

Les conséquences de présenter d'importants antécédents familiaux de cancer du sein

Christine Maheu

Les constations formulées dans le présent article sont tirées d'une étude qualitative au cours de laquelle des données ont été recueillies auprès de 20 femmes qui avaient reçu des résultats non concluants à la suite d'un test génétique pour mesurer une susceptibilité héréditaire au cancer du sein. Avant de parler de la signification qu'elles accordaient à leurs résultats, toutes les participantes ont décrit comment elles vivaient le fait de présenter d'importants antécédents familiaux de cancer du sein. Le présent article porte principalement sur l'expérience des femmes présentant des antécédents personnels et familiaux de cancer du sein. Pour ces femmes, de tels antécédents sont devenus une réalité qu'il est impossible d'ignorer. À partir des données recueillies, trois thèmes ont été dégagés : *anticiper un diagnostic de cancer du sein et y réagir, se protéger et protéger les autres, et avoir à subir davantage d'exams de dépistage du cancer*. Ces thèmes portent sur la réalité fondamentale qui entoure le fait de présenter des antécédents personnels et familiaux de cancer du sein, c'est-à-dire qu'il ne s'agit pas d'une situation isolée, mais plutôt d'une partie du parcours d'une personne dans le choix de se soumettre ou non à des tests génétiques en vue de déterminer une susceptibilité au cancer du sein.

Mots clés : cancer du sein, antécédents familiaux

Implications of Living With a Strong Family History of Breast Cancer

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The findings presented here are from a qualitative study in which data were gathered from 20 women who had received inconclusive genetic testing results for inherited breast cancer susceptibility. Before describing the significance, for them, of their genetic test results, all of the participants related what it was like to live with a strong family history of breast cancer. The focus of this article is the women's experience of living with a personal and strong family history of breast cancer. For these women, having such a history had become a fact of life that could not be ignored. Three themes were identified in the data: *expecting and dealing with a diagnosis of breast cancer*, *protecting oneself and others*, and *increasing exposure to cancer screening procedures*. These themes address the underlying reality that having a personal and family history of breast cancer is not an isolated situation but part of one's journey in choosing to undergo genetic testing for inherited breast cancer susceptibility.

Keywords: breast cancer, family history, qualitative, interpretive description

A family history of the disease is recognized as one of the most important risk factors for breast cancer (Emery, Lucassen, & Murphy, 2001; Yang & Lippman, 1999). A family history with the following characteristics indicates probable genetic susceptibility: breast and/or ovarian cancer in two or more first-degree relatives (mother, sister, or daughter); young age at diagnosis; and breast cancer appearing on the same side of the family, among same-blood relatives. Genetic testing for inherited breast cancer susceptibility is usually reserved for individuals who are assessed at risk of predisposition because of their personal and strong family history of the disease. Three types of result are possible with such genetic testing. The individual can be found to carry an inherited mutation and therefore receives a *positive* test result. When a mutation has been identified in a family, those family members who opt for testing can either be found to carry the familial mutation or be told that they do not carry it; hence, they receive the second type of result, a *true negative*. The third type of test result is *inconclusive*. The result is inconclusive when a mutation is not detected in individuals with a past personal cancer diagnosis, from a family at high risk of the disease, who have no prior identified familial mutation (Carter, 2001; Dorval et al., 2005).

The data-based literature shows that individuals from families with a strong history of breast cancer demonstrate intense interest in genetic testing (Bottorff et al., 2002). While this population shows great interest in genetic testing, it also exhibits a high degree of psychological cancer distress (Friedman et al., 2006). Zakowski et al. (1997) and Zakowski, Valdimarsdottir, and Bovberg (2001) report that individuals from families with a history of cancer are known to experience symptoms of general distress, to have frequent intrusive thoughts, and to at times deny their risk of cancer. According to Baum, Friedman, and Zakowski's (1997) theoretical model of stress and genetic testing for disease risk, individuals who perceive themselves to be at increased risk of cancer because of a strong family history but who have no identifiable mutations may experience stress similar to that exhibited by those who are found to carry an inherited cancer mutation. Baum et al. postulate that, for the former population, an inconclusive genetic test result does not reduce their uncertainty about the etiology of their cancer history, with the resultant distress adding to the distress from perceived risk of an inherited mutation. To date there have been few studies describing this unique pool of individuals: those who have a personal and family history of breast cancer and who have received inconclusive genetic test results for inherited susceptibility (Frost, Venne, Cunningham, & Gerritsen-McKane, 2004; Hallowell, Foster, Eeles, Arden-Jones, & Watson, 2004). In the large qualitative study on which this article is based, 17 of the 20 women interviewed took their inconclusive results to mean that there was still a possibility they carried a breast cancer mutation (Maheu & Thorne, 2008).

