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ORIGINAL RESEARCH

The Effect of BRCA Gene Testing on Family Relationships: A Thematic Analysis of Qualitative Interviews

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Abstract Discovery of mutations in the breast and ovarian cancer susceptibility genes *BRCA1* and *BRCA2* can have emotional consequences for both the tested individual and his or her relatives. This secondary analysis study investigated how *BRCA* testing impacts family dynamics and relationships. For the original study, a grounded theory inquiry, participants were recruited from a hereditary breast/ovarian cancer syndrome support website and open-ended interviews were performed asking about individual and family experiences after *BRCA* testing. All 12 participants whose interviews were included in the secondary analysis had a *BRCA* mutation. For the secondary analysis, thematic analysis was conducted and revealed three main themes characterizing the effect of *BRCA* testing on family relationships: 1. That the first in the family to have testing or seek genetic counseling takes on a special family role that can be difficult for them; 2. That discussions in the family often change; and 3. That individuals may feel more or less connected to certain family members. These changes seemed to relate to family cancer history, relationships, coping strategies, communication patterns, and mutation status. Genetic counselors might find it useful to explore these issues in order to prepare clients before *BRCA* testing and to support them through shifts in family dynamics after disclosure of results.

Keywords Qualitative research · BRCA · Hereditary cancer · Family relationships · Psychosocial · Genetic testing · Secondary analysis

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Introduction

A wealth of research since the early 1990s has focused on identifying and characterizing the genes involved in the hereditary predisposition to breast and ovarian cancer. Since early on in the search for and identification of the *BRCA1* and *BRCA2* genes, it has been recognized that testing for mutations in these genes has the potential for serious psychological consequences (Bredart et al. 1998). Several studies have investigated the psychosocial effects of *BRCA* testing (Watson et al. 2004; Hallowell et al. 2004; Van Dijk et al. 2004; Van Oostrom et al. 2003), but few of these have addressed the complexity that family cancer history and family members themselves bring to the decision to test and the response to the result.

The research that has focused on *BRCA* testing and the family generally suggests that whether or not family members are actually involved in the *BRCA* gene testing process, they may be intimately connected with this process, before, during and after their relative's decision to test is made.

Risk Perception

Even before genetic testing is considered, family cancer history seems to play a significant role in how an individual

perceives his or her own risk of cancer. Women in families with a strong cancer history are usually aware that they could follow the family pattern and develop cancer as well (Kenen et al. 2003a; Hallowell 1999; Forrest et al. 2003; Foster et al. 2002; Kenen et al. 2003b; Raveis and Pretter 2005; Hamilton et al. 2009). They tend to perceive their personal risk as higher if their relatives did not survive their cancer, if their mother or sister had cancer, or if they have a relative recently diagnosed (Kenen et al. 2003a; Foster et al. 2002; Raveis and Pretter 2005). Perceptions of their own risk are also subject to influence by 'stories' about the cancer in the family (Kenen et al. 2003b), which are crafted by members of the family and then become legacies ingrained in every new generation (Werner-Lin 2007). Saliency of personal risk can also wax or wane with cancer screening tests or by reaching or passing age milestones perceived to be important in the family cancer history (Kenen et al. 2003a; Werner-Lin 2007; Foster et al. 2002). Consequently, it appears that family history helps to create the context in which people consider having a *BRCA* gene test.

Genetic Testing Uptake

Once the option of *BRCA* testing is introduced, it seems that individuals often strongly consider the potential impact on family members when deciding whether or not to pursue testing (Foster et al. 2002; Kenen et al. 2003a; Kenen et al. 2006; Hallowell 1999; Goelen et al. 1999). For many, getting information for the family is an important reason to test (Hallowell 1999; Tercyak et al. 2007; Phelps et al. 2007; Foster et al. 2004; Daly et al. 2001; Goelen et al. 1999; Foster et al., 2002; Hallowell et al. 2003). Some go a step further and insist that testing is their duty, obligation or responsibility (Hallowell et al. 2003; Hallowell 1999; Kenen et al. 2003a) because their family members have a right to know their own risk for cancer (Hallowell 1999; Daly et al. 2001). As well, some individuals seem to approach testing as a chance to 'do right' by their families (d'Agincourt-Canning 2001) or to fulfill their obligations to care for family members (Hallowell et al. 2003), stay alive for them (Kenen et al. 2003a; Hallowell 1999; Foster et al. 2002), or honor their dying wishes that relatives get tested (Hallowell 1999; Kenen et al. 2003b). Therefore, the decision to have *BRCA* testing is often made not only in the context of a perceived personal risk for cancer but also by placing importance on the implications of genetic testing for family members.

Disclosure to Family

After receiving a test result, individuals need to decide whether or not to tell their family members. As expected, a common reason for deciding to disclose genetic information to family members is to provide them with information about their own risks and options for testing or management (Hughes et al. 2002; MacDonald et al. 2007; McGivern et al. 2004; Claes et al. 2003). Other reasons are that the family member had actively asked to be told or had already gone to genetic counseling or had genetic testing themselves (Claes et al. 2003), or to gain support or advice in next steps such as surveillance and surgical decisions (Hughes et al. 2002; McGivern et al. 2004) and informing other relatives (Goelen et al. 1999; Hallowell et al. 2005a). Common reasons for refraining from disclosing results to certain relatives are being out of touch or not emotionally close with relatives (Green et al. 1997; Forrest et al. 2003; MacDonald et al. 2007; Daly et al. 2001; Hughes et al. 2002; McGivern et al. 2004; Claes et al. 2003), or feeling concerned that the news of a *BRCA* mutation would alarm or upset others in the family. The latter often presents a dilemma to those who want to provide potentially life-saving information to their relatives without harming them emotionally (Hallowell et al. 2003; Hallowell et al. 2005b; Hallowell et al. 2005a; Green et al. 1997; Hallowell 1999; Bradbury et al. 2007; Hughes et al. 2002).

Tested individuals typically do disclose their results to at least one relative, whether the results are positive, negative, or inconclusive (Patenaude et al. 2006; d'Agincourt-Canning 2001; McGivern et al. 2004; Costalas et al. 2003). Some tell their family members shortly after receiving the test results (Hamilton et al. 2005; Hughes et al. 2002). Disclosures performed later tend to be more carefully planned (Hamilton et al. 2005). For instance, individuals think about which family member is best to disclose the test results to the rest of the family (Forrest et al. 2003; Blandy et al. 2003; McGivern et al. 2004; Hallowell et al. 2005b; d'Agincourt-Canning 2001), as well as whether certain family members are ready to hear the information (Hamilton et al. 2005; Hallowell et al. 2003; Bradbury et al. 2007). In addition, the setting and method of the disclosure is considered. For instance, some individuals choose to delay the disclosure until a family gathering for some other reason like a holiday (Bradbury et al. 2007; Green et al. 1997), sometimes to integrate the disclosure into the conversation that would be taking place already (Forrest et al. 2003; Green et al. 1997). Others decide to disclose the mutation and heritable cancer risks to family members gradually, over months or years (Bradbury et al. 2007). Individuals also tend to select *what* to disclose, sometimes omitting or altering information or downplaying the seriousness in an effort to prevent anxiety in their relatives (Daly et al. 2001; DudokdeWit et al. 1997; Hamilton et al. 2005, Kenen et al. 2006; Hallowell et al. 2005a). In an extreme case, there is one report of a woman who deliberately lied to her family about her positive mutation status in order to prevent her father from the guilt

of having passed the mutation to her (Loud et al. 2006). Disclosures may also involve the discussion of screening and surgery guidelines, risk of a mutation for family members, cost of testing, insurance discrimination and feelings about their own and their relatives' risks (Hughes et al. 2002; McGivern et al. 2004).

Reactions to Test Results

Not surprisingly, individuals who decide to have *BRCA* testing seem to react to their test result according to its ramifications for not only their own risk, management and self-identity (Kenen et al. 2003a), but also for family members. For instance, individuals identified to have a *BRCA* mutation sometimes feel distressed or guilty because they may have already passed the mutation on to their children (Kenen et al. 2003b; d'Agincourt-Canning 2001), or because their test result reveals that their siblings are also at-risk (Smith et al. 1999). Parents found to have a negative test, i.e., they do not carry a deleterious mutation, generally feel relieved that their children are not at risk (d'Agincourt-Canning 2001; d'Agincourt-Canning 2006). However, testing negative can sometimes result in guilt if other close family members had already been found to carry a mutation (d'Agincourt-Canning 2006), a phenomenon often referred to as 'survivor guilt'.

