

Telephoned *BRCA1/2* Genetic Test Results: Prevalence, Practice, and Patient Satisfaction

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Abstract While the traditional model of genetic evaluation for breast cancer risk recommended face-to-face disclosure of genetic testing results, *BRCA1/2* testing results are increasingly provided by telephone. The few existing studies on telephone genetic counseling provide conflicting results about its desirability and efficacy. The current study aimed to (1) Estimate the prevalence among genetic counselors of providing *BRCA1/2* genetic test results by phone (2) Assess patient satisfaction with results delivered by telephone versus in-person. A survey was sent to members of the Familial Cancer Risk Counseling Special Interest Group via the NSGC listserv and was completed by 107 individuals. Additionally, 137 patients who had received BRCA genetic testing results either by phone or in-person at UNC Chapel Hill Cancer Genetics Clinic were surveyed regarding satisfaction with the mode of their *BRCA1/2* results delivery. The genetic counseling survey revealed that the majority of responding counselors (92.5%) had delivered *BRCA1/2* genetic test results by telephone. Patients having received results either in person or by phone reported no difference in satisfaction. Most patients chose to receive results by phone and those given a choice of delivery mode reported significantly higher

satisfaction than those who did not have a choice. Those who waited less time to receive results once they knew they were ready also reported higher satisfaction. This study found supportive results for the routine provision of *BRCA1/2* genetic test results by telephone. Results suggest that test results should be delivered as swiftly as possible once available and that offering patients a choice of how to receive results is desirable. These are especially important issues as genetic testing becomes more commonplace in medicine.

Keywords BRCA1 · BRCA2 · Breast cancer · Genetic testing · Results disclosure · Phone counseling · Telemedicine · Telephone

Introduction

According to the American Cancer Society, breast cancer is the most frequently diagnosed cancer in women and the second leading cause of cancer deaths in women after lung cancer (American Cancer Society 2008). Approximately 180,000 new cases of breast cancer are diagnosed each year, with over 40,000 deaths from this disease (National Cancer Institute 2008).

Mutations in the *BRCA1* and *BRCA2* genes result in a substantially increased risk for developing breast and ovarian cancer. The normal gene products act as tumor suppressors and confer risk through mutation of the wild type allele and subsequent loss of function of tumor suppressor activity (Petrucci et al. 2005). Estimates for the penetrance of mutations in *BRCA1* and *BRCA2* have varied, but appear to confer a lifetime risk for women of up to 87% for developing breast cancer and up to 68% for the development of ovarian cancer (National Cancer Institute 2008). The prevalence of *BRCA1/2* mutations in the general

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U.S. population is 1–2 per 1,000, and genetic testing for these mutations has been available to the general public since 1996 (Petrucci et al. 2005).

Regarding *BRCA1/2* mutation testing, the current guidelines recommend testing be performed after provision of pre-test education by a trained genetics professional and formal client consent. The process of informed decision-making may or may not be completed in a single visit and final testing results should be provided in person by the same genetics professional who performed the pre-test counseling (American College of Medical Genetics Foundation 1999). Due to the nature of results disclosure and the fact that post-test counseling is a multi-step process, the NSGC recommends that this process optimally be done face-to-face. Elements included in this session are not only results disclosure, but discussion of impact of test results, medical management decisions, informing other relatives, encouragement of future contact with the clinic, and provision of additional resources and support services, all of which may be difficult to adequately address over the telephone or in a letter (Trepanier et al. 2004). Billing and legal issues may also pose barriers for genetic counseling by telephone (Ormond et al. 2000). For these reasons, current guidelines suggest telephone counseling for test result disclosure only in special circumstances, such as for clients who do not live locally or who are terminally ill. Consequently, most cancer programs in the past have required at least two clinic visits, a pretest visit and results disclosure visit, with some programs requiring additional visits (Schneider 2002).

The recommendations of The American College of Medical Genetics (1999) and NSGC have been influenced greatly by the protocol established for pre-symptomatic Huntington Disease (HD) testing (Baker et al. 1998; International Huntington Association and World Federation of Neurology Research Group on Huntington's Chorea 1994) which recommends three in-person visits with a genetics professional as well as psychological and neurological evaluations. Advocates for modeling familial breast cancer genetic testing protocols on the HD model have noted that genetic testing for both conditions is predictive in nature. However, crucial differences between the two diseases include the fact that preventive measures are available in the realm of breast cancer risk and that the population prevalence of mutations in *BRCA1/2* far exceed that of mutations in the HD gene. Given the increasing frequency of *BRCA1/2* genetic testing in routine clinical care, its established role as an important aspect of oncologic care for many women and little (if any) evidence of harm resulting from such testing, it is reasonable to re-examine the need for in-person results disclosure.

