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Abstract

The purpose of this retrospective study was to examine the process of communicating a positive *BRCA1* or *BRCA2* genetic test result to first, second, and third degree relatives. Participants were 38 women who tested positive for a deleterious mutation through the Cincinnati Children's Hospital Medical Center. Overall, 59% (470/803) of relatives were informed of the test result. The proportion of informed parents, siblings and offspring was nearly twice that of more distant relatives including nieces, nephews, aunts, uncles, grandchildren, and cousins (88% versus 45%). The method of communication differed by the gender of the relative, as did some of the topics discussed. The major barrier to communication was little contact and/or emotionally distant relationships. The study indicates that female mutation carriers act on a perceived duty to inform close relatives of their test result; however, there is a need for genetic counseling strategies that address communication with more distant relatives.

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Table of Contents:

List of Tables and Figures.....2

Introduction.....3

Methods..... 6

Results.....9

Discussion..... 13

References.....21

Appendix – Tables and Figures..... 25

List of Tables and Figures:

Table 1. Characteristics of respondents

Figure 1. Percent of relatives informed of the *BRCA1* or *BRCA2* test result

Figure 2. Percent of relatives informed based on gender and degree of relatedness

Figure 3. Percent of relatives informed by age of respondent

Figure 4. Percent of respondents and the method they used most often to inform male and female relatives of the test result

Table 2. Topics discussed with family members

Figure 5. Motivations for informing family members of the test result

Figure 6. Barriers to informing family members of the test result

Introduction:

Genetic information is rapidly becoming an integral part of clinical management for numerous medical conditions. The current medical model limits the delivery of genetic information to only the individual seeking services, when in reality the information has implications for the entire family. Because of confidentiality, privacy issues, and health care regulations, responsibility for sharing genetic test results with relatives falls on the index patient who may not be prepared or willing to assume this role. Understanding the determinants of communication about genetic test results will assist health care providers in addressing this critical issue.

Previous research studies have examined family communication about a variety of genetic conditions including cystic fibrosis (CF),[1,2,32] fragile X syndrome,[3] hemophilia,[4] balanced translocation,[5] and hereditary cancer syndromes.[6-15] Overall, the studies suggest that patients selectively inform relatives of test results and that patients wish to maintain control over dissemination of information to family members.[1] Studies on CF in particular show that adequate information is not always provided or understood during this communication process.[2] These data suggest that an evaluation of the content of disclosure to family members is also an important research focus.

The bulk of research on family communication with regard to genetic testing has focused on hereditary breast and ovarian cancer syndrome.[6-15,33] This is likely due to the increased availability and utilization of cancer genetic counseling services as well as the complex clinical and psychological issues related to testing for cancer risk.[16-20] Two major breast cancer genes, *BRCA1* and *BRCA2* are thought to be responsible for 5-10% of breast and ovarian cancer cases.[21,22] Mutations in these genes are inherited in an

autosomal dominant manner and confer an inherited predisposition to breast, ovarian, and other cancers.[23-28]

DNA-based testing for *BRCA1* and *BRCA2* cancer-predisposing mutations is available on a clinical basis for individuals at increased risk for hereditary breast and ovarian cancer. The identification of a disease-associated mutation allows for predictive testing in at-risk family members. A primary goal of risk assessment for hereditary cancer is to enhance screening and to increase awareness of options for risk reduction.[29] Choices available to individuals with an identified *BRCA1* or *BRCA2* mutation include surveillance, prophylactic surgery, and chemoprevention.[29,30]

The earliest studies of hereditary breast and ovarian cancer syndrome focused on communication of test results with siblings and children.[7-13] These studies found that the majority of male and female participants informed siblings and children of their test results, whether these results were positive, negative, or inconclusive.[6,8,10,12,13,33] Disclosure of results was highest to sisters and adult children, followed by brothers and minor children, suggesting that female relatives are more likely to be informed than male relatives.[8,10,12,13,33] A major limitation of these prior studies is that the participants were often from cancer family registries where communication about cancer in general could be increased, and multiple family members could be receiving genetic test results at the same time.