Disclosures of genetic risk information to family often seem to evoke unexpected and intense emotions (Speice et al. 2002). For those who are the first in the family found to have a *BRCA* mutation, disclosing the mutation to family members can be particularly burdensome or upsetting (Hallowell et al. 2005b; Costalas et al. 2003), as they sometimes feel guilty for 'being the bearer of bad news' (Hamilton et al. 2005; Kenen et al. 2006; McGivern et al. 2004; d'Agincourt-Canning 2001). Also, individuals may feel pressure to educate their family about genetic information that they may not themselves understand well (Kenen et al. 2006; DudokdeWit et al. 1997; Costalas et al. 2003). Family members who receive news of a mutation can feel shock, fear, resentment, sadness, guilt, anger and blame (Speice et al. 2002; Bradbury et al. 2007; Costalas et al. 2003). Strong emotions can manifest from concern about one's own cancer risk or a family member's risk (DudokdeWit et al. 1997), because grief or tension from a past loss of a family member is reactivated (DudokdeWit et al. 1997; Speice et al. 2002), because of the timing or method of the disclosure (Hallowell et al. 2005b), or because the disclosure happened at all (DudokdeWit et al. 1997). Additionally, some family members do not seem to understand or acknowledge the significance of the result (Bradbury et al. 2007; Costalas et al. 2003; Blandy et al. 2003; Speice et al. 2002). Others interpret a positive result not as upsetting but as an explanation for the strong family history of cancer (Bradbury et al. 2007). Likewise, some family members pay less attention to the news of the mutation than to a recent cancer diagnosis in the family (DudokdeWit et al. 1997). After learning of the *BRCA* mutation in the family, uptake of testing for informed high risk relatives was estimated to be 15% (29% for first degree relatives and 12% for nieces) in a study conducted by Blandy et al. (2003). In this study, 37% of families had no close relative requesting testing.

Impact on Family Relationships

To date, little research focus has been placed on how family *relationships* might change after disclosure of a *BRCA* test result. Existing studies report a range of effects on relationships. It appears that relationships can be weakened after *BRCA* testing when family members do not share the same coping styles or interest in testing (Speice et al. 2002; d'Agincourt-Canning 2001; McGivern et al. 2004; Blandy et al. 2003). Members of the family can start to feel isolated from each other, especially if they are not sure how best to communicate with each other, or if they feel they are in different life stages (Kenen et al. 2006; Hamilton et al. 2005; Werner-Lin 2007; Speice et al. 2002). Some families report family members who feel stigmatized and thus are very resistant to talking about the inherited breast/ovarian cancer in the family (Kenen et al. 2007). This can create 'areas of sensitivity,' further discouraging these conversations (Bradbury et al. 2007), and can cause other family members to feel shunned at family events by relatives not as open to discussing it (Speice et al. 2002). Family discord can also happen if family members disagree about the implications of the mutation for the family (Speice et al. 2002) or about who should be informed (Forrest et al. 2003). Individuals identified to have a *BRCA* mutation also describe their family lives as becoming 'dominated' by talk of cancer (Werner-Lin 2007).

On the other hand, discovering a mutation in the family appears to be able to positively influence some family relationships. Kenen et al. (2006) reported on a research participant who felt consoled upon finding that she shared the *BRCA* gene with her family members, because cancer then became a problem the family could address together (Kenen et al. 2006). d'Agincourt-Canning (2001) described that genetic testing allowed her research participants to strengthen interpersonal family ties due to the importance of the information that was disclosed. A study by Bradbury et al. (2007) reported that 22% of individuals reported a strengthening of a parent-child relationship. Also, McInerney-Leo et al. (2005) found that perceptions of family cohesion increased upon making a testing decision. Interestingly, in this study family cohesion seemed to increase even if the decision was to not pursue testing. However, several studies have reported no change in family functioning, relationships

or conflict (McInerney-Leo et al. 2005; Bradbury et al. 2007; Stroup and Smith 2007).

On the whole, relatively few investigations have focused on how *BRCA* testing might impact family relationships. Considering that changed family relationships could affect an individual's life substantially, in the current study we sought to investigate the impact of *BRCA* testing on family dynamics and the relationships between family members.

Methods

Qualitative Research Design of Original Study

The present work is a secondary analysis of an original study conducted by Hamilton and colleagues (Hamilton 2003). The purpose of the original study was to increase the understanding of individual and family experiences after *BRCA* testing or after discovering a high risk for hereditary breast/ovarian cancer. The results of the original study have been published (Hamilton et al. 2005; Hamilton and Bowers 2007).

For the original study, a qualitative research design was selected because it can uncover unexpected experiences of members of hereditary breast/ovarian cancer families, since it is the research participants themselves that lead the direction of the study. Qualitative research often aims to explore meanings of experience; it attempts to understand both what people do and why they do it (Beeson 1997). Qualitative inquiry may also place importance on all observations no matter how deviant from the 'norm' they are. Thus, health care professionals can use the results of this type of research to sensitize themselves to the breadth of potential feelings and experiences of their patients.

Participants for the original study were recruited by posting notices on a hereditary breast/ovarian cancer syndrome support website (FORCE: Facing Our Risk of Cancer Empowered: www.facingourrisk.org). All of the participants were members of families who had confirmed or probable hereditary breast/ovarian cancer syndrome. The study was performed under IRB approval and required informed consent from each participant.

Open-ended interviews were performed in the original study by Dr. R.J. Hamilton for 17 participants. Although phone or in-person interviews are the more traditional form of interviewing, email interviews have recently been introduced as an appropriate alternative (Hamilton and Bowers 2006). Therefore, depending on the participants' preferences, either phone or email interviews were done. Examples of initial questions asked in the interviews are included in Appendix A. Follow-up questions were also posed to the participants, by phone or email. Some participants were interviewed again approximately 3 years after their first interviews to follow the participants over time. Grounded theory was the qualitative method used for interviewing and interpreting the data in the original study (Hamilton et al. 2005).

Secondary Analysis-Present Study

a. Rationale

Although the research question of the original study concerned the overall experience of genetic testing it was noted that participants often seemed to want to discuss their families, particularly the impact on their families after testing. The participants were encouraged to discuss and expand on this topic during their interviews. Recognizing that these interviews had the potential to help us characterize the impact of *BRCA* testing on family relationships, we decided to perform a secondary analysis of the data for this purpose.

Secondary analysis in qualitative research is defined by Thorne (1994) as the analysis of data either beyond its original intent or in relation to new and extended inquiries. As reported in other qualitative studies (Deatrick et al. 1993; Knafl et al. 1995), asking a question different from that of the primary study may yield equally rich data with a different focus. It is often performed by individuals who were not involved in the initial study design or collection of data (Thorne 1994), permitting a different researcher to bring a different interpretive lens to the data.

Of all participants interviewed in the original study, only those who had participated in two interviews ($n=12$) — an initial interview and another approximately 3 years later– were chosen for this secondary analysis. Selecting participants for whom data were available across a period of time permitted different questions to be asked of the data than was done in the primary data analysis (Hinds et al. 1997). Prior to the analysis, personal identifiers were removed from interview transcripts and participants were given pseudonyms. Participants who were family members were given names that begin with the same letter (e.g. The R family includes Rebecca, Rachel and Raymond). Interview passages that were included in this report were subject to minor revision only when it was judged that the revision would not take away from the meaning of the passage but would promote understanding by the reader. The analysis for this study was done primarily by the first author, who was not involved in the collection or analysis of data in the original study. IRB approval was obtained for the secondary analysis.

b. Data Analysis

Thematic analysis was selected as the most appropriate method for the secondary analysis of the interview transcripts. Thematic analysis is a method for identifying,

describing, analyzing and reporting themes and patterns within data (Braun and Clarke 2006). It can be used to analyze data obtained under a number of qualitative theoretical frameworks, including grounded theory (Braun and Clarke 2006).

Since few qualitative studies had previously examined the effects of *BRCA* gene testing on family relationships, an inductive thematic analysis approach was chosen so that a cross-section of family effects related to *BRCA* gene testing could be explored. This data-driven approach was of benefit also because it allowed identification of unexpected themes in the data (Braun and Clarke 2006), rather than themes that were fundamentally modeled under our own prior theories or preconceptions (Braun and Clarke 2006). A review of relevant literature on the psychosocial impact of *BRCA* testing was performed prior to conducting the analysis to sensitize us to subtle concepts that may be present in the data but could be missed if not recognized prior to the analysis.

In performing the thematic analysis, all transcripts from the interviews conducted in the original study were read twice and preliminary notes were made. Initial coding of the transcripts was then performed with a goal to remain open to all possible interpretations. Codes either stored information about patient demographics (Richards and Morse 2007), or were far more analytical, representing links between the data and an idea (Richards and Morse 2007). Codes were made as descriptive of the participant's experience or thought as possible. For the parts of the transcript that explicitly addressed the family, line-by-line coding was used, in which at least one code is given to each phrase, line or sentence in the data set. The remaining transcript sections were read and connections with family relationships were noted.

After coding, we set out to identify themes. A theme in qualitative research is defined by Braun and Clarke (2006) as something that captures an important aspect of the data in relation to the research question, which for this study was "What is the impact of *BRCA* testing on family dynamics and the relationships between family members?" Representation of the theme across the data set is ideal but not necessary (Braun and Clarke 2006). In our study, identification and characterization of themes was a process. Initial ideas about themes were noted early on. Potential themes were described in writing throughout the initial stages of coding. After coding the interviews of the first few participants, codes were organized using QSR's Nudist Vivo software (version 7.0.281.0 SP4) into potential themes or subthemes depending on their content or motivation. Often, codes were classified under several themes. For each additional interview transcript, codes were organized into these preliminary thematic categories. At a few points throughout the analysis the codes were reorganized to better reflect the themes present in the data.

After all interviews were coded and all codes were classified under thematic headings, a visual map was created, consisting of themes and subthemes (Fig. 1). While not shown in Fig. 1, the connections between themes were also mapped. Creating this visual diagram aided in the appreciation of the intertwined nature of the data. From this, three major themes were selected to be the focus of this article.