Ideally, telephone counseling can provide a clinical service that is efficacious, convenient, cost-effective,

accessible and educational, while at the same time maximizing the efficiency of clinics so that they can serve a maximal number of patients. The service is becoming increasingly more prevalent for prenatal teratogen counseling and is a standard method for delivering prenatal genetic test results (Ormond et al. 2000). Genetic counselors surveyed in a previous study (Young 1993) indicated that the majority utilized the telephone to discuss and deliver information to patients, including delivery of normal test results, and were motivated to do so primarily by the patient's convenience. However, the majority of counselors in this study also reported never delivering inconclusive or abnormal test results over the telephone. Consequently, there is interest in determining the feasibility of providing high-quality counseling in the context of *BRCA1/2* testing results by telephone.

A number of studies have investigated various facets of telephone delivery of breast cancer risk information and *BRCA1/2* genetic testing results. These studies have previously shown that both in-person and telephone counseling and results delivery have been generally well-received (Helves et al. 2006; Klemp et al. 2005). One observed difference, however, was that patients who received breast cancer risk counseling by telephone found it more difficult to talk about their concerns with one woman reporting that her telephone counseling session was "easier to become distracted than face-to-face". Another difference found was that more women who received telephone counseling would rather have received in-person counseling (Helves et al. 2006). In regard to results delivery, no difference in satisfaction has been shown between those who receive *BRCA1/2* genetic testing results over the phone versus those who receive results in person (Chen et al. 2002; Jenkins et al. 2007). When given a choice as to how to receive results, most individuals choose to receive their *BRCA1/2* genetic testing results by telephone (Klemp et al. 2005). No previous studies have reported the prevalence of telephone usage to provide cancer genetic test results, nor the specific circumstances under which these test results are being provided by telephone. Thus, there is need for current and further investigation of telephone BRCA counseling and results delivery.

As discussed by Peshkin et al. (2008), genetic counseling and testing services are increasing, particularly as additional genes are characterized and additional genetic tests become clinically available. Exploring alternative methods to traditional face-to face genetic counseling is imperative to devise service delivery mechanisms which meet this increase in demand. One of these methods, utilizing the telephone for counseling and/or delivery of results, has been and is currently utilized, but there exists a lack of data about its use and effectiveness from patient and

provider perspectives. In this study we investigated the prevalence of provision of *BRCA1/2* results by telephone among genetic counselors in the U.S as well as patient satisfaction with the mode of results delivery at our institution. The results have broad implications for medical management as the impact of genetics continues to expand in medicine.

Methods

This study consisted of two parts, Current Genetic Counseling Practices and Patient Satisfaction. The study was conducted under the approval of the University of North Carolina Chapel Hill IRB and the University of North Carolina Greensboro IRB.

Part I: Current Genetic Counseling Practices

The first part of the study assessed the prevalence and circumstances accompanying delivery of *BRCA1/2* genetic test results by telephone in current cancer genetic counseling practices throughout the US. Members of the National Society of Genetic Counselors (NSGC) Familial Cancer Risk Counseling Special Interest Group (SIG) ($n=476$) were sent an invitation through their listserv to an electronic survey adapted to Survey Monkey. This group was selected to survey because it represents a large accessible population of practicing cancer genetic counselors.

The survey (see Appendix A) was developed by the researchers as an original measure and piloted to several practicing genetic counselors. The measure included questions to ascertain how many counselors currently deliver *BRCA1/2* genetic test results by telephone, what percentage of their patient population is receiving results in this manner, and the clinical and social circumstances of the patients for whom results are provided by telephone. A list of circumstances under which a counselor may or may not deliver *BRCA1/2* genetic test results over the phone was provided for negative and positive results. The counselor was then asked to answer whether they usually, sometimes, rarely, or never provide results over the telephone for each circumstance (Table 1).

Genetic counselors who responded that they do not deliver any *BRCA1/2* test results to patients were excluded from the study. One hundred seven qualifying individuals responded to the invitation by completing the survey yielding an estimated response rate of 22.5%. The survey was anonymous and submission of the survey was electronic.

