Genetic Testing without Genetic Counselors: Exploring the BRCA Testing Experiences of Patients with Breast Cancer

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ABSTRACT

Genetic Testing without Genetic Counselors:
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A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Margaret Hayes

Increasing numbers of patients are having genetic testing ordered by a healthcare provider without following the traditional model of seeing a genetic counselor for pre-test counseling. It is important, however, to ensure that patients receive complete and accurate information about genetic testing and its implications on their medical management and the medical management of their relatives.

The purpose of this qualitative study was to explore the experiences of breast cancer patients who had BRCA testing ordered without meeting with a genetic counselor. We interviewed four women who fit these criteria using a semi-structured interview guide to elicit information about the process they went through. We coded interviews based on questions asked of the participants and emergent themes. We analyzed the data looking for patterns and variations within these themes.

Three main themes emerged: (1) the value genetic testing holds for the participants in informing surgical options, assessing risk to family members, determining their future risk of cancer, and curiosity; (2) the emotions patients experience about the genetic testing
process in general and throughout the course of waiting for results, as well as receiving them; (3) patient interactions with providers and how they felt about what they should say and know about genetic testing.

Through this research we found that some providers are ordering genetic testing for patients without following the traditional model of a genetic counselor providing pre-test and post-test counseling. This study suggests that BRCA testing ordered by non-genetic counselor healthcare providers is not detrimental to patients. Recommendations for future research entail a new model where providers order genetic testing at the time of medical necessity and then a genetic counselor delivers follow-up counseling. This process would facilitate patients being adequately informed about genetic testing, while also unifying the approach and increasing the education of providers.

Keywords: breast cancer, BRCA1, BRCA2, genetic testing, genetic counseling, oophorectomy, mastectomy, FORCE
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INTRODUCTION

Increasing numbers of people are curious about what information their genes may hold, especially after a cancer diagnosis. According to the American Cancer Society (2013), breast cancer is the second leading cause of cancer deaths in women and ovarian cancer is the fifth. In the United States, the general population of women has a 12.8% risk of developing breast cancer and a 1.4% chance of developing ovarian cancer by the age of 90 (NCI, 2012).

There have been significant advances made in our knowledge of breast cancer over the years. Studies in the 1990s demonstrated the existence of an autosomal dominant form of breast and ovarian cancer risk, and led to the identification of two highly penetrant genes: BRCA1 on chromosome 17 (Hall et al., 1990) and BRCA2 on chromosome 13 (Wooster et al., 1994; Wooster et al., 1995). Women with a germ-line mutation in BRCA1 or BRCA2 have a 50-87% risk of developing breast cancer and a 27-44% risk of developing ovarian cancer by age 70 (Ford et al., 1994; Ford et al., 1998). These women also have an increased risk of developing a second primary breast cancer or ovarian cancer within 10 years of an initial breast cancer diagnosis (Metcalf et al., 2004; Metcalf et al., 2005). Other reports suggest a slight increase in risks of additional cancers for BRCA1 and BRCA2 mutation carriers as well (NCI, 2013). In addition to the individual’s cancer risks for women with a mutation, their family members are at significant risk of carrying the mutation (and thus the increased risk for cancer).
In the general population, the carrier frequency of inherited mutations in BRCA1 and BRCA2 is not relatively high (estimates range from 1 in 300 to 1 in 800), but these mutations are thought to account for up to 7% of breast cancer and 14% of ovarian cancer cases (Pal et al., 2005). Because only about 7% of women with breast cancer will be found to carry a BRCA1 or BRCA2 mutation, not all breast cancer patients are appropriate candidates for genetic testing. To help the healthcare provider, there exist professional guidelines that can aide in the identification of individuals that are appropriate for further genetics risk assessment (NCCN, 2014). It is suggested that those women meeting these specific criteria be referred for further genetics risk assessment by a genetics professional. Genetics professionals typically rely on expert opinion criteria to help guide them when offering genetic testing to individuals at risk of carrying a BRCA1 or BRCA2 mutation (NCCN, 2014).

Many professional organizations agree that individuals deciding to have genetic testing should undergo genetic counseling, since the importance of pre-test and post-test genetic counseling for cancer susceptibility testing is widely recognized (ASCO, 2003). Referral to a cancer genetic professional is also critical because the provider ordering the genetic testing must understand the complexities of the process and the appropriate interpretation of the test results. Practice guidelines exist to guide the genetic counselor in this process (Berliner et al., 2013; Riley et al., 2012). Figure 1 illustrates the typical course of the cancer genetic counseling process as described by Christinat and Pagani (2013).
Historically, the process of BRCA1 and BRCA2 genetic counseling and testing occurred after the completion of a woman’s treatment for her breast cancer. Women with a BRCA1 or BRCA2 mutation have a 40-60% risk of developing a contralateral breast cancer at some point in the future (NCI, 2013). Therefore, during their genetic counseling session, a genetic counselor would discuss the options of more aggressive screening for a second primary breast cancer (including annual mammography and breast MRI screening) (Kriege et al., 2004; Stoutjesdijk et al., 2001; Warner et al., 2004), and consideration of prophylactic...
Bilateral prophylactic mastectomy reduces the incidence of breast cancer in BRCA1 and BRCA2 mutation carriers by at least 90% (Rebbeck et al., 2004). In addition, after treatment for breast cancer is complete, women with BRCA1 and BRCA2 mutations should consider risk-reducing bilateral salpingo-oophorectomy (RRBSO), ideally between the ages of 35 to 40 years, or on completion of child bearing (Rebbeck, 2002; Rebbeck et al., 2002; Kauff et al., 2002), given the significant risk of developing ovarian cancer. Preventive oophorectomy can reduce the risk of ovarian cancer by 80% in BRCA1 and BRCA2 carriers (Finch et al., 2014). Neither of these surgical options, mastectomy or oophorectomy, would be recommended as intensely for a woman who does not have a genetic predisposition to develop breast and/or ovarian cancer. Thus, genetic testing can help clarify cancer management recommendations in women with a diagnosis of breast cancer.

