University of Cincinnati

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I, Justine A Snyder B.A., hereby submit this original work as part of the requirements for the degree of Master of Science in Genetic Counseling.

It is entitled:
Tools and Strategies That a BRCA Positive Population Considers to be Useful in the Result Disclosure Process to Family Members

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This work and its defense approved by:

Committee chair: Robert Hopkin, MD
Committee member: Erin Acra Mundt, MS
Committee member: Valentina Pilipenko, PhD

UNIVERSITY OF CINCINNATI
Tools and Strategies That a BRCA Positive Population Considers to be Useful in the Result Disclosure Process to Family Members

A thesis submitted to the
Graduate School
of the University of Cincinnati
in partial fulfillment of the
requirements for the degree of

Master of Science

in the Program of Genetic Counseling
of the College of Medicine

by

Justine A Snyder

B.A. San Diego State University
May 1989

Committee Chair: Robert Hopkin, MD
Abstract

BACKGROUND: Communicating positive test results to at-risk family members can be difficult for individuals with BRCA mutations. Previous studies have identified barriers to disclosure but have not addressed the utility of supportive materials in the disclosure process.

PURPOSE: The purpose of this study was to identify tools and strategies useful to patients with BRCA mutations in disclosing result information to family members.

METHODS: A questionnaire was mailed to 482 patients with BRCA mutations assessing their experience with disclosing test result information to their families.

RESULTS: Of the 177 completed questionnaires received, 90 participants reported that they told every at-risk biological relative about their BRCA positive test results. These 90 questionnaires were analyzed to assess the utility of specific tools and strategies provided by genetic counselors to assist patients with results disclosure. Participants felt that printed materials, such as a sample letter explaining that a BRCA mutation had been identified in the family was the tool expected to be most helpful in disclosing result information. The strategy expected to be most helpful in disclosing BRCA positive test result information was a step by step plan for informing family members. Eighty-seven participants, those who reported they did not tell one or more family members their results, reported reasons for non-disclosure including those that may be considered to be valid reasons, such as at-risk relatives who were under the age of 18 at the time the results were in active discussion, some family members did not wish to be informed of results, and relatives who previously received results by someone other than the participant.

CONCLUSIONS: Results from this study suggest that patients perceive a benefit from receiving a sample letter to distribute, as well as a clear plan to effectively communicate positive results to family members. It is recommended that both be provided by genetic counselors to all patients with BRCA mutations to increase the likelihood that results will be shared with family members. However, this study
found evidence that there are valid reasons some patients may choose not to disclose results to every at-risk relative. This finding requires additional investigation to be completely understood.

**Keywords**  BRCA disclosure · BRCA mutations and family · Cancer genetic counseling · Disclosure of genetic risk · Tools for communicating genetic risk
Acknowledgements

The members of my RAC (Dr. Robert Hopkin, Erin Mundt, Jody Wallace, Dr. Valentina Pilipenko, and Kristin Theobald) for their expertise and guidance through this process and insight into the development of the questionnaire. The Graduate Student Governance Association (GSGA) for supporting this research through grant monies. The use of REDCap for data management through grant support (UL1RR026314).
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Introduction

Genetic mutations in the *BRCA1* and *BRCA2* genes, the cause of Hereditary Breast and Ovarian Cancer syndrome (HBOC), are identified in 7% of breast cancer and 14% of ovarian cancer (Pal, et al., 2005). It is well documented that individuals with *BRCA1/BRCA2* mutations face substantially increased risks for developing cancer, with women having up to an 87% lifetime risk of developing breast cancer and up to a 44% lifetime risk of developing ovarian cancer (Ford, Easton, Bishop, Narod, & Goldgar, 1994; Ford, et al., 1998). Men with a BRCA mutation have up to a 10% lifetime risk of developing breast cancer and an increased risk for earlier onset prostate cancer (Brose, et al., 2002; Thompson, et al., 2002). It is essential that individuals with BRCA mutations disclose their results to their at-risk relatives to allow others the opportunity to better understand their own personal risk and to consider testing. As a result, at-risk relatives may have the opportunity to decrease their risk for cancer and improve long-term outcomes. Not only do family members benefit when results are disclosed, but potential benefits exist for the patient as well. By disclosing results to family members patients have benefited from gaining assistance with medical decision making, experiencing feelings of liberation, and increased emotional support (Hughes, et al., 2002; McInerney, Leo, et al., 2005; Wiseman, et al., 2010).

Although genetic counselors routinely communicate the importance of disclosing genetic risk information to family members, results are not always shared (Wagner Costalas., et al., 2003). Studies have shown as many as 62% of patients failed to share their results (Forrest, et al., 2003; Landsbergen, Verhaak, Kraaimaat, & Hoogerbrugge, 2005). Patients declining to share results with family members may not understand that their relatives could become angry and may resent not having been informed of a known predisposition for cancer (Forrest, et al., 2003).

