a population with a genetic disorder.

Objectives

1) to select an appropriate theory to organize the barriers and facilitators of family communication;

2) to update a previous systematic review[13] on barriers, facilitators and issues involved in disclosure of genetic test results to family members;

3) to use the evidence identified from the systematic review and the chosen theory to develop a framework for disclosure of genetic risk information between family members, as a way of organizing them and, potentially, as a way of creating a predictive model;

4) to use descriptive data from a sample of genetics patients to assess the initial applicability of the framework.

CHAPTER II

CHOICE OF THEORY

Aims and Objectives

The first step in applying the complex interventions framework is the choice of an appropriate theoretical model. The overall aim of this section is to select a theoretical framework which is suitable for use as a starting point for organizing published evidence on barriers to and facilitators of family communication of genetic information, in order to facilitate the choice, development, and evaluation of interventions to promote communication. The specific objectives are:

a) to identify the key criteria for selecting a suitable theory;

b) to identify potentially relevant theories and frameworks, drawing on literature from communication research and social cognition models.

c) to review each theory against the key criteria;

d) from those which meet the key criteria, to select the one which appears best suited to the overall aims of the current project.

Methods

Key criteria to guide the selection of a theory were developed through discussion with a health services researcher, a genetic counsellor, and a social psychologist. A range of theories were identified through consultation with a psychologist. Key references were reviewed to assess how well each theory met the selection criteria; the final decision was made through discussion within the research team of the theories which were the best fit with the criteria.

Results

Key Criteria

Five key criteria were identified:

1) Relevance to family communication of genetic risk information ("relevance")
2) Ability to incorporate a wide range of factors which underlie communication of genetic risk information ("range of factors")
3) Ability to delineate or categorize target behaviors amenable to intervention (e.g. theories which describe communication as a function of culture would not be useful, since culture is not a factor which can be easily modified) ("amenable behaviours")
4) Ability to predict whether communication will occur in varying circumstances ("predictive ability")
5) Evidence of validity ("validity")

Potential Theories for the Framework

Nine theories were considered, and are listed in Table 1. Five models frequently used in family communication research were examined. These types of models are used to describe the different types of interactions and communications within families, to assess familial functioning based on communication patterns or how situational experiences can alter the family and its communication. Social cognition models refer to how individuals make sense of social situations with a focus on individual cognitions/thoughts as processes intervening between observable stimuli and responses in real world situations (cognitions as key proximal (and modifiable) determinants of behaviour). Four of the most widely used and research social cognition models were also considered.

Table 1: Types of models considered for use in developing the family communication of genetic risk framework

Type of Model	Model name
Family Communication	
	Circumplex Model of Marital and Family Systems[37]
	Double ABCX model of Adjustment and Adaptation[38]
	The Beavers Systems Model[39;40]
	McMaster Model of Family Functioning[41]
	Family Communication Patterns[42;43]
Social Cognition	
	Health Belief Model[44]
	Protection Motivation Theory[45]
	Social Cognitive Theory[46]
	Theory of Planned Behaviour[47]

Communication models

The Circumplex Model of Marital and Family Systems

The Circumplex Model of Marital and Family Systems[37] is one of the two most widely researched and clinically used models. The circumplex model has its foundation in family systems and family development theories and focuses on three dimensions of family systems: cohesion, flexibility and communication. [48] Families with balances in cohesion, flexibility and communication are viewed as 'healthier' than families which are on the

extremes of these dimensions. This model has been used in research to assess family functioning and family change over time.

Double ABCX model of Adjustment and Adaptation

The double ABCX model is the other most widely researched communication model and, like the Circumplex model, was based on the work of Ruben Hill.[49] Focusing on adjustment and adaptation,[38] it was designed to explain how families adapt to stressful events. It is postulated that a family's response to a stressor is affected by the family's resources and the perception family members develop of the situation. Family adaptation is considered the outcome measure of the model, measured on a continuum from positive adaptation to maladaptation. This model is also used to assess family functioning, particularly in the context of stressful events.

The Beavers Systems Model

The Beavers Systems Model,[39;40] also assesses family functioning; however, it does so in terms of strengths and weaknesses within the family on two dimensions: family competence and family style. Family competence explains the extent to which a family structure will allow family members to be flexible and adaptable and includes the components of family structure (power, coalitions, closeness), family mythology, goal directed negotiations, autonomy, and family affect. Family style addresses the elements of family interaction and is based on a balance of internal and external family orientation. This dimension is made up of the components; dependency, conflict, proximity, attitudes toward outsiders, closeness, assertive versus aggressive qualities, positive versus negative feelings and scapegoating. The Beavers system model communication is used as a measure of the degree of functioning a family exhibits.

The McMaster Model of Family Functioning

The McMaster Model of Family Functioning [41] emphasizes six dimensions of family functioning: problem solving, communication, roles, affective responsiveness, affective involvement, and behaviour control. Each of these is divided into 'instrumental' (finances, living arrangements) and 'affective' (feelings and emotional experience types). The communication dimension specifically deals with how information is verbally exchanged and includes clarity and directness of communication. Thus, like the Beavers system model communication is used as a marker of the degree of functioning in a family.

Family Communication Patterns model

The Family Communication Patterns model[42;43] describes families' tendencies to develop fairly stable and predictable ways of communicating with one another. It suggests that two dimensions underlie the family communication patterns – conformity orientation and conversation orientation. Conformity orientation refers to the degree to which family communication stresses a climate of homogeneity of attitudes, values and beliefs, whereas conversation orientation refers to the degree to which families create a climate where all family members are encouraged to participate in unrestrained interaction about a wide array of topics. There are four types of family communication patterns – consensual (high in both conversation and conformity), pluralistic (high in conversation but low in conformity), protective (low in conversation but high on conformity) and 'laissez-faire' (low in conversation and conformity). This model appears useful in describing why the communication pattern in a family exists and defining the type of communication pattern within a family.

Social cognition models

Health Belief Model

The health belief model (HBM)[44] includes six determinants of behaviour: perceived susceptibility, perceived severity, perceived benefits, perceived barriers, health motivation and cues to action. The model suggests that health behaviours are more likely to be carried out if the individual perceives high threat of disease (high susceptibility and severity), if benefits can be derived from performing the behaviour, and if there are few barriers to performing the behaviour or a combination of these.[44] However, studies have shown[50;51] weak predictive validity of the HBM, most likely due to poor definition of constructs, lack of rules for combining constructs and lack of evidence for discriminant validity between the model's components and variables from other models.

Protection Motivation Theory

Protection motivation theory (PMT)[45] represents health behaviour as adaptive coping (beneficial to health) or maladaptive coping. It suggests that there is a threat appraisal process, which is determined by perceived vulnerability to and perceived severity of the threat. This appraisal increases protection motivation, unless there are advantages in performing the maladaptive behaviour. It is suggested that a second appraisal process, coping appraisal, is partly determined by the usefulness of the effect and confidence in one's ability to perform the behaviour (known as self-efficacy).

Social Cognitive Theory

Another well known motivational theory of behaviour is Bandura's social cognitive theory.[46] This theory states that self-efficacy and outcome expectancies (of the situation and action) are the two key determinants of behaviour. Situation-outcome expectancies are

based on the perception that some consequences are determined by the environment and thus are not under volitional control. Action outcome expectations are related to the belief that one's actions are instrumental to a particular outcome (self-efficacy related to confidence in one's ability to carry out a behaviour). Therefore, social cognitive theory proposes that, in order for someone to perform a health behaviour, the person must believe that he or she is at risk for acquiring a serious and severe negative health outcome, and believe that the benefits of performing the behaviour outweigh the costs of performing the behaviour.

Theory of Planned Behaviour

One of the most thoroughly researched theories is Ajzen's (1988) Theory of Planned Behaviour (TPB).[52-55] The TPB proposes that behaviour is predicted by the strength of an individual's intention to perform the behaviour. Thus, behavioural intention is determined by three considerations: attitude towards the behaviour, subjective norm (perceived social pressure) and perceived behavioural control over the behaviour. The theory has been used to predict behaviour in a wide variety of health-related and non-health related behaviours and has been shown to explain 39-41% of the variance in intention and 27-34% of the variance in behaviour in a range of studies.[53-55]

Final Choice of Theory

Table 2 describes each theory and whether it met each of the five key criteria set out, which was used to eliminate theories which did not meet the criteria. Two theories met all five criteria set forth and thus the final choice of theory between these was based practical aspects related to development of a survey instrument.

The communication theories are most applicable in research where factors related to family functioning are of interest. However, they may be less useful in the context of this

research, where the aim is not to foster change in a family unit or even understand the functioning of a family in terms of its communication, but rather to explain *why* and *how* communication of this type of information occurs within families.

Table 2: Rating of each potential theory according to five key criteria

Theory	Relevance	Range of factors	Amenable behaviours	Predictive ability	Validity
Circumplex Model of Marital and Family Systems[37]	-	+	-	-	+
Double ABCX model of Adjustment and Adaptation[38]	-	+	-	-	+
The Beavers Systems Model[39]	-	+	-	-	+
The McMaster Model of Family Functioning[41]	-	+	-	-	+
Family Communication Patterns model[42]	-	+	-	-	+
Health Belief Model[44]	-	+	+	-	-
Protection Motivation Theory[45]	-	-	+	+	+
Social Cognitive Theory[46]	+	+	+	+	+
Theory of Planned Behaviour[56]	+	+	+	+	+

From a clinical perspective, it is not reasonable to consider developing interventions which aim to somehow amend dysfunctional family relationships, nor is it the role of a genetic counsellor to do so; thus communication theories have limited use in this the context

of this research project, where the goal is to facilitate communication irrespective of the family situation. Other limitations to the application of these theories in the current study exist. Firstly, these theories offer little in terms of ability to predict whether 'communication' will occur, which is relevant to the development of interventions. Also, it is likely that the communication style of the family will be a key factor influencing *whether* an individual is disposed towards disclosing genetic information, and thus would likely act as a predictor in a behavioural theory model.

Behavioural theories generally offer explanations of the target behaviour and thus, potentially, a means to predict it, and are therefore better suited to the aims of this project. The theory of planned behaviour was selected as the most applicable model for this exploratory study for several reasons. Firstly, it is a robust, well-validated theory supported by extensive empirical data, including meta-analyses,[57;58] not only for the prediction of health behaviours, but a variety of non-health behaviours as well.[53] In contrast, the HBM and PMT deal specifically with health behaviours. While communication of risk information could be considered a health behaviour, it is qualitatively different from the typical health behaviours (e.g. exercising or eating a healthy diet) for which they were designed. Finally, the TPB was chosen over the social cognitive theory, both of which take into account non-health and health-related behaviour, for several reasons. In practice, the social cognitive theory is harder to operationalize than the TPB (Holly Etchegary, personal communication, Nov 2006); it has more variables (e.g. three different types of outcome expectations), so is less parsimonious than the TPB. The TPB allows for the influence of multiple external factors to be included within the model, whereas the SCT does not.

Finally, there is also a detailed guide to incorporating the TPB into instruments, developed specifically for use in health services research,[59] which would facilitate this project.

Theory of Planned Behaviour: Constructs and variables

As mentioned, the TPB suggests that behaviour is predicted by the strength of an individual's intention to perform the behaviour, and that intention is determined by an individual's attitude towards the behaviour, subjective norm (perceived social pressure to perform the behaviour) and perceived behavioural control over the behaviour.[47] The theory further elaborates that attitudes about a particular behaviour are formed from beliefs about the outcomes of the behaviour (behavioural beliefs), weighted by whether these outcomes are evaluated in a positive or negative manner (outcome evaluation). Depending on whether the behavioural beliefs are positively or negatively evaluated by the individual, the result will be a favorable or unfavorable attitude toward the behaviour. Subjective norm (SN) is a function of an individual's beliefs about the expectations of other important individuals or groups (normative beliefs) and the person's motivation to comply with these groups (motivation to comply). Perceived behavioural control (PBC) is the construct added to the Theory of Reasoned Action[60]to account for the fact that not all behaviours are 100% under volitional control. PBC is predicted by beliefs about factors likely to facilitate or inhibit the behaviour (control beliefs) and an evaluation of the power that each factor has to affect behaviour (power).[56;61] Other, demographic and personality type factors can have an influential role in forming attitudes, subjective norm and perceived control, which in turn can affect the occurrence of a behaviour. The theory also states that attitudes, subjective

norm and perceived behavioural control can change over time and are different between different situations and thus precise defining of the target behaviour is imperative.[56;61]

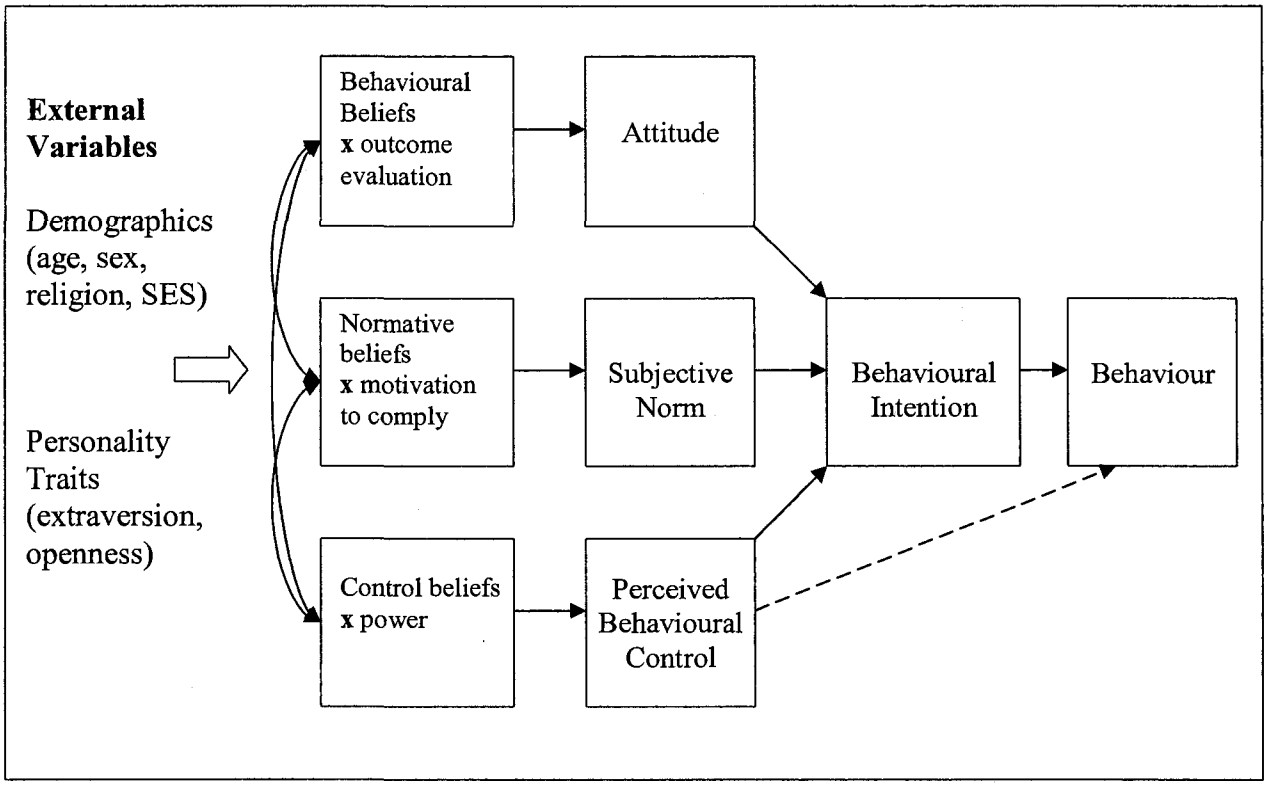


Figure 2: The Theory of Planned Behaviour [47]

CHAPTER III

POPULATING THE THEORY

Aims and Objectives

The overall aim of this chapter is to create a theoretical framework to guide the development of family communication interventions by populating the theory of planned behaviour with specific factors relating to communication of genetic risk identified in previous research studies. The specific objectives are:

1) to update a published systematic review on barriers and facilitators of disclosure of genetic risk information to family members; and
2) to map the barriers and facilitators on to the domains of the TPB to produce a framework specific to the communication of genetic risk information.

Methods

Systematic Review

A published systematic review[13] was available which described the evidence available to November 2003. This review was designed to update and extend this review.

Study Identification

A systematic electronic search strategy was used to identify all studies related to genetic risk information and family communication from January 01, 2003 to October 09, 2007. The search strategy was modeled on the previous review,[13] which covered the period up to November 2003. The search included years before 2003 in case the previous review missed relevant reports. Electronic databases searched were:

- MEDLINE (1966-Oct. week 1 2007)
- Medline In Process (1996-Oct 09, 2007)
- EMBASE (1980-2007 week 40)
- CINHAL (1982 – Oct. week 1 2007)
- PsycINFO (1806- Oct week 1 2007)
- Web of science (searched Oct 09, 2007)
- Sociological abstracts (searched Oct 09, 2007)
- Social services abstracts (searched Oct 09, 2007).

The date of the last search was October 09, 2007. Published studies were identified with the medical subject headings: *communication, disclosure, truth disclosure, self disclosure, family, family relations, parent-child relations, intergenerational relations, relative or sibling, genetic predisposition to disease, genetic diseases, inborn, genetic screening, genetic counseling, genetic services, heredity, hereditary disease, disease susceptibility.* The following combination of text words were used to supplement the medical subject heading search: *non-disclosure, family communication, family systems, family dynamics, family secrets, kinship, pre-selection, genetic risk, genetic test, genetic disorder, predictive test and genetic services.* Complete search strategies for each electronic database can be found in the Appendix A. Screening of citations of retrieved articles was also completed. Grey literature was not searched because of lack of access to relevant databases, and authors were not contacted because of the restricted time available. Studies included in the original review were included in the current analyses in order to be broad in scope and inclusive of all the evidence available to date on the subject.

Title and abstract screening

Titles and abstracts resulting from the search were independently screened by the candidate. They were retained for full text review if:

1) they were published in English;

2) genetics or inheritance were indicated as a subject in title or abstract; and

3) communication was indicated as a subject in title or abstract.

Studies were excluded at this stage only if the title and abstract clearly indicated that the focus was not communication between family members. The full texts of all remaining reports were obtained for the next stage of review, which was also conducted by the candidate.

Full text screening

Studies were retained in the review if they met the following criteria.

Population: people who had undergone genetic testing for any genetic disorder; people known to be at increased risk because of a family history of any genetic disorder.

Study aim: exploration, examination, or assessment of any aspect of communication between family members about genetic risk.

Design: any type of qualitative or quantitative design where a clearly defined research question was articulated.

Studies were excluded if they focused on disorders which have not been shown to have a genetic component. Narratives, commentaries, book chapters, and editorials were also excluded.

Data Abstraction

A standardized data abstraction form was developed in Microsoft Access (2003) and applied to all studies which met the inclusion criteria (Appendix B). The form was piloted and any changes made prior to commencement of data abstraction. The candidate extracted data on the following items: study author, title, date, journal, study design, verification of all eligibility criteria mentioned above, number of participants, country, recruitment, participants' characteristics, setting, study rationale, study methods, main findings, specific barriers and facilitators, implications and limitations.

Quality rating of the studies was not done because of the wide range of study designs included.

Data Analysis

Because of the wide range of study designs considered, quantitative synthesis was not considered. The findings are presented as a narrative review, summarized in a qualitative manner.

Mapping of family communication barriers and facilitators to TPB domains

Firstly, the barriers and facilitators ("factors") were listed by individual report included in the review. Secondly, they were reorganized into a list of individual, distinct, mutually exclusive factors ("distinct factors"), and each were labeled "B" or "F" and given a unique number. The original articles from which they were derived were noted alongside each distinct factor, to give a count of the frequency with which the same one was identified

or reported in different studies. Next, each distinct factor was mapped on to the appropriate TPB construct. For example, in reports that described how individuals felt they could not disclose genetic test results to other family members from whom they were estranged, the distinct factor (a barrier) would be included under the 'perceived behavioural control' construct of the TPB. The difficulty of contacting this family member, and the perception of a hostile response if contact was made, would lead to this behaviour being perceived by the individual in question as less within their control. This concept fits in the perceived behavioural control construct. Finally, within each construct of the TPB, the distinct factors were categorized into themes; for example, within the construct of 'subjective norms', distinct factors could potentially be classified into the themes of 'perceived pressure from other family members', 'perceived pressure from professionals', or 'perceived pressure from society'.

The interpretations of meaning and organization of distinct factors were subject to a validation process described below.

It was anticipated that individual barriers or facilitators might fall under several different constructs, because it can be difficult to differentiate between an attitude and a perceived behavioral control item. This is a commonly encountered issue with TPB.[59] For this project, where a factor could be placed in two constructs, it was included in both, in order to create the most comprehensive framework possible. Similarly, it was anticipated that some barriers and facilitators would not fit perfectly into any of the three constructs. The TPB also has a mechanism to account for factors which are not directly definable according to attitude, subjective norm or perceived behavioural control constructs, such as personality or culture. These are considered to be factors which contribute to forming

perceptions of attitude, subjective norms or perceived behavioural control[56] and are placed outside the defined constructs.

Validity of the Framework

A full-day workshop was organized in order to obtain relevant feedback on the development of the framework, to decide upon the most applicable and useful organization scheme, and ensure that the final framework chosen had face validity. For this purpose, members of the thesis committee (a social psychologist with expertise in the TPB (HE), a genetic counsellor with first-hand experience of communication issues in the clinical setting (CH), and a health services researcher with an interest in the field of family communication of genetic information (BW)) formed a panel to scrutinize the emerging data. In the morning session, the candidate presented the findings of the TPB framework in draft format to the panel. Factors which could have different interpretations were discussed and agreed upon by the panel. The afternoon session focused on verifying the interpretation of barriers from the literature, ensuring the correct placement of the factors within the TPB, and deciding which factors were the most clinically relevant encountered in the clinical setting. Input from the workshop session was integrated into the final version of the framework and additional input and final verification of the framework was done electronically at a later date.

Results

Systematic Review

Results of the systematic literature search are detailed in Figure 3. A total of 6231 citations were identified (1677 from MEDLINE, 537 from Medline Inprocess, 1230 from EMBASE, 935 from CINHAL, 702 from PsycInfo, 168 from Web of Science, 535 from sociological abstracts, 447 from social services abstracts, and 1 from citation snowballing). 1575 duplicates were removed leaving 4655 for the broad screen. 211 manuscripts potentially relevant to the current review were identified by independent review of abstracts and titles and retrieved for further examination. 173 studies were excluded for the following reasons (see Appendix C for citations): 1) genetics or inheritance was not the subject of communication, 2) communication was not between 'biological family members', 3) there was no defined research question (commentary, narrative, editorial, book chapter etc), 4) the paper/research did not offer any insight into the patient's or family's experience, or 5) the study was included in the previous review.[13] Based on the inclusion and exclusion criteria 38 articles were left for review, published between January 2003 and October 09, 2007.[2;17-19;62-95] The thirty-one articles found by Wilson *et al.*, (2003)[14-16;20-22;24-26;28;29;31;96-114] were also included (Appendix D).

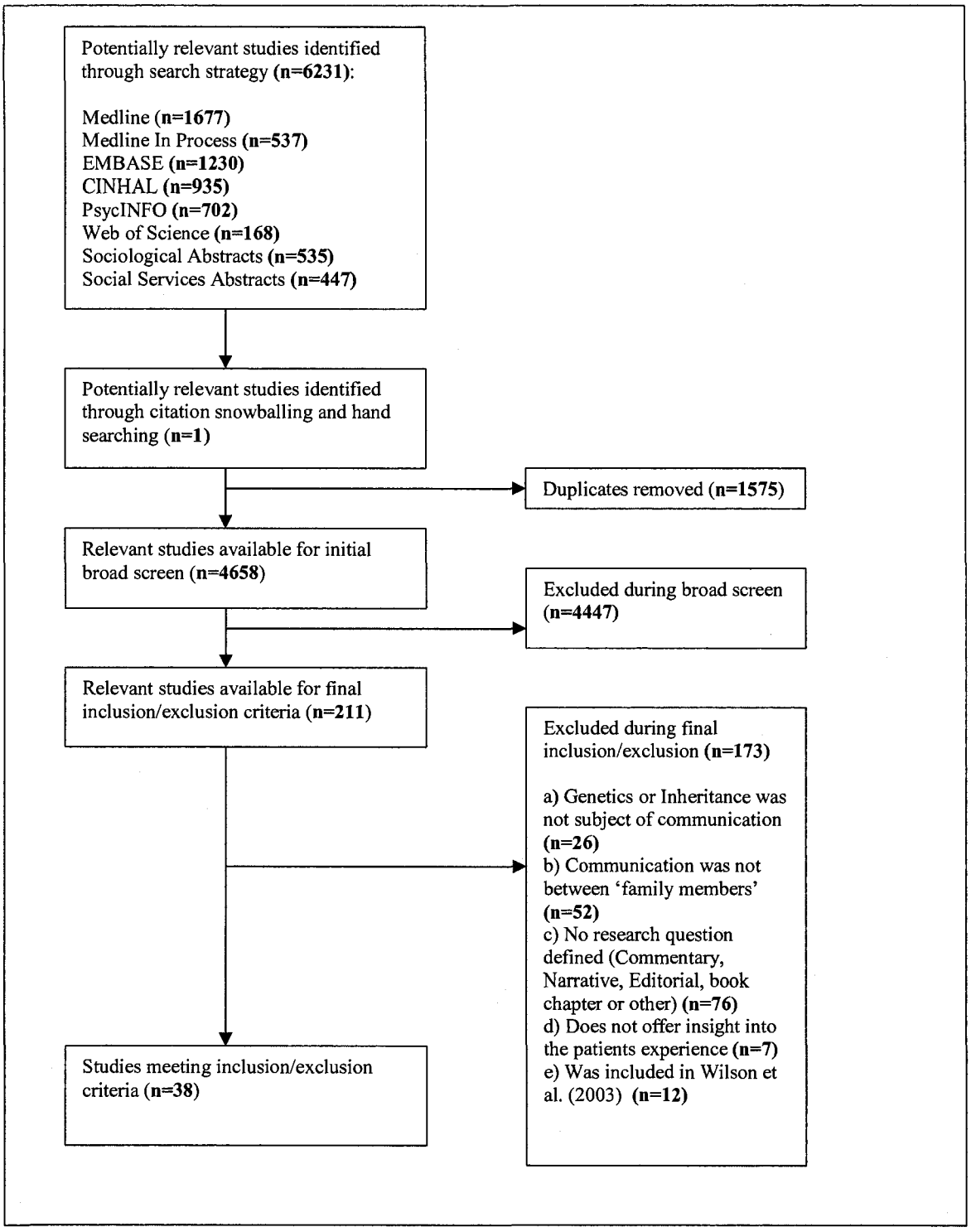


Figure 3: QUOROM[115] flow diagram outlining the results of the literature search and the selection of studies for inclusion in the review.

Study Characteristics

A detailed evidence table including the characteristics of the included studies is presented in Appendix E. Below, table 3 illustrates that the research to date has been predominantly qualitative and/or descriptive in nature, using semi-structured or in-depth interviews,[2;14;15;19;21;22;25;26;31;62;64;66;68-70;72-78;80;82;86;92;93;96;99;100;102-106;109-113;116] and questionnaire-based cross-sectional surveys.[2;14;16-18;20;22;24;28;29;63;65;81;83;84;87-91;94;95;97;101;107;108;113] Two evaluative studies were identified.[84;92] No randomized controlled trials were identified.

The number of published papers has increased substantially since the first review was published, from 31 in the period covered by the first review to 38 since 2003.

Table 3: Types of studies included in the systematic review

	Studies included in Wilson *et al.*.			**Studies included in update**		
Study Type	Author	Year	N	Author	Year	N
Evaluation of Tools	--	--	--	Van den Niewenhoff	2006	16
	--	--	--	McKinnon	2007	28
Interviews	Adelsward	2003	31	Berth	2006	76
	Claes	2003	63	Blase	2007	12
	Cox	1999	51	Bradbury	2007	128
	d'Agincourt Canning	2001	36	Daly	2003	26
	Duster	1999	369	ForrestKeenan	2005	56
	Fanos	1995	84	Foster	2004	15
	Fitzpatrick	1990	96	Gadzicki	2006	332
	Forrest	2003	37	Gaff	2005	12
	Green	1997	46	Gallo	2005	139
	Hallowell	2003	30	Gregory	2007	44
	Henneman	2002	18	Hallowell	2005	29
	Hughes	2002	43	Hamilton	2005	29
	Koehly	2002	36	Holt	2006	8
	Peterson	2003	39	Kasparian	2006	40
	Shakespeare	1993	42	Kenen	2004	21
	Skirton	1998	15	Kenen	2004b	21
	Smith	2002	305	Klitzman	2007	21
	Sorenson	2003	136	Lim	2004	47
	Suslak	1985	12	Mellon	2007	292
	Wagner Costalas	2003	162	Mesters	2005	30
				Van den Nieuwenhoff	2006	16
				Van den Nieuwenhoff	2007	20
				van Oostrom	2007	271
Randomized control trials	--	--	--	--	--	--
Surveys	Ayme	1993	289	Blandy	2003	30
	Bruce	2003	78	Bowen	2004	221
	Claes	2003	63	Farkas Patenaude	2006	273
	Denayer	1992a	183	Kohut	2007	105
	Denayer	1992b	109	Landsbergen	2005	50
	Julian-Reynier	1996	209	MacDonald	2007	122
	Julian-Reynier	2000	398	MacKinnon	2007	28
	Miesfeldt	2003	267	McGivern	2004	38
	Ormond	2003	48	Riedijk	2005	66
	Sorenson	2003	136	Schroy	2005	71
	Varekamp	1992	500	Segal	2004	31
	Wagner Costalas	2003	162	Taylor	2005	57
				Weiner	2005	717
Focus Groups	--	--	--	Mellon	2006	39
Prospective reporting	--	--	--	Clarke	2005	65
Prospective surveys	--	--	--	van Oostrom	2007	271
Questionnaire in prospective cohort	Lerman	1998	201	--	--	--
Ascertainment of chromosomal translocations	Wolff	1989	36	--	--	--

The most commonly researched genetic disorder, with regard to family communication, has been hereditary breast and ovarian cancer (HBOC) and its associated BRCA1 and BRCA2 mutations (Table 4). In total, 35 studies examined this disorder; 12 were from before 2003.[14-16;28;31;100;104;105;107;108;111;113] Six studies published after 2003 focused on huntington's disease (HD),[2;67;75;77;80;91] and 5 prior to 2003;[31;98;99;109;110] however, for two of these only abstracts were presented[98;109] and one was a thesis dissertation.[99] Seven published papers had a sample of cystic fibrosis families,[20;23-26;102] only one of which was done recently but did not focus on communication of genetic test results; it examined parents' communication about the disorder in general and the 'genetic nature of the condition' with their affected children.[72] This same study also included the disorders hemophilia, sickle cell anemia, thalassemia, phenylketonuria (PKU), Marfan syndrome, neurofibromatosis, and von Willebrand disease [72] (some not included in the table below because no relevant information was presented). Communication about HNPCC and colorectal adenoma risk was researched in seven studies,[19;21;62;71;81;89;106] and balanced chromosome translocations in four.[67;97;112;114] All research on balanced translocations was done prior to 2003, the only recent publication yielding information based on professional reports of patients' experiences of disclosure.[67] Research on communication and genetic risk has also covered hemophilia, hereditary melanoma,[76;88] duchenne muscular dystrophy,[103] hereditary hearing loss, [64] inherited high cholesterol (IHC), [92;93] and sickle cell anemia in varying degrees of depth.

Table 4: Genetic conditions identified in the systematic review

Disorder	Inheritance	Citations from Wilson *et al.*	Citations from Update
Childhood Onset			
Hearing loss (GJB2, GJB6)	Autosomal dominant		64
Cystic fibrosis	Autosomal recessive	20;24-26;101;102	
Sickle cell anemia	Autosomal recessive	102	
Duchenne muscular dystrophy	x-linked recessive	103	
Hemophilia	x-linked recessive	22;29	72;73
Chromosome translocations / balanced translocations	Complex inheritance pattern	97;112;114	67
Late Onset			
Hereditary breast and ovarian cancer (BRCA1/2)	Autosomal dominant	14;16;28;31;74;96;100;105; 107;108;111;113	2;17;18;63;65-70;74;77;78;82-87;90;94;116
Hereditary non-polyposis colon cancer (HNPCC, colorectal adenoma)	Autosomal dominant	21;106	19;62;71;81;89
Hereditary melanoma	Autosomal dominant		76;88
Huntington's disease	Autosomal dominant	31;98;99;109;110	2;67;75;77;80;91
Inherited high cholesterol (LDLR, APOB, PSCSK9, LDLRAP1)	Autosomal dominant or recessive		92;93

Thus, a wide range of different inheritance patterns have been examined including autosomal recessive (sickle cell disease, cystic fibrosis), x-linked recessive (hemophilia, duchenne muscular dystrophy), autosomal dominant (HBOC, HD, HNPCC, hearing loss, IHC and melanoma) and more complex inheritance patterns as seen with chromosome translocations. However, research has predominantly focused on autosomal dominant

disorders which appear in adulthood (HBOC and HD). Two were carried out on haemophilia,[72;73] one on chromosome translocations,[67] and one on cystic fibrosis[72] since 2003 indicating that research on recessive inheritance patterns and complex inheritance patterns has decreased. This also indicates that research on family communication and disorders with childhood onset are being researched less than in the past. Research on family communication of genetic risk information is sparse or non-existent for other disorders. There has also been an increase in the number of research studies examining disorders with a new-found genetic susceptibility and for which genetic testing has recently become available, such as melanoma and inherited high cholesterol.

Table 5: Potential recipients of risk information identified by the systematic review

Potential recipients	**Number of Studies**	**Citations**
First degree		
Any	13	17;18;20;28;63;65;84;87;89;92;93;109;112
Siblings	15	14;24;25;64;67;70;71;77;78;80;100;107;111;113
Parents	6	14;25;64;80;94;100
Children	19	14;66-68;70;72;74;75;77;78;80;90;94;100;107;108;110;111;113
Second degree		
Any	12	17;18;20;63;65;77;84;92-94;109;112
Grandparents	1	64
Uncles or Aunts	4	14;64;100;101
Nieces or Nephews	1	70
Third degree		
Any	8	18;20;63;65;77;84;92;112
Cousins	4	14;64;70;100
Members or members of family group not Specified	23	15;16;19;21;26;29;31;69;73;76;80;81;83;86;88;95;96;102;104;106;114;116

Note: one study can be included in several categories

Included studies varied in which family members were the targets for the disclosure (or the recipient of the information). In 13 studies the potential recipient for risk information was any first degree relative.[17;18;20;28;63;65;84;87;89;92;93;109;112] In 15 studies the recipient of the information was specified as siblings,[14;24;25;64;67;70;71;77;78;80;100;107;111;113] in 6 they were parents, [14;25;64;80;94;100] and 19 they were children. [14;66-68;70;72;74;75;77;78;80;90;94;100;107;108;110;111;113] Twelve studies examined any second-degree relative as the receiver of the information.[17;18;20;63;65;77;84;92-94;109;112] One study specified inclusion of grandparents [64] and another one for nieces and nephews. [70] Four studies looked at aunts and uncles as a specific category of a second-degree relative.[14;64;100;101] In 8 studies the recipient of the information was any third-degree relative[18;20;63;65;77;84;92;112] and in four were specified as cousins who were being examined as a recipient. [14;64;70;100] Some left the definition of family undefined to the reader, thus the precise family members which were included in the investigation of the study were not specifically known; examples of undefined family terms included 'extended family',[71;80] 'family',[15;19;26;29;31;62;69;73;78;81;85;88;102;104;106;114] 'wider family',[82] 'relatives',[76;96] 'biological family',[2] 'immediate family',[76;82] 'other relatives',[67;83;93] and 'at risk relatives'.[16;21;97;99] Some only looked at certain first-degree family members such as sisters and children,[83] first- and second- degree female relatives,[85] sisters only,[105] at risk female relatives.[22] Some looked at children only[68;72;75;90;108;110] or sons only.[103]

The majority of the findings on barriers and facilitators since 2003 are consistent those found by Wilson *et al.*, (summarized in chapter I). Appendix F describes the participants and recruitment of the included studies and Appendix G summarizes the main findings of all studies included in this review.

Barriers & Facilitators

Table 6 is a comprehensive list of all the barriers and facilitators of communication of risk supported by evidence reported by the included studies. A wide and diverse range of themes was identified (table 6). The most frequently identified themes were: the desire to protect relative(s) from negative consequences of genetic information, feelings of responsibility towards relatives, assessments of the relevance of the genetic information for relative(s), beliefs about the right of relatives to information (or the rights of relatives to decline information), and various factors relating to the nature, severity, and age of onset of the genetic condition in question.

Table 6: Barriers and facilitators identified in the systematic review

Study	Barriers	Facilitators
Included Studies from Systematic Review Update		
Berth 2006[62]*	• Age of recipient • Gender • Index patient vs. healthy consumer • Degree of relationship • Psychological distress • Family characteristics	• Age of recipient • Gender • Index patient vs. healthy consumer • Degree of relationship • Psychological distress • Family characteristics
Blandy 2003[63]	• Age of recipient • Being male • A family who believes cancer is a 'taboo subject' • Coping strategies like denial and blunting behaviours • Poor knowledge, misunderstandings, confusion between different risks, the persistence of false beliefs	• Being female • Having good family cohesion and support • Feeling responsible for the health of others
Blase 2007[64]	• Social/geographic distance • Perceiving the information as being irrelevant • Desire to protect • Family communication set-up does not allow for sharing with particular members	• Being a first degree relative • Feeling a duty or responsibility • Normal passive information sharing • Wanting to keep family up-to-date
Bowen 2004[65]	• None	• Being a female relative (especially a mother or sister)
Bradbury 2007[66]	• Young age of recipient • Might increase child's anxiety, fear or worry • Parent still coping • No medical reason for disclosure	• Older age of recipient (offspring) • desire to provide information and awareness • wanting the children to be tested • having to explain the family history or medical intervention
Clarke 2005[67]	• Significant life events (heroin addiction, imprisonment, major mental illness) • Adoption (even if able to contact) • Gender ambiguity and paternity (family secrets) • Bad family relationships • Desire to protect relative • Desire to avoid causing anxiety • Judgments as to whether their relatives personally needed the information or could cope with it • Problematic family dynamics	• None

Study	Barriers	Facilitators
	• Loss of contact • Fear that disclosure might disrupt a family member's marriage plans • Another relative does not allow the disclosure • Fear of being blamed • Feeling unable or unwilling to take responsibility for informing relatives • Still expressing strong emotion from their test result • Issue of privacy • Beliefs about whether it is better to know or not know	
Daly 2003[68]	• Fear that telling will cause stress on people and make the disease come earlier (myth) • Past experience with mother and family members dying makes them especially sensitive to the pain the disease can cause	• None
Farkas Patenaude 2006[87]	• Having an inconclusive test result (perception that telling would be of little or no use to relatives) • Older women less likely to tell mother and father • Lay understandings or incorrect information about inheritance of the disorder • Young age of recipient / child (understanding)	• Being female • Having a conclusive test result (yes/no) • Younger women were more likely to tell their test result to parents than older women • Brothers are told more often if the mutation is was in the paternal side of the family • If a family member has already had cancer (the lines of communication about the topic are already open)
Foster 2004[69]	• The family had different people with different ways of coping with information and a different desire to know information • Uncertainty as to what information should be told to whom; especially men have restricted information flow. • Women who were found not to carry the mutation had difficulties when talking to carriers and relatives with cancer • Not wanting to cause upset to oneself and others • Being a non-carrier and having to tell a carrier • Feeling guilty (in this instance about having promoted testing for others then finding out they themselves did not have the mutation) • Estranged relatives who might use the information against them.	• Feeling a responsibility to tell healthy relatives about genetic testing so that they could do something to modify their risk • A death of a family member (who would have told children) made the responsibility theirs • Open communication with family members
Gadzicki 2006[70]	• Inconclusive test result (no p value so uncertain whether it was significant relationship)	• Being female • Having a true negative result (shared with nieces and nephews more than carriers)
Gaff 2005[71]	• Believing the person will not be interested in the result	• None

Study	Barriers	Facilitators
	• Men tell less than women to extended family members • Lack of a good relationship • Lack of contact with the individual • Anticipated disinterest in testing or a combination or reasons • Believing the at risk relative could not undergo testing or if the relative was considered too young for surveillance • Social or geographical distance • They perceive the information to be irrelevant to that individual	
Gallo 2005[72]	• Young age of the recipient • Desire to protect (maintain their childhood) • Child is considered too immature • Fear of blame for passing it on	• None
Gregory 2007[73]	• Being an obligate carrier of hemophilia (i.e. father has hemophilia) • Time not being right (maturity of the child, not relevant at that point in their life)	• Being a non-obligate carrier (i.e. brother has hemophilia) • Life situations which make the information more relevant such as planning a pregnancy or being in a serious relationship
Hallowell 2005[74]	• An internal conflict between respecting their children's rights to information about their risks and their parental duty not to cause them needless anxiety • Situational factors: pregnancy, emotionally fragile • Feeling a responsibility to protect their children from anxiety provoking information • Child is less than 18 years of age • A pre-existing culture of discussion about cancer within family may account for disclosure method chosen	• Dislike of secrecy
Hamilton 2005[2]	• Age (as related to how relevant the information is) • Situational factors (in graduate school etc) • Personality factors (vulnerability, their anticipated receptivity of the information) • Mental and physical conditions • The harm the news of genetic test result might cause (especially if they misinterpret the information) • Causing distress to family member • Having to take care of the distressed person once they know • Not wanting to be the bearer of bad news (feeling guilty) • Not wanted to losing connections with family members who identify themselves as being 'at risk'	• None
Holt 2006[75]	• Desire to maintain hope and preserve emotional health of the	• Dislike of secrecy

Study	Barriers	Facilitators
	family • Desire to protect	• Previous personal experience of being held in the dark
Kasparian 2006[76]	• Worried about how family members may cope or react • Worried about the unwell, elderly or illness focused	• Desire to discuss the need for relatives to practice precaution adoption.
Keenan 2005[77]	• Not feeling a sense of responsibility • Believed they don't have the authority to tell over certain family members heads (i.e. A brother died, and the mother of his children was now responsible to tell the kids – not the uncle). • Not knowing that information was important for males • Past family conflicts • Contemporary notions of family (always changing – not stable structure) allows for feeling very different obligations to different family members	• Close emotionally • First degree relatives
Kenen 2004a[116]	• Fear of causing anxiety in others • Others won't listen (there is a different desire to talk about it) • One family member hides family history of illness • Difficult relationships • Desire to protect members from bad news (not wanting to bring back anger and grief) • Protect children's childhood	• Sense of responsibility to disseminate information
Kenen 2004b[78]	• Emotions about cancer get in the way of conversations • geographical distance • emotional distance • Wanting to protect children • Poor knowledge that men could be at risk • Blocked communication pattern (with sons in this instance)	• None
Klitzman 2007[80]	• Want to preserve their privacy or confidentiality • Stigma of the disorder • Desire not to burden others • The other does not want to know • The individual can't handle the information • Don't want to make others feel guilty • Past and anticipated interpersonal conflicts	• To receive support • Feel a responsibility to help others health • Feel they have a right o know their risk • Family members have children
Kohut 2007[81]	• No contact • Older age of the recipient (elderly relative) • Young age of the recipient • Living far away from relative • Family does not want to discuss the issue	• None

Study	Barriers	Facilitators
	• A desire not to worry them (protection)	
Landsbergen 2005[17]	• Relative had already been informed • Lack of contact with the family member • Information was too emotional • Thought that the information would be too difficult for the family member • Felt insufficiently informed (less often) • Was to difficult and to emotional (less often) • Less contact (with nieces and nephews in particular in this instance)	• None
MacDonald 2007[83]	• Concern about upsetting other • Recalling painful memories • Geographic distance • Information not useful • Difficult family relationships • Difficulty talking about cancer risk • Age differences • Lost touch with relatives	• Being female • Being a first degree relative
McGivern 2004[18]	• Not having the decision power to over rule parents decisions about nieces and nephews • Not wanting to upset the relative (less often) • Having difficulty coping (less often) • Not feeling close to the family member • Not being in contact with the family member • Being a second or third degree relative • Not knowing what to say (less often)	• Feel a duty to inform them of their risk • Having a pathological mutation test result • Using communication as a strategy to cope • The result was already expected by the family • Being a first degree relative
Mellon 2006[85]	• Fear that talking to them about the risk will make them avoid pursuing cancer risk information • Worry for causing children undue stress • Some participants expressed ambivalence regarding how to balance anxiety about testing with the desire to know • Lack of proven options for risk interventions caused added stress (not having a definite solution after finding out one's risk status was to stressful and would not be worth pursuing more knowledge and or testing) • Insurance discrimination • Cost of genetic testing	• None
Mellon 2007[86]	• Uncertainty about their reactions • Adding more stress	• Close relationship • Desire to keep family members informed