A recent study examined the process and content of communication between 43 women who underwent testing for *BRCA1* and *BRCA2* and their 81 sisters.[7] Overall, 85% of sisters were informed of the test result whether it was a positive or inconclusive result (i.e. no mutation was identified). The major motivation for disclosure was to

provide risk information and in the case of mutation carriers, to encourage testing. The major barriers for disclosing the test result were not being close to the sister and not wanting to upset her. For those carriers who disclosed results to sisters, topics most often discussed were the sister's risk of having the mutation, medical information about cancer, and preventive surgery recommendations. The major limitations of this study were that it dealt with a small number of subjects (13 carriers) and was limited to communication with sisters.

More recently, Claes et al. evaluated communication between 63 patients who were tested for *BRCA1* and *BRCA2* mutations and more distant relatives including children, parents, siblings, aunts, uncles, and cousins.[6] Again, the majority of patients informed some or all of their children and siblings of the test result whether it was conclusive or inconclusive. However, communication with distant relatives was more problematic. The major reason for not informing distant relatives was little or superficial contact with the family member. This study was an important first step in expanding the research on family communication about *BRCA1* and *BRCA2* genetic test results; however, these results need to be validated.

The aim of the current study was to provide additional information about family communication of *BRCA1* and *BRCA2* genetic test results. It is the second study to assess communication patterns beyond first degree relatives and is the first to examine communication with nieces, nephews, and grandchildren. This study describes the motivations, barriers, method and content of communication with family members. The current study is important because inadequate communication can lead to inaccurate risk perception for uninformed family members.

Methods:

Study design

A retrospective cohort study design involving women who had previously tested positive for a *BRCA1* or *BRCA2* gene mutation was used to examine each subject's communication with her family regarding the test result. Mutation carriers were identified from the Cincinnati Children's Hospital Medical Center Hereditary Cancer Program (HCP) patient database, which contains information from individuals seen since the establishment of the program in 1996.

Study sample

Eligible female participants had all obtained genetic testing for *BRCA1* and *BRCA2* through the HCP and had been found to carry a deleterious mutation associated with an increased risk for developing cancer. To our knowledge, eligible participants were also the first members of their families to undergo genetic testing for *BRCA1* and *BRCA2*.

A total of 78 women received a positive result for a disease-associated mutation in *BRCA1* and/or *BRCA2* through the HCP. Thirteen were excluded because they were tested after a mutation in the family had previously been identified. One woman was excluded because she assisted with the development of the study questionnaire. Of the remaining 64 women, one was deceased and one could not be located. Thus, the study questionnaire was mailed to 62 eligible subjects.

Procedures (data collection)

The institutional review boards of Cincinnati Children's Hospital Medical Center and the University of Cincinnati approved this study prior to implementation. Eligible subjects were contacted and invited to participate via mail. They received a cover letter explaining the aim of the study and the study's voluntary nature, a consent to participate in the study, and the Family Communication Survey, a four-page questionnaire designed by the authors to evaluate family communication about the subject's genetic test result. Subjects were asked to review the consent form and maintain it for their own records. The consent form and questionnaire were each given a unique study number that was used to prevent duplicate analysis of questionnaires.

All eligible subjects received a mailing in December 2002. A second mailing was sent to all subjects in January 2003. For each survey that was completed and returned, a $5 donation to a regional cancer-related organization was made.

Measures

The self-reported questionnaire consisted of 7 sections with a total of 27 close-ended questions and 2 open-ended questions. The close-ended questions assessed the proportion, identity and gender of relatives who were personally informed of the positive genetic test result, the motivations and barriers for communication to family members, and the method and content of the communication process. Questions eliciting demographic information about the respondent, the number of relatives who had

themselves undergone genetic testing, and which relatives were indirectly informed of the test results were also included.

Some questions from this survey were adapted from a telephone interview designed and utilized previously.[7] Prior to use, the survey was pilot tested by a *BRCA1* mutation carrier.

Data analysis

All data were initially entered into a spread sheet created on Microsoft Excel (2000). Demographic data regarding non-respondents were obtained from the Hereditary Cancer Program database (Microsoft Access 1997) and entered into the investigator database after removal of identifying information. Median household income data were estimated based on zip code and obtained from the 2000 U.S. Census.[31]

Data analysis was performed using the SPSS System for Windows, release 11.0.1. Relationships between categorical variables (nominal and ordinal) were examined by Chi Square analysis. Fisher's exact test was used for this analysis when a 2x2 table contained cells with expected counts less than 5. Independent samples t-test analysis was used for continuous, numeric variables. The general linear model of repeated measures was used to compare the proportion of first, second, and third degree relatives informed by each subject. The nonparametric signed ranks test was used to evaluate Likert scale questions. In all cases, p-values of 0.05 or lower were considered statistically significant.