Specific barriers to disclosure of genetic test results experienced by individuals with BRCA mutations have been identified in previous studies. Family dynamics including factors such as, divorce, lack of contact over time, generation gaps, age of children, and physical distance between relatives may impede result disclosure to at-risk family members (Aktan-collan, et al., 2007; Claes, et al., 2003; Forrest, et al., 2003; Hallowell, et al., 2005; Hallowell, Murton, Statham, Green, & Richards, 1997; Julian-Reynier, et al., 2000; Landsbergen, et al., 2005; Patenaude, et al., 2006; Speice, McDaniel, Rowley, & Loader, 2002; Wiseman, et al., 2010). Individuals with BRCA mutations may avoid telling family members because they are concerned that risk information could impact family relationships, causing either broken bonds between relatives, conflicts among family members, or fear of
feeling isolated (McInerney-Leo, et al., 2005; Speice, et al., 2002; Wiseman, et al., 2010). Numerous emotions like anger, guilt, anxiety, and feeling burdened by sharing results can also impact the decision regarding disclosure of test results (d’Agincourt-Canning, 2006; Mackenzie, Patrick-Miller, & Bradbury, 2009; Speice, et all, 2002; Wagner Costalas, et al., 2003; Wiseman, et al., 2010). Furthermore, individuals with BRCA mutations may fail to disclose because they do not feel as though they have the necessary skills for communicating complex information about treatment, management, prevention, risk probabilities, and genetic testing to family members (Fischhoff, et al., 1999; Smerenik, Mesters, Verweij, de Vries, & de Vries, 2009; Weiseman, et al., 2010).

As there are many perceived barriers patients encounter while attempting to disclose BRCA positive results to their family members, the genetic counselor may play an important role in preparing patients with materials to assist them with such challenges. Therefore, the purpose of this study was to identify tools and strategies useful to patients with BRCA1/BRCA2 mutations in disclosing result information to at-risk family members.
Methods

Study Participants

Eligible participants were men and women who received genetic counseling for positive BRCA1/BRCA2 test results from the Hereditary Cancer Programs at Cincinnati Children’s Hospital Medical Center, CCHMC, (a large, academic hospital) or St. Elizabeth Healthcare, SEHC, (a suburban, community hospital) from 1995 to July 31, 2011. In addition, eligible participants had to be able to understand English, be over the age of 18, and not be adopted. All participants meeting these requirements and whose current address information was available were invited to participate.

Study Questionnaire

A quantitative questionnaire (Appendix A) was developed to assess which tools (materials or instruments explaining BRCA1/BRCA2 mutations and associated risks) and which strategies (a plan or process outlining specific steps for the dissemination of results), utilized by genetic counselors, are most helpful in preparing patients for sharing results. The questionnaire was designed by the study investigators, to assess the sharing of BRCA1/BRCA2 positive results with family members, and was comprised of four primary areas of content including:

(1) participants’ experience with pursuing BRCA1/BRCA2 testing, (2) participants’ experience with the disclosure process including the utility of specific tools and strategies that were expected to help with disclosure, (3) participants’ experience with barriers encountered during disclosure which hindered sharing results with one or more family members, and (4) demographic information. The majority of questions were multiple choice with some questions allowing participants the option of an open-ended qualitative response.

Procedures

A letter (Appendix B) and study questionnaire were mailed to each eligible patient, inviting them to participate in this study. Each questionnaire had a unique identifying number that was used to track responses; however data was stored separately from personal identifying information. Those who did not respond to the first mailing received a second invitation approximately three weeks later. This study was approved by the Institutional Review Boards of Cincinnati Children’s Hospital Medical Center, St. Elizabeth Healthcare, and the University of Cincinnati.
Statistical Analysis

All study data received from the completed questionnaires was loaded into REDCap (Research Electronic Data Capture), a software package hosted by CCHMC. Categorical data were presented as frequencies and percentages. Contingency tables were created to assess participants’ perceptions of tools and strategies. Some responses, such as “other”, “not sure”, or blank, were not included in the analysis. Chi-square test (two tailed) was used to compare variables between those participants who shared their BRCA positive results with all identifiable at-risk family members to those who chose not to share their results with all identifiable at-risk family members. When less than five participants received a particular tool or strategy, Fisher exact test (two tailed) was performed. Statistical analysis was performed using JMP software (JMP, SAS, Institute Inc., Cary, NC).
Results

Study Population

The study questionnaire was mailed to 482 eligible participants and 177 completed questionnaires were included in data analysis assessing participants’ experiences with pursuing genetic testing and demographic information, for a response rate of 36.7%. Of the 177 questionnaires, only 90 of these questionnaires, participants reported having shared their BRCA positive results with all identifiable at-risk family members were analyzed to assess participants’ experience with disclosing results and the utility of specific tools and strategies provided by genetic counselors to assist patients with results disclosure. The remaining 87 questionnaires, participants reported they did not tell all such family members, were analyzed to assess specific barriers participants experienced that hindered telling one or more family members their BRCA positive results. No statistically significant differences between demographic variables were identified between these two groups (Table I).

Motivating Factors for and Experiences with Pursuing BRCA Testing

The first section of the questionnaire assessed all participants’ experiences with pursuing genetic testing for mutations in the BRCA1/BRCA2 genes. Of the 177 participants, 157 provided reasons for having BRCA testing including wanting to know if they had a mutation (73.2%), concern about cancer risk for family members (68.8%), and concern about one’s own cancer risk (62.4%).

Prior to receiving results, 92% of all participants reported that they told family members they were pursuing BRCA testing. When participants were questioned about how family members responded to their decision to pursue BRCA testing, of the 149 who provided one or more responses to the question, 86.1% found their family members to be supportive of their decision to test. In addition, 58.4% of all responders reported family members wanted to be informed of results. However, when this was analyzed between those who shared their results with family members and those who did not, 66.2% of those who told all family members their results reported that family members wanted to be informed of the results compared to only 50% of those who did not tell all family members. This comparison between those who told all of their family members their results and those who did not and their perceived desire of whether family members wanted to be informed of results was statistically significant (p=0.04).
Of those who reportedly did not tell family members they were pursuing testing, 23.8% reported that they wanted to receive results first, 13.1% did not want to worry family members, 6.2% did not feel it was their family’s business, 4.6% did not feel the time was right to discuss their pursuit of testing with family, and 1.5% reported fearing that they would be talked out of testing.