Recent studies have indicated that a woman’s knowledge of her genetic status at the time of diagnosis may influence treatment recommendations and decisions (Silva, 2008; Wevers et al., 2011). Some women who test positive for a mutation in BRCA1 or BRCA2 would undergo bilateral mastectomy at the time of diagnosis, not only to treat her existing breast cancer but also to reduce the risk of developing a second primary breast cancer. Because BRCA status may influence surgical treatment of breast cancer, women are now often offered genetic testing at the time of their breast cancer diagnosis. This process has been deemed ‘treatment-focused genetic counseling and testing’ (Meiser et al., 2008).

There has been concern that treatment-focused genetic testing (TFGT) may place additional psychological stressors on patients. A focus group of women with a BRCA1 or BRCA2 mutation reported that they felt there was no ‘right’ time for everyone in terms of
the timing of introducing the option of BRCA genetic testing. They preferred to be told that genetic testing was available if they wanted it at the time of diagnosis, rather than simply being offered the test (Ardern-Jones et al., 2005). Interviews with women diagnosed with breast cancer under the age of 50 revealed positive attitudes toward TFGT as well (Zilliacus et al., 2012). These women thought it was highly relevant, and that offering TFGT did not add an undue psychological burden. In another study, genetic counseling and testing offered to breast cancer patients between diagnosis and surgery did not cause short or long term distress, but did influence surgical treatment (Wevers et al., 2012). For women with breast cancer, genetic testing has historically been seen more as an after thought because they are dealing with treating the breast cancer they already have (Hamilton et al., 2009). Although testing was a secondary concern, patients have said that the most important information given during their genetic counseling session for influencing risk-reduction surgery decisions was their BRCA test results (positive or negative) (Ray et al., 2005). Hopwood (2005) also found that personally tailored risk information that addresses patients’ concerns increased their understanding of breast and/or ovarian cancer risk. All of these studies suggest that TFGT should be offered, but that it should be done in a supportive environment that is responsive to the issues of a patient’s psychological reaction and decision-making questions that may present in these situations.

While many professional organizations agree that genetic counseling be offered for women undergoing genetic testing, it is unclear as to who is the most appropriate healthcare provider to present this information at such a sensitive time point. It is widely accepted that patients identify physicians as one of their most important sources of information about their healthcare (Wonderlick & Fine, 1997). However, more recently,
patients have expressed a lack of confidence in their physicians having sufficient genetic knowledge and felt they had misguided them or prevented them from learning important risk information earlier (MacDonald et al., 2010). Given this absence of direction, various providers order BRCA genetic testing and “genetic counseling” is delivered by different healthcare professionals. We were interested in exploring the experience of women who are tested outside of the traditional model where a trained genetic counselor provides pre-test genetic counseling prior to ordering the genetic test.

The Mass General Cancer Center has a unique program that offers a Multidisciplinary Breast Cancer Clinic (“Multi Clinic”) for newly diagnosed breast cancer patients. Individuals are afforded the opportunity to consult with a surgical oncologist, medical oncologist, and radiation oncologist in one scheduled appointment. In a process that is unique to the Mass General Cancer Center, patient records are reviewed prior to their multi appointment by a certified genetic counselor to determine whether or not the patient is appropriate for discussion of genetic testing. If the patient's history meets the established criteria (Table 1) the patient is triaged to meet with a genetic counselor at the time of their Multi Clinic visit.

**Table 1. Mass General Center for Cancer Risk Assessment (CCRA) Genetics Triage**

- Early age onset breast cancer (<45y)
- Previous breast cancer or previous ovarian cancer
- A family member with two or more breast primaries, or breast and ovarian cancer
- A family member with breast cancer diagnosed ≤45y
- A family member with ovarian cancer
- A family member with male breast cancer
- Two or more individuals with breast cancer on the same side of a family (maternal or paternal)
- Ashkenazi Jewish descent
- Patient who is a member of a family with a known mutation in a cancer susceptibility gene
- Any male breast cancer
- Triple-negative cancer diagnosed <60y
Genetic counselors at the Mass General Center for Cancer Risk Assessment (CCRA) have noted that an increasing number of individuals presenting to the Multi Clinic are coming in with genetic testing already in progress – ordered by an outside provider. There is a paucity of data about whether or not this course of action is ideal. This study is directed toward exploring breast cancer patient experiences of BRCA genetic testing when a genetic counselor is not involved.
METHODS

Methodology

The Massachusetts General Hospital (MGH) and Brandeis University’s Institutional Review Boards approved our study protocol and materials. We utilized qualitative methods through interviews to better understand participants’ individual experiences.

Sampling Methods and Participant Recruitment

We recruited participants through the Multi Clinic and the Facing Our Risk of Cancer Empowered (FORCE) website. Subject inclusion criteria were as follows: 18 years or older, female, personal diagnosis of breast cancer, and BRCA genetic testing results or BRCA genetic testing in progress when the pre-test counseling was not performed by a genetic counselor. The CCRA genetic counselors identified those patients recruited at the Multi Clinic during their pre-screen of the patients for genetic testing. If a CCRA genetic counselor found a Multi Clinic patient’s non-genetic counselor provider had already ordered genetic testing, they approached that patient during their Multi Clinic appointment and invited her to participate in the study. To increase recruitment outside of the Multi Clinic, we posted a recruitment notice (Appendix A) on the FORCE website’s message board asking participants to be in contact with the interviewer if they met the inclusion criteria. A CCRA genetic counselor then contacted patients that responded to this recruitment notice and reviewed the inclusion criteria with them to determine whether or not the patient was eligible for the study. A CCRA genetic counselor also reviewed the consent form (Appendix B) with each study participant either in person or over the telephone, and obtained her
written consent. A CCRA genetic counselor collected demographics information (Appendix C) through medical records review (Multi patients) or telephone interview (FORCE participant). Once an individual agreed to participate in the study, the interviewer contacted them and coordinated a time to conduct the interview. Each participant received a $25 gift certificate in appreciation of her contribution.

