

MOTIVATIONS FOR PURSUING GENETIC COUNSELING TO EXPLORE THE
AVAILABILITY OF PANEL GENETIC TESTING: A PILOT STUDY

By

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ABSTRACT OF THE THESIS

Motivations for Pursuing Genetic Counseling to Explore the Availability of Panel

Genetic Testing: A Pilot Study

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Little is known about the factors that motivate or deter patients from pursuing updated genetic testing after previously receiving negative or uncertain results. The purpose of this study was to investigate which factors influenced patients' decisions to either accept or decline the invitation to return to the genetic counseling clinic to discuss updated genetic testing for breast and ovarian cancer. Four hundred forty-six individuals who had previously tested negative or had a variant of uncertain significance in *BRCA1* or *BRCA2* were mailed a letter informing them of the availability of breast cancer gene panels and inviting them to schedule an appointment with a cancer genetic counselor to discuss updated testing options. Patients who returned were asked to complete a survey about what motivated them to return, and patients who declined were given the opportunity to complete a survey detailing their reasons for declining. The response rate to the letter was 7.83% when we include all patients who returned for testing as well as patients who

responded to the survey for those not returning for updated testing. The surveys on motivations and deterrents required the patients to rate six different factors from 1 (not important) to 5 (very important) in their decision to return or not return for genetic counseling. In addition, patients were able to rank those factors from most important to least important in their decision. Patients were also given the opportunity to write-in other factors that may have influenced their decision. We discovered that the factors that most influenced patients to return were a desire to learn information for their family members and a desire to aid their own health. The main reasons patients declined the invitation were a perceived lack of benefit of updated testing and concerns about the cost of testing. This data provides valuable insight into the factors that motivate and deter patients to consider updated genetic testing and can shape how clinicians inform their patients about expanded genetic testing options.

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Introduction

Genetic testing for hereditary cancer syndromes has evolved rapidly over the past 25 years. Testing for mutations in *BRCA1* and *BRCA2*, which have been implicated in hereditary breast and ovarian cancer, has been commercially available since 1996 (Azvolinsky, 2013). As research has progressed, however, 110 genes have been implicated in increased susceptibility for breast cancer (Baxter et al., 2018). Genetic testing companies now offer multigene panels of genes related to increased breast cancer risk. Prior to the availability of panel testing, genetic testing was typically offered in a stepwise approach. Patients were most often initially offered genetic testing for the *BRCA1* and *BRCA2* genes, with the addition of other genes if initial testing came back negative.

Currently, there are eleven breast cancer genes with well-established clinical management guidelines, and thus it has been recommended that patients who previously tested negative for mutations in *BRCA1* and *BRCA2* consider updated genetic testing (Desmond et al., 2015). Studies have shown that approximately 11% of individuals who have previously tested negative for pathogenic *BRCA1* or *BRCA2* mutations have a pathogenic germline mutation in a breast cancer susceptibility gene identified through multigene panel testing (Yadav et al., 2017). Studies have also found that there is a higher diagnostic yield associated with using a single-tier, multigene panel approach rather than testing patients in a multi-tier approach as previously done (Yorczyk et al., 2015 and Tung et al., 2015). Thus, the clinical utility of updated genetic testing has been demonstrated.

While the clinical utility of multi-gene panel testing has been shown, it can still be difficult for providers to identify which patients need updated testing. The American Society of Breast Surgeons recently recommended updated testing for individuals who had negative *BRCA1/2* analysis in the past (Manahan et al., 2019). Additionally, NCCN guidelines state that multi-gene panels may be more efficient, cost-effective, and increase the yield of detecting pathogenic mutations in at-risk patients (NCCN, 2020). Despite physicians' recognition of these guidelines however, it remains difficult to predict which patients will return for updated testing and strategies need to be developed to inform patients of advancements in genetic testing.

Numerous studies have aimed to address patient motivations and barriers to pursuing *BRCA1/2* analysis. In a previous study regarding factors motivating the decision to undergo *BRCA1/2* testing, participants reported that their primary motivations were wanting information for their children, wanting to take better care of themselves, and gathering information for childbearing decisions (Lerman et al., 1994 and Lerman et al., 1996). Another study reported that a desire to aid cancer research was important to patients as well (Clark et al., 2000). In terms of barriers, numerous factors including concerns over insurability, cost, emotional concerns, time constraints, and confidentiality have been documented (Geer et al., 2001). Motivations and deterrents for genetic testing have not been studied in the context of patients who are considering updated genetic testing.

Several studies have also explored patient preferences regarding updates about improved genetic testing options. When patients were surveyed on their preferences to be re-contacted, Griffen et al. found that patients preferred personalized letters from their

genetics provider if there was information pertinent to them (2007). In addition, when patients were re-contacted regarding the availability of updated testing, the majority were pleased to have received the updated information (Arenas et al., 2018). Despite patients desiring written information, Hampel et al. has highlighted several logistical concerns of this approach, including maintaining updated contact information, maintaining a database of patient testing information, and the time-consuming nature of mailing letters (2009). Nevertheless, other studies have seen up to a 40% response rate from mailing letters to patients (Griffen et al., 2007). In addition to a lack of consensus on how to recontact these patients, there is also a lack of professional guidance in regards to what health-care provider's responsibility to recontact patients is. It is unclear who is responsible for maintaining open lines of communication, what information should be communicated, and how often patients and practitioners should be communicating about testing options (Hampel, 2009). Based on these somewhat discrepant results in previous research, it is important to investigate the utility of sending letters to inform patients about updated testing options.