Results

Characterization of the Sample

The analysis involved 12 participants who all tested positive for either a *BRCA1* or *BRCA2* mutation. These 12 participants came from a total of eight families, with two families from which three family members participated. All but one participant were female. Ages ranged from the mid-20s to the late-50s. Some participants had a personal history of cancer. Most participants were married or partnered, and most had biological children.

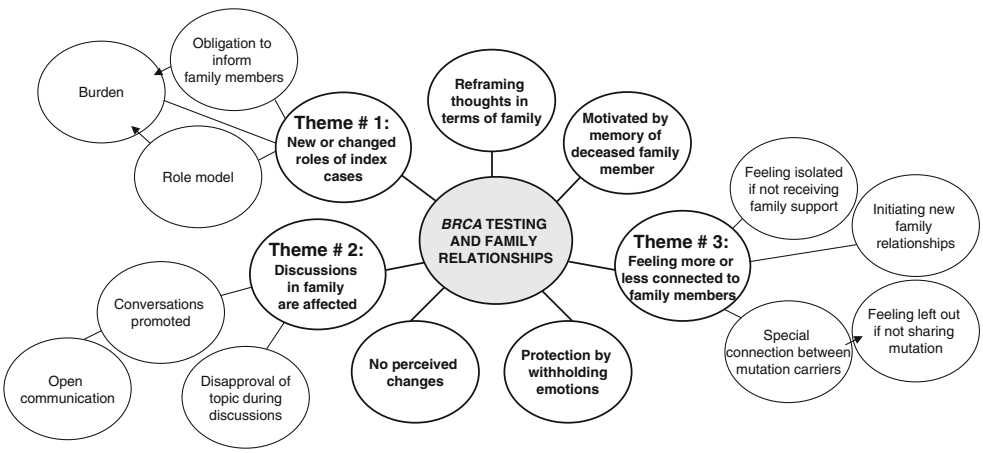

Fig. 1 A map of themes related to the effect of *BRCA* testing on family relationships. Themes were identified by thematic analysis in the secondary data analysis. The three main themes featured in this paper are highlighted and their subthemes are shown.

Themes Identified in the Analysis

The thematic analysis identified several themes and their components (i.e. subthemes) reflecting how testing positive for a *BRCA* mutation can impact family relationships, as shown in Fig. 1. Characterizing all of the subtleties and nuances of each one would have been unmanageable for one paper. Briefly, the themes that were not chosen to be a focus of this paper included: 1. Reframing thoughts in terms of family, such as thinking of oneself or one's own mutation status in terms of impact for their family; 2. Motivated by memory of deceased family member, such as thinking and acting as their relative would have wanted; 3. Protection of oneself, a relative or a family relationship by withholding emotional expression; and 4. No perceived changes in family relationships.

This article will focus on three other themes, each featured in Fig. 1, since they were commonly mentioned and thus easiest to characterize the available data. Importantly, as family relationships can be complex, so can the themes that were identified. Thus, although the three major themes described here have some distinct features from one another, they are also related, and they are related to the themes we chose not to highlight.

Theme # 1: New or Changed Family Roles for the Index Case

The first individual in the extended family to pursue genetic counseling or testing is often referred to as the 'index case' of the family. After genetic testing, most of the index cases in the current study seemed to take on a new role in their families. Some of the nuances of these roles will be described here.

a. Obligation to inform family members

Firstly, those index cases receiving the news that they have a *BRCA* mutation usually felt obligated to inform relatives. Interestingly, this obligation was also felt by other participants who fit the role of index case in some sense. For instance, Claire was the first in her family to attend genetic counseling, as she was concerned about her strong family history of breast and ovarian cancer. She had not been diagnosed with cancer so was not the appropriate person to test first in the family. However, she convinced relatives to pursue testing, and she organized a family meeting with the genetic counselor to discuss the likelihood of hereditary cancer in their family. After a *BRCA* mutation was found in some of her family members, she clearly feels responsible for her family's well-being:

> I was the first person in the family. I'm kind of the person who opened the can of worms. So, um, I felt that responsibility very heavily.

A similar sense of responsibility was expressed by Danielle, whose mother was actually the first in the family to have testing, but died only 2 weeks after learning that she carried a mutation. Danielle later had testing and discovered that she also carries the mutation. Danielle says:

> I feel this huge sense of responsibility to let extended family know to kind of you know, if they want to be tested they can be.

In this case, after her mother's death, Danielle seemed to have assumed the role of 'index case by proxy', taking on the task of disclosing the result to her relatives since her mother never had a chance to do this before her death. Interestingly, although this participant wanted her distant cousin to know she probably also carried the mutation (since this cousin had had both breast and ovarian cancer), she did not feel she had the right to actually tell her.

> I called my mom's cousin, who has breast and ovarian cancer and I was really worried about that because I … did not feel I had the right to say [that she has the mutation] and so wanted to tell her that my mom tested positive and that you know, I did and that uh, there's concern, kind of thing.

Thus, Danielle served the obligation she felt as the 'index case by proxy' to let this relative know of the test result without directly stating her assumption that the cousin has the mutation.

b. Acting on the responsibility to bring news to the family can be burdensome for the index case

Upon feeling this responsibility to inform family members of the *BRCA* mutation in the family, most study participants took the primary role in dispensing the information throughout the family. This role of 'bringer of news' was not always easy, however.

For example, several participants felt upset when disclosing the news of the mutation to their family. Many felt guilty during the disclosure, even though they thought that guilt was an irrational feeling. This sentiment is best expressed by Judy, who was the first in her family to have genetic testing, several years after a diagnosis of breast cancer.

> I almost had feelings that I had done something "wrong" and was confessing to them. Intellectually I know that is silly, but it was the feeling. Perhaps it was hard because it meant my breast cancer really was more than about me — and was now a factor for the whole family. And it was out of my control! Who wants to be the bearer of bad news?

Interestingly, some index cases appeared to feel guilty even when the news of the family mutation was welcomed by family members. For instance, Rachel, who chose to

have a prophylactic mastectomy upon learning she carried the *BRCA* mutation, recalls conversations she had with her aunt, the index case in her family, after her aunt sent a letter informing her of the mutation.

> Interviewer: And how was that letter received in the family?
>
> Rachel: Um, funny. I'll say by us it was received by my immediate family and my mom and my sister and I, very positively and grateful. My aunt herself…had a little bit of, um…you know, guilt at times…[stating] …'If I hadn't found out then they [nieces] wouldn't have to go through that, you know…God you're going through so much…. I can't believe what I started. I hope it didn't open up too much.' And me in response, 'Oh my God, no. You know, I wouldn't, I made the decision to follow through with it, I wouldn't trade it for anything.'

Thus, the index cases described in this study tended to feel a special obligation to inform relatives of their mutation, but found that doing so could be burdensome due to the enormity of the implications of the news for their family.

c. Index case as role model

Getting cancer and testing positive for a *BRCA* mutation created a different burden for another participant, Elise, who was also the first in her family to have testing. After her cancer diagnosis and her positive *BRCA* test result, she felt pressured to act as a role model to her family, especially her younger siblings.

> Interviewer: Can you talk some more about what it was like to be the "first" identified in your family and thus the one to tell the news? Do you think there has been any change in your relationships with your parents or siblings because of this?
>
> Elise: It sucks to be first. I feel like I always have to have the answers and be the voice of reason. (I'm the oldest child and have always been maternal towards my brothers and sisters). So even though I'm a basket case to my sweet husband— to my brothers and sisters I feel like I have to be at peace with all of this. If I'm not, how can I expect them to be. I don't want them to live scared because of this.

She even explains how she communicates with her siblings so that she doesn't alarm them.

> I wanted to be gentle with my brothers and sisters because I know that this type of risk is so difficult to deal with at such a young age. (Yes, the cancer patient was calming everyone else down. Doesn't that seem backwards).

In the first passage, this participant suggests that her established role as a maternal big sister might have encouraged her to act as a role model to her siblings in this situation. However, this particular participant's family history may also have provoked such pressure. Before her own breast cancer diagnosis in her late 20s, she did not know of any cancer history in her parents, aunts, uncles or cousins. However, upon investigation after her diagnosis, the family discovered a strong cancer history in distant relatives that was consistent with a hereditary cancer syndrome. When asked if it might have been the lack of cancer in close family that is forcing Elise to be a role model, she agrees.

> Interviewer: If you had known there was a "family history" of cancers in your family, do you think your response (i.e. feeling guilty) would have been different when the mutations were identified? How so?
>
> Elise: I think it would have been different. I would have had someone else to point to— or someone else to relate to. I feel like I am setting the "standard" here. i.e., you should feel this way when diagnosed, you should feel this way after chemo. It's all new to my family. If we would have known, it wouldn't have been such a shock. Everything has felt like a big giant snowball picking up speed— maybe if we would have had some idea we would have been on more even ground with all of this. I would definitely not feel like I have to be such a role model about this whole thing.

Interestingly, Elise's mother, Evelyn, acknowledges her daughter's special role in the family but does not suggest that it has been burdensome to her daughter.

> [Elise]'s been amazing because she's really been the one that has kept this um, almost all together and I even wrote her a letter just before her last chemo treatment just telling her how proud I was of her because she was the one we all took our cue from. She was the one that was always upbeat, um, very seldom did you see her down…I would just sit back in awe and say, this is my child, my child, and look at what she's doing.