Consequently, empirical research looking at the implications of living with a personal and family cancer history indicating a probable inherited genetic susceptibility seems warranted. We need to better understand how such implications create unique health and illness experiences in the context of clinical genetics. The present article addresses this gap by describing women's experiences of living with both a breast cancer diagnosis and a strong family history of breast cancer that indicates probable inherited susceptibility. The findings presented here are from a qualitative study with women who received inconclusive genetic test results (Maheu & Thorne, 2008).

Method

The study was guided by the interpretive description approach (Thorne, Reimer Kirkham, & MacDonald-Emes, 1997). This approach recognizes the contextual and constructed nature of the health and illness experiences of those who come into contact with clinical settings and relates how clinical context can influence an individual's subjective interpreta-

tion of their experience. The purpose of interpretive description is to identify common clinical phenomena that, within individuals' contexts, generate new understandings and new meanings of the phenomenon under study (Thorne et al., 1997).

Recruitment and Sampling Procedures

Recruitment took place within one Hereditary Cancer Program (HCP) in Canada. A sample of 21 women was drawn from a pool of 250 who spoke English, had a previous breast cancer diagnosis, had already undergone genetic testing for inherited breast cancer susceptibility, and were considered to have a strong family history of breast cancer (having met eligibility criteria for genetic testing that predicted a 10% to 20% chance of finding a mutation). The women were selected with the assistance of genetic counsellors and the HCP's educational nurse. They had been identified by the health professionals because of their openness during genetic counselling about their views on genetic testing for inherited breast cancer susceptibility and because of their divergent cancer backgrounds. First, the health professionals sought the women's permission to be contacted for research. Next, I contacted interested individuals to further describe the goal of the study and to seek their participation. Of the 21 women who were approached, one declined, as she was experiencing a second primary breast cancer. Once verbal consent was obtained, the 20 women were sent an information letter, the interview guide, and an informed consent form to be signed on the day of the interview. Clearance to conduct the study was obtained from the Behavioural Research Ethics Board at the University of British Columbia and from the HCP.

Data Collection

In this study, semi-structured interviews captured participants' experience of living with a personal and strong family history of breast cancer. Each interview began with the interviewer asking the woman to describe, in a story format with a beginning, middle, and end, her personal experiences with breast cancer and the experiences of others in her family. I strategically used this opening segment to help the participant feel comfortable telling her story in the open-interview format. During the course of the interview, I used prompts to guide the woman in exploring her experience of living with a personal and family history of breast cancer. These prompts included the following: *How has breast cancer affected your life? How did you feel when a family member was diagnosed with breast cancer? Did this influence your thoughts of your own risk or the risk of others in your family? Do you have an explanation for the appearance of breast cancer in you and in your family members?* All but three of the interviews took place

in the participant's home. The interviews lasted 60 to 90 minutes on average. All participants were asked to choose a code name to safeguard their anonymity. All transcribed interviews were transferred to QSR N5 software for qualitative analysis, for ease of data management and retrieval.

Data Analysis

Data collection and analysis were conducted iteratively over 1 year. The analysis consisted first of identifying key statements that spoke to the implications of living with a strong family history of breast cancer. These key statements were recurrent beliefs and ideas cited by the participants while telling their stories. These key statements served as initial identification of themes. While searching for themes in the interview data, I asked myself: *What led this participant to respond in this way? What am I hearing and not hearing? What is different and similar within the interviews conducted thus far?* I marked these insights as *outstanding questions* in my field notes, to be either tested or negated by the next interviewees.

The reflection allowed for clarification of other emerging themes. In subsequent interviews, the woman was asked to reflect on the meaning of emergent themes for her lived health and for her illness experiences with breast cancer. This strategy contributed to the validity of the findings. I also contrasted identified themes with individual and aggregate stories in order to assess how context influences and alters the experience of living with a personal and family history of breast cancer. After comparing the themes in the 20 different experiential contexts, I could see a pattern in the overall experience of deciding to undergo genetic testing for inherited breast cancer susceptibility. As one participant explained, the women had not "arrived at the decision to have genetic testing overnight" but had been led, by certain factors, to become open to this new technology. One major factor was the implications of having a personal and family history of breast cancer.