We identified former patients who previously underwent genetic testing for *BRCA1/2* and received a negative or VUS result, and invited them to return for a discussion regarding updated genetic testing using a multigene panel to assess their risk of hereditary breast and ovarian cancer. Through re-contacting these individuals using an informational letter, we invited them to return to the genetics clinic to discuss updated testing options. We tracked the patient response rate as a result of these letters to determine if sending letters is a reasonable option to communicate updated testing information to patients. In addition, we sought to understand both the barriers and

motivations to patients pursuing updated genetic testing and if any demographic factors served as predictors of patient motivations. We were the first study, to our knowledge, to assess these barriers and motivations in the context of updated genetic testing. Through this work, we hope to inform clinical genetics providers of some considerations that can help aid efforts to inform and motivate patients to consider medically advisable updated genetic testing.

Methods

The primary objective of this study was to determine the factors that motivate and deter patients from returning for genetic counseling to explore the option of updated panel testing for hereditary breast cancer susceptibility genes. The secondary objective was to investigate the utility of sending informational letters to notify patients of updated genetic testing options. The Hereditary Oncology Prevention and Education (HOPE) program at the Rutgers Cancer Institute of New Jersey (RCINJ) offers genetic testing to patients who are identified to be at risk for hereditary cancer syndromes. A list of patients who had genetic testing ordered by RCINJ through Myriad Genetics between the years 2010-2013 was obtained from Myriad Genetics as this was the timeframe prior to the widespread availability of panel genetic testing. The list was then filtered to include only individuals who had *BRCA1/2* testing and received a negative result or a variant of uncertain significance. Any patients who were recently seen for updated genetic testing were removed from the list. Additionally, individuals who had testing for only the three Ashkenazi Jewish founder mutations in *BRCA1/2* were excluded based on the assumption that they likely would not meet NCCN criteria for panel testing since they didn't meet criteria for full *BRCA1/2* testing in the past. The total number of patients left after these filters were applied was 446.

The proposed project was approved by the RCINJ Scientific Review Board and the Rutgers University Health Sciences Institutional Review Board. A total of 446 patients were mailed a letter through the US Postal Service detailing the availability of updated genetic testing and inviting them to call The HOPE Program to schedule a genetic counseling appointment and participate in this study (Appendix A). This letter

was written in a neutral tone so that patient motivations could be assessed without the influence the outside influence of a motivational letter. In this letter, patients were also given the opportunity to complete a survey if they did not wish to return for genetic counseling. A paper with a QR code to access this survey was provided with the letter. The QR code allowed participants to complete the survey digitally through Qualtrics, a secure web-based survey platform.

Patients wishing to have updated genetic testing were invited to call the HOPE program to schedule a genetic counseling appointment. Only those patients seen at RCINJ between October 2019 and January 2020 were included in the analysis for this study. Patients who responded to the study invitation letter after this time period or at affiliated clinical sites were still offered updated genetic counseling and testing but were not included in the analysis in this study. Additional patients who were not recruited through the letter but who previously had *BRCA1/2* testing only and were seen for updated testing during this time frame were also invited to participate in the survey on their motivations to obtain updated genetic testing. At the time of their appointment, informed consent for participation in the study was obtained, and patients were given a paper survey to complete to assess their motivations for returning. In the genetic counseling session, their medical history and family history were updated, genetic testing options were discussed, and a clinical sample for updated testing was collected if the patient consented to proceed.

Two surveys (one for patients who elected to return for updated testing and one for patients who declined) were developed by the investigators based on clinical expertise, review of the literature, and input from the scientific review board (Appendix

B & C). The surveys consisted of demographic information and a list of factors influencing their decisions to either pursue or decline updated testing. Participants were asked to rate each factor independently and then rank the factors in order from most to least important. Participants were also allowed to write in up to four of their own responses.

Data from the survey for those pursuing updated testing was transferred from paper surveys to Excel for analysis. Data from the survey for those who declined genetic counseling was compiled through Qualtrics and then exported to Excel. Data was deidentified by assigning each survey a unique identifier that was stored at the RCINJ. Statistical analysis was performed in SPSS 25 (IBM Corp. Released 2017. IBM SPSS Statistics for Windows, Version 25.0. Armonk, NY: IBM Corp.). Chi Square analysis was used. Statistical significance was considered at $p < 0.05$.

Results

Demographics

A total of 25 individuals participated in the survey to assess motivations for returning for updated genetic testing, all of whom were female. The mean age of the participants was 58 years old. The majority of participants were white (n=21, 84%) and had a college education (n=13, 52%). Additionally, the majority of participants were married (n=19, 76%). Eight individuals identified as Ashkenazi Jewish (32%) and one individual identified as Hispanic (4%). The specifics of the participant demographics are displayed in Table 1.