**Interviews**

We designed an interview guide (Appendix D) with open-ended questions to elicit the experiences of our participants in a conversational yet semi-structured format. This guide and the subsequent interviews included topics such as who ordered the patient’s testing, why they considered testing, how they felt waiting for results, what their results meant to them, and would they change anything about their genetic testing process. Prior to conducting interviews with participants, this interview was pilot tested with the primary investigator who has personal and professional experience with breast cancer patients.

We assigned each interviewee a unique identifier to ensure confidentiality. For consistency, the same investigator interviewed all participants using the same interview guide. However, the amount of time spent on each topic, the exact questions asked, and the order of topics was dependent on the experiences of each participant. The interviewer audio-recorded all interviews with consent from the participant, and the interview duration ranged from 10 to 30 minutes. A professional transcription service transcribed the interview recordings.

**Data Analysis**

We uploaded our data into Atlas.ti (version 7.0), a software program for qualitative analysis, and analyzed the data using electronic coding, as well as manual categorization.
To begin we coded sections of text, paragraphs, phrases, or views the participants expressed during their interviews. Then we manually categorized the original set of 71 codes into 13 code groups based on the interview guide. From these 13 categories, 3 major themes emerged: the value genetic testing holds for the participants, the emotions patients experience throughout the genetic testing process, and patient reactions to their interactions with providers. We analyzed the patterns and variations among individual responses within these themes.
RESULTS

Based on the inclusion criteria, the participants in this study were all female and had recently been diagnosed with breast cancer. Only one woman had two diagnoses of breast cancer. Demographics for participants are listed in Table 2.

<table>
<thead>
<tr>
<th>Participant #</th>
<th>Recruitment site</th>
<th>Age at diagnosis</th>
<th>Ordering provider</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Multi Clinic</td>
<td>43</td>
<td>Surgeon</td>
</tr>
<tr>
<td>2</td>
<td>Multi Clinic</td>
<td>44</td>
<td>Intake nurse</td>
</tr>
<tr>
<td>3</td>
<td>Multi Clinic</td>
<td>40</td>
<td>Surgeon</td>
</tr>
<tr>
<td>4</td>
<td>FORCE</td>
<td>44 &amp; 56</td>
<td>Gynecologist</td>
</tr>
</tbody>
</table>

The main themes that emerged from the interviews were the value of testing, patient emotions, and interactions with providers.

**Theme 1: Value of Testing**

The beginning of the interview gathered information on the genetic testing process of each participant. We asked questions about the process of how the BRCA test was initially offered and ordered. Participants spoke of their awareness of genetic testing before their provider offered it, and the process involved in being tested, including who informed them of and ultimately ordered the test. The reasons why participants considered testing echoed four main topics: helping to decide on surgical options, assessing risk to family members, determining future risk of cancer, and curiosity.

**Surgical options**

Only 25% of participants stated that one of their reasons for testing was that the results would impact the recommendations for surgery. However, 75% of participants
indicated that they did or did not have specific surgeries based on their genetic test results.

Table 3 lists the surgeries each participant elected to have performed and if they were influenced by their genetic test result.

Table 3. Surgeries

<table>
<thead>
<tr>
<th>Participant #</th>
<th>Staging of Breast Cancer</th>
<th>BRCA Status</th>
<th>Breast Surgery</th>
<th>Prophylactic Oophorectomy</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>Type</td>
<td>Influenced by gene test (Y/N)</td>
</tr>
<tr>
<td>1</td>
<td>1</td>
<td>Negative</td>
<td>Lumpectomy</td>
<td>Y</td>
</tr>
<tr>
<td>2</td>
<td>2</td>
<td>Negative</td>
<td>Bilateral mastectomy</td>
<td>N</td>
</tr>
<tr>
<td>3</td>
<td>2</td>
<td>Negative</td>
<td>Bilateral mastectomy</td>
<td>N</td>
</tr>
<tr>
<td>4</td>
<td>1</td>
<td>Positive (BRCA2)</td>
<td>Bilateral mastectomy*</td>
<td>N</td>
</tr>
</tbody>
</table>

*Surgery based on new diagnosis of breast cancer.

Patients reported that sometimes the results of genetic testing helped determine the best option for what surgery to proceed with. One participant said she had genetic testing because:

“*[M]y understanding was if I was positive then there would have been a potential recommendation to take the surgery a little bit further...”*

Since her test results came back negative, and had been ordered by her surgeon, she proceeded with a lumpectomy and did not have more invasive surgery.

**Risk for Another (non-breast) Cancer**

Fifty percent of participants identified the chances of developing ovarian cancer as a motivating factor when deciding to have testing. They reported that while going through a current cancer diagnosis, to hear that you may be at increased risk for it to happen again could be upsetting, but also informative:

“*[T]here is a link to ovarian cancer and there is not a screening process for that. And I think I, I guess when I was in that mode of hearing all of this stuff I wanted to hear everything and kind of make a holistic decision if I could.”*
Whether stated or not, all participants did (positives) or did not (negatives) have their ovaries removed based on the result of their genetic test.

**Risk to family**

Fifty percent of participants discussed the possibility that other family members, such as children and siblings, could be at risk of also carrying the mutation if they tested positive and that played a role in their decision to undergo genetic testing. Table 4 presents family history information gathered from participants.

<table>
<thead>
<tr>
<th>Participant</th>
<th># of children</th>
<th>Family history of cancer</th>
<th>Family history of HBOC cancers</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>1</td>
<td>+ (Colon)</td>
<td>-</td>
</tr>
<tr>
<td>2</td>
<td>3</td>
<td>+ (Breast and Ovarian)</td>
<td>+</td>
</tr>
<tr>
<td>3</td>
<td>1</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>4</td>
<td>2 (adopted)</td>
<td>+ (Ovarian)</td>
<td>+</td>
</tr>
</tbody>
</table>

One woman stated:

“Well the importance of the test was more or less that if you test positive for the gene you have a 50% chance of passing it on to your children and they can come from your mother or your father, and that is why I decided, because I have children.”

Another participant explained why she wanted the information:

“I have a lot of sisters, so for my family's knowledge I wanted to know.”