Study	Barriers	Facilitators
	• Previous family conflict • Emotional distance • Young age of the recipient	• Receive emotional support • Relying on knowledge from the individual • Concern for family members at potential risk
Mesters 2005[19]	• Young age of the recipient (understanding) • Bad first attempt at disclosure • Anticipated rejection of message by biological family member • Do not feel it is their responsibility (in this instance, to inform cousins because it is their mom and dads role) • Conflicts between family members • Conflicts about the subject already existing in the nuclear family • Conflicting opinions about disclosure between the carrier and partner • Poor understanding of who it is important to tell (Thinking it is only important to tell first degree relatives) • Being unaware there is a family history because others hid information.	• Felt they had a moral obligation • Anticipated regret • External cues such as medical organizations/professionals telling you to tell family members (gave people the feeling that what they were doing was ok) • Perceived susceptibility is inflated because someone else has already had or died from it
Riedijk 2005[88]	• An individual who is emotionally motivated would be less likely to inform their family about an unfavorable test result • Emotionally motivated people had unrealistic perceptions of what caused melanoma and had more anxiety	• None
Segal 2004[90]	• Young age of the recipient • Not wanting to cause worry when there was no concrete information or tangible solutions (only for 2 kids) • Concerns about insurance discrimination, feeling confused about whether or not to disclose • Being worried • Feeling ill-prepared to disclosure the genetic test result and its implications • Gender	• Belief that they had the right to know • Concern for children's risks
Taylor 2005[91]	• Distant relationship	• Close relationship • Being female
Van den Nieuwenhoff 2006[92]	• Thinking informing relatives about a genetic risk was a burden • Not having personal contact (distant relatives)	• written materials for patients and relatives (external cues) • cue to action • feeling a responsibility to tell
Van den Nieuwenhoff 2007[93]	• Desire to protect relatives from psychological harm at that moment • Relatives who were already distressed by other life events and children whose parents wanted to prevent them from worrying	• practitioners advice • moral duty to inform (driven by strong family bonds, perceptions of relatives susceptibility, perceived severity of IHC, the possibility of treatment, and consistency with ones values and or integrity)

Study	Barriers	Facilitators
	about their own and their parents health • Not feeling a responsibility (reflecting cultural norms regarding the boundaries of familial responsibility) • Lack of contact between relatives • Anticipated the message would be rejected • Ignorant of the possible risk for distant relatives • Superficial knowledge about close relatives risks • Misconceptions about the mode of transmission • Inadequate risk knowledge (stemming from professionals advice being restrictive or open to multiple interpretations, contradictory messages from health care professionals, physicians who may have contributed to the IPs opinions that their IHC was not serious or important for the family, general criteria used by professionals for diagnosis and treatment of (inherited) high cholesterol levels varied as time went on, and public opinion that lifestyle is the cause of high cholesterol levels) • Denial • Low self-efficacy regarding disclosure	• altruistic desire or perceived obligation to protect relatives from avoidable physical harm • roles within the family (the responsible one..) and self concept in family relationships (viewing oneself as helpful to relatives) • enhance the probands status within the family (being the one to tell and help, making themselves seem less responsible for their disorder, or shifting focus onto how they are helping instead of an alcoholic) • reduce IPs anticipated negative emotions of grief or guilt if someone becomes ill later (regret) • cultural norm of responsibility for close relatives • fear of losing relatives later • receive emotional support
Weiner 2005[95]	• None	• Being female • Having a test
Included Studies from Wilson *et al*		
Adelsward 2003[96]	• Desire not to harm	None
Claes 2003[14]	• Close relatives more likely to be informed than distant relatives • Assumed other relatives were going to tell or had already told • Felt the information was not important for them (inconclusive) • Little and or superficial contact • Person-specific reasons (e.g. not information an aunt because of her age, not informing the daunts daughter because their was a lack of contact)	• Feeling the information was important to the relative • Feeling the individual needs to take preventive action • Having obtained diagnostic testing • Having a close relationship • Having a conclusive result (distant relatives were more often informed if the result was conclusive) • Younger (younger relatives informed distant relatives more)
Cox 1999[99]	• Fear of stigmatization	• Being female
d'Agincourt Canning 2001[100]	• Worried to cause anxiety • Don't want to be the bearer of bad news • Afraid the persons may hold the information against you • Guilt (for passing on the disorder)	• Feeling a sense of responsibility • Feeling that relatives had a right to know • Being female

Study	Barriers	Facilitators
Duster 1999 [102]	• Fear of employment and insurance discrimination	• None
Fanos 1995[25]	• Avoidance of guilt (for not being at risk when a sibling is affected) and envy (by a sibling who is affected) • Fear of blame • Fear of loss of interpersonal desirability (one more thing to bring into a relationship) • Erroneous thinking (ex. thinking that because they are like their sibling they have the gene mutation and thus, do not need testing) *barriers apply to transmission of genetic information – not specifically genetic test results	• Having open communication style in family
Fitzpatrick 1990[103]	• Culture	• None
Forrest 2003[31]	• Not having contact with family member • Not feeling they have the authority to tell • Not feeling they have the responsibility to tell • Key family member has died (who normally would have the authority and responsibility) • There are two communication patterns in one family (pragmatics and prevacators) • Family tensions • Young age of recipient (children) • Having different coping styles in a family • Having an uncertain message or not being able to make sense of their risk • Thinking they do not need to know • Pre-existing family conflict that cannot be overcome • Desire to protect family members	• Being female • Major life events are facilitators in some instances because the right time for them to know • Parents thought it was the right time in terms of health decisions or life decisions. • Wanting to obtain more information about the family and disease
Green 1997[15]	• *In getting information for the initial counseling session:* • Geographically and emotionally distant relationships • The person was old or ill • Not wanting to upset people with painful memories • Not getting along with a stepparent or an in-law • Not being close with siblings because of a big age gap • Having lost touch with a 'branch' of the family • Being male exacerbated the problem • *In disseminating information after the clinic session:* • Desire not to upset • Social or geographical distance	• *In disseminating information after the clinic session:* • Believing the family member has a right to know • Having a leader in the family who feels that it is their role to ensure that everyone is told
Hallowell 2003[104]	• Some were in moral conflict about not wanting to harm their	• Wanting to provide information for making health choices

Study	Barriers	Facilitators
	relatives but wanting them to have the information • Desire to avoid causing anxiety in other members	• Feeling like they had a responsibility
Henneman 2002[26]	• Situational factors (pregnancy) • Not planning on having children at the moment • Problems talking openly in general • They felt others would not support the idea of abortion	• None
Hughes 2002[105]	• Inconclusive test results (not clear as to why reported less often) • Not being close to the family member • Thinking they will misinterpret their information • Lack of opportunity to provide test results information to the relatives • Not wanting to upset the relative • Experiencing guilt and anxiety (less important) • Concerned about confidentiality (less important)	• Wanting to provide risk information to relative • Wanting emotional support • To get advice about medical decisions (?)
Julian-Reynier 1996[16]	• Difficult family relationships • Lack of perceived usefulness • The unpleasantness of the message • Desire to avoid causing a disturbance • Likelihood the message would be rejected	• None
Julian-Reynier 2000[28]	• None	• Being female • Older age of recipient • Having had cancer or experience in the family with cancer • Being a sibling
Koehly 2002[106]	• None	• Being a first degree relative • Being female • Being a mother (a key person) • Being a spouse • Getting along • Listening to each other and take each others advice • Close relationship
Lerman 1998[107]	• Distant relationship (from a case study) • Established patterns of communication in family (from a case study) • Occurrence of other life circumstances (new marriage, pregnancy, chronic illness) (from a case study)	• Being female
Ormond 2003[20]	• Family history of CF • Young age of recipient (children)	• Desire for social support • Having a close social relationship

Study	Barriers	Facilitators
	• Social and geographical distance • Life circumstances (i.e. done having children, being unmarried)	• Thinking that they should know the information for future decisions in life and marriage and reproduction • Being close biologically i.e. First degree relatives • Being a maternal relative • Having a child already affected with the disease (Because they have to explain the illness of the child)
Peterson 2003[21]	• Not feeling they have the authority to tell • Not feeling it is their responsibility to tell	• None
Shakespeare 1993[109]	• A person acts to deliberately block information transmission	• None
Skirton 1998[110]	• Perceptions of the disease • Perceptions the individual has to information • Desire to protect • Retaining control over the lives of their children • Delay telling in order to protect the child from anxiety for as long as possible • Lack of skills for conversations or disclosure	• Desire to act honorably and respectfully towards their children • Desire to give the children an opportunity to plan and make informed decisions
Smith 2002[111]	• Not being close • Carrier status (can influence depending on gender) • Thinking they already know • The topic 'never came up' • Young age of the recipient • Worry of discrimination – health and employment (for telling co-workers)	• None
Wagner Costalas[113] 2003	• None	• Being female • Having a positive test result
Wolff 1989[114]	• Not personally knowing relative • Not being able to contact relative • Fear of psychological and social consequences of positive test • Denial • Coping mechanism (to deal with stigmatization)	• None

* direction of influence of each variable not specified, only an abstract presented (not included in framework due to lack of evidence and identification of direction of effect

Organization of the Barriers according to the TPB

From the systematic review, all of the barriers listed by author were reorganized into a list of distinct factors, with no repetition. The citation of each paper which mentioned each barrier or facilitator was noted in a separate column to ensure that the number of times the barrier or facilitator was supported by evidence was noted. Next, each barrier and facilitator was placed within the appropriate construct of the TPB. Tables 7a through 7c organize the barriers from the review into the attitude, subjective norm and perceived behavioural control domains of the TPB. Each table shows how the different barriers and facilitators identified in the literature relate to its underlying theme and which papers included that barrier or facilitator. Using the attitude construct as an example, 'a desire to protect' is one category within that TPB construct. This category encapsulates the barriers: not disclosing to a relative due to fear of causing them upset and pain (protecting others), not disclosing to a relative due to a fear that the individual will blame the messenger (protecting oneself) and not disclosing to children at an early age to allow them to have a happy childhood (protecting others). For a more extensive elaboration of all the specific factors which are encompassed in each category under each TPB construct see Appendix H.

In total all of the barriers and facilitators of genetic risk information disclosure to family members identified in the systematic review were placed into five attitude categories, three subjective norm categories, and four perceived behavioural control categories. Other influential factors involved in forming a person's attitude, perceived behavioural control and subjective norm for this specific behaviour identified in the literature include culture, past experience, personality, disease severity and gender. These factors impact perceptions and beliefs which in turn alter the importance of each construct in performing the behaviour.

Table 7a: Barriers and facilitators relating to the attitude domain of the TPB

Category	Interpretations in literature	Citations
Desire to Protect	Wanting to protect oneself from guilt, blame, reactions of family members, ending of a relationship	2;18-20;25;26;29;63;65;67;69;73;75;78;80;82;83;85;86;93;97;100;105;107;110;114;116
	Wanting to protect other family members from pain, anxiety, distress, emotional turmoil, and discrimination	2;15-20;25;26;28;29;31;63;64;66;67;69;71-76;78;80;81;83;85;86;90;91;93;97;100;103-106;108;110;111;114;116
Perception of Relevance	There is an actual increased relevance of the information for the individual because they may be a future care giver (spouse / GP)	28
	There is a perceived decrease in relevance because the disease is not fatal, the individual is to young to be affected by the news, the individuals life situation (mental illness or pregnant) is more important than the genetic message, or there is no family history of disease	15;16;20;22;26;29;31;63;64;66;67;73;76;81;87;93
	Interpretation of a test result (Inconclusive test results can be interpreted as meaningless, conclusive mutation positive test results as very relevant, conclusive mutation negative as not relevant or very relevant)	14;20;21;31;70;87;105;113
	Low perceptions of the other family members' interest in the information (thinking they would not want to be informed or that they have already been informed)	14;26;66;72;73;110;111

	High perceptions of the other family members' interest in the information (because they have been affected by the disorder, they have witnessed a strong family history of the disorder, they are at a high risk for the disorder, there are methods to decrease risk, or because they may be an important decision making point in their life – reproduction, marriage, sexual activity or family myths)	14;19;21;28;31;64;66;69;73;76;77;83;87;106;110;111
	Low perception of relevance due to misinformation (Misunderstanding the information, lay beliefs about inheritance and family myths Misunderstanding information about inheritance and myths)	15;19;63;77;85;87;93
Perceptions of Responsibility	Not feeling responsible for telling	67;93
	Feeling a responsibility or duty to pass on the genetic risk information	18;19;63;64;68;69;77;80;92;93;100
Perceptions of 'Right to Know'	Feeling that genetic information belongs to other family members	64;74;80;86;100;110
Usefulness of Communication	No use in informing because they anticipate their relative will be disinterested in testing or will reject the message	16;71;81;93
	No use in informing because their family member would not have access to testing (financial or no services)	71
	Personal use in telling because the teller may be able to gain useful information (ex. about family history, or about symptom management)	86

Table 7b: Barriers and facilitators relating to the subjective norm domain of the TPB

Category	Interpretations in literature	Citations
Pressure from Family Members	Taboo and shame	15;29;63;69;93;106;107
	Conforming to established family or gender roles (as the communicator or 'kin keeper)	15;69;93;106;107
	Others pressure teller not to discuss topic	26;67;70
	Familial lines of authority (ex. only parents tell sensitive information to their children)	21;31;64;106
Pressure from Professionals	External cues from professionals (informing you who to tell, reminders etc)	19;31;92;93;97
Pressure from Society	Openness about discussing in society	21;31;69
	Taboo and shame	29;63

Table 7c: Barriers and facilitators relating to the perceived behavioural control domain of the TPB

Category	Interpretations in literature	Citations
Family Dynamics and Relationships	Unhealthy family relationships	14-16;19;31;67;69-71;77;80;83;86;106
	Lack of contact with person	14;15;17;21;29;31;71;81;83;92;93;100;114
	Close social relationship	14;19;20;64;83;86;87;91;106
	Superficial relationship or not being close/not emotionally close	15;18;20;29;64;83;86;105;106;111
	Geographical distance	15;20;64;78;81;83
	Disorder is already openly communicated in the family	31;82;87
	Adoption	18
	Generally good communication, and supportive, and take each others advice, and generally get along	63;73;74;86;106;107
Communication Skills	Having a complex message to convey (Inconclusive test results and other complex inheritance and genetic information)	14;23;24;31;70;72;87;105;112;113
	Don't think they have communication skills (or do not communicate in general) or not know what to say	18;26;93;110
Ability to Understand	Complexity of information (Conclusive vs. Inconclusive test results)	14;23;24;31;70;72;87;105;112;113
	Older age (greater ability to understand)	66
Coping Skills	Having difficulty coping with own problems/diagnosis/test result	18;31;66

Framework of family communication and genetic information

The final framework developed using the TPB and the literature review is depicted in Figure 4 below. The framework explains how the barriers and facilitators in disclosing genetic test results and genetic risk information to at-risk family members might interact in terms of the TPB. This model is, in effect, a TPB model of the behaviour of disclosure of genetic test results and thus should function the same as the generic TPB in terms of prediction of intention and behaviour. The framework generalizes the specific barriers identified in the systematic review by their underlying themes.

Thus, the factors within each TPB construct should act in a positive or negative way to form an individual's attitude, PBC and SN, which in turn, should predict their intention to perform the behaviour. Factors influential in forming any three of the constructs (their attitude, PBC and SN) with regard to disclosure may be in conflict, may all be positive or may all be negative. Some, all, or none of those particular factors might apply in the formation of each construct for any given individual. In keeping with the TPB, intention is construed as the sum of all three constructs [47;59;61]. The different weight each construct carries for each individual should be different because attitudes, PBC and subjective norm, and the factors which influence their formation, will be different between people and situations. Similarly, factors which influence those three concepts are also different between people and behaviours.

In the case of disclosure of genetic test results to at-risk family members, this framework predicts that an individual's attitude towards performing the behaviour is potentially formed by one or more of the following factors: whether the individual feels a desire to protect the relative in question and/or herself or himself from harmful news,

perception of the relevance of the genetic risk information for the relative, perception of his or her responsibilities regarding disclosure, assessment of the relative's right to the genetic information, and perception of how useful the information will be to that relative. Four themes emerged which affect an individual's perceived behavioral control: a) the family dynamics and relationships within the family, b) the individual's communication skills, c) whether the individual has the ability to understand complex genetic information, and d) whether the individual has skills to cope with his or her own bad news as well as the reactions of others

In terms of the pressure that an individual feels to perform (or not perform) the behaviour (i.e. communicate or not), the literature identified three sources of pressure: a) other family members, for example a relative who wants to control who knows what, or a receiver of information who does not want to know; b) genetic professionals or other health professionals, and c) society as a whole in the form of the "appropriateness" of discussing such topics.

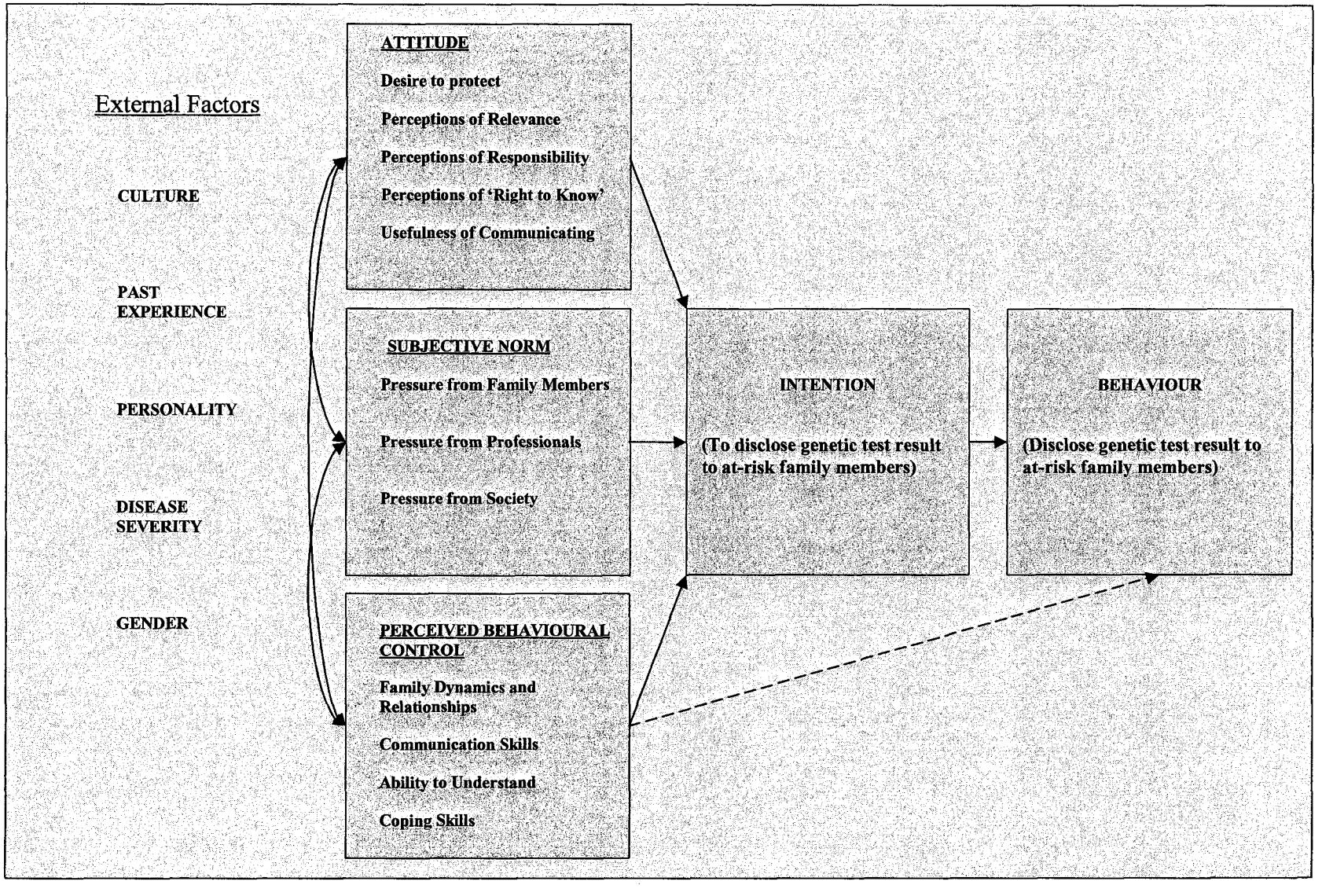


Figure 4: Theory of Planned Behaviour Framework of Family Communication of Genetic Risk Information

All three constructs and factors can influence each other to determine whether the individual will have the intention to do the behaviour and in turn, perform the behaviour. For example, an individual might have an overall attitude that she has a responsibility to tell, and that her relative has a right to the information; however she is wary of telling her relative because she does not want to cause worry. Therefore, her attitude might be slightly positive towards telling. However, she might also perceive that, within her family, talking about these types of topics is unacceptable, so she feels less able to actually tell a relative, even though she thinks she can communicate the message effectively and cope with the relative's anticipated (painful) response. Thus the subtle pressure and norms from her family members produce a negative perceived behavioural control and negative subjective norm. Overall, the strength of her attitudes, perception of her control over the behaviour and the pressure to tell or not tell will determine if she intends to tell the relative. In this example, this individual will not disclose because she feels the social pressure from her family against communication very strongly even though she believes that the relative has the right to know and she has a responsibility to disclose the information.

Things that are influential in forming a person's attitude, perceived behavioural control and subjective norm for this specific behaviour include culture, past experience, personality, disease severity and gender. The theory suggests that the weight of each of these factors is also going to be different between situations and people.

CHAPTER IV

PRELIMINARY TESTING OF THE FRAMEWORK

Aims and Objectives

After finalization of the theoretical framework explaining the barriers and facilitators to communicating genetic risk information to family members, a cross sectional survey was designed in order to test the face validity of the framework in a patient population. The objectives of the survey were to:

1) examine, in a preliminary manner, the relevance and applicability of the framework in a population who had undergone genetic testing and who would have at-risk family members.

2) test the feasibility of using a survey based approach which incorporates the framework, as a way of generating specific insights into barriers and facilitators of communication in genetic patient populations.

3) assuming an adequate response to the survey, to test the actual predictive power of the framework.

4) gain insight into the communication process and experience of a population which has yet to be examined in the literature.

Methods

Study Sample

The sample population consisted of adults who had undergone genetic testing at the CHEO genetics clinic for myotonic dystrophy, type 1 (DM1) within the previous ten years. The CHEO genetics clinic is one of nine regional genetics clinics in Ontario for genetic assessment, diagnosis, counseling and testing. It serves adult and children from eastern Ontario and western Quebec as well as an outreach clinic to those in Northern Ontario.

DM1 is a hereditary autosomal dominant, multi-system disorder which affects skeletal and smooth muscle, the eyes, heart, endocrine system and central nervous system. Anticipation, i.e. the occurrence of a lower age of onset and more severe manifestations of an inherited disease with each successive generation, is a feature of DM1.[117] Clinical presentation includes myotonia (sustained muscle contraction), muscle weakness and wasting, cataracts, and variable cognitive dysfunction. Cardiac conduction abnormalities of varying degrees of severity are common. These defects are a significant cause of early mortality in individuals with DM1, and less commonly, cardiomyopathy may occur.[118] Clinical findings range from mild to severe and have been categorized, by age of onset and predominant symptoms, into three somewhat overlapping phenotypes: mild, classic (encompassing childhood and adult DM1), and congenital. [119;120] DM1 was chosen as the disorder to investigate in this study for several reasons: 1) because it has never been studied and displays anticipation and different phenotypes which had the potential to produce information on topics not yet understood, 2) because it was one of the few populations at the CHEO genetics clinic not recently involved in research studies and 3) because there was a large number of eligible participants.

A genetic counsellor (CH) screened the genetic clinic's database of patient demographics (DEMOG) to determine the number of patients seen for counselling and testing of DM1 from January 1st 1997 to December 31st, 2006. The search strategy for the DEMOG database included: all records with 'myotonic' in the referring diagnosis field plus all records in which the 'seen by' field included a genetic counsellor or geneticist plus all records with birthdates after March 1st 1989. Two hundred and sixty-one records for the identified patients were collected and reviewed by the genetic counsellor (CH) to determine whether the patients were eligible for inclusion in the study. Only patients who had a molecular genetic test for DMI were included. Patients who were identified as too vulnerable to participate (e.g. undergoing prenatal diagnosis for DM1 in a current pregnancy, parents of a child recently born with congenital DM1), who had molecular tests prior to January 1 1997, or who were likely too cognitively affected by the disorder to complete a survey, were excluded. Male and female patients with both positive and negative test results were included in the sample.

Recruitment

In order to maintain patient confidentiality, an arm's length recruitment method was used. After identifying the potential eligible participants, a CHEO employee who was not part of the research group, assembled the research packages and affixed labels with names and addresses to the envelopes. Thus, the researchers were never privy to any participant identifying information and only had access to study ID numbers. All patients from the genetics clinic determined to be eligible to participate were sent a study package. Included in the package was an invitation letter from the director of the CHEO genetics clinic, which

explained the study and that their participation was voluntary; an information form explaining the study in detail; the 15 page survey; and a pre-paid return envelope (See Appendix I, J and K for invitation letters, information forms and survey). Participants were also given the option to complete an online version of the survey.

The information form explained that if they did not wish to participate in the study they had three options: 1) they could call the research coordinator of the study (MW) and have their name removed from the mailing list; 2) they could send back a blank survey and their name would be removed from future mailings or; 3) they could ignore the correspondence. They were informed that they would at most receive three surveys by mail. Surveys were mailed approximately every three weeks; the first mail-out occurred August 08, 2007. Completing and returning the survey indicated their informed consent.

Instrument development: part 1

The cross-sectional survey consisted of two parts. The first part was designed to test the validity of the framework and was created as a TPB survey. Standard methods for TPB survey construction were used, guided by a manual designed for the purpose.[59] Not all barriers and facilitators listed in the framework could be included into the survey because the length of the survey would be impractical. The genetic counsellor (CH) reviewed the barriers in the framework and identified the ones she saw most frequently in clinical practice, and these were the barriers and facilitators examined in the survey.

Defining the behaviour

To create a TPB survey, the behaviour of interest must first be appropriately defined. The TACT acronym is often used to define the behaviour in terms of its target, action, context and time.[59] The behaviour of interest in this study was 'disclosure of genetic test

results to at-risk family members by patients who have had genetic testing for DM1 and received a conclusive result'. The target was 'at-risk family members', the action was disclosure of their genetic test result, the context was patients who underwent genetic testing and received a positive or negative test result for DM1. Time is left undefined, implicitly being the time between receiving their test result and the date they filled out the survey.

This particular behaviour was difficult to define since there are potentially many family members for whom communication of the genetic information would be relevant. We observed the 'principle of compatibility',[121] designed to ensure that all measures in the survey would have the same level of generality[59] (for example, a question referring to 'at-risk family members' and then a question referring to 'your at-risk brother' would not be compatible). The definition 'at risk family members' was too broad a target because 1) respondents might not be aware of every family member at risk; and 2) they might answer different statements thinking of different family members, thus making the data uninterpretable. It was not feasible or reasonable to create a survey exploring disclosure to each individual family member because some families are very extensive. Thus, in order to promote clarity, minimize respondent burden, and ensure validity, we decided to allow each participant to select a relative to whom they had not disclosed their genetic test result, and indicate the nature of their relationship. Respondents were directed to answer all the relevant questions with that one person in mind. This would allow the participant to define the behaviour in a way which was meaningful. See Appendix K for wording of the introduction to the survey and the specific instructions

Measurement of TPB constructs

The TPB construction manual[59] describes how to use the barriers and facilitators identified for a specific behaviour (in this case, disclosure of genetic risk information) to formulate questions about the participant's attitude (an attitude statement), subjective norm (a subjective norm statement), perceived behavioural control (perceived behavioural control statement) and intention. Attitude, subjective norm and perceived behavioural control factors were measured by both direct and indirect means; and each method of measurement had a specific format for phrasing the statement(s) for each construct. The use of a 7 point scale in which participants circled the number that best represented their opinion was used for each question, with different endpoints. The length of the scale is irrelevant in measuring the constructs[61]; a seven point scale was chosen because it allowed participants to have neutral response (4 out of 7 point scale) to any given question.

Behavioural Intention

Measurement of generalized intention to tell a family member about a genetic test result is achieved through three items which have good internal consistency. These items ask whether the individual 'plans to', 'will' and 'intends' to perform the target behaviour, which, in this case, was 'tell your family member about your genetic test result'.

Attitude statements

Direct measurement of attitude uses bipolar adjectives (pairs of opposites) which are evaluative in nature.[59] Four items were used following a single stem which defined the behaviour as disclosure. For example, the stem for this behaviour is 'talking to my family member about my genetic test result would be:' and this was followed by four different

bipolar adjectives ('very harmful' to 'very beneficial', 'very frustrating' to 'not at all frustrating' etc.).

Indirect measurement of attitude was also measured using one statement about their behavioural belief (what they believe would happen if they performed the behaviour) and a corresponding statement about their outcome evaluation (whether they felt that the outcome would be good or bad). For example, one important factor identified from the literature review was whether the individual felt that the information would be useful for their family member in making personal health decisions. Thus, to measure their behavioural belief about the usefulness of the information to their relatives, the survey item was 'talking to my family member about my genetic test result would give them information that would be useful for them when making their own decisions about their health' (strongly disagree to strongly agree) and the corresponding outcome evaluation statement was, 'for me, telling my family member information that is useful for them when making health decisions is: (desirable to undesirable). The indirect attitudes assessed in the survey were a) usefulness of information, b) relevance of information, c) causing worry and concern to family members and d) responsibility to tell family members.

Subjective norm statements

Direct measurement of subjective norm is typically done with three items which assess an individual's perceptions of pressure from others to perform the behaviour.[59] The statements: 'it is expected of me that...', 'people who are important to me want me to...', and 'I feel under social pressure to' [perform the target behaviour] were used in this survey.

Only one indirect statement was included because the literature indicated only that the opinion of the genetic counsellor might be important in communication to family

members.[19;71;90] Indirect measurement of subjective norm was based on one statement which measured the source of the social pressure (normative belief) to perform the behaviour, and another which measured the respondent's motivation to comply with that source (motivation to comply).

Perceived behavioural control statements

PBC was measured directly with three statements which measure self-efficacy to perform the behaviour and respondents' assessment of their ability to control performing the behaviour. The statements included: 'For me, talking to my family member is' (very difficult to very easy), 'I am confident I could talk to my family member about my genetic test result' (strongly disagree to strongly agree), and 'the decision to talk to my family member about my genetic test result is out of my control' (strongly disagree to strongly agree).

A total of five PBC factors identified in the systematic review as being important to family communication of genetic information were assessed in this survey. As with indirect measurement of the other constructs, indirect measurement of PBC involves two statements. One statement measures the participant's belief about the amount of control he or she has over performing the behaviour (control beliefs) and the other measures the perceived power that factor has in influencing the behaviour. For example, the systematic review in chapter 3 identified family conflict as an important factor inhibiting disclosure. The manual would indicate to ask this indirect attitude statement as two questions: 1) 'If I did not get along with my family member it would make it (more difficult to less difficult) to talk to them:' and have the participant choose a number on a 7-point scale from more difficult to less difficult; and 2) 'I do not get along with my family member', again having the participant choose a

number on a 7-point scale from 'strongly disagree' to 'strongly agree'. The other four PBC factors measured were a) emotionally distant relationship between family members, b) understanding of genetic information, d) physical distance between family members and d) denial of possible increased risk running in the family.

Instrument Development Part 2

One objective of the survey was also to collect data on the communication issues specific to patients with DM1. The TPB section of the survey examined the potential barriers faced by respondents who did not tell at least one at-risk relative about their genetic test result, therefore it was important also to examine the experience of telling an at-risk relative and their overall experiences with the process in general. This was done with a series of closed and open-ended questions focusing on aspects of disclosure that were identified as important from the literature as well as the clinical experiences of the genetic counsellor. This examination of aspects of communication which had already happened fell outside the TPB, which specifically focuses on the intention to perform a behaviour in the future.

Instrument Piloting

The survey was piloted on a small group of patients (n=10) from the CHEO genetics clinic. A genetic counsellor approached some of her patients, who had already undergone testing and counseling, and asked if they would participate in a survey pilot. The patients had a variety of genetic disorders and test results. Ten pilot surveys were sent out to those who agreed with a letter from the genetic counsellor or their doctor explaining the survey. Two surveys were returned with comments and were used to make minor changes to the final survey.

Data Analysis

Analysis was performed using SAS (v 9.1). Descriptive statistics were used to describe the responses to the survey and the population. Responses to open-ended questions are summarized qualitatively. The initial plan was to examine relationships between categorical variables (nominal and ordinal) with Chi Squared or Fisher's exact test, to use cronbach's alpha to test for internal consistency between responses to TPB statements for each construct, and to use regression analysis to test the predictive power of the model. However, the low response rate to the survey in general, and to the TPB section in particular, did not allow this (see Results).

The Children's Hospital of Eastern Ontario (CHEO) research ethics board approved the research project (Appendix L).

Results

Of the 261 DM1 genetics patients identified from the genetics clinic database as being potentially eligible for the study, 25 were excluded because they were duplicate files, a file for a fetus (picked up by the search string since the fetus is associated with the mother's demographics) or a file for a spouse of someone with DM1 (i.e. seen for counselling but not testing). Twenty three files were excluded because the genetic test on file was conducted to 'rule out' myotonic dystrophy in a patient with suggestive symptoms, the diagnosis of DM1 in a family member was never confirmed in the file or the patient had other additional congenital anomalies. 16 were excluded from the sample because the patient was an affected fetus or someone with developmental disorder (severity of disease would make completing the survey difficult cognitively). In 18 of the files there was no genetic test done or was performed prior to January 1, 1997. In three files there was no genetic

counselling done in the clinic (referred samples only). Thirty of the files were missing. Thus, the total number of eligible participants was 139. Of the surveys mailed to these, 40 were returned marked 'incorrect address'. Therefore, 99 respondents received the survey and were eligible to complete it. Twelve of the 99 participants declined to participate; 28 participants responded to the survey, 24 by mail and 4 by electronic survey and; 59 did not reply. Thus, the response rate for the cross-sectional survey was 28.3%.

The characteristics of the sample are presented in table 8. The age distribution of participants ranged from 20 to over 70 years of age. The greatest number of participants was in the 40-49 years of age range. The sample was predominantly married females whose first language was English. Most participants had completed a college education. 42.9% of the sample had negative test genetic test results for myotonic dystrophy and 25% identified themselves as having a mild case of DM1 with a lower repeat number. 81.5% of the sample had at least one other family member who had DM1 and 46.4% had symptoms at the time of answering the survey. The median time since they learnt their genetic test result was 5 years.

Table 8: Characteristics of participants

Variable		N	%
Age	39	9	33.3
	40-59	10	37.0
	60 +	8	29.6
Sex	Male	9	32.1
	Female	19	67.9
Marital Status	Not married	5	17.9
	Married	19	67.9
	Divorced /separated / widowed	4	14.3
Education	High school or less	10	37.0
	Community college or technical college	11	40.7
	University Degree and more	6	22.2
Ethnic Group*	Canadian	19	
	French	12	
	English	7	
	Irish	8	
	Italian	5	
	Other	6	
First Language	English	19	70.4
	French	8	29.6
Self-reported test result	Positive	6	24.0
	Mild (low repeats)	7	28.0
	Negative	12	48.0
Symptoms at time of survey?	Yes	13	50.0
	No	13	50.0
Other family members with DM1?	Yes	22	84.6
	No	4	15.4
Median time since test result		5 years	

*numbers total > 28 because respondents were asked to 'check all that apply'

For each type of relative, respondents reported higher rates of disclosure than non-disclosure, the lowest disclosure rate being over 50 percent. Every participant disclosed his or her genetic test result to at least one relative. Disclosure was most likely to have been made to mothers, sisters, brothers, and fathers, and least likely to have been made to male cousins, nephews, uncles, nieces and aunts (table 9). Female cousins (third-degree relative) were more likely to have been disclosed to than all second degree relatives.

Table 9: Genetic test result disclosure according to relative type

Relative Type	N with relative	N told	%
Mother	16	16	100
Father	12	10	83.3
Sister	19	18	94.7
Brother	21	19	90.1
Daughter	12	8	66.7
Son	16	11	69.0
Aunt	25	17	68.0
Uncle	23	13	56.5
Niece	19	12	63.2
Nephew	18	10	55.5
Female cousin	27	17	70.4
Male cousin	27	15	55.5
TOTAL	235	166	70.6

With regard to first-degree family members rates of disclosure are much higher than rates of non-disclosure. Disclosure to children was lower than any other first-degree relatives. Disclosure rates to aunts, uncles, nieces and nephews were lower than to first-degree family members. Uncles and nephews were disclosed to less often than aunts and nieces. Percentage disclosed to was 56.5% and 55.5% for uncles and nephews and 68% and 63.2% for aunts and nieces respectively.

All participants reported having at least one male and one female cousin. Rates of reported disclosure were still higher with cousins than rates of non-disclosure (15 male and 19 female cousins were disclosed to); however, non-disclosure occurred more frequently with these groups than with other relative types; 12 male cousins and 8 female cousins were not told about the respondent's test result.

Communication about genetic test results occurred more frequently with female than male relatives. Overall, respondents had communicated their genetic test result to more than

half of available relatives, 90/118 females and 79/117 males. In total, 14 respondents had disclosed genetic information to all available at-risk family members, and 14 had not. This limited the sample size for the TPB analysis to 14 (those who had not disclosed), and precluded the use of cronbach's alpha to test for internal consistency or regression analysis to test predictive validity of the TPB. The low total sample size also resulted in several cells of the 2 x 2 tables with expected counts of < 2 which invalidated the use of Chi-squared and Fisher's exact test.

Table 10: Types of relatives about whom the respondent chose to answer TPB questions

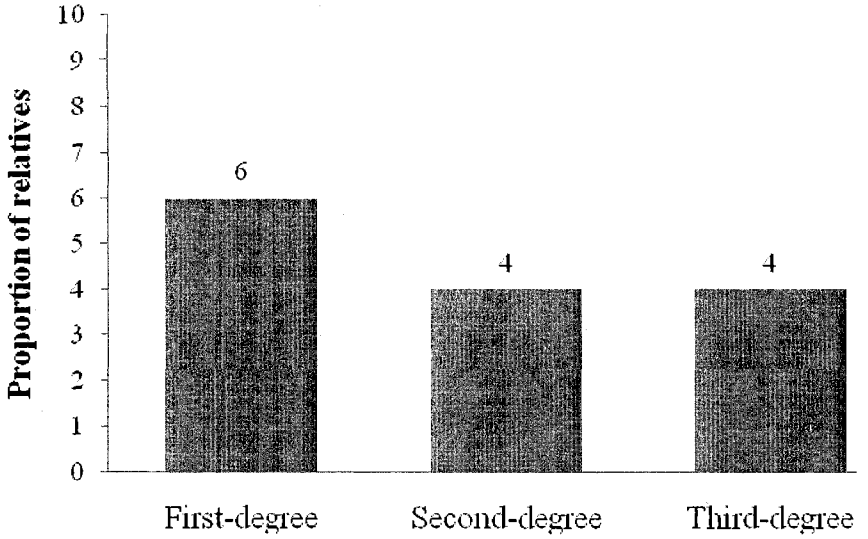
	Family member chosen: non-disclosure section of survey		Family member chosen: disclosure section of survey	
Relative	**Total N = 14**	**%**	**Total N = 28**	**%**
Mother	1	7.1	10	40.0
Father	1	7.1	0	0
Sister	1	7.1	4	16.0
Brother	2	14.3	1	4.0
Daughter	1	7.1	8	32.0
Son	0	0	1	4.0
Aunt	2	14.3	1	4.0
Uncle	1	7.1	0	0
Niece	0	0	0	0
Nephew	1	7.1	0	0
Female cousin	1	7.1	0	0
Male cousin	3	21.3	0	0
Total	14	100%	25	70%

Table 10 presents the type of family member about whom respondents chose to respond for the TPB section of the survey. In this section, respondents were asked to choose one family member to whom they did not disclose their result and answer it thinking of that person only. For the other section, respondents were to choose one family member to whom they did disclose, and answer it thinking of that person. Participants chose to answer the survey part concerning disclosure most often about their mother (40%), daughter (32%), and

sister (16%). The relative type chosen most often to answer the survey section regarding non-disclosure (TPB section) was male cousin (21%), aunt (14.3%) or brother (14.3%).

Figure 5 shows the frequency with which respondents chose to answer the two survey sections on disclosure and non-disclosure about, categorized by degree of relative. In this sample, respondents chose to focus on first-degree relatives most often for the survey section on non-disclosure and disclosure. Second-degree and third-degree family members were chosen as survey subjects more often for non-disclosure compared to disclosure (Figure 5b).

a) Non-disclosure



b) Disclosure

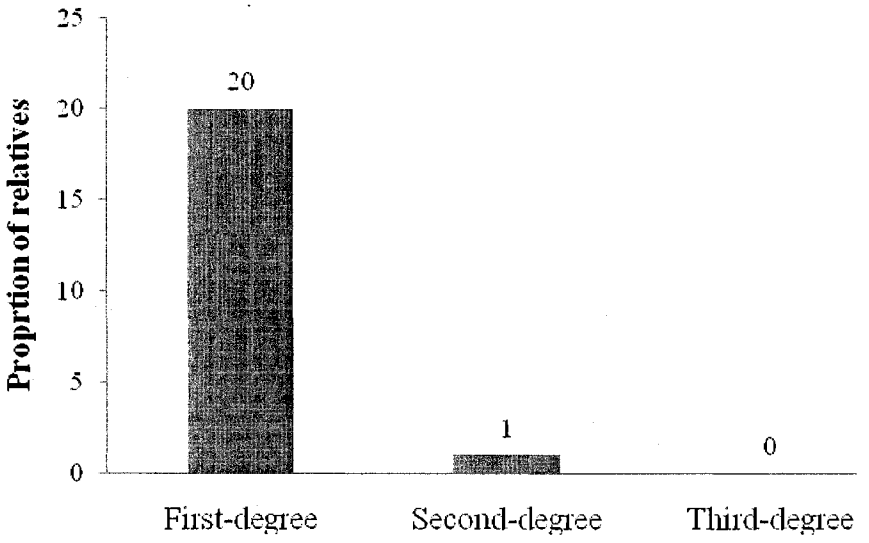


Figure 5 (a, b): Proportion of first-, second- and third-degree family members respondents answered each survey section about

Table 11 presents data on the likelihood of having told all available at-risk relatives by selected respondent characteristics. The sample size is too small to permit meaningful assessment of associations.

Table 11: Relationship between age, gender, test result and time since test result and likelihood of having disclosed to all available at-risk relatives

Respondent characteristic		Total N	N (%) who told all relatives
Age (years)	< 40	9	6 (67)
	40-59	10	4 (40)
	≥ 60	8	3 (37)
Gender	Male	9	5 (55)
	Female	19	9 (47)
Self-reported test result	Negative	12	4 (33)
	'mild'	8	4 (50)
	Positive	5	4 (80)
Time since test result (years)	≤ 5	17	6 (35)
	6-9	4	2 (50)
	≥ 10	6	5 (83)

There was no obvious association between the age or gender of the participants and whether or not they told their genetic test results to all at-risk relatives.

The same amount of respondents told all their family members regardless of what their genetic test result was. Of those who did not tell all of their at-risk family members a higher proportion of them received negative test results.

The majority of respondents have known their test results between 0 and 5 years. There was a lower proportion of respondents who told all at risk relatives of their genetic test result (35%). As time since receiving their test result increased the proportion of all at-risk relatives being told also increased.

Relatives to whom respondents did not disclose (TPB results)

As noted before, the maximum possible sample size for this analysis was 14, limited to the respondents who had not disclosed genetic information to at least one available at-risk relative. Table 12 provides the details of the median scores for each item in the TPB section of the survey, organized by construct. Respondents' scores relate to disclosure with the relative they each selected, to whom they had not yet told the genetic test result. A score of low was assigned if the value fell within the first third of the possible values, moderate if it was in the middle third and high in the top third. The overall median score for the generalized intention to disclose (3.33/7) was moderate. The median direct attitude score (3.88/7) was moderate, and the median indirect attitude score was also moderate. These relate to the respondents perceptions of: relevance of the information for the family member, their responsibility to tell relatives, the usefulness of the information for the relatives, and whether the information will cause the relative worry and concern. The median direct subjective norm score (concerning the pressure respondents felt from society and to perform the behaviour) was low, while the median score for indirect subjective norm, (perceived pressure specifically from their genetic counsellor and the respondents' desire to comply with the genetic counselor) was moderate. Participants' median perceived control over disclosing to the selected family member was moderate, whereas the indirect measurement of this construct was low. Indirect PBC measured the factors: emotional distance, ability to understand genetic information, physical distance from the family member, conflict with the family member and the impact of denial.

Table 12: TPB items scores

TPB item	Median †	N	Interpretation
Generalized Intention	3.33 [7]	14	Intention to disclose genetic test result to chosen relative is *moderate*
Direct Attitude	3.88 [7]	14	Attitude towards disclosing to chosen relative is *moderate*
Indirect Attitude	72.5 [196]	14	Attitude towards disclosing to the chosen relative is *moderate*
Direct Subjective Norm	2.00 [7]	14	Perceived social pressure to disclose to chosen relative is *low*
Indirect SN	24.00 [49]	13	Perceived social pressure from genetic counsellor to disclose to chosen relative is *moderate*
Direct PBC	4.00 [7]	14	Perception of control over disclosing to the chosen relative is *moderate*
Indirect PBC	41.00 [245]	13	Perception of control over disclosing to the chosen relative is *low*

† Numbers in [ ] indicate the highest possible total for each construct

Table 13 displays the TPB constructs, item statements used in the survey (and to calculate the TPB scores in table 12), number of participants who responded to each statement and the median score for that statement.