Age	Responses (n)	Percentage
<50	5	20%
50-60	9	36%
60-70	7	28%
70-80	4	16%
TOTAL	25	Average = 58
Race	Responses (n)	Percentage
White	21	84%
Native American	1	4%
Black	1	4%
Asian	1	4%
Middle Eastern	1	4%
Education Level	Responses (n)	Percentage
Some college	5	20%
College	13	52%
Graduate/Professional	7	28%
Marital Status	Responses (n)	Percentage
Single	1	4%
Married	19	76%
Divorced	4	16%
Widowed	1	4%
Personal Cancer History	Responses (n)	Percentage
Yes	22	88%
No	3	12%

Table 1. Participant Demographics. Participant demographics were recorded to assess age, race, education level, and marital status and are represented as the number of participants in each category as well as percentage of respondents (n=25).

A total of three individuals completed the survey for individuals who were not returning for genetic testing. To protect the confidentiality of these individuals, specific demographic breakdowns will not be provided, however the majority of the participants were white, and the mean age of those who responded was 57.3 years old. Additionally, all three participants had not completed college.

Response Rate

Letters were mailed to 446 individuals who had previously had a negative or a VUS test result on *BRCA1* and *BRCA2* genetic testing. Sixty-three letters were undeliverable due to out of date addresses, leaving 383 letters that were presumed to be delivered to eligible patients. Of these 383 letters, 27 individuals (7.05%) returned for updated genetic testing at RCINJ or one of the affiliate sites during the specified study enrollment period. Since the study was IRB approved only at the RCINJ site, only those seen at RCINJ (20/27) were eligible to complete the survey on motivating factors at the time of their appointment. An additional five participants who did not receive a letter through this study completed the survey on motivating factors. These individuals had previously had *BRCA1/2* testing and were referred to RCINJ for updated testing. Of the 25 individuals who received a survey, all 25 completed the initial task of rating the importance of motivating factors, and 12 completed the ranking task. Three individuals (0.83%) returned surveys detailing their reasons for not returning for updated testing via a Qualtrics-based survey. The breakdown of response rates is detailed in Figure 1.

