Genetic Testing for Hereditary Breast and Ovarian Cancer: Responsibility and Choice

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Genetic testing for hereditary breast-ovarian cancer has become an important part of clinical genetics practice. Although considerable work has focused on the psychological impact of this technology, there has been little research into the moral implications of genetic information on hereditary cancer families. In this article, the author examines moral issues related to individuals’ decisions to seek or decline testing. In-depth interviews with 53 participants make up the core of the research. Analysis of participants’ accounts illustrates how the decision to be tested (or not) interconnects with moral agency and aspects of self (embodied, familial-relational, and civic self). The findings form the foundation for inquiry into conceptualization of moral responsibility, autonomy, and choice. They also provide insight that might assist clinicians to understand more fully the needs and responses of those who seek genetic testing for hereditary breast-ovarian cancer.

**Keywords:** moral agency; responsibility; choice; autonomy; genetic testing; hereditary cancer

The early detection of women at high risk provides a promising approach for reducing the high incidence and mortality associated with breast and ovarian cancer. . . . The isolation of the BRCA1 genes now allows us to proceed with the identification and understanding.

—Lisa Cannon-Albright and Mark Skolnick (1996, p. 2)

The potentially sick, potentially vulnerable and potentially stigmatized are not an esoteric group of people. They are all of us waiting for our harmful genes to be identified.

—Regina Kenen (1994, p. 53)

The quotations above reveal the complex, and often contradictory, reactions to genetic testing for hereditary breast-ovarian cancer. Hailed by some as a major medical breakthrough, enthusiasts have purported that information gained from

**AUTHOR’S NOTE:** I thank foremost the women and men from hereditary breast-ovarian cancer families who so willingly shared their experiences with me. I also thank Patricia Baird, Susan Sherwin, Carolyn Ellis, Jan Atkinson-Grosjean, and the anonymous reviewers for their helpful comments on different versions of this article and to acknowledge the contributions and invaluable support of the Hereditary Cancer Program, BC Cancer Agency, Vancouver, British Columbia. This research was supported by the Canadian Breast Cancer Foundation, the Huntington Society of Canada, and the Earl and Jennie Lohn Foundation under grants held by Dr. Michael Burgess, Chair of Biomedical Ethics, University of British Columbia, and Dr. Douglas Horsman, Director of the Hereditary Cancer Program, BC Cancer Agency.

QUALITATIVE HEALTH RESEARCH, Vol. 16 No. 1, January 2006 97-118
DOI: 10.1177/1049732305284002
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testing for BRCA1/2 mutations will facilitate early intervention, allowing women to take measures that might ultimately save their lives (Foulkes & Narod, 1998; Muto, 1997; Olopade, 1997; Ponder, 1997). Others have viewed genetic testing as far more threatening, promising to usher in a new era of discrimination and to control people in untoward ways (Finkler, 2000; Lippman, 1998). Still others have described genetic technology as in its infancy and have argued that far more research is needed to determine how genetics interacts with other causal factors in triggering both hereditary and nonhereditary cancers (Burke, Press, & Pinsky, 1999). Yet missing from these discussions are the perspectives of the women (and sometimes men) who seek BRCA1 and BRCA2 testing. How do their experiences accord with these enthusiastic or cautionary tales? Namely, how does the availability of genetic testing and information about genetic risk affect women’s understandings of self and their everyday lives?

The purpose of this article is to contribute to the understanding of the complex moral and social issues generated by genetic testing for hereditary breast-ovarian cancer. Ethicists and others have raised hard questions about privacy, the confidentiality of information, and the threat of genetic discrimination (Henderson, 2001; Knoppers & Godard, 1998; Sommerville & English, 1999; Surbone, 2001). However, it is important to balance these theoretical concerns with people’s subjective experiences with genetic testing. Evaluating the moral and social effects of testing requires that we go beyond abstract discussion to examine the life circumstances in which people make their decisions and live with genetic information. It invites us to explore the specific experiences, values, commitments, and meanings that women and men bring to genetic testing. We also need to examine the local social, political, and life circumstances in which genetic information is made available and used. Little research has been done in these areas.

This article is based on a qualitative study designed to examine the various dimensions of people’s moral experiences with genetic testing for hereditary breast-ovarian cancer. Here, I explore a question central to the research: Why do women and men seek or decline genetic testing for hereditary breast-ovarian cancer? Although there is a large body of literature examining this question from a psychological and clinical perspective (Balmana, Stoffel, Emmons, Garber, & Syngal, 2004; Brain et al., 2000; Capelli et al., 1999; Jacobsen, Valdimarsdottir, Brown, & Offit, 1997; Julian-Reynier, Eisinger, Chabal, et al., 1998; Julian-Reynier, Eisinger, Vennin, et al., 1996; Kelly et al., 2004; Lerman, Daly, Masny, & Balshem, 1994; Lerman, Narod, et al., 1996; Lerman, Seay, Balshem, & Audrain, 1995; Phillips et al., 2000; van Asperen et al., 2002), less is known about the moral aspects of genetic decision making. Drawing from in-depth interviews, my purpose is to describe and analyze the moral territory involved in individuals’ decision making concerning genetic testing. Namely, how do people understand genetic testing with respect to their sense of self and moral agency? In doing so, I hope to broaden ethical discussion by providing insight into how genetic testing affects people’s everyday moral lives.

Before I proceed, however, I wish to clarify the terminology used. Scholars frequently use the terms morality and ethics interchangeably. To be clear, I take as my starting point a characterization of morality offered by Walker (1998). Morality, she wrote, ”tells us something deep and central about how to live” (p. 3). It refers to those values that shape the way we see ourselves and how we ought to live our lives. Embedded in, and fashioned by, particular relationships and social settings,
morality comprises shared understandings that inform our beliefs, motivations, and choices. It is central to how we understand concepts such as trust, kindness, respect, obligation, blame, accountability, and responsibility. Moral agency refers to the notion of individuals making self-determining moral choices and acting on these (Rodney, Brown, & Liaschenko, 2004). Moral agency comprises both what a person believes is the right action or decision and his or her rationale for doing so, and is important when considering experiences related to self-concept (Doane, 2004). Ethics, on the other hand, encompass the reflective, systematic, and normative study of morality. It is concerned “with value questions about human conduct” (Sherwin, 1992, p. 35). Taking my lead from Walker, I describe how choices concerning genetic testing intersect with people’s constructions of their moral selves.