While not stated as a reason she considered testing in the first place, one woman shared that she now knows all-too-well how impactful her results can be for her siblings:

“[M]y younger sister has never had anything. [...] She went and was tested and she also is BRCA2, and then in December she had her ovaries out and she actually has ovarian cancer. And so she is in chemo right now, but it was literally-- I mean we are very grateful for the testing. I would have been okay. [...] She wouldn't have been [...] she already had it in both ovaries and it had spread to her appendix, but it had not spread to any lymph nodes. By the time they found it in her it would have been a death sentence is what her oncologist is telling us.”
Curiosity

Participants without a significant family history of breast and/or ovarian cancer are driven to get genetic testing for the sake of collecting information.

One woman stated she wanted the testing:

“Just for my own knowledge...I just was kind of curious of where the breast cancer came from, because I have no family history.”

One woman almost declined testing, but her curiosity made her change her mind:

“I mean at that point I was seriously clueless and it was just I could have gone either way. I mean I look back on it and I’m very, very grateful that I got the testing, but at that moment I could have literally gone either way. I remember sitting there going, ‘I’ll just wait a year.’ And then something just kind of, I said, ‘Let’s just do it, get it done, whatever.’ And that was it.”

Theme 2: Patient emotions

The next section of the interview aimed to understand the different emotions that a patient experiences throughout the genetic testing process. We asked participants questions about the first time genetic testing was brought up for discussion, the waiting period between testing and receiving results, and what their result meant to them.

The Waiting

One hundred percent of participants remembered their provider informing them how long it would take to get results. Participants reported that the timeframes their providers quoted varied from as short as two weeks, to as long as five weeks. The real results were reported to some more quickly than expected, with the shortest turn-around time being nine days. Some tests took even longer than anticipated (often due to insurance issues), at almost two months.

Seventy-five percent of the participants stated they were not overly anxious or worried while waiting for their test results. These women reported that they were
preoccupied with surgery, or believed their risk of testing positive was low. One woman explained that:

“[T]he best way I can describe it in my head is like a checklist. You feel like you have a lot of concerns and you don’t know what is going to happen, and you just, I think-- For me anyway, my whole process was very much based on faith and my outlook was based on faith, and you kind of just whatever it was going to be it was going to be. It’s already kind of determined and I’ll deal with it when I get it. So yup, it was-- But once you get it it was like, “Okay, now I’m past that. Now what’s next?””

Feelings about the genetic testing process

One hundred percent of participants were content with the way they received their results. Seventy-five percent of participants had their results disclosed over the telephone, while 25% had an appointment to hear their results. Those participants who received a telephone call reported that this was preferable to waiting for an appointment. One woman said she requested a telephone call because:

“Waiting for an appointment before you get your results, that’s not good. Then you’re just waiting longer.”

What testing results mean

We asked participants to describe what testing either positive or negative meant to them, and 50% of participants said it was a relief to know that they were negative. They reported that the test results meant that they did not necessarily need to have as aggressive or additional surgeries. One participant had mixed emotions about testing negative since she still had an extensive family history of breast and ovarian cancer:

“It’s great. I mean, well, it’s complicated what it means to me, because there is still something wrong with my genes that we can’t identify, so it’s a blessing and a curse. It’s mixed emotions about it, because okay good, I’ve got a 50% chance I’m not going to get cancer recurrence, but what don’t I know, you know what I mean, like-- I don’t know. I mean it’s a relief, but then it’s not. It’s a relief because okay, so this, what we do know is not the case, so we’re back to what we don’t know.”

For the participant whose test result was positive, she felt:
“It was a fork in the road. It was a complete life changer.”

Based on her results, this participant had a bilateral salpingo-oophorectomy and began increased breast screening. While she did not plan to have a bilateral mastectomy, a second breast cancer diagnosis after her first MRI (along with additional screening issues) prompted her to have a bilateral mastectomy.

**Theme 3: Interactions with providers**

When BRCA testing occurs outside of the traditional genetic counseling model, the role of information giving falls in the hands of the ordering health professional. Some of the participants felt that they were well informed, either by their health care provider or from personal gathering of information. However, what and how much information was relayed to each patient about genetic testing varied greatly, except that all participants seemed to understand the need for additional surgery if their test result came back positive. None of these women chose to follow-up with a genetic counselor after their genetic testing, even when offered at the end of the interview.

**What providers say matters**

Participants shared what they found helpful to know before moving forward with the testing. They reported that most of the information shared had to do with facts about the testing procedure (e.g., that it would involve a blood draw, and how long it would take to get results). Participants recalled that there was also some discussion about recurrence risks, medical management, insurance, and the type of results one might receive; however this information was not consistent across participants.

One participant reported she liked the fact that her doctor was not reassuring about her results:
“I don’t know if [my doctor] weighed in and said, “Oh you’re probably fine.” I don’t believe she did, which I actually appreciated, because I’d rather not be told I’m fine and then find out otherwise, because I think that-- I’ve had a couple of instances where some of the doctors have said, “Oh this should be just nothing.” And it turned out the opposite. And I know that they’re trying to be reassuring, but if they’re wrong I think it just kind of gives you a double whammy.”

What providers should know: make it personal

With the final questions of the interview, we asked participants to think back on their whole genetic testing process. Upon reflection, participants said that they would not change their experience or have done anything differently, except if there was a way to get results back more quickly. Seventy-five percent of participants shared final thoughts on what they would like healthcare professionals to know about their experience. One participant felt that the providers she interacted with went above and beyond her expectations:

“The information you get can be overwhelming, but everyone is patient and they listen and they answer all of your questions no matter what and no matter what the question. Everyone is well informed. It’s just very impressive.”

Another participant spoke highly of her experience and physician:

“I felt like my experience was very personal and very-- I felt like [my doctor] was very personal, very thorough, very kind. I think empathy has got to be involved because of everything you’re going through. And I think that, I mean I have experienced nothing but that, which is why I think it was important for her to call me as opposed to somebody running down a checklist of, “Hey, here are the people you have to call with results today.” It’s not as personal as having [my doctor] call me back was very personal. I was surprised. I was very surprised that it was her that called me, but very grateful.”