Findings

The 20 women ranged in age from 41 to 70. More than half were married and more than half had a relatively high level of education. All except one were Caucasian. Nine of the 20 had received a breast cancer diagnosis while under the age of 40. Twelve had a mother diagnosed with breast cancer and seven had a sister diagnosed with breast cancer. For 11 of the women, three or more family members in the last two generations, on the same side of the family, had been diagnosed with breast cancer.

Three themes associated with the implications of living with a family history of breast cancer ran through the interviews: *expecting and dealing with a diagnosis of breast cancer*, *protecting oneself and others*, and *increasing expo-*

sure to cancer screening procedures. Each of these themes will be discussed separately.

Expecting and Dealing With a Diagnosis of Breast Cancer

Having been in close contact with family members who had developed breast cancer, the women had come to expect that they would get the disease themselves at some point. One woman, Juniper, had seen her mother develop breast cancer while Juniper was still a preteen. She described her own experience: "I basically prepared myself to have cancer all those years. It was like I knew that probably at some point I would have cancer." The following contextual description of Juniper's life reveals how she became comfortable with the word "cancer."

When Juniper's mother was diagnosed with breast cancer at the age of 45, she pulled Juniper and her sister out of school so they could look after their younger brother, feeling that she was no longer able to do so herself. Juniper said that her mother went into a depression and showed the many facets of the illness to her three young children. The children became their mother's only support system. Juniper stated that her mother's mental pain was more difficult for the children to deal with than her physical pain, even when she showed them the gruesome scars from her radical mastectomy.

Like her mother, Juniper was diagnosed with breast cancer at the age of 45. She explained that, although she had not expected to get breast cancer one day, she felt that she had already experienced the diagnosis through her mother's cancer, and she did not fear it; in fact when Juniper received her breast cancer diagnosis, she did not experience intense shock.

This one participant expressed the cancer risk awareness of many of the others. The women explained that it was difficult to let go of their perception of increased cancer risk. They feared a recurrence or another primary cancer, knowing that this was a real possibility, as they had seen it happen to other family members. The women described how they looked to past generations for possible scenarios of how breast cancer would strike their generation — themselves as well as their family members. Those with daughters also worried that they would be diagnosed with breast cancer. Interestingly, one woman reported that her daughter did not fear cancer but had come to accept it as inevitable. Another participant, Donna, related a conversation between herself and her daughter:

My daughter said, "I know what happened to my grandmother and to you. I expect it to happen to me, but I don't really want to think about

that now." My daughter has kind of accepted that this is something that's going to happen. She's just waiting for the other shoe to drop.

The women's awareness of their family disease and their learning to live with the heightened risk help to explain why many of them were comfortable with the word cancer and saw it as the family norm. The women explained that it was an accepted topic of conversation at family gatherings, triggered either by a recent diagnosis or by the many reminders of breast cancer in the homes of family members. Those reminders, they said, became part of their family history. One woman made an analogy between the expectation of breast cancer diagnoses in her family and society's expectation of car accidents because there are so many cars on the road: "There's always the thought that . . . someone else in the family will be diagnosed."

Nineteen of the 20 women said that their perceived high risk made their cancer diagnosis less stressful than it otherwise might have been. The one exception, Erika, had immigrated to Canada relatively young and had little contact with her extended family. Her mother had not been diagnosed with breast cancer and was unaware of any other breast cancer diagnoses in the family except for her grandmother. Erika herself had been diagnosed at 35.

In Stephanie's case, conversely, both her mother and her grandmother had been diagnosed with breast cancer in their early 30s. Stephanie commented, "What else was I to assume?" Stephanie was diagnosed with precancerous cells in her cervix at 20, received a second cancer diagnosis at 35 when she developed uterine cancer, and was diagnosed with breast cancer at 50. Stephanie not only had come to expect a diagnosis of breast cancer one day, in light of her family history of cancer, but had become comfortable with the word cancer.

Although some of the women had become comfortable with the word cancer, two said that they experienced difficulty with losing their breast. They explained that, although images of breastless women were not strange to them, they still felt ill at ease:

Having most of my adult life seen my mother with no breast, it wasn't as if I didn't know what it was going to look like. You get used to the word cancer and are almost comfortable with it, even when one gets a diagnosis. But what has been harder is losing the breast. It's like there [are] two different issues happening. I had a harder time with losing my breasts [than with] having cancer.