This situation also exemplifies that the burden felt by an individual after genetic testing may not be recognized by their family members. Moreover, it appears that her mother might be reinforcing the pressure her daughter is feeling.

Theme # 2: Genetic Testing Affects Discussions in the Family

The second major theme identified in the study was that a positive *BRCA* test result can impact discussions between family members, in a variety of ways.

a. Testing promotes conversation between family members

For some individuals, receiving the news of a *BRCA* mutation in the family resulted in more talk about cancer, cancer risk, or associated topics like prophylactic surgeries.

For example, genetic testing served as a catalyst for opening up Naomi's relationship with her father, which had been strained since Naomi lost her mother to breast cancer.

> My dad and I have always had a pretty stilted relationship and a hard time talking about my mom—in a lot of ways having the concrete issues of the testing process to discuss made the whole issue more approachable. I also definitely felt his approval and relief that I was doing something about the cancer thing, so that has made our relationship somewhat more comfortable.

Likewise, Elise feels that the genetic testing she pursued following her cancer diagnosis opened up discussion about cancer and genes in her family. However, for her, these conversations sometimes have had a negative effect.

> My family is no longer as lighthearted as we once were. Genetic discussions happen all the time. I feel like there is absolutely no escaping this disease… Every time I pick up the phone someone else is calling to talk about my breasts.

Importantly, this is the same participant who felt forced to be a role model to her family, as described above. Interestingly, Elise's sister, Emily, does not perceive that genetic testing has fueled family discussions significantly.

> The family doesn't talk about the mutation directly, except when the testing was done. Since the cancer is such a big part of our life, we don't really discuss the what-ifs of everyone else.

It is possible that these different perceptions could be because Elise, the index case and first in the immediate family to get cancer, is likely at the center of most genetic conversations in the family while Emily, never diagnosed, may not be so intimately involved in such discussions. Their mother, Evelyn, sees genetic testing as having mostly a positive impact on their family discussions.

> Interviewer: What do you think the impact of having these predictive genetic tests in your family has been?
>
> Evelyn: I think that all and all it's been a very positive, um, it's given us a focus away from so much Elise having the cancer and saying OK, this is why Elise had the cancer, and again we'll do what we can do to try and not have it.

Thus, testing positive for a *BRCA* mutation seems to have the potential to open up discussions about cancer and genetics in the family, and this may be welcomed or unwelcome all within the same family. An important observation is that different members of the same family can perceive the impact on their discussions differently.

Genetic testing for *BRCA* mutations promoted conversations for family R as well. Family R included a nuclear family that was already very close and talked about cancer frequently even before genetic testing, due to the strong history of breast cancer on Raymond's, the father's, side of the family.

> Interviewer: When you all are together is [cancer] a common topic of conversation or not so much anymore or….
>
> Raymond: No, it's real common. Every single conversation. It always has been. Now of course it's much more personal.
>
> Interviewer: Now when you say it always has been… before testing what was the conversation?
>
> Raymond: …we didn't talk about it quite that much.

In this family, genetic testing has encouraged Raymond to be more in touch in general with his two daughters, who are in their 20 s and who both tested positive for the mutation.

> I think of them much more frequently now that um, this kind of news has happened and we communicate more regularly than we did before and that's, we've always been very close, but I just kind of have this feeling of, of I don't know, just maybe the time that I have with them is, really means a whole lot more to me now.

One of his daughters, Rebecca, noted a similar change in her relationship with her father, remarking that her father phoned her and her sister much more often now than he did before their genetic testing experience.

Thus, for some families, genetic testing can promote discussions not only related to cancer risk but general conversations as well. The reactions to these conversations may be positive or negative, and may be shared by family members or not.

b. Disapproval of topic during family discussion

Even when conversations in general were promoted, some participants reported that there were at least some relatives who clearly did not want to talk about the family *BRCA* mutation. They let other family members know of their wish in various ways. One way was by directly telling them. Claire experienced an example of this.

> And one niece actually after I had told the family about the results sent message through her mother, my sister, that um, I had mentioned it, now once I told about it I could, um, basically damn well shut up now… She was really quite adamant that she did not

> want, and she said something to the effect of 'I don't want this coming up every time the family gets together. We don't need to talk about it.'

Family members also used subtle, indirect messages to discourage talk of the mutation. For instance, Danielle describes feeling this message from her siblings.

> I mean they never bring it up now, I'm the one who has to bring it up. That's you know, I don't think they really want me to, anymore. There's certainly a subtle message. If I bring it up they'll talk about it but there's definitely a subtle, I guess a picture, 'this is not what we want to talk about.' And they won't ever bring it up. They'll ask me how I'm doing. But, and lots of ways want to know that, oh, everything's just fine. You know, they just don't want to know, don't want to uh delve into it too much.

Her siblings' hesitancy to talk about the mutation has been difficult for Danielle. She says:

> Um, my sisters don't like to talk about it, don't want to deal with it, the two sisters that have chosen not to be tested. And um that's very difficult for me because I need to talk about this and that's my way of coping.

Yet Danielle still seemed to want to keep her siblings informed about *BRCA* and cancer risks. She adapted to her siblings' hesitancy by choosing methods to inform them other than directly talking to them about it.

> And um, and I also am hesitant…it's a fine line and I don't want to push my sisters away…I get a sense if I continue to push this, that it's gonna really impact our relationship negatively and I don't want to do that…I mean…they know what the risk is and they know what to do, they know who to contact. Um, we are fortunate enough [in their region] to have a support group…you know I send the newsletters and that kind of thing, let them know when the speakers are happening…I think that's my work now instead of to keep bringing it up.

A very similar family dynamic was present in Claire's family. In this family, Claire's siblings were supportive around the time of her prophylactic mastectomy, but were very hesitant to bring up related topics at other times.

> We got together and nobody brought it up once. For me it was so weird because it was just so important to me at the time. A couple times that happened. I just have heard, you know, about the family having the gene or something, and then we'd get together and we'd be talking about the weather, you know, it was like they, they didn't want to bring it up, or didn't know what, or just, I don't know, or didn't want me to bring it up. So it felt very odd for me because to me, I did want to discuss it with them and they weren't that interested.

In the R family, although several members were very open to each other about the mutation and cancer risk management, certain family members were not. Like these other two families, some family members did not acknowledge the mutation or even Rachel's recent prophylactic mastectomy at family gatherings. Rachel thinks these relatives were in denial.

> Interviewer: Do they talk about it at all at family gatherings?
>
> Rachel: Yeah, but not unless um, the genetic issue, not unless my dad um, or [aunt] who's the one that sent out the letter, she brings it up. I mean the other [family members] that haven't got tested or anything, it's a nonissue. I mean, that's the way it presents. Maybe in their private home, um, you know, there's some other discussion that I'm not aware about, but it's not discussed openly with the extended family. No. And it's, my classification on the outside is a big fat denial.

Interestingly, Rachel's father, Raymond, feels that their extended family actually does talk about the mutation and cancer risk quite a bit. He also provides a reason for why Rachel might be particularly sensitive.

> And so um, my wife and I were um supporting my family and they do talk about it, they do discuss it, they're not afraid or ashamed of it, and Rachel is saying no. But I think Rachel is, she, she's still processing a lot, um, you know she's 28, and, and she's had uh, the bilateral mastectomy and she's not finished with her therapy and um, she has uh, um, she's not happy with the way things are going because she has a lot of pain, and fatigue, she's a little misshapen, and you know, so there's a lot of questions out there. So I think she's a little bit on guard.

Overall, the study data suggest that in families in which a *BRCA* mutation has been identified, there may be individuals who would prefer to ignore it. These people can make their feelings known by avoiding talk about the subject all together. This can create conflict within the families, especially when other family members value open discussions. Again, members of the same family may have different perceptions about how *BRCA* testing has affected the family dynamics.

c. Open communication with family

However, as indicated above, some of the conversations occurring after testing involved open, candid communication about the *BRCA* mutation.

Open communication could be detrimental, as evidenced by an exchange between Danielle and one of her sisters, after Danielle tested positive for the mutation and her sister tested negative. Danielle says:

> She felt this huge sense of relief and said things in the beginning that were inappropriate and very hurtful, like 'I can look at my daughter in my, her eyes and know she's OK.' You know, things like that, that uh, she didn't mean.

For the most part, however, open communication was perceived in a positive manner by the participants in the study. For instance, Rebecca speaks about how happy she is that she can be candid with her nuclear family, even when some of their more extended family members are reluctant to talk.

> I've, I choose not to let it bother me because I think you know, I'm the only one that really needs to worry about this, and my immediate family is clued in and we talk about it all the time. Everyday we see each other, every time we're on the phone we talk about it. And so um, I mean there's other people that are going to be in denial about some of these issues.

Participants whose extended family was resistant to discussion shared that they were forthright about the topic with their own children. For instance, Danielle believed that sharing information with her own young daughters about the mutation and about her own prophylactic mastectomy would prepare them to cope with the cancer risk they may face in adulthood.