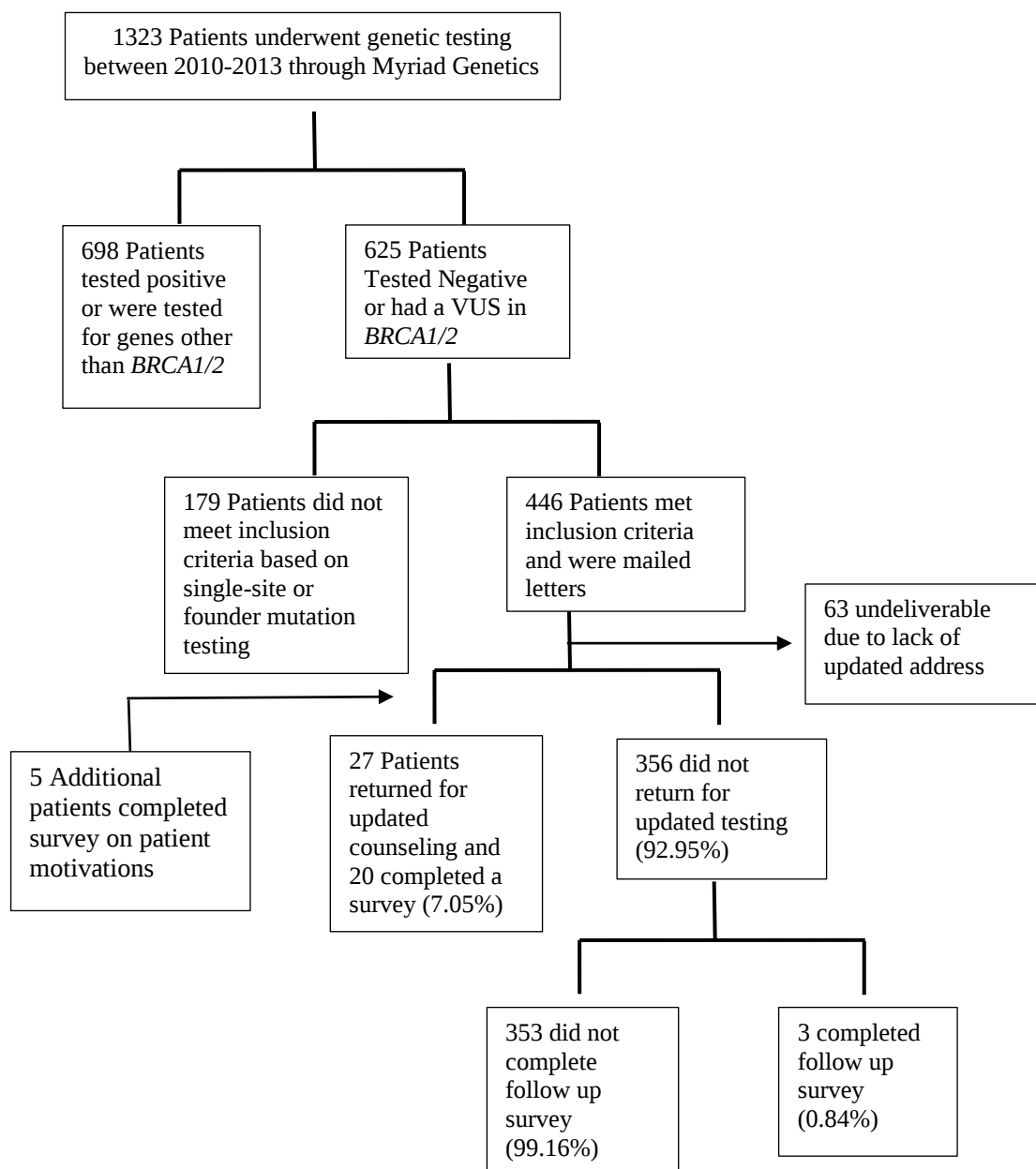


Figure 1. Breakdown of Participants. Over 1000 patients underwent genetic testing through Myraid between 2010-2013. Of these, 446 tested negative or had a VUS in *BRCA1/2* and met testing criteria. Of the 446 letters mailed, 383 were delivered and 27 individuals returned for genetic counseling (7.05%) and three responded regarding the reasons they did not wish to have updated testing (0.84%).

Motivations for Returning for Genetic Counseling

The 25 survey participants were asked to rate six factors that may have influenced their decision to pursue genetic counseling to learn more about updated panel testing. The six factors were (1)*Desire to take care of my health* (health), (2)*Desire to learn risk for my children and/or other family members* (family), (3)*Desire to plan for the future* (future), (4)*Childbearing decisions* (childbearing), (5)*Reduce anxiety/uncertainty* (anxiety), and (6)*My doctor encouraged me to* (doctor). Each factor was rated on a scale from 1 (not important) to 5 (very important). The frequency of ratings for each factor are displayed in Figure 2. All 25 participants rated *Desire to learn risk for my children and/or other family members* as 5, or very important. The other factors had variation within the participant ratings, *however desire to plan for the future* was also rated very highly, with 92% (N=23) of participants rating the factor as 5, or very important. The factor with the lowest importance score was *childbearing decisions* with only 12% rating this factor as very important. Childbearing decisions was rated as significantly less important than all other factors ($p<0.01$).

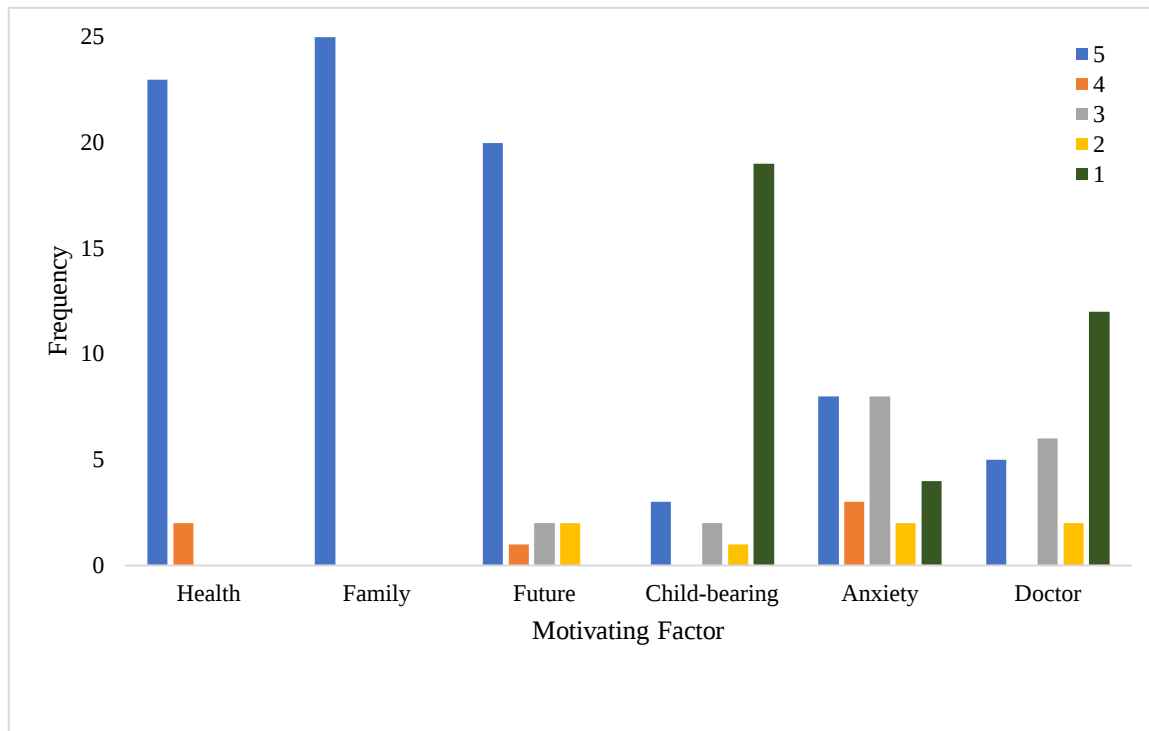


Figure 2. Importance of Motivating Factors. Bars represent the number of patients who rated each factor from 5 (very important) to 1 (not important). N=25

When asked to rank the six given factors in order from most important (1) to least important (6), 12 of the 25 participants completed this task (48%). As shown in Figure 3, the distribution of ranking for each factor varied. 75% of participants ranked *personal health* as first or second most important. 92% ranked *family health* as first or second most important. 17% of participants selected *plan for future* as their first or second choice, and 17% chose *reduce anxiety* as their first or second choice. None of the participants selected *child-bearing decisions* or *doctor's encouragement* as their first or second most important factor that motivated them to return for genetic counseling.