There was no statistically significant difference in telling family members results based on having received follow-up by a genetic counselor, specifically a phone call inquiring about the patient’s experience including any potential obstacles with dissemination of results. Fifty-two percent of those who shared results with all identifiable family members received such follow-up by their genetic counselor compared with 49% of those who did not share with all family members. Most of the participants (78.3%) who responded that they did not recall receiving follow-up from their genetic counselor reported that they would not have wanted to be contacted. Of the 18 participants who responded that it would have been helpful to receive follow-up, 64.3% would prefer to have it occur 3-4 weeks after receiving results.

Ninety-two percent of all participants reported being informed by their genetic counselor of the importance of sharing BRCA test results with their family before testing. Over 96% of participants reported being told after receiving results. However, only 51% of participants responded that they shared their BRCA positive test results with every identifiable at-risk family member.

**Barriers Perceived when Sharing BRCA Positive Result Information with Family Members**

Those who reported that they did not share their results with all identifiable at-risk family members (n=87) were asked to provide specific barriers that hindered telling one or more of their family members their result information. The most commonly reported barrier was family dynamics (51.7%). When asked to explain this further, 47.1% reported that a lack of contact over time with their relatives inhibited their ability to communicate their results. An additional 23% reported that physical distance made it difficult for them to discuss results. Less common hurdles included having large age gaps between the participant and the other family members (11.5%), a feeling of wanting to protect family members from bad news (3.4%), and a divorce or remarriage in the family (4.5%).
Other recognized barriers to disclosing BRCA positive results to family members chosen by study participants included that it was difficult to identify which family members needed to be told (5.7%), it did not feel like it was the right time to disclose the information (3.4%), the result information was not any of their family’s business (2.3%), and participants were not comfortable discussing result information (1.1%). In addition, two participants (2.3%) reported feeling that one or more emotions, such as guilt, vulnerability, burden, anxiety, anger, fear, sadness, worry, or overwhelming thoughts made it too emotional to share result information. Participants who reported that result disclosure would impact family relationships (3.4%) were especially concerned that bonds between family members could be broken and that conflicts could arise as a result of the information. Finally, a perceived lack in specific skills recognized by participants as a barrier (2.3%) included one or more feelings, such as they did not have the ability to answer their family members’ questions, they could not explain BRCA test result information, they did not know how to initiate the conversation with family, and they did not have a good understanding of the BRCA result information themselves.

Reasons participants provided for non-disclosure that may be considered to be valid included family members were under the age of 18 and therefore too young to be told (22%) and relatives were told by someone else in the family first (23%).

Motivating Factors for Disclosure and Tools and Strategies Provided by Genetic Counselors Expected to be Helpful in Sharing BRCA Positive Test Results

Responses to questions about disclosure experience and specific tools and strategies that would be helpful in disclosing to family members were analyzed from the 90 participants reporting having told all identifiable at-risk family members. The desire to inform family members that they were at risk for having a \textit{BRCA1}/\textit{BRCA2} mutation was the motivating factor for sharing results most often reported by this population (94%). Additional responses included informing family members about: the need to consider BRCA testing (78%), the need for increased cancer screenings (76%), specific cancer risks (71%), and cancer risk-reducing strategies (51%). Less frequently motivating factors for sharing were 33% hoped to gain emotional support from their family members and 25% needed their family’s assistance with making medical decisions. The majority of those who told disclosed results to family members within two weeks of receiving their results (88%). Seventy-nine percent reported that their family
members seemed to understand the result information. However, when asked about what information was difficult for family members to understand, 17% reported the inheritance information, 14% reported the genetic information, 12% reported prophylactic surgical options, and 10% reported the cancer risks.

From a list of tools, participants were asked to report all tools provided by their genetic counselor to assist with disclosure to family, as well as the single, most helpful tool aiding in the disclosure process (Table II). The tool received most often by those who shared results with all identifiable family members was the verbal explanation by the genetic counselor (91.4%), describing risk information to be disseminated to family members, yet only 14.9% reported that it would be the most helpful tool. On the contrary, only 38.3% of participants received a sample letter to distribute to family notifying the recipient that a BRCA1/BRCA2 mutation had been identified in the family, although, this was the tool perceived to be the most helpful by those who received it (22.6%). This was closely followed by written information and/or a brochure about BRCA-related risks which was received by 90.1%, and identified as the most helpful tool by 21.9%.

Table III provides the strategies for disclosure received and the strategy expected to be the most helpful of all strategies by those who shared their BRCA positive results with all identifiable at-risk family members. Encouragement by the genetic counselor for the patient to openly communicate to at-risk family members their pursuit of genetic testing and their willingness to share results, thus interested family members could contact the patient for the test result outcome, was the strategy received most often (70.5%). Approximately 38% of participants were provided a step by step plan for informing family members of results with 41.2% of those who received it expecting it to be the most helpful strategy.
Discussion

Disclosure

This study found that the three main reasons participants shared their BRCA positive information were to inform family members: they were at-risk for having a BRCA mutation, they should consider BRCA testing, and that there is a potential need for increased cancer screenings. Clearly, patients were more concerned about the health of their relatives based on their understanding of the associated cancer risks rather than personal gains they may receive, such as emotional support from relatives and assistance with making medical decisions. Nevertheless nearly half of the study participants failed to disclose results to some relatives they perceived as being at-risk.