Four other women spoke of losing a part of themselves and their femininity when they had their breasts removed as part of their cancer treatment. They described the event as a disruption of their self-identity;

living with breastlessness or with reconstructed breasts forced them to rebuild their self-identity. For two other women, however, breast removal was a positive event. These participants explained that they had, as they put it, finally got “rid of it” — the source of their fear and anxiety. Gilligan said that her mastectomy decreased her personal risk of cancer while permitting her to take on more risk in other aspects of her life, such as her professional life.

The accounts above concern how women dealt with their own cancers. Coming from families with a high prevalence of cancer, they also had to learn to deal with cancer diagnoses and deaths among family members. More than three quarters of the women explained that they had to deal with disrupted plans due to new diagnoses or deaths. These are further implications of living with a strong family history of cancer. The participants said that when they made plans, unlike people with no family cancer history, they often had to ask, *What if someone gets a diagnosis?* For example, Gladys said that her own vacation plans and those of her eldest sister were sadly derailed when the middle sister received her fourth cancer diagnosis. For Emma, disrupted plans meant broken dreams. The following is Emma’s biographical account of broken dreams after her two sisters died from breast cancer. It is constructed from segments of her interview.

It was only recently that Emma was able to speak about the death of her sisters. Emma had planned to grow old with them. Instead, she found herself nursing both of them to their deaths from breast cancer. One of Emma’s sisters had been like a mother to her. When this sister received her diagnosis, Emma and her husband decided to sell their home and move closer to her. Soon after the move, Emma took her sister into their new home, where she cared for her until the end. Following the death of her two sisters, Emma was diagnosed with breast cancer as well, at the age of 43. At the time of the interview she was still suffering from survivor guilt.

Protecting Oneself and Others

Individuals who believed that cancer was the norm in their family viewed themselves as the family guardian. They made it their responsibility to oversee the cancer screening behaviours of their siblings and sometimes of extended family members as well. Who was guarded in the family and who was not seemed to depend on one’s geographical and emotional proximity to the guardian. Some women explained that, as family guardian, they had genetic testing on behalf of family members who did not meet the eligibility criteria for testing, such as having a previous cancer diagnosis. In their role as family guardian, these women

expressed a need for ongoing professional support to keep up to date with information on breast cancer.

For many of the women, receiving an inconclusive genetic test result left them uncertain about the etiology of their cancer. To understand it, the women compared themselves to others in their families, seeking family risk factors for breast cancer. Three women specifically posed an interesting question: If they could not determine why they had developed breast cancer in the first place, how could they know whether they were being diligent enough to prevent another cancer? Many of the participants, perhaps as a result of being family guardians and aspiring role models, were proactively trying to reduce their risk of breast cancer. Erika reported becoming highly aware of anything that could be carcinogenic; at one point she had almost stopped eating for fear of consuming carcinogenic agents. Victoria had decided to grow an organic garden in the summer and to avoid all non-organic vegetables and fruits during the rest of the year.

Increasing Exposure to Cancer Screening Procedures

The women's accounts suggest that they had gone through much more cancer screening and testing than undergone by the average woman. As one woman explained, "I understand that my lumpy breasts are not the same as your lumpy breasts." The participants commented that, in their case, a suspicious lumpy breast or fibrous cyst would be investigated more thoroughly because of their family cancer history. For some of the women, before they received their first diagnosis of breast cancer they had already experienced much cancer screening and testing:

By the time I was barely 30 years old I had already experienced my first mammogram. By the time I was 40 I had [had] about five or six done. And by the time I was 41 I had [had] one breast aspirated for a fibrous cyst. Yes, I had been thinking about cancer for a long time. Then you also have those "worrisome mammograms" that tend to lead to other mammograms soon after.

As well, a breast cancer diagnosis in the family often served as a sharp reminder to others to get screened, whether or not they were due for their regular checkup. One woman described how her breast cancer diagnosis prompted her three sisters to have their mammograms redone. As a result two sisters were told that their mammograms were normal and the other was found to have a suspicious lump; this led to more tests, which revealed a malignant tumour.

Participants explained that a strong family history of breast cancer made them eligible for clinical trials. A few said that they were at times solicited and encouraged to participate in clinical trials because of their

family history. Some women linked their eligibility for trials and their eligibility for genetic testing; just as they had easily met many of the criteria for different clinical trials, they easily met criteria for genetic testing for inherited breast cancer susceptibility.

All but three of the women interpreted meeting eligibility criteria for genetic testing as confirmation of their high cancer risk and their likelihood of carrying a genetic mutation. About half of the sample explained that agreeing to be genetically tested was a way for them to gain control over what they saw as their “chronic illness.” Approximately a third perceived their high cancer risk as a constant in their lives, concluding that living with high cancer risk was like living with a chronic illness.