The survey was available for 4 weeks, and an electronic reminder letter was issued to the NSGC Cancer listserv approximately 2 weeks after the first letter was issued to increase participant response. All quantitative data were

analyzed and reported using frequencies calculated by the Survey Monkey program.

Part II: Patient Satisfaction

The second part of the study was designed to assess patient satisfaction with the mode of *BRCA1/2* genetic test results delivery. Surveys were mailed to 379 patients who had undergone *BRCA1/2* genetic testing through the University of North Carolina Chapel Hill (UNC-CH) Cancer Genetics Clinic. A subset of patients, those who were African-American, were not invited into this study because they had previously been invited to participate in a concurrent study. The remaining patients were therefore randomized alphabetically by last name to participate in this study. The study sample included patients who did not have a choice as to how to receive their results (mostly patients seen prior to 2005 when the practice of the UNC Chapel Hill Cancer Genetics Clinic was to deliver all results in person) and patients who were provided a choice of telephone or in-person results disclosure (practice that began in 2005).

The patient survey (see Appendix B) was created as an original measure after reviewing several genetic counseling satisfaction surveys posted on the NSGC website (Lea 1996; Middleton and Rodriguez 1997; Rhee-Morris 2006). The measure was used to assess an overall satisfaction score of the results session experience for each individual. Participants were provided with a list of statements pertaining to their results session and asked to strongly disagree, disagree, agree, or strongly agree with each statement using a summative Likert scale to calculate a total satisfaction score. These scores were then divided by the amount of the answered items to produce an average satisfaction score for each individual. These averaged scores were then used to compare satisfaction between those who received their results in-person versus those who received their results over the telephone. Satisfaction scores were also compared between those patients who reported that they had a choice as to how to receive their results versus those who reported not having had a choice. As the goal of our study was to evaluate satisfaction in the context of mode of results delivery and choice of mode of results delivery, for those individuals who may not have recalled how they received their genetic test results or whether they had a choice in their mode of results delivery, the option of "unsure" was provided for these questions to allow these responses to be eliminated in data comparison and analysis.

To test the accuracy of patients' recall of their results, surveys were color-coded depending on the type of result the patient received: positive (55 individuals), negative (307 individuals), or variant of unknown significance (17 individuals). Participants were asked to report their result and responses were checked for accuracy, without identi-

Table 1 Reported Frequency of *BRCA1/2* Test Results Delivery According to Type of Result and Circumstance

	POSITIVE % Respondents (N)					NEGATIVE % Respondents (N)				
	Usually	Sometimes	Rarely	Never	Total N	Usually	Sometimes	Rarely	Never	Total N
When patients are unable to return to clinic for results due to illness or travel issues	27.5% (25)	13.2% (12)	20.9% (19)	38.5% (35)	(91)	34.8% (32)	27.2% (25)	26.1% (24)	12.0% (11)	(92)
When patients refuse to return to clinic for results	16.7% (15)	5.6% (5)	14.4% (13)	63.3% (57)	(90)	20.9% (19)	11.0% (10)	18.7% (17)	49.5% (45)	(91)
When patients choose to have their results delivered by telephone	26.9% (25)	7.5% (7)	15.1% (14)	50.5% (47)	(93)	29.0% (27)	14.0% (13)	17.2% (16)	39.8% (37)	(93)
When results are for the first person being tested in a family	20.2% (19)	10.6% (10)	19.1% (18)	50.0% (47)	(94)	26.9% (25)	11.8% (11)	18.3% (17)	43.0% (40)	(93)
When results are for someone tested from a family with a known familial mutation	21.3% (20)	10.6% (10)	19.1% (18)	48.9% (46)	(94)	25.8% (24)	20.4% (19)	21.5% (20)	32.3% (30)	(93)
When I am confident that the patient comprehended the information discussed during pre-test counseling	24.5% (23)	7.4% (7)	20.2% (19)	47.9% (45)	(93)	30.4% (28)	13.0% (12)	25.0% (23)	31.5% (29)	(92)
When, in my clinical judgment, I believe that my patient is not at risk for an intense emotional response to the result	24.5% (23)	8.5% (8)	14.9% (14)	52.1% (49)	(94)	29.7% (27)	14.3% (13)	22.0% (20)	34.1% (31)	(91)
When I feel confident that I will have an opportunity to follow up with the patient face to face	27.2% (25)	12.0% (11)	19.6% (18)	41.3% (38)	(92)	29.0% (27)	18.3% (17)	26.9% (25)	25.8% (24)	(93)

fying individuals, by matching their response to the color-coded survey they were sent.