Landsbergen, et al. suggested that BRCA carriers should receive follow-up from their genetic counselors to ensure result information is being transmitted to all at-risk family members and to identify specific barriers that may be impeding dissemination (2005). There was no difference in telling family members results based on having received or not received follow-up from their genetic counselor. Therefore, based on the results from this study one cannot recommend counselor follow up as an effective intervention to increase result disclosure to relatives unless the contact is accompanied by other strategies or tools that may increase the effectiveness of the follow up contact.

Even though participants were aware of the importance of disclosure, 49% chose not to tell one or more of their family members their results. This is in agreement with previous findings (Hughes, et al., 2002; Julian-Reynier, et al., 2000; Patenaude, et al., 2006; Wagner Costalas, et al., 2003). It is of interest that of those who did not tell one or more of their family members their results, only 50% responded that their family members wanted to be notified of the results. Although not previously considered a barrier to disclosure, it would seem that perception of relative’s interest in learning of the result may play an important role in curtailing dissemination of result information. If a patient perceives that family members are not interested in or are resistant to receiving result information, perhaps dialogue between the patient and genetic counselor is necessary to determine the basis of this perception. In addition, factors impacted dissemination of results, such as 23% reported their relatives were told by someone else and 22% reported family members were under the age of 18 and therefore, too young to be told. Although it is clear that the intentions of sharing results existed as concern for family members’ cancer risks motivated the majority of patients to test and their genetic counselors explained the importance of sharing results, it
is apparent that specific barriers that inhibit sharing exist and must be addressed during post-result counseling. It is also important that some of the barriers are potentially valid; for example, disclosing results to a family member who has stated that they prefer not to receive them is potentially violating that relative’s autonomy and should be approached with great care.

**Barriers to Disclosure**

This study supports previous findings that family dynamics can create significant barriers to communicating BRCA positive results to family members. Specific barriers found to be most problematic amongst this population were the lack of contact patients had with family members and the physical distance which separated patients from family members. This suggests the importance of equipping patients with BRCA mutations with specific tools and strategies to assist them with the results disclosure process as challenges are encountered.

**Tools and Strategies Provided by Genetic Counselors to Assist with Disclosure**

The sample letter was the tool expected to be the most helpful of all tools provided in assisting with disclosure of BRCA positive results to family member by those who received it. Although these participants were not asked to elaborate on why they expected the sample letter to be so helpful, several potential benefits of its utility with disclosure exist. A sample letter could help remedy many reported barriers encountered by patients with BRCA mutations when disclosing to relatives, such as geographical distance, limited contact, emotional closeness, and previous family conflict, due to its ease of use by mail (or email) rather than having direct contact with each family member. Both the sample letter and written information, which was expected to be the second most helpful tool, can include a detailed explanation of genes, inheritance, risks, resources, and the importance of pursuing genetic testing, information which has previously been described as being difficult for family members to understand. A detailed sample letter and/or written information such as this, would allow for the accurate communication of this information, removing the burden from the patient. Based on participant responses in this study, patients with BRCA mutations prefer tools like the sample letter and written information that can be passed on to family members.
Although the tool received most often was a verbal explanation of BRCA-related risks, only eleven of the seventy-four participants in this population who received a verbal explanation expected it to be the most helpful tool in the disclosure process. Certainly the verbal explanation is an integral part of the result process in this population based on the number of participants who received it; however, data from this study suggests that the verbal explanation cannot be relied upon as a sole means of facilitating communication of risk to family members. Thus, it is recommended that the verbal explanation be provided in conjunction with a supportive tool.

The strategy perceived by patients to be most helpful in the disclosure process was a step by step plan to approach family members for results dissemination. Patients may not know how to proceed with disclosure and would benefit from receiving a detailed plan of how best to utilize tools in the disclosure process, suggesting that simply supplying an informative tool to disseminate to relatives may not be enough. Thus, it is suggested that a step by step plan should accompany an effective tool to maximize the ease for sharing BRCA positive results with those at-risk.

Limitations

The majority of this population received their BRCA results four or more years ago, suggesting possible recall bias (Table I). Although the questionnaire was piloted to several members of the general population, there were structural problems that impacted analysis. Many participants provided more than one answer to questions pertaining to tools and strategies where only one choice should have been selected. Those responses were excluded from analysis, therefore reducing statistical power.

Research Recommendations

Future studies should better define the content of specific tools provided by genetic counselors to assist patients with BRCA mutations in the disclosure process, such as a sample letter. In addition, it is necessary to explore why patients with BRCA mutations expect specific tools to be helpful and to assess their expectations of a step by step plan for results disclosure. Finally, it is important to gain an understanding of why some patients with BRCA mutations perceive that family members do not want to be informed of results and why some patients are met with resistance by family members when attempting to disclose.
Implications for Practice

A combination of tools provided to BRCA positive patients, including a sample letter to distribute to family members and printed literature on BRCA1/BRCA2 testing, in combination with a step by step plan for disclosure is the suggested best approach to increase the likelihood that results will be shared with family members. Thus, it is clinically appropriate for genetic counselors to incorporate such an approach into practice.
Bibliography


Vos, J., Gomez-Garcia, E., Oosterwijk, J., Mendo, F., Stoel, R., van Asperen, C., Jansen, A., Stiggelbout, A., & Tibben, A. (2010). Opening the psychological black box in genetic counseling. The psychological impact of DNA testing is predicted by the counselee's perception, the medical impact by the pathogenic or uninformative BRCA1/2-result. *Psycho-Oncology, DOI:10, 1002/pon.1864.*