Table 13: TPB items, anchors, number of participants responding and median score

TPB construct	Items	Anchor	N	Median
Intention				
	I will talk to my family member about my genetic test result	Very unlikely / very likely	14	4.0
	I plan to talk to my family member about my genetic test result	Strongly Disagree / Strongly Agree	14	3.5
	I intend to talk to my family member about my genetic test result	Strongly Disagree / Strongly Agree	13	3.0
Direct Attitude				
	For me talking to my family member about my genetic test result would be:	Very harmful / very beneficial (for me)	14	4.0
	For me talking to my family member about my genetic test result would be:	Very comfortable / very uncomfortable (for me)	14	4.0
	For me talking to my family member about my genetic test result would be:	Very frustrating / not at all frustrating (for me)	14	6.0
	For me talking to my family member about my genetic test result would be:	Very useful / not at all useful (for me)	14	3.0
Indirect Attitude				
Behavioural belief 1	My genetic test result is important for my family member	Strongly disagree / Strongly agree	12	4.0
Outcome expectation 1	For me, talking to my family member about things that are important for them to know would be:	Very undesirable / very desirable	14	6.0
Behavioural belief 2	Talking to my family member about my genetic test result is my responsibility	Strongly agree / Strongly disagree	14	5.5
Outcome expectation 2	For me, carrying through my responsibilities is	Very undesirable / Very desirable	13	6.0
Behavioural belief 3	Talking to my family member about my genetic test result would give them information that would be useful for them when making their own decisions about their highest	Strongly disagree / strongly agree	14	6.0
Outcome expectation 3	For me, telling my family member information that is useful for them when making health decisions	Very undesirable / very desirable	14	6.5

Behavioural belief 4	If I told my family member about my genetic test result I believe it would cause him or her a lot of worry and concern	Strongly disagree / strongly agree	13	3.0
Outcome expectation 4	For me, causing a lot of worry and concern for my family member is	Very undesirable / Very desirable	14	6.0
Direct SN				
	I feel under social pressure to talk to my family member about my genetic test result	Strongly disagree / strongly agree	12	2
	People who are important to me ant me to talk to my family member about my genetic test result	Strongly disagree / strongly agree	12	2
	It is expected of me that I talk to my family member about my genetic test result	Strongly disagree / strongly agree	13	2
Indirect SN				
Normative belief 1 (pressure)	My genetic counsellor expects me to talk to my family member about my genetic test result	Strongly disagree / strongly agree	14	4
Motivation to comply 1	What my genetic counsellor thinks I should do matters to me	Strongly disagree / strongly agree	14	5.5
Direct PBC				
	For me, talking to my family member about my genetic test result is	Very difficult / very easy	14	4.5
	I plan to talk to my family member about my genetic test result	Strongly disagree / strongly agree		
	The decision to talk to my family member about my genetic test result is out of my control	Strongly disagree / strongly agree	13	4
	I am confident I could talk to my family member about my genetic test result	Strongly disagree / strongly agree	14	5
Indirect PBC				
Control belief 1	I do not feel emotionally close with my family member	Strongly disagree / strongly agree	14	5
Power 1	If I was not emotionally close with my family member it would make it ___ to talk to them about my genetic test result	More difficult / less difficult	12	3
Control belief 2	Genetic risk information is hard to understand (for me)	Strongly disagree / strongly agree	13	3
Power 2	If I found genetic risk information hard to understand it would be ___ to talk to my family member about my	More difficult / less difficult	14	3.5

	genetic test result			
Control belief 3	I live far away from my family member	Strongly agree / strongly disagree	14	4
Power 3	If I was living far away from my family member it would make it ___ to talk to them about my genetic test result	More difficult / Less difficult	13	3
Control belief 4	I do not get along with my family member	Strongly disagree / strongly agree	13	1
Power 4	If I did not get along with my family member it would make it ___ to talk to them	More difficult / less difficult	13	2
Control belief 5	My Family member is in denial about his or her genetic risk	Strongly agree / strongly disagree	12	3
Power 5	If my family member was in denial about his or her genetic risk it would make it ___ to talk to them	More difficult / less difficult	14	1.5

Relatives to whom respondents did disclose

Table 14: Importance of each reason to the decision to disclose to the family member

Reason	N (%) very important	N (%) important	N (%) neither	N (%) not important	N (%) very unimportant
'you have a close relationship with the family member'	21 (84)	1 (4)	1 (4)	1 (4)	(1) (4)
'you have no conflicts or tensions with this family member'	12 (50)	4 (17)	5 (21)	1 (4)	(2) (8)
'you thought that the person could also give you social support in dealing with your test result'	12 (48)	2 (8)	7 (28)	3 (12)	(1) (4)
'you thought the person was emotionally capable of handling the information'	16 (64)	3 (12)	4 (16)	1 (4)	(1) (4)
'you thought that the information was relevant for the person'	18 (72)	2 (8)	4 (16)	0 (0)	(1) (4)
'you thought you might regret it if you didn't tell the person'	14 (56)	2 (8)	3 (12)	2 (8)	(4) (16)
'you thought you had a responsibility or duty to tell the person'	14 (56)	4 (16)	4 (16)	0 (0)	(3) (12)
'you thought you would feel guilty later if you didn't tell the person'	10 (41.7)	2 (8.3)	7 (29)	1 (4)	(4) (17)

Table 14 presents data on the respondents' reasons for disclosing their genetic test result to the family member they selected. In general, most responses tended towards the 'important' end of the scale. The most important reasons appeared to be a close relationship with that person, followed by believing that the information was relevant for the family member, and believing that they were emotionally capable of handling the information. The

two reasons of apparently lowest importance were thinking that they would feel guilty later and thinking that they could get social support from that family member.

Open questions on factors which made disclosure difficult:

Respondents offered a range of open comments on factors which helped or hindered disclosure. The most common factor mentioned was the experience of the family watching so many family members suffer and die, making the topics of genetics and myotonic dystrophy very emotional, sensitive and difficult to discuss. One participant suggested that coping with the death of loved ones made it more difficult to talk about genetic test results.

The perceptions of other family members regarding myotonic dystrophy appeared to be an important barrier to communication about genetic test results. For example, one respondent indicated a relative's view that the myotonic dystrophy risk information was not applicable or relevant to them, and another respondent reported that a family member did not think that the disorder was serious or something to be concerned about. Other factors mentioned included thinking that the illness did not exist, not being concerned because the respondents themselves did not have symptoms, and, in one family, a belief on the part of several members the disorder was actually caused by a virus and had no genetic basis. One participant perceived that some relatives were not sympathetic and did not understand why an affected person couldn't do all that a healthy person could (either suggesting that they didn't believe that this individual had DM1 or that the disease did not exist). Additionally, three participants reported that their family members were in denial and did not elaborate further.

Table 15: Summary of themes identified in qualitative responses

Factors making disclosure difficult:
• Death of family members (the topic of DM1 is sensitive) • Guilt (for passing it on to others or for not being a carrier) • Denial • Family member did not wish to know about their risk • Family member did not think that the risk information was applicable to them • Family members did not think the disorder was serious or something of concern • Distant relationship (not knowing where they are or their address or seeing them for over 20 years)
Factors which made disclosure easier:
• Having a negative test result or very mild symptoms • Information or letters from the genetics clinic • Having support from their family (spouse was mentioned as vital) • Having an 'open relationship' with their family • Others have been diagnosed, have symptoms or a positive genetic test result and the topic is already open in the family • The family feeling as though they are going through the testing and experiences together
Other reasons for disclosing to the chosen relative
• Important for family planning • Important that family member begin monitoring for symptoms and taking good care • Importance of knowing increased when a daughter became pregnant (2 participants) • Urge family members to get tested • To get more information and compare symptoms • Desire to be open and honest (values)
Other relevant points mentioned:
• One believed that second degree relatives do not have a right to know their test results • Several individuals indicated that they would like the genetics clinic to contact relatives for them if possible • Myths about the disorder exist and hinder communication (the disorder is a virus, or doesn't even exist) • A common reason for having the test was to help others know their future

Guilt was also frequently mentioned as a factor which made the experience more difficult. This guilt related either to feeling guilty about passing on the genetic disorder to others and guilt for not carrying the gene when so many other family member do. One

person noted that while being emotionally close with someone made it easier to disclose their genetic test result to, it also made it more difficult in the fact that the guilt was very strong for not having the gene when a close sister did.

Distant family relationships, socially or geographically, were identified as being important impediments to communication. Respondents found it difficult to disclose to family members they haven't spoken with for 10-20 years, some did not even know their address or what country they were currently living in.

Particular situations in the life of certain family members can act to either facilitate or hinder disclosure of the genetic test result, for example, the unstable mental state of one family member, and parents acting as though they were being blamed for the disorder.

Open questions on factors which made disclosure easier

A commonly mentioned factor which was important in easing the disclosure process was the services provided by the genetics clinic, including the information provided in the counselling session by the counsellor, and the letters written for the participants by the counsellors.

Certain family characteristics also seemed to make it easier for some to discuss the topic. Two respondents felt it was easy to talk about because they were all experiencing the disorder together as family members got sick and others got tested, thus they were all open and curious about the situation of other members. The fact that others already had the disorder or a test made it easier for two respondents to tell others because the topic had been opened up in the family by another person already. Open, supportive and interested family was mentioned as a factor that made the experience easier for three participants.

Several participants mentioned that because their test result was negative (i.e. do not have myotonic dystrophy) or that they only had mild DM1 it was easier for them to disclose their result to others, because they felt in a way that they were giving good news.

Additional reasons for disclosing to the family member and comments

Respondents added that family planning was important for them in telling other family members so they could plan their life and future generations as well as to start monitoring for symptoms and take good care of their heart and eyes. Two women told their pregnant daughters their test result so they could choose whether to have the child with knowledge of the risk for myotonic dystrophy. One respondent told other family members to urge them to get tested themselves. Less commonly mentioned factors included feeling the family member had rights to the information, a desire to be open and honest, to obtain information about others symptoms to compare their own symptoms and worry that they would end up in the same situation as other family members.

Several participants suggested that the reason for their getting the genetic test result was to provide information on risk to other family members so they would be able to know their future better.

One participant was astonished that the survey would even suggest that it was his responsibility to inform second degree relatives about his genetic test result. He believed that they did not have rights to that information, he was worried about his privacy, and thought if they were entitled to any information it would be that he had testing – definitely not the result. He was also worried about family members telling employers and insurance

companies. Similarly, one participant believed that aunts, uncles, nieces, nephews and cousins are not defined as family and thus do not have a right to know the genetic test result.

Several participants requested that the genetics write letters (anonymous and not) to the people they could not disclose to due to no contact or because of concern for privacy. A few reported feeling very worried about their children and their future since they do not have symptoms yet but the concerns for their health are prevalent themes in their life.

Other factors examined

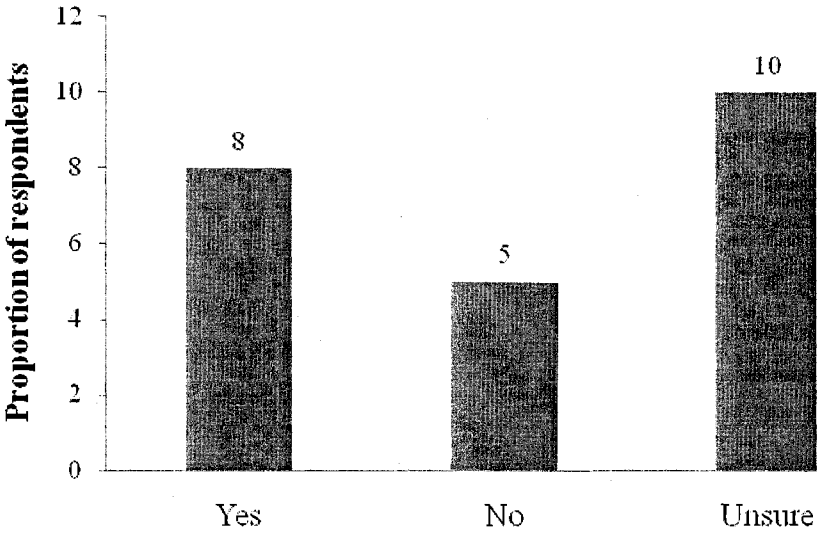


Figure 6: Proportion of respondent who reported that their genetic counsellor informed them to which family members to disclose their genetic test result.

Of 23 respondents, eight reported that their genetic counsellor had indicated which family members it was important to speak with about the genetic test result, five reported that their genetic counsellor did not talk with them about this, and ten were unsure. Twenty one participants indicated that they were not ambivalent about disclosing their genetic test results to their family member and six did feel some ambivalence (not shown).

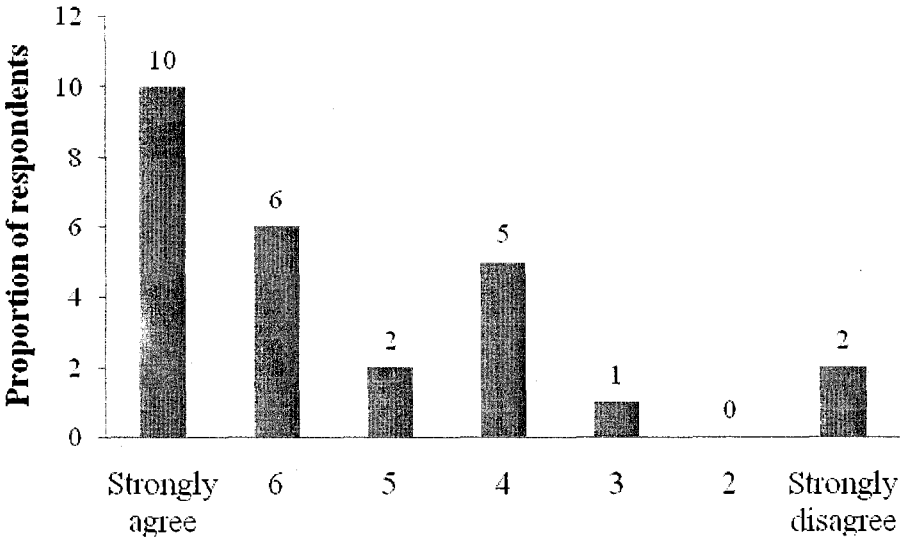


Figure 7: Respondents agreement with the statement 'myotonic dystrophy is a serious problem'

Eighteen of 26 participants tended to agree or strongly agree that myotonic dystrophy was a serious problem, while five were neutral and three disagreed. Regarding communication patterns within respondents' families, ten of 28 respondents indicated that communication patterns in general were 'open', and eight reported them to be 'closed'; ten of 27 reported 'open' communication patterns about health problems and seven reported 'closed'.

CHAPTER V

DISCUSSION

The complexities surrounding family communication of genetic risk information are great. In addition to the difficult nature of the subject, families themselves as functioning social units are characterized by individual norms, interactions, rituals and behaviours which exist regardless of knowledge of genetic risk. Not surprisingly then, the introduction of complicated and sensitive information into a complex and dynamic relational structure may face diverse barriers. There is evidence that, in the clinical setting, genetic counselors and patients sometimes encounter issues to do with disclosure of genetic test results to family members. The intention of this research study was to gain an understanding of the barriers and facilitators of communication of genetic information within a family in the light of a behavioural theory; the ultimate aim being to provide a solid foundation for the development of appropriate interventions. This is the first study to explicitly conceptualize the communication of genetic risk information as a behaviour, and to lay the groundwork in such a detailed way for the development of tools and interventions which can assist those with communication issues. This is also, to our knowledge, the first study to examine the communication of genetic risk information in a population of families at risk of myotonic dystrophy. This research is the first step from knowledge generation to problem solving in the realm of family communication of genetic risk information.

Overview of Key Findings: Review and Framework

The published evidence on disclosure of genetic risk information between family members indicates that the majority of individuals who encounter the clinical genetics setting do go on to disclose their genetic test result or risk information to (at least) some family members. It is clear that those who delay or fail to disclose such information generally have a desire to tell their at-risk relatives about their risk, as shown by the consistently recurring theme of having a 'moral duty' and responsibility to provide family members with information that is relevant to their health. It is uncommon for direct refusal to disclose, and this usually occurs in situations where families already have conflict or atypical circumstances. Usually it is in relation to second and third degree relatives, and/or the relatives with whom a patient has little contact, where disclosure is more of an issue. Barriers are related to whom, when and how to do the disclosing rather than whether to disclose; from an ethical perspective, this is an encouraging finding, as it suggests that interventions can be directed towards promoting a behaviour which follows through on a decision, rather than the decision itself.

At the core of disclosing genetic information to family members are basic beliefs and perceptions about responsibility and rights. In deciding whether to disclose, these are potentially altered by several factors: the person's situation, personality, feelings of guilt, the test result (degree of 'good' or 'bad' news), the characteristics of the disease, their own understanding and interpretation of the information and ability to explain this to others (communicate this effectively), and their beliefs about whether telling a relative is useful. The factors are numerous and very variable between patients, which underscores the challenge of this issue within the clinical context. Thus, it is important for genetic

counselors to be able to assess the specific issues faced by their patient as well as having tools available that might help them in their situation. This suggests a possible role for decision aids to help genetic counselors select appropriate interventions for individual clients.

The results must be interpreted in light of the limitations of the studies included in the review. Most of the studies on this topic are qualitative, or are quantitative with small sample sizes, and their generalizability to the general population of genetic patients is not certain. The majority of the research done on family communication and genetic risk has focused on adult onset, autosomal dominant inherited disorders, thus the full spectrum of the issues for other genetic disorders may not be captured within this review.

There are other limitations to the review. Because of time constraints, only one reviewer did the initial screening of articles, the final selection for inclusion and exclusion, and data abstraction. There was no attempt to search grey literature or make contact with authors to identify other. Only English language studies were included which limits conclusions which can be drawn regarding genetic information communication issues in different cultures.

The information from the systematic review informed the creation of a framework of communication of genetic risk information. The barriers and facilitators identified were fit with the TPB constructs to describe how an individual's attitude towards a behaviour, their beliefs about pressures from others, and their belief in their ability to control the behaviour will act to determine their intention to communicate with family members and thus, the occurrence of that behaviour. Preliminary evidence on the framework's face validity and applicability was explored in a cross sectional survey in a specific patient population.

Overview of Key Findings: Cross-sectional survey

The aims of the cross-sectional survey were to a) test the face validity of the framework of communication of genetic information, based on the TPB, in a patient population and, b) to gain insight into the communication process and experience of a population which has yet to be examined in the literature. The later will be addressed first.

The survey findings relating to myotonic dystrophy are consistent with studies of populations with other disorders reported in the literature. Overall, this sample had relatively high rates of disclosure, and first-degree relatives were more likely to have been informed than more distant relatives. This trend has been reported in studies of other disorders (including hereditary breast or ovarian cancer, Huntington's disease, HNPCC, Cystic fibrosis, and Hemophilia).[18;19;63;70-73;75;77;83;87] Some respondents to this survey indicated that disclosing their genetic test result was made easier by the fact that knowledge about the disease was already open in the family, either because others had already been tested, individuals have passed away from the disorder, or that the family in general had open and supportive relationships between members. Just over half of the sample rated their family communication pattern in general as 'open', potentially contributing to the high communication about myotonic dystrophy. The openness of communication has been found to be important in other studies. Holt *et al.*, (2006) contrasted two HD families with open and closed communication patterns and found that there those with open patterns appreciated the openness whereas the children in families with closed patterns retrospectively wished for an open pattern. This stresses the importance of family dynamics in influencing individuals' experiences with genetic information.

Our findings relating to barriers and facilitators are consistent with other published literature on a range of genetic disorders. We found that disclosure was highest to first-degree relatives and lower for second and third degree relatives. It is likely that this kind of information would be considered most relevant for first-degree family members, and that first-degree family members tend to be socially closer than other relative types. On average, female cousins were more likely to be told than any type of second-degree relative, perhaps reflecting greater communication in general with female relatives or a sister-like closeness of female cousins to each other in some families.

Communication between female relatives was slightly higher than between males in this sample (although statistical significance could not be measured); however males in this sample also displayed generally high disclosure rates. Several reasons for this gender difference have been postulated including, the role of women being particularly focused on caring for the family's health,[100] women being more communicative than men by nature[68;71] or the tendency of studies in this area to focus on disorders of particular relevance to women such as HBOC.[74] Myotonic dystrophy affects both males and females, and thus closer disclosure rates between males and females could be expected. However, myotonic dystrophy genetic risk information is not completely 'gender neutral'. The genetic mutation for myotonic dystrophy displays anticipation, i.e. as the disorder passes from generation to generation, the age of onset can be earlier and disease severity worse. Thus, genetic risk information has important implications in family planning and decisions surrounding termination of pregnancies and birth.

The most important reasons in deciding to disclose to their family member identified were a) having a close relationship with the family member, b) perceiving that the

information was relevant for them, c) believing that they were capable of handling the information and d) feeling a responsibility to tell them about their genetic risk. These items are also consistent with previous literature.[13] The significance of these results is that they indicate factors that may be amendable to intervention: educating patients on who needs to know (regardless of closeness) their test result, and about which information is relevant for different family members; preparing patients for the possible reactions of family members; and using decision aids to help patients work out their own views on whether, and who, to tell. Open comments from the survey all focused on reasons which would help the family member they told, for example: telling results because daughters became pregnant and they needed to know, telling so that the family member was more careful with their health and monitored more carefully for symptoms, or telling so that they could urge others to get tested, and know their risk. These factors have also appeared, although less often, in the literature on the topic.

Factors which made disclosure more difficult were also examined and were also consistent with past findings[13]. Often the topic of the disease and genetics is very sensitive to families who carry the mutation because they have witnessed the illness and death of loved ones. Disclosure was viewed as more difficult in this population because they were still coping with family member deaths and the topic was emotional and upsetting for the family. Another reason alluded to several times was the situation where family members did not want to know or refused to know the information. For example, participants encountered family members who had different desires for knowledge of their genetic risk, and others who refused to accept that the disease existed or was genetic in nature, others who were in denial. This illustrates that the few who were not disclosed to were

'exceptional' cases, where the types of interventions described above would either not be appropriate (as individuals have a right not to know their genetic risk status) or would not address the issue of family members in denial or with 'non-rational' thinking. This leads to an important point from a clinical and intervention perspective - there are clearly instances where neither the family member who underwent testing nor a genetic professional should intervene, to respect the rights of relatives, or because of the possible psychosocial harm that might be caused.

Although we could not test the predictive power of the TPB constructs, the theory section of the survey nevertheless yielded some interesting results. Taking the results at face value, this sample appeared to have a 'moderate' intention to tell their chosen family member about their genetic test result, which could indicate either some lingering ambivalence about telling them, or perhaps they haven't acted on an intention. The attitude towards disclosing to this family member was also 'moderate' on average, and there appeared to be no perceptions of strong external pressures to disclose. Interestingly, the subjective norm variable related to pressure from a genetic counsellor was 'moderate', which is consistent with usual clinical practice, since genetic counsellors are generally expected to inform patients that disclosure is a good idea, but to do so in a non-directive manner. The importance of the role of the genetic counsellor is underscored by the median score for the indirect subjective norm which suggests that assistance from the genetic counsellor might have an important part in disclosure. The sample's perceived control over telling their chosen family member was 'moderate' to 'low'. This suggests feelings of some degree of control over communication, but barriers which are preventing them from doing it. This is supported by the open comments offered by respondents. In summary, in this

sample, attitudes towards communicating appeared to be positive but there were no strong pressures pushing this and possibly not insignificant barriers which prevent them from doing it.

The study has provided some preliminary evidence of the face validity of the framework, but this must be interpreted cautiously because of the small sample available for analysis and the low overall response rate. The survey findings are consistent with those of other studies. These preliminary results indicate that it is worth examining further the usefulness of the framework as a way of meaningfully describing the barriers and facilitators of communication of genetic risk information in general.

Since this project began, other models addressing the issue of genetic risk communication have been published. Klitzman *et al.*,[80] created a model based on communication between those with a Huntington disease genetic test result and their immediate and extended family: it described decisions involved in the process included what to tell, how to tell, why tell, and when to tell. Similarly, van den Nieuwenhoff *et al.*,[93] created a model of factors with affect family disclosure of the genetic risk for inherited high cholesterol based on the divisions of community and public policy, institutional, interpersonal and intrapersonal factors. Both of these models provide evidence of the validity of the framework developed in this project, because the factors identified within each are included within the taxonomies created in our TPB-based model. However, to our knowledge, this model is currently the only one based on behavioural theory, and which has the potential for prediction of behaviour at the individual level.

Limitations

Several limitations of the current study must be acknowledged. The survey was retrospective in nature and thus may be influenced by recall bias; however, a good portion of the survey items related to the present, and decision making up to this point in time. Prospective studies would offer richer insights into the processes of communication. The sample was restricted to patients with a single genetic disorder. The results cannot be considered generalizable, although it should be noted that this was not the primary intent of the survey. Unfortunately, the arms length recruitment strategy also severely restricted an assessment of the number of families this sample represented – the data are almost certainly hierarchical (respondents within families), but we had to treat the respondents as independent observations.

The low response rate means that we cannot rule out the possibility that respondents were more likely than non-respondents to have communicated genetic information, and the high disclosure rates we observed may be unrepresentative. We were also less likely to capture information on the sub-group of patients most severely affected by the disorder due to impairments in cognition. Again, privacy concerns meant that there was no way to assess this. The low response rate may also be in part because of the length of the survey which may have caused respondent burden (the survey was 15 pages long, and took no more than 20 minutes to complete).

There were a higher proportion of English speaking participants than Francophones. We were unable to provide the survey in French, because of the challenge of truly translating the concepts, words, and phrases meaningfully for a different cultural group.[122] Although we obtained a professional translation of the questionnaire, we were not confident

in its validity and decided to abandon the attempt at a French language version because of the time constraints for this project. The approach to designing a TPB survey demands very particular wording for many items. Also, it is challenging to be succinct and unambiguous when asking about an inherently complex area such as communication and family dynamics. However, all supporting documents were provided in French with an explanation of why a French survey was not available at this time. Unfortunately, this limits confidence in the findings of the survey, as myotonic dystrophy is common in the French Canadian population, and we were unable to capture representative data from this population. This may have also played a role in the low response rate because eligible participants, who could only speak, write or understand French would not be able to complete the survey. It also begs the question of the validity of responses from francophone participants answering in their second language.

The sole use of one key informant to decide upon the theory to base the framework on is also a limitation of this research. Ideally, a literature review of potential theories or consultation with more experts would have been conducted. Unfortunately, the time restraints on the project and feasibility of obtaining expert advice from several sources prevented the use of these methods.

On balance, the consistency of the findings with other research indicates that further development and piloting would be worthwhile.

An important issue in designing a survey based on the TPB is the definition of the behaviour. Previous research clearly indicates that the more specific and clearly defined the behaviour of interest, the better the explanation of its variance by the TPB constructs.[47;59] The nature of the 'behaviour' of communicating genetic test results to family members is

complicated. In order to keep a survey manageable in terms of length and literacy level, we had to limit the behaviour to communication with a single relative, to whom the respondent had not yet disclose the genetic test information. Because this was a retrospective exercise and likely the participants had already told some relatives, we could not prescribe one (or several) specific relative(s) because they may have already told that relative making the survey irrelevant for them. The next best approach was to allow the respondent to select a relative about whom they answered questions, and to tell us who that relative was. We are therefore extrapolating responses about one relative to a general pattern of communication, which may not be valid.

Future approaches to this issue might include designing a prospective, longitudinal approach (although the issue of the survey itself acting as an intervention would need to be considered), and using innovative survey techniques to capture data. For example, an electronic, algorithm-based survey application which presented one question at a time might make the entire exercise easier for a respondent, less daunting, and possibly more comprehensive.

Conclusions

The systematic review revealed an extensive, and expanding, literature on the topic of family communication and genetic risk information. There are gaps in research which need to be further explored, particularly with respect to recessive disorders and those with more complex inheritance patterns. However, there appear to be consistent observations in the published research base which may be useful in informing the development and evaluation of interventions to facilitate communication. Based on this literature, and the exploratory survey, we suggest a theoretical framework to describe communication of

genetic information as an individual behaviour, and present very preliminary evidence which supports its validity.

Future directions

The theoretical framework requires further evaluation and possible refinement in a wider range of study populations, with careful attention to the design and implementation of the survey itself. The findings from this project may provide, even at this stage, the groundwork for the future development and exploration of interventions to facilitate communication of genetic risk information, including decisions about whether or not to disclose to relatives.

If these results were replicated in a larger, prospective study it would suggest that interventions which focused on the perceived behavioural control component of intention formation and behaviour are in need (the results indicated that there was moderate to low perceived control over disclosure in this sample). These types of barriers may be more amendable by intervention than attitudes or pressure from important others. Educational materials could focus particularly on reasons why others may want to know, information on the disorder, and lists of additional community resources. Educational materials can be tailored to family members of the tested in order to facilitate understanding and negation of myths about the disorder. Several participants from the survey found the information provided by the genetic counsellor to be a useful aid in disclosing to family members to ensure that the information was correct and as a means to bring up the topic – various templates of letters and indirect methods for passing on information may help overcome some barriers. Interventions should be developed to assist patients in learning coping skills for dealing with their test result and for dealing with the reactions of family members.

Furthermore, educational type interventions which can be used in the clinical setting or sent home with individuals could also target attitudes about disclosure as well as the pressure they feel not to disclose. Finally, the potential for use of decision aids in clinical genetics is great. These can help clients who have not made a settled decision to tell or not tell make decisions which are in line with their values and help participants make informed choices which they feel comfortable with.

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APPENDICES

APPENDIX A: Systematic review search strategies

Medline (1966-Oct week 1 2007 - search date: Oct. 09)
References = 1677
1. Communication/
2. disclosure/
3. truth disclosure/
4. self disclosure/
5. (non-disclosure or nondisclosure or disclosure or (family adj communication)).mp.
6. family/
7. family relations/
8. parent-child relations/
9. intergenerational relations/
10. ((family adj system$) or (family adj dynamic$) or (family adj secret$) or kinship or pre-selection).mp. [mp=title, original title, abstract, name of substance word, subject heading word]
11. genetic predisposition to disease/
12. genetic diseases, inborn/
13. genetic screening/
14. genetic counseling/
15. genetic services/
16. ((genetic adj risk$) or (genetic adj susceptibility) or (genetic adj screening) or (genetic adj test$) or (genetic adj disorder) or (predictive adj test$) or (genetic adj counseling) or (genetic adj services) or (hereditary adj disease$) or (genetic adj disease$) or (genetic adj predisposition)).mp.
17. or/1-5
18. or/6-16
19. 17 and 18
20. limit 19 to yr="2003 - 2007"

Medline Inprocess (1996 – Oct. 09/ 2007)
References= 539
1. Communication/
2. disclosure/
3. truth disclosure/
4. self disclosure/
5. (non-disclosure or nondisclosure or disclosure or (family adj communication)).mp.
6. family/
7. family relations/
8. parent-child relations/
9. intergenerational relations/
10. ((family adj system$) or (family adj dynamic$) or (family adj secret$) or kinship or pre-selection).mp. [mp=ti, ot, ab, nm, hw]
11. genetic predisposition to disease/
12. genetic diseases, inborn/
13. genetic screening/
14. genetic counseling/
15. genetic services/
16. ((genetic adj risk$) or (genetic adj susceptibility) or (genetic adj screening) or (genetic adj test$) or (genetic adj disorder) or (predictive adj test$) or (genetic adj counseling) or (genetic adj services) or (hereditary adj disease$) or (genetic adj disease$) or (genetic adj predisposition)).mp.
17. or/1-5

18. or/6-16
19. 17 and 18
20. limit 19 to yr="2003 - 2007"

Embase (1980-2007 week 40 - search date: Oct. 09/07)
References = 1230
1. Interpersonal communication/
2. self disclosure/
3. (non-disclosure or nondisclosure or disclosure or (family adj communication)).mp.
4. Family/
5. relative/ or sibling/
6. ((family adj system$) or (family adj dynamic$) or (family adj secret$) or kinship or pre-selection).mp.
7. genetic predisposition/
8. disease predisposition/
9. genetic risk/
10. genetic disorder/
11. genetic screening/
12. genetic counseling/
13. familial disease/
14. familial cancer/
15. heredity/
16. ((genetic adj risk$) or (genetic adj susceptibility) or (genetic adj screening) or (genetic adj test$) or (genetic adj disorder) or (predictive adj test$) or (genetic adj counseling) or (genetic adj servic$) or (hereditary adj disease$) or (genetic adj disease$)).mp.
17. 1 or 2 or 3
18. 4 or 5 or 6 or 7 or 8 or 9 or 10 or 11 or 12 or 13 or 14 or 15 or 16
19. 17 and 18
20. limit 19 to yr="2003 - 2007"

CINAHL (1982-Oct. week 1 2007 - search date: Oct 09/07)
References = 935
1. communication/
2. communication barriers/
3. truth disclosure/
4. self disclosure/
5. family/ or exp extended family/
6. family relations/
7. parent-child relations/
8. sibling relations/
9. disease susceptibility/
10. genetic screening/
11. genetic counseling/
12. genetics/
13. heredity/
14. hereditary diseases/
15. (non-disclosure or nondislcosure or disclosure or (family adj communication)).mp.

16. ((family adj system$) or (family adj dynamic$) or (family adj secret$) or kinship or pre-selection).mp.
17. ((genetic adj risk$) or (genetic adj susceptibility) or (genetic adj screening) or (genetic adj test$) or (genetic adj disorder$) or (predictive adj test$) or (genetic adj counseling) or (genetic adj service$) or (hereditary adj disease$) or (genetic adj disease$) or (genetic adj predisposition)).mp.
18. 1 or 2 or 3 or 4 or 15
19. 5 or 6 or 7 or 8 or 9 or 10 or 11 or 12 or 13 or 14 or 16 or 17
20. 18 and 19
21. limit 20 to yr="2003 - 2007"

PsycInfo (1806 – Oct. week 1 2007 – search date: Oct. 09/07)
References = 702
1. Communication/
2. Interpersonal Communication/
3. exp parent child communication/
4. communication barriers/
5. Self Disclosure/
6. (non-disclosure or nondisclosure or disclosure or (family adj communication)).mp.
7. Family/
8. Family relations/ or sibling relations/
9. Kinship/
10. family members/
11. ((family adj system$) or (family adj dynamic$) or (family adj secret$) or kinship or pre-selection).mp.
12. Genetics/
13. Genetic Disorders/
14. Genetic Counseling/
15. Genetic Testing/
16. Heritability/
17. At Risk Populations/
18. Predisposition/
19. "Susceptibility (Disorders)"/
20. risk factors/
21. ((genetic adj risk$) or (genetic adj susceptibility) or (genetic adj screening) or (genetic adj test$) or (genetic adj disorder) or (predictive adj test$) or (genetic adj counseling) or (genetic adj services) or (hereditary adj disease$) or (genetic adj disease$) or (genetic adj predisposition)).mp.
22. 1 or 2 or 4 or 5 or 6
23. 7 or 8 or 9 or 10 or 11 or 12 or 13 or 14 or 15 or 16 or 17 or 18 or 19 or 20 or 21
24. 22 and 23
25. limit 24 to yr="2003 - 2007"

Web of Science (search date: Oct. 09/07 (limited by 2003-2007))
[Science citation index expanded (SCI-EXPANDED) 1970-present, Social Sciences Citation Index (SSCI) 1970 - present, Arts and Humanities Citation Index (A&HCI) 1975-present)]
References = 168

#4 168 #3 AND #2 AND #1
DocType=All document types; Language=All languages; Databases=SCI-EXPANDED, SSCI, A&HCI; Timespan=2003-2007

#3 93,449 TS=("genetic counseling" or "genetic counselling" or "genetic screening" or "genetic risk*" or "genetic predisposition" or "disease susceptibility" or "genetic susceptibility" or "genetic test*" or "genetic disorder*" or "genetic service*" or "hereditary disease*" or "genetic disease*" or "hereditary" or "risk factor*" or "familial disease*" or "heredity")
DocType=All document types; Language=All languages; Databases=SCI-EXPANDED, SSCI, A&HCI; Timespan=2003-2007

#2 >100,000 TS=(family or "extended family" or "family relation*" or "sibling relation*" or "family system*" or "family dynamic*" or "family secret*" or "kinship" or "pre-selection" or "parent-child relation*" or "relative")
DocType=All document types; Language=All languages; Databases=SCI-EXPANDED, SSCI, A&HCI; Timespan=2003-2007

#1 50,976 TS=(communication or disclosure or "family communication" or "communication barriers" or "nondisclosure" or "non-disclosure" or "self disclosure" or "truth disclosure")
DocType=All document types; Language=All languages; Databases=SCI-EXPANDED, SSCI, A&HCI; Timespan=2003-2007

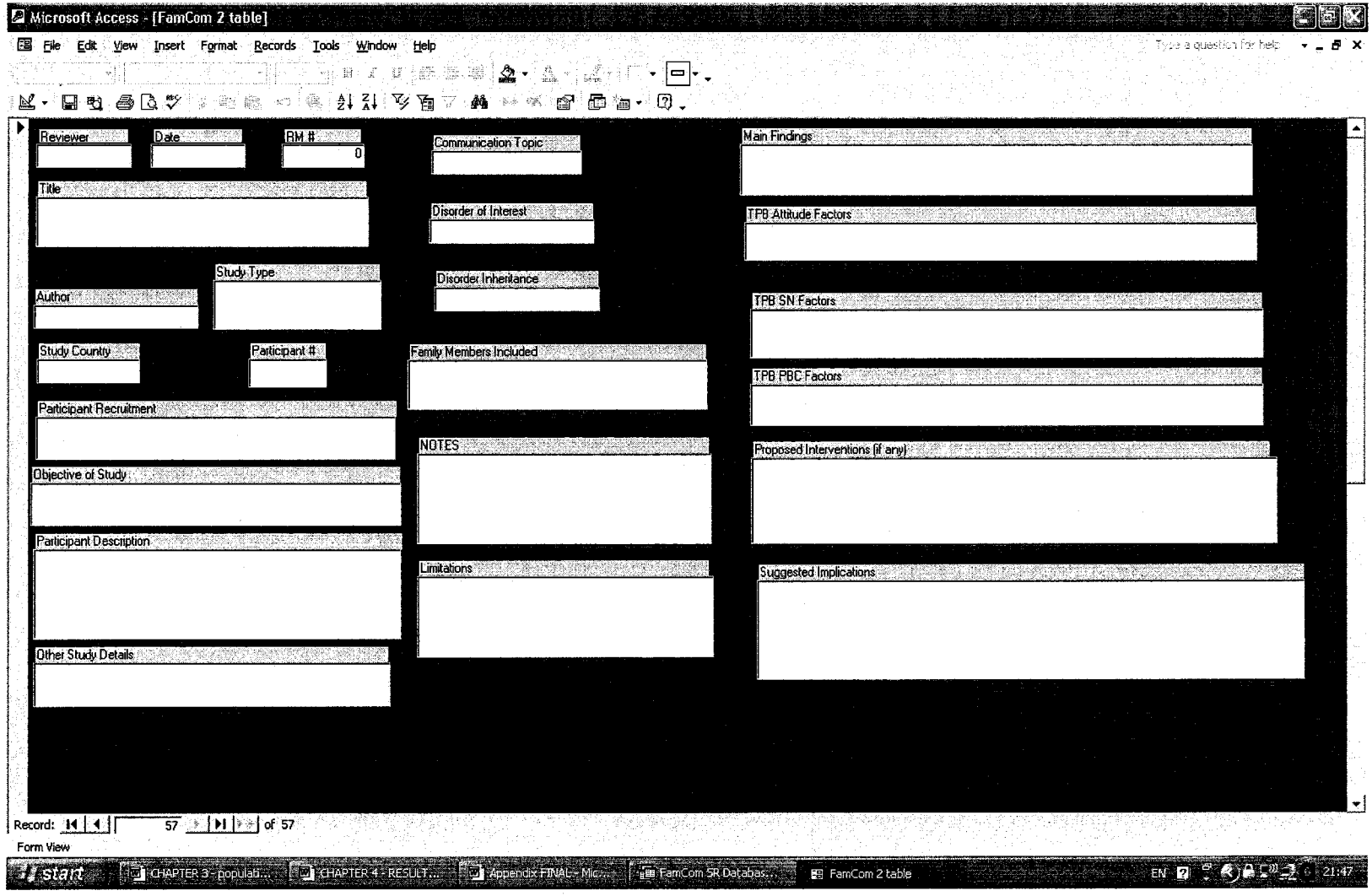
Sociological Abstracts (search date: Oct. 09/07 (limited by 2003-2007))
References = 535

genetic or genetic testing or counseling (DESCRIPTORS)
OR interpersonal communication or blank or blank (DESCRIPTORS)
AND parent child relations or family relations or family (DESCRIPTORS)

Social Services Abstracts (search date: Oct. 09/07 (limited by 2003-2007))
References = 447

genetic or genetic testing or counseling (DESCRIPTORS)
OR interpersonal communication or blank or blank (DESCRIPTORS)
AND parent child relations or family relations or family (DESCRIPTORS)

APPENDIX B: Data abstraction form



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APPENDIX E: Characteristics of included studies

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
Articles included from 2003 to Oct 09, 2007								
Berth 2006	Interview	Germany	76	• Not specified • Not specified • Males and females	Genetic counselling and test results	-HNPCC, Lynch-Syndrome -Adult-onset -Autosomal dominant inheritance (A.D)	'relatives'	- abstract; limited information on subjects and methods
Blandy 2003	Survey	France	30	• Clinic-based sample • Affected • Females	Genetic risk information	- BRCA1/2 - Adult-onset - A.D.	1 , 2 , 3	- small sample size - no verification with relatives
Blase 2007	Semi-structured phone interview	California, USA	12	• Clinic-based sample • Parents of affected children • Males and females	Genetic test results	- hearing loss (GJB2, GJB6 genes) - early onset - A.D.	1 , 2 , 3	-small sample size -unrepresentative sample -Majority of participants had a positive test result - no verification with relatives
Bowen 2004	Survey: baseline & post counselling -part of a larger study	Washington, USA	221	• Community-based sample (Ashkenazi Jewish decent) • Unaffected participants (no personal history of breast or ovarian cancer, and did not have a family history indicative of a family history) • Females	- General communication - health communication - breast cancer risk communication	- HBOC - Adult-onset - A.D.	mother, father, sister, grandparent, aunts, uncles, children, grand-children	- volunteer sample - measurement instrument for communication not validated
Bradbury 2007	Semi-structured telephone interviews	Chicago, USA	128	• Clinic-based sample • Unaffected and Affected parents (unaffected children) • Males and Females	Genetic test result and risk	- BRCA1/2 - Adult-Onset -A.D.	offspring	- retrospective design (recall bias) - unrepresentative sample - potential response bias - no children interviews to verify parents perceptions
Clarke 2005	Prospective reports on the	UK and Australia	65	• Genetics clinic-based sample • Unaffected and Affected	Genetic test result	- HD, HBOC chromosome translocation,	'at-risk family members'	- relies on professionals understanding of clients' reasons for nondisclosure

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
	frequency of non-disclosure			participants (adult, pediatric and reproductive referrals) • Males and Females		Mendelian conditions - adult onset, children - A.D and complex inheritance		- only allows for 12 month period for disclosure to occur - completeness of professional reports is questionable
Daly 2003	counselling process/ interviews	Ireland	26	• Genetics clinic-based sample • Affected and Unaffected • Males only	Genetic test result	- BRCA1/2 - Adult-onset - A.D.	spouse and offspring	- small sample size - qualitative
Farkas Patenaude 2006	Cross-sectional survey	Boston, MA	273	• Clinic/Hospital-based sample • Affected and Unaffected with HBOC • Females only	Genetic Test result	- BRCA1/2 - Adult-onset - A.D.	1	- measurement instrument for communication not validated (potential for measurement error) - Intervention arm participants had more training with disclosure (may mean communication in this population is higher than normal) - no verification of participant self-reports -examined short time frame (4 months) for disclosure to occur - wide 95% CI for OR's (may indicate N not big enough to detect differences)
Forrest Keenan 2005	Interviews	Aberdeen, UK	56	• Clinic-based sample • HBOC: All unaffected, all female • HD: Unaffected and affected, male and female	Genetic test result	- HBOC and HD - Adult-onset - A.D.	'at risk family members'	-small sample size -selected, unrepresentative sample limits generalizability -volunteers
Foster 2004	Interviews	UK	15	• Genetics clinic-based sample • Unaffected • Females only	Risk and results	- BRCA1/2 - Adult-onset - A.D.	'at risk family members'	- small sample size -no verification of participant reports - qualitative

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
Gadzicki 2006	Structured telephone interviews - part of a larger study	Germany	332	• Genetics clinic-based sample • Unaffected and Affected • Females only	Risk and results	- BRCA1/2 - Adult-onset - A.D.	1 , 2 , 3	- paper is in the form of a letter to the editor (details of study design missing) - no verification with relatives
Gaff 2005	-Telephone interviews	Australia	12	• Hospital-based sample • Affected • Males and Females	Genetic Test result	- HNPCC - Adult-onset - A.D.	'at risk family members'	- small sample size - retrospective and subject to recall bias - selection bias
Gallo 2005	-Semi-structured interviews -part of a larger study	Chicago, USA	139	• Hospital-based sample • Caregivers of affected children • Males and Females	- Genetic disorder - management of condition - genetic nature of condition	various	children	- small sample size - retrospective - children's perspective was not measured - parents were only measured at one point in time
Gregory 2007	Semi-structured interviews -part of a larger study	UK	44	• Hospital-based sample • Affected and unaffected (carriers) • Males and Females	-genetic condition and genetic transmission	- Hemophilia - children - X-linked recessive	Nuclear family	- some interviews were held with the parents and their adult child which may have influenced the ability of the child to recount their own story of how they learned of their definite or possible carrier status - family member verification not done - small sample
Hallowell 2005	Semi-structured interviews	UK	29	• Genetics clinic-based sample • Unaffected • Males (and their spouse and eldest child)	-Genetic testing of father -the risk implications for offspring	- BRCA1/2 - Adult-onset - A.D.	Biological offspring	- small unrepresentative sample - snowball method used to enroll other family members meaning the initial contact decided who was enrolled -retrospective -sample was gender skewed -used two interview type methods