BREAST CANCER AND SUSCEPTIBILITY TESTING

It has long been thought that cancer is genetic, meaning that the transformation of a normal cell to an invasive and malignant growth is due to changes in the DNA. However, researchers have also been aware that certain cancer syndromes such as breast, ovarian, colon, and prostate cancer cluster in some families. Indeed, current estimates indicate that between 5% and 10% of breast and ovarian cancer cases are due to an inherited predisposition to the disease (King, Rowell, & Love, 1993).

The discovery of mutations in two genes related to hereditary breast and ovarian cancer, \textit{BRCA1} and \textit{BRCA2}, offers the opportunity to identify individuals within families who have inherited a strong disposition to develop these diseases (Miki et al., 1994; Wooster et al., 1995). Available data suggest that women who inherit a mutated copy of either gene have a 45% to 65% risk of getting breast cancer by 70 years of age (Antoniou et al., 2003). The same mutations confer an 11% to 39% risk of developing ovarian cancer by age 70. These inherited mutations are also associated with a 59% risk for premenopausal breast cancer and an increased risk for second primaries (new breast and/or ovarian cancers as opposed to recurrent disease) (Easton, Ford, & Bishop, 1995). The incidence of breast cancer in male mutation carriers is extremely low, with mainly \textit{BRCA2} mutations posing an elevated risk (Svedlov et al., 2000). Men who inherit mutations in this gene have approximately a 6% lifetime risk of getting breast cancer (Easton, Steele, et al., 1997). However, there is some evidence to suggest that \textit{BRCA1} and \textit{BRCA2} mutations put men at a slight increased risk for developing other cancers, such as prostate cancer (BCLC, 1999).

Genetic testing for breast cancer susceptibility has been advanced in the hope that it will reduce cancer morbidity and mortality. Knowledge of a positive mutation status might encourage identified carriers to engage in cancer surveillance, avoid exposure to triggering mechanisms, or make decisions about other options (prophylactic surgery, chemoprevention), which can reduce the risk of getting breast-ovarian cancer (Rebbeck et al., 2004; Warner et al., 2004). Accordingly, genetic testing is seen as a technology that might assist individual decision making with regard to health. It is based on a medical and bioethics model that views the individual as rational, independent, and autonomous. Although theorists debate the exact definition of autonomy, respect for this principle is usually interpreted as the competent patient’s right to make choices and direct his or her health care based on personal values and beliefs (Sherwin, 1998). From a clinical standpoint, autonomy
is supported through standardized protocols that emphasize genetic counseling and informed consent (Burgess, Knoppers, & Laberge, 1999). In pretest counseling, individuals discuss with genetic counselors the possible benefits and the potential harms of receiving genetic information and must evaluate whether the risks are worth taking based on their personal notion of welfare. Traditional views of autonomy idealize a concept of self as a free, self-sufficient, and rational chooser (MacKenzie & Stoljar, 2000).

Over the past two decades, however, scholars have challenged the individualistic bias dominating conventional views of autonomy. They have argued that individuals can become themselves only within a network of social relations and that social relations are actually constitutive of self (Baier, 1985; Friedman, 1997; McIntyre, 1984; Nedelsky, 1989; Sherwin, 1998; Taylor, 1989). In their critiques, they have called for a reconceptualization of autonomy as a characteristic of persons “who are emotional, embodied, desiring, creative and feeling, as well as relational, creatures” (MacKenzie & Stoljar, 2000, p. 21). The term relational autonomy has been adopted to portray this concept. Like other theoretical positions, relational autonomy is not a monolithic term but encompasses a range of perspectives; the common thread, however, is the conviction that persons are socially embedded and that an individual’s identity is formed within the “context of social relationships and is shaped by a complex of intersecting social determinants” (MacKenzie & Stoljar, 2000, p. 4). Relational autonomy supports a view of moral personhood that recognizes and gives value to relations of interdependence, solidarity, and community (Friedman, 1997; Meyers, 2000). It purports that human beings develop and enact their autonomy in a social world and that human relationships and community are central to the realization of autonomy (Baier, 1985; Friedman, 1997; Meyers, 2000; Nedelsky, 1989). Thus, the focus of relational approaches is to explore the implications of subjective, intersubjective, and social dimensions of selfhood in relation to moral and political conceptions of autonomy (MacKenzie & Stoljar, 2000).

Reflection on decision making in the context of genetic testing also calls attention to the reciprocity between autonomy and moral agency. In recent philosophical literature, theorists have argued that moral agency is more than isolated individuals engaging in rational actions to deal with ethical problems. Rather, moral agency is better understood as “individuals acting in a web of interconnectedness with others in specific contexts” (Varcoe, Rodney, & McCormick, 2003, p. 959). Walker (1998) further described moral agency as inclusive of identity, relationships, and values, as well as people’s understandings of their and others’ responsibilities. “In making each other accountable to certain people for certain states of affairs, we define the scope and limits of our agency, affirm who in particular we are, show what we care about” (p. 16). Thus, the responsibilities we assume intersect with who we think we are or who we want to be. Responsibility toward others is critical in shaping interactions and shaping lifeworlds. Indeed, responsibility toward others and choice are grounded in self-identity (Walker, 1998). As Nelson and Nelson (1995) put it,
Although clearly shaped by familial, social, and historical conditions and thus not always under voluntary control, the choices we make and the responsibilities we assume can be seen as a reflection of what matters to us and who we are in moral terms.