Results:

Response rate and characteristics

Of the 62 study questionnaires that were mailed, 39 were completed and returned (62.9%). The majority were returned in the first mailing (n=31). Because the second mailing was to all 62 subjects instead of only to non-respondents, it was possible for subjects to return the survey in both mailings. Using the survey identification number, we found that one individual did return two questionnaires. Her second questionnaire was excluded from all analyses.

Respondents (n=38) and non-respondents (n=24) did not differ significantly by race, religion, education, age, time since testing, whether the woman had cancer, or median household income as determined by zip code. The median household income for both respondents and non-respondents ($47,973) was slightly higher than Ohio's median income of $40,956.[31]

Although our intent was to identify study participants who were the first in the family to be tested, this was not true for all respondents. Two sisters who each responded were seen together for initial counseling and genetic testing. They received their positive test results simultaneously and were both included in the analysis. Two respondents indicated that they were not the first family members to be tested. Analyses were done with and without these two subjects and no significant differences were found; therefore, reported data included their surveys.

Characteristics of respondents are summarized in Table 1. The mean age of the respondents was 48.1 years with ages ranging from 23 to 77 years. The average amount

of time that had passed since receiving the test result was 2.4 years with time ranging from 5 months to 6 years.

Relatives informed or not informed of the genetic test result

Thirty-seven of the 38 respondents personally informed at least one at-risk relative of the test result. The one respondent who did not inform any family members was adopted and was not able to locate any biological relatives.

The 37 subjects who shared results with at least one family member reported having 803 living relatives including parents, children, siblings, nieces, nephews, grandchildren, as well as aunts, uncles, and cousins from the affected side of the family. Overall, 470 of the 803 relatives (59%) were informed of the test result.

Sisters were most likely to be informed with 100% of sisters being told (n=63) (see Figure 1). Respondents reported informing 68 of 73 brothers (93%). The 5 brothers not informed were from 3 different families. Respondents reported having 15 mothers, 12 fathers, 40 sons, 47 daughters, 36 aunts, 35 uncles, 82 nieces, 96 nephews, 40 grandchildren, 130 female cousins, and 134 male cousins. On average, each respondent reported having 21 at-risk relatives.

Respondents reported sons, daughters, nieces, nephews, and grandchildren of any age. Fifteen of the 31 sons informed of the test result (48%) were less than 18 years old at the time of disclosure. Of the 35 daughters informed, 12 (34%) were less than 18 years of age when informed.

Aunts, uncles and cousins were reported for only the side of the family at risk for the mutation. Sixty-six percent of respondents reported that the mutation originated on

their maternal side of the family (n=25), 18% reported paternal side (n=7), and 16% were unsure (n=6). If the subject was unsure, relatives from both sides of the family were reported.

First-degree relatives (children, siblings, and parents) were informed of the test results more often than second or third degree relatives (aunts, uncles, nieces, nephews, grandchildren, and cousins) (88% versus 45%; p=0.02). The difference between informed males and females in each category was not significant (see Figure 2).

Seventeen of the 37 respondents (46%) informed less than 50% of their relatives of the test result. Those that informed less than 50% of relatives did not differ from those that informed greater than 50% of relatives by age, race, income, marital status, years since testing, whether they were affected with cancer, or *BRCA* mutation type. There was an inverse relationship between the subject's education level and the proportion of relatives informed. Participants who were educated beyond college were less likely to inform a majority of relatives. Religion was also found to differ between the two groups. Catholics were more likely to inform a majority of relatives as compared to other religious groups (p=0.037).

Of the 37 respondents who informed at least one relative of the test result, 49% were less than 50 years old (n=18), and 51% were 50 years or older (n=19). The younger women had fewer sons, daughters, nephews, and grandchildren than the older women. Older women were significantly more likely to inform their daughters, sons, and nieces of the test result (see Figure 3).

Method and content of communication to family members

Methods used most for communicating the test result to male and female relatives are summarized in Figure 4. Nine percent of respondents used another method to inform male relatives such as indirectly informing the male relative via another family member (see Figure 4).