Results were analyzed using SPSS. The satisfaction scores were checked for normality using One-Sample Kolmogorov-Smirnov Test and found to lack normality. Therefore, all comparisons of satisfaction scores between groups were made using nonparametric tests such as Kruskal-Wallis Test, Man-Whitney Test, and Chi-Square analyses. Cross-Tabulations and frequencies were used to describe variables of the study population.

Results

Part I: Current Genetic Counseling Practices

Of the estimated 476 genetic counselors belonging to the Cancer Special Interest Group, 107 individuals responded to the invitation by completing the survey for a response rate of approximately 22.5%. Eight of the respondents (7.5%) reported that they had never delivered *BRCA1/2* genetic test results to patients by telephone and were not asked to answer any further questions.

Frequency of Phone Results Disclosure

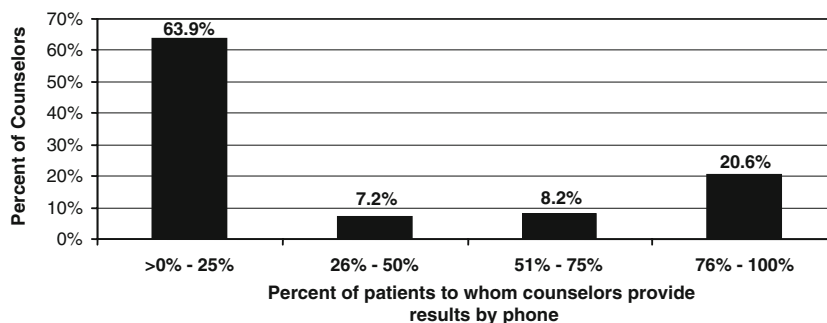
Of the 99 individuals who had delivered results by telephone in the past 12 months, the majority (63.9%) reported that 25% or less of their patient population received their *BRCA1/2* genetic testing results in this manner (Fig. 1).

Participating genetic counselors were asked to estimate how often they provided telephoned results in the past 12 months according to the *type* of result (Fig. 2). Counselors provided phone results “sometimes or usually” for positive, negative, or variant results 33.0%, 55.7% and 33.7% of the time, respectively. While there was no difference between the frequency of phone results among patients with positive or variant results ($p=0.9187$), both positive and variant results were given significantly less often over the telephone than were negative results (Positive vs. Negative: OR=0.39, CI 95% 0.22–0.70, $p=0.0015$; Variant vs. Negative: OR=0.40, CI 95% 0.23–0.73, $p=0.0022$.)

Circumstances Surrounding Phone Disclosure

Genetic counselors were provided with a list of possible circumstances under which one might or might not deliver *BRCA1/2* genetic test results over the telephone and were asked for each circumstance whether they would “usually, sometimes, rarely or never” provide results by phone. In the positive result scenario, the most common preference was to *never* deliver this type of result by telephone, regardless of the circumstance provided (Table 1). From a list of possible circumstances under which a genetic counselor might give negative results, the most commonly chosen answer was also to *never* deliver this type of result over the telephone (Table 1). However, in specific circumstances, such as: “When patients are unable to return to clinic due to illness or travel issues” and “When I feel confident that I will have an opportunity to follow up with the patient face

Fig. 1 Proportion of *BRCA1/2* Tested Population to Whom Genetic Counselors Gave Results by Phone.



to face” the highest percentage of individuals (34.8% and 29.0% respectively) answered that they *usually* would deliver these negative test results over the telephone.

Genetic counselors were also given the opportunity to provide open-ended responses regarding circumstances under which they might deliver positive and/or negative results over the telephone. Respondents’ reasons for providing phone results included: exceptional types of situations, routine practice by their institution, delivery of preliminary results while BART© (an additional testing method offered by Myriad Genetics Laboratory to high risk patients who receive negative results) results are pending, and delivery of BART results.

Part II: Patient Satisfaction

The primary goal of Part II in this study was to determine patient satisfaction at our institution with respect to how their *BRCA1/2* results were delivered. The response rate for patient surveys was 36.1% (137/379). Demographics of responders are shown in Table 2.