<table>
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<th>Characteristics</th>
<th>Told All Family Members</th>
<th>Did Not Tell All Family Members</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>%  (n)</td>
<td>%  (n)</td>
</tr>
<tr>
<td>Participants</td>
<td>51 (90)</td>
<td>49 (87)</td>
</tr>
<tr>
<td>Female</td>
<td>87 (77)</td>
<td>91 (79)</td>
</tr>
<tr>
<td>Age 40 or over</td>
<td>80 (71)</td>
<td>85 (74)</td>
</tr>
<tr>
<td>Married</td>
<td>87 (77)</td>
<td>82 (71)</td>
</tr>
<tr>
<td>Caucasian</td>
<td>99 (87)</td>
<td>98 (84)</td>
</tr>
<tr>
<td>Ashkenazi Jewish Ancestry</td>
<td>14 (10)</td>
<td>13 (10)</td>
</tr>
<tr>
<td>Some college or higher education</td>
<td>83 (71)</td>
<td>90 (76)</td>
</tr>
<tr>
<td>BRCA mutation maternally inherited</td>
<td>64 (47)</td>
<td>70 (46)</td>
</tr>
<tr>
<td>Personal history of cancer at testing</td>
<td>45 (40)</td>
<td>51 (43)</td>
</tr>
<tr>
<td>First in family to test</td>
<td>48 (42)</td>
<td>52 (43)</td>
</tr>
<tr>
<td>4 or more years since received results</td>
<td>58 (51)</td>
<td>51 (43)</td>
</tr>
<tr>
<td>Received genetic counseling from</td>
<td></td>
<td></td>
</tr>
<tr>
<td>CCHMC</td>
<td>80 (70)</td>
<td>82 (69)</td>
</tr>
<tr>
<td>Received genetic counseling from SEHC</td>
<td>20 (17)</td>
<td>18 (15)</td>
</tr>
</tbody>
</table>

Table I The demographic distribution of those who told all identifiable at-risk family members their BRCA positive results and those who did not tell all identifiable at-risk family members their BRCA positive results.
<table>
<thead>
<tr>
<th>Tool</th>
<th>Received (%) of total</th>
<th>Expected Best Tool (%) of total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Written information and/or a brochure about BRCA-related risks</td>
<td>73 (90.1%)</td>
<td>16 (21.9%)</td>
</tr>
<tr>
<td>A sample letter explaining results to distribute</td>
<td>31 (38.3%)</td>
<td>7 (22.6%)</td>
</tr>
<tr>
<td>A video or DVD about BRCA-related risks</td>
<td>3 (3.7%)</td>
<td>0 (0.0%)</td>
</tr>
<tr>
<td>A website that family members could access</td>
<td>13 (16.1%)</td>
<td>2 (15.4%)</td>
</tr>
<tr>
<td>A local support group</td>
<td>41 (50.6%)</td>
<td>0 (0.0%)</td>
</tr>
<tr>
<td>A verbal explanation of BRCA-related risks</td>
<td>74 (91.4%)</td>
<td>11 (14.9%)</td>
</tr>
<tr>
<td>Genetic counselor contact information for family members</td>
<td>44 (54.3%)</td>
<td>2 (4.6%)</td>
</tr>
<tr>
<td>Another BRCA mutation carrier to talk to</td>
<td>1 (1.2%)</td>
<td>0 (0.0%)</td>
</tr>
<tr>
<td>A therapist</td>
<td>1 (1.2%)</td>
<td>0 (0.0%)</td>
</tr>
</tbody>
</table>

Tools for disclosure received and the tool expected to be the most helpful of all tools by those who shared their BRCA positive results with all identifiable at-risk family members.

*As reported by those who shared BRCA positive result information with all available at-risk family members.

*After removing responses "not sure", "other", and if the question was left blank.
<table>
<thead>
<tr>
<th>Strategy</th>
<th>Received (% of total)</th>
<th>Expected Most Helpful (% of total)</th>
</tr>
</thead>
<tbody>
<tr>
<td>A step by step plan for informing family members</td>
<td>17 (38.6%)</td>
<td>7 (41.2%)</td>
</tr>
<tr>
<td>A meeting family members could attend to hear results</td>
<td>12 (27.3%)</td>
<td>3 (25.0%)</td>
</tr>
<tr>
<td>Genetic counselor told family members with consent</td>
<td>9 (20.5%)</td>
<td>0 (0.0%)</td>
</tr>
<tr>
<td>Held a webcast with family members</td>
<td>0 (0.0%)</td>
<td>0 (0.0%)</td>
</tr>
<tr>
<td>Asked patient to repeat back information for understanding</td>
<td>4 (9.1%)</td>
<td>1 (25.0%)</td>
</tr>
<tr>
<td>Encouraged patient to tell family so they could contact patient for results</td>
<td>31 (70.5%)</td>
<td>9 (29.0%)</td>
</tr>
<tr>
<td>A family therapist was provided</td>
<td>0 (0.0%)</td>
<td>0 (0.0%)</td>
</tr>
</tbody>
</table>

Strategies for disclosure received and the strategy expected to be the most helpful of all strategies by those who shared their BRCA positive results with all identifiable at-risk family members.

*As reported by those who shared BRCA positive result information with all available at-risk family members.

*After removing responses "not sure", "other", and if the question was left blank.
Dear Participant:

We would like to learn about your experience in talking to your family members about having a BRCA mutation. Your input will be helpful in evaluating current genetic counseling practices. We are conducting a study to identify tools and strategies that men and women who have a BRCA mutation consider helpful in preparing them for sharing genetic test results and associated cancer risks with family members. You were selected to participate in this study because you have an identified BRCA mutation and you received genetic counseling from Cincinnati Children’s Hospital Medical Center or St. Elizabeth Healthcare.