What providers should know: education is essential

All participants received some information on genetic testing before it was ordered, but not all health professionals knew what to do once the results came back. This situation arose for one participant, prompting this reaction:

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“Well, I think sometimes the education among healthcare professionals is probably not where it should be. On the oncologist and surgeon side when you’re at that level it’s taken extremely seriously and treated appropriately, but ... many of the nurses that treated me did not even know what BRCA was.

[...]
I think the genetics side of it has gone a lot further. I do think that the oncologists for sure are very well educated and as are the surgeons, but the normal medical care professional ... most of the time don’t know what you’re talking about, and you have to kind of explain it to them, especially if it’s something that should be in your records for them.”

She felt that educating providers about genetic testing was critical:

“there is not enough people who are checking family history and people understanding that there is, that this genetic test is really, really important. And so I think that needs to-- They need the education down a little further.
[...]
I think there has to be more of a unified approach as far as that goes, as far as testing and treating people and notifying and all of that. I just think that there is just not enough education right now. I don’t think people understand how important it is.”
DISCUSSION

In this study, we interviewed four women with breast cancer, who had BRCA genetic testing without meeting with a genetic counselor, in an attempt to gather information about the genetic testing process from their perspective. The themes and subthemes that emerged indicate what motivated these women to have genetic testing, the process they went through, and what was important to them about the genetic testing process. The information gathered can help develop ways that healthcare providers can improve this process for patients. Participants noted that their motivations were primarily for surgery decision-making, that they appreciated when health professionals made the experience individualized to the patient, and that increasing education of healthcare providers about the meaning of genetic testing could better the process in the future.

Motivations for testing

Participants in this study had genetic testing because their provider informed them of this option and explained that it would have implications for their medical management. The provider in each participant’s experience raised the topic of genetic testing, even with those women who reported having prior knowledge of genetic testing unrelated to their personal diagnosis. Patients reported that often the results of genetic testing helped inform their surgical decisions. The test results impacted all of the women’s surgical choices. While dealing with their current diagnosis of cancer is the main priority for these patients, previous studies have also found that patients’ desire to have the testing ordered as quickly as possible is important so that they can determine with what surgery to proceed
(Hamilton et al., 2009). One of the women who underwent testing used the test results to choose lumpectomy instead of bilateral mastectomy and another woman used the test results not to change her breast cancer surgery choice (she opted for bilateral mastectomy regardless of the results), but was able to avoid an oophorectomy because she did not have a BRCA mutation. The influence of genetic test results on clinical management and surgical options is evident also in the woman whose breast cancer occurred 12 years ago who was wavering in her choice to pursue genetic testing. While it did impact her medical management (she decided to have an oophorectomy based on the presence of a BRCA mutation), she was far removed from her breast cancer diagnosis and having genetic testing did not seem to be something that was as imminent to her. Healthcare providers should note that the impact of genetic test results remains a clear priority for patients regardless of the timing of the testing.

Another incentive for women to have genetic testing was their concern for their family members. If a person is found to have a BRCA1 or BRCA2 mutation there is a 50% chance that they will pass it on to each of their children. In most cases, brothers and sisters of a person with a mutation also have a 50% chance of having the mutation. Additionally, other family members may be at risk to have the mutation. The particulars of inheritance if the patient tested positive were possibly not expressed to all participants since only 50% of them mentioned it as a motivation for undergoing genetic testing. While this information may not be pertinent for patients to understand before their surgery or treatment, it is a topic that needs to be thoroughly explained to each patient at some point after they receive their genetic testing results. Healthcare providers should note that the familial implications
of genetic test results are important to patients, and that these implications must be addressed for the full benefit of genetic testing to be realized.

**What patients want in the testing process**

Individualized care is highly appreciated by patients, and has also been proven to decrease concerns and increase understanding of risks (Hopwood, 2005). When a patient’s current provider orders the genetic testing, they feel as though a personal approach is being taken. Through this process, patients are kept connected to someone they already know and have a relationship with during a delicate time. Patients may see their provider’s office as the supportive environment needed for this treatment focused genetic testing (TFGT) (Ardern-Jones et al., 2005; Wevers et al., 2012; Zilliacus et al., 2012), and would rather not introduce a new provider into their situation at that time. Again, it is all about their diagnosis and what steps need be taken to address their current condition. It is important to note, however, that some patients reported that their own physician might not have the expertise to deal with the nuances of genetic testing (MacDonald et al., 2010). For this reason, it is important for providers to recognize that appropriate follow up with a genetics specialist is needed.

The specific information important to patients at the time of testing is mostly related to logistics surrounding their results (including insurance coverage and timeframe of results return), and recommendations for surgery. The insurance approval process can differ between insurance companies causing the turn-around time of the test results to vary. Patients expressed their desire to get their results back as soon as possible, and many appreciated that their results were returned over the telephone. Therefore, even if their insurance delayed the testing procedure, the patient still received their results as soon as
they were available. Then, armed with this knowledge, the patient proceeded with the most appropriate surgical option. It is important that healthcare providers recognize the need for rapid communication of test results.

In the traditional genetic counseling model, when genetic counselors provide the pre-test genetic counseling, the information shared with patients follows practice guidelines (Berliner et al., 2013; Riley et al., 2012). Other healthcare providers may not follow this standardized approach. Arguably though, other information typically reviewed during pre-test genetic counseling, such as family history, risks to family members, and risk of recurrence, is not as important to the patient at the time as the implications on their clinical management. Healthcare providers must recognize, however, that the implications the test results have on these other matters must be dealt with after the imminent treatment of the patient’s current diagnosis is complete.

**Post-results information**

Today the accessibility of genetic testing is growing rapidly. However, not all providers are as knowledgeable about the details of the identification of appropriate testing candidates, the genetic testing process, and the interpretation of the test results.

Identification of appropriate testing candidates is important (NCCN, 2014). One instance where this could have helped was with one of our participants who was diagnosed with breast cancer 12 years ago and should have been offered genetic testing much earlier. As she said herself:

“It may have helped both my sister and I had I found out a year earlier...”

Women in similar situations may be missing out on getting genetic testing because it was not offered when they were first diagnosed with cancer, but now current criteria stipulates
they should consider testing. Healthcare providers may not be aware of these updates to testing guidelines. It is imperative that healthcare providers are kept up to date on such changes to medical care.