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
Hamilton 2005	Interviews -part of a larger sample	15 US States, Canada, Denmark	29	• Community-based sample • Not specified whether affected or unaffected • Not specified – definitely females in sample	-Genetic test result	- HBOC and HD - Adult-onset - A.D.	Not specified, likely all at risk	- small sample - all those at risk for HBOC tested positive - retrospective - did not have family member verification or perspectives -different interview methods used
Holt 2006	Semi-structured interviews	USA	8	• Convenience sample (2 families known by researchers) • Both families had affected fathers and at-risk children • Males and Females	Genetic risk	- HD - Adult-onset - A.D.	children	- convenience sample - small sample size - no observation of family dynamics because all interviews were separate - symptomatic family member in both families was male - subjects were volunteers
Kasparian 2006	Semi-structured telephone interviews	Australia	40	• Hospital/clinic based sample • Affected and unaffected • Males and Females	Hypothetical genetic testing	- Hereditary Melanoma - Adult-onset - A.D. (can be spontaneous)	'Family'	- small, unrepresentative sample (convenience sampling) - hypothetical
Kenen 2004	Interviews -part of a larger study	UK	21	• Clinic-based sample • Unaffected (with no family member having test but have a family history of HBOC) • Females only	Risk due to family history	- HBOC - Adult-onset - A.D.	'all family'	- small sample size - qualitative -only talking about risk – no tests results
Kenen 2004b	Interviews	UK	21	• Clinic-based sample • Unaffected (with no family member having test but have a family history of HBOC) • Females only	Risk due to family history	-HBOC -Adult-onset - A.D.	Women friends, spouses, brothers, sisters and children	- small sample size - same sample as Kenen 2004
Klitzman 2007	In depth interviews	New York, USA	21	• Clinic-based sample • Affected and unaffected	Genetic test result	-HD -Adult-onset	'All family'	-small sample size - interviewed them at one

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
				• Males and females		-A.D.		point only
Kohut 2007	Survey	Ontario, Canada	105	• Population-based sample • Affected • Males and females	Genetic test result	-HNPCC - Adult-onset - A.D.	'family'	- unrepresentative sample - most participants have not been identified as HNPCC mutation carriers therefore, results are hypothetical - only registry members
Landsbergen 2005	Cross sectional survey	Netherlands	50	• Hospital-based sample • Affected and Unaffected • Females only	Genetic Risk Information about mutation	- BRCA1/2 - Adult-onset - A.D.	1 , 2	- retrospective - small sample size - experiences not validated with family members - Use 'getting a genetic test' as a proxy to indicate that the family member has been informed. Other 'informed' patients may decide not to do testing.
Lim 2004	interviews	Australia	47	• Population sample • Unaffected • Females only	Genetic test result	- BRCA1/2 - Adult-onset - A.D.	'wider family'	- small sample - not culturally generalizable - qualitative - only relates to process of telling
MacDonald 2007	Surveys: longitudinal study - Part of a larger study	California, USA	122	• Clinic based sample • Affected and unaffected • Females only	Genetic/cancer risk	BRCA1/2 Adult-onset A.D.	1	- high % of cancer survivors which limited findings differentiated by cancer history. - small sample size - unrepresentative population
MacKinnon 2007	Survey : pre & post intervention	Vermont, USA	28	• Clinic-based sample • Affected and unaffected • Males and females	Genetic testing and results	- BRCA1/2 - Adult-Onset - A.D.	1 , 2 , 3	-small sample size - self selected, well educated, high income group of primarily women
McGivern 2004	Cross sectional survey	Cincinnati, USA	38	• Hospital based • Affected and Unaffected • Females only	Genetic test result	- BRCA1/2 - Adult-onset - A.D.	1 , 2 , 3	- potential for response bias - retrospective - small sample size - nieces and nephews may have been told but not by the index person - Several sections of survey

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
								were left blank by lots of participants
Mellon 2006	Focus groups	Michigan, USA	39	• Community and Clinic-based sample • Affected (who selected an unaffected relative) • Females only	Inherited cancer risk information	- BRCA1/2 - Adult-onset - A.D.	1 , 2 unaffected female relatives	- small sample size - restricted to women - cancer survivor self-selected the female relative to recruit into the study, communication was likely already open.
Mellon 2007	Interviews - part of a larger study	Detroit, USA	292	• Population-based sample • Affected and unaffected • Females only	BRCA1/2 risk	BRCA1/2 - Adult-onset - A.D.	'family members'	- abstract; limited information on sample and methods
Mesters 2005	Semi-structured interviews	Netherlands	30	• Clinic sample • Affected and unaffected • Males and Females	Hereditary predisposition	- HNPCC - Adult-onset - A.D.	'at-risk family members'	- small sample size - retrospective
Riedijk 2005	Cross sectional survey	Netherlands	66	• Hospital-based sample • Affected and Unaffected • Males and Females	Carrier status	- Hereditary Melanoma - Adult-onset - A.D. (can be spontaneous)	1 , 2 , 3	- small sample size - retrospective - self-selected sample
Schroy 2005	Telephone survey	Boston, USA	71	• Hospital-based sample • Affected (with colorectal adenomas) • Males and Females	Risk of Colorectal adenoma	- Colorectal adenoma -Adult-onset - A.D.	1	- small sample size - retrospective - potential response bias - no verification from family members - disagreement in screening guidelines may have altered whether physicians discussed FDR risk
Segal 2004	Survey	Ontario, Canada and Melbourne, Australia	31	• Hospital or clinical- based sample • Affected and Unaffected by cancer (all mutation carriers) • Females	Genetic test result	- BRCA1/2 - Adult-onset - A.D.	offspring	- limited to female BRCA1/2 carriers - small sample size - retrospective
Taylor 2005	Cross sectional	Australia	57	• Community Based • Unaffected	HD Risk	- HD - Adult-onset	Spouse, family of	- small sample - unrepresentative sample

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
	survey -part of a larger study			• Males and Females		- A.D.	spouse, GP, friends, acquaintances, priest, employer, bank	
Van den Nieuwenhoff 2006	Plus-minus method for evaluation Semi structured interview	Netherlands	16	• Community/clinic based sample • Affected and unaffected (relatives) • Males and Females	Genetic risk	-Inherited High Cholesterol (IHC) -Adult onset -A.D. or A.R.	1 , 2 , 3	- small sample, unrepresentative - highly self selected group
Van den Nieuwenhoff 2007	Semi-structured interviews	Netherlands	20	• Community/clinic based sample • Affected • Males and females	Genetic risk and preventive measures	-Inherited High Cholesterol (IHC) -Adult onset -A.D. or A.R.	1 , 2 , 3	-Unrepresentative sample -Recall bias -No verification with relatives
van Oostrom 2007	Survey	Netherlands	271	• Clinic/Hospital- based sample • Affected and unaffected • Males and females	Familial communication style about hereditary cancer	- BRCA1/2 and HNPCC - Adult-onset - A.D.	- partners, children, parents, siblings, second degree relatives	- did not take into account difference between BRCA1/2 and HNPCC in analysis - no family member verification
Weiner 2005	Survey - part of a larger study	USA	717	• Community-based sample • Not clearly stated, assume unaffected • Males and females	Genes and health	N/A	'family'	- not culturally diverse population - was part of a larger study and thus couldn't investigate the topic of conversation in depth
Articles included up to November 2003 (from Wilson et al, 2003)								
Adelsward 2003	Interviews in counseling	Sweden	31	• Clinic-based sample • Not clearly stated, assume affected and unaffected • Males and Females	Genetic risk information and family history information	- 'Hereditary cancer' -Adult-onset - A.D.	All	- small sample size - qualitative
Ayme 1993	Survey	Marseille,	289	• Clinic-based sample	Genetic risk	- balanced	At-risk	- assumes that because people

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
		France		• Affected (index patient, and Unaffected family members) • Males and Females		chromosome relocation -children -complex inheritance patterns	family members	didn't get karyotyping that the information was not communicated to them by the index case - no direct measurement of communication -retrospective chart review and questionnaire review
Bruce 2003	Not stated	Aberdeen, Scotland	39 cou ples	• Not stated	Family history of HD and risk	- HD - Adult-onset - A.D.	Spouse/ partner	- abstract only; no methods or data presented
Claes 2003	- Semi-structured interview and; - structured questionnai re	Belgium	63	• Clinic-based sample • Affected • Males and Female (majority females)	Diagnosis, genetic testing and test result	- BRCA1/2 - Adult-onset - A.D.	1 , 2 , 3	- retrospective (long period between test result receipt and study commencement) - self-selected sample, response biase - content of communication not explored or verified
Cox 1999	In depth interviews	BC Canada	51	• Clinic-based sample • Unaffected • Males and Females	Genetic information, test result	- HD - Adult-onset - A.D.	Spouse /partner, parents, siblings, children, grandmothe r, nephew	- no men learned that they had inherited the mutation – gender differences difficult to interpret. - snowball recruitment strategy – self-selected sample
d'Agincourt Canning 2001	Interviews -part of a larger study	Canada	36	• Clinic-based sample • Not clear whether affected or unaffected or both • Males and Females	Genetic test result	- HBOC - Adult-onset - A.D.	'At-risk' family members	- small number of male participants does not allow for generalization - small sample size - unclear description of methods and study design -qualitative
Denayer 1992a	survey	Leuven, Belgium	183	• Hospital-based sample • Unaffected (had affected nieces and nephews) • Males and females	- Genetic information and diagnosis of disorder	- Cystic Fibrosis - children - A.R.	-the parents of the CF child (disclosing to aunts	- retrospective - did not verify with other family members - did not evaluate content of communication

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
							and uncles)	
Denayer 1992b	survey	Leuven, Belgium	109	• Hospital-based sample • Unaffected (had affected nieces and nephews) • Males and Females	None – this study assesses the aunts and uncles knowledge of genetic aspect of CF and evaluation of their risk	- Cystic Fibrosis - children - A.R.	n/a	- assume that information was not given to aunts and uncles because they don't have high knowledge (do not verify with parents whether they told the aunts and uncles, and paper does not clearly refer to a question about whether the aunts and uncles were asked who told them the information). - depend on parents cooperation to give addresses of aunts and uncles - No family member verification - did not evaluate content of communication -retrospective
Duster 1999	Interviews	USA	369	• Clinic and community-based sample • Males and Females • Unaffected (as they are discussing carrier testing)	Genetic nature of disorder	- Cystic fibrosis and sickle cell anemia - children - A.R.	n/a	- little methodological detail and participant information - book chapter which gives little reference to findings of research (evidence-based) studies.
Fanos 1995	Interviews	Oakland and Boston, USA	84	• Hospital-based sample • Males and Females • Unaffected (as they are obtaining carrier testing)	Testing for carrier status	- Cystic fibrosis -children -A.R.	'family members'	- qualitative - retrospective
Fitzpatrick 1990	Structured interviews	Dublin, Ireland and Massachus etts, USA	96	• Hospital-based sample • Males and Females • Parents of Affected boys and Affected boys	- General communication -communication about the disorder	- Duchenne Muscular Dystrophy - children -x-linked recessive	Parents to children (affected) and to spouses	- small numbers of participants - findings are specific to the two locations - findings may reflect broad cultural differences rather than unique to DMD - parents knowledge and child's knowledge was not

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
								measured
Forrest 2003	Semi-structured interviews	Aberdeen Scotland UK	37	• Clinic-based sample • Males and Females • Unaffected HBOC • Affected and Unaffected HD	Test results	-Huntington's Disease - HBOC - Adult-onset - A.D.	'at risk' family members	-no males in the HBOC sample - selected sample - in the HD group fewer mutation carriers than non carriers participated which may reflect difficulties in communication for the carriers.
Green 1997	Interviews	Cambridge UK	46	• Clinic-based sample • Females • Unaffected	- Family history info. - post counseling risk information	- HBOC - Adult-onset - A.D.	All family members	- small sample size - qualitative - more maternal side relatives than paternal (overrepresented)
Hallowell 2003	In depth interviews	UK	30	• Clinic-based sample • Females only • Affected (had breast or ovarian cancer)	Genetic test results	- HBOC - Adult-onset - A.D.	'at risk' family members	- small sample - qualitative - retrospective
Henneman 2002	Semi-structured interviews	Netherlands	18	• Clinic-based sample • Males and Females • Affected (with CF carrier status only)	Carrier status test result	- Cystic Fibrosis - children - A.R.	'at risk' family members	- small sample size - retrospective study - qualitative
Hughes 2002	Telephone interview	USA, and Toronto, Canada	43	• Clinic/hospital-based sample • Females only • Affected	Genetic test result	- BRCA1/2 mutation - Adult-onset - A.D.	Sisters	- small sample - no verification with other relatives about their reactions - the intense counseling process could have facilitated communication
Julian-Reynier 1996	Survey	France	209	• Clinic-based sample • Females only • Affected and unaffected	Genetic testing for BRCA1/2 mutation and genetic risk	- BRCA1/2 mutation - Adult-onset - A.D.	'at-risk' family members	-Unrepresentative sample (more highly educated, restrictive eligibility criteria)
Julian-Reynier 2000	Survey	France	398	• Clinic-based sample • Females only	Genetic test result	- HBOC - Adult-onset	parents, siblings,	-Hypothetical results (they haven't received them yet)

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
				• Affected and unaffected		- A.D.	children, spouse and GP	may differ from actual actions.
Koehly 2002	Interview	Texas, USA	36	• Clinic/hospital-based sample • Males and Females • Affected (patients)and unaffected (family members)	Genetic counseling and testing	- HNPCC - Adult-onset - A.D.	'All' -defined by participant (includes friends and coworkers)	-small number of families - Participants may be highly self-selected because they had also agreed to participate in another research study. - the recruitment process may have influences their decision to participate in testing and may have prompted greater communication about genetic testing - low response rate -retrospective
Lerman 1998	Survey in prospective cohort	USA	201	• Clinic-based sample (study one was family-based and study two was individual) • Males and Females • Not clearly stated whether affected or unaffected	Test results	- BRCA1/2 mutation - Adult-onset - A.D.	Spouse, brother, sister, children	- lacking description of recruitment and study sample
Miesfeldt 2003	Survey	Virginia, USA	267	• Hospital-based sample • Females only • Affected	Breast cancer risk (focus was on opinions and beliefs)	- HBOC - Adult-onset - A.D.	offspring	- proportion of women from target population who agreed to participate was small. -Unrepresentative sample -did not measure who they told
Ormond 2003	Survey	Chicago, USA	48	• Hospital-based sample and general population screening sample • Males and Females (carriers of CF gene) • Affected with carrier status • One group had a family history of CF and one did	Carriers status information	- Cystic Fibrosis - children - A.R.	1 , 2 , 3	- small sample size - low response rate in both groups - differences in education and religious affiliations between groups - data was not independently verified with relatives - differences between methods of counselling

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
				not.				between the two groups was not examined
Peterson 2003	Interviews (telephone) -part of a larger study	USA	39	• Hospital-based sample • Males and females • Affected and Unaffected	-perceptions of HNPCC -genetic testing -genetic risk	- HNPCC - Adult-onset - A.D.	1 , 2 , 3 , spouses	- retrospective and subject to recall bias - Self-selected sample who participated in another related study. - recruitment method may have made it more likely for these families to communicate about these topics.
Shakespeare 1993	interviews	Leicester	42	• Not stated • Not stated • Not stated	Hereditary nature of the disease	- HD - Adult-onset - A.D.	Not specified	- Abstract: little detail on participants and recruitment
Skirton 1998	Semi structured interview	UK	15	• Not stated • Males and females • Affected and unaffected (spouse) and at-risk	Genetic risk	- HD - Adult-onset - A.D.	Children	- limited description of methods, recruitment etc - small sample -non representative sample
Smith 2002	Interview (telephone) - part of a larger study	Utah and southern Idaho, USA	305	• Hospital-based sample • Male and female • Not stated	Test result	- BRCA1 mutation -Adult-onset - A.D.	Sisters, brothers, daughters, sons, coworkers, health insurance provider, life insurance provider,	- unrepresentative sample
Sorenson 2003	-Survey - telephone interviews - part of a larger study	USA	181	• Clinic-based sample • Affected patients were male and female, unaffected relatives were only female. • Affected and unaffected relatives	Carrier test result	- Hemophilia A - children - x-linked recessive	'at-risk' family members	- DNA based carrier test education, testing and post test genetic counseling were offered free to at risk relatives which may have increased interest and discussion.

Author	Study	Country	N	Sample (community based, hospital based, affected or not, gender)	Topic	Disorder (late- onset, children etc)	Relatives	Study Limitations
								- self-selected sample
Suslak 1985	Interview	New Jersey, USA	12	• Clinic-based sample • Male and Female (family) • Affected by carrier status	Discovery of genetic problem /carrier status	- Balanced translocation -children and adult carriers -complex	-parents and siblings	- small sample size - retrospective
Varekamp 1992	survey	Netherlands	500	• Clinic-based sample • Females only • Affected (with carrier status) and unaffected (with carrier status)	Hereditary nature of hemophilia -genetic testing and counselling	-Hemophilia -children -x-linked recessive	'at- risk' family members	- unrepresentative sample
Wagner Costalas 2003	Telephone survey /interview	USA	162	• Clinic-based sample • Females only • Affected and unaffected	Genetic test results	- BRCA1/2 mutation - Adult-onset - A.D.	Adult children and siblings	- no validation of accuracy of self-reports - number of probands with negative, positive or indeterminate results was disproportionate -low number of participants who tested negative
Wolff 1989	Ascertainment of chromosome translocation	Germany	36 f	• Clinic or hospital-based sample (registry?) • Males and Females index patients • Affected (index patients with carrier status)	chromosome translocations carrier status	- Balanced Chromosome relocations -children and adult carriers -complex	All family members	- limited description of recruitment and sample - No description of whether results were drawn from participant or assumed by researcher.

APPENDIX F: Participant description and recruitment strategy for included studies

Author	Study Objective	Participants	Recruitment
Berth 2006	Investigate the extent and determinants of family communication about genetic counselling	- 64.5% female, 35.5% male - Mean age = 41.8 years	-76 people who were interested in knowing their carrier status were assessed two weeks prior to genetic counselling. -Provided data on family cohesion, family adaptability, and psychological distress.
Blandy 2003	1) describe, in BRCA+ families, the diffusion of information by index cases, testing participation among first and second degree relatives and first cousins 2) assess understanding of the information by index cases and their satisfaction with the testing process 3) determine the factors associated with a higher/lower diffusion of information and testing participation among high-risk relatives	-All participants were women with breast /ovarian cancer (contacted between April-September 1999). -They were regularly followed at the Institut Cure. - Positive for a BRCA1/2 mutation and had been informed about their result at least 10 months before the survey. -Mean age of the women was 52 years -93% had breast cancer and 7% ovarian cancer; 70% and 30% had a BRCA1 or BRCA2 mutation, respectively. -For all index cases, the time spent since the information about the BRCA results was over 8 months -21 were married, 2 were single and 7 were divorced, separated or widowed, -18 had boys and 23 had girls -12 had less than high school education and 18 had greater than high school education	-Two questionnaires were completed -The living index cases were contacted by mail, including an introductory letter about the survey, its objectives, a consent form to participate and the first questionnaire. -The completed self-administered questionnaire was returned anonymously by mail in a stamped addressed enveloped. -A reminder was sent when no answer was received after two weeks, followed by a second complete envelope after another two weeks. -The second questionnaire was completed by telephone interviews with the women who had given their consent to participate.
Blase 2007	Identify if, how and why parents of young children with hearing loss who have had GJB2/GJB6 genetic testing share the genetic test information with family members	- Study sample consists of parents of a young child diagnosed with hearing loss who underwent GJB2/GJB6 genetic testing. -19 individuals (42%) responded to the letter of invitation. -Twelve of those responses (63%) were individuals interested in participating, seven declined participation. -The sample was composed of 12 participants: 10 females and 2 males. Representing 10 independent families -7 participants received a GJB2/GJB6 test results that explained their child's hearing loss (positive test result), four participants received a test result that did not explain their child's hearing loss(negative test result), and 1 received an inconclusive test result.	-Recruitment focused on parents who were participating in the University of California, Los Angeles (UCLA) Genetics of Hearing Loss study, which offers genetic counseling and genetic testing g for GJB2 and GJB6 to infants/toddlers with apparently non-syndromic hearing loss who are under the age of three years at no change to the family - Families are typically referred to the UCLA genetics of hearing loss study by their audiologist or early intervention teacher. Genetic counseling in the UCLA Genetics of Hearing Loss study is provided at enrollment and at result disclosure using a semi-structure format. - Parents received a copy of the test result and a written summary of the result disclosure counseling session. Individuals were eligible to participate in this study if they met all of the following criteria: 1) have a child with apparently non-syndromic hearing loss that underwent GJB2/GJB6 genetic

Author	Study Objective	Participants	Recruitment
		- majority were female (83.3%), Caucasian (58.3%), college-educated (66.7%) with an average age of 33.4 years and median family income greater that 95,000. -The majority of the children of these participants were female (70%) with average age of enrollment of 10.8 months (SD=9.3m). - There was a positive family history of hearing loss, defined as having congenital through early adulthood onset in first, second or third degree relatives of the infant in 40% of the families represented in the sample (5) although all participants were hearing. -There were few demographic differences between participants with a positive family history and those with a negative family history. -There were no male participants in the group who received a negative test result.	testing and counseling as part of their participation in the UCLA Genetics of Hearing Loss study 2) gave permission to be contacted about future studies on their consent form 3) at least 18 years old and 4) English speaking or American sign language -Forty six eligible individuals, 16 received positive test results, 26 received negative test results, and 4 received inconclusive test results -contacted by mail with a letter of invitation about the study and a contact form. Individuals were requested to return the contact form indicating whether or not they were potentially interested in participation in the study. -Those who did not respond to the first mailing within a two week time period received a second letter of invitation. -Interested participants were contacted to schedule either a phone interview or a fact to face interview.
Bowen 2004	Understand how women perceive their risk of breast cancer and how family communication relates to coping, health-related quality of life and health outcomes. -Report on how family communication and support regarding breast cancer risk affects interest in genetic testing	- women 18-74yrs (47 years average) - had at least one third degree relative of Ashkenazi Jewish descent -No personal history of breast or ovarian cancer -no family cancer pedigree indicative of an inherited autosomal dominant susceptibility -lived within 60 miles of Seattle, Washington -72.7% were married or living with a partner, 90% had completed college, 62% received some graduate education. Majority had moderate level of income and 48% had a household income of 70,000 USD.	- Local chapters of national Jewish orgs. , area synagogues, area newspapers, local libraries, community centers, community schools and social groups. -Announcements placed in newsletters, posting of study flyers and brochures, verbal announcements at organization gatherings, flyers in mailings of organizations, researcher visit to organization functions
Bradbury 2007	Describe the prevalence and experiences of parental communication of BRCA results to children under the age of 25 years old. Attempt to describe parental disclosure experiences among BRCA1/2 mutation carriers and evaluate impressions of the impact of disclosure over time	-42 BRCA mutation carriers from 32 unique families and 86 children. - Median age of parents was 45 (range 28-66) -37 were female and 5 were male - 27 had a BRCA1 mutation, 14 had BRCA2 mutations and BRCA1 and BRCA2. -39 were white, 1 black and 2 Hispanic - 36 were married, 5 divorced and 1 widowed - 15 had college degree, 11 had a graduate education, 8 had some college and 8 had a high-school degree. - 23 had a personal history of cancer and 19 did not. - 17 had mastectomy and 31 had an oophorectomy. - 9 people had 1 child, 32 had 2-4 and 1 had more	Patients were recruited through the University of Chicago Cancer Risk Clinical. Participants were BRCA1/2 mutation carriers who had previously enrolled in a registry for cancer-prone families, provided consent to be contacted in the future, and had at least one child younger than 25 at the time of parent genetic testing. -Potential participants were contacted by telephone, and verbal consent was obtained. Interviews were conducted by two trained research assistants over a 12-month time period (February 2004 to February 2005). -Of 171 BRCA1/2 mutation carriers enrolled onto the Cancer Risk Clinic, 112 were deemed ineligible for the study. Sixty-two mutation carriers did not have children who met the inclusion criteria, 26 had not signed consent allowing re-contact, 11 were

Author	Study Objective	Participants	Recruitment
		than 5 -median age of child when parent underwent testing was 12 years (5-24 years). - 30 children were less than 10, 16 kids were 10-18, 13 were 14-17, and 27 were 18-25. - There were 53 female children and 33 male children.	deceased, nine had insufficient contact information and four had not received, or did not understand, their test results. Forty-three of the 50 (73%) eligible participants completed the telephone survey.
Clarke 2005	Determine the frequency of non-disclosure of genetic information to relatives, the circumstances in which these situations arise and the actions taken by the genetic health professionals.	-A total of 65 cases of non-disclosure were reported. - Disorders reported included Huntington's Disease, chromosome translocations, HBOC, other mendelian conditions	-All patients in the 14 regional genetic services clinics from 2000-2001 - A total of 12 regional genetic services in the UK and two in Australia collaborated in this study - Researchers had ethics approval to report non-identifiable information without patient consent.
Daly 2003	- report on the process and outcome of 26 males from 10 families who underwent predictive testing for mutations in the BRCA1/2 genes from their research program.	- 26 men from 10 different families -Of the 26 who had predictive testing they represented 60% of those men within the families who had received information regarding this option -Age range was 28-82 (median 55) -Five men had cancer before entering the study Three of these were from the same family One had uretic and prostate cancer and one had prostate cancer and the third had invasive squamous cell cancer at two sites. Other men had prostate and breast cancer -three men did not have children, one of these was considering having children -24 men had spouses and 23 had 89 offspring. - family size ranged from one to 11 children (median 3). -Fifty of the offspring were female and 39 male -the relationship of the original mutation carrier to the male participant was a cousin in 15 participants, a daughter in 3, a sister in 1, and brother in 2, a nephew in 3, a niece in 1 and one was the index case.	-Men who had testing between 1996 and 2002 in an 'Irish setting' -location not specified
Farkas Patenaude 2006	-Examine sharing of BRCA1/2 results with first degree relatives by 4 months post disclosure	- 18 years or older - has had a personal or family history of breast or ovarian or other cancer consistent with BRCA1/2 heredity	-Women who enrolled in a randomized study of two genetic counselling interventions (Genetic Risk Awareness and Cancer Education) at the Dana-Farber Cancer Inst. and collaborating hospitals from Dec 1998 to July 2000

Author	Study Objective	Participants	Recruitment
	-determine which factors predict informing FDRs of BRCA1/2 results	- had documentation of cancer diagnosis - all white women, 93% had at least some college education -60% had either breast or ovarian cancer - Age ranges: less than 40, 41-50, greater than 50 were all equal representation. -273 participants were from 244 families -204 participants were the first in the family to undergo BRCA1/2 testing - Majority of participants were married, and half the sample reported an annual household income of $95000 or more. -25% had positive test, 12% had a negative test and 63% had an inconclusive test.	
Foster 2004	- Considers social role expectations within the family and how these may influence communication about genetic testing and associated risks. - Explore how women communicate with their relatives about genetic testing -Investigate communication between healthy women, who had a genetic test for BRCA1/2 and their relatives both before and after the test result.	- median age was 46 years (33-62) -7 women had one daughter and 5 had one son, 6 had two daughters and 3 had two sons. -Age of children ranged from 3-40, median was 21 years -All were white British women -13 maternal line of transmission and 2 had paternal line -10 had breast cancer only in the family -5 had both breast and ovarian cancer in the family -The number of affected first degree relatives ranged from 0-3 - 5 of the 15 were found to carry a BRCA1/2 mutation	-A consecutive series of healthy women from families with HBOC attending a genetics clinic for predictive testing - A known BRCA1/2 mutation in the family (an affected family member had been tested and identified as a carrier of a BRCA1/2 mutation) - they provided blood for the BRCA1/2 analysis -Over 18 years -unaffected with breast and/or ovarian cancer at the time of the test -no psychiatric history
Gadzicki 2006	Gain deeper insights and to identify factors that influence the communication process within the family	- 65% of their participants were affected with breast cancer -35% of participants carried a BRCA1 or BRCA2 mutation.	-Women from the German Cancer AID Consortium Study on HBOC, included 11 centres who underwent genetic testing and obtained their result at least six months previously
Gaff 2005	-Explore the experiences of probands with HNPCC when informing relatives that predictive genetic testing is available -obtain their perceptions of the utility of genetic	-Seven women and five men were interviewed -All except one participant was participating in regular surveillance for colorectal cancer -Respondents were older than non-respondents (median age 55 vs. 49) -Respondents had more cancer diagnoses per individual than non-respondents	-Patients from a familial cancer clinic at a metropolitan tertiary referral hospital who had been diagnosed with a HNPCC related cancer by genetic testing between 1997 and 2001 - had been diagnosed with an HNPCC related cancer - had a diagnostic genetic test saying they carried the pathogenic gene mutation causing HNPCC - were asked by genetics clinic to inform at risk relatives that

Author	Study Objective	Participants	Recruitment
	counseling and communication aids in this process. -impacts of gender on communication in a situation where both men and women have a high risk of developing and inherited cancer	- Respondents were more likely to be female than non-respondents - median time from test result to interview was 31 months (7-66) for respondents -Median time from test result to interview for non-respondents was 36 months (8-36)	predictive testing was available -had no other family members identified with a mutation at the same time of result disclosure - All except one participant were participating in regular surveillance for colorectal cancer
Gallo 2005	-To examine parents beliefs and strategies related to sharing information about a genetic condition with their affected and unaffected children	- participants came from 86 families -all participants were either parents or caregivers of the child -child was biological to at least one parent or caregiver - the children with genetic conditions were school aged or adolescents at the time of the interview and were within two years of their regular classroom by age. -The parents reflected a range of ages from 22 to 57 years (median =40) and had varied educational backgrounds. - more than half the sample was white Most were protestant or catholic -The children with genetic conditions ranged in age from 3.7 years to 15.9 years -45 of the children were female -The healthy siblings ranged from 4 months to 34 years, and some did not reside in the household. -15 of the families had at least two living children with the genetic condition - 19 reported income of less than 20,000 and 25 reported income greater than $100,00	-Parents of children with various genetic conditions and primary caregivers were recruited from three clinical sites in Chicago - project director contacted parents regarding their interest in participating in the study, either through direct contact at the clinical site or after they had expressed interested after receiving a letter from the site's clinical director.
Gregory 2007	Explore communication of families where one individual has haemophilia	None given	-Participants were recruited using the records of a comprehensive care centre of one UK hospital. The centre has operated for some years under the current UK guidelines, testing of non-obligate carrier status when the girls is judged to be Gillick competed, but also testing for coagulation status in at risk females during early childhood. Male index cases and mothers of sons with haemophilia were identified from the patient records. Between September 2004 and December 2005 information sheets were dispatched, giving details

Author	Study Objective	Participants	Recruitment
			of the study and inviting participation. In total 44 interviews were carried out, comprising seven male index cases, 2 wives, 19 obligate carriers, 13 non-obligate carriers and three women where the origin of the condition appeared to be a de novo mutation Two participants were interviewed twice, with a year's gap between the interviews in order that progress in disclosing information to their daughters could be followed
Hallowell 2005	- Exploration of the way in which information about predictive BRCA1/2 testing and its implication for children is disseminated within the families of at risk men who undergo genetic testing -describes the patterns of information disclosure and the justifications for using such strategies -document communication problems	-current partners and eldest adult children of male carriers and non-carriers -must have been >18 years -no diagnosis of cancer or mental illness -partners were normally the biological mother of the children in the family -there were 17 men (5 carriers, 12 non-carriers), 3 partners (three carriers and five non-carriers) and 4 children (daughters of non-carriers) -in nine families where more than one individual was interviewed: five couples, three family groups and one father-daughter pair. -white European or Ashkenazi Jewish descent -socio-economically unrepresentative of the British population but representative of the population undergoing predictive testing -Median age of fathers was 55 years (39-75) -Median age of partners was 51 (45-60) -Median age of children was 25 (19-37) - 15 men were married or living as married and 2 were divorced or widowed -the fathers family cancer history ranged from 3-7 members The mean time since receipt of results was 26 months (8-74)	-In two regional genetics centres in the UK families – male carriers and non carriers, their partners and eldest adult child were asked to participate -a letter from the clinician responsible for their care was sent to all eligible men -partners and adult children were recruited using snowballing methods. The mail patient was given a letter to pass on to his partner and eldest son/daughter
Hamilton 2005	-describe the experiences of people disclosing genetic test results for HD and HBOC to biological family members	-participants lived in 15 US states, Canada, and Denmark - 24 had completed predictive genetic testing and 5 who had decided against testing. - Of the 24 tested 14 were tested for BRCA1/2 and 10 for HD. -All of the 14 HBOC were tested as positive. -Of the 10 HD testees four had fully penetrate mutations and one had a reduced penetrance mutation.	-Purposive sampling -Recruited through an advertisement in a regional magazine in the Midwest US and posting notices on HBOC and HD support group web sites. -Recruitment took place over 4 months - theoretical sampling procedures

Author	Study Objective	Participants	Recruitment
		- Of the five not tested, two were at risk for HD and three were at risk for HBOC.	
Holt 2006	- to understand the process of disclosure to children, in the context of predictive genetic testing for HD. - Explore the contrasting choices of two sets of HD parents regarding disclosure of genetic risk status to their children. - explore the children's (now adults) lived experience growing up with the different disclosure choices.	Family 1: five members, four of whom participated in the study. The father was 47 and symptomatic for HD. The mother was 45 and not at risk. They had two biological daughters. The 19 year old as considering testing and was eligible to do so. The middle daughter was 16 years and was also considering testing and would be eligible shortly. The youngest daughter was too young for the study and was adopted. This family's family history with HD went back four generations and with each generation, the age of symptom presentation had decreased. The great-great grandfather became symptomatic in old age, (90's), the great grandfather in his 70s, and the grandfather in the 50s and the dads the 30s. -Family2: 4 members. The father was 49, and was symptomatic with HD, the Mom was 47 and not at risk. The son was 21 and the daughter was 19. Both kids were 50% at risk, and considering predictive genetic testing. This family had a family history with HD for four generations on dad's side, with each generation experiencing symptoms at a younger age: 60years, then 50 years, and 30 years. And a paternal nephew had testing and is positive in his early 20's and is not symptomatic.	- Convenience sample of two families selected through personal contact. -Family one volunteered for research involving HD - Family two was recruited as a result of a discussion of the research study at an HD convention -Participants had to have a family history of HD, at least one at risk family member had to be actively considering the use of predictive testing for themselves at the time of interview and the family have to live close enough for an interview. One at-risk person in the family who was considering predictive testing
Kasparian 2006	To document anticipated emotional, behavioral, cognitive, and familial responses to both mutation carrier and non carrier status following hypothetical genetic testing for melanoma susceptibility. Aimed to provide preliminary qualitative data on differences in views for a) those with a strong family history of melanoma compared to those without b)	-Sample comprised of forty participants, recruited into one of four equally sized groups on the basis of family history (individuals from families with a high density of melanoma versus those without a family history of melanoma) and disease status (those with and those without, a previous diagnosis of melanoma). -During interviews with two participants (one from Group 1 and one from group 2) it was discovered that predictive genetic testing for melanoma risk had already been undertaken and a test result received. -All participants were 18 or older and were excluded if they were currently under active treatment for locally advanced or metastatic melanoma.	-Data were collected from four equally sized samples of Australians who differed in terms 1) prior melanoma diagnosis and 2) family history. -Individuals with a family history of melanoma who had either previously been treated for melanoma or not , were ascertained via the Westmead Institute for Cancer Research/University of Sydney centre of the Genetic Epidemiology of Melanoma study. This is part of the international GenoMEL consortium, a multidisciplinary study of the genetic epidemiology of melanoma. Multiple-case melanoma families (at least three relatives with a past diagnosis of melanoma) have been ascertained from south eastern Australia to the Sydney arm of this study for over 16 years through either; i) a family member who attended the Sydney melanoma unit, the Victorian melanoma service, of other clinics,

Author	Study Objective	Participants	Recruitment
	those with a previous diagnosis of melanoma compare d to those unaffected by melanoma and c) males versus females. Sought to comment on potential differences between anticipated responses to genetic testing for melanoma risk compared to actual responses to testing for hereditary breast and colorectal cancers.	-Only English. -62 study invitation letters were mailed to prospective participants. -5 declined participation for reasons such as recent death of a loved one, ill health, and disinterest. Follow up was discontinued once 40 interviews (10 interviews per group) had been conducted. -Age ranged between 22 and 80 years. Mean age was 48. -19 were male and 21 were female. -19 had post school qualifications and 21 had school qualifications only -26 were married, 14 were not married -31 worked indoors, 6 worked outdoors and three spent even time indoors and outdoors. - 31 had biological children and 9 did not.	for treatment of melanoma ii) referral from health professionals such as clinical geneticists or dermatologists or occasionally iii) self-referral after media publicity. Data on family structure, cancer history, illness characteristics, skin phenotype, other melanoma risk factors, and genotype were collected. Patients who had received treatment for melanoma in the previous year and who had no family history of the disease were recruited by their surgeon, via the melanoma clinic at Westmead Hospital, a major clinic within the Sydney Melanoma Unit network. A sample of unaffected participants with no family history of melanoma was recruited by asking participants to nominate one friend or colleague of the same six and similar age who might be willing to take part in the study. Participants then approached this nominated person for permission to forward their contact details to the research team. Once this had taken place, individuals were contacted by the researchers in an identical manner to all other study participants. Purposeful sampling. Similar numbers of male and female participants were recruited to the sample, and all participants were from distinct families. A letter of invitation, signed by either the Principal Investigator of the Genetic Epidemiology of Melanoma study of the treating surgeon, was sent to selected individuals accompanied by a study information sheet and consent form. Individuals who did not decline participation were subsequently contacted to obtain informed consent and arrange a time for participation in the study.
Keenan 2005	-Explores the communication in families at risk of two late-onset genetic conditions. -Examine who takes responsibility for telling -Examine from the family perspective	- 29 participants had HD in their family and 27 had HBOC in their family - HD group: 16 individuals who had attended counseling, 12 partners and one other relative - HBOC group: contained 20 women who had received counseling and seven partners - urban and rural populations - All but six of the participants were 35 years old or older. -Mean age of the HD participants was 46.8 (26-65) -Mean age of the HBOC participants was 40.5 (25-62) -69% of the HD participants and 86% of the HBOC participants were married or living with their partner -94% of the HD participants had siblings while 62% of the HBOC group had siblings.	Those who attended genetic counseling for HD and HBOC from the outpatient genetic counseling clinic at Aberdeen Royal Infirmary in Scotland. -Purposive sampling to recruit a wide range of demographic characteristics and risk statuses. -Theoretical sampling to invite individuals who were know to have experienced communication difficulties in their families as well as those who had experiences of more open communication - Clinical geneticists identified appropriate potential participants and then sent personal letters inviting them to take part in the study. Partners were invited through the participant.

Author	Study Objective	Participants	Recruitment
Kenen 2004	- investigate how family context and genetic clinic context affect family communication patterns - how family communication patterns and family scripts influence the dissemination of genetic information and the sharing of feelings about genetic inheritance in families -How family members communicate information concerning breast/ovarian cancer in the family -the concept of family scripts can be used to explain some of these verbal interchanges and the influence of two social contexts in which members operation – the family context and the cancer genetics clinic context.	- twenty one women participated ages 24-61. - 13 were married, four were separated or divorced, one had a partner, one was engaged and two were single - 8 women had a bachelors degree or higher, seven had some additional training after their O or A levels -All women were either currently working or had paid employment some time in their lives - 6 had no children - of those who had children , five women did not have any daughters. - two stated no religion, nine belonged to the Church of England, four were Catholic and nine came from Jewish backgrounds One woman was Afro-British background and 20 were white	-Women from a busy, major clinical and research cancer center in UK from Jan to June 2000 -women had to never had breast/ovarian cancer -the woman is either from a family that has not undergone genetic testing or the women is unaware of any relatives' results -It is the woman's first visit to the breast/ovarian cancer genetics risk clinic -There are two or more cases of breast and or ovarian cancer in the family -The woman is 18 years or older and English -The interviewer did not have to travel more than 2 hr each way - eligible women received an information sheet a response form and envelope at their clinic appointment,
Kenen 2004b	-Investigate the influence of significant others on the communication patterns and interrelationships within families with a history of breast/ovarian cancer.	Same as above	Same as above
Klitzman 2007	To understand what, when, why, whom to and how people disclose HD genetic test results	21 participants 12 were male and 9 were female all but three participants were white 8 participants were mutation positive, 4 were mutation negative and 9 did not get tested 11 were married, 9 were single, one was divorced 8 had four years of college, 2 had less than four years of college, 4 had high school or less, 6 had some graduate education 3 had 3 children, 13 had zero, 2 had 2 children and 3 had one child	Potential participants were approached by staff at an HD clinic. Attempts were made to ask all people who underwent pre-symptomatic testing and a range of others if they would participate

Author	Study Objective	Participants	Recruitment
Kohut 2007	To survey participants of the Ontario Familial Colon Cancer Registry (OF CCR) regarding their duty to warn family members about a hereditary nonpolyposis colorectal cancer (HNPCC) – causing mutation, on opinions about personal responsibility, barriers to communication and the responsibility of health professionals, particularly if the participant was unwilling or unable to inform relatives.	A total of 105 OFCCR participants returned the survey, with a response rate of 46.3%. Five surveys were returned incomplete because the participants were no longer at that address and five other participants were reported deceased. Of the high-risk probands with no mutation identified, 52.4% responded to the mailing. Of the mutation carriers, 50% responded.	A one page survey was mailed to 227 participants in the OFCCR, a population based registry of colorectal cancer patients in Ontario, Canada. Individuals diagnosed between 1997 and 2000 aged 20-74 years were recruited. Participants received genetic counseling and were offered genetic testing. At the time of mailing the survey, 19 were identified as carriers of an HNPCC causing mutation and 21 were classified as 'high risk' based on meeting Amsterdam or modified Amsterdam criteria, but had test results suggesting the absence of a mutation or had not yet received results. The survey was mailed to an 197 additional probands classified as 'intermediate risk' based on family history of HNPCC related cancer, personal history of colorectal cancer at or before age 35 years, certain tumor pathology or Askenazi Jewish heritage.
Landsbergen 2005	Determine uptake rates for genetic testing in families diagnosed with a deleterious germline BRCA mutation and to explore possible facilitating and debilitating factors in the dissemination of the genetic information from index patients to relatives, with respect to cognitive emotional and behavioural factors.	- mean age was 49 years (31-71) - 90% were married - 80% had children - Of the children, 74% were daughters with a mean age of 20 (3-39). - Mean age of becoming aware of genetic risk was 41 years (+/-13). - Mean age of testing was 44 years (28-64) - Mean time period between disclosure and the study was 4 years (2-8 years) - Breast cancer occurred in 62% of participants and the mean age at diagnosis was 40 (+/-8). - 12% had a medical history of ovarian cancer with a mean age of diagnosis of 54 (+/-6). - 42% had both a bilateral prophylactic mastectomy and oophorectomy - 22% of the women chose breast cancer and ovarian cancer surveillance (not surgery) - The total number of family members of the participants diagnosed with breast cancer was 145. 91 family members died from breast cancer. 45 were diagnosed with ovarian cancer and 31 died. There were 14 families in which no one died from breast cancer, 26 families in which no women died from ovarian cancer and 9 families in which nobody died from either form of cancer.	Women who were the first BRCA1/2 mutation carrier in a family were identified from the registry of the Human Genetics Department of the University Medical Centre Nijmegen between 1995 and 2001.

Author	Study Objective	Participants	Recruitment
Lim 2004	Sought to discover the emotional and social impact of receiving results of genetic testing for HBOC by documenting the experiences of women participating in a registry of high risk breast cancer families and who had received either a positive and negative result for BRCA1 or BRCA2 or the ATM gene as well as those women who declined to know their test result.	- participants came from 11 study sites across Australia - 23 women were mutation positive - 24 women were mutation negative - Age ranged from 24-76 with a median age of 48 years -Fifty seven percent had no postsecondary school qualifications -The majority were married and had one or more children - The median time since receiving results was 13 months, ranging from 1 month to over 5 years	All unaffected women enrolled in 'kConFab' with available test results between 2001 and 2002 are eligible. kConFab is a research group established in 1996 to coordinate the collection of genetic, epidemiological, and clinical data in Australian families with dominantly inherited susceptibility to breast cancer. Women are eligible for kConFab if they have a family history consistent with a dominantly inherited susceptibility to breast cancer, and come from a family that meets the kConFab eligibility criteria. To be eligible for the kConFab study the participant had to have two or more carriers in the family (first or second degree from the informative side of the family) or have all of the following: i) four or more cases of breast or ovarian cancer (on the same side of the family) or two or more cases of breast or ovarian cancer in the same or adjacent generation, if at least one of these cases is high-risk and ii) two or more living, affected family members and iii) four or more living first or second degree unaffected female relative of affected cases. - All unaffected women enrolled in kConFab were eligible for participant in the kConFab psychosocial study. Women were recruited wither on entry into the main study or at approximately three years follow-up when their clinical information was being updated. Invitations to participate in the study were mailed from the coordinating research center, along with a detailed information and consent form, questionnaire and reply paid envelope. - Participants were recruited through 11 study sites across Australia. The current study focuses on a sub sample of the kConFab psychosocial study – unaffected women for whom mutation results were available, recruited between August 2001 and July 2002. and had received a predictive test where a family specific mutation had already been delineated in another family member
MacDonald 2007	To explore family communication of cancer risk of women before and by six months post-genetic cancer risk assessment regardless of whether BRCA testing was performed, and factors affecting this	Majority of women were between 40 and 60 years, white, married college graduates, Christian and had children. Approximately two thirds of the women had at least one living child aged 18 years or more (64% were daughters and 60% were sons), 81% had at least one child aged between 13 and 17 years (86% were daughters, 77% were sons) and 20% had a child aged	Data were previously collected between August 1999 and December 200 from standard self-administrated health and family history surveys and medical records. The study survey was mailed to women before their initial GCRA appointment and again at 1 month and 6 months post GCRA. Participants in the parent study were similar in demographics to the same number non-participating study-eligible women with and without a personal and or family history of breast or ovarian cancer.