Respondents reported that many topics were discussed with relatives who were informed of the test result (see Table 2). Topics most often discussed with both male and female relatives included the family history of cancer, the reasons for testing, and the risk for that relative to also have the mutation. Respondents were more likely to discuss surgery guidelines, feelings about the test result, and insurance discrimination with female relatives. The number of responses to these questions ranged from 10 to 34 because some respondents did not inform certain relatives of the result, and some left portions of the question blank.

Motivations and barriers for communication to family members

The most important reasons for discussing the test result with family members were: 1) to inform them of their risk, 2) to fulfill a duty to inform, and 3) to suggest testing (see Figure 5). Advice on treatment, emotional support, and suggesting testing were significantly less important when communicating test results to male relatives as compared to female relatives. The number of responses ranged from 28 to 37 for this question.

The major barriers to communicating the test result were not being close to and not being in contact with relatives (see Figure 6). There were no significant differences

based on the genders of the relatives not informed. The number of responses ranged from 10 to 15 for this question.

Relatives who were indirectly informed of the test result

Sixty-one percent of respondents said that they did not directly inform some relatives of the test result because they depended on another family member to do it. Of the 22 who stated this, 8 specified that they let their siblings decide about informing nieces and nephews. Six respondents specified that they depended upon their mothers to inform brothers, aunts, and/or uncles of the test result. One woman said that her female cousins informed male cousins. 7 did not elaborate on their response.

Number of relatives who were tested

On average, respondents reported that 2.1 female relatives and 0.6 male relatives were tested for the familial *BRCA* mutation. Overall, 37% of the informed female relatives (n=209) and 11% of the informed male relatives (n=219) underwent genetic testing after learning of the result. Some of the informed relatives were less than 18 years of age so testing would not be offered to them.

Discussion:

An initial genetic counseling session inevitably involves taking a family history so that the collection and discussion of medical information immediately extends to individuals beyond the patient. A positive genetic test result implicates family members even further because they are at risk for having the same mutation. Family

communication about hereditary cancer is especially important because it may determine whether or not family members take steps to learn more about their cancer risks and ultimately make decisions about cancer detection and prevention.

This study examined the communication patterns of thirty-eight women who tested positive for a deleterious mutation in the *BRCA1* or *BRCA2* gene. The goal was to describe communication of the positive test result to first, second, and third degree relatives. This included the method, content, motivations, and barriers of communication. Participants communicated their test result to a large majority of first degree relatives, but significantly fewer second and third degree relatives were informed.

It was not surprising that so many first degree relatives were informed of the test result because these relatives have a 50% risk of also having the mutation. Cousins are at lower risk than second degree relatives (12.5% versus 25%), but on average, equal numbers of cousins and second degree relatives were informed. In many families, cousins are similar in age and may be informed because participants feel close to them.

The proportion of informed first degree relatives was higher than what has been reported in other studies, and the difference in gender was not as large as reported in other studies where females are significantly more likely to be informed.[6,10,12,33] This may be because we only included women with a true positive *BRCA1* or *BRCA2* test result, whereas other studies have included individuals with indeterminate or inconclusive results.[6-13,33] It has been shown that patients who test positive are more likely to share their result than those who test negative or are found to have a variant of unknown clinical significance.[6,7,33]

Participants with less education and a religious affiliation that was Catholic were more likely to inform family members of the test result. First, it is important to recognize that the small numbers in this study prevent broad generalizations about these findings. Second, statistical significance is not equivalent to clinical significance. Lastly, the possibility for confounding factors cannot be eliminated. It is possible that these factors appeared to be significant but were actually related to other undescribed variables.

Study participants discussed some different topics with male versus female relatives and used different methods to inform them as well. Overall, participants showed good judgment as to which topics were most important to discuss with relatives. It was not surprising that respondents discussed insurance discrimination and preventive surgery guidelines with fewer males than females, as these issues are less relevant to men. Feelings about the test result were discussed with female relatives more often than male relatives. As seen in other studies, communication with close, female family members may be a strategy used to cope with the positive test result.[7,37]

Most subjects informed female relatives of the test result in person, while male relatives were informed over the phone, in person, or by other means of communication. Participants wanted more emotional support from female relatives; therefore, the difference in communication methods may indicate that they were most comfortable discussing their feelings about the result face-to-face. The findings may also suggest a greater sense of importance or urgency when informing females of the mutation. To further address this issue, it would be useful to identify the amount of time that passed between receiving the test result and informing male versus female relatives.