Patient Satisfaction by Mode of Results Delivery

In general, most individuals were highly satisfied with the delivery of their results, reporting an average

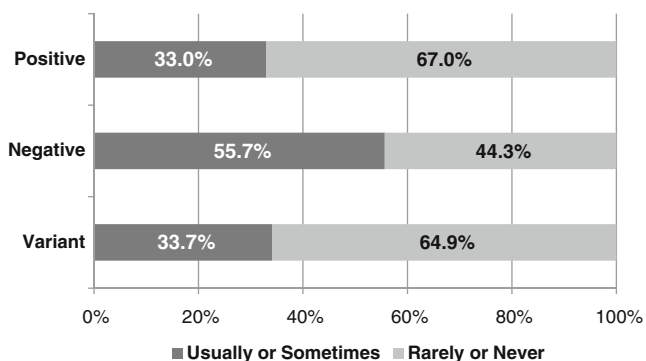


Fig. 2 Frequency of *BRCA1/2* Test Results Delivery by Telephone According to Type of Result.

satisfaction score of 3.6 on a four point scale ($n=135$). Notably, there was no significant difference in satisfaction between patients who received their results over the telephone and those who received their results in clinic (Table 3). No other factors, including the type of result, whether a patient was tested for a familial mutation, or the length of the results session, significantly correlated with patient satisfaction.

We also queried patients as to whether they would have rather received their results in a different way (Table 4). Of the 51 patients who received their results by phone, 50 responded to this question and five of them (10.0%) indicated that they would have preferred to receive their results in person. Of the 82 patients who received results in person, 79 responded and 11 of them (13.9%) reported that they would have preferred to receive their results by phone. There was no statistically significant evidence that a preference for receiving results in a different way was associated with the type of result delivered ($p=0.51$).

Patient Satisfaction by Time Waiting for Results

Patients were also asked to indicate how long it took for them to receive their *BRCA1/2* genetic test results once they knew the results were ready by checking one of several options provided (see Appendix B). The amount of time a patient waited for results once they knew the results were ready was significantly related to their satisfaction ($p<0.001$). Patients who waited 1 month or longer for their results were less satisfied than those who received their results in less time.

Patient Satisfaction Regarding Choice of Mode of Results Delivery

Those patients who had a choice of how to receive their results (those seen after 2005) reported a significantly higher mean rank of satisfaction ($p=.024$) when compared with those patients who reported not having a choice (those seen prior to 2005).

Table 2 Patient Characteristics

Race/Ethnicity	%	N
Caucasian	93.4	128
American Indian	0.7	1
Other	1.5	2
Missing	4.4	6
Age		
20–29	2.2	3
30–39	13.9	19
40–49	21.9	30
50–59	30.7	42
60–69	14.6	20
70–79	5.1	7
80–89	0.7	1
Missing	10.9	15
Gender		
Male	4.4	6
Female	93.4	128
Missing	2.2	3
Education		
Less than high school	0.7	1
High school	5.8	8
Some College/Technical school	20.4	28
College	27.7	38
Some Graduate/Professional school	13.1	18
Graduate/Professional school	28.5	39
Missing	3.6	5
Religion		
Catholic	11.7	16
Jewish	16.8	23
Protestant	61.3	84
Other	3.6	5
None	2.2	3
Missing	4.4	6
Marital Status		
Single	7.3	10
Living w/ partner	2.2	3
Married	75.9	104
Separated	1.5	2
Divorced	8.0	11
Widowed	1.5	2
Missing	3.6	5
When Received Results		
Prior to 2005	24.8	34
After 2005	75.2	103

Lastly, cross tabulations were utilized to assess patient accuracy in recalling the result that was provided to them. Of the 136 individuals who reported a result, 132 (97.1%) accurately recalled their result. Four (2.9%) individuals,