Author	Study Objective	Participants	Recruitment
	communication - assess if there are differences in the type of family member selected for communication of cancer risk comparing the baseline with six months post - GCRA - Are there differences in perceived barriers to this communication at the same time points? - Are there differences in these two variables at the same time points by demographic, personal or family history of breast and/or ovarian cancer or BRCA status	between 6 and 12 (7% were daughters, 13% were sons). Respondents and non-respondents were similar in demographics (age, ethnicity/race, education, marital status, children and religion). And personal/family history of breast or ovarian cancers. 69 (83%) of the 83 cancer survivors had a family history of breast or ovarian cancer (five cancer survivors had a family history of both cancers. Only 3 (8%) of the 39 women in the no cancer history group had a family history of ovarian but not breast cancers. 66 women (54%) had a personal or family history suggestive of hereditary breast cancer or, in some cases, breast/ovarian cancers. Of these 50 had full sequencing of both BRCA genes and 16 Ashkenazi Jewish women had testing for the three founder mutations responsible for approximately 95% of BRCA-related breast/ovarian cancer is Ashkenazim. Sixteen women (24.2%) were found to carry a deleterious BRCA mutation, including two Jewish cancer survivors and one of the three women without a personal cancer history. Eighty three women (68%) had a personal history of breast cancer or ovarian cancer	Eligibility for the parent study consisted of being a woman with a personal and /or family history of breast or ovarian cancers who was referred for GCRA at the CSPPN cancer center clinic in Los Angeles County, Orange County, or Santa Barbara County and consented to participate before being seen for GCRA. The clinics serve a predominantly white insurance-covered population. The current study sample consisted of women accrued to the parent study who responded to at least one survey item assessing cancer risk communications with family members and perceived barriers to this communication before and by six months post GCRA. Of the 224 eligible women, post GCRA data were missing for 9 because 7 never presented for GCRA and 2 died. Of the remaining 215 women, 135 responded to the item assessing risk communication pre and post GCRA. Responses to the perceived barriers item were reported by 122 women at both time points. Demographics and cancer histories were similar to non participating study eligible women, to women in the parent study who did not meet eligibility for this study and to the underlying CSPPN clinic population
MacKinnon 2007	Evaluate the impact of a retreat designed to update and support BRCA1/2 carriers and their families in a rural region, through changes in health behaviours and psychological distress before and 6 months after the retreat	Individuals eligible for the questionnaire study included those participants who were carriers of a deleterious BRCA mutation or variant of uncertain significance and their biologic relatives as well as participants from high risk families and their biologic relatives. Forty-one participants from twenty two different families attended the retreat. Eleven individuals came to the retreat alone, while the remaining 30 came with at least one other family member. Eighty percent of participants were women and one half of participant had a prior history of cancer. Fifty-four percent of participants were carriers of a known BRCA mutation. Fourteen of these had a prior history of cancer, while eight were unaffected with cancer at the time of the retreat. Twelve participants had not undergone genetic testing at the time of the retreat and	Patient from the Familial Cancer Program - a multidisciplinary healthcare service coordinated by the Vermont cancer center in alliance with the University of Vermont College of Medicine and Fletcher Allen Health Care. The program provides services in medical oncology, genetic counseling, molecular diagnostics, pathology, and psychology, genetic counseling and testing services to the entire state of Vermont and seven counties in upstate New York. Individuals who underwent genetic counseling and testing through the Familial Cancer Program of the Vermont Cancer Center and had at least one family member who had been identified as carrying a BRCA1 or BRCA2 mutation were invited to a one day retreat,. Individuals with variants of uncertain significance in the BRCA genes and high risk families in which no known BRCA mutation identified but a family history consistent with hereditary breast and ovarian cancer were also invited. The cancer genetic program at Dartmouth Hitchcock medical enter

Author	Study Objective	Participants	Recruitment
		for eleven of these individuals, a BRCA mutation had been identified in another family member. Two participants had undergone testing fro the BRCA mutation in their family and were found to be negative. One participant had undergone testing fro BRCA1/2 and no deleterious BRCA mutation had been identified, but her family history was suggestive of hereditary breast and ovarian cancer. Thirty seven of the 41 retreat participants were eligible for the questionnaire study. Thirty four out of 37 completed the baseline questionnaire for a 92% response rate. Twenty eight individuals completed both the baseline and follow up questionnaire for an overall response rate of 76%. Of those that completed the baseline questionnaire, that average age was 48.	was provided invitations for the BRCA positive families as well. Approximately 80 individuals were invited; each individual was sent three additional invitations to distribute to their family members.
McGivern 2004	To provide additional information about family communication of BRCA1 and BRCA2 genetic test results. -Examine communication with nieces, nephews and grandchildren. - To describe the motivations, barriers, method and content of communication. -To examine gender differences	- Only included women with a deleterious BRCA ½ genetic test result. - respondents did not differ from non-respondents on race, religion, education, age, time since testing, whether the woman had cancer or household income. -Mean age was 48.1 years, median was 47 years, and ages ranged from 23-77 years. -An average of 2.4 years had passed since receiving the test results with the time interval ranging from 5-72 months. - Majority of women were Caucasian, married and had a college degree. -79% were affected with cancer and 21% were not. - 76% had the BRCA1 mutation and 18% had the BRCA2 mutation. 5 had both.	- Women mutation carriers were identified from the Cincinnati children's hospital medical center hereditary cancer program patient database since 1996. - Eligible female participants had all obtained genetic testing for BRCA1/2 through the HCP and had been found to carry a deleterious mutation associated with an increased risk for developing cancer. Eligible participants were the first members of their families to undergo genetic testing.
Mellon 2006	-To explore participant's knowledge of cancer risk 2) to explore the perceptions and concerns of women and a family member regarding inherited cancer risk information 3) explore communication patterns within the family regarding inherited cancer risk	- only women who had breast and/or ovarian cancer and their first or second degree unaffected female relative -three different types of focus groups were utilized 1) cancer survivors and family members together 2) cancer survivors only and 3) family members only. - 9 focus groups were conducted (20 cancer survivors and 19 family members; two related survivors brought one member). Five focus groups (n=21)consisted of both survivors and their family	Sample selection included risk assessment criteria used by genetic professionals to identify at risk families. Criteria for eligibility included women: 1) with a diagnosis of breast and/or ovarian cancer 2) over 18 years of age 3) determined to be at possible increased inherited risk based on the following criteria: breast cancer diagnosis before age 50, or ovarian cancer at any age, or two primaries (breast and ovarian cancer or two breast primaries) and a family history of one other family member with breast or ovarian cancer diagnosed at any age 4) alter, oriented and able to understand and speak English and 5) have an unaffected

Author	Study Objective	Participants	Recruitment
	information and 4) identify potential factors that may affect their decision making about seeking inherited cancer risk information.	members, two groups of only cancer survivors (n=9) and two groups of only family members (n=9) - the majority of family members in the study were either sisters or daughters with other relationships also represented (mother, niece, granddaughters). - of the 19 dyads (n=39) there were 27 Caucasian and 12 African American women. - Of the group of survivors, 18 had breast cancer and two had ovarian cancer. - All cancer diagnoses were between 1994 and 2001 and only the two women with ovarian cancer were still in treatment. - Two women had experienced a recurrence (one breast and one ovarian) - The survivors reported undergoing a variety of treatments, including surgery, chemotherapy, radiation, or a combination of two or more treatment modalities.	female relative also willing to participate. Survivors were asked to invite a female relative of their choice to participate in the study providing she met the following criteria: 1) either first or second degree relative (mother, daughter, sister, aunt, grandmother, niece) unaffected by a cancer diagnosis 2) over 18 years of age 3) alert, oriented and able to understand and speak English Recruited through 1) cancer center clinic 2) breast and ovarian cancer support groups 3) network of cancer center community outreach office and 4) surveillance, epidemiology and end results cancer registry database in southeaster Michigan. Flyers, mailing were provided to contact persons at the clinic, support groups and community outreach sites. Recruitment though the SEER dataset consisted of obtaining contact information on African-American and Caucasian women who met eligibility requirements in the years 1997-2002 and contacting the physician of record for approval to contact patient.
Mellon 2007	Explore who survivors and unaffected female relatives would most likely talk to about inherited cancer risk and reasons for selection of these individuals	146 breast and/or ovarian cancer survivors and 146 unaffected female relatives	Participants were randomly selected from the National Cancer Institute Surveillance Epidemiology and End Results Cancer Registry in southeastern Michigan.
Mesters 2005	Investigate people's perspective regarding informing one's biological family on the hereditary predisposition for HNPCC	-8 males and 22 females -Average age was 53 years (15-69). -Eleven respondents had confirmed carriers status	Recruitment from the registration database of the Netherlands Foundation for the Detection of Hereditary Tumors. A STOET employee randomly selected patients to contact for interest in participation.
Riedijk 2005	1) assess which reasons non participants had in their decision not to be tested and the relative importance of each of these reasons 2) assess anxiety levels amongst non participants and how anxiety was related to the motives for non-participation 3) assess the relevance of family	- median age was 39.9 (SD=11.4) ranging from 18-85 years. -Half of the participants were male (34). - All met the inclusion criterion of knowing that the genetic test for p16-Leiden was available to them. - 39 respondents had completed high school and 24 had completed post high school secondary or vocational education -Forty-eight were married and 38 had children - 41 respondents attended the pigmented lesions clinic.	-Department of dermatology of the LUMC provided the addresses for 184 at risk individuals for melanoma in their database that went for counseling and testing there. -They contacted the remaining 328 at risk individuals and invited them to participate in the study. 93 individuals did not respond at all, 64 indicated that they did not seek testing because their parent had been tested negative and 29 indicated they waited for their parent to be tested first, 6 they did not have correct addresses, 3 were deceased, 3 indicted they were currently being tested or about to be tested 3 said they were not part of p16-Leiden families and 2 were traveling abroad.

Author	Study Objective	Participants	Recruitment
	dynamics, 4) Assess the relationship between risk knowledge and understanding of causes of hereditary melanoma and the decision to be tested.		- The final sample consisted of 66 respondents
Schroy 2005	1) assess knowledge about familial risk among newly diagnosed adenoma patients 2) define the relationship between awareness and risk communication and 3) delineate responsibilities for communicating risk to FDRs. -Assess knowledge about familial risk and risk communication among colorectal adenoma patients.	-54 were from Boston Medical centre and 17 were from Harvard Vanguard Medical Associates - ages 39-59 median 54 years - 69% males -65% non-white Hispanic -The number of responders per participating endoscopist ranged from 1 – 21 (median 4) -Indications for colonoscopy included: screening (24%), family history of colorectal cancer (23%), polyp at screening sigmoidoscopy (21%), positive fecal occult blood testing (14%), rectal bleeding (8%), personal history of colorectal cancer or polyps (7%) or symptoms other than bleeding (3%)	-Consecutive sample of patients, <60 years of age with colorectal adenomas diagnosed between march 1999-march 2000 at the Boston Medical Centre, an urban university based hospital or Harvard Vanguard Medical Associates -Potential subjects were identified from each institution's pathology database and contacted by mail within 6-12 weeks of their colonoscopy.
Segal 2004	To research disclosure process in BRCA1/2 positive families	All women were carriers, all women had children, all of the carriers were from different families. - 26 came from Ontario and 5 were from Melbourne, Australia -All of the carriers were from different families. Twelve carriers received a semi-structured questionnaire by mail and nineteen carriers were administered their questionnaires by telephone. The mean age of the participants was 47.7 (34-59) and the mean age of the disclosed group of 50.9 (6.6) was significantly older than the non-disclosed group of 43.3 (4.6) -Twenty six of the women were married, three were separated or divorced, one was widowed and one was a single mother. -Seventeen carriers had previously diagnosed with cancer, thirteen carriers were healthy and the proportion of affected and healthy carriers did no differ between disclosed and non disclosed. -Fourteen carriers completed high school, five carriers	- In the first phase of the study, data was collected through semi structured interviews conducted with eight mothers who carried mutations in the BRCA gene, recruited though the Familial Breast Cancer Research Unit at the Sunnybrook and Women's College Health Sciences Centre in Toronto, Canada. This was used to design the semi-structured questionnaire used in this study. -Convenience sample of maternal BRCA1/2 carriers who had participated in a group therapy study designed for female carriers from the Familial Breast Cancer Research Unit at the Sunnybrook and Women's College Health Sciences Centre in Toronto

Author	Study Objective	Participants	Recruitment
		completed college, seven carriers completed their undergraduate degrees, and three completed their postgraduate degree. -Ethnicity was similar across groups with 2/3 of Anglo-Saxon origin and 1/3 of Ashkenazi origin -Twenty participants had two children, five participants had three children, five participants had one child, and one participant was an aunt who has one niece and one nephew. The mean age of the children was 18.6 years (4-38). Eight carriers have only sons, nine carriers have only daughters, and fourteen carriers have both sons and daughters.	
Taylor 2005	Investigated a range of attitudes and perceptions of men and women who were at risk for Huntington disease. Looking at gender differences for and explanation for differential uptake of predictive genetic tests – there appear to be differences in how men and women perceive and respond to the genetic risk for HD.	-45 of 57 respondents were at 50% risk of HD and 5/57 were at 25% risk. -Seven had undertaken predictive testing and were excluded from the analysis -The mean and median age of all respondents was 38.8 years and 7 years, respectively.	Potential respondents were accessed via the mailing list of the local Huntington's Disease association after eliminating those known to not be at risk.
Van den Nieuwenhoff 2006	Describe the role of written information as support for the identification of carriers of mutation for late onset disorders illustrated by using Inherited high cholesterol (IHC) Describe how information packages influenced the decision of index patients to disclose genetic information to their relatives (and the quality of disclosure) and the way the packages influenced the decision of relatives to consult their family	All 16 were of Dutch origin Age ranged from 34-67 years with a mean age of 49 years. All but one index patients had children The IHC diagnosis was made 4-28 years ago (mean of 16 years). Five index patients did not know the type of their IHC, while three index patients reported having Familial hypercholesterolemia (FH) The age of relatives ranged from 18 to 65 years (mean of 42 years). Five relatives had children.	Sixteen members of the target group participated: eight index patients and eight relatives. The participants were selected from a list of the Bloodlink Foundation consisting of 113 index patients and 21 relatives who had indicated willingness to participate in research projects. Bother groups were stratified on gender and on age (below and above 45 years of age). Random samples were selected within the chosen strata. Inclusion criteria: 16 years or over, diagnosed with lCHC by a physician or being a blood relative of a diagnosed person, receipt of the information package in the last 5 months, and a thorough command of the Dutch language. Within the recruited index patients and relative, no members of the same extended family were allowed. Selected individuals were first contacted by phone. When participation was confirmed orally, participants received a complete information package by mail, accompanied by an instruction on how to perform their evaluative task.

Author	Study Objective	Participants	Recruitment
	physician for a serum cholesterol test, as a first step to genetic diagnosis.		
Van den Nieuwenhoff 2007	Describe family disclosure concerning inherited high cholesterol Determine the reason index patients inform their adult blood relatives of their risk, how disclosure occurs and what is disclosed	All participants were of Dutch origin 11 were females age ranged from 27-70 (mean age for women was 51 and for me was 49) four women and 8 men had children Six had low educational attainment (less that senior secondary vocational education Seven had attained moderate levels of education (from senior secondary vocational to pre university education) and seven had completed higher education (higher professional or university education). Eleven IPs were diagnosed with FH, three with FCH; six did not know the exact IHC variant. The length of time from their date of diagnosis ranged from 18 years to a few months. All had received documentation regarding family disclosure in the last year Apart from one father and one son, there were no family member among the interviewees	Potential participants had been diagnosed with IHC, had experience informing relatives about IHC, were 16 years or older, were fluent in Dutch Recruited through the Blood link Foundation, a Dutch patient organization for hereditary CVD, which distributes IHC information packages to adult IPs. IPs were invited to complete and return a questionnaire indicating their willingness to participate in future research and their desire to receive other pertinent information. Purposively selected 20 subjects for a total of 105 IPs who had indicated their willingness to participate. Participants were stratified according to gender, age, number of information packages requested, by occasional/spontaneous remarks concerning their difficulties with disclosure Selected IPS were informed about the study by phone and were reassured that their participation would be confidential and would not affect their medical care. If they indicated their continued willingness to participate, they received written confirmation, information abut the project, an informed consent form and a request to sketch a simple family tree prior to the interview. Of the 20 IPs originally selected, 19 agreed to participate and won was no longer interested. One stated that his father had performed the main informing role within the family. Through this participant, they contacted the father, who was willing to participate, bringing the total number of participants back to 20.
van Oostrom 2007	To investigate: 1) whether genetic testing for cancer susceptibility affects family relationships, 2) the nature of the effect on family relationships and 3) whether nuclear family functioning, differentiation from parents, communication style regarding hereditary cancer within the family and perceived familial support are predictive of adverse	-participants belonged to 96 different BRCA1/2 and HNPCC mutation families - no significant differences were found between HNPCC and BRCA1/2 participants regarding pretest genetic risk, cancer status uptake of genetic testing and demographics. - 175 participants were getting BRCA1/2 testing and 96 were getting HNPCC testing. - the BRCA1/2 group was all females and the HNPCC had 64 females and 32 males. -161 of the 175 BRCA1/2 participants had a DNA test and 92 of the 96 of the HNPCC participants had a DNA-test	Recruitment took place from January 2003 to October 2004 at the Rotterdam Family Cancer Clinic of the Erasmus MC, the Center of Human and Clinical Genetics of the Leiden University Medical Center and the Department of Medical Genetics of the University Medical Center Groningen (only individuals at risk of HNPCC). - Applicants for genetic susceptibility testing for known familial pathogenic gene mutation in BRCA1/2 or in a HNPCC related gene (MSH2, MLH1, MSH6) - aged 18 and over - Applicants were eligible for the study if they had a relative with an identified gene mutation, irrespective of cancer status and of the decision to proceed with genetic testing -excluded those not proficient in Dutch

Author	Study Objective	Participants	Recruitment
	consequences on family relationships after genetic testing.	-Twenty-one patients were affected by cancer: 19 had finished treatment six months to 28 years ago; two were diagnosed after having completed the second measurement and were excluded from the third measurement. - Mean age of those with BRCA1/2 testing was42.5 (12.2) and those with HNPCC testing was 41.0 (13.3) -61 of the 175 BRCA1/2 participants were mutation carriers, 27 of the 96 HNPCC participants were mutation carriers.	Males from BRCA1/2 mutation positive families were excluded because they are not at high risk of developing cancer themselves
Weiner 2005	–Attempts to answer how often families communicate about genes and health? And; - do race, biological sex, attitudes about response efficacy, test experience, or media exposure about genetic health issues impact family communication regarding genes and health?	- southern participants comprised 65.7% of the sample (n=471) and northern participants comprised 27.3% of the sample (n=195). -online data comprised 7.1% of the data -132 African American men, 135 African American women, 155 European American men, and 295 European American women. - the age of participants ranged from 18-73 years (m=29.58 SD=10.42) - education levels varied from less than high school (2.9%), high school diploma (16.9%), some college (26.2%), college degree (29.7%), vo-tech degree (4.2% and advanced degrees (17.6) -most people had not taken a genetics course in college (83.8%) - The majority of participants did not have children (62.2%) and 23.8% had either one or two children -11.9% of the sample had three or more children.	Data were collected in community-based setting in four geographic locations: southeastern town near a large land grant university b) southeastern, metropolitan city and a northeastern town located near a large state university and a northeastern metropolitan city. - At the southeaster locations, researchers collected data at a health fair, restaurants, churches, retailers, barbershops, beauty parlors, and airport, Laundromats, and the university - At the northeastern locations, researchers collected data at a train station, bus station, outlet mall, and business office as well as at a large, land grant university.
Adelsward 2003	- demonstrates how persons who are seeking information about their own health are dealing with doctors claims for information about others. - discusses the moral dilemmas surfacing in clinical practice.	-In 23 of the consultations, the person seeking information was a woman, in four a man and in four several family members were present. -Five of the persons seeking information were recorded twice, at a first and second consultation. - The diseases most frequently discussed (26/31) were breast, gynecological and colon cancer.	Patients who went for genetic assessment of cancer risk since 1991 in a clinic in Sweden - interested persons came in contact with the clinic through direct contact, referrals by other physicians or through a relative with either a cancer diagnosis or previously determined hereditary risk -A physician met with the person seeking information and drew up a genogram
Ayme 1993	Assess the importance of how individuals within families actually behave with respect to transmitting carrier information and non-	-289 individuals with balanced chromosomal rearrangements had 1816 at risk relatives - 808 of 1816 were karyotyped - In 52 families (18.4%) not even the parents of the index case was karyotyped.	All carriers of a balanced chromosomal abnormality ascertained in their area (clinic in Marseille) from 1975 to 1989 were included. - The reasons for the referral to cytogenetic analysis were: spontaneous abortions (35%), sterility (5.9%), abnormal child

Author	Study Objective	Participants	Recruitment
	disclosure	- In 41 families (14.5%) the exploration was completed in all the family member potentially at risk -In 63.2% of families less than half of their at risk member had been karyotyped -The chromosomal rearrangement found in the 289 index cases were Robertsonian translocation (90), reciprocal translocations (151) and inversion (48).	(48.8%), and prenatal diagnosis (10.7%). - A follow-up questionnaire was sent by mail every 2 years to all consultees - The inversion of chromosome 9, considered to be almost a normal variant, were excluded (144 cases)
Bruce 2003	To investigate the effects of presymptomatic testing for HD on the marriages and intimate relationships of 39 couples and explore the additional effect of having kept secret a family history of HD prior to marriage	Not stated	Not stated
Claes 2003	-Focus on family communication initiated by affected patients who have had a diagnostic genetic test for HBOC. -Gather systematic data on the extent to which these patients informed close and distant relatives about their personal diagnosis of cancer, the genetic test and its result. - Gain insight into the motive of patients to communicate the information. - Evaluate whether uptake of predictive testing is associated with informing other relatives or not. -Evaluate the level of knowledge of patients in order to see whether these patients have the ability to disseminate correct information when communicating with	- Mean age was 52.7 (SD=10.4) - The majority were married (78%) and had children (87%) - Half had higher education levels (48%) - Most had a person history of breast cancer (83%) - 11% had a personal history of ovarian cancer and 6% had had breast as well as ovarian cancer. - 41% (n=17) belonged to a family where a BRCA1/2 gene mutation had been detected in one or more affected relatives (conclusive) - 59% belonged to a family with an inconclusive familial genetic test results - the majority of participants were contacted directly (79%) - median time between the blood draw and the study interview was 28 months (range: 10-74 months) and the blood draw occurred between 1994 and 1999. - The median time between the notification of the test result by the genetic center and the interview was 17 months (6-29). The notification the test result had taken place between 1997 and 2000.	- Patients who had a diagnostic genetic test at their center and had a result and direct contact with the genetics clinic were sent a letter invitation to participate. - The second group had no contact with the clinic so their physician was asked permission to contact the patient and then were contacted in the same way as the direct contact group. - eligibility criteria: 1) personal history of breast and/or ovarian cancer 2) a family history of the cancers (two or more other affected relatives) 3) a diagnostic genetic test for HBOC carried out in their centre (no matter the result) 4) Dutch-speaking

Author	Study Objective	Participants	Recruitment
	relatives.		
Cox 1999	- how predictive testing candidates and their families arrive at an inter-subjectively meaningful interpretation of the experience of predictive testing - focus on patterns of communication about genetic information within the sphere of familial and social interactions.	- 102 in depth interviews with sixteen participants in predictive genetic testing, thirty-three of their family members and/or close friends and one couple where the wife was at risk but did not want to proceed with predictive testing. - The 51 participants represented seventeen different families who lived in both urban and rural settings in BC. - There were 11 spouses/partners, 3 parents, 4 sisters, 5 brothers, 1 daughter, 2 sons, 5 'other family', 2 friends, and 2 other (woman who did not want testing and her husband). Total of 35 family members, 17 were male and 18 were female -Candidate information: 12 female, and 4 males, mean age of 41.8 (23-57), 4 were single, 10 were married and 2 divorced and now living common-law - 6 had no offspring, 1 had one, 4 had two children, 3 had three children, 1 had four children, and 1 had five children. Total of 27 offspring. -Most candidates were formally fairly well educated but two had not completed high school and 10 had done vocational training, college or university. - 6 females and 4 males did not have the gene. 2 females had an intermediate allele, 4 females did have the gene.	-Predictive Testing Research Group in Medical Genetics at UBC which is affiliated with fourteen other genetics centres in Canada -participation in the predictive testing program: 1) must have confirmed family history of HD and an apriori risk of 50% or 25%, 2) be able to provide informed consent and 3) not have been given an established clinical diagnosis of HD - A community based protocol was developed to meet the needs of people who lived a big distance from the genetics centre at UBC. - recruitment occurred during the candidate's first visit to the genetics centre and the first round of interviews was schedules to occur prior to the clinical disclosure of results. - All eligible candidates attending their first counseling session at the local genetics clinic during the Nov. 1992-Sept 1994 were invited to participate. Where feasible, candidates were encouraged to invite their spouse or partner and unaffected parent. Siblings and other relatives and/or close friends were also invited to participant - It was up to the candidate to decide which family members they wished to invite to participate and in no case were family members interviewed in the absence of the PT candidate's participation in the study.
d'Agincourt Canning 2001	To examine how does gender intersects with moral responsibility and the duty to disclose genetic test results to others. - who takes primary responsibility for disclosure and communication of genetic information within the family? - is this a gendered activity? -Does disclosure of genetic risk information impose particular burdens or costs, and if so, what forms might	- 31 participants were women, 5 participants were men	Recruited through the genetic counseling center at the Cancer Agency, Vancouver where they had been referred to receive genetic counseling for hereditary breast or ovarian cancer.

Author	Study Objective	Participants	Recruitment
	these take? Is choice constrained by the sharing of genetic information or does this task help women/men fulfill their responsibilities within particular relationships and better cope with cancer risk?		
Denayer 1992a	- To evaluate the transfer of information about the hereditary aspect of CF from CF parents to their relatives. - Do relatives make use of this information when confronted with decisions about pregnancy planning, - are they are informed or not about the new possibility of carrier identification and how do they feel about it.	-32 families - Patients age ranged from six months to 18 years, with 41% in the age group 10-15 years, 25% in 5-10years, 7% in 15-20 years and 4% in 0-5 years. - health situation covered the entire range. The modified Shwachmann scores had a mean value of 70 with 47% of the patients between 80 and 95. In four of the CF families there were two CF patients, while in three other families a second (or first) patient had already deceased. In one family a nephew of the patient had CF. - The families could be very restricted or very large (1-16 relatives). -54% were aunts and 46% were uncles. They were recruited as often from the maternal family as from the paternal family. - The age of the respondents vried between 20 years and 58 years, with a mean of 38 years. The distribution of educational levels and religious convictions seemed to be comparable to the data for the general population. A majority (88%) was married, while 12% was single. One fourth of the study population (25%) had almost daily contacts with the CF patient while 16% had weekly contacts. One third saw him one or two times a month and 23% saw him less than monthly. Three percent had almost no contact with the CF patient. - Almost 86% of the respondents said they gave, at least occasionally, some kind of practical help to the CF-family while 26% of the total group did so rather frequently.	- Study was set up in close collaboration with the pediatrics department of the University Hospital of Leuven. The parents of all 46 CF patients who attended the clinic in the period May-December were asked to cooperate and give the addresses of their brothers and sisters. -Only six of them immediately refused to cooperate. Eight other families intended to cooperate but did not return the names addresses of their relatives. The received the list with the addresses of aunts and uncles belonging to 32 families.
Denayer 1992b	Aims at evaluating whether the group of relatives aged	-27 families -55 aunts of CF patients and 54 uncles of CF patients.	- Study was set up in close collaboration with the pediatrics department of the University Hospital of Leuven. The parents of

Author	Study Objective	Participants	Recruitment
	less than 40 years makes use of information about genetic transmission of CF when confronted with decisions about pregnancy planning, how they evaluate the burden of raising a CF child, whether they are informed or not about the new possibilities of carrier detection and prenatal diagnosis, and whether they intend to use them.	- Ages of respondents ranged from 20-40 years (20-25 years =7%, 26-30 years=25%, 31-35 years =33%, 36-40 years = 35%) - 78% were married, 22% were single divorced or widowed. - the distribution of educational levels and religious convictions was comparable to the data for the general population: 15% had primary school, 27% had less than high school, 35% had high school, 19% had greater than high school, and 4% had a university degree. - 76% had children – 125 in total. One fourth of these children were less than five years old, 44% were 5-10 years old, 23% were between 11 and 15 years old and 7% were older than 15 years. 20% of responders expressed the intention to have more children in the future, 18% were still undecided and the others did not want anymore children. - the patients age ranged from 6 months to 18 years, with 37% in the age group 10-15 years. - health situation covered the entire rand, the modified Shwachman scores, had a mean value of 67 (SD=20.2) with 44% of the patients between 80 and 95. One fourth of the study population had almost daily contacts with the CF-patient while 16% had weekly contacts. One third saw him one or two times a month and 23% saw him less than monthly. Three percent had almost no contact with the CF patients. 86% of the respondents said they gave some kind of practical help to the CF family -26% did so frequently	all 46 CF patients who attended the clinic in the period May-December were asked to cooperate. 32 families agreed to give the addressed of their brothers and sisters. - Only used respondents who were less than 40 for the data analysis. Same sample as Denayer 1992 above.
Duster 1999	-To reveal cultural and social-structural variations in perspectives - to compare the variable penetration and meaning of genetic information in two populations differentiated by	- no description given	Study started in summer of 1992 in clinical setting, but fanned out to support groups and advocacy organizations servicing individuals and families with a special interest or concern about either sickle cell anemia or cystic fibrosis. Interviews were conducted with men and women who have, or who have had, a persons with one of these conditions in the family.

Author	Study Objective	Participants	Recruitment
	the socially designated categories of 'race' - address who families in which genetic risk is known to exist respond to information about this risk		
Fanos 1995	To identify factors motivating or interfering with the pursuit of carrier testing adult CF siblings.	- 54 CF siblings and 30 spouses - all Caucasian - participants age ranged from 18 to 55 - 26 brothers and 28 sisters - Twelve of the sibs were single, 40 were married, 2 were divorced - Median number of children was 1 - median household income was $60,000 - median education for both siblings and their spouses was a college degree - of the 54, 45 had been tested and 9 had not been tested at the time of the interview	- All adult CF siblings and their spouses who had been tested for CF carrier status between 1987-1992 through the genetics division at the Children's Hospital, Oakland - Participants were contacted by the clinic that had referred their patients for testing to the hospital. - 7 other respondents were provided by Children's Hospital, Boston.
Fitzpatrick 1990	To compare the patterns of communication, and the use of professional support systems in Irish and American families with sons with DMD	- 34 Irish boys - 23 American boys	- The parents of boys with Duchenne muscular dystrophy were approached. - The patients were attending the muscular dystrophy services of the central remedial clinic Dublin, and the children's hospital Boston. Families from Boston were selected to match as closely as possible for social class, age, ambulatory status and presence of learning disability in the sons from Ireland. - Boys and their parents were interviewed separately - Control groups were selected from each culture because seeking professional support is likely to be culturally biases irrespective of the presence of chronic illness. The Irish control group was obtained from the school registers of a primary and secondary school in the vicinity of the Central Remedial Clinic. The American control group could not be obtained in the same way because the study was carried out during the school vacation period so it was obtained from children attending the Comprehensive Child Health Programme and the Sports Medicine Clinic at Children's Hospital, Boston and comprised of health boys attending for routine pediatric checkups or with minor sports injuries. The boys in both control groups were selected to match the DMD boys in their culture for age and social class.
Forrest 2003	Explore the barriers and	- 16 HD participants	- Participants were drawn from the patient population of the

Author	Study Objective	Participants	Recruitment
	facilitators in family communication about genetic risk. - explore and synthesize the factors which influence whether patients share information with relatives about late-onset disease and with whom they share it.	- 21 HBOC participants - 19 partners were interviewed (12 HD, 7 HBOC) but 7 separately from their partners -No male relatives from any of the HBOC families had recently attended the genetic clinic so this subgroup was not available to approach - Of the 16 HD participants , 11 had spouses, eight of whom took part. Of the additional Five HD partners who had been invited, four decided to take part. -In the HBOC group on one with a negative presymptomatic predictive test responded to the invitation. -Mean age in the HD group was 46.8 (26-65), and mean age in th HBOC group was 40.5 (25-62). - In the HD group there were 8 males and 8 females and in the HBOC group there were 21 females. - All participants described their ethnicity as white - in the HD group, 69% lived with their partner or were married , 69% were parents of children, 94% had siblings, 25% had post secondary education and 62.5% were in full or part time employment - In the HBOC group, 86% were married or living with a partner, 81% were parents of children, 95% had siblings, 62% had post secondary education and 90% were in part of full time employment.	regional genetic service in Aberdeen. Patients were identified from clinic records of those who had attended for predictive testing for HD and those who had sought advice about a family history of HBOC. A personal letter of invitation was sent by a clinician between 2000 and 2001. Purposive sampling and theoretical sampling to ensure a wide variety of participants and experiences. - It was asked whether partners could be interviewed separately
Green 1997	Attempt to address the following questions, drawing on a prospective study of women attending a genetic counseling clinic because of a family history of HBOC: With whom do probands communicate? What determines whether information is passed on? What do probands see as their obligations?	-All Caucasian - 42 married, 3 divorced and one not married - 12 had worked in a profession with some medical connection - 23 (52%) had completed full-time education by the age of 16, 11 (25%) had further education up to the age of 18, and the remaining ten (23%) were educated beyond age 18. - 46 women reported a total of 132 relatives affected by breast or ovarian cancer (mean 2.9, 1-8) and a further 77 affected by other cancers - only one women reported no maternal relatives with breast or ovarian caner. - 13 women reported cancers in paternal relatives and only four reported breast or ovarian cancer in paternal relatives.	Sample was recruited from the cancer family history clinic in Cambridge UK. Between Feb 1994-Feb 1995 anyone who was offered an appointment to discuss breast, ovarian or both cancers, but who did not have cancer themselves was invited to take part. - consent forms were sent with a letter explaining the research two weeks before the clinic appointment. Women were only contacted directly by the researchers when the form had been returned.

Author	Study Objective	Participants	Recruitment
		- Most commonly reported relatives affected by breast or ovarian cancer were mother, sisters, maternal grandmother and maternal aunt. - The majority of the sample did not have living mothers: 14 had died of ovarian cancer, 8 of breast and 5 from other causes. - Women whose mothers were still alive were slightly younger than women whose mothers had died of breast or ovarian cancer - Women whose mothers had died of other causes were considerably older - Of the 19 surviving mothers, five had themselves had breast cancer and five had had ovarian. Six were well but four were terminally ill. - Four women had no living siblings - Thirty had at least one living sister and 30 had at least one living brother. Six women had no children and seven were pregnant with their first child -Of the other 39 – 31 had at least one daughter and 29 at least one son. -Most daughters were still children but 16 were over 16 years of age.	
Hallowell 2003	To describe how women, previously diagnosed with breast/ovarian cancer, perceive their role in generating genetic information about themselves and their families	-Ten women were BRCA1/2 mutation carriers, 12 had not been found to carry a known BRCA1/2 mutation and 8 were awaiting results. - Median age was 54 (39-71) - Twenty seven women had previously been treated for beast cancer, two for ovarian cancer and one for an unspecified gynecological cancer - Time since the most recent diagnosis of cancer ranged between six months and 31 years (median five years) - 23 women had children - 12 were educated until the age of sixteen, six until they were eighteen, four had further education or professional qualifications and ten had a graduate or postgraduate degree. - Three women had worked in a medically related occupation at some time in their lives -18 women had a maternal family history of breast or ovarian cancer	Participants were recruited by a genetic nurse specialist when they attended the cancer genetic clinic or by letter from the consultant in charge. Data collection occurred between November 2000 and June 2001.

Author	Study Objective	Participants	Recruitment
		- Seven had a paternal family history and in five cases it was not clear whether the mutation had been inherited by the paternal or maternal line - A total of 119 relatives were reported as affected with cancer (range 1-10, median four per family) - 26 women had at least one first degree relative affected with either breast, ovarian, endometrial, prostate cancer.	
Henneman 2002	- to investigate the personal experiences of carrier couples identified by testing in CF families before the birth of an affected child in order to 1) explore their experiences with genetic carrier testing 2) explore the effects of prospective risk on reproductive plans and family relations and 3) to assess whether improvements are necessary in the provision of genetic counseling and services for these couples.	- All carrier couples were identified two to eight years before they were contacted to participate in the study. - in seven of the couples , one partner had a family history of CF and in two of the couples both partners had CF in their family - at the time of the interview, the women ranged in age from 28-36 years, with a mean age of 33 years. The mean age of the men was 34.3 years (30-38). - Only one couple went to church regularly	CF carrier couples who went to the DNA Diagnostic Laboratories of Groningen and Rotterdam in the Netherlands in 1997 were prospectively identified by medical records. All couples requested DNA testing due to a positive family history. The couples were contacted by their clinical geneticist and asked to participate in the study. Interviews were held between 1998 and 1999. Partners were interviewed together.
Hughes 2002	-describe the process and content of communicating BRCA1/2 test results to sisters, and to evaluate whether the proband's carrier status influenced communication outcomes. - describe motivations for communicating or not communicating BRCA1/2 test results to sisters and to describe specific topics that were discussed -to evaluate whether the carrier status of the proband had an influence on their	-female probands who received BRCA1/2 test results and completed the family communication questionnaire during the one-month follow-up telephone interview. - All probands were women and affected with breast or ovarian cancer - 63% of probands were less than 50 years of age and 74% were married. 77% were employed, and 74% were college graduates. 56% had household incomes of $750000 or higher. Thirty-seven percent of probands were BRCA1/2 mutation carriers and 64% received uninformative test results. - All probands had at least one sister Probands reported communication outcomes for a total of 81 sisters and on average, had 2 sisters. The mean age of sisters was 49 (SD=13.5) and 16% of	Participants were adult women who received BRCA1/2 test results through the genetic testing research programs located at the Georgetown university medical center in Washington, DC and the women's college hospital in Toronto, Canada. - To be eligible to participate in these programs, participants had to have a minimum 10-20% prior probability of having a BRCA1/2 mutation. All participants in the study were the first index family member affected with breast or ovarian cancer to undergo genetic testing and receive BRCA1/2 test results. Participants had to of had at least one sister. -After referral to the genetic testing research programs at the medical centers participants were contacted by telephone to determine eligibility using a phone interviews. Eligible participants were invited to participate in a pretest education and counseling session. Then were offered to give a blood sample for testing and underwent their counseling procedures. 1 month post

Author	Study Objective	Participants	Recruitment
	communication motivations and content.	them had a personal history of breast or ovarian cancer.	test they were administered a telephone interview.
Julian-Reynier 1996	- document the attitudes towards breast cancer predictive testing of women with a first degree relative with cancer and towards the transmission of information about the cancer risk to their relatives	- 124 healthy women and 85 women with cancer - Among the cancer patients 89.4% (n=76) had breast cancer alone. 5.9% had cancer of the breast and other sites (bowel, ovary, multiple) - 4.7% had ovarian cancer alone - Mean age was 47.8 years (SD=11.9), mean age of the cancer patients was significantly higher (53.4 SD=10.8) than healthy patients (44.2, SD 12.0) - 128 of the 209 had less than or a high school education. 180 of the 209 had children. - The oncogeneticists stated that in 78% (n=163) of all the cases, a genetic risk to cancer was likely to be in the families - 13.9% were unlikely to have a genetic risk of cancer and in 8.1% the oncogeneticist could not determine whether the risk was present or not.	- Six participating regional cancer centers were selected : Marseille, Toulouse, Lille, Nantes, Paris/St Cloud, Clermont-Ferrand) - inclusion criteria: presence of breast cancer running in the family, either in the patient or in a at least one first degree relative when she was not affected herself. All the healthy cases included had at least one first degree relative with breast cancer. All the affected ones had a first degree relative with cancer and when the consultand herself was not affected she had at least one first degree relative who was. -Investigators recruited female cancer patients and healthy patients who were attending the cancer genetic clinics for the first time between Jan 1994 and Jan 1995 - participants were mailed a standardized questionnaire the week after the consultation. A reminder was send after two weeks then a complete package.
Julian-Reynier 2000	-to describe women's theoretical attitudes towards family disclosure of breast cancer genetic tests results and to investigate the factors (medical, familial, socio-demographic, and psychological) related to the diffusion patterns observed. - report on the a priori feelings of a group of patients as to whether or not they would inform their first-degree relatives about eh results of their genetic tests if they turned out to be positive	- 211 healthy clients and 187 patients affected by cancer - Mean age was 43.6 years but the mean age of the cancer patients (47.9, SD=11.7) was significantly higher than mean age of healthy clients (39.8, SD=11.4). - 133 (71.1%) of cancer patients had a genetic risk running in the family, 160 (75.8%) of healthy participants had a genetic risk running in the family. - 113 (65.7%) of the cancer participants had been very much affected by caner in a close friend or relation while 157 (79.3%) of the healthy participants had been very much affected by cancer in a friend or close relative.	- Five French cancer genetic clinics participated. Each of the clinics deals with at least 60 cancer genetic clients every year. -Only women were included in the study and the criterion used to select the participants was having breast/ovarian cancer running in their family, either in the patient herself or in at least one first or second degree relative. All participants had to of attended the cancer genetic clinics between January and December 1996 before any biological tests had been carried out.
Koehly 2002	1) describe the composition of familial networks 2) characterize the patterns of family functioning and genetic counseling and testing discussions within the	- the 36 members were from 5 extended families who had previously been found to carry HNPCC-predisposing mutations. 58% of these were female. - family members included those who had been diagnosed with HNPCC syndrome caner, unaffected individuals who are at risk of carrying a mutation ,	-Colorectal cancer patients at the University of Texas MD Anderson Cancer center were invited to participate in a research protocol that offered genetic testing and counseling for HNPCC. When patients were identified as mutation carriers, their at-risk relatives were invited to participate. Individuals were asked to participate in the genetic testing protocol were also invited to

Author	Study Objective	Participants	Recruitment
	families. 3) how these patterns, or the familial culture were related to discussing genetic counseling and testing among family members. 4) defined the characteristics of individuals may assume responsibility in educating family members about their genetic risk, reminding family members to comply with high risk screening recommendations and convincing family members to participate in genetic counseling and testing.	and their spouses (35% of all respondents were spouses). - 27 relatives and 9 spouses took part (36) - the sample included those who had undergone genetic counseling and testing as well as those who had not. The five families are referred to as Family A, Family B, Family C1, Family C2, and family D. Family C2 and C1 represent two separate by related families. - All respondents from family B share biological ties with each other, and between 36% and 67% of the relationships among respondents from other families have a biological link - On average, each respondent has approximately 21.7 individuals named as family, 20% of whom are not based on biological or marital connections but rather are friends - The respondents family networks were family balanced in terms of gender distribution except for one family (C2).	participate in a concurrent longitudinal study of psychosocial aspects of HNPCC genetic counseling and testing. HNPCC mutation-positive families were identified for the qualitative study if a) the proband had participated in the longitudinal psychosocial study and b) the family included at least five living members estimated to be at 50% risk of carrying a mutation who were at least 18 years of age and could speak English. Invitations to participate in the longitudinal study were mailed to 63 biological relatives and 31 spouses whom they were given permission to contact. Of those invited, 45 biological relatives and 11 spouses had participated in the long study. 27 relatives and 9 spouses agreed to take part in the telephone interviews.
Lerman 1998	- Addresses family disclosure of information about genetic testing for cancer susceptibility. - present data on the determinants and outcomes of disclosure of BRCA1/2 genetic information within hereditary breast cancer families.	None given	Study 1: prospective cohort study. - Male and female members of hereditary breast cancer families who participated in early genetic linkage studies contributing to the isolation of the BRCA1/2 genes. Letters of introduction were mailed to family members to inform them that their breast cancer susceptibility gene in their family had been identified and that genetic counseling and testing are available. - Consenting family members were asked to participate in a baseline telephone interview to assess demographic characteristics, risk factors, and psychosocial well being. Individuals interested in genetic counseling and testing had the opportunity to participate in a pre-test education session. Most of these were conducted with the extended family. - Those who elected to receive their BRCA1/2 test results did so after completing additional written consent forms and participating in individual genetic counseling. - They followed mutation carriers, non-carriers and decliners of testing for one year - the data on family communication is based on the one month follow up assessment. Took place Creighton University and was family-based study.