The major reason for informing relatives of the test result was to notify them of their risk. Most women in our study had been diagnosed with cancer, and some underwent genetic testing to aid in treatment decisions. However, when a woman has already had cancer and has been treated surgically, the test result may not have a major impact on medical management. Therefore, some women undergo testing in order to clarify cancer risk for other family members.[36] It was not surprising that participants felt a strong duty to inform relatives of this increased risk since some were tested primarily for this reason.

A theme that emerged from our open-ended questions was that some participants found no difficulty with the communication process since for many of them, the test result was not unexpected. For example: "*...I was not shocked by the positive test result I received for BRCA1, therefore, it was not difficult to inform my relatives, this testing was more for confirmation of what we suspected was a genetic mutation.*"

However, there were barriers to informing relatives of a positive test result with the major one being a physically or emotionally distant relationship. One respondent noted, "*...There is a big difference between telling your children and your extended family. I am very open with my children, they know what is going on and stay informed. I am not close with my extended family. They know their own cancer history.*" Not wanting to upset the relative, not knowing what to say, and having difficulty coping were not identified as major barriers, but some participants did deal with these issues when informing certain relatives. For example: "*The most difficult part was telling my brothers, since we had already lost my mother to breast cancer.*" and "*Since I was the*

first to take the test, I started feeling like I was responsible for the gene and was apologetic to my family..."

Previous studies confirm that communication of a *BRCA1* or *BRCA2* test result can be hindered by a superficial relationship or lack of contact with a relative.[6,7,14] This is a legitimate barrier, and it may be difficult for health care providers to develop strategies to help patients overcome it. Nonetheless, professional policy suggests that providers may have an obligation to the at-risk relatives of the patients they see.[35] This obligation could be fulfilled by not only emphasizing to patients the importance of sharing the information, but also helping them to develop strategies for effective communication, despite the family rifts that may exist.

Further research should focus on how health care providers can develop more successful strategies for promoting family communication. Different approaches will likely be required for different family members, and providers might be better able to assist patients if they understand the established communication patterns and relationships in the family.[38] Communication about a genetic test result is selective rather than universal. Pre-test counseling could focus on family structure so that problem areas can be predicted. For example, if the patient has not told family members about her personal cancer diagnosis, it is unlikely that she will inform them of a hereditary cancer syndrome.[6] Once a problem area is recognized, the patient and professional can develop strategies such as providing literature for relatives or making plans to inform relatives indirectly via other family members.

Another important direction for future research is to evaluate the process of communication from the relative's perspective and to determine how the relative utilizes

this information. In our study, participants reported that a minority of informed relatives underwent genetic testing, and some felt frustrated when their relatives did not act on the information provided to them. Respondents wrote, "*...Some [of my siblings] hesitated to get testing and that was hard for me – I wanted them to be proactive.*" and "*My sisters and my female cousin have all chosen to ignore the situation. One sister doesn't want to know anything. Other sisters have taken no action. One has gotten [breast] implants!*"

A final suggestion for future research is to determine the best way to inform and counsel children and young adults about increased cancer risk. Older women in our study were more likely to inform children, grandchildren, nieces, and nephews of the test result because older women, on average, have older children in their families. Nonetheless, we know that some children informed of the test result were less than 18 years of age at the time of disclosure. This is interesting because there is no increased risk for childhood malignancy in individuals with a *BRCA1* or *BRCA2* mutation, nor is there an imminent medical benefit such as increased screening or behaviors to reduce cancer risk.

A recent study found that the majority of breast cancer survivors felt that children should be provided with personal cancer risk information between the ages of 13 and 18 years.[34] The motivation for choosing these ages is not clear and could conflict with policy statements and the clinical practice of postponing genetic testing for adult-onset disorders until adulthood.[35] It will be important to understand the psychological impact of providing genetic risk information to children and young adults, whether this impact is a positive or negative one.

There were limitations to this study that must be recognized. First, there was the potential for response bias in that non-respondents may have informed fewer relatives. Second, the study was done retrospectively which means there was potential for recall bias. Third, the small number of participants, although similar to numbers used in other studies,[6,7,9] may have prevented us from detecting important predictors of family communication. Fourth, the content, motivations, and barriers of communication were reported by gender of the relative rather than for each relative individually. This generalization may have concealed differences that were based on the identity of the relative. For example, the desire to obtain emotional support may be higher when informing sisters versus female cousins, but this would not be reflected in our data.