> Danielle: I have two beautiful daughters, 8 and 10.
>
> Interviewer: And how are they doing with all of this?
>
> Danielle: Well, they're, it's interesting, you know, [husband] and I have chosen to be, you know, pretty open about this. They don't understand the genetics and that's an OK thing, although, you know we've talked about, they know the words and the gene and that I'm doing this to prevent getting cancer…It's kind of healthy and uh, you know, they see me uh, they see my incisions, they're seeing me you know, as it heals…and uh, in fact on the whole I think it's been really positive and I don't regret how we've dealt with it. They've come on the internet with me when we were looking at [breast] reconstruction and making decisions and decisions like, they're, 'oh look at this one, this one look really good, mom,' and you know, 'dad come see this one,' so I think that's pretty healthy, to deal with it that way. And then like, they may walk this path…and I want them to be uh, you know, whatever they see that you know, if they know this isn't the end of the world, this is one small part in the whole picture…

Open communication in the family can also occur between only certain members, for instance between female *BRCA* mutation carriers, as in the R family. This observation will be discussed shortly.

d. No or few perceived changes, or did not mention it

Although the subject of family discussions emerged in conversations with participants, there were interviews in which this subject was not mentioned or participants did not perceive a change in family discussions or dynamics after genetic testing. These individuals tended to focus their interviews on their decision-making regarding health management options or their involvement in volunteer peer support organizations.

Theme # 3: Feeling More or Less Connected To Family Members

The last major theme discussed here is that after *BRCA* testing in their family, some individuals felt more connected or less connected with certain family members.

a. Feeling a special connection with fellow mutation carriers

Certain study participants expressed that, after receiving their test result, they felt a sense of belonging or a special connection with other family members carrying the mutation. For instance, Rebecca, who underwent predictive genetic testing in her mid 20s after several aunts, her father and her older sister had tested positive for the family mutation, says:

> And so we are, you know, mature and adult, and have been adults through most of the time [her aunts] have been sick, and so again, it's always the topic of discussion and so I feel like an instant sense of belonging and so you know, again because it's not a club I want to belong to but it almost made me feel very welcome, very like, well you know, at least I have this wonderful network.

In fact, this same participant felt so strongly about the connection with other family members that she actually felt relieved that she tested positive, so that she and her sister could go through the experience together.

> This is horrible of course, but I kind of feel like, in a really weird way I felt really, really relieved in a very bizarre way because I had it and my sister did too… We're so close…I almost felt like, 'Well, of course I've got this because she's got this and we do everything together.' And so it was, was easy in a bizarre way to feel, 'Well, I'm not alone and she's not alone and this is just, it's just worked out great.'

Later, she says:

> We've got an amazing bond in addition to all the other things, already being sisters and friends, we have this thing that's brought us so much closer…

Another participant, Naomi, who was the first to undergo testing in her extended family, spoke about how testing positive for a mutation made her feel more connected to some of her deceased family members.

> My 3rd uncle, Nathan, who was my favorite (and my mom's favorite brother — no coincidence, I'm sure), I believe would've "gotten" it, maybe it even would've helped him (as it has me) make sense of his mother's and sister's deaths. I even think about whether it might have helped his health — perhaps (and this is total conjecture) there was a twinge of pain in his stomach that he ignored, that he wouldn't have if he'd known about the mutation. So in that sense I have a sort of melancholy, belated sense of identifying with him.

Thus, some individuals who discover they have a *BRCA* mutation can find themselves identifying more or having a special connection with certain family members, either living or deceased.

b. Feeling left out when not sharing the family mutation

This special connection can make others in the family feel isolated in their own families. For instance, in this study relatives who were not at risk of the mutation, like in-laws, as well as the men in the family, who carry a substantially lower cancer risk than women, often felt helpless or like an outsider in the family. This was perceived by Elise when she said, "I think that my dad feels a little like my brother — left out kind of." This feeling was especially dramatic in the R family. Here Raymond talks about a family gathering in which a 'clique' of female mutation carriers made others feel left out.

> And right after that, we went to a family gathering because it was a baptism of my um, one of my youngest nieces, and all the um, *BRCA2* uh, victims were in a, club, in a little clique talking. And my wife and my, the sister who's never had been tested felt ostracized. And they admitted it, they felt like they were not included in this conversation. They didn't have anything to add, they didn't have anything to uh talk about, they felt they couldn't talk about it so much and they very much felt like they were not in the club. And it was a really strange situation…That um may not be the best news, but to all in the, in the group together and they know certain things and they have certain feelings that nobody else is going to have. Um, I wasn't in that discussion only because they were talking about prophylactic mastectomies, and oophorectomies, and uh, things that weren't going to relate to me so much. Uh and so, but I mean it was like instant cancer club. It was bizarre.

The female family member mentioned here who had not had a cancer diagnosis or genetic testing felt left out since her sisters had all had cancer and were mutation carriers. In fact, one reason she had been reluctant to pursue *BRCA* testing was because she felt if it was confirmed that she did not carry the mutation, she would not feel comfortable with her sisters, or her sisters would be angry. Finally, she did pursue testing, with the blessing of her sisters. Members of the R family talk about this family member.

> Rebecca: She said something like, 'Sometimes I feel because you know a couple of the other girls have cancer, and I don't, I feel like they have a closer bond than I have with them. And so sometimes I feel left out,' she said, 'like when going through something they call each other and they don't necessarily call me because they don't think I can understand.'
>
> Raymond: So after I got my test results she went and did it. Um, partly to, because I think she ostracized that demon, and when admitted that she was afraid that she would be negative, wouldn't feel comfortable being not in the club.

Thus, sharing the mutation and/or sharing the cancer experience associated with it could connect family members but could also isolate others in the family.

c. Feeling isolated when not receiving wanted family support

For many testing positive for the mutation, family support played a significant role in whether they felt connected or isolated in their family. Rebecca seems to have foreseen this when she wanted to test positive so that she could support her sister, Rachel, who had already tested positive for the mutation.

> Raymond: And [Rebecca] would have felt terrible if she had tested negative because then she couldn't be as uh, as supportive and understanding and sympathetic with her sister.

Danielle provides another example, by describing the isolation and hurt she felt when she did not receive support from her family. Danielle is the only one in her family who tested positive, other than an 80 year old uncle.

> Danielle: I didn't expect my friends to understand on the same level. But I did expect my sisters to understand. Either they didn't. Or they weren't able to give me the support I needed. I accept that and I stop looking for it. But the hurt is still there on some level but not to the same intensity. I try to accept them

> for who they are not what they can give. Truly, I don't think that they have any idea.

As Danielle articulates, it is hard to tell if this lack of support was because her sisters could not empathize with her since they had not tested positive or because they lacked the skills to support her at all. In fact, sometimes, different coping mechanisms or management decisions were the reason some family members were not as supportive as others wanted. For instance, Claire discusses that even though she and her sister both tested positive for the mutation, she felt less connected to her because her sister did not understand Claire's decision to have a prophylactic mastectomy.

> There are two situations where I think family bonds have been affected more strongly. One is, um, with my sister who actually ironically tested positive as well, when she and I made different decisions about mastectomy there was a period of time where I felt a lack of connection. I felt like she tried, was trying to be supportive to me, but honestly she wasn't. She didn't understand why I was doing this, she thought I had gone too far. She grieved the loss of my breasts for me, I think. I mean, I did too, but, um, she thought it was quite draconian. And so we kind of like, we lost touch at some point over this. Um, and I think maybe that's kind of getting where we're moving past that now, but there's certainly a period where I felt quite you know, like I wasn't interested in being very close to her because I didn't feel a sincere support.

Negative reactions of family members, as well as the tendency of certain family members to avoid discussing the topic all together, as outlined previously, can be perceived as a similar lack of support, having the potential to make other family members feel isolated.

Naomi, the participant for whom testing positive made her feel more connected to some of her deceased relatives, also felt less connected to other relatives after testing. She explains that this feeling was based on her prediction that they would have coped with knowledge of the mutation differently from her.

> I've never really identified with (or even known) my surviving cousins–going through testing, learning my status, becoming a part of the "world" of people who have a *BRCA* mutation makes me feel even more separate from them. The two uncles that I never got along with, even though they are dead, I feel sort of similar about—like they would never have "gotten" this, or taken it seriously in any way, and this is just another example of how unlike them I am.

Thus, this study provides several examples of when family coping strategies and support seem to make family members feel more or less connected to their relatives.

d. Genetic counseling process as a catalyst for initiating new relationships with relatives

For one participant, Evelyn, genetic testing served to not only change existing relationships but also served as a reason to kindle new relationships with relatives she never knew. This participant did not know her biological father or any of his relatives before this process. However, through researching her family health history to prepare for genetic counseling, she has met many relatives and has become friends with them, something she perceived as a positive outcome.

> However, the up side is that I met many cousins I never knew about. They all knew about me. Several of us are becoming great friends. We are the same ages, we look alike, we are having fun together. So I have to think that my biological father is smiling down on us.

Thus, this research shows that genetic testing has the potential to make individuals feel closer or more distant from certain relatives. These effects can be due to a special connection felt by sharing the mutation, to the types of coping strategies used in the family and the associated support from family, and the establishment of contact with long-lost relatives.

How are These Three Main Themes Connected?

As is likely evident throughout the preceding sections, all three major themes are intricately connected to each other. For instance, disclosing information as part of the role of an index case can incite or influence genetic discussions in the family. In contrast, the types of genetic conversations in the family can impact the burden felt by the index case. As well, the receptivity of family to discussions can cause individuals, including index cases, to feel more or less connected to each other. Likewise, feeling a connection to or a separation from family after testing can influence the dynamics of relationships, having an effect on family discussions at large. Thus, any change in one domain after genetic testing is likely to influence the others, having potential for dramatic change in family dynamics.