Author	Study Objective	Participants	Recruitment
			Study 2: prospective cohort study. - clinic-based study conducted at the Lombardi Cancer Center at Georgetown University medical centre. Study design similar to study 1 but the testing process flows through the initial proband who is the gateway for providing access to other family members. All counseling and testing is conducted on an individual, rather than family basis.
Miesfeldt 2003	- Examine women's concerns regarding their children's risk for disease, perceptions of their children's concerns, and beliefs regarding whether, at what age and how children should be informed about issues regarding potential hereditary breast cancer risk.	Women from Virginia who had been diagnosed with breast cancer prior to age 50 between 1994 and 1997. Women were categories into two groups: women with suspected hereditary breast cancer and women with presumed sporadic breast cancer. -90% of the sample was Caucasian - 7% had some high school education, 23% had graduated high school, 20% had some college and 6% had technical school graduate, 27% were college graduates and 18% had a graduate degree. -77% of the same were married, 14% separated, 3% divorced, 6% single - 87% of participants had one or more children and 13% had none. - 18% had income between 20,000 and 39 999, 20% between 40K and 59999, 18% were between 60k and 79999 and 29% were greater than 80 000.	- Participants were recruited from all 71 Virginia medical facilities reporting breast cancer cases in women under age 50 to the Virginia Cancer Registry between 1994 and 1997. - women were contacted for participated by the individual hospitals where they were diagnosed. This resulted in a two step sample recruitment process. Step one involved hospital recruitment. A detailed information packet describing the study and requesting the hospitals participating in recruiting participants was sent by our study coordinator to registry representatives at the 71 medical facilities. Of those nearly half (34) of the hospitals agreed to assist. Virginia cancer registry staff then sent each participating medical facility a list of the names of its breast cancer cases diagnosed at age less than 50 years and enrolled in the registry between 1994 and 1997. - Step 2 of recruitment involved registry personnel at each medical facilities sending an informed consent and family history questionnaire to all 2959 women with breast cancer that the facility had enrolled in the Virginia cancer registry. Women were asked to send the informed consent and completed family history questionnaire back to the medical facility. Each medical facility compiled the signed informed consent and completed family history questionnaire received from 314 women who agreed to participate in the study. Each was assigned a study code that as placed on her informed consent and completed family history questionnaire when received from the hospital. All 314 participated were subsequently mailed the Knowledge, attitudes and beliefs questionnaire.
Ormond 2003	- Addresses whether the presence of a family history of CF affects carrier status information dissemination patterns and the uptake of carrier testing in at-risk relatives, and which factors	- 30 participants in group A (parents of children with CF), 18 surveys were returned from group B for a total of 48 valid surveys. - No significant difference between the two groups with regard to age, gender, or percent married. - 37.4 was the average age of the total population. 65% of the sample was female, 92% of the sample	- Group A was obligate carriers identified through a CF clinic population at Rush Presbyterian St. Luke's medical center in Chicago. Both mothers and fathers of with children ages 2-10 were recruited. - Fifty families were included in this study population, and assuming intact families, questionnaires were mailed for both parents to complete individually. These families had been

Author	Study Objective	Participants	Recruitment
	facilitate or hinder information dissemination.	were married or in a committed relationship. - there were significant differences between the two groups with regard to religious affiliation and education. - 74% of the participants in group A had college or postgraduate education while 100% of group B did. 85 % of the total population had college or postgraduate education. - Living relatives of study participants included at total of 287 first-degree relatives (71 parents, 110 siblings, 12 half-siblings, 94 children), 186 second-degree relatives and 529 third-degree relatives.	counseled about the reproductive risks to themselves and their relatives by a pulmonologist or by a genetic counselor associated with the CF clinic. - Group B was CF carriers identified through general population screening at Northwestern Memorial Hospital in Chicago between Jan 1998-August 2000. - NMS offered CF screening to all Caucasian prenatal patients seen for genetic counseling to all patients requesting CF screening and individuals of Ashkenazi Jewish ancestry. All patients seen for carrier screening received a short genetic counseling session prior to screening and a follow-up letter with their results. Those who were determined to be carriers were also counseled about the reproductive risks to themselves and to relatives and received a generic letter to share with family members regarding their carrier risk and the availability of screening. Between Jan 1998 and Aug 2000 approximately 250 individuals were screened for CF and 850 were screened for the AJ panel.
Peterson 2003	- describe how information about the identification of an HNPCC gene mutation was disseminated in 5 families - when and under what circumstances this information was shared -how family members reacted to and acted on this information. - what facilitates and inhibits family communication and individual behaviour with regard to HNPCC genetic counseling and testing	-39 members of five families representing 30 separate house holds -Of the 29 biological relatives 27 had counseling, 25 donated blood and 23 received the test -Thirteen of those who received results were mutation carriers -For those participants who had undergone genetic testing at the time of the interview the median length of time between receipt of their test results and their interview was 15 months (1-44). -Three probands were men and two were women. -Of the spouses who were interviewed seven were married to mutation carriers. -None of the families had participated in cancer genetics research prior to the study. -Non-participants were more likely to be male and less likely to have completed genetic counsel ling, testing or other interviews. - Family 1: had three generations represented with 7 females, 4 who completed college, 6 biologically related family members, 2 with cancer history, 1 who was never married or divorced, Family 2: 2 generations represented, 5 females, 3 completed college, 8 biologically related, 1 with a cancer	-This study was part of a multiphase study of psychosocial aspects of genetic counseling and testing for HNPCC which offered free genetic counseling and testing for HNPCC. -they identified five families who had a t least one member recruited to participate in the multiphase psychosocial study and who met the following criteria: the family included the proband who had previously undergone genetic counseling and testing through the clinical research protocol, and the family included at least five members estimated to be at 50% risk of carrying a mutation who were at least 18 years old and could speak and read English. -Among the five families, they had permission to contact 63 biological relatives (including probands and 31 of their spouses. - Of the 63 biological relatives they invited to participate 29 agreed to take part, of the 31 spouses invited, 10 agreed to take part -Letters of invitation were sent to probands, at risk relatives and spouses. Phone calls were made to provide additional information to participants.

Author	Study Objective	Participants	Recruitment
		syndrome history, and 3 were never married or divorced. Family 3: represented 1 generation, had three females, 6 who completed college, 6 biologically related members and 4 who where never married or divorced. Family 4: had two generations represented, three females, 1 who completed college, 3 who were biologically related, 1 who had a cancer syndrome history, and Family 5: represented 2 generations with six females, 0 completed college, 6 were biologically related, 2 had a history of cancer, 1 was never married or divorced.	
Shakespeare (1993)	Not stated	- 24 at risk and 8 not at risk members of HD families	Not stated
Skirton (1998)	-study the process of disclosure of risk information from parents to their children to examine the difficulties surrounding this area	- 6 males and 9 females - 8 at risk, 5 spouse, 2 affected - Twelve subjects were parents who had adolescent or younger children at risk of Huntington's disease. -Three subjects were adults at risk of this condition who had a parent in the cohort. -Mean age of participants was 41.7 years (22-66)	-Subjects were selected from families affected by Huntington's Disease. -The sample of 15 was chosen to include a range of subjects with respect to age, disease status, gender and social class.
Smith 2002	-Report the probabilities that individuals tested for a BRCA1 gene mutation communicate their genetic test results to sisters, brothers, daughters, sons, co-workers, health insurance providers.	- All Caucasian - 62% were female and 38% were male - Mean age was 44 - Mean education level was 14 years of schooling - Median household income was 37500 - 56% of the women were employed and 77% of the men were employed. - 29% of the females and 28% of the males tested positive for the BRCA1 gene mutation.	Data collected from kindred which comprised 120 nuclear families. Kindred members have an elevate risk of carrying a specific BRCA1 gene mutation. - Adult women and men were recruited into a longitudinal study regarding the behavioural and psychosocial consequences of genetic testing. If the individual agreed to participate, a baseline survey was done. Then the individual met with genetic counselors at the University of Utah Health Sciences Center to discuss the advantages and disadvantages of being tested for the BRCA1 gene mutation. They then had the test and chose whether to receive their test, and did a post test phone interview one week after learning results and four months later.
Sorenson 2003	-extent to which there was discussion of genetic carrier testing for hemophilia A before they offered recently developed DNA-based testing to at-risk women -the roles of relatives in the women's decisions to have DNA-based testing	-majority of patients had severe hemophilia (69%) and were not the only one in their family with the disease (63.5%). - Just over two thirds of patients were Caucasian (67.8%) and three fourths were protestant (76.5%) -majority of men (50%) were not married due to young age - The 98 women relatives came from 23 kinships -There was great variability in the number of eligible	- Two populations: patients and their at-risk relatives. - All patients were followed at two large southeastern hemophilia clinics. Before a regularly scheduled clinic visit, the patient/parent received a letter from the clinic. At the next clinic visit a research genetic counselor approached the patient, reviewed the study and asked to consent. Then they were administered a questionnaire. - Staff contacted eligible at risk female relatives who completed a baseline telephone interview and one at 6 and 12 months later. -Once patients were informed that the mutation in their family had

Author	Study Objective	Participants	Recruitment
	-which family members did the women communicate the results of their DNA based testing -whether the tested women have concerns about communicating carriers test results to relatives, including their children.	relatives identified in the kinships, ranging from 1 to 38 eligible relatives. - the rate of eligible relative test acceptance within these kinships ranged from 18% to 100% -43.9% were between 18 and 35 years old, 41.8% were between 36 and 55 years old and 14.3% were 56 or older. - Most were married (68.4%) - 52% reported high school or less, 28.6% reported having some college education and 19.4% had more than college. -Large majority were Caucasian (81.6%) and indicated they were protestant (77.6%) -Of the 94 relatives just over three fourths were not planning any pregnancies (78.4%), only 3% were unsure about their reproductive plans and 18.4% were going to have more children.	been identified, staff worked with participants to facilitate contact with at risk female relatives. -At-risk relatives were first contacted by the family member with a packet of information. Once the family contact provided the counselor with the telephone number of an interested relative they contacted the woman to tell them amore about the study and let her deicide if she would have genetic counseling and testing. Counselling and blood drawing for mutation testing were done in the woman's home or a mutually convenient location.
Suslak 1985	-determine whether individuals counselled for translocation carrier status transmitted information of their carrier status to appropriate relatives.	- Seven of the 12 families were identified by birth of a chromosomally unbalanced child, three by chromosome tests for habitual abortion and two by amniocentesis performed for advanced maternal age. - Eight had Robertsonian translocations and four had reciprocal translocations. - the 12 identified balanced translocation carriers had a total of 36 surviving sibs and 21 surviving parents.	Families with balanced translocations were collected from the genetic files at the New Jersey Medical School - the 12 participants were interviewed 1-9 years later by a genetic counselor who had not been involved in the original counselling.
Varekamp 1992	This paper discussed the diffusion of genetic counseling and carrier testing among potential clients, and the influence of the social network on this diffusion. It pays special attention to the influence of family networks on genetic counseling.	- on average, the respondents named 3.5 persons as the size of their personal network - the family network was much larger than the friends network, on average 2.7 for the family , including the partner, against 0.7 for friends. More than half of the respondents did not name any friends or other apart from kin -the women ranged in age from 18 to 38 years. - 24% lived with their parents, 14% lived alone 47% were married and 14% were living with a partner - 37 % had children - most of the respondents had a nearby hemophiliac relative: a father or a brother or a son. - most women know about the possibility of carrier testing (86%). Almost everyone had a positive attitude toward carrier testing and genetic counseling	Hemophilia patients in the Netherlands (where this information was gotten from was not specified) supplied the researchers with addresses of women who were obligate or possible carriers of hemophilia.

Author	Study Objective	Participants	Recruitment
		(70% said it was very useful and 28% said useful). Almost half of the respondents were tested (45%). One third was not opposed to abortion because of hemophila (32% said not opposed, 18% said no opinion) and 50% were opposed	
Wagner Costalas 2003	-To understand what percentage of probands receiving BRCA1 or BRCA2 results actually inform their at-risk relatives, - how the information is communicated to other family members	162 surveys were conducted by a genetic counselor (n=127), social worker (n=17), health educator (n=6) over a 28 month period. -147 (91%) women were Caucasian, 79% had some college experience. -Median age of the proband was 41 years (18-77). -Of the 162 who participated 34 (21%) tested positive 6 (3.7%) tested negative and 122 (75.3%) had indeterminate results. - 62% of the sample was of Ashkenazi Jewish decent. -The median age of the respondents was 50 years (24-77) -40%% (n=65) had a cancer diagnosis and 60% (n=97) had no cancer diagnosis. - the 162 probands had 444 adult at risk relatives: 259 adult siblings and 185 adult children. - 112 were adult sisters (median age 46 (25-73)), 147 were adult brothers (median age 46 (25-77)), 92 were adult daughters (median age 34 (18-53)) and 93 were adult sons (median age 33 (18-77)).	Recruited from a multidisciplinary family risk assessment program at the Fox Chase cancer center. Participants who were identified as having a possible hereditary family history were offered the option to participate. Recruitment occurred from March 1997 to July 1999. - Three months after disclosure of the genetic test result, the genetic counselor or social worker called the proband to administer the telephone survey. Voluntary consent was obtained over the phone
Wolff 1989	- report on the results of an investigation in 36 families with inherited balanced translocations, with special reference to the completeness of the family investigation, causes of incompleteness, attitudes of family members to the family investigation and sharing information within families.	- the majority of probands were registered through a child with an unbalanced translocation (20/36), followed by families with multiple abortions (8/36), the finding of a balanced translocation in a child with dysplasias and/or mental retardation (4/36) and the coincidental findings of a balanced translocation at amniocentesis based on the indication of increased maternal age (3/36). In one family, infertility of the male partner was the indication for chromosome analysis.	All families or carriers of a balanced chromosome abnormality ascertained in the area by 1986, found in a registry (not specified). There were different reasons for referral to cytogenetic analysis. Chromosomal analyses were performed. Starting from one identified balanced carrier, they actively tried to get information about and contact with the families of both parents and the family doctors. Tests were then done on willing family members.

APPENDIX G: Summary of main findings from included studies

Study	Summary of Main Findings of Included Studies
Berth 2006	• On average, the participants informed 3.4 family members. In many cases, participants did not inform relatives who were identified being at increased risk for developing HNPCC. • Disclosure of information was influenced by age, gender, patient vs. health consumer, degree of relationship, psychological distress, and family characteristics (cohesion)
Blandy 2003	• The diffusion of genetic information to living, high-risk, adult relatives was generally high (75%), especially among first-degree relative (87%), nieces (86%), and nephews (88%). It was still high among second-degree relatives (76%) band among first cousins (63%). Except in the case of daughters, who were informed more frequently than sons (94% vs. 72%) the diffusion of information was just as frequent among male members as among female members. • The average testing participation among informed high risk relatives was only 15%, with rates of 29% for first degree relatives and 12% for nieces. Of the total of 22 aunts and uncles, one-third was informed, but none were tested. In 11 families, no close relatives of the index case requested the test. • Statistically significant differences were found between males and females in terms of those going for testing. • Understanding of the risk of a genetic predisposition to breast cancer was very poor recall and understanding of the mode of transmission of the risk. The study participants still overestimated the lifetime risk of breast cancer for the general population and underestimated the risk for a woman with a BRCA1/2 mutation. • 67% thought that it would be important to have a written document summarizing the information given to them at the first genetic counseling visit. 47% thought that it would be important to have time to think before taking the test, 63% would have liked another visit after the result. 93% of the cases would certainly have chosen to participate; one would probably have taken the test and only one would certainly not have decided to participate in BRCA1/2 testing. 80% were certainly recommended to their relatives that they take the test. The other 6 patients would probably recommend the test. None of them would not recommend the test. • Two variables found to be significantly and positively associated with a higher percentage of testing decision-making among first-degree relatives were 1) greater family support and 2) better knowledge of the risk of genetic transmission by the women. • There were a number of favorable and unfavorable factors for diffusion of information and testing participation by relatives: 1) personal characteristics of the relatives, such as age and gender (children may be too young, and mothers too old) , males appear to be difficult to inform and to motivate, whereas the fact of being a female relative appeared to be favorable in the diffusion patterns; 2) family characteristics - cohesion and support was the most important thing and positively correlated with the diffusion of information and testing decision making by relatives. A family representation of cancer as a taboo subject did not help index cases to inform their relatives; 3) Psychological characteristics of the recipients. Coping strategies, such as denial or blunting behavior, were barriers for the diffusion of information in the family 4) personal characteristics of the index case – if they felt responsible for the health of others they would spread the information, and incoherent answers, misunderstandings, and confusion between different risks, and the persistence of false believes prevented accurate transmission, the doctor-patient relationship was very important as well. 5) Difficulty for the index case to accept that the relatives refuse genetic testing.
Blase 2007	• Of the 12 participants, 75% reported sharing with at least one family member that their child was having genetic testing to try to determine the cause of the hearing loss. • Usually they shared with first degree relatives • Reasons for sharing included emotional/psychosocial support, to update and keep the family involved, implications for family members and passive information distribution • In most cases where parents shared, the news was greeted with questions and concerns regarding perceived benefits and harms. Perceived

Study	Summary of Main Findings of Included Studies
	benefits responses captured comments indicating curiosity and hopes for an explanation of the hear ling loss. More frequently observed were responses categorized as perceived harms and included comments regarding procedural and psychosocial issues. Of these the most common theme dealt with the psychosocial issues of blame/guilt, denial in accepting hearing loss, and mistrust of modern science. One response indicated concern about procedure • Social or geographical distance and lack of motivation were the primary reasons for not sharing about the genetic process with selected family members. Other reasons for selective non-disclosure included irrelevance, family communication set up and prematurity of the information • After receipt of the test result: all participants reported sharing the results with at least one family member, regardless of test result. In all cases, participants shared their child's genetic test results with participant's first degree relatives (parent, sibling) and in addition a third of the participants shared with grandparents, aunts and uncles or cousins. • Reasons for sharing included for emotional psychosocial reasons, to update and keep family involved or passively shared the information , having a duty to inform (for positive test results only) • For participants receiving a positive test result, the difficulty may have stemmed from trying to explain autosomal recessive inheritance to an unsophisticated audience and in those cases participants tended to either give their family members a copy of the test results or to explain the results in literal, procedural terms. • None of the participants who had a negative or inconclusive test result felt that it was easy to explain the test results to family members. • Regardless of the test result it was not uncommon for relatives to raise questions, comments or concerns about the genetic information an the nature of their concerns was clarified as practical or psychosocial. • Practical concerns included what the risk was to other family members, positive test results brought questions or concerns about the testing procedure and results, ramification of the information to the child and research opportunities. Negative or inconclusive test results brought questions of concerns about other explanations of hearing loss. And racial/ethnic differences in the testing. • Psychosocial concerns involved concerns of the origin if the genetic material or emotional well being • 8 of 12 participants opted not to share the genetic test results with at least one family member. Regardless of the test result, the family members that participants most commonly chose not to share with were in laws and extended family; however, a majority of participants also chose not share with more closely related family members such as siblings and parents • Reasons for not sharing were based on social/geographic distance, irrelevance of the results, a desire to protect, family communication set up .
Bowen 2004	• General communication and health communication was more frequent than breast cancer risk communication • Communication about breast cancer risk was reported with very low frequency across all relative types (community sample) • Mothers and sisters were talked with more often than fathers, brothers and children about breast cancer risk information • Age, number of supporting relatives and tendency to seek personal support were predictors of communication about breast cancer risk • Interest in genetic testing was predicted by a tendency to seek social support less frequently
Bradbury 2007	• 55% of parents reported discussing hereditary risk of cancer with at least one child. 49% of the 86 offspring learned of their parents genetic test results or the hereditary cancer risk • most parents disclosed immediately, 30% reported delaying communication to at least one offspring. The primary reasons parents gave for delays included waiting for the child to get older, for the parent to accept the information, or to decide how to use the information for themselves and to share it personally. • The majority of adult and late adolescent children learned of the hereditary risk of cancer or their parent mutation. The mean age o children who were disclosed to was 17.95 (standard deviation, 4.85). Although disclosures to young children were rare, two children younger than 10 were told of their parent's BRCA mutation or the hereditary risk of cancer, one as young as 7. • Factors associated with disclosure included older child age, female parent sex, parent history of prophylactic surgery, and less formal parent education.

Study	Summary of Main Findings of Included Studies
	• Offspring age was strongly associated with disclosure; the majority of adolescent and adult children learned of the familial mutation or the hereditary risk of cancer • Most disclosures were intentional, one child learned of the parent's BRCA mutation inadvertently. Of those who did not disclosure because they felt their child was too young, the majority (90%) had at least one child younger than 10. • 45% of parents reported that they themselves were the most important person in assisting them with the decision to disclosure. 43% said 'my spouse' and 5% reported a health professional. • Reasons for disclosure included: provide access to information and awareness, so the children could be tested, to explain the family history of cancer, for the grandchildren, to explain the parent's medical intervention , because ti was the appropriate age to tell, unintended disclosure • Reasons for not disclosing included: children were to young , it might increase childes anxiety or fear of getting cancer, might increase child's worry about parent's health , unnecessary given the child's awareness of the family hsiotyr, parent still coping eith the test result, no medical reason to disclose. • Parents reported that some offspring did not appear to understand the significance of the information and some had initial negative reactions to disclosure. Physician and genetic counselor involvement in parent decisions to disclose were low.
Clarke 2005	• A wide range of mendialian and chromosomal encountered disclosure issues, but were identified in less than 1% of consultations in twelve months • 39 cases of parents failing to give information to adult offspring, 22 cases where siblings or other relatives were not given information and 4 cases where information was withheld from partners • Significant life events can diminish the immediate importance of the genetic information (ex. Imprisonment, heroin addiction) • Most often reported reason for non-disclosure was due to concern and desire to protect their relatives rather than poor family relationships. • Many cases of nondisclosure were due to practical difficulties encountered by the clients in managing the disclosure rather than from their wish not to inform their relatives. • There were no reports of professionals disclosing information to relatives without the consultant's permission but there were several instances of active offers to facilitate disclosure and persuasion.
Daly 2003	• Majority of men communicated their positive result to their family freely • 3 instances of non-disclosure occurred and were related to prior bad experience witnessing close family members with the illness. • A spouse of one of the men experienced an adverse psychological reaction to the news but the risk information was still passed onto their offspring. • The main motivation for seeking testing was concern for their children, and they did not express significant self-health concerns
Farkas Patenaude 2006	• Majority of participants had informed the majority of their first degree relatives by four months. Fewer than 9% reported not telling any FDRs by 4 months • For all test results, disclosure ranged from 71% - 87% to parents and siblings of the participant • 71% of brothers and 86% of sisters had been informed directly of their sister's result • 29% of brothers had not been informed • The only category o of FDRs win which 100% of living relatives had been informed of the test result were the mothers, sons >18 years, daughters>18 years of women who received true negative results. • Females were told more often than men in all test result groups, as well as female children were told more than male children • Children less than 13 years old were told less often than children older than 13 and 30. • Conclusiveness of result (true negative and true positive vs. inconclusive) appeared to affect whether sisters were told. • Participants were more likely to inform their brothers if the mutation was inherited through the paternal line

Study	Summary of Main Findings of Included Studies
	• Older women were less likely to tell their result to their mother and father than women younger than 40 years • When a father had experienced cancer the proband was more likely to tell him the test result.
Forrest Keenan 2005	• Interviews confirmed that disclosure was a problem for some participants and it was evident that there were important communication issues common to HD and HBOC participants • It was felt that it was the parent's responsibility to tell (adult or young) children about their risk. And in both groups health professionals were sometimes needed in a supporting role to legitimize or clarify risk information. • There were a few key people in families who pass on genetic knowledge to other family members. The pivotal person self identified themselves as the one who bears responsibility for others younger then themselves. There may also be pivotal people who prevent communication. • Female participants often took responsibility for disseminating risk information and for initiating contact with the genetic clinics. • Some participants were aware of potentially 'unknowing' relatives but did not feel it was their responsibility to pass on the information. • Lay constructions of family and kinship may influence individuals' perceptions of genetic risk, attitudes towards testing and communication patterns in families. Participants had very different notions of who is and who is not family, and about their responsibilities and obligations towards different family members, and that is highly dependent upon their own individual and familial experiences. • Information tended to be passed on to first degree relatives and if it wasn't it was due to past family conflicts. Second and third degree relatives were told selectively – mainly those who they felt emotionally close to or had some contact with. • Family communication is influenced by who people feel obligated to tell but this is tempered by the condition. • Participants could feel very different responsibilities and obligations towards different family members, and that this was highly dependent upon their own individual and familial experiences. • It was more common for HBOC to actively seek out more distant relatives. • There was so evident indicating that the male HD participants felt a moral responsibility to tell close and distant family members of their risk.
Foster 2004	• Tensions between these women's (social) roles of kin keeper (maintaining family relationships), health-promoter (taking care of the health of family members) and support-seeker (desiring support upon hearing their results) impacted who, when and how they told about their risk status • There was a sense of moral duty to tell family members about risk and test results which conflicted with a desire to respect family member's right not to know this information and a desire to protect family members from potentially upsetting information • Differences in coping style and desire for information between different family members presented difficulties in disclosure. • Changes to structure of family following deaths from cancer affected the availability of information and increased reliance on a small number of surviving relatives for information. • Information dissemination was a process and the task of providing information (or full information) was often delayed, involved a face to face meeting or delegation of the responsibility to another relative. • They considered age, feelings and their right not to know unpleasant or unwanted information. • Telling children depended on the appropriate time • Spouses and friends provided support to the women when they sometimes felt they didn't have anyone to talk to • Women spoke with women more often
Gadzicki 2006	• Female relatives were informed more frequently than male relatives • 85% of sisters and 77% of daughters were informed about the test results compared with 57% of brothers and 58% of sons • Non-significant trend that mutation carriers disseminate test results to distant relatives less frequently than true negative • Uninformative test results were generally reported less frequently within the family than a true positive or negative result

Study	Summary of Main Findings of Included Studies
	• Mutation carriers reported significantly more frequently that they faced conflicts with family member about how to deal with the test results compared to negatives • 10% of all women tested were asked by at least one family member not to disclose the test result
Gaff 2005	• All respondents told some at-risk family members about testing by directly information 72 at risk unaffected members • Average number of family members informed was six per family (3-9) • Virtually all siblings and children were informed within 48 hours of the proband receiving their result • 11 of 12 respondents failed to communicate with one or more members of their extended family (2^{nd} degree or more distant) • When family members and their offspring were at risk, respondents felt that their responsibility had been discharged when they told the parents of the children. • Men only informed first-degree relatives, while women informed only some members of their extended family • Two men sought assistance from women • Responsibility to nieces and nephews was passed onto their parents when siblings were told. • Women reported that it was normal for family members to communicate about these issues where men did not. • People were motivated by the desire to help family become aware of choices that could improve their health and the availability of predictive testing was seen as good news. • Family reactions included: interest, disinterest, denial, shock, fear and trepidation. • Counselling on who to inform and how was mixed in opinions in usefulness, however; the booklet and post clinic summary letter were reported as helpful, in ensuring that family members had accurate information • Men found the genetic counseling about who to inform found it very helpful where the women did not • At risk family members who were not informed were usually socially or geographically distant or the information was perceived as irrelevant to that individual.
Gallo 2005	• There were four types of disclosure of information about the condition and its inheritance to children: Openly sharing, selectively sharing, not sharing, and using conversations between parents and health care professionals to inform their children. • Open conversation and selective conversation occurred most commonly. Very few did not tell them anything or used the medical professional approach. • Parents initially focused on sharing information on the management of the conditions and tried to be positive about the disorder by not dwelling on the negative consequences of inheritance of a mutated gene. • Most parents agreed that their children needed to know this information as soon as they began to understand in order to accept the condition and its management and to avoid shock or disappointment. • Selective sharers did not share information that they thought would unnecessarily frighten or worry their children – such as possibility of shortened life or life-threatening conditions and uncertainty about future symptoms and problems • Parents who did not share evaluated their children as being 'too young' or immature to understand the information. Most were younger than 5, one was younger than 8. • One instance of a grandmother raising a child did not disclose because she didn't understand the information and could not explain it to her. • Most parents wanted to be open and honest based on their assessment of the children's level of understanding. • Reframing strategies like comparing the child's condition to a perceived more serious condition were used. • Parents focused on the normalcy of their situation by adopting a flexible approach to information sharing that emphasized incorporating the condition into the usual routines of the family and the child. Life-shortening or life-threatening conditions such as cystic fibrosis or sickle cell anemia, were more likely to cause parents to selectively share information related to death and dying

Study	Summary of Main Findings of Included Studies
	• 20% of parents were given advice by health professional • 80% reported not receiving any advice from health professionals on how to talk with children about genetic inheritance.
Gregory 2007	• Families frequently made specific contrasts between the type of information about haemophilia that they obtained from the clinic, and that generated within the family. The clinic was seen as giving formal medical information about the disease, genetic basis and risks in pregnancy of producing a son with haemophilia or a daughter with carrier status. The family was seen as providing a context in which the reality of living practically with the condition could be appreciated. • There was only one case where the father had fully discussed genetic transmission patterns with his daughters • Discussion within the family normalized haemophilia within the context of everyday life, in contrast to the family of relative gravity of information received from a doctor • Families with non-obligate carriers: information tended to emerge over a period of time, and as the context made t relevant. Often using plain language like 'your blood is special'. Families report giving information in small packages over an extended period, in contrast to the more formal constraints of communication in the clinic. Many referred to answering questions from their children as and when events occurred within the family – such as a n accident or a visit to the hospital. Because haemophilia was such a normal part of their lives, there was no perceived need to discuss it as a specific issue: it was present as an everyday reality. • Obligate carrier families: where information had not been shared with daughter, this more commonly occurred in those families where the father had haemophilia than those where a son was affected. Fathers were less likely to have accidents and need emergency treatment, and girls were not required to be especially careful with their father in the same way as they would be with a brother. In such circumstances, it was more likely that the daughter would be told on a particular managed occasion, rather than growing up with an understanding of the illness and her own carrier status that spring from the reality of that illness. In one account a fathers condition was completely hidden and combined with a lack of overt discussion meant that the daughter reached teenage years without any awareness of the disease • It appears that there is an issue of finding the right time to impart information for some. Timing of communication is of crucial importance, both in delivering the information and in the readiness of the recipient fully to absorb the information and its implication for themselves. • Age and relative maturity is an issue which needs to be considered before disclosure. Relevance of the information for their life stage was also important. Although the disclosure of carrier status means that they know the facts of inheritance of the condition it may not really impact until they are at a stage where it has some practical significance – like when getting pregnant. Another closely related factor that was mentioned in the interviews was the need for information to be communicated by people who knew the individual well enough to judge whether or not they had listened and understood what they had been told. • A crucial aspect of family communication about haemophilia was that it concerned not only the facts and the practical management of the condition, but also the communication of family values about the condition and assurances that it could be dealt with.
Hallowell 2005	• Communicating the information about genetic testing to children was seen as a shared parental responsibility • There was evidence that ongoing discussions about health implication s for offspring were more likely to take place between children (daughters especially) and mothers. • Parents deliberately managed the timing and content of disclosure within their immediate family. • No family informed their children about the father's intention to undergo predictive testing until after it had been decided he would take the test or until after he received his result. • Three communication strategies arose: 1)complete openness, 2)limited disclosure, 3) total secrecy • Majority of parents opted for limited disclosure – involvement in testing was not necessarily hidden, but the disclosure of certain facts about testing was limited a priori. Casually mentioning it in passing and keeping the full implications of potential test results to themselves until after they received the result was also done. • Balancing the children's right to information about their risks with their parental duty not to cause them needless anxiety. Age of children

Study	Summary of Main Findings of Included Studies
	played some part in how communication was managed. • Parents were more likely to limit disclosure about the implications of mutation positive results or keep the fathers involvement a secret if children were younger than 18 years. • Daughters were more likely to be told prior to testing than sons, and those children that parents regarded as emotionally fragile in general or at that particular time were less likely to be told beforehand than those they considered emotionally robust • Not all involved parties were satisfied with the communication method. Some regretted the way they told or did not tell their children and some children resented being left in the dark for such a long time or did not understand the full meaning of the test results.
Hamilton 2005	• Most participants engaged in three processes when disclosing a) they considered the effects on themselves and family members b) they selectively disclosed particular aspects of the information they had received and c) they planned the timing of the disclosure. • Participants assessed their family member's vulnerability and receptivity to the information. Assessments of vulnerability and receptivity were related not to the disease or test outcomes but to life situation and personality. • Siblings were also assessed by considering the mental and physical conditions and the harm the news of the genetic test result might cause. • Some experienced a loss of connectivity with family members after receiving a negative result because it was the 'risk' and 'uncertainty' of disease that connected the family together • Disclosure content differed between HBOC and HD families, with HD being more selective in what they disclosed and topics discussed • Participants at risk for HBOC discussed more information about genetics, risk information and treatment option, meaning of the test result regarding the need for others to be tested, the obligations to children, treatment options and their consequences and the potential effect on reproductive choices. • Timing of disclosure was related to the readiness of the teller and receiver of the information. The only reason disclosure was delayed was if the participant was planning to disclose results in a particular setting (family gathering etc). • HD negative participants expressed concerns about the potential harm of disclosure more so than HD-positive because they believed that the sibling would perceive an increase in their own risk as a consequence of one sibling having a negative test. • More distant siblings were told only after deliberate planning while close siblings were told almost immediately.
Holt 2006	• The family which chose to disclose risk status: father with HD felt resentful that he learned of HD in his family through a pamphlet left out in his house. His family had no communication about it, as a result he was very open with his family and saw genetic testing as a tool to help his generation and the next make the best decisions possible based upon knowledge and information. Family meetings were the central forum for discussions. The middle daughter suffers from depression because of knowing what the family is going through. The eldest daughter is considering predictive testing for herself; she agrees with her father that predictive testing is a tool that will empower her in a variety of way regarding her health status and her future planning. Regardless all children agree that the open /supportive disclosure communication style was the right decision. • The family which chose non-disclosure: parents decided not to tell about the family history and their own risk status to children until father started to show symptoms. Entire family believes that testing is only useful for diagnosis of present symptoms – not predictive testing. They believed that knowledge about having the disorder would disrupt the emotional and psychological health of the whole family. The preservation of hope played a major role in not telling the children. The children felt pressure by their parent's beliefs and did not want to undergo testing in order to protect their parents and not upset them. They children echoed their parent's beliefs about genetic testing. All children expressed a preference for an open/supportive communication regarding HD within the family. • Individuals considering predictive genetic testing for themselves are influenced by the dynamics of the family unit. All four children displayed evidence of direct familial influence on their current thinking regarding predictive genetic testing. • Desire to protect is a central issued when nondisclosure is chosen.
Kasparian	• Most commonly anticipated emotional response to being identified as a mutation carrier in groups 1,2 an 3 was calm acceptance. At least one

Study	Summary of Main Findings of Included Studies
2006	third in each of these three groups also expected to be a carrier. For some the personal importance of such information appeared to outweigh any anticipated negative affect. Those in group 4 acknowledged the potential to feel depressed, worried, scared, and or shocked above all other emotions (group 1 = diagnosed and family history, group 2 = no diagnosis, family history, 3= diagnosis, no family history, 4= neither family history or diagnosis). Group four also made reference to potentially having mixed feelings about a positive genetic test result, feeling fearful of melanoma and perceiving themselves as sick or defective. More males than females anticipated calmly accepting the status of carrier. Second, a greater number of women reported that they would feel depressed or worried about their risk if they were found to be a carrier. More males than females perceived themselves as carriers, and expected to receive a positive test result. • Although half of the sample believed that genetic testing would not have any impact on the quality of family relationships, the sharing of carrier status information, particularly with immediate family was a major theme. Importantly, participant's anticipated family communication to be based on pre existing patterns of inter-relating within the family unit. Females emphasized the need to disclose a positive test result to family members and to discuss the need for relatives to practice precaution adoption. Some mentioned the need for support, and a similar number expressed worry about the way some family members may cope with the participant's result. • Small subset of participants expressed a sense of uneasiness about disclosing to family members. This subset of predominantly female participants discussed their desire to keep their genetic test results private, for fear of the way other family members might react, particularly if their relatives were unwell, elderly, or tended to be illness focused. Interestingly, all but one of the participants within this subset also said that they did not intend to pursue genetic testing for melanoma risk. • A small subset of participants said that being identified as a carrier would stimulate careful thinking about childbearing. All but one thought that being a carrier would not prevent them from having kids. • Participants though that being a non-carrier would evoke positive emotional responses – relieved, happy, satisfied, and/or lucky. • Some who already had melanoma diagnosis would find a negative test as hard to believe. • Sizable majority of participants thought that even if they were a non carrier their current precautionary practices would be vigilantly maintained. They had been habituated. • Some participants did appear to consider the idea of reducing precautions when outdoors and in the sun. This group still valued taking precaution. • At least one participant from each group without a family history of melanomas said they would increase the amount of time their bodies could be exposed to the sun. It was not shown in groups 1 and 2. • Most commonly anticipated cognitive response to non-carrier status was that, despite genetic testing results, on was still susceptible to developing melanoma. Different factors contributed to this belief – participants' multi-factorial view of melanoma causation, multiple past melanoma diagnoses, awareness of non melanoma skin cancers, and the importance of being a good role model for children within the family. Spontaneous disclosure of this belief was equal across all groups except group 3. Some from group three thought that in the presence of a negative result their diagnosis could be considered a fluke.
Kenen 2004a	• Communication patterns that members of families with HBOC history used included: 1) open and supportive 2) directly blocked (hang up phone, refuse to answer, agree not to talk about cancer) 3) indirectly blocked (not responsive, appear uncomfortable) 4) self censored (pro-active, fear of causing anxiety and 5) use of third parties (intermediaries, sounding boards). • Open and supportive communication characterized by: most women had some family members to whom they could speak freely – sisters or female cousins. This did not mean that all of their family was. Some members were reticent or hostile. • Directly Blocked: extreme differences at what could be openly discussed. Their lives were intertwined but had difficult relationships • Indirectly Blocked: family members sent out signals instead of overtly cutting off conversations. They were unresponsive or felt uncomfortable. Men often sent the blocking signal. Hiding past family cancer illness was another way that communication was indirectly blocked

Study	Summary of Main Findings of Included Studies
	• Self-Censored: there were reactive people who sensed that someone didn't want to talk and therefore pulled back. Proactive self-censurers will not talk about the cancer to specific members of the family for fear of causing anxiety. • Use of third parties: some asked an intermediary to contact the persons • Most families were open to discussion about cancer in the family, but even the predominantly open ones included members who wanted to avoid the issue. • A combo of personality differences, differences in values regarding openness of family communication, and differences in reactions to the family context and genetic clinic context lead to various approaches to discussion, or avoiding discussing, their feelings about the family history of breast / ovarian cancer.
Kenen 2004b	• Friends of women with cancer genetic risk war not considered family but were considered easier to talk to because frequently family members found their emotions about cancer got in the way of discussing the topic. Some friends the women considered to be 'like sisters'. • There was a much wider range of sister relationships than friend relationships. Most of the women had close relationships with sisters but couldn't necessarily talk about their risk for developing breast/ovarian cancer. Some had completely open discussions while other conversations were truncated because some sisters were fatalistic. • Less conversation with brothers. Brothers appear to be in a space apart which it comes to discussing the breast/ovarian cancer in their families. Particularly if a loved member had died. Some women found them unresponsive. The women didn't know why but they think that either men don't think about the cancer very much, are less interested, less honest or less verbal. • Partners were supportive on the whole but a few could not talk to them about everything because they could feel vulnerable and upset when faced with the possibility of their loved ones becoming ill and dying • Most women indicated that their partners were supportive but added qualifications (ex. He is very supportive but I can't talk to him about my fears) • Mothers worried about protecting their children and found it difficult to decide what to tell and when to tell their children about the cancer that runs in the family. Some did not know that men could carry the genetic mutation. • Authors suggest multiple viewpoints for understanding men in the genetic risk and counseling setting.
Kohut 2007	• 94.3% agreed with the statement that 'if I am told I have a HNPCC mutation, it would mean that my family members could also have an increased chance to develop colon cancer' • 93.3% of all survey respondents thought that it was their duty to tell all at risk family members and 95.2% thought that it was their duty to encourage regular screening examinations. All nine mutation carriers who responded agreed that they had a duty to tell family members about the mutation and about their possible risks of cancer • 96.2% indicated that they would tell their family if they were diagnosed as having cancer. The total number of respondents who would tell their family members about a negative HNPCC result was 87.6%. The proportion of respondents who thought that difficult relationships would prevent them from informing family members about an HNPCC causing mutation was 18.5% .28.9% indicated that lack of contact would prevent disclosure. Most participants denied having issues telling family members. All 15 respondents identified particular relatives whom they would not be able to tell and specified the reasons. • A total of 40 participants (38.1%) provided written comments, revealing three major themes. The majority (n=27) strongly believed that disclosure of test results to family members is important and is a personal responsibility. Some respondents indicated that they would want their relatives to be informed, even if directly though their doctor without their permission. Two believed strongly that disclosure should only be through the patient who is tested. Seventeen additional respondents were hesitant to contact family members owing to communication barriers or lack of contact. The strongest responsibility was felt towards immediate family or relatives with whom they were in contact.
Klitzman 2007	• Individuals struggle with what to tell and disclosed a range of information. • A few disclosed nothing to certain family members, but these tended to be asymptomatic

Study	Summary of Main Findings of Included Studies
	• Coded or partial information may be divulged. Others revealed only that HD was a possibility even if it had in fact been diagnosed • Within families, overt deceit can be encouraged and defended; older family members may dissemble to offspring who were now in their 40s and still did not know. • In part, families vary a priori in styles and norms of communications. Indirect of implicit or nervous humor was documented • Delaying information rather than permanently prohibitions can build on longstanding histories of silence over multiple generations arising in part from general family communication styles and denial or minimization • How ones parents discuss HD can shape how one later talks about it to one's own children • Disclosure may transpire immediately and unhesitatingly because it is fully expected within the culture and implicit norms of a family. • With children, partial and incomplete information may also be given over several years generating misunderstandings. Incomplete information can foster misunderstanding about what was not being said • Disclosure may become unavoidable and essential for coping optimally once symptoms appear • Many individuals disclosed because of perceived responsibility to foster the health of family members enabling these others to pursue appropriate medical evaluation and family planning. • Tensions emerged because the information could burden the recipient. These feelings often stemmed from other prior experiences and interactions, and desire to avoid troubling elderly parents already overwhelmed by the effects of HD. • People didn't tell parents because they didn't want them to feel guilty • Parents justify not disclosing to children because the want to avoid burdening them • The fact that HD causes psychiatric symptoms also impeded disclosure. Individuals made assessments that family members because of age and psychological fragility could not handle the information • Even without symptoms, being at risk can be difficult because psychiatric symptoms can cause discrimination and generate additional fear, representing loss of control and not generally being fully treatable • Strained or estranged relationships can impede disclosure. • They took into account whether the family member would want to know the result. • Decisions on when to disclose were shaped by two sets of time frames – that of the life cycle of the family member who is told, and that of the medical course of the affected members • Marriage, engagement, or having children were important points in life to tell • Decisions of what and when to tell also shaped each other. Dilemmas of at what age and juncture to disclose a parent's disease are important since when a parent disclosed the child is learning of his or her own risk. Young children may feel overwhelmed. • Extended family members were not told because they were long lost relatives. • With unetended relatives, rules were unclear since patterns of relationships are more fluid and less socially prescribed • Individualsy may not tell because they are not close socially even if they are biologically • After disclosure difficult questions arose too – wiether to pressure family members to be tested. The issues involved in testing family members included: family members vary in their interst in testing, individuals can encourage others to test to knowothers risk, individuals can choose not to push because of a family member's vulnerability, becaue of other's rights, individuals can dissuade others from testing, individuals face decisions of when to disucss and or encourage testing.
Landsbergen 2005	• The mean % of eligible first and second degree relatives per family not tested for the BRCA mutation was 64% (7-100%). • In 14 families, 20% or fewer of the family members were tested • In 34 families, 20% or more of the family members were tested • 62% of index patients (n=31) did not inform one or more family members about the mutation detected in the family.

Study	Summary of Main Findings of Included Studies
	• Reasons included: they had already been informed by other family members (52%), lack of contact (42%), the information could be to emotional (26%) and the message would be too difficult for the family member (23%). • The barriers 'felt insufficiently informed', 'was to difficult' or 'was too emotional' was mentioned less often. • The participants commented that 16% of their relatives did not have the test because it was a man without children, 7% because they were too young and 4% because they were not financially able. Emotional reasons for not talking the test included: fear (37%) and avoidance (16%). Two women wanted to complete their family before getting the test. • The low genetic uptake group retrospectively expressed a desire for more support in the communication with family members. Emotional problems blocking informing relatives were only reported by index patients from families with a low genetic uptake. Index patients of families with a low genetic uptake informed their parents less often after disclosure and do so less often personally.
Lim 2004	• Women reported the impact of receiving a genetic mutation result for themselves, and the impact on their relationships with their partner and wider family. • Most women reported no significant impact on their relationship with their partner, most had a supportive partners. • The wider family was also generally supportive but some had neutral reactions (they were used to cancer in the family) and a few were unsupportive. • A few women were selective in who they told • Both those with positive and negative results spoke of the difficulty of dealing with other family member's results and emotions at the same time. • Some mutation positive and negative women didn't tell their families or were selective in who they told about their results. • Knowing their mutation status was seen as an advantage because it removed uncertainty and gave a sense of control, opportunity to prepare mentally and emotionally as well as increased access to screening programs and surgical options were available to them. • Advantages of negative test results include peace of mind, relief for their children and reduction in anxiety. • Majority cited no changes or impact on their lifestyle since receiving their result. Some positive mutation women mentioned taking better care of their general health, weight, diet fitness and stress. A few younger women described it as a life changing experience, a reassessment of priorities and renewed appreciation for life. • The majority of women cited no disadvantages; four women experienced intrusive thoughts and felt a loss of innocence. A mutation negative worried about becoming to complacent. • Of those who did not want to know their results three reported not wanting to become stressed out but the constant thought of getting cancer if they were identified as carriers and wanted to live life as it came. Two cited insurance implications • Conclusions: the impact of genetic testing extends further than the individual to the family and social context.
MacDonald 2007	• Cancer risk communication increased with all types of first degree relatives pre to post genetic cancer risk assessment, but the degree of change was non significant • Most women (91%) discussed cancer risk with female FDRs and their spouse/partner (86%), whereas only 62% did so with male FDRs and far fewer indicated doing so with other relatives (primarily reported as aunts or cousins). The composite communication score did not significantly differ pre to post genetic cancer risk assessment. • Changes in all of the communication barriers were reported pre to post GCRA but the degree of change was non significant. • Some women (5-32%) reported pre and post GCRA cancer risk communication barriers, mainly geographic distance, loss of contact, and difficult family relationships. • Most women indicated that discussing cancer risk provided useful information to their relatives and the difficulty talking about cancer risk did not prevent these discussions. • The composite communication barrier score did not significantly differ pre to post GCRA.