Another critical aspect of morality is the intersection between embodied engagement of self and moral agency. In my usage of embodied self, I focus on those aspects described by Abel and Browner (1998) to include constructions of self based on a woman’s experiences of her body and its physical nature. The embodied self takes into account how embodiment affects how we see ourselves, how we live, and the decisions we make. Bodily experience is of central importance in making one empirically aware of one’s personal self and is a source of moral sensitivity and knowledge (Doane, 2004). I concur with Church (1997) that selves are not separable from or identical to bodies, but “are created and sustained by the establishment of particular sorts of interconnections” (p. 8). The body does not make the self, but the body (well, ill, able, disabled, etc.) influences understandings of self. Thus, illness can set limits on or reveal new possibilities for the self through the body, as shown by the classic writings on illness and identity (Bury, 1982, 1991; Charmaz, 1995). Similarly, the “bodily” nature of genetic risk might influence the way in which women think about their bodies and, consequently, their sense of present and future self. Likewise, it might generate new kinds of moral understanding. Strong perceptions, sensations, and physical responses can inform ethical practice by providing insight into the moral significance of a particular event (Doane, 2004; Raingruber & Kent, 2003). To recognize this is not to dismiss the importance of rationality but to understand embodiment as a source of knowledge and authenticity (Howson, 1998).

Bringing these different aspects to bear, a relational and embodied approach to autonomy and moral agency suggests that we need to rethink traditional discussion regarding genetic testing and its focus on the autonomous chooser. Clearly, a decision to have genetic testing is not made in isolation but is shaped by other life experiences, circumstances, responsibilities, and commitments. The latter, in turn, will affect how genetic information is interpreted and acted on. Thus, a broader ethical analysis requires that we examine specific decisions within the context that influences, and sometimes constrains, those decisions. We ought to be concerned about the effects of genetic testing on understanding of self and self in relation to others. In particular, do choices concerning genetic testing influence self-concept and relationships with others? Do they facilitate enactment of moral agency?

Although few researchers have examined directly the moral concerns experienced by people seeking genetic testing, some qualitative studies point to the complexity of this issue. Research by Hallowell and colleagues (Hallowell, 1999; Hallowell, Foster, Eeles, et al., 2003) and Foster, Watson, Moynihan, Jones, and Eeles (2002) indicates that responsibility to others serves as a primary motivation for women to undergo BRCA1/2 testing. These investigators cautioned that the obligations to obtain this information for family might, in fact, pose a constraint on autonomous choice related to genetic testing. Goelen, Rigo, Bonduelle, and de Greve (1999) and Julian-Reynier, Eisinger, Chabal, et al. (1998) also found that many patients have themselves tested for the benefit of family. Likewise, several studies have shown that men who have a family history of breast-ovarian cancer frequently undergo testing for the sake of the children (Daly et al., 2003; Goelen et al., 1999; Julian-Reynier, Eisinger, Chabal, et al., 1998; Liede et al., 2000; Lodder et al., 2001).
General altruism has also been reported as a reason for testing (Bernhardt et al., 1997; Geller, Strauss, Bernhardt, & Holtzman, 1997). Most previous research, however, has focused on genetic decision making without exploration of the deeply personal, relational, and embodied process of moral agency and self. I aim in the present study to contribute to understanding in this area.

STUDY METHODS

The work reported here was part of a larger qualitative study, directed by ethnographic methodology (d’Agincourt-Canning, 2003). In contrast to research methods that focus on people’s behavior without regard for what such behavior means to those engaged in it, ethnography is committed to trying to understand the meanings people make of their experiences in everyday life (Emerson, Fretz, & Shaw, 1995; Hammersley & Atkinson, 1995). Epistemologically, it is based on the position that as people interact, they create their social realities and derive meanings about events in their lives (Blumer, 1969; Mead, 1934). Consistent with this approach, this study started from the perspectives of women and men who were eligible for and/or who had undergone genetic testing for hereditary cancer. It was designed to explore the reasons participants had for seeking or declining hereditary cancer genetic counseling and/or testing and the meaning they gave that experience.

Data were derived from individual interviews and field observations collected from 1998 to 2001, after evaluation and approval by the University of British Columbia research ethics board. In keeping with formal ethical approval, I obtained informed consent prior to all interviews. Participants were initially recruited through a hereditary cancer program to which they had been referred for genetic counseling for breast-ovarian cancer because of their personal and/or family histories of the disease. These participants then aided further recruitment by contacting other family members who were eligible for, or considering, genetic testing. Beeson and Doksum (2001) have called this form of sampling cascade sampling. It begins with the index person or carrier, who refers the researcher to other biological relatives or significant others. I approached 59 people, and 6 either declined to participate or did not respond to a letter or phone call of invitation. Those who declined gave no reason for their decision. The final sample comprised 53 persons (45 women and 8 men).

Study Participants

Participants ranged in age from their early 20s to over 60. The majority was married and had children. Educational background varied: 38% had completed high school, and 49% had obtained further vocational training or university.

Thirty-nine (34 women and 5 men) of the 53 participants chose to undergo testing. Of these, 14 women had already been affected by breast-ovarian cancer; the remainder (20 women and 5 men) were cancer free but considered at high risk for carrier status based on family history. Four participants from the same family (1 with breast cancer, 3 without) had sought testing but were waiting for the index test results before proceeding further. Six participants refused testing, even though they
met eligibility criteria. Of these, 2 women previously had breast cancer, and 4 were cancer free. In addition, I interviewed 4 individuals (2 women, 2 men) who were spouses of participants who underwent testing.

Data Collection and Analysis
In-depth interviews made up the core of this research. Participants lived in various locales throughout western Canada, including rural and nonurban regions. With two exceptions (done by phone), the interviews were conducted in the participants’ homes or another place of their choosing. The interviews were tape-recorded with prior consent of all participants and transcribed verbatim.