Table 3 Comparison of Satisfaction Between Various Groups

Grouping Variable	Specific Variables	n	Mean Rank Satisfaction (of 100)	
Telephone v. Clinic results (<i>n</i> =131)	Telephone	51	69.8	<i>p</i> =.344
	Clinic	80	63.6	
Reported result (<i>n</i> =129)	Negative	109	64.3	<i>p</i> =.613
	Positive	20	68.8	
Given a choice of how you received results (<i>n</i> =101)	Yes	37	59.3	<i>p</i> =.024
	No	64	46.2	
Tested for familial mutation (<i>n</i> =122)	Yes	34	59.7	<i>p</i> =.716
	No	88	62.2	
Time waited for results once you knew they were ready (<i>n</i> =125)	None	25	78.7	<i>p</i><.001
	1 day	7	80.2	
	1 week	36	73.3	
	1 month	40	44.8	
	> 1 month	17	53.8	
Length of results session (<i>n</i> =133)	15 min	59	68.3	<i>p</i> =.546
	30 min	50	68.2	
	1 h	22	58.6	
	>1 h	2	91.5	

Statistical significance calculated by Mann Whitney & Kruskal Wallis Tests

however, did not accurately recall their provided result. Three of these four individuals had received a result which involved a variant of uncertain significance. Of the four patients who did not accurately recall their result, three had received their results in-person (Table 5).

Discussion

Provision of *BRCA1/2* genetic testing results by telephone is not rare among genetic counselors, but is provided to a minority of patients. The majority of counselors report that they deliver such results by telephone to less than 25% of their patient population. Even in exceptional circumstances,

Table 4 Assessment of Whether Patients Would Have Preferred a Different Mode of Results Delivery

Reported Result N(%)	Received Results By Telephone—Rather In Person? (<i>N</i> =50)		Received Results In Person—Rather Over Phone? (<i>N</i> =79)	
	YES	NO	YES	NO
Negative	5 (10.0%)	36 (72.0%)	9 (11.4%)	55 (69.6%)
Positive	0 (0.0%)	7 (14.0%)	1 (1.3%)	11 (13.9%)
VUS	0 (0.0%)	2 (4.0%)	1 (1.3%)	2 (2.5%)
Total	5 (10.0%)	45 (90.0%)	11 (14.0%)	68 (86.1%)

VUS Variant of unknown significance

Table 5 Scenarios of Four Patient Respondents whose Actual and Reported Results were Discordant

	Actual Result	Reported Result	Mode of Results Delivery
Patient 1	Negative	Positive	In person
Patient 2	Negative	VUS	Telephone
Patient 3	Negative	VUS	In person
Patient 4	VUS	Negative	In person

a large proportion of counselors would still *never* consider delivering these results by telephone. Although counselors are more likely to deliver negative (as opposed to positive) results over the telephone, the majority of counselors would still encourage patients to come into clinic to receive their results in all circumstances.

In our study, patients themselves exhibited no difference in satisfaction when their results were delivered over the telephone versus in clinic. These results are consistent with previous study results (Chen et al. 2002; Jenkins et al. 2007) and suggest that provision of results by telephone can be an acceptable practice in the setting of cancer genetics.

Two other findings were important with respect to patient satisfaction. First, patients who received their results within 1 week of knowing that their results were available were significantly more satisfied than those that had to wait a month or longer for their results. Thus, when dealing with a test like *BRCA1/2* which often takes several weeks to be performed, providing results by telephone may be one way to increase patient satisfaction with the testing and counseling process. Second, our results demonstrate that patients who were given a choice of how they wanted to receive their results were more satisfied with their results session than those who were not given a choice. While the field of genetic counseling has focused on the pros and cons of telephone delivery of results, the current study suggests that perhaps the conversation should shift to discussing whether clinics should offer the option of how to receive one's results instead of prescribing the mode of delivery. Such deference to patient autonomy is fully in keeping with the tradition of genetic counseling in which patient autonomy and choice have been of paramount concern. The results of this study support the notion that it is the autonomy to choose that impacts patient satisfaction, not the specific mode of delivery.

While the number of individuals correctly recalling the result provided to them is encouraging (~97%), it should be noted that a few individuals inaccurately recalled their specific result. However, as previously mentioned, three of these four individuals' results were variants of unknown clinical significance. These variant results may have been reclassified since the time of initial results disclosure or

deemed low enough risk to have been considered negative. Therefore, it is uncertain whether these three individuals actually inaccurately recalled their result. The fourth individual who inaccurately recalled the test results actually received a negative result but recalled having received a positive result. Again, it is unclear from the information we have whether this is an example of incorrect comprehension on the part of the patient or if, for example, the negative was considered to be a false negative. However, future studies aimed at investigating patient comprehension of variants of unknown significance may be warranted. While no firm conclusions can be gleaned from the four instances of apparent misunderstanding of patients regarding their results, it is worth noting that in three of the four cases of incorrect recollection, the patients had received results in-person.