Discussion

This study represents one of the few to investigate how testing positive for a *BRCA* mutation can impact relationships among family members. Most participants described some alteration in family dynamics after testing. The changes were perceived as both positive or negative and seemed to depend on the participant's perception of the

family history of cancer, prior relationships among the family, emotional coping strategies of relatives, value placed on particular communication patterns, and sharing or not sharing the family's *BRCA* mutation. It appears that as *BRCA* testing impacts one aspect of a relationship between family members, a ripple effect occurs, influencing other aspects as well.

Conclusions

Theme # 1: New or Changed Family Roles for the Index Case

Studies have shown that individuals often decide to undergo *BRCA* testing to provide information for their family, some even feeling that their family has a right to know this information. Thus, it was not surprising that index cases in the current study tended to feel an obligation or responsibility to inform their relatives of their *BRCA* mutation. The burden they felt when disclosing the information to their families is also consistent with the findings of previous studies (Kenen et al. 2006; McGivern et al. 2004; d'Agincourt-Canning 2001; Hallowell et al. 2005b; Costalas et al. 2003). Importantly, the current study demonstrates that the special role of the family's index case can be transferred to others in the family under certain circumstances. For instance, one study participant assumed the obligation and burden of informing relatives of the mutation after her mother, the first in the family to have *BRCA* testing, passed away from cancer.

Another participant, after being the first in her family to have cancer and test positive for a *BRCA* mutation, felt pressured to explain genetic information and demonstrate for her siblings how to cope emotionally with cancer and genetic testing. In the literature, there are reports of tested individuals feeling burdened by the need to accurately explain the implications of their test results, especially when they themselves do not fully grasp the concepts (Kenen et al. 2006; DudokdeWit et al. 1997; Costalas et al. 2003). However, to our knowledge, there have been no reports of individuals feeling burdened because they are pressured to model emotional coping for their family in the context of *BRCA* testing. Importantly, this participant may have felt this special type of pressure at least partly because she had not been aware of a cancer diagnosis in any family member. Thus, she had no prior family experiences from which to model her own actions or emotions, and instead took on the responsibility of being a model for the rest of her family. The detection of a *BRCA* mutation may have intensified this pressure since it implicates that close family members will likely need guidance at some point, after their own cancer diagnoses or genetic testing.

The new roles that index cases tend to take on after testing positive for a *BRCA* mutation may also be in part due to something special about index cases themselves. One study in particular suggested that index cases think differently than their relatives (Loader et al. 2004). In Loader et. al.'s study individuals interested in genetic testing (i.e. index cases) had to recruit a family member to also undergo testing. It was found that the index cases tended to perceive their own emotional and general health as worse than did their relatives. For their perception of their general health, this was true even when the index cases were healthy and their relatives had cancer. As well, following genetic counseling and testing, breast cancer worry fell for most recruited relatives but remained steady for most index cases. These trends suggest that those individuals who initiate a genetic consult and testing are more anxious about their health and are more preoccupied by breast cancer worry than are some of their relatives. It is possible then that this psychological profile of index cases could at least partially explain the burden they feel after genetic testing. For instance, perhaps their burden derives from predicting that their relatives will feel as anxious and worried as they do with the news of the mutation, when in reality this may not actually be the case. Future study could further investigate the reasons index cases tend to feel burdened in their new family role.

Theme # 2: Genetic Testing Affects Discussions in the Family

As mentioned, several studies have commented on the types of discussions in families after *BRCA* testing, however few made them a primary focus. The current study reveals that, for some families, genetic testing serves as a catalyst for conversations about cancer or for more general contact between family members. These conversations can improve a relationship when they are welcomed or can upset individuals when they are unwelcome. Similarly, it may be common in families for some members to be resistant to discussing cancer or genetic testing, and their reluctance can sometimes isolate or hurt the family members who want to talk about it. Those wanting to talk however do tend to find some relatives to be open with, commonly their children, spouse, or biological relatives with whom they feel a special connection. Open communication can be used to prepare family members like children for the cancer risk they may face in their future.

These results are reminiscent of a study in which Kenen et al. (2004) interviewed unaffected women with a family history of breast and/or ovarian cancer after attending a cancer genetics consult. In contrast to the current study, these women had not had genetic testing, and were not aware of testing results for any of their relatives. Despite this difference, the study by Kenen et al. (2004) identified a very similar range of family communication patterns as the

current study. This would suggest that learning about the potential for a family *BRCA* mutation during a genetic counseling session might result in similar types of family communication as actually receiving the news of carrying a *BRCA* mutation. Perhaps, in both situations, individuals in the family draw from the family scripts, coping mechanisms and heuristics that they have established throughout their lifetimes.

Theme # 3: Feeling More or Less Connected to Family Members

Many participants in the current study described that, after genetic testing, they or other relatives felt more or less connected to certain family members. Sometimes a special connection arose from sharing the *BRCA* mutation with relatives. Something similar has been observed in a study by Duncan et al. (2008) for young people that had genetic testing for HD or familial adenomatous polyposis (FAP), another cancer predisposition syndrome. In the current study, family members who did not share the mutation tended to feel left out or less connected to their family. This has been noted in individuals testing negative for the gene for HD (Sobel and Cowan 2003; Sobel and Cowan 2000). Notably, Sobel and Cowan (2003) described one individual who attempted suicide because her negative test result made her feel disconnected from her siblings, who were already displaying the signs of HD.

The current study also establishes that individuals testing positive for a mutation may be at particular risk of feeling isolated in their family. This has been seen several times before in *BRCA* families (Phelps et al. 2007; Speice et al. 2002) and in families with HD or FAP (Sobel and Cowan 2000; Sobel and Cowan. 2003; Duncan et al. 2008). Specifically, isolation can happen when relatives are not able to empathize and provide the support needed due to different coping skills or management decisions. A recent study by Bakos et al. (2008) found that women testing negative for a known familial *BRCA* mutation recognized this and tended to avoid talking about their mutation status so that their relatives who had tested positive would not feel isolated in the family.

Also, for one participant in the current study, meeting distant relatives while researching her family cancer history was a very positive experience as it allowed her to feel more connected to her family as a whole. However, as Carlsson and Nilbert (2007) point out in their study of families undergoing genetic testing for hereditary non-polyposis colorectal cancer (HNPCC), meeting long-lost relatives may not always be welcomed, and sometimes researching the family cancer history can reveal family secrets like the existence of step/half siblings and extra-marital children.

General Observation

In the families from which more than one member participated in this study, there were several examples of scenarios in which relatives had different perceptions about family dynamics. This was true even though the family members asserted that they had a very close relationship with each other.

Implications for Genetics Practice

Genetic counselors are in the position to assess each client's family situation and identify the potential for shifts in family dynamics after genetic testing. For instance, genetics professionals might consider inquiring about staging or prognosis of affected patients since this information could help recognize whether the role of index case might shift from their client to another relative if the client passes away. As well, recognizing the potential for a special pressure on clients who are the first in their family to have cancer will become increasingly important, since individuals with little known family history of cancer are presenting more commonly to genetics clinics due to the recognition that particular family structures, such as having few women in the family, can mask the presence of a *BRCA* mutation (Weitzel et al. 2007) and thus not everyone who has a *BRCA* mutation has a dramatic family history of cancer. In addition, genetics professionals can investigate the existing connections between family members in the context of the cancer history and any prior genetic testing. Counselors might review with the patient the relationship he or she has with each relative, as well as the coping skills each relative tends to use under stress or a serious situation, in order to try to identify in which relationships there may be conflict or emotional distance after testing.

The next step would be preparing clients for their relatives' reactions to a genetic test result as well as potential shifts in family dynamics. A possible strategy might be to brainstorm with clients about potential relationship changes, what change would mean to them, and how it might impact them after testing. As well, as seen in this study, family members can have different perceptions about changes in family dynamics, so genetics professionals could draw upon this observation and offer to teach skills to families or provide a physical venue for family members to discuss their feelings openly with one another. Encouraging such communication might prevent feelings of isolation in the family and might promote awareness of the difficult new family roles that some family members assume after testing.

Counselors might also focus on follow up of clients and their families weeks and months after testing in order to support and guide them through changes in family

dynamics. Family or individual therapy may be a useful referral for some of these families.

In order to prepare genetics professionals for these types of interventions, training programs should consider how to address the types of psychosocial concerns observed in our study and others. Ideally, both the classroom and the clinical domains of their education programs could include relevant training. Specific interventions could include a renewed focus on family counseling skills in training programs as well as routine instruction and practice in the use of genograms to gather information about existing family relationships (Daly et al. 1999; Eunpu 1997). Genograms are an established tool in the field of family therapy, and in hereditary breast/ovarian cancer families they have been shown to provide valuable insight into the nature of one's social world and grieving processes, as well as a starting point from which to formulate interventions (Peters et al. 2006). Eventually, the efficacy of counseling interventions should be assessed in order to determine how well genetics professionals are addressing concerns about *BRCA* testing and the family.