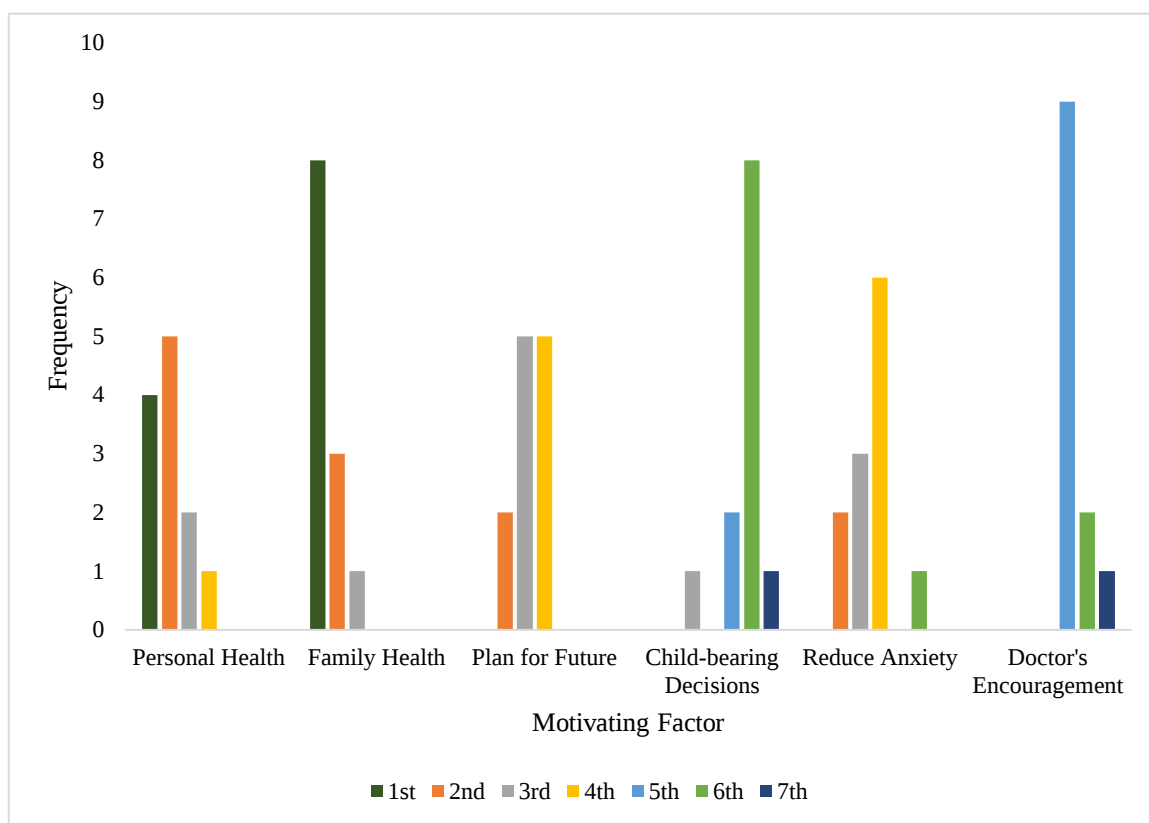


Figure 3. Ranking of factors that motivated participants to pursue genetic counseling to learn more about panel testing. Bars represent the frequency of participants who ranked each factor as 1st-7th. N=12

Additional Motivating Factors

Participants were given the opportunity to write in additional factors that motivated them to return for genetic counseling. Five participants provided responses which were analyzed and grouped based on the theme of the response. Two participants responded that a *desire to aid research* motivated them to return for genetic counseling. Another two participants cited a *desire to help others in the future* as a motivating factor, and one participant cited *family encouragement* as a motivating factor.

Survey Answers by Demographic Factors

Chi square analysis was performed to determine if there was an association between various demographic factors and the rating of the motivations for genetic testing.

Age was not found to be a significant predictor of participant's rating of anxiety as a motivation for pursuing genetic counseling ($\chi^2 = 1.646$, $df=1$, $p = 0.199$). Age was also not found to be a significant predictor patient's desire to plan for the future as a motivation for genetic counseling ($\chi^2 = 1.504$, $df=1$, $p = 0.220$). Age was however a significant predictor of patient's rating of childbearing as a motivation for genetic counseling. Women less than 50 were more likely to rate childbearing as very important than women over the age of 50 ($\chi^2 = 10.795$, $df=1$, $p = 0.001$).

Deterrents to Returning for Updated Genetic Counseling

Participants were asked to rate six factors that may have deterred them from pursuing genetic counseling after receiving the letter detailing the availability of updated panel testing. The six factors were (1)*Concern about insurability of self/family members* (insurance), (2)*Cost* (cost), (3)*Do not think I will benefit* (benefit), (4)*Do not have the time* (time), (5)*Worried about privacy/confidentiality* (privacy), and (6) *Worried about emotional impact of test* (emotional impact). *Cost* and *perceived lack of benefit* were both rated as very important factors with 2/3 of the participants rating them as a four or a five. *Insurance*, *privacy*, and *emotional impact* were all rated of lesser importance with none or one participant rating these as very important.