Analysis involved an interpretive process and was guided by constant comparative techniques (Glaser & Strauss, 1967). Initially, I grouped interview segments into preliminary categories based on interview questions (e.g., family history, experiences with cancer, reasons for seeking testing, personal understanding of risk). As more data were collected and coded with descriptive phrases or words, these categories were revised. Comparison of differences and similarities within and between categories and subcategories (as well as field notes and reflective inquiry) enabled further refinement, clarification of meanings, and the development of conceptual themes (Strauss & Corbin, 1990). I managed the data using NUD*IST qualitative software.

FINDINGS
Inductive analysis revealed three thematic categories in relation to choice and self-understandings: the embodied self, the familial-relational self, and the civic self. These categories referred to the manner in which participants viewed genetic testing when thinking about their physical selves (embodied self), their families’ health and well-being (familial-relational self), and their general relationship to unknown others (civic self). Narrative accounts about specific situations illustrating these themes are presented for both those who accepted and those who declined testing.

Accepted Testing
The Embodied Self
For most study participants, breast-ovarian cancer posed a real physical threat to their health, their bodies, and their sense of future self. Based on lived experiences of seeing it in family members, many equated breast or ovarian cancer with death. The notion that death from breast cancer is inevitable might reflect, in part, the historical reality of the disease before more successful treatments became available. With new and better treatments, we might begin to see intergenerational differences in the way or how soon a family member affected with cancer dies (Rosenbaum & Roos, 2000). However, it also reflects the concrete reality that most participants had faced. In recent years, medical and activist discourse has shifted to emphasize that
breast cancer is not fatal; the message that if breast cancer is detected early enough, women can survive the disease is commonly heard (Lerner, 2000).

For many participants, this message just did not hold true. Lorraine (all names are pseudonyms), who is a public health nurse and was diagnosed with breast cancer at age 47, explained it this way:

The survival rate [from breast cancer] is really good and they [physicians] are quite confident if they find it early people survive. It’s just in our family that was never our experience. Everybody who got it died quite quickly actually... Aunty M. and Aunty E. it seemed like within a year or two after diagnosis they had both died. So my experience, my family experience isn’t that you do well and you survive.

It is important to add that Lorraine did not reject the medical paradigm. Rather, she sought medical treatment that she considered would best meet her needs. She was very aware of her family’s cancer legacy (both breast and ovarian cancer) and the risk it imposed on her. She used experiential knowledge as a basis for deciding to pursue prophylactic surgery even as biomedical information shaped that knowledge.

I just knew that I had to do this. I don’t know why I knew, but I knew I had to do it and I had to get going on it and not keep waiting and waiting.

It was during the “prophylactic surgery” (a bilateral mastectomy) that a 1.8 cm tumor and cancerous cells were found throughout Lorraine’s right breast. The cancer had been previously missed on both clinical exam and mammography. Lorraine later underwent genetic testing when it became available. She said it confirmed what she already knew: “I mean I knew, being a nurse, there’s something wrong with the family. Like why is everybody dying of cancer?” She had a BRCA1 mutation.

All of the 17 women diagnosed with breast-ovarian cancer viewed cancer as a persistent threat. As other studies have shown, uncertainty about the future and fears of recurrence were ever present and motivated them to seek testing. They sought genetic information about hereditary risk, in part to guide further medical management. They perceived genetic testing as providing information that would enable them to protect themselves. Haunted by images of the past, these women said they would do anything they could to prevent cancer from recurring. With the exception of one woman, who was in the terminal phase of her disease, information about BRCA1/2 mutation status was instrumental in helping affected participants decide whether to pursue prophylactic surgery. Sandra, who was diagnosed with breast cancer at age 37, put it this way:

You know having gone through that [breast cancer] it was easy to make the decision that I didn’t want to do it again and take that chance. And I think seeing that my mom had it twice and that my aunt had it twice, only reassured me that I was making the right decision to get tested and then making the decision to have the surgery.

Most affected women viewed information gained from genetic testing as allowing them to make choices about medical management that might improve their survival. In that sense, genetic testing was viewed as empowering. Martha wanted to be tested and learn her genetic risk status, because, as she put it, “knowing gives
you more control.” The ability to know her genetic risk status offered her some control over an uncertain disease. Marlee, who was diagnosed with breast cancer at age 33, also talked about genetic testing in terms of knowledge, awareness, and prevention.

My mom’s doctor spoke to her quite a bit about it [genetic testing] after finding out about me. So yeah, it just went from there, but no it was/ it interested me right away. It interested my mom right away. . . . To me the more I can know the more I can help myself . . . right?

Some of those who did not have breast-ovarian cancer but were at high risk based on family history expressed similar sentiments. Brenda’s decision to be tested was also influenced by her family history, which included several aunts and cousins as well as her mother who had died from breast cancer. At the time of our interview, her older sister (then age 48) was struggling with ovarian cancer after having had breast cancer when she was 30. A BRCA1 mutation had already been found in several members of her family, including Brenda’s sister with cancer. Several times during our interview, Brenda talked about genetic testing as providing her with specific information about her body and the opportunity for choice. In her words,

At least we know. This wasn’t something that we could find out and do at all before. And I think the more people that find out that they can do this, um, the more informed decisions that they can make, you know? We need to know as much as we can about our bodies.

Brenda’s comments fit into a larger discourse shared by many participants: that knowledge is good, and knowing cannot be worse than not knowing. Not only is genetic information relevant to informed decision making, it allows one to understand, cope with, and possibly take control of what is happening. As Martha stated,

I needed whatever information I was going to receive, and I was . . . grateful is the wrong word, but it’s, I think that the value of having this testing for women cannot be underestimated.

Decision making is typically understood as a process that involves reflection about the implications and outcomes of a specific action or event, yet a few participants spoke of being just “too curious” not to be tested. In each of these instances, a mutation had already been found in their family. Their decision to seek testing did not reflect contemplative choice as it is typically constructed—that is, the weighing of alternatives and balancing of perspectives—but was more of a gut reaction. They said as soon as they learned about the familial mutation, they just had to know whether they carried it or not. They gave little consideration to what the implications of this information might be. As Marilyn, age 28, put it,

As soon as I found out about the gene, all I wanted to do was get tested. Because I just had to know. I don’t know [whether it’s] because I am stubborn and nosy, but I had to know.