Study	Summary of Main Findings of Included Studies
	• Risk communications were not significantly influenced by demographics, cancer history, BRCA status, or GCRA. • The lack of statistically significant support for their hypothesis that interfamilial cancer risk communications and communication barriers would influence by GCRA an other factors they believe that several finding shave clinical relevance and provide further support for similar observations in the literature. • Women discussed cancer risk with three types of family members, both before and b six months after their clinic visit. Spouse/partner and female first degree relatives and sisters. • Barriers to cancer risk communication included: concern about upsetting others, recalling painful memories, geographic distance, information not useful, difficult family relationships, difficulty talking about cancer risk, age differences, lost touch with relatives.
MacKinnon 2007	• Everyone who filled out the baseline questionnaire stated that they had conversations with family members about genetic testing prior to attending the retreat. Spouses and sisters were the most commonly spoken to relatives. A quarter of these individuals stated that the results of genetic testing had changed relationships within their family and most said the information brought the family closer together. • At follow up – the majority of attendees continued to have discussions with family members about issues related to BRCA. Three individuals reported changes in family relationships since the retreat. Two were positive: one woman's parent understood better what their children were going through and this made them all closer, and one mother felt more educated an able to talk with her daughter. One experienced a negative change in relationship: her father refuses to discuss it and it upsets her. • The majority had good health and practiced healthy lifestyle behaviours at baseline. At follow up, of those who completed both sets (14/28) made changes in their lifestyle following the retreat and over half said it was due to information they received at the retreat. • 17 of 28 participants who completed both the pre and post retreat questionnaires reported that they increased their cancer screening, initiated chemoprevention, completed prophylactic surgery or are considering prophylactic surgery in the future. 12 of the 17 individuals reported that their decisions were made based on information received at the retreat. • At baseline 53% of the group reported having been to a therapist to discuss their personal and or family history of cancer and 24% reported taking anti anxiety or anti depressant medication. • The retreat did not change distress levels, avoidance scores or intrusive thoughts.
McGivern 2004	• All participants informed at least one of their at-risk biological relatives of the test result. • 37 subjects reported having 803 living relatives including children, parents, siblings, nieces, nephews, and grandchildren as well as aunts, uncles and cousins from the affected side of the family • 470 of 803 (59%) living relatives of the proband were personally informed of the test result. 100% of sisters and 93% of brothers were told. • Children under 18 years were informed less than children over 18 years. • First degree relatives were informed of the test result more often than second or third degree relatives. • Aunts, uncles and cousins were reported for only the side of the family at risk for the mutation. Sixty-five percent of the respondents reported that the mutation originated on the maternal side of the family, 27% reported the paternal side and 8% were unsure. • First degree relatives (children, siblings and parents (were informed of the test result more often than second or third degree relative s (aunts, uncles, nieces, nephews, grandchildren and cousins. • The different between informed males and females in each category was not significant • 17 of the 37 respondents informed<50% of their relatives. Those that informed <50% of their relatives did not differ from those that informed >50% of relatives by age, race, income, marital status, years since testing, whether they were affected with cancer, or BRCA mutation type. • Methods used for communication included: telling in person, over the phone, letter or email or indirectly through someone else • Males were more likely to use the indirect method. • Topics of discussion: surgery guidelines, feelings about result, insurance discrimination, screening guidelines, cost of testing, family history of cancer, reasons why tested, risk of a mutation. Topics varied by gender.

Study	Summary of Main Findings of Included Studies
	• Motivations for telling family members: inform them of their risk; fulfill a duty to inform, to suggest testing. • Barriers: not being close to and not being in contact with relatives were major barriers • 61% said that they did not directly inform some relatives because they depended on another family member to do it. • 8 of 22 let their sibling decide about informing nieces and nephews • 6 of 22 depended on their mothers to inform brothers, aunts, uncles. • Respondents reported that 2.1 female relatives and 0.6 male relatives were tested for the familial BRCA mutation. • Overall, 37% of the informed female relatives and 11 % of the informed male relatives underwent genetic testing after learning of the result.
Mellon 2006	• There was diversity in family communication patterns between and within families– there were family members who wanted to know and talk frequently about the cancer while others avoided any discussion. Large number of dyads indicated that they talked openly about cancer; this was a way of coping and being able to handle the cancer effectively. • Openness in communication extended to sharing information across the internet, web based information and email. But generally talking decreased over time. • Some participants indicated difficulty in talking about the cancer – referring to it as the pink elephant in the living room and talking about it made it real. • Some felt that cancer was too private of a topic to discuss. • Few participants had in depth understandings of mutated genes, such as BRCA1/2. Uncertainty and myths about cancer were evident (such as believing that cancer skips generations and that men cannot have breast cancer nor pass it to their children) • Some felt decision making about cancer risk information was a personal decision while others saw it as a family affair. Some women would make decisions independently, but family members would influence so of this decision making. Several patients and family members shared they would seek out risk information on behalf of their children. There were a few who did not talk or participate in conversations about health decisions. • Barriers: concern that fear of getting cancer would stop them or their family members from pursuing cancer risk information. Avoidance of dwelling on possible genetic linkage was a common response, along with sensitivity about sharing information with family members • The family's communication and relationship patterns accounted for the level of family involvement in decision making regarding cancer risk information.
Mellon 2007	• For all participants who were married, husbands were identified first by a majority of women. Following in priority ranking, survivors selected daughters, health professionals and sisters, whereas unaffected female relatives preferred mothers, sisters, and then health professionals. • Preliminary themes for communication with select individuals included closeness of the relationship, keeping family members informed, receiving emotional support, relying on the knowledge of the individual selected (particularly health care professionals), and concern for family members a potential risk. • Themes for not discussing cancer risk with individuals in their networks included uncertainty about reasctions, adding more stress, previous family conflict, emotional distance, and young age of family members.
Mesters 2005	• Reasons to inform one's family included feeling a moral obligation to do so and anticipated regret if they did not tell. • External cues such as medical organization/professionals saying that they should tell the family were helpful • If someone has already died from it in inflated their perceived susceptibility and increases likelihood that they will tell others • If there was conflict there was less chance they would be told • All interviewees responded that all family members that should receive the information had received it but the definitions of family members and who should know varied. • Telling was usually executed as pure and simple notifying family members about the hereditary cancer and the possibility to get a hereditary

Study	Summary of Main Findings of Included Studies
	test. • If there was a bad first attempt at disclosure the individual was less likely to inform others, but if they had a few good experiences then a bad ones they would continue to tell. • Some people did it on their own, some approached family members together with a partner or another relative, a family member other than the index patient informed the family, some choose a domino approach or it just happened (no predetermined plan). • Strategies use to inform included just phoning them, a personal visit and mail a letter drawn up by the clinic. • Reactions ranged from shocked and angry and taking it out on the messenger to a confirmation of something they already knew.
Riedijk 2005	• Out of 65 respondents, 11 had had melanoma • 17 respondents indicated that one or more first-degree relatives were/had been affected • 15 had one or more affected second-degree relatives • 27 respondents had one or more third-degree affected relatives • Out of 59 respondents 57 reported having positive attitudes towards the test offer, 83% of whom thought it was good to be kept informed • 38 indicated that it would have been a missed opportunity if they had not been informed and 23 stated that now at least they could make their own decision • One individual indicated rather not to have been informed • 42 respondents felt their relatives should be informed as well and 22 had no opinion regarding the matter and one individual thought relatives should not be informed. • 63/66 reported that in their family everybody could decide about genetic testing individually and 58 felt that they could openly discuss p16-leiden matters and health risks in general in their families. 40 felt that the family should be informed about an unfavorable test result. 24 had preselected mutation carriers in the family and 20 strongly valued the opinions of their relatives regarding genetic testing where 32 did not feel they needed to agree with family members about genetic testing. • 39 people did not want to participate in testing because they were happy with life as it is and 26 were worried about the possible difficulties obtaining a mortgage. The least important reasons for non-participation were the negative impact an unfavorable test result would have on their relationship and being advised bay a doctor not to be tested. • Having children and high anxiety both predicted whether they would participate in testing • Being older and having children increased likelihood of participation in hereditary melanoma testing whereas having anxiety decreased the likelihood of participation in HM testing.
Schroy 2005	• All responders knew they had undergone either a colonoscopy or signoidoscopy but only 75% said that a polyp had been removed during their examination. Only two responders were told that their polyp was an adenoma - benign growth, precancerous group or precancerous lesion. 31% did not know. • Few (n=25) participants were aware that their first degree relatives were at increased risk of colorectal cancer • Of the 25 who were aware of the familial implications of their diagnosis most reported that they had informed one or more at risk relatives about their diagnosis and told them to contact their primary care provider or arrange for screening or to tell their primary care physician at their next visit that a close relatives was recently diagnosed with colorectal polyps. • Regardless of whether they informed FDRs or not, nearly all knowledgeable responders felt that screening would not be difficult for their at risk relatives and in general most felt it was the patients responsibility to notify at risk relatives • This study suggests that physicians are not educating adenoma patients < 60 about the familial risks either because there is a complete lack of communication, ineffective communication or a lack of awareness.
Segal 2004	• Ages 20-29 were disclosed to by carriers most often. 30+ years was the second highest percentage disclosed to. Ages 8-14 and 15-19 were tied for third highest level of disclosure from parents to offspring.

Study	Summary of Main Findings of Included Studies
	• Of Those who did not disclose, 37% had kids who were between 8-14 and 29.2% had kids who were 0-7 and 25% had ages 15-19 • The mothers who did disclose decided to tell because they were concerned for the child's risks and a belief that their offspring had the right to know. They also wanted to increase their children's knowledge about the gene. • Of those who disclosed 44.4% had done it immediately after receiving their results, 5 disclosed within a week, 2 carriers disclosed between 1 and 4 months after receiving the test result. One disclosed between 1 and 2 years and another between 2 and 3 years. One chose not to answer. • Most perceived the most appropriate age to disclose was between 19-15 (35%). 7 thought it was between the ages 14-17 and 6 thought it was 18 years old. 5 said it depends on the maturity of the child and individual difference. One carrier felt that children should only be told after they are married and one said that children should never be told about a positive BRCA1/2 mutation in a parent. • Most who did not disclose had children less than 18 years of age and did not want to tell them because they felt they were too young. 2 cases of non disclosure occurred with children over 18 years of age. And the reasons for not telling said were that fear of increased worry without being able to offer concrete information or tangible solutions. • 66.7 % of the children disclosed to expressed significant concerns about their mothers future health as well as concern for their testing options. Older children tended to approach the situation in a more logical manner, want more information and facts and show more concern for their mother. Younger expressed more worries, and showed a stronger desire for testing and possible prevention. • 67.7% felt their family communication was very open, 8 described it as moderately open and 2 as somewhat open. There were no differences in perceived openness of communication between those who disclosed and those who have not yet disclosed. • Majority of women indicated that the disclosure conversation content would includeL their children's feelings, their child's percentage risk, whether or not the child should be tested, health concerns, preventative methods and the carriers reason for being tested. • Most women disclosed to their children without direct support from their partner. • 12 of 15 nondisclosures (80%) said that they have not created a plan to tell their children. Three have created a plan surrounding circumstances of the conversation but not the content and one had a plan but didn't want to share it. • Similar proportions of disclosures and non disclosures indicated that they would have appreciated further follow-up meetings with a genetic counsellor around disclosure content and process. • Family counselling, peer support groups of carriers and their children, professionally lead support group, educational forum for families, and informational pamphlets on how to disclose and cope were all mentioned as being potentially helpful. • Those who did not disclose were more interested in receiving individual counseling, educational videos, email newsletter's focusing on specific strategies and techniques for transmitting information about genetics and the risk of disease compared to those who had already disclosed.
Taylor 2005	• 64% of the sample was negatively inclined towards predictive genetic testing for HD overall. • Of those who believed they could cope with predictive testing a greater proportion were female than male • 60% of respondent (3/57) reported disclosing their HD risk to others with relative ease, with women reporting greater readiness than males. • The closer the social relationship the more likely respondents were to disclose their HD risk. • Spouses/partner, family of spouse/partner and good friends were told most often by males and females. Women were more likely to tell their general medical practitioner. • Majority did not disclose to their employers and none disclosed to their bank/lending institution, • Majority reported that their HD family history or genetic risk had not resulted in discriminatory treatment, although 54% feared discrimination and felt vulnerable. • Male respondents were three times more likely to fear disadvantage than females.
Van den	• The option to distribute information packages within at risk families was highly valued by index patients and by relatives. Both groups

Study	Summary of Main Findings of Included Studies
Nieuwenhoff 2006	preferred a fact to fact distribution, accompanied by an account of patients own experiences with IHC • All of the index patients informed their relatives and six out of eight relatives in the present study consulted a family physician for a diagnostic cholesterol test as a result of the information package • Nearly all index patients only informed their children sisters and brothers, whereas second-degree and third degree relatives were also at elevated risk. • Informing relatives about a genetic risk was still felt as a burden and index patients searched for rationales to avoid doing so. As a result, index patients generally informed no more than their first-degree relatives, the disclosure to more distant relatives was left to other family member. • Some recognized that distant relatives were at risk but only felt the duty to inform their close relatives (particularly their children) • The information packages seem to have had a function in reducing barriers to inform relatives, however the packages were generally not sent to more distant relatives with whom index patients were not frequently in touch, nor were they personally handed over. • The barrier 'anticipated the message would be rejected' was overcome by including written information. Some index patients revealed the exception that their relatives might sooner reject the oral information than the information package of trustworthy experts. However, threw were also doubts about the significance of this expected rejection. Index patients strongly believed that they had the obligation to inform at least once and that is was to the receiver of the information to accept or reject the information. • Packages focused attention on IHC and have been an external cue to action with first degree relatives. Index patients admitted that they had not thought of their second or third degree relatives. • Relatives generally accepted the content and the way genetic risk information was distributed, using a combination of written and personal communication. • The family packages presumably help to reduce some of the barriers to visiting a family physician but maybe increased others. By devoting attention to insurance, the packages might have induced fear for insurance discrimination which could be a barrier to test uptake. • A strong motive for relative to go for a diagnostic cholesterol test probably was the fear of dying from a heart attack and the opportunity to prevent this. • The family packages were reported to be a strong cue to action for the majority of relatives. • The family packages stimulated index patients with IHC to inform their relives and most relatives consulted a physician. Only first degree relatives were informed.
Van den Nieuwenhoff 2007	• Reasons for disclosure varied from simple compliance with a professional's advice to more complex moral, social and emotional motivations. Index patients (IPs) were influenced by familial, institutional and communal contexts • Practitioner's advice or a written request from the patient organization were important for disclosure • Nearly all perceived a strong moral duty to inform relatives, especially their adult children and siblings. This duty was driven by interpersonal factors, strong family bonds and intrapersonal considerations such as perceptions of relatives susceptibility, the perceived severity of IHC, the possibility of treatment and consistency with one's values and or integrity • There is an altruistic desire or perceived obligation to protect relatives from avoidable physical harm induced disclosure as well. The wish to protect others was based on strong positive interpersonal relationships. • Roles within the family influenced disclosure likelihood. Viewing oneself as being open or helpful to relatives and perceived characteristics of familial relationships, such as openness were reasons for disclosure. • In some cases, disclosure was egoistically motivated and influenced interpersonal factors because being the first to know of a familial risk enhanced their status in the family. Ex. an alcoholic who could now focus the families attention on how she was becoming healthy and helping others to be healthy, a man who's family held him responsible for his CVD had an increased sense of social acceptance. • Three IPs postponed disclosure because they felt obliged to protect relatives for psychological harm at that moment. These adults were already

Study	Summary of Main Findings of Included Studies
	distressed by other life events and children who se parents wanted to prevent them from worrying bout their own and their parents health • No interviewees intended to permanently withhold genetic information however temporary postponement may result in permanent nondisclosure when the IP becomes accustomed to living with the IHC SECRET or when communication between an IP and the relative ceases. • IPs generally refrained from disclosing to second and third degree relatives as they believed that parents were responsible for informing their own offspring (reflecting cultural norms) • Lack of contact with relatives made it strange to suddenly contact estranged relatives • IPs did not attempt to disclose because they expected their message to be rejected (more often with second and third degree relatives) • About half had little confidence in their ability to effectively explain IHC, due to a perceived lack of medical knowledge. Lower self-efficacy seemed related to a lower number of relative approached. • Disclosure usually happened on the individual level and during accidental encounters as it was not perceived as being urgent. • Most IPs viewed the disclosure as one discrete event and did not want to involve themselves anymore in their matters • Two stated they would have liked to have had more assistance from knowledgeable professionals (not necessarily physicians) when announcing ICH. • IPs generally began disclosure by explaining the reason for their conversation, in particular the IPS diagnosis or a professional's advice to inform relatives. Some IPS apparently used the doctor's request as justification for the disclosure or for the recommendation to get tested. Then the facts about IHC were given. They discussed the threat, severity and familial predisposition or susceptibility. • Only five literally had said to some relatives that they should have their cholesterol checked. Most IPs refrained from persuading relatives to get tested because this would appear to threaten relative's autonomy which is highly valued in the Dutch culture. Some used covert persuasion attempts by deliberately stressing the information regarding severity
van Oostrom 2007	• About a third of participants reported positive changes in one or more relationships with family members. 18.8% reported negative changes in one or more relationship, 3.5% reported one or more familial conflicts and 13.1% reported one or more difficult situations in the family due to genetic testing. • Gene mutation carriers reported significantly more positive changes in relation to their parents and second degree relatives than non-carriers. • Positive changes in family relationships due to genetic testing included: feeling closer, improved communication, more understanding and support, more appreciation of the other, relief of a negative test result • Negative changes in family relationships are familial difficulties due to genetic testing: more emotional distance, guilt feelings, secrecy, does not want genetic testing, other relational problems. • Participants who felt less free to communicate about hereditary cancer related issues within the nuclear family at baseline also reported more frequently adverse effects on these relationships and tended to report more familial difficulties. • Participants who felt less free to talk about hereditary cancer with siblings and parents reported more adverse consequences in these relationships and more family difficulties. • Family communication style regarding hereditary cancer was selected to predict adverse consequences in the family of origin and difficult situations or conflicts. • Individuals feeling more reluctant to talk about hereditary cancer with relatives reported more familial difficulties and unwanted changes in relationships. Families characterized by a rigid family structure in combination with unconnected family bonds were found to be more susceptible to negative changes in nuclear family relationships. Also, families with very loose and flexible family structures in combination with very close relationships were less well equipped to cope with the stress of genetic testing. • Open communication between relatives should be stimulated because a lack of open communication may be an important determinant of familial adverse effects.

Study	Summary of Main Findings of Included Studies
Weiner 2005	• Families from the general public are not yet talking about whether to get genetic testing (607/717) said they had never discussed this in their families said they had never discussed this in their families). • 60 indicated that they had had such discussion but selfdom and 37 indicated discussing genetic testing often in their families. • Families do tend to discuss prenatal testing but it is still not a commonly discussed topic (446/717) • Those with genetic test experience are more likely to talk about genetic testing and women discussed it more than men. • Exposure to movies that show how genes affect human health was related to discussing the topic of 'genes and health' • Having a high response efficacy, television viewing and newspaper readership were related to discussions of medical history in the family • European Americans reported more than African Americans that they discuss family medical history and encourage family member to behave in healthy ways.
Wilson 2004	• A total of 30 studies were identified which included the genetic conditions: Huntington's disease, hereditary breast and ovarian cancer, hereditary non-polyposis colorectal cancer, balanced translocations and recessive and sex-linked disorders such as CF, haemophilia, and Duchenne muscular dystrophy. • Factors which are influential in the communication about genetic risk with relatives can be thought of in terms of: disease factors, individual factors, family factors and socio-cultural factors. • Different forms of inheritance can influence whether information is passed on to members. Different inheritance can mean different risks for family members, whether they know about the disease already, how they found out about the disease in their family and risk perceptions. • Genetic risk information is not always certain or easily understood, this may play a role in who tells what to whom. • The availability of preventative interventions and risk management options can also alter people's perceptions of risk and desire to protect individuals from harmful information. • Individual factors like feelings of guilt, blame, fear and defense mechanisms such as denial can close communication pathways. Social support may be a prime mover for communication. Personality and perceptions of risk are important factors which will influence with whom one feels should be communicated with. • The nature of family relationships and dynamics play a large role in how risk information is spread. Relatives who are emotionally and socially close tend to hear this information more often. Distant relatives often get this information less systematically and more selectively. • The communication pattern of a family in general will impact the ease with which information is passes. • Barriers such as divorce, separation, adoption, large age gaps between siblings and pre existing family rifts are also influential. • The 'roles' people play within a family can influence who is seen as having authority to tell such information as well as who normally takes the lead in situations like this. • There is evidence that certain family members can function to either increase or hinder family communication and may represent an exercise of control or protectiveness. • Incorrect information, family myths and myths about inheritance can skew the truth of the genetic risk and influence who is told and who is not • Socio-cultural factors such as gender, stigma or taboo associated with a disease or topic, concerns about discrimination may influence communication. • Cultural beliefs about inheritance and family structures promote pre-selection and promotion of certain communication pathways.

APPENDIX H: Framework creation Process

Table 1: Step 1 - Listing all Barriers and Facilitators from Literature and assigning numbers

#	Factors which act as Barriers	References	#	Facilitating Factors	References
1	Culture (e.g. European Americans more open communication than African Irish less than American, cultural beliefs about responsibility to family members)	Fitzpatrick 1990*, Weiner 2005, Van den Nieuwenhoff 2007	1F	Culture (e.g. certain cultures feel more responsibility towards family)	Van den Nieuwenhoff 2007, Weiner 2005
2	Fear of insurance or employment discrimination, Stigmatization	Klitzman 2007, Mellon 2006, Taylor 2005,Wolff 1989	2F	Exposure to genetic information in the media	Weiner 2005
3	Openness about discussing disease in society	Forrest 2003, Peterson 2003	3F	Acting honorably and respectfully towards children	Klitzman 2007, Skirton 1998
4	Taboo, shame	Blandy 2003, Varekamp 1992	4F	Feeling they have the right to know the information	Blase 2007, Klitzman 2007, Mellon 2007, d'Agincourt-Canning 2001, Hallowell 2005, Skirton 1998
5	Different coping styles between family members	Blandy 2003, Foster 2004, Kenen 2004	5F	Having a feeling of responsibility and duty to inform or moral obligation to tell	Blandy 2003, Blase 2007, Klitzman 2007, Foster 2004, Mesters 2005, McGivern 2004, Daly 2003, Keenan 2005, Van den Nieuwenhoff 2006, Van den Nieuwenhoff 2007, Green 1997, d'Agincourt-Canning 2001
6	Conflicting views about genetic testing, or communication of family history or abortion	Foster 2004, Henneman 2002	6F	Conflicting roles of women: kin keeper, health promoters, support seekers	Foster 2004
7	Different desire for information (about health and genetic risk)	Foster 2004	7F	Anticipated regret	Van den Nieuwenhoff 2007, Mesters 2005
8	Wish to protect and preserve a relationship	Foster 2004	8F	Appropriate time to tell (sexually active, stable relationship)	Foster 2004, Gregory 2007, Forrest 2003, Skirton 1998

#	Factors which act as Barriers	References	#	Facilitating Factors	References
9	Protection of their feelings, not wanting to upset someone (yet or at all)	Blase 2007, Klitzman 2007, Kohut 2007, Kasparian 2006, Foster 2004, Gallo 2005, Hallowell 2005, Clarke 2005, MacDonald 2007, McGivern 2004, Mellon 2007, Kenen 2004, Kenen 2004b (especially with kids), Segal 2004, Holt 2006, Van den Nieuwenhoff 2007, Skirton 1998, Green 1997, Hughes 2002, d'Agincourt-Canning 2001, Forrest 2003	9F	Close social relationship	Taylor 2005, Patenaude 2006, Mellon 2007, Koehly 2003, Ormond 2003
10	Information perceived as too emotional	Landsbergen 2005, MacDonald 2007	10F	Younger age of mother (there is no evidence suggesting why this may be the case therefore not in the framework)	Claes 2003
11	Moral dilemma: conflict between wanting to protect and wanting to help	Hallowell 2003	11F	Being Female daughters, mothers, sisters, nieces, maternal relatives	Blandy 2003, Mellon 2006, Gadzicki 2006, Taylor 2005, Patenaude 2006, Kenen 2004, Bowen 2004, Kenen 2004b, Keenan 2005, MacDonald 2007, Lerman 1998, Claes 2003, Julian-Reynier 2000, Wagner Constalas 2003, d'Agincourt-Canning 2001, Smith 2002, Koehly 2003, Fanos 1995, Ormond 2003, Sorenson 2003, Varekamp 1992, Forrest 2003
12	Conflicting roles of women: kin keeper (protector), health promoters (protector), support seekers	Foster 2004	12F	Older mothers more likely to inform daughters	Julian-Reynier 2000, Smith 2002
13	Wanting to respect the needs of others (respecting their right not to know)	Klitzman 2007, Foster 2004	13F	Siblings, spouses and GPs	Julian-Reynier 2000

#	Factors which act as Barriers	References	#	Facilitating Factors	References
14	Age (too young, under age 18)	Blandy 2003, Foster 2004, Gallo 2005, Gaff 2005 (for surveillance etc), Kohut 2007, Mesters 2005, Mellon 2007, Segal 2004, Miesfeldt 2003, Smith 2002, [Koehly 2003], Ormond 2003	14F	Conclusiveness of test	Patenaude 2006, Claes 2003
15	To immature (not able to comprehend information)	Gallo 2005, Gregory 2007, [Skirton 1998], Henneman 2002	15F	Mutation positive more likely to communicate than inconclusive or negative	Claes 2003, Wagner Constalas 2003
16	Not wanting to spoil childhood and to protect them	Gallo 2005	16F	Cancer is already open in the family (therefore it is easier to talk about it or the family is already familiar with the disease)	Patenaude 2006, Lim 2004, Forrest 2003
17	Guilt (survivor guilt or guilt for passing on disease, guilt for making mothers feel guilty about passing on the gene etc.)	Klitzman 2007, Foster 2004, Hamilton 2005, Hughes 2002, d'Agincourt-Canning 2001, Fanos 1995, Varekamp 1992	17F	Being a survivor of the disease	Peterson 2003
18	Avoidance of resentment (when other doesn't have carriers status)	Foster 2004	18F	Potentially curable disease	Peterson 2003
19	Denial	Blandy 2003, Van den Nieuwenhoff 2007, Ayme 1993, Wolff 1989, Fanos 1995	31F	X-linked inheritance (more likely to be discussed with females)	Sorenson 2003
20	Avoidance of envy of sibling who is not carrier	Fanos 1995	19F	Perception of relevance for members	Kasparian 2006, Patenaude 2006
21	Unhealthy family relationships (estranged relatives, conflicts)	Klitzman 2007, Foster 2004, Gadzicki 2006, Gaff 2005, Mesters 2005, Clarke 2005, Keenan 2005, MacDonald 2007, Mellon 2007, Claes 2003, Green 1997, Julian-Reynier 1996, Koehly 2003, Forrest 2003	20F	A brother is more likely to hear about a sisters BRCA1/2 result if the mutation is inherited through the paternal side (reflecting the myth that if its runs in the paternal line it has more relevance for men)	Patenaude 2006
22	Lack of contact with person (or not knowing how to contact)	Kohut 2007, Gaff 2005, Landsbergen 2005, MacDonald	21F	Strong family history (there is more fear and the	Mesters 2005

#	Factors which act as Barriers	References	#	Facilitating Factors	References
		2007, Van den Nieuwenhoff 2006, Van den Nieuwenhoff 2007, Claes 2003, Green 1997, d'Agincourt-Canning 2001, Peterson 2003, Wolff 1989, Varekamp 1992, Forrest 2003		information is perceived as more important)	
23	Geographical distance	Blase 2007, Kenen 2004b, Kohut 2007, MacDonald 2007, Green 1997, Ormond 2003	22F	Experienced cancer (themselves or someone close to them)	Julian-Reynier 2000
24	Adoption (communicator is adopted)	McGivern 2004	23F	External cues (a physician or counselor telling you to do it)	Mesters 2005, Van den Nieuwenhoff 2006, Van den Nieuwenhoff 2007
25	Superficial relationship or not being close	Blase 2007, McGivern 2004, MacDonald 2007, Mellon 2007, Green 1997, Hughes 2002, Smith 2002, Koehly 2003, Ormond 2003, Varekamp 1992	24F	Counselor's role (in providing information and facilitating discussion with others)	Ayme 1993, Forrest 2003
26	Misunderstanding information about inheritance or myths, not being informed appropriately about risk	Blandy 2003, Mellon 2006, Van den Nieuwenhoff 2007, Green 1997	25F	First degree relatives (relates to social closeness and perception of relevance of information)	Blase 2007, Mesters 2005, MacDonald 2007, Claes 2003, Koehly 2003
27	Not knowing who it is important to tell (thinking its only important for nuclear family)	Blandy 2003, Mesters 2005, Van den Nieuwenhoff 2007	26F	Generally good communication in the family (supportive, take each others advice, and generally get along, share information)	Blandy 2003, Mellon 2007, Koehly 2003
28	Lay attitudes and beliefs about family	Blandy 2003, Keenan 2005	27F	Open communication style in family (dislike of secrecy)	Hallowell 2005, Lerman 1998, Koehly 2003
29	Fear of hearing information about risk, fear of disease blocks communication in family, fear of being responsible for problems in offspring	Mellon 2006, Kenen 2004a, Kenen 2004b, MacDonald 2007, Wolff 1989	28F	Feeling it as an avenue to obtain social support	Klitzman 2007, McGivern 2004, Bowen 2004, Mellon 2007, Van den Nieuwenhoff 2007, Lerman 1998, Ormond 2003
30	Fear of creating conflict in the family	Ayme 1993	29F	Lay attitudes and beliefs	Keenen 2005

#	Factors which act as Barriers	References	#	Facilitating Factors	References
				about family	
31	Fear of being blamed for disease or bringing bad news	Clarke 2005, d'Agincourt-Canning 2001	30F	Someone who takes the leadership communicator role or a key family member, family roles	Van den Nieuwenhoff 2007, Lerman 1998, Green 1997, Koehly 2003
32	Fear of psychological and social consequences of being tested positive	Wolff 1989	32F	Older age of child	Bradbury 2007
33	Anticipated loss of connectivity with family (those families who define themselves by their risk status)	Hamilton 2005	33 F	Being a non-obligate carrier (ie. brother has disorder – haemophilia)	Gregory 2007
34	Anticipating the message would be rejected	Kohut 2007, Van den Nieuwenhoff 2007, Julian-Reynier 1996	34F	Relying on knowledge from the individual	Mellon 2007
35	Anticipated disinterest in testing	Gaff 2005	35F	enhance the probands status within the family (being the one to tell and help, making themselves seem less responsible for their disorder, or shifting focus onto how they are helping instead of an alcoholic)	Van den Nieuwenhoff 2007
36	Being male makes it harder to communicate the result to others and to have information communicated to you (men's ability to cope with test result may not be as good as women's)	Blandy 2003, Mellon 2006, Taylor 2005, Green 1997, Wagner Constalas 2003			
37	No interventions to help the person once they know their risk	Mellon 2006			
38	Pressure from others not to tell	Gadzicki 2006			
39	One person acts as a 'banner' of communication or discourages others about disclosure (because don't think its important)	Clarke 2005, Henneman 2002			
40	Inconclusive test results (uncertainty)	Gadzicki 2006, Claes 2003, Huges 2002 (uninformative), Wagner Constalas 2003, Forrest 2003, Patenaude 2006			

#	Factors which act as Barriers	References	#	Facilitating Factors	References
41	Mutation positive or carrier positive less likely to inform second degree relatives. There is no tentative explanation as to why; therefore it will not be included in the framework.	Gadzicki 2006			
42	Non-carrier (HNPCC)	Peterson 2003			
43	Clinical condition is life shortening or life threatening (more likely to give selective information to kids)	Gallo 2005			
44	Balanced translocations	Suslak 1985			
45	Non fatal disease (therefore people see it as less relevant to discuss)	Sorenson 2003			
46	Complexity of information	Patenaude 2006, Gallo 2005, Denayer 1992 and Denayer 1992b			
47	Perception of relevance for members is low (they aren't having children yet, they don't think some family members need to know)	Blase 2007, Gregory 2007, Patenaude 2006, Green 1997, Julian-Reynier 1996, Henneman 2002, Varekamp 1992, Forrest 2003			
48	No family history of disorder so they think their risk is lower and it's not as relevant.	Ormond 2003			
49	Being emotionally motivated person. This is a reflection of personality	Riedijk 2005			
50	Relative didn't have access to testing (country or insurance)	Gaff 2005			
51	Bad First Attempt at disclosure	Mesters 2005			
52	Believing they had already been informed by other relatives	Claes 2003, Smith 2002			
53	Personality characteristics of the person (emotionally fragile etc)	Klitzman 2007, Hallowell 2005, Fitzpatrick 1990			
54	Persons situational factors (heroin abuse, mental illness, imprisonment, pregnant, marriage plans, old or ill)	Blandy 2003, Clarke 2005, Kohut 2007, Kasparian 2006, Van den Nieuwenhoff 2007, Green 1997, Henneman 2002			
55	Unable or unwilling to take responsibility for	Clarke 2005			

#	Factors which act as Barriers	References	#	Facilitating Factors	References
	informing				
56	Not feeling able to cope with others emotional reactions plus your own reactions	Lim 2004			
57	Having difficulty coping with own problems/diagnosis/test result	McGivern 2004, Forrest 2003			
58	Not knowing what to say	McGivern 2004			
59	Not wanting to increase worry without being able to offer concrete information or tangible answers.	Segal 2004			
60	Believe receiver will not perceive the information correctly (they will think inconclusive is no risk or at higher risk)	Hamilton 2005, Hughes 2002			
61	Peoples own beliefs of the impact of the test on their own life influence what they believe how and whether others will use the information. From a case study, therefore not included in framework.	Lerman 1998			
62	Whether the individual will want the news	Hamilton 2005			
63	Bad lived experiences with Cancer (or other disease)	Daly 2003, MacDonald 2007			
64	Preserve Hope that HD won't be part of the family	Holt 2006			
65	Higher education less likely to disclose to children (higher education gives had a stronger desire to protect because they know how bad the information is)	Lerman 1998			
66	Don't think they have communication skills (or do not communicate in general)	Van den Nieuwenhoff 2007, Skirton 1998, Henneman 2002			
67	Retaining control over family members lives	Skirton 1998			
68	Older women less likely to inform their mothers	Julian-Reynier 2000			
69	Unpleasantness of message (relates to wanting to protect them)	Julian-Reynier 1996			
70	Not feeling they have the authority (inappropriate to tell nieces and nephews and cousins without parents)	Blase 2007,Peterson 2003, Koehly 2003, Forrest 2003			

#	Factors which act as Barriers	References	#	Facilitating Factors	References
71	It never came up	Smith 2002			
72	Someone in the family has died from the disease.	Fanos 1995, Forrest 2003			
73	Wanting to protect their own privacy and confidentiality	Klitzman 2007			
74	Being an obligate carrier (ie. father has the disorder)	Gregory 2007			
75	Unsure of how the family member will react	Mellon 2007			
76	Viewing the act of disclosing as a burden to oneself	Van den Nieuwenhoff 2006			
77	Not feeling it is their responsibility to tell	Van den Nieuwenhoff 2007			

Table 2: Step 2 - Underlying meanings of barriers and facilitators derived from literature organized by TPB construct

A) ATTITUDE FACTORS

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
2	Fear of insurance or employment discrimination, stigmatization for family member	Fear is a reflection of their concern for their family member and a desire to protect them	Desire to protect (other)	3F	Acting honorably and respectfully towards children	Reflects their values towards their children and their right to have health information By telling their children they are respecting their child's right to have ownership over their own health information.	Right to know
5	Different coping styles	This refers to how some prefer not to discuss sensitive topics while others reach out for support. In both cases 'a desire to protect' is evident. One protects themselves by not facing the problem and hindering communication while the other faces it and reaches out for support (protecting themselves). It is pointed out that Men's coping style is different than womens	Desire to protect oneself	4F	Feeling they have the right to know the information	Reflects values about rights to/ownership of health information	Right to know
6	Conflicting views about genetic testing, or communication of family history or abortion	Often different family members have different views about the appropriateness of	Desire to protect (oneself or others)	5F	Having a feeling of responsibility, duty to inform or moral obligation to tell	Reflects their values about responsibility, duty and obligation	Responsibility, duty or moral obligation

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
		genetic testing, and abortion. Also, differing communication patterns about health and ways of coping are common. These differences can lead to conflicts which then lead to wanting to protect oneself and others.					
7	Different desire for information	Relates to the concept of 'right not to know'. If someone would prefer to live without knowledge of their genetic risk, not informing them is respecting their rights and protecting them from information they don't want to know.	Desire to protect	6F	Conflicting roles of women: kin keeper (protector), health promoters, and support seekers	Related to an internal conflict between wanting to protect those you love (and yourself through support) and 'protection' through providing them with health information	Desire to protect
8	Wish to protect and preserve a relationship	Self-explanatory	Desire to protect (other and oneself)	7F	Anticipated regret (note: this factor has been noted in the literature to potentially be a separate variable in the TPB, this notion will be tested with out survey)	Related to ones desire to prevent themselves from regretting not telling if something bad later happens to their family member	Desire to protect (oneself)
9	Protection of their feelings, not wanting to upset someone (yet or at all)	Self-explanatory	Desire to protect (other)	8F	Appropriate time to tell individual (i.e. sexually active, stable relationship)	The appropriate time to tell an individual is usually one where the information would be of use to the person in planning their life. This reflects the tellers perception of relevance of the information	Relevance

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
10	Information perceived as too emotional	Related to fear of upsetting them and afraid of their reaction, which in turn reflects a desire to prevent harm to another	Desire to protect (other)	12F	Older mothers more likely to inform daughters (because daughters are older)	Older age of children coincides with life decisions where genetic information may be wanted and may help in planning for the future. The tendency for mothers to tell children when they are older reflects their perception of relevance of the information for their children.	Relevance
11	Moral dilemma: conflict between wanting to protect and wanting to help	Delaying or not telling because of this moral conflict reflects a desire to protect them from potentially harmful information but at the same time wanting to 'protect' them by giving them the information	Desire to protect (other)	13F	Siblings, spouses and GP	Specific family members will need to know about your genetic test result regardless of whether they are at risk (i.e. your spouse will need to know because they may be your caregiver, siblings would give social support and a GP needs to know for future health encounters)	relevance
12	Conflicting roles of women: kin keeper (protector), health promoters (protector), support seekers	Related to an internal conflict between wanting to protect those you love (and yourself through support) and 'protection' through providing them with health information	Desire to protect (others and oneself)	14F	Conclusiveness of test	If someone has a conclusive positive test the information is more relevant for other family members because they are at higher risk (as compared to inconclusive tests).	Relevance

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
						Similarly, conclusive negative tests may be relevant to other family members because it can alleviate worry	
13	Wanting to respect others need to 'not know' that information	Relates to the notion of 'right not to know'. If it is known (or assumed) that an individual would prefer to be ignorant of their genetic risk status then 'not telling' is reflecting a desire to protect them and respect them	Desire to protect (through respect)	15F	Mutation positive more likely to communicate than inconclusive or negative	A positive test result would have more serious implications for family members therefore would be communicated more because it is more relevant	Relevance
14	Age: being to young	Often children are told of their genetic risk status at an older age in an effort to 'preserve' their childhood and allow them to live a normal child's life without worries of morbidity or mortality. This reflects a caregivers desire to protect a child.	Desire to protect (other)	17F	Being a survivor of the disease	Likely, having experienced and survived the disease will alter ones perception of risk and severity of the disease. This in turn increases their perception of the relevance of the risk information to others who may be at increased risk.	Relevance
15	To immature (not able to comprehend information)	The maturity of person will affect their ability to comprehend the information and thus the relevance of that information for the person	Relevance	18F	Potentially curable disease	When there are interventions available to cure the disease (such as with breast cancer) it is often more to be spoken about because there is a greater sense of control over the disease since	Relevance

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
						action can be taken to prevent morbidity and death. This can make the information more relevant to certain family members compared to a disease where they can do nothing to prevent it	
16	Not wanting to spoil childhood and to protect them	Often children are told of their genetic risk status at an older age in an effort to 'preserve' their childhood and allow them to live a normal child's life without worries of morbidity or mortality. This reflects a caregivers desire to protect a child.	Desire to protect (other)	19F	Perception of relevance for members	If the information is perceived as relevant to another family member it is more likely to be communicated to them	Relevance
17	Guilt (survivor guilt, guilt for passing on a 'faulty gene', and guilt for making other family member feel guilt about their role in passing on the gene)	By not telling certain family members about their genetic status it helps avoids some of the negative repercussions such as oneself feeling guilt or playing a role in making another person feel guilty. Thus, not telling is reflecting a desire to protect that person or oneself.	Desire to protect (other or oneself)	20F	A brother is more likely to hear about a sisters BRCA1/2 result if the mutation is inherited through the paternal side (reflecting the myth that if its runs in the paternal line it has more relevance for men)	Even thought this information is incorrect it acts to facilitate communication because the 'teller' believes the information to be more relevant to male family members.	Relevance
18	Avoidance of resentment (when other doesn't have carriers status)	A desire to protect oneself from the negative emotional reactions of others or your own	Desire to protect (oneself)	21F	Strong family history	There is more fear and the information is perceived as more important	Relevance

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
		negative emotional reaction					
19	Denial	Denial allows a person to ignore information in an attempt to protect themselves from emotional pain they cannot handle	Desire to protect (oneself)	22F	Experienced cancer	Experience with a disease will affect perceptions of severity of disorder and likely increase the perception of relevance of the information for others who may be able to prepare for or prevent the disorder	Relevance
20	Avoidance of envy of sibling who is not carrier	A desire to protect oneself from the negative emotional reactions of others or your own negative emotional reaction	Desire to protect	25F	First degree relatives	Close family are often perceived to be more genetically linked than extended family, therefore, telling these members would be more important. Also, because they are close and the disorder might impact them directly (ie. Have to take care of them) which would make the information more relevant for them.	Relevance
26	Misunderstanding information about inheritance and myths	Misinformation will affect who the teller thinks the information is relevant for (i.e. the myth that breast cancer is only a female disease, may result in some male family members who are at risk not knowing their risk status)	Relevance	28F	Feeling it as an avenue to obtain social support	The desire to help oneself by obtaining social support from family members often acts as a facilitator of communication of genetic risk (often the people feel most socially close with are their first degree	desire to 'protect'

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
						relatives, thus also contributing to why first degree relatives are informed more often than distant)	
27	Not knowing who it is important to tell – thinking its only important for nuclear family	This kind of misinformation will affect who the teller things the information is relevant for (they may not tell extended family members who are also at increased risk)	Relevance	31F	X-linked inheritance (more likely to be discussed with females)	Because x-linked disorders appear more often in sons and the mother is usually a carrier with a defective X, if she passes on her defective X and the dad passes on a Y to create a son then the son will have the disorder – if the dad gives his X then the daughter will only be a carrier. Therefore, women may be perceived as more relevant to tell because it has implications for whether they have health children.	Relevance
28	Lay attitudes and beliefs about family	This is a cultural influence which inadvertently affects perceptions of who is at risk for disease (i.e. a son who behaves similarly to a father may be thought to get the disorder later in life if the father had it. Based only on the lay belief 'like father like son'	Relevance	32F	Older age of child	Coincides with life decisions where genetic information may be wanted and may help in planning for the future. The tendency for mothers to tell children when they are older reflects their perception of relevance of the information for their children.	Relevance

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
29	Fear of hearing information about risk (fear of disease blocks communication in family, fear of being responsible for problems in offspring)	Fear of the disease or reactions of others reflects a desire to protect oneself from emotional pain	Desire to protect (oneself)	34F	Relying on knowledge from the individual	Gaining more knowledge of the family history or how to deal with the disease is important to some who have undergone or will undergo genetic testing	Usefulness of communicating
30	Fear of conflict in the family	Reflects a desire to protect oneself and others from emotional pain and conflict	Desire to protect (oneself and others)	35F	enhance the probands status within the family (being the one to tell and help, making themselves seem less responsible for their disorder, or shifting focus onto how they are helping instead of an alcoholic)	Self explanatory	Desire to protect oneself
31	Fear of being blamed for disease or bringing bad news	Reflects a desire to protect oneself from emotional pain and conflict	Desire to protect				
32	Fear of psychological and social consequences of being tested positive	Reflects a desire to protect oneself and others from emotional pain and conflict	Desire to protect				
33	Anticipated loss of connectivity with family	Some families plagued by a disorder define their family and members in terms of their shared risk. Those who later find themselves not to be at risk often feel they not longer share that family bond and are no longer connected with their family. Therefore, it reflects a desire to	Desire to protect				

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
		protect oneself and others from emotional pain and conflict					
34	Anticipating the message would be rejected	If one anticipates that a family member will not accept a message, the act of communicating the risk would be useless.	Usefulness of communication				
35	Anticipated disinterest in testing	If one anticipates that a family member will not respond to the information in a health promoting or information obtaining manner, the act of communicating the risk may be perceived as useless	Usefulness of communicating				
37	No interventions to help once you know your risk	If there are no interventions for the family member to do anything about their risk, communication may be impeded in an attempt to prevent anxiety and distress to their family member for the time where they can do nothing.	desire to protect				
40	Inconclusive test results	Inconclusive test results are slightly more difficult to interpret because you are still at risk but not population risk and not the risk defined by the other mutations. This impacts people	Relevance				

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
		perceptions of relevance because often they mistakenly think they are in the clear.					
42	Non-carrier (HNPCC)	If one is found to be a non-carrier their perception of the relevance of their test result to others is lessened because there is nothing bad going to happen to children.	Relevance				
43	Clinical condition (life shortening, life threatening) – more likely to give selective information to kids	If the disorder is more severe full communication may be less likely in an effort to prevent the family member (often children and selective information sharing in this case) from emotional pain	Desire to protect				
45	Non fatal disease	Because death is not likely people may view the information as less relevant to discuss with others	relevance				
47	Perception of relevance for members is low	Self-explanatory (i.e. if the family member isn't having children yet then they don't need to know the information because there is no one at risk.)	Relevance				
48	No family history of disorder	Since the disorder is not prominent within the family (likely this is the first case) the person	Relevance				

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
		may think others risk is lower and the information is not as relevant for them.					
50	Relative didn't have access to testing (country or insurance)	Knowing that a family member may not be able to verify their risk the utility of telling that member may appear to be less.	Usefulness of communicating				
52	Believing they had already been informed	Mistakenly believing that family members already have been told about your result decreases perceptions of relevance of communicating	Relevance				
53	Personality characteristics of the person (emotionally fragile etc)	Knowing that a family member often does not deal well with bad information can prevent some from informing	Desire to protect				
54	Persons' situational factors (heroin abuse, mental illness, imprisonment, pregnant, marriage plans, old or ill)	This can relate to both a desire to protect someone who is already in a vulnerable life situation, and the relevance of the information for the person at the life-point they are at.	Desire to protect, relevance				
55	Unwilling to take responsibility for informing	Self-explanatory	Not feeling responsible				
56	Not feeling able to cope with others emotional reactions plus your own reactions	Predicting someone else's emotional reaction to the information results in some not disclosing	Desire to protect oneself				

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
		because they don't want to deal with that reaction					
59	Not wanting to increase worry without being able to offer concrete information or tangible answers.	Self - explanatory	Desire to protect				
60	Believe receiver will not perceive the information correctly [they will think inconclusive is no risk or at higher risk]	Misinterpretation of inconclusive test results as 'no risk' or very high risk has been documented. This can result in greater or lesser preventative activities or greater emotional distress. If a proband believes the person will misinterpret the result they may be less willing to tell in an effort to protect them.	Desire to protect				
62	Not sure whether the individual will want the news	Relates to the concept of 'right not to know'. If someone would prefer to live without knowledge of their genetic risk, not informing them is respecting their rights and protecting them from information they don't want to know.	Desire to protect or desire to respect wishes				
64	Preserve Hope that HD won't be part of the family	Receiving a definite test result takes away hope for life because you know you will eventually get the disease and die.	Desire to protect				

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
		In this case, it is sometimes better to be left in the unknown to preserve a little hope.					
67	Retaining control over family members lives	It has been documented that some members will not tell others because they think they can predict what their 'choices' (such as abortion or not getting married) will be afterward. In not telling, they prevent the person from making a 'bad decision' which they think will eventually hurt the person. Ultimately, it is a mechanism of protection.	Desire to protect				
68	Older mothers less likely to inform their mothers		Desire to protect & relevance				
69	Unpleasantness of message	relates to wanting to protect them	Desire to protect				
72	Someone in the family has died from the disease.	After death of a family member, often emotions are still running high especially about the subject of the disorder that the member died from. If the person is perceived to be too emotional to handle the results of the genetic test result then the information may be	Desire to protect (from emotional pain)				

#	Barriers from review	Elaboration	Underlying theme	#	Facilitators from review	Elaboration	Underlying theme
		withheld to protect them.					
73	Wanting to protect their own privacy and confidentiality	Relates to the desire to protect oneself (either from others judgment, or just maintaining their sense of rights to information of oneself)	Desire to protect				
74	Unsure of the family members reaction	Their reaction could lead to pain, stress for the teller and/or the receiver of the information	Desire to protect (others and oneself)				
75	Viewing the act of disclosure as a burden to oneself	Self explanatory	Desire to protect (oneself)				
77	Not feeling it is their responsibility to pass on the information	Self explanatory	Not feeling responsible				

2) SUBJECTIVE NORM FACTORS

#	Barrier from review	Elaboration	Underlying theme	#	Facilitator from review	Elaboration	Underlying theme
3	Openness about discussing disease in society	Societal pressures and norm can influence the likelihood of discussion on certain topics	Pressure from society	6F	Conflicting roles of women: kin keeper (protector), health promoters, and support seekers	Pressures from society and families to fulfill traditional women's roles as a communicator and kin keeper.	Pressure from family and society
4	Taboo and shame	Societal and family norm can influence the likelihood of discussion on certain topics	Pressure from family and society	23F	External cues – like Dr's telling you to do it	This relates to pressure from professionals to disclose	Pressure from Genetic counselors and professionals
38	Pressure from others not to tell	This relates to pressures from family members to behave a certain way	Pressure from family members	24F	Counselor's role	This relates to pressure from professionals to disclose	Pressure from genetic counselors and professionals
39	One person acts as a 'banner' of communication , or discourages others to disclose	This relates to pressures from family members to behave a certain way	Pressure from Family members	30F	Someone who takes the leadership communicator role or a key family member	Roles within families can influence whether there is a 'communicator' or not and who tells what information.	Pressure from families
70	Not feeling they have the authority (inappropriate to tell nieces and nephews and cousins without parents)	Roles within families can influence whether there is a 'communicator' or not and who tells what information.	Pressure from family member not to take that role because it is theirs.				