Discussion

This study explored the experiences of 20 women who had grown up in families with a strong history of breast cancer. Each participant had received a breast cancer diagnosis. The women’s knowledge of their strong family history of breast cancer appears to have provided them with time to adapt to their own risk and that of others in their family. Analysis of the women’s experiences of living with a family history of cancer revealed contexts for their experience of genetic testing for inherited breast cancer susceptibility. It became apparent in the larger investigation of which the present study was a part that the decision to undergo genetic testing did not take place in a vacuum but occurred in the context of the women’s lives; for example, the women attached meanings to living with a personal and strong family history of breast cancer.

In their experience of expecting a diagnosis of and dealing with breast cancer, all of the women described how they became comfortable with the word cancer. Kenen, Ardern-Jones, and Eeles (2003) theorize that this ease mimics coping strategies among individuals with chronic illness. Kenen et al. describe the expectation of disease onset as “living with a chronic risk perspective.” The participants in the present study experienced two types of chronic risk: the risk of breast cancer diagnosis, and the risk of inherited mutation of breast cancer. Although theories of uncertainty in illness have been developed (Mishel, 1988, 1990), they pertain to chronic illness, not chronic risk. Therefore, much remains to be discovered about the impact of living with chronic disease risk and with an (unconfirmed) inherited risk of a disease.

For the participating women, having a personal and family cancer history was a concrete and constant reminder of their increased risk of the disease. The women were reminded of the risk when other family members received a cancer diagnosis or when they saw photographs of family members who had been diagnosed in the past. Because of their

heightened risk of cancer, the participants in the present study and in the study conducted by Hallowell et al. (2004) commented that they could conceptualize their futures only as the eternal present — that is, plans had to be moved forward because *What if a family member is diagnosed with cancer?* In the context of the study by Hallowell et al. and the present study, the concept of chronic risk for a disease could be said to encompass disruptions in one life, uncertainty about one's future, and uncertain timetable of one's life (Hallowell et al., 2004).

One grave consequence of cancer distress is a tendency to avoid cancer screening. Although this tendency was not the focus of the present study, the women's accounts indicated that they both were offered and accepted *more* cancer screening as opposed to *less*. Hallowell et al. (2004) suggest that accepting one's increased cancer risk serves to reduce anxiety, which in turn can positively affect adherence to cancer screening and interest in genetic testing. Perhaps those participants for whom cancer in the family was the norm and part of their upbringing were adapting to their increased risk of breast cancer and were therefore less likely to exhibit cancer-specific distress. The interviews revealed that cancer was a frequent topic of conversation at family gatherings. Nonetheless, the perception of increased risk among one's family members can lead to uncertainty and distress (Baum et al., 1997; Lerman et al., 1993; Schwartz et al., 2002).

The themes identified in this study show that having a personal and family history of breast cancer is not an isolated situation but part of a woman's journey in choosing to undergo genetic testing for inherited breast cancer susceptibility. For example, when the women spoke about being family guardians, they were acknowledging the breast cancer expertise they had acquired — expertise that enabled them to decode ambiguous information about breast cancer risk. Future research could investigate what specific needs and supports are required for individuals to be effective family guardians. Such support could be virtual or in person.

The main themes derived from the experiences of the 20 participants in living with a personal and strong family history of breast cancer, along with their individual stories, could be used to guide oncology nurses in applying aggregate knowledge to individual cases (Thorne et al., 1997; Thorne, Reimer Kirkham, & O'Flynn-Magee, 2004). As with Clements et al. (2007), this study explored concepts associated with living with a personal and strong family history of breast cancer; it focused on identifying emotional and cognitive experiences of this history, as opposed to quantitatively measuring the intensity of the experience. Further research is needed to compare this experience with the experience of living with

other prevalent cancer diagnoses for which there is probable inherited genetic susceptibility.

Conclusion

The identified themes of living with a personal and family history of breast cancer provided context for the 20 participants who underwent genetic testing for breast cancer susceptibility. The findings show that gathering information on family history of cancer as part of initial genetic counselling can be used not only to guide assessment of cancer risk and risk of a probable inherited mutation, but also to increase our understanding of individuals' reaction to a strong family history of breast cancer. Whether they work in the community, in cancer screening programs, or in cancer genetics programs, health professionals can use the themes presented here as prompts when assessing whether individuals have learned to cope with a family history of cancer.

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