Carolyn also spoke about testing in this way. Like most participants, she had experienced the immediacy of cancer. Indeed, one of her earliest memories was
seeing her aunt’s scarred chest after a mastectomy. Strongly aware of her own risk, Carolyn (now age 21) had been doing breast self-exams since the age of 15.

Once I understood everything that was going on and that there was this gene and everything, I wanted to know right away. So, not so much that it changed my life or anything, but just so I know what’s going on with my body.

The Familial-Relational Self

Although genetic information provided the foundation for gauging individual risk and guiding medical decisions, responsibility toward others proved equally important in participants’ decision making. In fact, very few participants considered undergoing genetic testing just for themselves. Because genetic risk is shared by all members of a biological descent, genetic information was generally perceived as a benefit to all kin. Participants often wanted testing so that they could provide children or other family members with information relevant to their risk status. Mothers who had been afflicted with breast or ovarian cancer, in particular, wanted to spare their children a similar fate. For example, the words of this participant illustrate how she viewed genetic knowledge as personally valuable, but getting information for other family members provided an equally strong impetus to undergo testing.

I wanted to get tested more for my kids. And for Alice, she’s the youngest [sister in the family]. She’s like my best friend, Alice and I. So yes, I kind of wanted to find out not more so for myself, but just to see if they would possibly have the gene or that I have passed it onto my children.

Martha’s comments also illustrate the link between testing and responsibility to other family members.

I felt that it [genetic testing] would also be important for me, but this was a secondary thing . . . the primary thing I was thinking about is risk for my family.

The few women I met who were in the terminal phases of their illness viewed their participation in genetic testing as almost a duty. Obviously, at this stage, information about genetic status provided no clinical utility for them, but these women framed their discussions of genetic testing in terms of its greater value to their daughters, granddaughters, and future family. They thought that genetic information would empower their daughters by placing them in a better position to screen and manage their health. Ellen was quite emphatic about this point. When I talked to Ellen at her home, she had been fighting breast cancer for 2 years and had been recently assigned to palliative care.

I was, as a mother, thinking it was important for me to know, important for my children to know and understand and to have preparedness for it.

For Barbara, getting genetic information for her children was the only reason for obtaining her test results. Because of her strong family history of cancer, Barbara perceived the disease not as a neutral probability but as a salient and ever-present threat in her life. After her younger sister died from breast cancer, Barbara underwent prophylactic mastectomy. Later, a BRCA1 mutation was identified in other family
members. She then decided to learn if she carried the mutation for the sake of her children.

I have children, so, you know, to me it would just naturally follow that you would do that [have testing], so you would be armed with the knowledge for your children. And for me, knowing that I don’t have the gene is wonderful, ’cause now I don’t have to worry about my children…. If I had tested positive, then I would have had my ovaries removed as well. I mean that would be the, you know, the carry on with that. But for me I needed to know simply because of my children. I mean, now they don’t need to be tested.

With no history of male breast cancer, all 5 men who underwent testing did so out of responsibility to their daughters and granddaughters. David’s response is typical in this regard:

I had my daughter and my granddaughters that could be affected if I have the gene. It doesn’t necessarily mean that they would also have the gene, but then they could make some intelligent choices on them/ on their own if I were to be tested. And if I had the gene, I could tell them, say, okay, you know, it’s up to you. You can just make your own decisions now. I have the gene and that there’s a possibility that I may have passed it on to you, now you can at least make some, some choices. And it was on that basis that I was tested.

Bill, whose mother and other relatives died from breast or ovarian cancer, said that he had “no choice” but to participate in testing. Because he was a man, genetic testing for the BRCA1 mutation had little impact on his health management. His decision to undergo testing stemmed from a sense of duty to provide his daughters with this information. He felt that they should know whether they could inherit the gene mutation from him.

Having daughters, I didn’t even hesitate a bit to do it. I figured for me to find out, it had to be so.…. When [my cousin] phoned and said about this study, I didn’t have to think about it.…. I said I’ve got no choice having daughters. That’s the way I looked at it.

Feelings of genetic responsibility can play out in another way, however. Some participants felt so responsible for carrying a genetic mutation that they pressured other family members to get tested. Likewise, a few participants felt coerced to undergo testing by well-meaning relatives who thought this information was in the best interests of the participants’ health. Not interested in the information herself, for example, Kelly said that she had little choice but to learn about genetic testing.

Contrary to the notion that genetic testing provides choice, Kelly felt just the opposite. Her mother and aunts had obtained certain medical information that affected her life. This posed new problems for her. She felt forced to live with information that she would rather not have and from which she could not escape.
I think it was no big deal to them [mother and aunts], but they didn’t think about what it was going to do to their kids and their grandkids. Because this is a never-ending thing now. Like we opened a box that’s never going to close, like it’s an open door to forever. Like I said, once you open that door you can’t ignore what’s behind it.

Kelly’s comments are instructive, in that they remind us that facts (e.g., information that a genetic mutation exists in one’s family) are not received neutrally. People interpret them differently according to their understandings, life context, and experiences. Moreover, genetic testing differs from other medical tests, in that one person’s decision to seek genetic testing will affect the choice of and medical facts about others. As Kelly states, information about a familial BRCA mutation has implications not just for her mother and herself but also for future generations. Whether Kelly decides to have genetic testing or not, her choice is affected by the knowledge that she has a 50% chance of carrying a mutation associated with breast and ovarian cancer.

People’s decisions to undergo testing can affect family members’ choice to know in other ways as well. For example, Veronica (who did not have cancer) asked her mother to undergo genetic testing because of their extensive family history of the disease. Multiple members had died from breast and/or ovarian cancer. Yet Veronica’s mother, who also had been affected by breast cancer twice, refused to be tested. Veronica was disappointed in her mother’s decision but proceeded to be tested herself anyway. She could be tested without her mother’s participation, because a specific mutation had already been identified in other close relatives. In doing so, however, Veronica obtained information not just about her genetic status but about her mother’s as well. Testing revealed that she carried the same mutation as her mother’s sisters, meaning that her mother must have had it and passed it on to her.