When a person is diagnosed with breast cancer at a young age (<45 years old), some agree that it is best for them to see a cancer genetics specialist because there are other gene mutations that can cause an increased risk for breast cancer (NCCN, 2014). For example, our participant who has a significant family history of breast and ovarian cancer, but tested negative for a BRCA mutation may be at risk for other hereditary cancer syndromes. If she had met with a genetic counselor additional testing may have been offered. The genetic testing process is changing rapidly, even for professionals in the field of genetics. Healthcare providers must recognize that referral to a genetics professional is often in the best interest of the patient so that the patient’s genetic testing is complete and most appropriate.

Not all providers who order genetic testing know how to interpret the results once they are returned (Brierley, et al., 2010; Brierley, et al., 2012). One participant in our study found herself being passed on to other healthcare professionals when her test result came back positive. The ordering provider, her gynecologist, only was prepared to deal with the implications the testing had to his specialty. This woman has also explained what her BRCA status means to some of her general practitioners. Healthcare providers must recognize that referral to a genetics professional is often in the best interests of the patient so that the most up to date information on genetic testing is communicated to the patient.

After receiving their test results, patients continue to need support (Berliner et al., 2013; Riley et al., 2012). Whether they are positive or negative, it is important to ensure
that the patients are adequately informed about future risks of cancer and to family members. One difficulty in this approach will be ensuring that all individuals that undergo genetic testing are referred properly, and that they follow through with post-test counseling. It is important to note that none of the participants in this study saw a genetic counselor, even when offered the opportunity at the end of the interview. One woman mentioned that she could have met with a genetic counselor, but she would have waited two months for the appointment and that was too long for her. Therefore, these healthcare providers can improve the well being of patients and the experience of genetic testing by being familiar with known factors of hereditary cancer, and follow-up recommendations for test results when follow up genetic counseling is not occurring. Making genetic counseling more immediately available to patients is imperative.

Limitations and Future Research

The greatest limitation of this study was a small sample size. Future studies should include a larger sample with uniformed demographics, as well as comparison to controls (women who did have pre-test genetic counseling performed by a genetic counselor). Furthermore, the participants were recruited mainly from one institution. The experiences of patients in other geographic areas would undoubtedly be a valuable addition to the study findings. Standardized measures of knowledge or psychological reactions were not utilized in this study but would enhance the information gathered from participants. Not only speaking to patients about their experiences, but also talking to healthcare providers about ordering genetic testing would enrich the information on this topic. As stated previously, none of the participants in this study met with a genetic counselor. If they had, it would have helped to understand and identify other disparities and gaps in patient
knowledge. Future research should include development of models to ensure best practice. The material gathered through this study can begin to inform questions for a survey that would further investigate the experiences of patients with other cancer diagnoses who have had genetic testing ordered outside of the traditional genetic counseling model and would allow these findings to be applied in a broader context.
CONCLUSION

The purpose of this study was to explore the experiences of women who have been diagnosed with breast cancer and have received BRCA testing without meeting with a genetic counselor. Participants felt that they received adequate information about genetic testing before proceeding. However, there were details that are important, such as medical management recommendations and implications for family members that were not told to all participants. With the expansion of genetic testing awareness in the general population, the education about these procedures needs to be understood by all providers in a general sense so that their patients do not perceive their medical care to be disjointed.

Based on the genetic testing experiences of these participants, this study suggests BRCA testing ordered by non-genetic counselor healthcare providers is not unfavorable or detrimental to patients. However, a genetic counselor can greatly enhance the process by providing back up to other healthcare providers so that information can be clarified in the long term. These healthcare providers can also improve the well being of patients and the experience of genetic testing by being familiar with known factors of hereditary cancer, and follow-up recommendations for test results.
REFERENCES


APPENDICES

Appendix A: FORCE Recruitment Notice

Have you undergone genetic testing recently? Was your testing performed by someone other than a genetic counselor? If you answered YES to these questions, we are interested in speaking with you!!

Margaret Hayes, a graduate student in the Brandeis Genetic Counseling Program, together with colleagues at Massachusetts General Hospital, is researching actual experiences/perceptions of persons undergoing genetic testing. Your expertise on this topic is important because genetic testing is becoming more widespread and healthcare professionals want to make sure that we are appropriately caring for patients. The study involves a telephone interview, which should last approximately one hour. Participants receive a $25 gift card as a token of appreciation. If you are interested and think you may be eligible, please email Margaret Hayes at mhhayes@brandeis.edu. Thank you for your consideration!
Appendix B: Consent Form

Partners HealthCare System
Research Consent Form

General Template
Version Date: February 2010

Subject Identification

Protocol Title: EXPLORING THE BRCA1/2 GENETIC TESTING EXPERIENCES OF BREAST CANCER PATIENTS WHO DO NOT MEET WITH A GENETIC COUNSELOR BEFORE ORDERING TESTING

Principal Investigator: Kristen Shannon

Site Principal Investigator:

Description of Subject Population: Breast Cancer Patients

About this consent form

Please read this form carefully. It tells you important information about a research study. A member of our research team will also talk to you about taking part in this research study. People who agree to take part in research studies are called “subjects.” This term will be used throughout this consent form.

Partners HealthCare System is made up of Partners hospitals, health care providers, and researchers. In the rest of this consent form, we refer to the Partners system simply as “Partners.”

If you have any questions about the research or about this form, please ask us. Taking part in this research study is up to you. If you decide to take part in this research study, you must sign this form to show that you want to take part. We will give you a signed copy of this form to keep.

Why is this research study being done?

This research study is being done to explore the BRCA1/2 genetic testing experiences of breast cancer patients. That is, what was the process that they went through to get their genetic testing and how did they feel about it. We are asking you to take part in this research study because you have been diagnosed with breast cancer and have undergone BRCA1/2 genetic testing.

How long will I take part in this research study?