Psychosocial complexity of *BRCA* testing can also be referenced by genetics professionals when establishing or continuing relations with primary care providers (PCPs). Recent marketing efforts by genetic testing laboratories have placed pressure on PCPs to order genetic tests and interpret the results. However, there is concern that PCPs in busy practices have neither the time nor the specialized training to address the psychosocial implications of such testing. Effort could be directed toward educating PCPs of genetic counseling services and targeting them as a referral agent, underlining the potential for PCPs to save time by referring their patients to genetics.

Study Limitations and Future Research

a. Recruitment and selection bias

Since the original study involved in-depth interviews, it might have attracted the individuals who were more willing to discuss their experience with *BRCA* testing. Using the FORCE website to recruit for the study may have also selected for individuals who tended to be open or proactive about their cancer risk, since they may have been visiting the site to participate in the online support community or to review the extensive information featured on the site about risk management options. Attempts were made to counter this bias by asking participants if they thought their family members would be interested in participating in the study. However, if family relationships were already strained by *BRCA* testing, participants might have been unwilling to ask their family members to participate. Thus, this bias may have been unavoidable in some cases, and participants were asked to richly describe interactions with their family members in order to gain the most accurate information as possible about their relationships with relatives that are unwilling to participate. Future studies involving several members of the same family could be performed in order to better understand the differing viewpoints that seem to be occurring among family members.

Another limitation was that all participants in the study had tested positive for a *BRCA* mutation. Further studies should address the experiences of individuals testing negative for a mutation, both in families in which a mutation had been identified as well as in families in which no prior family member had been found to have a mutation. This would be useful in appreciating the full spectrum of experiences that members of tested families can have. Bakos et al. (2008) provide a preliminary description of the experience of women testing negative for a known *BRCA* mutation in the family.

b. Limitations of secondary data analysis

The grounded theory and open-ended interview methodology used in the original study promoted a rich description of the experience that the participants chose to focus on. However, the original study did not specifically focus on family relationships after *BRCA* testing and if this topic was not the focus of some participants, little insight into the dynamics of their families could be gleaned from the secondary analysis. For example, a few study participants spoke almost exclusively about their experiences with management choices like surgeries or their involvement in community-based organizations. When asked about if or how *BRCA* testing had influenced their relationships with their families, they would either deny that there was any influence or they would answer with a short statement only. It is hard to discern whether they really did not perceive any change within their families or if they did perceive a change but chose not to discuss it. In situations where there really were no or only minimal changes in family dynamics, a possible next question would be *why?* Future studies could try to outline the profile of families whose dynamics are reportedly not seriously affected by genetic testing, so that genetics professionals could use this information to identify families at high risk for serious negative changes in family relationships, and try to minimize these upsetting effects.

Another limitation in the interview process was that not all participants were asked in their second interview whether and how their relationships with their families had changed since the first interview. Thus, from the current secondary data analysis, it is hard to describe any trends in how families might incorporate the knowledge of a mutation into the way they interact with each other in the long term. We are planning on following the same cohort of

participants for several more years, so we will use the current study to prompt the participants to comment on how family dynamics may have changed over time.

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APPENDIX A: Initial questions for original study

1 Go back with me to when you first started thinking of having a predictive genetic test and walk me through your thinking.
2 Think back to when you first started hearing conversations in your family about cancer. What was being said? Who said it? What effect did that have on you—on thinking about your own health?
3 What went in to your thinking about having the genetic test? Did you talk to anyone? Who? How did you decide who to talk to and who not to?
4 What about the positive result created a change in how you thought about your own health, your own self, your family?

References

Bakos, A. D., Hutson, S. P., Loud, J. T., Peters, J. A., Giusti, R. M., & Greene, M. H. (2008). BRCA mutation-negative women from hereditary breast and ovarian cancer families: a qualitative study of the BRCA-negative experience. *Health Expectations, 11*(3), 220–231.

Beeson, D. (1997). Nuance, complexity, and context: qualitative methods in genetic counseling research. *Journal of Genetic Counseling, 6*(1), 21–43.

Blandy, C., Chabal, F., Stoppa-Lyonnet, D., & Julian-Reynier, C. (2003). Testing participation in BRCA1/2-positive families: initiator role of index cases. *Genetic Testing, 7*(3), 225–233.

Bradbury, A. R., Dignam, J. J., Ibe, C. N., Auh, S. L., Hlubocky, F. J., Cummings, S. A., et al. (2007). How often do BRCA mutation carriers tell their young children of the family's risk for cancer? A study of parental disclosure of *BRCA* mutations to minors and young adults. *Journal of Clinical Oncology, 25*(24), 3705–3711.

Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative Research in Psychology, 3*, 77–101.

Bredart, A., Autier, P., Audisio, R. A., & Geragthy, J. (1998). Psychosocial aspects of breast cancer susceptibility testing: a literature review. *European Journal of Cancer Care, 7*, 174–180.

Carlsson, C., & Nilbert, M. (2007). Living with hereditary non-polyposis colorectal cancer; experiences from and impact of genetic testing. *Journal of Genetic Counseling, 16*(6), 811–820.

Claes, E., Evers-Kiebooms, G., Boogaerts, A., Decruyenaere, M., Denayer, L., & Legius, E. (2003). Communication with close and distant relatives in the context of genetic testing for hereditary breast and ovarian cancer in cancer patients. *American Journal of Medical Genetics, 116A*, 11–19.

Costalas, J. W., Itzen, M., Malick, J., Babb, J. S., Bove, B., Godwin, A. K., et al. (2003). Communication of BRCA1 and BRCA2 results to at-risk relatives: a cancer risk assessment program's experience. *American Journal of Medical Genetics. Part C, Seminars in Medical Genetics, 119C*, 11–18.

d'Agincourt-Canning, L. (2001). Experiences of genetic risk: disclosure and the gendering of responsibility. *Bioethics, 15*(3), 231–247.

d'Agincourt-Canning, L. (2006). A gift or a yoke? Women's and men's responses to genetic risk information from *BRCA1* and *BRCA2* testing. *Clinical Genetics, 70*, 462–472.

Daly, M., Farmer, J., Harrop-Stein, C., Montgomery, S., Itzen, M., Costalas, J., et al. (1999). Exploring family relationships in cancer risk counseling using the genogram. *Cancer Epidemiology, Biomarkers & Prevention, 8*, 393–398.

Daly, M. B., Barsevick, A., Miller, S. M., Buckman, R., Costalas, J., Montgomery, S., et al. (2001). Communicating genetic test results to the family: a six-step, skills-building strategy. *Family and Community Health, 24*(3), 13–26.

Deatrick, J. A., Knafl, K. A., & Guyer, K. (1993). The meaning of caregiving behaviors: Inductive approaches to family theory development. In S. L. Featham, S. B. Meister, J. M. Bell & C. L. Gillis (Eds.), *The nursing of families: Theory/research/education/practice*, pp. 38–45. Newbury Park, CA: Sage.

DudokdeWit, A. C., Tibben, A., Frets, P. G., Meijers-Heijboer, E. J., Devilee, P., Klijn, J. G. M., et al. (1997). *BRCA1* in the family: a case description of the psychological implications. *American Journal of Medical Genetics, 71*, 63–71.

Duncan, R. E., Gillam, L., Savulescu, J., Williamson, R., Rogers, J. G., & Delatycki, M. B. (2008). "You're one of us now": young people describe their experiences of predictive genetic testing for Huntington disease (HD) and familial adenomatous polyposis (FAP). *American Journal of Medical Genetics Part C (Seminars in Medical Genetics), 148C*, 47–55.

Eunpu, D. L. (1997). Systemically-based psychotherapeutic techniques in genetic counseling. *Journal of Genetic Counseling, 6*(1), 1–20.

Forrest, K., Simpson, S. A., Wilson, B. J., van Teijlingen, E. R., McKee, L., Haites, N., et al. (2003). To tell or not to tell: barriers and facilitators in family communication about genetic risk. *Clinical Genetics, 64*, 317–326.

Foster, C., Watson, M., Moynihan, C., Jones, A., & Eeles, R. (2002). Genetic testing for breast and ovarian cancer predisposition: cancer burden and responsibility. *Journal of Health Psychology, 7*, 469–484.

Foster, C., Evans, D. G. R., Eeles, R., Eccles, D., Ashley, S., Brooks, L., et al. (2004). Non-uptake of predictive genetic testing for *BRCA1/2* among relatives of known carriers: attributes, cancer worry, and barriers to testing in a multicenter clinical cohort. *Genetic Testing, 8*(1), 23–29.

Goelen, G., Rigo, A., Bonduelle, M., & De Greve, J. (1999). Moral concerns of different types of patients in clinical *BRCA1/2* gene mutation testing. *Journal of Clinical Oncology, 17*(5), 1595–1600.

Green, J., Richards, M., Murton, F., Statham, H., & Hallowell, N. (1997). Family communication and genetic counseling: the case of hereditary breast and ovarian cancer. *Journal of Genetic Counseling, 6*(1), 45–60.

Hallowell, N. (1999). Doing the right thing: genetic risk and responsibility. *Sociology of Health & Illness, 21*(5), 597–621.

Hallowell, N., Foster, C., Eeles, R., Arden-Jones, A., Murday, V., & Watson, M. (2003). Balancing autonomy and responsibility: the ethics of generating and disclosing genetic information. *Journal of Medical Ethics, 29*, 74–83.