She’s dead now so, and she was only...6 7 when she died [from breast cancer]. So she’s, she wasn’t that old but she had chosen not to have the genetic testing done. I had encouraged her to do that but she wouldn’t. And um, even when she found out that I’d had it done and I did have the BRCA 1 gene, she still would /she was still in denial and felt that it didn’t matter, didn’t mean that she had it.

This example shows how genetic testing can lead to the disclosure of one’s relatives’ risk status by another. It might also lead to coercion and an impingement on choice for some. Clearly, as Veronica’s situation shows, people might be told information about their genetic risk that they do not want to know. We need to guard against coercion, but at the same time, it is important to recognize there might be situations in which people are blocked from obtaining information that they might view as beneficial to them. Indeed, some participants felt that certain family members had abdicated their responsibilities by declining the test. Leslie described her brother’s refusal to undergo testing as cruel. She and two of her sisters had been tested, and all three were found to be BRCA1 mutation carriers. Despite the family’s extensive history of breast-ovarian cancer and now information about a genetic defect, he refused to be tested, even though he had an adult daughter and, more recently, a “brand-new” granddaughter. In Leslie’s words,

I think it’s very irresponsible. I mean if he doesn’t have it, he doesn’t have to worry about worrying his kids about it. If he does, she’d [his adult daughter] better get
tested pretty soon. It’s ridiculous. I think it’s very irresponsible, if you have something like that and you can, you know, make sure. ‘Cause I mean you’re giving your kid no option to have themselves checked, have themselves have any preventative stuff if they have to, or testing that they should have. It’s horrible. I think it’s very cruel.

The Civic Self

Also embedded in participants’ narratives was a strong sense of altruism. Several participants expressed a desire to be tested, not just for themselves and their families but for all women. They perceived a duty to do anything they could to advance medical science. Women who were affected with breast cancer, as well as those who had a family history but were disease free, expressed this concern. Although an extension of the relational self, I use the term civic self to describe this response. It allows me to make a distinction between actions taken with regard to family (known others) and actions take out of concern for society at large (unknown others).

For example, Claire perceived little immediate benefit from genetic testing for herself. She agreed to participate in testing only because she thought it might prove beneficial to breast cancer research. Genetic testing for hereditary cancer began under a research protocol in Canada. Participants were aware that their test results were contributing to research as well.

The advantage is just information to the people doing cancer research. That is the only reason I said yes [to the testing]. The larger your sample size, the better your results. … If our family is showing a lot of this, there is a good chance that we would have these genes that could help somebody’s research project and provide answers down the line for some other people, maybe even for us.

Ross also talked about the desire to contribute to research for the benefit of others. A family member had asked him to participate in testing because of their shared history of the disease. She had been found to be a BRCA1 mutation carrier. Initially, he was reluctant to participate. His sister-in-law had recently died of breast cancer, and he just wanted to distance himself from anything to do with cancer. Despite his struggles with these memories, however, he felt an obligation to participate. He did not see the test as much use to himself or his immediate family but hoped that his participation would benefit others. In his words,

I guess [I thought] if it could help, if it could help with a cure, or help their further their knowledge, that was a the reason to go. … In my mind, I felt that the more they found out, the more they know, the more they are going to get to the bottom of it.

Declined Testing

The Embodied Self

A small group (6 women) declined testing: Of the 6 who declined, 2 had experienced breast cancer, and the other 4 were unaffected by cancer but eligible for testing based on the existence of a known family mutation. The primary reason women gave for declining testing is that it would not alter their screening behavior or medical management. All 6 study participants recognized that their family and/or
personal history put them at increased risk for cancer but felt that their current measures of health care surveillance were adequate. Like those who underwent testing, they saw themselves as highly susceptible to the disease, yet this group perceived little or no benefit to having genetic information. They said that the information would make no impact on their embodied selves. Sheila put it this way:

We are very aware of it. It’s not like it’s taken us by surprise. I mean my mom has lived with it for over thirteen years and we knew about my aunt [died from ovarian cancer]. So for the last twenty years we’ve been very aware of it. And we, I think we’ve taken the appropriate steps to not/ I mean yes, to protect ourselves and also to know early on if there’s anything. . . . So this wouldn’t change anything for me if I was confirmed that I had this breast cancer gene or this defected gene. It wouldn’t change anything for me. It wouldn’t change my lifestyle. It wouldn’t change what I am doing. It doesn’t change my predisposition to having the disease.

Another participant expressed a similar view:

The genetic testing, I would sort of be willing to do it if they have something that could alter the genes or kill it or, I don’t know, do something. But they don’t know. They cannot at this point as far as I know/ there is no way that they could do any-thing. It’s just finding out that’s it there.

These women’s accounts indicate that they did not reject a genetic mutation as the underlying etiology of breast-ovarian cancer. Rather, they did not seek testing on the basis that it could not provide any practical benefit for them. Unlike those who sought testing, decliners did not think genetic testing offered them any further choice. They did not reject genetic testing outright but evaluated it in terms of the benefit it afforded them. Similar to those who were tested, they approached genetic testing pragmatically.

In addition to being of little practical use, all 6 participants declined genetic testing on the basis that it would cause them increased anxiety. Instead of providing information that would enable them to take action, they saw genetic testing as potentially distressing. Rather than adhering to the notion that information is power or that it is better to know than to not know, those who declined perceived genetic information as something that would affect their lives adversely. Learning their carrier status did not offer the promise of control but posed a potential risk. Women with breast cancer, as well as those who were unaffected by the disease, expressed concern about the impact a positive test result would have on their emotional well-being. The following comments help illustrate this point.