3) PERCEIVED BEHAVIOURAL CONTROL FACTORS

	Barrier from Review	Explanation	Underlying theme		Facilitator from review	Explanation	Underlying theme
21	Unhealthy family relationships	estranged relatives and conflicts between relatives can alter whether a person will allow themselves to be contacted or whether an individual can actually contact a member	Family Dynamics/ relationships	9F	Close social relationship		Family Dynamics/ relationships
22	Lack of contact with person (or not knowing how to contact)	Often in families there are extended relatives where there is little contact and one does not know how to get a hold of this person or where they live any more	Family Dynamics/ relationships	14F	Conclusiveness of test	Less complex and easier to explain	Ability to understand
23	Geographical distance	If a family member lives far away geographically it is less likely that this type of information will be communicated to them. Partially because of lack of closeness but also because some see this as information which should be communicated in a face-to-face manner	Family dynamics/ relationships	16F	Cancer is already open in the family	It is easier to talk about the subject because it has already been spoken about within the family. This factor boils down to how the family interacts and deals with health issues, and whether their communication about such issues	Family Dynamics/ relationships

	Barrier from Review	Explanation	Underlying theme		Facilitator from review	Explanation	Underlying theme
						is open.	
24	Adoption	If the communicator is adopted they do not know how to get a hold of family etc.	Family dynamics/ relationships	25F	First Degree Family Member	This reflects the social closeness of family members and the regular communication and information sharing that occurs in families.	Family Dynamics/ relationships
25	Superficial relationship or not being close/not emotionally close	An individual may not feel capable of having a potentially emotional discussion with someone they don't feel close to. Or want to approach someone when the conversation will be uncomfortable. Most likely due to existing relational habits (ie. Not talking about stuff)	Family dynamics/ relationships	26F	Generally good communication, and supportive, and take each others advice, and generally get along		family dynamics
40	Inconclusive test results (uncertainty)	The genetics and explanation of inconclusive test results are more complicated than conclusive results. Not being able to fully understand what the result means for the proband will affect	Ability to understand/ Communication skills	27F	Open communication style in family (dislike of secrecy)		Family dynamics

	Barrier from Review	Explanation	Underlying theme		Facilitator from review	Explanation	Underlying theme
		communication to others					
44	Balanced translocations	The genetics behind some disorders is very complicated and difficult to understand and explain.	Ability to understand, Communication skills	32F	Older age of offspring	Often children are not told of their risk status because they are too young to understand or grasp the implications of the risk. Therefore, as a child gets older the likelihood of disclosure increases	Ability to Understand
46	Complexity of information	The genetics behind some disorders is very complicated and difficult to understand and explain	Ability to Understand, Communication skills	33 F	Being a non-obligate carrier (ie. brother has disorder – haemophilia)	Because the brother has the illness, the management of the disorder and implications of the disorder are open and visible. Questions regarding the disorder are likely to occur and the sibling is likely to know about inheritance etc	Family Dynamics/ Relationships
57	Having difficulty coping with own problems/diagnosis/test result	Some people do not discuss initially because they themselves cannot	Coping skills				

	Barrier from Review	Explanation	Underlying theme		Facilitator from review	Explanation	Underlying theme
		handle the information and just 'can not' do it					
58	Not knowing what to say	Self-explanatory	Communication skills				
66	Don't think they have communication skills (or do not communicate in general)	Self-explanatory	Communication skills				
74	Being an obligate carrier (ie. father has the disease)	Fathers tended to talk to their daughters less about their disorder, so the carrier status of the females was not discussed	Communication skills, Family Dynamics/ Relationships				

D) OTHER INFLUENCIAL VARIABLES AND DEMOGRAPHIC VARIABLES

	Barrier From Review	Notes		Facilitator from Review	Notes
1	Cultural Norms		1F	Cultural Norms	
23	Geographical distance	Does not fit into the TPB and would be interesting to investigate whether it is actually physical distance or social closeness which affects communication	2F	Media	Relates to knowledge and awareness, can assess whether they have been exposed a lot or not.
36	Being male	May impede communication (ability to cope with test result not as good as women, harder to communicate a result to a male) Plan on investigating whether this is true in our population	11F	Being Female (daughters, mothers, sisters, nieces, maternal relatives)	May facilitate communication. Plan on investigating if it has to do with social closeness, support, or gender roles
41	Mutation positive or carrier positive	Plan on investigating whether communication problems are associated more or less with each status	20F	For males – if the mutation is in their family line it makes it easier for them to talk	May facilitate communication. Probably won't investigate because it is not very common in literature
49	Being emotionally motivated person.	This is a reflection of personality	29F	Lay attitudes and beliefs about family (culture)	
51	Bad first Attempt at disclosure	Past Behaviour			
63	Bad lived experiences with Cancer	Perception of disease severity			
65	Higher education less likely to disclose to children	Check to see if education influences any factors.			
71	It never came up	Reflects ambivalence which is possibly another construct in the TPB therefore it will be measured in the demographics section.			

Table 3: Step 3 – Organization of Factors under its Underlying Theme within the TPB constructs

ATTITUDE THEMES				
Desire to protect oneself and others	**Perception of Relevance**	**Perception of Responsibility**	**Evaluation of 'Right to know'**	**Usefulness of Communicating**
2. Fear of insurance / employment discrimination, or stigmatization of a family member	15. To immature to comprehend the information	55. Unwilling to take responsibility for informing	**3F. Acting honorably and respectfully towards children**	34. Anticipating the message would be rejected
5. Different coping styles need to verify in original papers	26. Misunderstanding information about inheritance and myths	77. Not feeling it is their responsibility to tell others	**4F. Feeling they have the right to know the information**	35. Anticipated disinterest in testing
6. Conflicting views about genetic testing, or communication of family history or abortion		**5F. Having a feeling of responsibility, duty to inform or moral obligation to tell**		50. Relative doesn't have access to testing (not available in their country, do not have insurance)
7. Different desire for information				**34F. Relying on family members for information on family history or disease management etc.**
8. Wish to protect and preserve a relationship				

ATTITUDE THEMES

Desire to protect oneself and others	Perception of Relevance	Perception of Responsibility	Evaluation of 'Right to know'	Usefulness of Communicating
9. protection of their feelings, not wanting to upset someone (yet or at all)	27. Not knowing who it is important to tell – thinking its only important for the nuclear family			
10. Information perceived as too emotional	28. Lay attitudes and beliefs about family			
11. Moral dilemma: conflict between wanting to protect and wanting to help	40. Inconclusive test results			
12. Conflicting roles of women: kin keeper (protector), health promoters (protector), support seekers	42. Non-carrier of the mutation			
13. Wanting to respect others need to 'not know' that information	45. Non fatal disease			
14. Recipient of information is to young	47. Perception of relevance for members is low			
16. Not wanting to spoil childhood and to protect them	48. No family history of disorder			
17. Guilt (survivor guilt, guilt for passing on a 'faulty gene', and guilt for making other family members feel guilt about their role in passing on the gene)	52. Believing they have already been informed			
18. Avoidance of resentment (when the other individual	**8F. Appropriate time to tell individual (i.e. Sexually**			

ATTITUDE THEMES

Desire to protect oneself and others	Perception of Relevance	Perception of Responsibility	Evaluation of 'Right to know'	Usefulness of Communicating
doesn't have carrier status)	**active, stable relationship)**			
19. Denial	**12F. Older mothers more likely to inform daughters**			
20. Avoidance of envy of a sibling who is not a carrier	**14F. Conclusiveness of test**			
29. Fear of hearing information about risk (fear of disease blocks communication in family, fear of being responsible for problems in offspring)	**15F. Mutation positive more likely to communicate than inconclusive or negative**			
30. Fear of conflict in the family	**17F. Being a survivor of the disease**			
31. Fear of being blamed for disease or bringing bad news	**18F. Potentially curable disease**			
32. Fear of psychological and social consequences of being tested positive	**19F. Perceptions of relevance for members**			
33. Anticipated loss of connectivity with family	**20F. A brother is more likely to hear about a sisters BRCA1/2 result if the mutation is inherited through the paternal side (reflecting the myth that if it runs in the paternal line it has more relevance for men)**			
37. No interventions to help once you know your risk	**21F. Strong family history**			
43. Clinical condition (life shortening, life threatening)	**22F. Experienced cancer**			
53. Personality characteristics of the	**25F. First degree relatives**			

ATTITUDE THEMES

Desire to protect oneself and others	Perception of Relevance	Perception of Responsibility	Evaluation of 'Right to know'	Usefulness of Communicating
recipient (emotionally fragile etc)				
54. Persons situational factors (heroin abuse, mental illness, imprisonment, pregnant, marriage plans, old or ill)	**13F. Siblings, spouses and GPs are more likely to be told**			
56. Not feeling able to cope with others emotional reactions plus your own reactions to the result	**31F. X-linked inheritance (more likely to be discussed with females)**			
59. Not wanting to increase worry without being able to offer concrete information or tangible answers	**32F. Older age of offspring**			
60. Believe the receiver of the information will not perceive the information correctly (they will think inconclusive is not risk or a much higher risk)				
62. Not sure whether the individual will want the news				
64. Preserve hope that HD won't be part of the family				
67. Retaining control over family members lives				
68. Older mothers are less likely to inform their mothers				
69. Unpleasantness of message				
72. Someone in the family				

ATTITUDE THEMES

Desire to protect oneself and others	Perception of Relevance	Perception of Responsibility	Evaluation of 'Right to know'	Usefulness of Communicating
has died from the disease				
73. Wanting to maintain your privacy and confidentiality				
74. Unsure of the family members reaction to the information				
75. Viewing the act of disclosure as a burden to oneself.				
7F. Anticipated regret				
6F. Conflicting roles of women: kin keeper, health promoter and support seeker				
28F. Feeling it is an avenue to obtain social support				
35F. enhance the probands status within the family				

SUBJECTIVE NORM THEMES		
Pressure from Family Members	**Pressure from Professionals**	**Pressure from Society**
4. Taboo and Shame	**23F. External cues: a Dr. telling you it is important to communicate to certain people**	3. Openness about discussion disease in society
38. Pressure from others not to tell	**24F. Counselor's role**	4. Taboo and Shame
39. One person acts to 'ban' communication or discourages others to disclose		***6F. Conflicting roles of women***
70. Not feeling they have the authority (appropriate to tell nieces and nephews and cousins without parents)		
6F. Conflicting roles of women		
30F. Someone who takes the leadership communicator role or a key family member		

PERCEIVED BEHAVIOURAL CONTROL THEMES			
Family Dynamics and Relationships	**Communication Skills**	**Ability to Understand**	**Coping Skills**
21. Unhealthy family relationships	58. Not knowing what to say	40. Inconclusive test results	57. Having difficulty coping with own problems/diagnosis/test result
22. Lack of contact with person (or not knowing how to contact)	66. Don't think they have communication skills (or do not communicate effectively in general)	44. Balanced translocations	
23. Geographical distance	74. Being a female obligate carrier (ie. father has disease)	46. Complexity of information	
24. Adoption		**14F. Conclusiveness of test**	
25. Superficial relationship or not being close/not emotionally close		**32F. Older age of offspring**	
74. Being an obligate carrier (ie. father has the disorder - haemophilia)			
9F. Close social relationship			
16F. Cancer is already open in the family			
25F. First degree family member			
26F. Generally good communication, supportive and take each others advice, and generally get along.			
27F. Open communication in family (dislike of secrecy)			
34F Being a non-obligate carrier (ie. brother has disorder – haemophilia)			

APPENDIX I: Participant Invitation Letter (English and French)

Children's Hospital of Eastern Ontario
Centre hospitalier pour enfants de l'est de l'Ontario

[Date] ID # _____

[Name of potential participant]
[Address 1 of potential participant]
[Address 2 if required]
[City, province, postal code]

Dear [Name of potential participant],

I am writing to you as the Chief of the Division of Genetics to tell you about a research study based at the Children's Hospital of Eastern Ontario (CHEO). The project is about how families talk about genetic test results. It has been approved by the CHEO Research Ethics Board. The research team is led by Dr. Brenda Wilson from the University of Ottawa. Christina Honeywell, a genetic counsellor from CHEO, is also one of the investigators.

They are interested in how people talk about genetic test results with their family. They would like to know what makes it hard to talk about genetic test results with different family members. CHEO helped the research team find people for the study. The staff at CHEO used the hospital database to create a list of people who received counseling between 1997 and 2007. This is how you have been identified. The list contains your name and address only. The research team will never be given your name or address.

The study is a short English language survey which is included in this package. The 'Participant Information Form' gives more details about the project and explains why this survey is not yet available in French. Please read the information form before deciding whether or not to take part.

If you would like to take part, please fill in the survey and return it to Miriam Wiens in the postage-paid envelope provided. Please keep the information form.

401 Smyth Road, Ottawa, ON K1H 8L1, Canada
Tel: (613) 737-7600 www.cheo.on.ca

Making a difference in the lives of children and youth

401, chemin Smyth, Ottawa (ON) K1H 8L1, Canada
Tél. : (613) 737-7600 www.cheo.on.ca

Faire une différence dans la vie des enfants et des adolescents

.../2

We understand if you do not want to take part. If this is the case, you can:

1. Return a blank survey in the envelope provided – we will take you off the mailing list for this study, or

2. Phone Miriam Wiens (research coordinator from the University of Ottawa) at (613)562-5800 ext 8716 – we will take you off the mailing list for this study, or

3. Ignore this letter and the reminders that follow. We will only send you three letters. If you ignore all three you will be taken off the mailing list for this study.

Even if you do not take part, please keep the information form.

You are under no obligation to the Children's Hospital of Eastern Ontario or the University of Ottawa. Thank you for taking the time to read this letter.

Yours Truly,

Dr. Judith Allanson MD
Chief, Division of Genetics
Children's Hospital of Eastern Ontario

Children's Hospital of Eastern Ontario
Centre hospitalier pour enfants de l'est de l'Ontario

[Date] N° d'id. _____

[Nom du participant potentiel]
[Adresse 1 du participant potentiel]
[Adresse 2 au besoin]
[Ville, province, code postal]

Madame, Monsieur,

Je vous écris à titre de chef de la Section de génétique pour vous parler d'un projet de recherche effectué au Centre hospitalier pour enfants de l'est de l'Ontario (CHEO). Ce projet porte sur la manière dont les familles parlent des résultats de tests de génétique. Il a été approuvé par le Conseil d'éthique en recherche du CHEO. L'équipe de chercheurs est dirigée par la D^re^ Brenda Wilson, de l'Université d'Ottawa. Christina Honeywell, conseillère en génétique du CHEO, fait partie des chercheurs.

On s'intéresse à la manière dont les gens parlent des résultats de test de génétique au sein de leur famille. On aimerait savoir ce qui rend si difficile la tâche de parler des résultats de test de génétique pour les différents membres de la famille. Le CHEO a aidé l'équipe de chercheurs à recruter des participants à l'étude. Le personnel du CHEO a utilisé la base de données de l'hôpital pour préparer une liste de gens qui ont reçu des services de counseling entre 1997 et 2007. C'est pour cette raison que vous avez été identifié. La liste contenait votre nom et votre adresse seulement. L'équipe de chercheurs ne sera jamais mise au courant de votre nom ou de votre adresse.

Cette étude consiste en un court questionnaire en anglais inclus à cette trousse. Le formulaire d'information pour les participants fournit de plus amples détails sur le projet et explique pourquoi le questionnaire n'est pas encore disponible en français. Veuillez lire le formulaire d'information avant de décider si vous désirez ou non y participer.

Si vous souhaitez participer, veuillez remplir le questionnaire et le renvoyer à Miriam Wiens dans l'enveloppe affranchie fournie à cet effet. Veuillez conserver le formulaire d'information.

Nous comprenons que vous ne voudrez peut-être pas y participer. Dans ce cas, vous pouvez :

4. renvoyer un questionnaire en blanc dans l'enveloppe fournie, et nous vous retirerons de la liste d'envoi de cette étude, ou

5. téléphoner à Miriam Wiens (coordinatrice de la recherche à l'Université d'Ottawa), au 613-562-5800, poste 8716 et nous vous retirerons de notre liste d'envoi, ou

6. ne pas tenir compte des envois – nous ne vous enverrons que trois questionnaires au total puis nous vous retirerons de notre liste.

Même si vous n'y participez pas, veuillez conserver le formulaire d'information.

Vous n'avez aucune obligation envers le Centre hospitalier pour enfants de l'est de l'Ontario ou l'Université d'Ottawa. Merci d'avoir pris le temps de lire cette lettre.

Veuillez agréer mes salutations les meilleures.

D^{re} Judith Allanson MD
Chef, Section de génétique
Centre hospitalier pour enfants de l'est de l'Ontario

APPENDIX J: Information form for participants (English and French)

Family Communication about Genetic Disorders

Investigators:
Dr. Brenda Wilson, Associate Professor, Faculty of Medicine, Department of Epidemiology and Community Medicine, University of Ottawa, brenda.wilson@uottawa.ca, (613) 562-5800 ext. 8261.

Christina Honeywell, Genetic Counselor, CHEO, Division of Genetics, Assistant Professor, Faculty of Medicine, Department of Pediatrics, University of Ottawa, CHoneywell@cheo.on.ca, (613) 737-7606 ext. 3474.

PARTICIPANT INFORMATION FORM

You are being invited to take part in a research project. This information form describes the study and what will be asked of you if you agree to take part.

Purpose of the Research

Genetic testing is now available for many genetic disorders. If a genetic disorder is found in one person some of their family members may be at risk for the disorder too. However, many families find it hard to discuss genetic test results. We want to find out more about this. We also want to work out ways that will help people talk to their family about genetic test results.

The purpose of this research is to understand how people communicate with family members about genetic test results. This information will be used to help people who want to talk to family members but find it hard.

Description of Participant Requirements

If you agree to take part in this project, you will be asked to complete the short survey. The survey is included with this package. It should take, at most, 20 minutes to complete. The questionnaire will ask about your feelings and experience with talking to different family members about your genetic information. We are trying to understand what makes talking about genetic test results hard. If you would like to take part, fill in the survey by yourself. Mail it back to us in the postage-paid envelope included in this package.

We regret that we are unable to offer a French version of this survey. It is very important to our study that the questions we ask you are clear in their meaning and have a scientific basis. This allows us to analyze the answers in a scientifically appropriate way. Unfortunately, surveys do not always translate well between different languages and cultures. The technique for this survey is based on an approach developed in English. However, it is beyond the scope of this project to produce a version in French to the same standard. We do not wish to exclude the Francophone community. If you feel you understand the English version, we would like you to take part. You are welcome to complete any written answers in French. We apologize if you are unable to understand the English version and thank you for your time in reading this material.

If we do not receive the survey back from you in about two weeks, we will send you a letter and another copy of the same survey. If you do not want to participate, you can:

(i) return a blank questionnaire – we will take you off our mailing-list; OR

(ii) phone Miriam Wiens (research coordinator from The University of Ottawa), at (613) 562-5800 ext 8716, we will take you off our mailing-list ; OR

(iii) ignore the mailings – we will only send you three surveys in total and then take you off our list.

Potential Risks and Benefits

Information about you will be kept confidential at all points in the study. To safeguard your confidentiality, no information which could lead to your identification will leave the CHEO genetics clinic. All information that comes to us from CHEO will have details about who you are removed before they send it. This means that it will be totally anonymous.

There is a small chance that some of the survey questions may cause you distress, because it requires you to think about you and your family's experience with illness. Dr. Mario Cappelli, a CHEO psychologist, will provide mental health services to any participant who needs such help.

There are no direct benefits from taking part in this study. However, your opinions may be used to help individuals and families who will use genetic services in the future.

Withdrawing from the Project

You are under no obligation to take part in this study. Your decision to participate or not in this study will not affect the care you receive at CHEO. You may withdraw from the project at any time without fear of penalties to you.

Confidentiality

Because we will not have your name, your identity will not be revealed to the research team or in any publication or presentation of this study. Any personal information about you that leaves the hospital will converted into a code so you cannot be identified by name.

If you choose to take part, when your questionnaire is received, your responses will be entered into a computer for analysis. Since the questionnaire information is coded, personal details which could identify you (such as your name) will not appear in the computerized data. The surveys themselves will be kept in a secure location. Only the investigators and their research team will have access to the surveys. Once the study is done, the codes will be permanently removed from computerized data so that they can never be linked with the individual participants.

Questions
If you have any questions about the research project, you may contact the researchers at the emails or phone numbers at the top of the page 1. They will be happy to talk to you about this study.

The Research Ethics Board (REB) is a group of people from scientific and non-scientific backgrounds that reviews research studies. Its goal is to ensure the protection of the rights and welfare of people involved in research. You may contact the Chair of the REB for information regarding patient's rights in research studies at 613-737-7600 x 3272, although this person cannot provide any health-related information about the study.

By completing and returning the survey you are consenting to have read this information sheet.

Communications familiales concernant les troubles génétiques

Chercheurs :
D[re] Brenda Wilson, professeure agrégée, Faculté de médecine, Département d'épidémiologie et de médecine sociale, Université d'Ottawa, brenda.wilson@uottawa.ca, 613-562-5800, poste 8261.

Christina Honeywell, conseillère en génétique, CHEO, Section de génétique, professeure adjointe, Faculté de médecine, Département de pédiatrie, Université d'Ottawa, CHoneywell@cheo.on.ca, 613-737-7606, poste 3474.

FORMULAIRE D'INFORMATION POUR LES PARTICIPANTS

On vous demande de bien vouloir participer à un projet de recherche. Ce formulaire d'information décrit l'étude et c'est ce qu'on attendra de vous si vous acceptez d'y prendre part.

Objectif de la recherche

Des tests génétiques sont maintenant disponibles pour dépister de nombreux troubles génétiques. Si un trouble génétique est diagnostiqué chez une personne de la famille, certains des autres membres de cette famille courent également le risque de présenter ce trouble. Cependant, de nombreuses familles ont de la difficulté à discuter des résultats de tests génétiques. Nous voulons nous renseigner davantage à ce sujet. Nous voulons aussi trouver des moyens d'aider les gens à parler aux membres de leur famille des résultats de tests de génétique.

L'objectif de ce projet de recherche est de comprendre comment les gens communiquent avec les membres de leur famille concernant les résultats de test de génétique. Ces informations seront utilisées pour aider les gens qui veulent à parler aux membres de leur famille mais ont de la difficulté à le faire.

Description des exigences pour les participants

Si vous acceptez de participer à ce projet, on vous demandera de remplir un court questionnaire. Le questionnaire est inclus à cette trousse. Il devrait vous prendre au plus 20 minutes à remplir. Le questionnaire vous pose des questions concernant vos sentiments et vos expériences en matière de discussion des informations génétiques avec les différents membres de votre famille. Nous tentons de comprendre ce qui rend si difficile la discussion des résultats de test génétique. Si vous voulez y participer, répondez aux questions par vous-même. Renvoyez-nous le questionnaire par la poste dans l'enveloppe affranchie inclus dans cette trousse.

Nous regrettons de ne pas être en mesure de vous offrir une version française de ce questionnaire. Il est très important pour notre étude que les questions posées aient une signification claire et soient fondées sur des bases scientifiques solides. Ceci nous permet d'analyser les réponses de manière scientifique appropriée. Malheureusement, les questionnaires ne se prêtent pas toujours à une bonne traduction entre les différentes langues et les différentes cultures. La technique utilisée pour ce questionnaire a été développée en anglais. De plus, préparer une version française du même niveau de qualité dépasse la portée de ce projet. Nous ne souhaitons pas exclure la communauté francophone. Si vous pensez comprendre la version anglaise nous vous demandons de bien vouloir participer au sondage. Vous pouvez si vous le désirez répondre fournir des réponses écrites en français. Nous vous prions de nous excuser si vous ne comprenez pas la version anglaise et vous remercions de votre temps passé à lire ce matériel.

Si nous ne recevons pas votre questionnaire dans environ deux semaines, nous vous enverrons une lettre et un autre exemplaire du même questionnaire. Si vous ne voulez pas y participer, vous pouvez :

(iv) renvoyer le questionnaire non rempli – nous vous retirerons de notre liste d'envoi, OU

(v) téléphoner à Miriam Wiens (coordinatrice de la recherche à l'Université d'Ottawa), au 613-562-5800, poste 8716 et nous vous retirerons de notre liste d'envoi, OU

(vi) ne pas tenir compte des envois – nous vous enverrons que trois questionnaires au total puis nous vous retirerons de notre liste.

Risques et avantages possibles

Les informations vous concernant seront conservées de manière confidentielle à toutes les étapes de l'étude. Pour sauvegarder le caractère confidentiel de vos renseignements personnels, aucune information pouvant aboutir à votre identification ne quittera la clinique de génétique du CHEO. Toutes les informations nous provenant du CHEO ne comporteront aucun détail vous concernant puisque toutes les informations personnelles seront retirées avant l'envoi. Ceci signifie qu'elles seront entièrement anonymes.

Il y a un petit risque que certaines des questions du questionnaire vous causent de la détresse, parce qu'elles vous obligeront à réfléchir à votre expérience et à l'expérience de votre famille face à la maladie. Le D[r] Mario Cappelli, psychologue du CHEO, fournit des services de santé mentale à tous les participants ayant besoin d'une telle aide.

Participer à cette étude ne vous procurera aucun avantage direct. Cependant, vos opinions pourraient être utilisées pour aider les personnes et les familles qui auront recours à des services de génétique à l'avenir.

Retrait du projet
Vous n'êtes aucunement obligé de participer à cette étude. Votre décision de participer ou non à cette étude n'affectera pas les soins que vous recevez au CHEO. Vous pouvez vous retirer du projet à n'importe quel moment sans que ceci entraîne des pénalités pour vous.

Confidentialité
Comme nous n'aurons pas votre nom, votre identité ne sera pas révélée à l'équipe de chercheurs ni dans aucune publication ou présentation reliée à cette étude. Toutes les informations personnelles vous concernant quittant les murs de l'hôpital seront converties en un code pour que l'on ne puisse pas vous identifier par votre nom.

Si vous choisissez de participer à cette étude, quand on recevra votre questionnaire, vos réponses seront entrées en ordinateur pour analyse. Comme les informations du questionnaire sont codées, les détails personnels qui pourraient permettre de vous identifier (comme votre nom) ne paraîtront pas dans les données informatisées. Les questionnaires seront conservés en lieu sûr. Seuls les chercheurs et l'équipe de recherche auront accès aux questionnaires. Une fois l'étude terminée, les codes seront retirés en permanence des données informatisées pour qu'elles ne puissent jamais être reliées aux participants individuels.

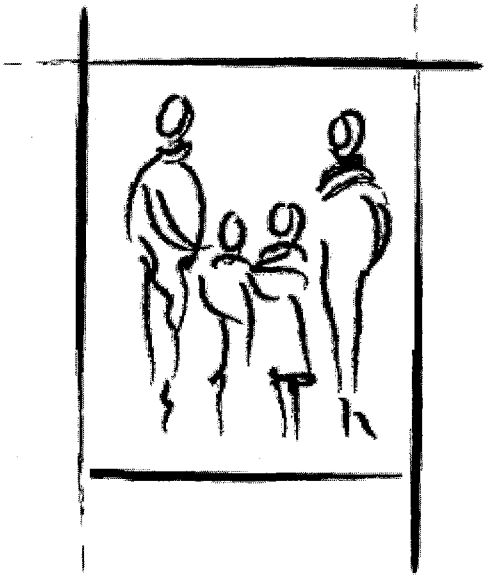
Questions
Si vous avez des questions à poser concernant ce projet de recherche, vous pouvez contacter les chercheurs aux adresses courriel ou numéros de téléphone indiqués en haut de la page 1. Ces personnes se feront un plaisir de vous parler de cette étude.

Le Conseil d'éthique de la recherche (CER) est un groupe de personnes de formation scientifique et non scientifique qui passe en revue les projets de recherche. L'objectif est de veiller à protéger les droits et le bien-être des personnes participant aux projets de recherche. Vous pouvez contacter le président du Conseil d'éthique de la recherche pour obtenir des informations concernant les droits des patients participant à des projets de recherche en composant le 613-737-7600, poste 3272, bien que cette personne ne soit pas en mesure de vous fournir des informations reliées à la santé concernant cette étude.

En remplissant et en renvoyant le sondage, vous confirmez que vous avez lu ce feuillet d'information.

APPENDIX K: Family Communication and Genetics Survey

FAMILY COMMUNICATION & GENETICS STUDY



GENESENS
DE LA DÉCOUVERTE À LA SANTÉ
TRANSLATING DISCOVERIES INTO HEALTH

CHEO Children's Hospital of Eastern Ontario
Centre hospitalier pour enfants de l'est de l'Ontario

uOttawa
L'Université canadienne
Canada's university

If you prefer to do an electronic version of this survey go to:

www.fam-com.com

Your password is: communication
Your username is your three digit id code to the right ➔

We have sent you this survey because you had a test for myotonic dystrophy. This blood test might have been any time in the last 10 years. People have these tests for different reasons – sometimes because they are showing symptoms and sometimes because other people in their family have myotonic dystrophy.

We do research projects on how genetic information affects family life. In this study, we want to learn about how easy or hard it is for people to talk to other family members about genetic tests.

This survey has questions about talking to, and not talking to, family members about genetic test results. There are no right or wrong answers. We are trying to learn about your opinions and experiences. We believe that information like this will help us design better services in the future for people who have genetic testing.

As well as answering the questions, feel free to tell us other things about your experiences with your family and testing if you wish. You can write extra comments on the back of the survey.

We would like to thank you in advance for taking the time to answer this survey.

GENESENS

If you prefer to do an electronic version of this survey go to:

www.fam-com.com

Your password is: communication
Your username is your three digit id code on the cover page.

SECTION 1: INTRODUCTION

Please read and answer each question, even if it seems similar to one you have already answered.

PLEASE FOLLOW THESE 2 STEPS:

STEP 1: Look at the list of relatives in **Box A**. Put a check (✓) beside all the people you **DID NOT TELL** your myotonic dystrophy test result to.

Cross out any relatives that you do not have.

(For example, I have no niece, so I would cross it off the list: ~~☐ at least one niece~~).

Box A

☐ mother	☐ at least one sister	☐ at least one daughter	☐ at least one aunt	☐ at least one niece	☐ at least one cousin (female)
☐ father	☐ at least one brother	☐ at least one son	☐ at least one uncle	☐ at least one nephew	☐ at least one cousin (male)

STEP 2: Now look at the list of relatives in **Box B**. Put a check (✓) beside all the people you **DID TELL** your myotonic dystrophy test result to.

Cross out any relatives that you do not have.

Box B

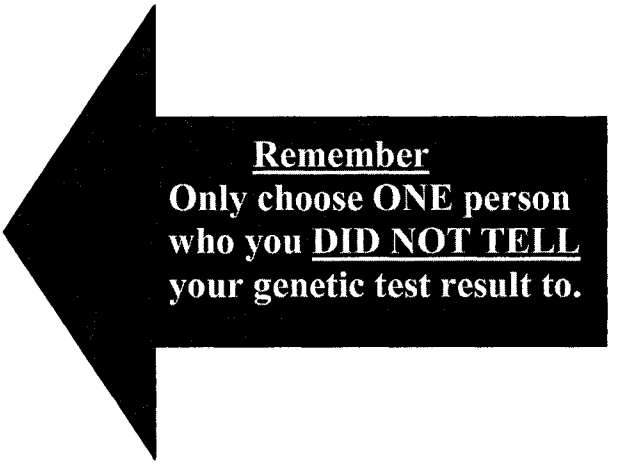
☐ mother	☐ at least one sister	☐ at least one daughter	☐ at least one aunt	☐ at least one niece	☐ at least one cousin (female)
☐ father	☐ at least one brother	☐ at least one son	☐ at least one uncle	☐ at least one nephew	☐ at least one cousin (male)

SECTION 2: QUESTIONS ABOUT A PERSON YOU HAVE NOT TOLD

- Look back at **Box A**. Choose **ONE PERSON ONLY** you put a check beside. Answer the questions in Section 2 thinking about this person.

The person I have chosen to answer Section 2 about is:

- ☐ my mother
- ☐ my father
- ☐ a sister
- ☐ a brother
- ☐ a daughter
- ☐ a son
- ☐ an aunt
- ☐ an uncle
- ☐ a niece
- ☐ a nephew
- ☐ a cousin (female)
- ☐ a cousin (male)



If you did not check anyone in BOX A, go straight to SECTION 3.

Please circle the number from 1 to 7 that is closest to your opinions or feelings about each statement (example: if you feel that talking to your family member about things that are important for them to know is 'VERY UNDESIRABLE' you would circle the number 1).

For me, talking to my family member about things that are important for them to know would be:

Very Undesirable 1 2 3 4 5 6 7 Very Desirable

For me, talking to my family member about my genetic test result is:

Very Difficult 1 2 3 4 5 6 7 Very Easy

I do not feel emotionally close with my family member.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

My genetic counsellor expects me to talk to my family member about my genetic test result.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

Talking to my family member about my genetic test result is my responsibility.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

I will talk to my family member about my genetic test result.

Very Unlikely 1 2 3 4 5 6 7 Very Likely

If I found genetic risk information hard to understand it would be _____ to talk to my family member about my genetic test result.

More Difficult 1 2 3 4 5 6 7 Less Difficult

I live far away from my family member.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

Talking to my family member about my genetic test result would give them information that would be useful for them when making their own decisions about their health.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

What my genetic counsellor thinks I should do matters to me.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

I am confident I could talk to my family member about my genetic test result.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

I plan to talk to my family member about my genetic test result.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

I feel under social pressure to talk to my family member about my genetic test result.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

The decision to talk to my family member about my genetic test result is out of my control.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

For me, telling my family member information that is useful for them when making health decisions is:

Very Undesirable 1 2 3 4 5 6 7 Very Desirable

I do not get along with my family member.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

For me, talking to my family member about my genetic test result would be:

a) Very Harmful (for me) 1 2 3 4 5 6 7 Very Beneficial (for me)

b) Very Comfortable (for me) 1 2 3 4 5 6 7 Very Uncomfortable (for me)

c) Very Frustrating (for me) 1 2 3 4 5 6 7 Not at all Frustrating (for me)

d) Very Useful (for me) 1 2 3 4 5 6 7 Not at all Useful (for me)

People who are important to me want me to talk to my family member about my genetic test result.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

It is expected of me that I talk to my family member about my genetic test result.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

For me, causing a lot of worry and concern for my family member is:

Very Undesirable 1 2 3 4 5 6 7 Very Desirable

For me, carrying through my responsibilities is:

Very Undesirable 1 2 3 4 5 6 7 Very Desirable

If I told my family member about my genetic test result I believe it would cause him or her a lot of worry and concern.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

If my family member was in denial about his or her genetic risk it would make it ____ to talk to them.

More Difficult 1 2 3 4 5 6 7 Less Difficult

If I did not get along with my family member it would make it _____to talk to them.

More Difficult 1 2 3 4 5 6 7 Less Difficult

Genetic risk information is hard to understand (for me).

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

I intend to talk to my family member about my genetic test result.

Very Unlikely 1 2 3 4 5 6 7 Very Likely

My family member is in denial about his or her genetic risk.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

If I was not emotionally close with my family member it would make it ____to talk to them about my genetic test result.

More Difficult 1 2 3 4 5 6 7 Less Difficult

If I was living far away from my family member it would make it _____to talk to them about my genetic test result.

More Difficult 1 2 3 4 5 6 7 Less Difficult

My genetic test result is important for my family member.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

Please go to section 3.

SECTION 3: QUESTIONS ABOUT A PERSON YOU HAVE TOLD

- Look back at **Box B** on page 3 and **CHOOSE ONE PERSON ONLY** you put a check beside. Answer the questions in Section 3 thinking about this person.

The person I have chosen to answer Section 3 about is:

- ☐ my mother
- ☐ my father
- ☐ a sister
- ☐ a brother
- ☐ a daughter
- ☐ a son
- ☐ an aunt
- ☐ an uncle
- ☐ a niece
- ☐ a nephew
- ☐ a cousin (female)
- ☐ a cousin (male)

Remember
Only choose ONE person who you DID TELL your genetic test result to.

If you did not check anyone in Box B, go straight to SECTION 4.

1. Please rank whether the statements listed below were important in your decision to tell your family member about your test result. For each statement choose a number from 1 to 5.

	Very Important		Neither important nor unimportant		Very Unimportant
	1	2	3	4	5
a) You have a close relationship with the person	☐	☐	☐	☐	☐
b) You have no conflicts or tensions with the person	☐	☐	☐	☐	☐
c) You thought that the person could also give you social support in dealing with your test result	☐	☐	☐	☐	☐
d) You thought the person was emotionally capable of handling the information	☐	☐	☐	☐	☐
e) You thought that the information was relevant for the person	☐	☐	☐	☐	☐
f) You thought you might regret it if you didn't tell the person	☐	☐	☐	☐	☐
g) You thought you had a responsibility or duty to tell the person	☐	☐	☐	☐	☐
h) You thought you would feel guilty later if you didn't tell the person	☐	☐	☐	☐	☐

2. Please tell us about any other reasons or issues that were important to you when you decided to tell this family member about your genetic test result.

_____

_____

_____

_____

3. On a scale of 0 to 100%, what do you think is the chance that, someday, this family member will develop myotonic dystrophy?

☐ %

4. How likely do you believe it is that this family member will get myotonic dystrophy someday?

No chance 1 2 3 4 5 6 7 Certain to happen

SECTION 4: QUESTIONS ABOUT YOUR EXPERIENCES AND OPINIONS

1. On the whole, does your family have an open or closed communication pattern? Please choose one.

☐ my family has an open communication pattern
☐ my family has a closed communication pattern

2. Does your family have an open or closed communication pattern about personal health problems? Please choose one.

☐ my family has an open communication pattern
☐ my family has a closed communication pattern

3. Did your genetic counsellor give you advice on who you should talk to about your genetic test result?

☐ Yes

☐ No

☐ Unsure

4a. Was there anything that made it hard to talk to certain family members?

Please explain:

4b. Was there anything that made it easy to talk to certain family members?

Please explain:

5. Having myotonic dystrophy is a very serious problem.

Strongly Disagree 1 2 3 4 5 6 7 Strongly Agree

6. For how many years have you known the result of your genetic test? If you have known for less than a year, how many months?

_____years OR _____months

7. Can you remember the result of your test?

□ Yes □ No □ Unsure

8. If yes, what was the result? (optional)

_____

9. When you had your test did you have symptoms of myotonic dystrophy?

□ Yes □ No □ Unsure

10. Do you currently have symptoms of myotonic dystrophy?

□ Yes □ No □ Unsure

11. Does anyone else in your family have myotonic dystrophy?

□ Yes □ No □ Unsure

12. Did you have mixed feelings about telling your family members about your genetic test result?

□ Yes □ No

13. Are you adopted?

□ Yes □ No

14. Do you have anything else to add about your experience with telling family members or anything about your particular situation?

_____

_____

_____

_____

_____

_____

_____

SECTION 5: QUESTIONS ABOUT YOU AND YOUR FAMILY

What is your age in years?

☐ <20 ☐ 20-29 ☐ 30-39 ☐ 40-49 ☐ 50-59 ☐ 60-69 ☐ 70+

What is your sex?

☐ Male
☐ Female

What is your current marital status?

☐ Single
☐ Not married, in a steady relationship
☐ Married
☐ Divorced or separated
☐ Widowed

What is the highest level of education that you have completed?

☐ Less than high school
☐ High school
☐ Community college, technical college, or CEGEP
☐ University degree – undergraduate
☐ Professional degree (e.g. education)
☐ Post graduate degree
☐ Other education or training (please specify):

_____

To which ethnic or cultural groups do your ancestors belong ?(check all that apply)

☐ Canadian
☐ French
☐ English
☐ German
☐ Scottish
☐ Black
☐ North American Indian
☐ Metis
☐ Inuit/Eskimo
☐ Chinese
☐ Jewish
☐ Polish
☐ Portuguese
☐ Irish
☐ Italian
☐ Ukrainian
☐ Dutch(Netherlands)
☐ South Asian (East Indian, Pakistani, Punjabi, Sri Lankan, etc.)
☐ Other *(please specify)*

_____

What is your first language?

☐ French ☐ English

How would you rate your experience filling out this survey?

Easy									Hard
1	2	3	4	5	6	7	8	9	10
☐	☐	☐	☐	☐	☐	☐	☐	☐	☐

PLEASE RETURN YOUR SURVEY IN THE POSTAGE PAID ENVELOPE PROVIDED.

THANK YOU VERY MUCH FOR TAKING THE TIME TO FILL IN THIS SURVEY!!

It's not too late! If you prefer to do an electronic version of this survey go to:

www.fam-com.com

Your password is: communication
Your username is your three digit id code on the cover page.

APPENDIX L: Approval of REB application

Children's Hospital of Eastern Ontario
Centre hospitalier pour enfants de l'est de l'Ontario

CHEO RESEARCH ETHICS BOARD APPROVAL – EXPEDITED REVIEW

Principal Investigator:	Christine Honeywell
Proposal Number:	#07/14X
Protocol Title:	Family Communication about Genetic Disorders: Developing a Framework for Effective Interventions
Department or PSU:	Genetics
Approval date:	March 23, 2007
Valid Until:	March 22, 2008
Documents reviewed and approved:	Research protocol submitted January 15, 2007

This is to notify you that the Children's Hospital of Eastern Ontario Research Ethics Board has granted approval to the above named research study on the date noted above. Your project was reviewed under the expedited stream, which is reserved for projects that involve no more than minimal risk to human subjects.

During the course of the research, no deviations from, or changes to, the protocol or consent form may be initiated without prior written approval from the REB. Further, investigators are asked to report the following to the REB:

- Proposed changes to the study procedures (including the recruitment strategy, inclusion criteria, etc.);
- Concerns or issues that arise in conducting the research;
- Changes to the consent documents and advertisement notices;
- Changes to the investigators who assume responsibility for the study; &
- An annual report.

Wishing you success in your project.

Regards,

Dr. Carole Gentile, C.Psych.
Chair, Research Ethics Board

CG/smeh 23/03/07
c.c. Pat Brazeau, Manager, CHEO RI
Dr. Judith Allanson, Genetics

This is an official document. Please retain the original for your file *version 11/2003*

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