Your participation will be limited to participating in a phone interview, designed to explore your experience with BRCA1/2 genetic testing. The interview should take approximately one hour.
What will happen in this research study?
Your medical records will be reviewed to obtain demographics, your family history information, details about your diagnosis with breast cancer, as well as details about your treatment for breast cancer. You will be contacted approximately a week after receiving your genetic test results and asked to participate in a study interview over the phone. The interview will take approximately one hour to complete. The interview will be audio recorded and the recording will be kept secured until transcribed and then will be destroyed. We anticipate that 10 women will participate in this study.

What are the risks and possible discomforts from being in this research study?
There are risks to taking part in any research study.

The risks of this study are expected to be minimal. You may experience psychological distress when you complete the interview. You do not have to answer any question that makes you feel uncomfortable.

What are the possible benefits from being in this research study?
Taking part in this research study may or may not benefit you. We hope the information learned from this research study will provide more information about the process of genetic testing. It is hoped that exploring patient experiences with genetic testing will allow for a more appropriate process in the future.

Can I still get medical care within Partners if I don’t take part in this research study, or if I stop taking part?
Yes. Your decision won’t change the medical care you get within Partners now or in the future. There will be no penalty, and you won’t lose any benefits you receive now or have a right to receive.

Taking part in this research study is up to you. You can decide not to take part. If you decide to take part now, you can change your mind and drop out later. We will tell you if we learn new information that could make you change your mind about taking part in this research study.
What should I do if I want to stop taking part in the study?

If you take part in this research study, and want to drop out, you should tell us. We will make sure that you stop the study safely. We will also talk to you about follow-up care, if needed.

It is possible that we will have to ask you to drop out before you finish the study. If this happens, we will tell you why. We will also help arrange other care for you, if needed.

Will I be paid to take part in this research study?

You will not be paid to participate. As a token of our appreciation, we will send you a $25 gift card.

What will I have to pay for if I take part in this research study?

Although study funds will pay for certain study-related items and services, we may bill your health insurer for, among other things, routine items and services you would have received even if you did not take part in the research. You will be responsible for payment of any deductibles and co-payments required by your insurer for this routine care or other billed care. If you have any questions about costs to you that may result from taking part in the research, please speak with the study doctors and study staff. If necessary, we will arrange for you to speak with someone in Patient Financial Services about these costs.

What happens if I am injured as a result of taking part in this research study?

We will offer you the care needed to treat any injury that directly results from taking part in this research study. We reserve the right to bill your insurance company or other third parties, if appropriate, for the care you get for the injury. We will try to have these costs paid for, but you may be responsible for some of them. For example, if the care is billed to your insurer, you will be responsible for payment of any deductibles and co-payments required by your insurer.

Injuries sometimes happen in research even when no one is at fault. There are no plans to pay you or give you other compensation for an injury, should one occur. However, you are not giving up any of your legal rights by signing this form.
Partners HealthCare System
Research Consent Form

General Template
Version Date: February 2010

If you think you have been injured or have experienced a medical problem as a result of taking part in this research study, tell the person in charge of this study as soon as possible. The researcher's name and phone number are listed in the next section of this consent form.

If I have questions or concerns about this research study, whom can I call?

You can call us with your questions or concerns. Our telephone numbers are listed below. Ask questions as often as you want.

Kristen Shannon, MS, CGC is the person in charge of this research study. You can call her at 617-726-9337 M-F 9-5 with questions about this research study.

If you want to speak with someone not directly involved in this research study, please contact the Partners Human Research Committee office. You can call them at 617-424-4100.

You can talk to them about:
- Your rights as a research subject
- Your concerns about the research
- A complaint about the research

Also, if you feel pressured to take part in this research study, or to continue with it, they want to know and can help.

If I take part in this research study, how will you protect my privacy?

During this research, identifiable information about your health will be collected. In the rest of this section, we refer to this information simply as “health information.” In general, under federal law, health information is private. However, there are exceptions to this rule, and you should know who may be able to see, use, and share your health information for research and why they may need to do so.

In this study, we may collect health information about you from:
- Past, present, and future medical records
- Research procedures, including research office visits, tests, interviews, and questionnaires
Partners HealthCare System
Research Consent Form

General Template
Version Date: February 2010

Who may see, use, and share your identifiable health information and why they may need to do so:

- Partners research staff involved in this study
- The sponsor(s) of this study, and the people or groups it hires to help perform this research
- Other researchers and medical centers that are part of this study and their ethics boards
- A group that oversees the data (study information) and safety of this research
- Non-research staff within Partners who need this information to do their jobs (such as for treatment, payment (billing), or health care operations)
- The Partners ethics board that oversees the research and the Partners research quality improvement programs.
- People from organizations that provide independent accreditation and oversight of hospitals and research
- People or groups that we hire to do work for us, such as data storage companies, insurers, and lawyers
- Federal and state agencies (such as the Food and Drug Administration, the Department of Health and Human Services, the National Institutes of Health, and other US or foreign government bodies that oversee or review research)
- Public health and safety authorities (for example, if we learn information that could mean harm to you or others, we may need to report this, as required by law)
- Other: none

Some people or groups who get your health information might not have to follow the same privacy rules that we follow. We share your health information only when we must, and we ask anyone who receives it from us to protect your privacy. However, once your information is shared outside Partners, we cannot promise that it will remain private.

Because research is an ongoing process, we cannot give you an exact date when we will either destroy or stop using or sharing your health information.

The results of this research study may be published in a medical book or journal, or used to teach others. However, your name or other identifying information will not be used for these purposes without your specific permission.

Your Privacy Rights

You have the right not to sign this form that allows us to use and share your health information for research; however, if you don’t sign it, you can’t take part in this research study.
You have the right to withdraw your permission for us to use or share your health information for this research study. If you want to withdraw your permission, you must notify the person in charge of this research study in writing. Once permission is withdrawn, you cannot continue to take part in the study.

If you withdraw your permission, we will not be able to take back information that has already been used or shared with others.

You have the right to see and get a copy of your health information that is used or shared for treatment or for payment. To ask for this information, please contact the person in charge of this research study. You may only get such information after the research is finished.

**Informed Consent and Authorization**

**Statement of Study Doctor or Person Obtaining Consent**

- I have explained the research to the study subject.
- I have answered all questions about this research study to the best of my ability.