Counseling kind of established that there isn’t really anything you can do about it . . . right? That it’s important to maintain that the level of you know the mammogram and all that stuff . . . right? . . . It didn’t seem to be information that was going to, that you were going to do anything about to make it any better, so why find out about it to make you feel worse? (Participant affected by breast cancer)

I am sure there are millions of people walking around with the gene who don’t know it or who do know it and it doesn’t affect their life. Like you think it might be something, gene testing might be something that might swallow a person’s thoughts up, you know, because they know they have the gene for cancer and then they just sit idling waiting to receive cancer and then cancer becomes this consuming thing in your life. I don’t really want that, you know. (Unaffected participant)
The Familial-Relational Self

Like those who had accepted testing, those who declined discussed choices concerning testing not solely as individuated or autonomous agents but also in connection to others. For the most part, this group of participants saw genetic information as having the potential to cause excessive worry and emotional distress. Furthermore, they perceived information gained from genetic testing as posing this risk not just to themselves but to other family members as well. Thus, women who declined testing, like those who accepted, sought to act in the best interests of themselves and their families in making their decision. Leanna described her concern for her sisters:

If you found out that you have it, then your anxiety level, you know, would just / especially if you haven’t gone through it [cancer]. I mean for my sisters, right? For me, you know, well I’ve been through it, but for my sisters who haven’t been through it, the anxiety level would just, I mean it would just / you would now be almost like a person who’s been diagnosed with cancer because you’re diagnosed with a breast cancer gene. . . . I mean, I think / yeah if you have found out that you have the breast cancer gene it would really be negative. It would be hard to distinguish the breast cancer gene from breast cancer.

Reda, who did not have cancer, expressed a similar viewpoint:

You have this information that I don’t know if you, if you / if people should have. If they know how to monitor it, you know? I think that, you know, a couple of members in my family if they found out that they had the gene. I think it would just, like I am really worried about my sister, you know, because I think that if she found out that she had the gene she’d panic.

Embedded in these comments is the ideology of genetic determinism: that those who have the mutation are destined to get cancer, even though this result indicates only a probability, not a certainty, of developing cancer (Lippman, 1998). Genetic counselors go to considerable lengths to clarify this misunderstanding, yet family history and experiential knowledge play a vital role in how biomedical knowledge is understood. Some participants viewed themselves as predestined to get breast cancer, even though they chose not to be tested. Genetic determinism is not unique to this group but reflects a larger social discourse that links genetic risk to inevitability (Finkler, 2000; Kenen, 1994; Lippman, 1998). Indeed, many participants who sought testing expressed the view that a genetic mutation would eventually lead to cancer, yet knowing whether they carried a mutation and taking action gave those tested hope, whereas “not knowing” gave those who declined testing hope. They reached different decisions, but the two groups of participants were alike, in that they saw their decisions connected to the well-being of others as well as themselves.

The Civic Self

Although the small number of participants makes it impossible to draw conclusions, only 1 woman who rejected testing configured her response in relation to responsibilities engendered by a civic self. Most participants framed their decision to decline testing in terms of the neutral or negative impact it would have on them
and their families. These women did not express the desire to aid medical science as a whole. Their response should not be too surprising, given that those who declined testing looked at the technology as having the potential to cause more harm than good.

DISCUSSION

To date, bioethics has relied heavily on the language of principles and rights, as well as social good, in assessing the morality of actions or policies. Although concerned with protection of the patient, bioethicists tend to approach problems from the physician’s vantage point (Hoffmaster, 2001). Indeed, ethics work in genetics has emphasized clinicians’ responsibilities for the effects of tests or information and patient autonomy for decisions made within genetic counseling and regarding health care. This approach has been criticized for its individualistic orientation toward testing and its emphasis on clinical service approaches (Burgess, 2001). It provides little basis for understanding the moral experiences of individuals and how genetic information is assimilated into their lives. It also fails to recognize patients’ concerns about family relations in their analysis of moral issues (Burgess & d’Agincourt-Canning, 2001).

In this study, I set out to explore participants’ reasons for seeking or declining genetic testing. Participants’ accounts indicate that this choice is not just an objective activity based on an individual interest alone but is a process that emerges from and is constructed in relationship with others. Choices concerning testing involve decision making about personal health but also intersect with understandings of responsibility to children, extended family, and unknown others. An individual’s decision to be tested (or not) also affects the choice of others. What distinguishes hereditary diseases from other disorders are their implications for family members (Lerman, 1997). Indeed, within genetics, people might see their selves inscribed onto the lives of others. The reverse is also true.

The findings presented demonstrate that decision making about genetic testing involves an integration of different elements of self. These aspects of self refer to the manner in which participants view genetic testing when thinking about their own physical health (embodied self), their family’s health and well-being (familial-relational self), and the welfare of society in general (civic self). Although separated for analytic purposes, clearly these aspects of self are not distinct entities but tightly integrated. Furthermore, they are not static. Expressions of self and, accordingly, the choices people make might shift and change as circumstances within the family, within relationships with others, or within one’s life change.

Generally, most participants talked about genetic testing as providing choice. Some people valued genetic information both for the sake of knowing and for the control over their lives that this knowledge implied. Many hoped to put uncertainty to rest, wishing to lessen their worry or to avoid surveillance programs if they were to test negative. They saw genetic testing as enabling regardless of who initiated it. Central to these accounts were women who perceived cancer as a tangible threat to their embodied selves. They accepted the privileged discourse of science, because it responded to the threats they felt from the possibility of cancer. That does not mean they did not feel constrained by the options available but that genetic testing held out the best hope for improved survival. In other words, they approached genetic
testing pragmatically. This response is not unique to genetic testing. Focusing on the complexity of women’s relationship to medical technology, Lock and Kaufert (1998) have written, “For by force of the circumstances of their lives, women have always had to learn how they may best use what is available to them. If the apparent benefits outweigh the costs to themselves, and if the technology serves their own end, then most women will avail themselves of what is offered” (p. 3, emphasis in original).