Hallowell, N., Foster, C., Eeles, R., Ardern-Jones, A., & Watson, M. (2004). Accommodating risk: responses to BRCA1/2 genetic testing of women who have had cancer. *Social Science & Medicine, 59*, 553–565.

Hallowell, N., Ardern-Jones, A., Eeles, R., Foster, C., & Lucassen, A. (2005a). Communication about genetic testing in families of male *BRCA1/2* carriers and non-carriers: patterns, priorities and problems. *Clinical Genetics, 67*, 492–502.

Hallowell, N., Ardern-Jones, A., Eeles, R., Foster, C., Lucassen, A., Moynihan, C., et al. (2005b). Men's decision-making about predictive *BRCA1/2* testing: the role of family. *Journal of Genetic Counseling, 14*(3), 207–17.

Hamilton, R. J. (2003). *Experiencing predictive genetic testing in families with Huntington's disease and hereditary breast and ovarian cancer.* Unpublished Dissertation, Madison, WI.

Hamilton, R. J., & Bowers, B. J. (2006). Internet recruitment and e-mail interviews in qualitative studies. *Qualitative Health Research, 16*(6), 821–835.

Hamilton, R. J., & Bowers, B. J. (2007). The theory of genetic vulnerability: a Roy model exemplar. *Nursing Science Quarterly, 20*(3), 254–265.

Hamilton, R. J., Bowers, B. J., & Williams, J. K. (2005). Disclosing genetic test results to family members. *Journal of Nursing Scholarship, 37*(1), 18–24.

Hamilton, R. J., Williams, J. K., Bowers, B. J., & Calzone, K. (2009). Life trajectories, genetic testing, and risk reduction decisions in 18–39 year old women at risk for hereditary breast and ovarian cancer. *Journal of Genetic Counseling*, doi:10.1007/s10897-008-9200-1.

Hinds, P. S., Vogel, R. J., & Clarke-Steffen, L. (1997). The possibilities and pitfalls of doing secondary analysis of a qualitative data set. *Qualitative Health Research, 7*(3), 408–424.

Hughes, C., Lerman, C., Schwartz, M., Peshkin, B. N., Wenzel, L., Narod, S., et al. (2002). All in the family: evaluation of the process and content of sisters' communication about *BRCA1* and *BRCA2* genetic test results. *American Journal of Medical Genetics, 107*, 143–150.

Kenen, R., Ardern-Jones, A., & Eeles, R. (2003a). Living with chronic risk: healthy women with a family history of breast/ovarian cancer. *Health, Risk & Society, 5*(3), 315–331.

Kenen, R., Ardern-Jones, A., & Eeles, R. (2003b). Family stories and the use of heuristics: women from suspected hereditary breast and ovarian cancer (HBOC) families. *Sociology of Health & Illness, 25*(7), 838–865.

Kenen, R., Ardern-Jones, A., & Eeles, R. (2004). We are talking, but are they listening? Communication patterns in families with a history of breast/ovarian cancer (HBOC). *Psycho-Oncology, 13*, 335–345.

Kenen, R., Ardern-Jones, A., & Eeles, R. (2006). "Social separation" among women under 40 years of age diagnosed with breast cancer and carrying a *BRCA1* or *BRCA2* mutation. *Journal of Genetic Counseling, 15*(3), 150–162.

Kenen, R. H., Shapiro, P. J., Hantsoo, L., Friedman, S., & Coyne, J. C. (2007). Women with *BRCA1* or *BRCA2* mutations renegotiating a post-prophylactic mastectomy identity: self-image and self-disclosure. *Journal of Genetic Counseling, 16*, 789–798.

Knafl, K. A., Ayers, L., Gallo, A. M., Zoeller, L. H., & Breitmayer, B. J. (1995). Learning from stories: parent's accounts of pathways to diagnosis. *Pediatric Nursing, 21*(5), 4122–4415.

Loader, S., Shields, C. G., & Rowley, P. T. (2004). Impact of genetic testing for breast-ovarian cancer susceptibility. *Genetic Testing, 8*(1), 1–12.

Loud, J. T., Weissman, N. E., Peters, J. A., Giusti, R. M., Wilfond, B. S., Burke, W., et al. (2006). Deliberate deceit of family members: a challenge to providers of clinical genetics services. *Journal of Clinical Oncology, 24*(10), 1643–1646.

MacDonald, D. J., Sarna, L., van Servellen, G., Bastani, R., Giger, J. N., & Weitzel, J. N. (2007). Selection of family members for communication of cancer risk and barriers to this communication before and after genetic cancer risk assessment. *Genetics in Medicine, 9*(5), 275–282.

McGivern, G., Everett, J., Yager, G. G., Baumiller, R. C., Hafertepen, A., & Saal, H. M. (2004). Family communication about positive *BRCA1* and *BRCA2* genetic test results. *Genetics in Medicine, 6* (6), 503–509.

McInerney-Leo, A., Biesecker, B., Hadley, D. W., Kase, R. G., Giambarresi, T. R., Johnson, E., et al. (2005). *BRCA1/2* testing in hereditary breast and ovarian cancer families II: impact on relationships. *American Journal of Medical Genetics, 133A*, 165–169.

Patenaude, A. F., Dorval, M., DiGianni, L. S., Schneider, K. A., Chittenden, A., et al. (2006). Sharing *BRCA1/2* test results with first-degree relatives: factors predicting who women tell. *Journal of Clinical Oncology, 24*(4), 700–706.

Peters, J. A., Hoskins, L., Prindiville, S., Kenen, R., & Greene, M. H. (2006). Evolution of the colored eco-genetic relationship map (CEGRM) for assessing social functioning in women in hereditary breast-ovarian (HBOC) families. *Journal of Genetic Counseling, 15*(6), 477–489.

Phelps, C., Wood, F., Bennett, P., Brain, K., & Gray, J. (2007). Knowledge and expectations of women undergoing cancer genetic risk assessment: a qualitative analysis of free-text questionnaire comments. *Journal of Genetic Counseling, 16*(4), 505–14.

Raveis, V. H., & Pretter, S. (2005). Existential plight of adult daughters following their mother's breast cancer diagnosis. *Psycho-Oncology, 14*, 49–60.

Richards, L., & Morse, J. M. (2007). Coding. In *Readme first for a user's guide to qualitative methods* (2nd ed., pp. 133-151). Thousand Oaks, CA: Sage Publications, Inc.

Smith, K. R., West, J. A., Croyle, R. T., & Botkin, J. R. (1999). Familial context of genetic testing for cancer susceptibility: moderating effect of siblings' test results on psychological distress one to two weeks after *BRCA1* mutation testing. *Cancer Epidemiology, Biomarkers & Prevention, 8*, 385–392.

Sobel, S., & Cowan, D. B. (2000). Impact of genetic testing for Huntington disease on the family system. *American Journal of Medical Genetics, 90*, 49–59.

Sobel, S., & Cowan, C. B. (2003). Ambiguous loss and disenfranchised grief: the impact of DNA predictive testing on the family as a system. *Family Process, 42*(1), 47–57.

Speice, J., McDaniel, S. H., Rowley, P. T., & Loader, S. (2002). Family issues in a psychoeducation group for women with a *BRCA* mutation. *Clinical Genetics, 62*, 121–127.

Stroup, A. M., & Smith, K. R. (2007). Familial effects of *BRCA1* genetic mutation testing: changes in perceived family functioning. *Cancer Epidemiology Biomarkers & Prevention, 16*, 135–141.

Tercyak, K. P., Peshkin, B. N., DeMarco, T. A., Farkas Patenaude, A., Schneider, K. A., Garber, J. E., et al. (2007). Information needs of mothers regarding communicating *BRCA1/2* cancer genetic test results to their children. *Genetic Testing, 11*(3), 249–255.

Thorne, S. (1994). Secondary analysis in qualitative research: issues and implications. In J. M. Morse (Ed.), *Critical issues in qualitative research methods* (1st ed.), pp. 263–279. Thousand Oaks, CA: Sage Publications, Inc.

Van Dijk, S., Van Asperen, C. J., Jacobi, C. E., Vink, G. R., Tibben, A., Breuning, M. H., et al. (2004). Variants of uncertain clinical significance as a result of BRCA1/2 testing: impact of an ambiguous breast cancer risk message. *Genetic Testing, 8*(3), 235–239.

Van Oostrom, I., Meijers-Heijboer, H., Lodder, L. N., Duivenvoorden, H. J., van Gool, A. R., Seynaeve, C., et al. (2003). Long-term psychological impact of carrying a BRCA1/2 mutation and prophylactic surgery: a 5-year follow-up study. *Journal of Clinical Oncology, 21*, 3867–3874.

Watson, M., Foster, C., Eeles, R., Eccles, D., Ashley, S., Davidson, R., et al. (2004). Psychosocial impact of breast/ovarian (BRCA1/2) cancer-predictive genetic testing in a UK multi-centre clinical cohort. *British Journal of Cancer, 91*, 1787–1794.

Weitzel, J. N., Lagos, V. I., Cullinane, C. A., Gambol, P. J., Culver, J. O., Blazer, K. R., et al. (2007). Limited family structure and *BRCA* mutation status in single cases of breast cancer. *Journal of the American Medical Association, 297*(23), 2587–2595.

Werner-Lin, A. V. (2007). Danger zones: risk perceptions of young women from families with hereditary breast and ovarian cancer. *Family Process, 46*(3), 335–349.