This study found supportive results for the routine provision of *BRCA1/2* genetic test results by telephone, and was designed to assess counselor practice and patient satisfaction with this mode of results delivery. However, there were a number of limitations to the current study. Counselor and patient response rates were not high (22.5% and 36.1% respectively), and therefore cannot be assumed to be entirely representative of these populations. Selection bias cannot be ruled out and the exclusion of African American individuals due to a concurrent study also limits the generalizability of results. Reliance on estimation and recall for both counselors and patients also limits this study since genetic counselors were asked to estimate past and present practices in regards to delivery of *BRCA1/2* genetic test results by telephone. Patients were asked to recall their experiences with receiving their results, which in some cases occurred greater than 3 years ago. However, this specific limitation was addressed in the survey design by giving patients the option to answer "unsure" to many of the questions, allowing those responses to be eliminated in data comparisons and analysis.

Another limitation of this study stems from the fact that some individuals received results before 2005 (those who were not given a choice in their results method) and were compared with a group of later patients who were given an option. Time-based factors could have affected patient satisfaction responses that were not related specifically to the mode of disclosure. However, the stable nature of the clinic and personnel throughout both periods of time should have minimized such concerns.

Conclusion

As genetics increasingly permeates medicine, genetic testing will become commonplace. It is critical that our field continue to explore optimal means of delivering

results with an eye towards maintaining professional standards, accuracy and patient satisfaction, while at the same time seeking maximal efficiency. The results of this study suggest that results should be delivered as swiftly as possible once they are available, and that offering patients a choice of how to receive results is desirable. If future studies confirm that patients are more satisfied with choice, and that there is little difference in patient comprehension and retention of knowledge by telephone versus in person, then the option of results by telephone, perhaps with the option of a follow-up session if desired, should become the standard mode of delivery for most genetic testing results. Such considerations are of great importance as medicine

struggles to incorporate an increasing volume of genetic data into daily practice while maximizing efficiency and appropriately caring for the increasing number of patients receiving genetic testing.

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Appendix A

Counselor Survey:

1. Have you (under any circumstance) ever delivered *BRCA1/2* test results to your patients by telephone?

Yes No I don't deliver *BRCA1/2* test results to patients

If you answered "YES" to the previous question, please continue. Otherwise, you are done with the survey.

2. In the past 12 months, what percent of your *BRCA1/2* tested patient population received their test results by telephone?

>0% - 25%
 26% - 50%
 51% - 75%
 76% - 100%

3. In the past 12 months, how often have you given each of the following *BRCA* genetic testing results by phone?

	Usually	Sometimes	Rarely	Never
POSITIVE				
NEGATIVE				
VARIANT OF UNCERTAIN SIGNIFICANCE				

4a. For each circumstance listed below please indicate how often you have delivered **POSITIVE** BRCA1/2 test results by phone over the past 12 months?

	Usually	Sometimes	Rarely	Never
When patients are unable to return to clinic for results due to illness or travel issues				
When patients refuse to return to clinic for results				
When patients choose to have their results delivered by telephone				
When results are for the first person being tested in a family				
When results are for someone tested from a family with a known familial mutation				
When I am confident that the patient comprehended the information discussed during pre-test counseling				
When, in my clinical judgment, I believe that my patient is not at risk for an intense emotional response to the result				
When I feel confident that I will have an opportunity to follow up with the patient face to face				

In the past 12 months, are there other circumstances under which you have delivered **POSITIVE** BRCA1/2 test results by phone?

4b. For each circumstance listed below please indicate how often you have delivered **NEGATIVE** *BRCA1/2* test results by phone over the past 12 months?

	Usually	Sometimes	Rarely	Never
When patients are unable to return to clinic for results due to illness or travel issues				
When patients refuse to return to clinic for results				
When patients choose to have their results delivered by telephone				
When results are for the first person being tested in a family				
When results are for someone tested from a family with a known familial mutation				
When I am confident that the patient comprehended the information discussed during pre-test counseling.				
When, in my clinical judgment, I believe that my patient is not at risk for an intense emotional response to the result				
When I feel confident that I will have an opportunity to follow up with the patient face to face				

In the past 12 months, are there other circumstances under which you have delivered **NEGATIVE** *BRCA1/2* test results by phone? Please Specify

5a. For each circumstance listed below, please indicate how often you have encouraged people to come in to clinic to receive **POSITIVE** *BRCA1/2* test results in the past 12 months.