The decision to participate in this study is completely voluntary. Whether you choose to participate or not will not affect your future care at these, or any other medical centers. Furthermore, all responses are confidential and will not be linked to your name nor will your name be used in any publications as a result of this study. This study has been approved by the Institutional Review Boards of Cincinnati Children’s Hospital Medical Center, St. Elizabeth Healthcare, and the University of Cincinnati, ensuring eligibility for human research.

Your personal experience of having a BRCA mutation and deciding if and how to tell your family members is important in helping us to identify useful materials to distribute to families and to create plans that help BRCA carriers prepare for disclosure of results. Men and women identified in the future as BRCA carriers may benefit from improvements based on your participation in this study. Enclosed you will find a questionnaire that should take approximately 15 minutes to complete. If you consent to participate in this study, simply complete the questionnaire and promptly return it in the enclosed postage paid envelope. Do not include your return address or name on either. If you decide not to participate, no action is necessary. If you have any questions about the study or you would like any BRCA resources, please contact Justine Snyder at the number provided below.

Sincerely,

Justine Snyder, Graduate Student, BA
Primary Investigator
University of Cincinnati
Division of Human Genetics
(513) 636-5839

Jody Wallace, CGC, St. Elizabeth Healthcare
Kristin Theobald, CGC, St. Elizabeth Healthcare
Erin Mundt, CGC, Cincinnati Children’s Hospital Medical Center
This is a questionnaire identifying tools and strategies that patients consider helpful in preparing them for sharing positive BRCA genetic test results and associated cancer risks with their at risk family members.

1. **Before testing, BRCA testing was offered by my (choose one):**
   - [ ] Genetic counselor
   - [ ] Gynecologist
   - [ ] Oncologist
   - [ ] Breast surgeon
   - [ ] Family doctor
   - [ ] Not sure
   - [ ] Other (please specify) _______________________________________________

2. **I received my BRCA genetic counseling from (choose one):**
   - [ ] Cincinnati Children’s Hospital Medical Center
   - [ ] St. Elizabeth Healthcare
   - [ ] Other (please specify) _______________________________________________

3. **Before testing, my genetic counselor explained the importance of sharing my BRCA test results with my family (choose one).**
   - [ ] Yes
   - [ ] No
   - [ ] Not sure
   - [ ] I did not meet with a genetic counselor before BRCA testing

4. **After receiving my genetic test results, my genetic counselor explained the importance of sharing my BRCA test results with my family (choose one):**
   - [ ] Yes
   - [ ] No
   - [ ] Not sure

5. **I chose to have BRCA testing, because (choose all that apply):**
   - [ ] I was concerned about my cancer risk
   - [ ] I was concerned about the cancer risk for my family
   - [ ] I wanted to know if I had a mutation
   - [ ] I was pressured by my family to test
   - [ ] I was encouraged by my physician to test
   - [ ] I wanted to have a better understanding of my medical management
   - [ ] A family member asked me to
   - [ ] Not sure
   - [ ] Other (please specify) _______________________________________________
6. I received my BRCA genetic test results (choose one):

- By phone
- By phone with an in-person follow up appointment
- In person
- By mail
- Not sure
- Other (please specify)__________________________________________

7. Before receiving my BRCA test results, I told family members that I was having BRCA genetic testing (choose one):

- Yes
- No
- Not sure

8. I did not tell some or all of my family members that I was going to have BRCA testing because (choose all that apply)?

- I did not want people to talk me out of testing
- I wanted to wait for the BRCA test results
- I did not want to worry people
- I did not feel it was any of their business
- It was not a good time to have this conversation
- I did tell all of my family members
- Not sure
- Other (please specify)__________________________________________

9. When I told my family member(s) I was going to have BRCA testing the responses I received were (choose all that apply):

- Most were supportive of my decision to test
- Most did not support my decision to test
- Most wanted me to tell them the results when I received them
- Most did not want me to tell them the results when I received them
- I did not tell any of my family members I was having BRCA testing
- Not sure
- Other (please specify)__________________________________________
10. When answering these questions, start at the top of the column and circle appropriate responses as you go down the column. Then go on to the next column.

<table>
<thead>
<tr>
<th>Relative</th>
<th>Total number of this type of relative in your family on the side of family with the BRCA mutation</th>
<th>Number of relatives (from the first column) to whom I told my BRCA test result information</th>
<th>It was difficult for my family member to understand my results</th>
<th>This is how I told my BRCA test result information to my family</th>
<th>Number of relatives who went on to have BRCA testing</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>First Degree Relatives:</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mother</td>
<td>0 1</td>
<td>0 1</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1</td>
</tr>
<tr>
<td>Father</td>
<td>0 1</td>
<td>0 1</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1</td>
</tr>
<tr>
<td>Sister</td>
<td>0 1 2 3 4+</td>
<td>0 1 2 3 4+</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1 2 3 4+</td>
</tr>
<tr>
<td>Brother</td>
<td>0 1 2 3 4+</td>
<td>0 1 2 3 4+</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1 2 3 4+</td>
</tr>
<tr>
<td>Male child</td>
<td>0 1 2 3 4+</td>
<td>0 1 2 3 4+</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1 2 3 4+</td>
</tr>
<tr>
<td>Female child</td>
<td>0 1 2 3 4+</td>
<td>0 1 2 3 4+</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1 2 3 4+</td>
</tr>
<tr>
<td><strong>Second Degree Relatives:</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Aunt</td>
<td>0 1 2 3 4+</td>
<td>0 1 2 3 4+</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1 2 3 4+</td>
</tr>
<tr>
<td>Uncle</td>
<td>0 1 2 3 4+</td>
<td>0 1 2 3 4+</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1 2 3 4+</td>
</tr>
<tr>
<td>Niece</td>
<td>0 1 2 3 4+</td>
<td>0 1 2 3 4+</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1 2 3 4+</td>
</tr>
<tr>
<td>Nephew</td>
<td>0 1 2 3 4+</td>
<td>0 1 2 3 4+</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1 2 3 4+</td>
</tr>
<tr>
<td>Grandmother</td>
<td>0 1</td>
<td>0 1</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1</td>
</tr>
<tr>
<td>Grandfather</td>
<td>0 1</td>
<td>0 1</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1</td>
</tr>
<tr>
<td>Cousin</td>
<td>0 1 2 3 4+</td>
<td>0 1 2 3 4+</td>
<td>Yes</td>
<td>P I M F O</td>
<td>0 1 2 3 4+</td>
</tr>
</tbody>
</table>
11. I received the following materials, or tools, from my genetic counselor to use in helping me inform my family members about the BRCA test result information *(choose all that apply)*:

- Written information and/or a brochure about BRCA-related risks
- Sample letter to distribute to my family explaining BRCA-related risks that had been identified
- Video or DVD about BRCA-related risks
- A website that my family could go to learn about BRCA-related risks
- A local support group for people who are BRCA positive
- I was given a verbal explanation of BRCA-related risks
- Genetic counselor contact information for family members
- Another BRCA carrier for me to talk to
- A therapist for me to talk to
- I did not receive any of these materials or tools
- Not sure
- Other (please specify) ____________________________________________________

12. Of the tools listed in question 11, which one was the most helpful for you in disclosing BRCA result information to your family *(choose one)*:

- Written information and/or brochures about BRCA-related risks
- Sample letter to distribute to my family explaining BRCA-related risks that had been identified
- Video or DVD about BRCA-related risks
- A website that my family could go to learn about BRCA-related risks
- A local support group for people who are BRCA positive
- A verbal explanation of BRCA-related risks
- Genetic counselor contact information for family members
- Another BRCA carrier for me to talk to
- A therapist for me to talk to
- I did not receive any of these materials or tools
- Not sure
- Other (please specify) ____________________________________________________

13. A genetic counselor could have better prepared me for sharing BRCA test result information with my family members by providing the following tools *(choose all that apply)*:

- Written information and/or a brochure about BRCA-related risks
- A sample letter to distribute to my family explaining BRCA-related risks that had been identified
- A DVD about BRCA-related risks to give to my family
- A website that my family could go to learn about BRCA-related risks
- A local support group for people who are BRCA positive
- A verbal explanation of BRCA-related risks
- Genetic counselor contact information for family members
- Another BRCA carrier for me to talk to
- A therapist for me to talk to
14. Of the tools listed in question 13, which one would you expect to be most helpful in disclosing BRCA test result information to your family (choose one):

- The written information and/or a brochure
- The sample letter
- The DVD
- The website
- The local support group
- The verbal explanation
- The genetic counselor contact information
- The BRCA carrier to talk to
- The therapist
- Not sure
- Other (please specify)

15. My genetic counselor provided the following strategies to help me share my BRCA test result information with my at risk family members (choose all that apply):

- A step by step plan for informing my family members
- A meeting that my family members could attend
- My genetic counselor told my family members the risk information for me, with my consent
- Held a webcast with my family members to tell them risk information
- Asked me to repeat BRCA test result information back to my genetic counselor to ensure my understanding
- Encouraged me to tell my family I was testing so they could contact me if they wanted to know my BRCA test result information
- Provided a family therapist
- My genetic counselor did not provide any of these strategies
- Not sure
- Other (please specify)

16. My genetic counselor could have better prepared me for sharing my BRCA test result information with my family members by (choose all that apply):

- Providing a step by step plan for informing my family members
- Holding a meeting that my family members could attend
- Telling my family members the risk information for me, with my consent
- Holding a webcast with my family members to tell them risk information
- Asking me to repeat BRCA test result information back to my genetic counselor to ensure my understanding
- Encouraging me to tell my family I was testing so they could contact me if they wanted to know my BRCA test result information
- Providing a family therapist
17. Of the strategies provided by a genetic counselor which were listed in question 16, which one would you expect to be the most helpful in disclosing BRCA test result information to your family (choose one):

- Providing a step by step plan for informing my family members
- Holding a meeting that my family members could attend
- Telling my family members the risk information for me, with my consent
- Holding a webcast with my family members to tell them risk information
- Asking me to repeat BRCA test result information back to my genetic counselor to ensure my understanding
- Encouraging me to tell my family I was testing so they could contact me if they wanted to know my BRCA test result information
- Providing a family therapist
- Not sure
- Other (please specify)______________________________________________

18. From the time I received my BRCA test result information to the time I shared this information with my family was (choose one):

- 0-2 weeks
- 3-4 weeks
- Over 1 month but less than 6 months
- Over 6 months
- I did not share my test result information because I did not have any family to share with
- I did not share my test result information
- Not sure

19. I told my spouse my BRCA test result information (choose one):

- Yes
- No
- I did not have a spouse

20. Reasons I shared my BRCA test result information with my family (choose all that apply):

- To inform my family members that they were at risk for having a BRCA mutation
- To inform my family members about their specific cancer risks
- To inform my family members about the need for increased cancer screenings
- To inform my family members about cancer risk reducing strategies
- To inform my family members that they should consider BRCA testing
- To gain emotional support from my family
- To get help making medical decisions
- I did not share genetic test result information because I did not have any family to share with
21. I shared my BRCA test result information with every living biological family member on the side of the family my BRCA mutation was inherited from (choose one):