______________________________  _______________________
Study Doctor or Person Obtaining Consent  Date/Time

**Statement of Person Giving Informed Consent and Authorization**

- I have read this consent form.
- This research study has been explained to me, including risks and possible benefits (if any), other possible treatments or procedures, and other important things about the study.
- I have had the opportunity to ask questions.
- I understand the information given to me.

**Signature of Subject:**

I give my consent to take part in this research study and agree to allow my health information to be used and shared as described above.
Partners HealthCare System
Research Consent Form

General Template
Version Date: February 2010

Subject Identification

Subject ______________________________ Date/Time ______________________________

Consent Form Version: 1
Appendix C: Demographics Sheet

Study # ________________
Patient initials: __________
Completed by: ______

Demographics

1. Age at diagnosis _______ years
   a. Prior cancer history (cancer type, age at diagnosis) ______________________

2. Race
   1  Caucasian
   2  African American
   3  Hispanic or Latin
   4  Native American
   5  Asian American
   6  Other (please describe):

3. Ethnicity
   Maternal: ________________
   Paternal: ________________

4. Marital Status
   1  Married
   2  Single
   3  Divorced
   4  Domestic Partner
   5  Widowed

5. # Children: ______________

6. # 1st & 2nd degree relatives affected: ______________

Diagnosis

7. Time interval between diagnosis and genetics appt: ________________ days

8. Staging: I₁/ I₁₁ / III₁ / IV₁ / DCIS₁

9. Tumor size: __________

10. Receptor Status:
    a. ER = + / -
    b. PR = + / -
    c. HER2 = + / -
11. Adjuvant chemo: Y₁ / N₂

12. Neoadjuvant chemo: Y₁ / N₂

13. Surgery discussed at time of multi:

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>Lumpectomy</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>2</td>
<td>Mastectomy</td>
</tr>
<tr>
<td></td>
<td>3</td>
<td>Bilateral Mastectomy</td>
</tr>
</tbody>
</table>

**Genetic testing**

14. BRCA1/2 ordering provider

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>Surgeon</th>
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<tbody>
<tr>
<td></td>
<td>2</td>
<td>Oncologist</td>
</tr>
<tr>
<td></td>
<td>3</td>
<td>Primary Care Provider</td>
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<tr>
<td></td>
<td>4</td>
<td>Ob/Gyn</td>
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<tr>
<td></td>
<td>5</td>
<td>Other (specify):</td>
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</table>

15. BRCA1/2 testing lab

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>Myriad</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>2</td>
<td>Ambry</td>
</tr>
<tr>
<td></td>
<td>3</td>
<td>Other (specify):</td>
</tr>
</tbody>
</table>

16. BRCA1/2 testing ordered

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>Sequencing</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>2</td>
<td>Sequencing + del/dup (BART)</td>
</tr>
<tr>
<td></td>
<td>3</td>
<td>Ashkenazi panel</td>
</tr>
<tr>
<td></td>
<td>4</td>
<td>Single site</td>
</tr>
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</table>

17. Specimen type:

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>Blood</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>2</td>
<td>Buccal</td>
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</table>

18. Was additional genetic testing ordered?

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>Yes (specify)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>2</td>
<td>No</td>
</tr>
</tbody>
</table>

19. BRCA1/2 genetic test results

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>Mutation</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>2</td>
<td>No mutation</td>
</tr>
<tr>
<td></td>
<td>3</td>
<td>VUS</td>
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</tbody>
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Appendix D: Interview Guide

Introduction:
Thank you for agreeing to participate in this study. I am conducting this interview to understand the actual experiences/perceptions of persons undergoing genetic testing. Your expertise on this topic is important because genetic testing is becoming more widespread and healthcare professionals want to make sure that we are appropriately caring for patients.

I am going to ask you a series of questions related to your experience with genetic testing. There are no right or wrong answers. If you do not have an answer to a particular question or would rather not answer a question, just tell me and we’ll move on. With your permission, I will be recording the interview so that I do not miss any information. I may also take a few notes during the interview. Afterward, the interview will be transcribed by a confidential transcription service. Your name or any personal identifying information will not be associated with or used in our final report.

The interview will take about one hour. Your participation is voluntary. I would like to give you a $25 gift card as a token of our appreciation.

Do you have any questions before we start?

Great, then let’s begin!

Questions:
1. Could you describe your diagnosis for me? (Prompts: type of breast cancer, bilateral vs. unilateral…)
   a. How was your cancer found?
   b. Have you spoken with a surgeon/do you have a treatment plan set up?
      Where in your treatment are you?
2. Can you tell me how you first found out about genetic testing?
   a. Was it something that you learned about on your own or did someone introduce it to you? (Prompts: your physician, the oncologist…)
   b. How was it brought up? (Prompts: at the end of an appointment…)
   c. What did that feel like?
   d. Did you have questions for the ordering health professional?
3. Can you describe the genetic testing process?
   a. Was your blood drawn or saliva taken?
   b. Can you tell me about the forms (if any) that you needed to sign?
4. What do you remember being told to you about the test? (Prompts: importance, risks, benefits…)
5. What did you find helpful to know before you proceeded with testing?
6. Can you tell me the reasons why you decided to get testing? (Prompts: surgical decisions, other treatment, family…)
7. (What were your results?) Tell me, what does testing “positive” or “negative” mean to you?
8. Think back, can you describe how you felt while waiting for your results? (Prompt: anxious, uncertain…)  
9. Can you describe how you received your test results? (Prompt: phone call, mail…)  
   a. What was that like?  
   b. Would there be a better process?  
10. What actions will you take/have you taken based on your test results? (Prompt: surgery, surveillance…)  
   a. Would you speak to/share them with anyone?  
11. Now that you have completed the genetic testing process, is there anything that you would have liked to have gone differently with regard to your experience?  
   a. Would you have done anything differently?  
12. Is there anything else that you would like healthcare professionals to know about your experience?

We have now reached the end of the interview. Thank you for you time. How was the interview experience for you? Do you have any questions for me?

At this time I would encourage you to schedule an appointment with an MGH genetic counselor to discuss your testing results. They are also available as a resource if this interview experience has caused any emotional distress.