	Usually	Sometimes	Rarely	Never
When, in my clinical judgment, I anticipate that my patient is emotionally unable to handle the results				
When results are for the first person being tested in the family				
When results are for someone tested from a family with a known familial mutation				
When I am unsure if the patient comprehended the information discussed during pre-test counseling.				

In the past 12 months, are there other circumstances under which you have encouraged patients to come it to clinic to receive **POSITIVE** BRCA 1/ 2 results? Please specify.

5b. For each circumstance listed below, please indicate how often you have encouraged people to come in to clinic to receive **NEGATIVE** BRCA1/2 test results in the past 12 months.

	Usually	Sometimes	Rarely	Never
When, in my clinical judgment, I anticipate that my patient is emotionally unable to handle the results				
When results are for the first person being tested in the family				
When results are for someone tested from a family with a known familial mutation				
When I am unsure if the patient comprehended the information discussed during pre-test counseling.				

In the past 12 months, are there other circumstances under which you have encouraged patients to come it to clinic to receive **NEGATIVE** BRCA 1/ 2 results? Please specify.

Appendix B

**Telephone vs. In –Person Genetic Testing Results:
Participant Survey**

1. How long has it been since you received your BRCA genetic testing results?

Please choose the closest amount of time.

Less than 1 month *1-6 months* *6 months-1 year* *1-3 years* *More than 3 years*

2. What was your test result?

Positive *Negative* *Variant of Unknown Significance* *Unsure*

3. Were you tested for a mutation that was already found in your family?

Yes *No* *Unsure*

4. How did you receive your test results?

By Telephone *Came in to Clinic* *Unsure*

5. Were you given a choice whether to receive your results over the phone or in clinic?

Yes *No* *Unsure*

6. How long did it take from the time you knew the results were ready to the time when you received the results? Please choose the closest approximate amount of time.

Immediately *Same day* *Within one week* *Within 1 month* *More than 1 month*

7. Whether you received your results by telephone or in clinic, how long was the session during which you received your results?

15 minutes *30 minutes* *1 hour* *More than 1 hour*

Experience with Receiving your BRCA Genetic Test Results

Please circle your response

	Strongly Disagree	Disagree	Agree	Strongly Agree
a. Once I knew my results were ready, I was able to get them in a reasonable amount of time	1	2	3	4
b. An adequate / clear explanation was given with results	1	2	3	4
c. I felt a connection with my counselor	1	2	3	4
d. Results were conveyed in a sensitive manner	1	2	3	4
e. All of my concerns were addressed	1	2	3	4
f. The length of the results session was appropriate	1	2	3	4
g. I was encouraged to call the counselor if I had questions later	1	2	3	4
h. I felt I could talk about my reaction to my test results	1	2	3	4
i. I found it easy to communicate with my genetic counselor	1	2	3	4
j. Getting my results was convenient	1	2	3	4
k. I was satisfied overall with the way my results were delivered	1	2	3	4

Please complete this section only if you received your results by PHONE:

Would you rather have gotten your results in person? Yes No

Did you come in for an in-person follow up session after the phone session?
 Yes No

Where / on what phone did the results session take place?

Home Phone

Work Phone

Cell Phone (please specify where you were, e.g. in the car, at home, etc.) _____

Other (please specify) _____

I Don't Remember

Please complete this section only if you received your results IN CLINIC:

Would you rather have gotten your results over the phone? Yes No

Everyone:

Your Gender: Male Female Your Age: _____

What is the Highest Level of School
You Have Completed?

Less than high school

High School

Some College / Technical School

College

Some Graduate / Professional School

Graduate or Professional School

What is your Religious Background?

Buddhist

Catholic

Hindu

Jewish

Muslim

Protestant

Other (Please Specify) _____

None

What is your ethnicity?

Caucasian

African American

American Indian

Asian

Hispanic

Other (please specify) _____

What is your marital status?

Single

Living with Partner

Married

Widowed

Divorced

Separated

Do you have any children under the age of 12? Yes No

Do you work outside the home? Yes No

If yes, how many hours/week are you employed? _____ (Enter Number)

Please add any comments about your preferences
for receiving your results by telephone or in-person:

Thank you for taking the time to complete this survey!!

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