- Yes
- No
- I did not have any family to share BRCA result information with

If you answered yes or that you did not have any family to question 21, you can skip questions #22, #23, #24, #25, and #26

22. I chose not to share my BRCA test result information with one or more of my family members because (choose all that apply):

- Family dynamics (for example: protect family, divorce, distance lived from each other, lack of contact, remarriage, age gaps, under age 18) If selected, answer question 23
- The test result information was too emotional for me (including: guilt, vulnerable, burdened, anxious, angry, afraid, sad worried, jealous, overwhelmed) If selected, answer question 24
- I was concerned about how the test result information would impact family relationships (for example: envy, isolation, broken bonds, conflict) If selected, answer question 25
- I felt that I did not have the skills to share genetic test result information (for example: couldn’t explain the information, didn’t understand the information, didn’t think I could answer questions, didn’t know how to start the conversation) if selected, answer question 26
- I did not know that I needed to tell my family members
- I did not know which family members needed to be told
- I did not think it was any of their business
- Family member(s) were told by someone else
- It was not the right time
- I am not comfortable discussing my personal information with family members
- Not sure
- Other (please specify)__________________________

23. Family dynamics caused me not to tell one or more of my family members about my BRCA test result information (choose all that apply):

- I wanted to protect my family members from bad news
- Divorce(s) in the family
- Distance we lived from each other
- Lack of contact over time
- Remarriage in the family
- Large age gaps within the family
- Family members were under the age of 18
Family dynamics were not an issue for me in disclosing BRCA test result information
Not sure
Other (please specify)___________________________________________

24. It was too emotional for me to tell one or more of my family members the BRCA test result information because the test result information made me feel (choose all that apply):

- My family would think that it was my fault that this BRCA mutation was found
- Guilty that I may have passed the BRCA mutation on to my family members
- Vulnerable
- Burdened with disclosing
- Anxious
- Angry
- I was in denial
- Afraid
- Sad
- Worried
- Jealous because my family did not have an identified BRCA mutation at this time, like I did
- Overwhelmed by decisions I had to make about my own health
- Emotions were not an issue for me in disclosing BRCA test result information
- Not sure
- Other (please specify)___________________________________________

25. I did not tell one or more of my family members the BRCA test result information because I had the following concerns about how the BRCA test result information would impact family relationships (choose all that apply):

- Envy for those among my family that may test negative
- Isolation for those who test positive among my family
- Broken bonds among my family
- Conflict among my family
- Family relationships were not an issue for me in disclosing BRCA test result information
- Not sure
- Other (please specify)____________________________________________

26. I felt I did not have the following skills to communicate the BRCA result information (choose all that apply):

- I did not feel that I could explain this BRCA test result information
- I did not have an understanding of the BRCA test result information
- I did not feel that I had the ability to answer my family’s questions
- I did not know which of my family members were at risk for inheriting the mutation
- I did not know how to start this BRCA test result conversation with my family
- Necessary skills were not an issue for me in disclosing BRCA test result information
- Not sure
- Other (please specify)___________________________________________
27. My genetic counselor followed up with me to see how disclosure to my family was going (choose one):

- Yes
- No
- Not sure

28. If you answered no or not sure to question 27, would you have liked a genetic counselor to follow up to see how disclosure to your family was going (choose one):

- Yes
- No
- Not sure

29. If you answered yes to question 28, when would the follow up from the genetic counselor have been most helpful (choose one):

- 1-2 weeks after receiving results
- 3-4 weeks after receiving results
- 5-6 weeks after receiving results
- Other (please specify)__________________________________________

30. After explaining my BRCA result information to my family members, they had a difficult time understanding (choose all that apply):

- The genetic information
- The inheritance information
- The cancer risk information
- The prophylactic surgical options
- My family members did not have a difficult time understanding any of the information
- I did not share BRCA result information because I did not have any family to share with
- I did not share BRCA result information with my family
- Not sure
- Other (please specify)__________________________________________

31. Gender

- Female
- Male

32. Age

- 18-29
- 30-39
- 40-49
- 50 or over
33. **Marital Status**

- Married
- Single
- Widow
- Divorced

34. **Ethnicity (the side of the family that the BRCA mutation is on)**

- Caucasian
- African-American
- Hispanic
- Asian
- Other (please specify)____________________________

35. **Ashkenazi Jewish**

- Yes
- No
- Not sure

36. **Annual Income Level**

- $29,999 or less
- $30,000-$74,999
- $75,000 or over

37. **Highest Educational Level**

- Less than high school graduate
- High school graduate or GED
- Some college or Associate’s Degree
- Bachelor’s Degree
- Graduate Degree or higher

38. **Insurance at Time of Testing**

- Private individual
- Group
- Medicaid
- Medicare
- Uninsured

39. **My BRCA mutation came from**
☐ My mother’s side of the family
☐ My father’s side of the family
☐ Not sure

40. At the time of BRCA testing, I had a personal history of cancer

☐ Yes (please specify type of cancer(s))____________________________________
☐ No

41. I was the first in my family to have BRCA testing

☐ Yes
☐ No
☐ Not sure

42. Time since receiving my BRCA test results

☐ Less than 1 year
☐ 1-3 years
☐ 4-5 years
☐ Over 5 years

43. My BRCA mutation type is

☐ BRCA1
☐ BRCA2

Thank you for participating in this study by completing this questionnaire. Please return this completed questionnaire in the postage paid envelope provided. Do not include your name or address on the questionnaire or return envelope.