When asked to rank the six given factors in order from most important (1) to least important (6), 33% chose concerns about *insurance* as the top answer, 33% chose *lack of benefit*, and 33% chose fear of an *emotional response*. For the second most important factor, 33% selected concerns about *insurance*, 33% selected *cost of testing*, and 33% selected *lack of perceived benefit*. For the third most important factor, 33% selected *lack*

of time, 33% selected *concerns over privacy*, and 33% selected an additional factor that they wrote in.

Additional Deterring Factors

Participants were given the opportunity to provide additional factors that influenced their decision to not pursue the invitation for updated genetic testing. These factors were analyzed and grouped relating to themes. Two additional responses were supplied. One factor was categorized as *lack of perceived benefit*, as the individual was receiving appropriate cancer treatment and had no children who she perceived would benefit from her results. The other response was categorized as fear of emotional impact as the participant cited “Don't think I want to know if things are gonna get worse at some point” as her reason for declining the invitation for genetic counseling.

Discussion

The considerations for offering patients updated genetic testing as technologies and our understanding of the genetics of breast cancer evolve is a uniquely complex problem. With the recent change in genetic testing guidelines for breast cancer based on the American Society of Breast Surgeons, there has been much discussion about who should have genetic testing and how the information about genetic testing should be communicated to them. Not only do at-risk patients need to be identified, but they must also be contacted in an effective way which speaks to their individual motivations to undergo genetic testing (Griffin 2007). Through this study, we investigated factors that motivate patients who previously tested negative or had a VUS in *BRCA1/BRCA2* to return for genetic counseling and discuss updated testing options. We also investigated factors that may deter patients from pursuing this opportunity. We found the main factors motivating patients to return were a desire to learn risk for their children and/or other family members and a desire to take care of their health. Some factors that may prevent patients from considering updated testing include concerns about the cost, fear of the impact of results on their insurability, lack of a perceived benefit, a lack of time, concern over their privacy, and a fear of the emotional impact that receiving test results would have on them.

Understanding the reasons that patients do and do not want updated genetic testing is important for creating strategies for re-contacting patients. Previous studies have identified patient motivations for initial testing, however we identified that these motivations differ from motivations for updated testing (Lerman et al 1994). While individuals considering initial testing placed a strong emphasis on reducing their anxiety,

childbearing decisions, and planning for the future, patients who were considering updated testing placed the emphasis more heavily on getting information for their families and for their own personal health. This difference may be related to the ages of patients who are receiving this testing. The patients surveyed through this study were older at the time of re-testing than initial testing, and therefore, their motivations differ and may shift more towards the family that they have already created as opposed to a family that they are planning. Additionally, patients may not feel as strongly that undergoing updated testing will reduce their anxiety because they have already had testing that was unable to explain their personal or family cancer history.

Deterrents to updated genetic testing were similar to the deterrents for patients initial genetic testing, however it is difficult to extrapolate this to the larger population of patients who did not pursue updated testing due to the low survey response rate. For initial testing, patients cited fear of the impact on insurability, the potential cost of testing, lack of benefit, and the emotional impact to be the main factors that deterred them. For updated testing, patients felt similarly, however the most highly rated factor was a lack of perceived benefit, followed by cost and a lack of time.

One consideration regarding why patients may have felt more strongly that they would not benefit from this updated testing is that they had already received genetic testing results that did not alter their management in the past as they were either negative or uncertain. Additionally, their concerns over the cost of testing and the time they would devote to genetic counseling and testing may be rooted in their previous genetic testing experiences in which they may have received a high bill for testing or felt as though genetic counseling was a waste of their time.

Within our participant cohort, those who responded to the survey to detail what deterred them from updated testing were of a lower educational background compared to those who returned for testing. This may have also influenced their responses as individuals with a lower level of education may have understood and been able to recall less information from their initial genetic counseling visit. Based on their educational status, they may have also been from a lower socioeconomic status which could have influenced their concerns over the cost of genetic testing (Aikens and Barbarin, 2008). When re-contacting patients, it may be prudent to highlight examples of how updated testing may benefit the patient despite previous negative results and provide more detailed information on the cost of testing to alleviate these concerns.

Re-contacting Patients

Previous studies have reported implementation of different methods to re-contact patients and provide them with updates about testing availability. Studies have found that patients prefer to be contacted via letters, and therefore, we used letters to contact our eligible patient population (Romero Arenas et al. 2018). The response rate from mailing letters to patients in our study was overall 7.83% when both those who came in for updated testing and those who responded to the secondary survey are considered. This response rate does not reflect all patients who may have been motivated to pursue updated genetic testing after receipt of the informational letter, as patients may have received genetic counseling through a different institution or may have responded after the window of data collection for this study.

The letter that was developed for use in this study was written in a neutral tone as to not influence the responses of study participants. Previous studies have created letters

that provide more information to patients about the benefits of updated testing (Romero Arenas et al. 2018). Through the use of these motivational letters, clinicians saw up to a 40% response rate to informational letters. In the future, studies could compare the response rates when patients receive a letter written in a neutral tone versus a letter that provides more information and is written in a more motivational tone.