The homogeneity of the study population may cause some to question the generalizability of the study results. In the year 2000, only 20% of households in Ohio reported a yearly earning of $75,000 or more,[31] yet the majority of respondents in this study reported this household income range. In the same year, 55% of Ohio residents were married, 47% were educated at the college level or beyond, and 85% were Caucasian.[31] Although our study participants may not reflect the general population, they are typical for women who receive genetic counseling for hereditary breast and ovarian cancer through our Hereditary Cancer Program.[39]

In conclusion, the present study of family communication about positive *BRCA1* or *BRCA2* genetic test results provides an overview that is more extensive than other studies done on this topic. Female *BRCA1* or *BRCA2* mutation carriers informed most siblings, offspring, and parents of the test result; however, communication of the result to distant relatives including aunts, uncles, nieces, nephews, grandchildren, and cousins was

more problematic. It is essential that family structure and established communication patterns be explored prior to testing so that strategies to overcome communication barriers are more successful after testing. Although many professionals and patients believe that confidentiality should be the highest priority, current case law is not completely consistent with respect to "duty to warn" issues.[35,40,41] The ethical and legal dilemma of breaching patient confidentiality to warn relatives at high risk of inherited diseases can be avoided if professionals succeed at helping patients overcome communication barriers.

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Appendix:

Table 1. Characteristics of respondents (n=38)

Characteristic	Category	%	n
Race	Caucasian	97	37
	Native American	3	1
Marital status	Married	92	35
	Divorced or separated	5	2
	Single	3	1
Highest education level	Elementary school	3	1
	High school	10	4
	Some college	24	9
	College degree	37	14
	Graduate or beyond	18	7
	No response	8	3
Annual household income	$\leq$ \$25,000	8	3
	> \$25,000 but < \$75,000	29	11
	$\geq$ \$75,000	61	23
	No response	3	1
Religion	Catholic	47	18
	Jewish	18	7
	Protestant	29	11
	Other	3	1
	No response	3	1
Affected with cancer	Yes	79	30
	No	21	8
Genetic mutation	*BRCA1*	76	29
	BRCA2	18	7
	Both *BRCA1* and *BRCA2*	5	2