Similarly, most participants did not acquiesce to genetic technology unwittingly; rather, they were trying to resist this disease in the best way they thought possible. Only a few women embarked on testing without giving it much thought. For a few participants, however, genetic testing by others denied them the opportunity to choose. These participants stressed the familial implications of genetics and felt that this information (and, accordingly, its burdens and benefits) was thrust on them without their consent. Still other women who came from high-risk families decided not to seek testing on the basis that it would not change their medical management. They did not challenge genetic information, for the most part, but felt constrained by the narrow range of medical options available to them. They also feared the results of testing would be emotionally disturbing. These findings are in accord with other investigators’ results (Geer, Ropka, Cohn, Jones, & Miesfeldt, 2001).

Although participants’ choices related to testing reflected practical concerns about the embodied self, the self in relation to others was also instrumental in these decisions. Participants considered genetic testing in relation to the welfare of the family and their responsibilities to specific relatives. Some women believed that testing could give their daughters, sisters, and other female relatives information that would be useful for them. Referring to their own experiences with breast cancer, some hoped that genetic information would give all family members greater control over the disease. Still others acquired genetic information with the hope it would allow them to fulfill their role-based (parenting) responsibilities. Many also expressed a desire to aid other women and society in general, which is consistent with other researchers’ observations (Geller et al., 1997; Hallowell, 1999; Tessaro, Borstelmann, Regan, Rimer, & Winer, 1997).

Similarly, study participants who declined testing voiced concern about the impact that this information would have on family members. They also felt responsible for their families but thought that a positive result would do more harm than good. They perceived the family as a unit and felt a responsibility to that unit. This suggests that we should be careful about dichotomizing decision making about genetic testing into accepters and decliners. Although the outcomes differed, participants drew on similar aspects of the moral self and perceived responsibilities to others in making their decisions.

Genetic testing is enacted within discursive practices (medical, ethical, and legal) that give primacy to individualistic models of autonomy, rational decision making, and choice. Similar to Hallowell et al.’s (2003) work, results from this study suggest that a relational concept of self might provide a better framework for understanding the complexities raised by genetic testing for hereditary breast-ovarian cancer. Participants did not view their decision to seek testing in isolation from everyone else. Rather, obtaining genetic information allowed them to express their identity as embodied selves as well as selves-in-relation (Held, 1993), that is, as mothers, daughters, sisters, fathers, and citizens. Choice did not appear to be autonomous in an individualistic sense but instead existed along a continuum of self-care...
and concern for others. The decision to pursue genetic testing reflected a choice arrived at in relation to the context of family and community. Moreover, this choice had implications not just for the individual but also for past, present, and future generations.

In the context of genetic testing, emphasis has been placed on the role of individual self-determination and choice. In fact, some authors (Foster et al., 2002; Hallowell, 1999; Hallowell et al., 2003) have expressed concern that consideration of others in making decisions about genetic testing serves as a “constraint” on personal autonomy and choice. Hallowell et al. (2003) wrote,

The familial nature of genetic information compromises the possibility of making an autonomous decision about genetic testing on two counts. First, an individual’s DNA test results have direct implications for biologically related kin and second, the persons who undergo testing have social obligations to these kin. (p. 78)

Although I do not deny that family nature of genetic information might influence decision making concerning genetic testing, it does not follow that individuals’ choices are not autonomous. Findings from this study clearly indicate that choice is not just an objective activity based on an individual interest alone but is a process that emerges and is defined in relation to others. Choice reflected the participants’ values and goals not as isolated individuals but as moral agents. This was evident in both women who chose to pursue testing and those who declined. To view all obligations or responsibilities as constraints on a person’s autonomy is to neglect the social and moral fabric of people’s lives. What people seek from testing will be moderated by self-interest as well as social and family relationships, including responsibilities toward others.

It is also important to recognize, however, that there can be a dark side to responsibility. Although some women might embark on testing for the benefit of their family, others might experience their efforts as oppressive and denying them choice. Not everyone perceives the ability to choose as a good thing, especially if there is no cure or definite means of prevention (Kenen, 1994). Furthermore, it is important to recognize that expectations and judgments of responsibility are not just individually determined but evolve out of particular social practices that characterize social life (Walker, 1998). Responsibilities are passed down to us by a society that sees responsibility in certain ways. Thus, one can question whether a decision or action taken out of responsibility for others is truly autonomous if the factors that shape responsibility are overlaid by oppressive norms.

CONCLUSIONS

The accounts presented in these findings make apparent some of the “everyday” moral issues arising from genetic testing for hereditary breast-ovarian cancer. They aid understanding of how the choice to seek genetic testing (or not) intersects with aspects of self, moral agency, and relational responsibilities to other. Viewing choice as embedded in particular social locations and relationships shifts ethical discussion from the abstract to people’s everyday experiences. It acknowledges the need to explore the potential benefits and harms of genetic testing from the vantage point of those living with hereditary disease.
These findings also offer insight that might help clinicians understand more fully the needs and responses of those who seek genetic testing for hereditary breast-ovarian cancer. In the context of genetic counseling and patient education, knowledge of the moral concerns of individuals might aid clinicians in identifying information that is relevant to the individual and should be dealt with more thoroughly. For example, knowing that many women seek testing not just for themselves but also for their children suggests that clinicians should pay special attention to whether testing and genetic information can, in fact, help the person fulfill his or her relational responsibilities to family (Burgess & d’Agincourt-Canning, 2001). Also relevant to genetic counseling and patient education is the fact that feelings of responsibility might lead to coercion of others. This implies that providers should also be prepared to counsel clients about the negative effects of pressuring family members to be tested.

Finally, this research draw attention to the broader familial, social, and political context in which genetic testing occurs and, by doing so, suggest avenues for future research. The influence of social factors (e.g., age, ethnicity, socioeconomic status, and/or location) on agency and choice with respect genetic testing is an area that warrants future investigation.

NOTE

1. Although men can carry BRCA1 or BRCA2 mutations, at the present time, most of the people undergoing genetic testing for hereditary breast-ovarian cancer are women.

REFERENCES


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