However, the low uptake rate seen through this study may suggest that the effort to recontact patients is not proportional to the benefit of this endeavor. Numerous resources are used to recontact patients including time, staff, and money. If sending letters is not an effective way to re-contact patients, then clinics may need to develop alternative methods to inform patients of changes in genetic testing, if they chose to re-contact patients at all. In order to increase the response rate, clinics may consider following letters with phone calls to answer any questions that the patients may have and to help facilitate scheduling the appointments to coordinate the updated testing. Previous studies that originally contacted patients through postal mail and then followed-up with a phone call to patients who did not respond saw a 9% increase in response after the addition of the phone call (Traina et al., 2005). In addition, based on the findings in this study, clinics can focus patient information on the ability of updated testing to provide information that may benefit their family's health as well as their own personal health.

Predictors of Patient Motivations

The data was analyzed to determine if there were any demographic characteristics that could serve as predictors of patient motivators. Within the patients surveyed in this study, we found that younger women were more likely to rate childbearing as very important (4 or 5) than women over the age of 50, suggesting that younger women may

be more motivated by childbearing decisions than older women. This trend is in agreement with the idea that younger women are still considering childbearing, while older women may not consider this anymore. In addition, we investigated if patients with higher education levels may have different factors that motivated them to consider updated testing when compared to those with lower education levels. Again, patient education was not found to be a significant predictor of patient motivations for any of the factors that were investigated. Interestingly, however, all patients who returned for updated testing had received at least some college education, suggesting that individuals who elected to respond had high levels of education, and possibly education level is a predictor of response to informational letters. The lack of differences in patient motivations based on demographic factors, if representative of the larger patient population, may suggest that a similar approach can be taken when re-contacting patients regardless of demographic factors. Future research would be needed to explore this hypothesis given the extremely limited sample size in this study.

Limitations

Several limitations exist within this study. One limitation is that this study was carried out at a single cancer center, and therefore, the responses from participants in this study may not be able to be extrapolated to different populations. In addition, the small sample size in this study was a significant limiting factor. Participants may not have represented the entire population of patients who received the informational letter. Patients who responded to the letter may have self-selected based on prior information on genetic testing, education level, or other factors that led to a non-representative participant pool. Another limitation of this study was the limited information that we

were able to obtain on the patients who did not respond to the letters. It was difficult to draw conclusions about the differences between those who were interested in updated genetic testing and those who were not due to the skewed participation rates in the two arms of the study. The individuals who did not respond to any of the surveys leaves a large proportion of patients whose feelings towards updated testing were not obtained and may signify a demographic that was not represented through this study.

Future Directions

Understanding patient motivations to pursue updated genetic testing will continue to be important as genetic testing evolves, and more patients become eligible for updated testing. Future research may consider designing a study with multiple clinical sites to determine if there are differences in patient motivations based on regions of the country as well as other demographic factors. In addition, to make any conclusions, a larger sample size would be needed to ensure that the results represent the population of patients who are eligible for updated testing. A larger effort to elicit the opinions of those who decline the invitation for updated testing must be undertaken to understand deterrents to updated testing and to determine if there are differences between those that do and do not wish to have updated genetic testing. Capturing the opinions of those who do not wish to undergo updated genetic testing may best be captured by physicians as they see patients and discuss testing options with them. In addition, future studies may utilize different contact methods such as using emails, electronic charts, phone calls, physician recruitment, or newsletters with the goal of increasing patient response rates.

Another important area of research involves the development of materials to use to recontact patients. Future research may investigate the optimal language to use when re-contacting patients about updated testing through focus groups to elicit direct patient feedback. Researchers may consider developing multiple versions of patient educational materials to determine how to best educate patients about updated testing practices.

Recommendations for Clinical Practice

Many barriers exist in re-contacting patients. Clinics who wish to maintain contact with patients regarding updated testing will have to develop systems that allow for efficient patient contact. Importantly, clinics must maintain updated patient databases to allow for re-contact. In the future, clinics may consider web-based applications such as electronic charts or emails to deliver testing updates to patients to avoid the complications of using patient addresses.

While there is still no consensus on if clinics have an obligation to re-contact patients regarding updated testing, it is prudent that each cancer center that offers genetic testing develops their own policies and guidelines on this topic. If clinics decide to re-contact patients, they must ensure that efforts are taken to inform all eligible patients and provide them with the necessary information about new genetic testing options.

Understanding patient motivations can help guide clinics in their development of re-contacting strategies to motivate patients to take advantage of ever-expanding genetic testing options. It is important for patients to maintain their autonomy when deciding if updated testing is right for them, however by creating educational materials that speak to reasons that patients may want additional testing, we may be able to reach more patients and make life-saving diagnoses not only for our patients, but for their families as well.

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Appendix A

Date

Dear Jane A. Doe:

Greetings from The Hereditary Oncology Prevention and Evaluation (HOPE) program at the Rutgers Cancer Institute of New Jersey. You had genetic testing for hereditary breast cancer before 2013 at the Cancer Institute of New Jersey (CINJ). When you were previously tested at CINJ, you were only tested for 2 genes (BRCA1 & BRCA2) that can lead to an increased risk of cancer, as that was the only test that was available. We can now offer testing of over 30 different genes that may impact your cancer risk management. This updated testing can give you more information about your personal cancer risks as well as potential risks to your family members. If you chose to have the updated genetic testing, you will be financially responsible for this testing, however the cost of testing will not exceed \$250. The cost of testing may be covered by some insurance companies.