Figure 1. Percent of relatives informed of the *BRCA1* or *BRCA2* test result

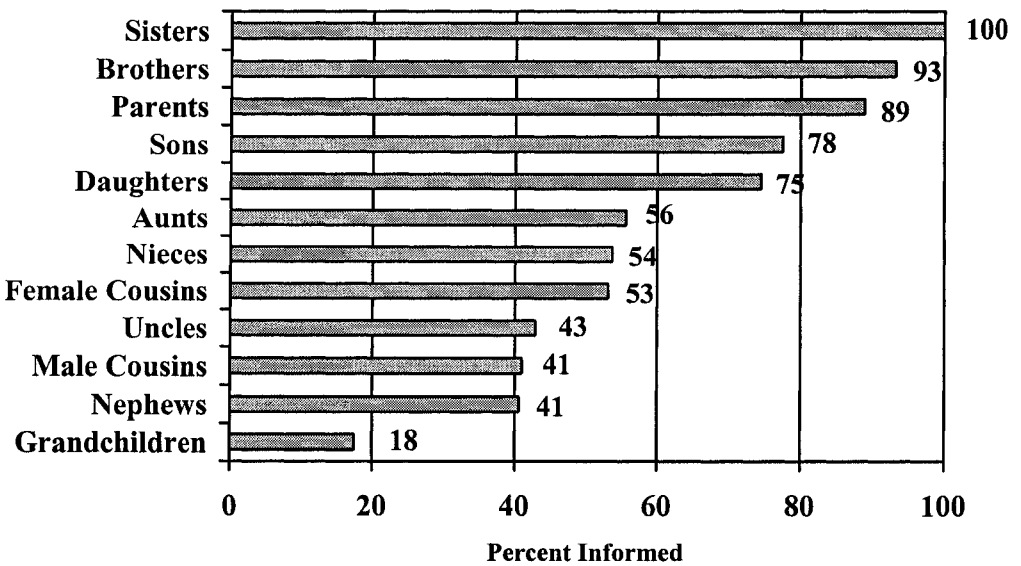
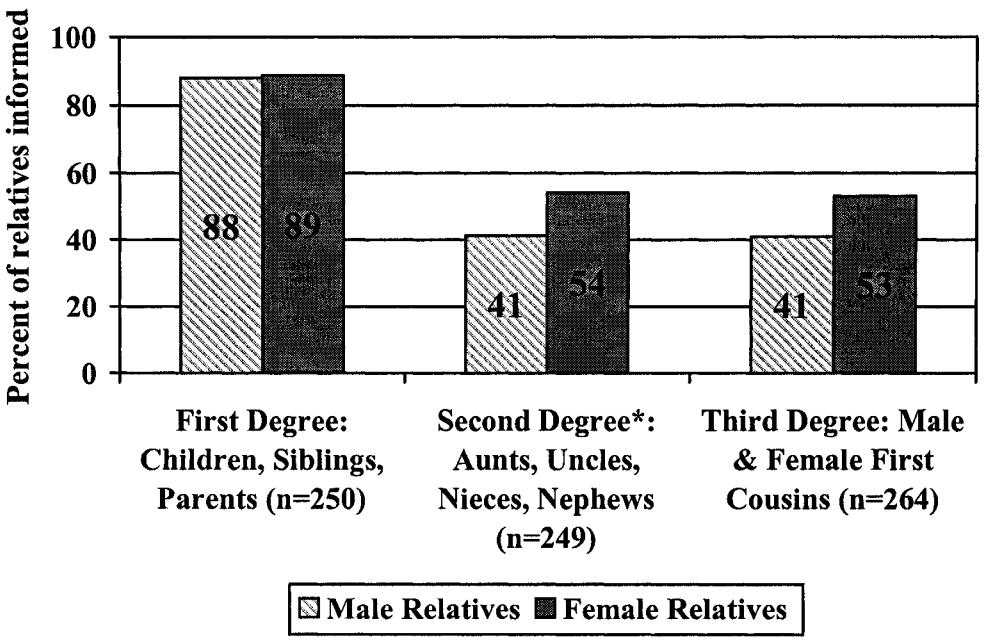
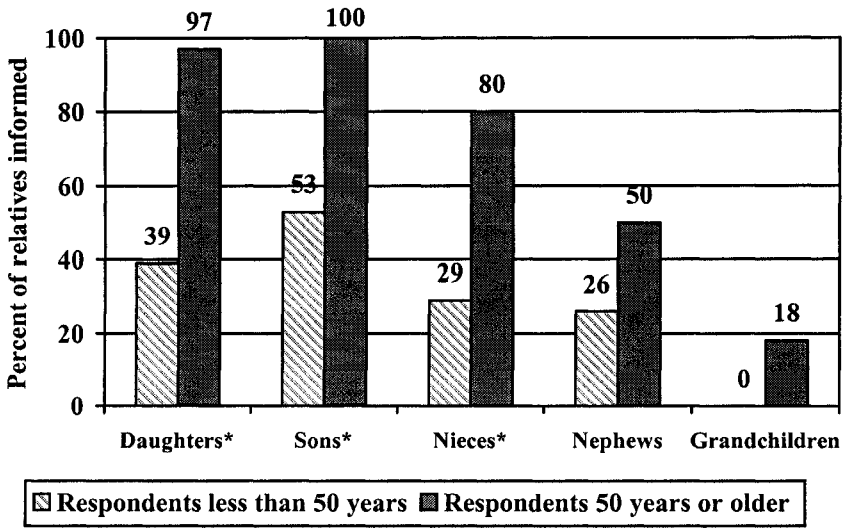


Figure 2. Percent of relatives informed based on gender and degree of relatedness



* Grandchildren are excluded from Figure 2 because the genders of the grandchildren were not elicited

Figure 3. Percent of relatives informed by age of respondent



* Indicates statistical significance between groups at p=0.05

Figure 4. Percent of respondents and the method they used most often to inform male and female relatives of the test result