If you are interested in coming back to CINJ to learn more about the updated genetic testing, you will also be eligible to participate in a research study. The study will look at the factors that motivate patients to return for genetic counseling to discuss updated testing options.

The confidentiality of your medical information is very important to us. Personal identifiers will be used for the study only and not shared. Participation in this study is voluntary and your participation will in no way affect your relationship with CINJ. You may have the newer genetic testing without participating in the study.

To learn more about updated genetic testing options, please call and schedule an appointment at the HOPE Program at 732-235-7110 for genetic counseling. Before your appointment, please review the consent form and survey for the research study which will be provided to you along with other important pre-appointment paperwork. This will be provided after you schedule an appointment for genetic counseling.

If you do not wish to schedule an appointment to discuss updated testing, we ask that you please complete a short survey to help us understand why you do not wish to return for a genetic counseling session. You can access this survey through the QR code or link that are found on the following page. We greatly value your feedback!
Thank you for your consideration.

Sincerely,

Hereditary Oncology Prevention and Evaluation (HOPE) Program
The Cancer Institute of New Jersey
195 Little Albany Street, Suite 1135
New Brunswick, NJ 08903
(732) 235-7110

Appendix B

Age: _____

Race: Please Specify

White

Black or African American

Native American or American Indian

Asian / Pacific Islander

Other: _____

Ethnicity

Hispanic or Latino

Yes

No

Do you have any Jewish ancestry?

Yes

No

Education: What is the highest degree or level of school you have completed?

Less than high school

Some high school

High school

Some college

College

Graduate/Professional degree

Marital Status:

Single

Married/Domestic partnership

Widowed

Divorced

Separated

Have you previously been diagnosed with cancer? If yes, please specify

Yes. (If yes, example Breast Cancer, age 42)

No

Please list any family members who have been diagnosed with cancer, including their relation to you, their diagnosis, and age at diagnosis (Ex: Maternal Aunt- Breast Cancer, 42)

Thank you for your participation. How did you hear about the availability of updated genetic testing? (Select all that apply)

- ☐ Letter from CINJ
- ☐ Information from oncologist
- ☐ Information from primary care physician
- ☐ Information from another health care provider (Please specify): _____
- ☐ Friends or family
- ☐ TV, radio, newspaper, magazine, internet
- ☐ Other (Please specify): _____

Consider each of the following factors which may have influenced your decision to return for genetic counseling. Please circle a number from 1 (not important) to 5 (very important) that describes how important each factor was in your decision to return for genetic counseling. Additionally, in the right-hand column, please rank the following factors in order from most important (1) to least important (10) in your decision to return for genetic counseling. You may write in up to 4 additional factors that influenced your decision:

Factor

Rank (1-10)

Desire to take care of my health	_____
1 2 3 4 5	
Desire to learn risk for my children and/or other family members	_____
1 2 3 4 5	
Desire to plan for the future	_____
1 2 3 4 5	
Childbearing decisions	_____
1 2 3 4 5	
Reduce anxiety/uncertainty	_____
1 2 3 4 5	
My doctor encouraged me to	_____
1 2 3 4 5	

Write in additional factors on the lines provided below:

_____	_____
1 2 3 4 5	

1	2	3	4	5
---	---	---	---	---

1	2	3	4	5
---	---	---	---	---

1	2	3	4	5
---	---	---	---	---

Appendix C

Age: _____

Race: Please Specify

White

Black or African American

Native American or American Indian

Asian / Pacific Islander

Other: _____

Ethnicity

Hispanic/Latino

Yes

No

Do you have any Jewish ancestry?

Yes

No

Education: What is the highest degree or level of school you have completed?

Less than high school

Some high school

High school

Some college

College

Graduate/Professional degree

Marital Status:

Single

Married/Domestic partnership

Widowed

Divorced

Separated

Have you previously been diagnosed with cancer? If yes, please specify

Yes (If yes, example Breast Cancer, Age 42)
No

Please list any family members who have been diagnosed with cancer, including their relation to you, their diagnosis, and age at diagnosis (Ex: Maternal Aunt- Breast Cancer, 42)

Have you previously undergone updated genetic testing?

Yes

No

If yes, please do not complete the following portion of the survey

Consider each of the following factors which may have influenced your decision not to return for genetic counseling. Please circle a number from 1 (not important) to 5 (very important) that describes how important each factor was in your decision to not return for genetic counseling. Additionally, in the right-hand column, please rank the factors in order from most important (1) to least important (10) in your decision to not return for genetic counseling. You may write in up to 4 additional factors that influenced your decision:

Factor
Rank (1-10)

Concern about insurability of self/family members

1 2 3 4 5

Cost

1 2 3 4 5

Do not think I will benefit

1 2 3 4 5

Do not have the time

1 2 3 4 5

Worried about privacy/confidentiality

1 2 3 4 5

Worried about emotional impact of test

1 2 3 4 5

Write in additional factors on the lines provided below:

1 2 3 4 5

1 2 3 4 5

1 2 3 4 5

1 2 3 4 5