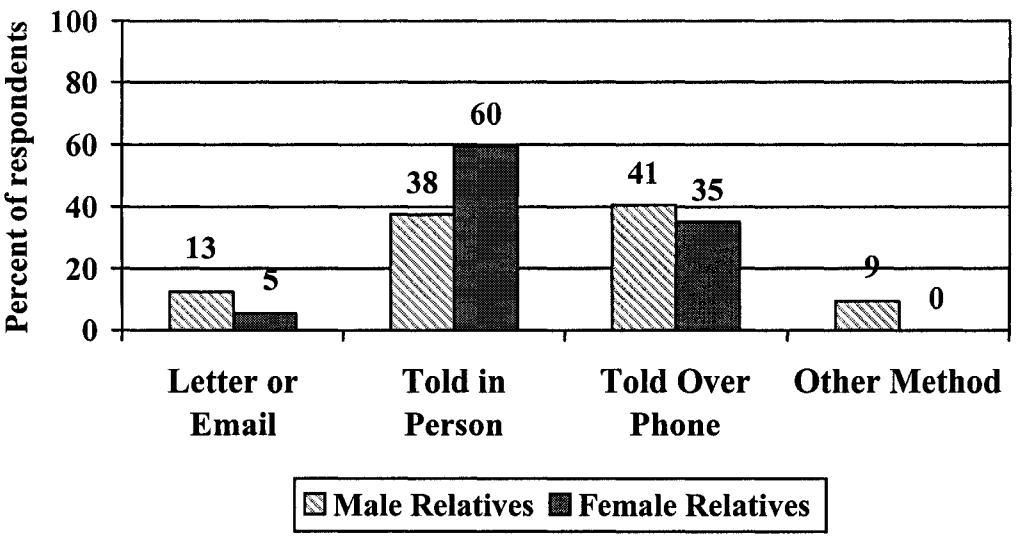
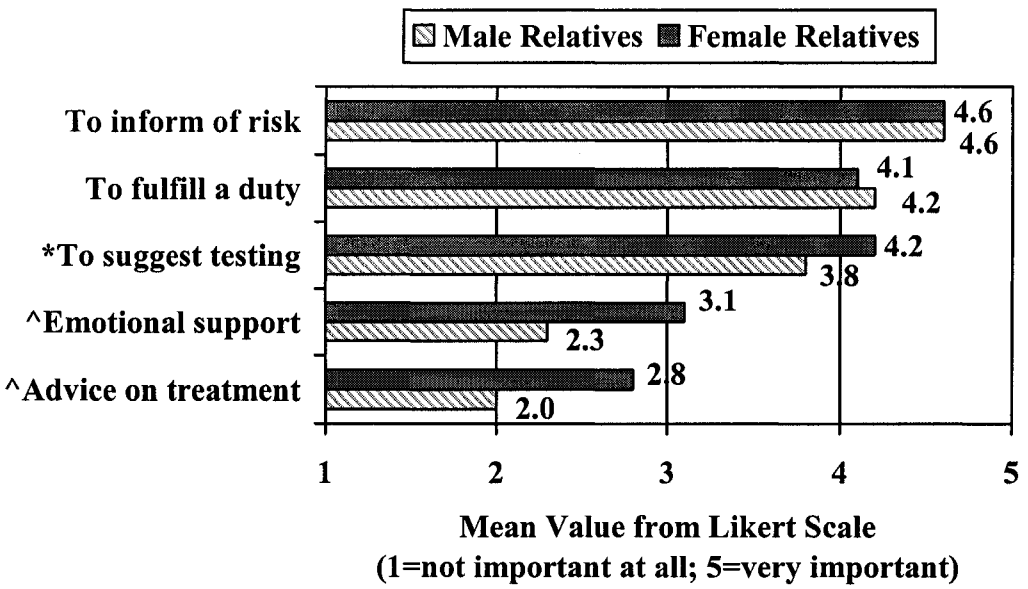


Table 2. Topics discussed with family members

Topic discussed	Percent who discussed topic with MALE relatives	Percent who discussed topic with FEMALE relatives	p-value
Surgery guidelines	40	78	0.003
Feelings about result	62	89	0.011
Insurance discrimination	68	89	0.022
Screening guidelines	67	81	0.083
Cost of testing	68	80	0.083
Family history of cancer	90	97	0.161
Reasons why tested	87	94	0.161
Risk of a mutation	83	91	0.326

Figure 5. Motivations for informing family members of the test result



* Indicates statistical significance at p=0.05
^ Indicates statistical significance at p=0.01

Figure 6. Barriers to informing family members of the test result