Published in final edited form as:

Genet Test. 2005 ; 9(3): 261–268. doi:10.1089/gte.2005.9.261.

Hereditary Nonpolyposis Colorectal Cancer Family Members' Perceptions about the Duty to Inform and Health Professionals' Role in Disseminating Genetic Information

Rebecca D. Pentz¹, Susan K. Peterson², Beatty Watts², Sally W. Vernon³, Patrick M. Lynch², Laura M. Koehly⁴, and Ellen R. Gritz²

¹Winship Cancer Institute, Emory University, Atlanta, Georgia ²The University of Texas M.D. Anderson Cancer Center, Houston, Texas ³The University of Texas Health Science Center at Houston School of Public Health, Houston, Texas ⁴National Human Genome Research Institute, NIH, Bethesda, Maryland

Abstract

This study's aim was to ascertain hereditary nonpolyposis colorectal cancer (HNPCC) families' views on the duty to inform with particular focus on the role of health professionals in disseminating familial genetic information. Eighty members of 16 families with a clinical or molecular diagnosis of HNPCC completed qualitative interviews regarding views on family members' right to know and who should disseminate familial genetic information. Most indicated that everyone in the family should know about the presence of a mutation in the family, with family members themselves being the preferable informant, supported by health professionals who were seen as helpful in overcoming barriers. All but one respondent indicated that if a parent did not test and presumably did not inform his/her child about the family mutation, the child should be informed by other family members or by a health professional. Many were attuned to confidentiality concerns, but judged them to be outweighed by the importance of family members knowing about the mutation and undertaking proper surveillance. Respondents were more private about the disclosure of individual results to other family members, clearly distinguishing personal results from familial genetic information. These families with a hereditary colon cancer syndrome favor open sharing of genetic information within the family, and desire the supportive involvement of health care professionals in disseminating genetic information.

Introduction

The Health Professional stands at the nexus of policies regarding the dissemination of information about inherited risks for developing diseases, such as cancer, within families.

© Mary Ann Liebert, Inc.

Address reprint requests to: Rebecca D. Pentz, Ph.D., Winship Cancer Institute, Emory University, 1365 C. Clifton Road, N.E., Room C 3008, Atlanta, GA 30322, Rebecca.Pentz@emoryhealthcare.org.

Preliminary findings from this work were reported in poster form: 2003 American Society of Clinical Oncology Annual Meeting, Chicago IL: Pentz RD, Peterson SK, Watts B, Vernon S, Lynch P, Gritz ER. Hereditary nonpolyposis colorectal cancer family members' perceptions of the duty to disclose family mutations. May 2003. Abstract 3431.

Tension exists between the role as caregiver to an individual patient and the duty to warn others in a patient's family of potential harm. Governmental agencies, professional organizations, and the courts offer decisions and policies about this duty to warn and the role of the health care professional in disseminating information, yet little has been heard from individuals and families affected by a genetic susceptibility to disease. Interview research, such as that reported here, provides a forum for hearing from affected families.

The guidelines of the American Medical Association (Council on Ethical and Judicial Affairs, 2003) and the American Society of Clinical Oncology (2003) advise physicians to inform and encourage their patients to disseminate family genetic information as a means of meeting responsibility to family members. Commentators emphasize the burden and impracticality of requiring any more of health professionals (Mertz *et al.*, 1998; Offit *et al.*, 2004) and the threat to patients' confidentiality (Sulmasy, 2005). Some point out that physicians may not be aware of family dynamics and therefore cannot determine if disclosure is less harmful than nondisclosure (Rothstein, 1997). Also, the burden on a physician to find and contact at-risk family members may be overwhelming. This position is further bolstered by privacy legislation, including the Healthcare Insurance Portability and Accountability Act (HIPAA) that forbids any disclosure of personal healthcare information without patient authorization except in cases of "serious and imminent threat to the health or safety of a person or the public (45 CFR §160.510(j))." Yet, others continue to emphasize the familial nature of genetic information (Wachbroit, 1996). The family covenant model (Doukas and Berg, 2001) stipulates that it is the duty of the patient (Weijer, 2000) and permissible for the physician (Knoppers, 2002) to share genetic information with at-risk family members. Wachbroit (1993) further argues that the family, rather than the individual, is the patient for hereditary diseases; therefore, the health professional's patient care obligations extend to all family members and include a duty to share genetic information with the family. Parker and Lucassen (2003) counter that if serious harms are to be avoided, the model for sharing genetic information, individualistic or familial, must be decided on a case by case basis. A recent World Health Organization (WHO) report states, "Genetic information is not the sole property of individuals, but is shared among family members... . It would be unethical to conceal genetic information from its owners, who include blood relatives with genes for a disorder (Wertz *et al.*, 2003)." The WHO report follows the recommendations of the American Society of Human Genetics, which allows disclosure without consent in strictly limited circumstances: (1) the patient persistently refuses to disclose to family members; (2) harm to family members is highly likely, serious and foreseeable; (3) the at-risk relative(s) is identifiable; (4) the disease is preventable or treatable or early monitoring reduces risk; and (5) the harm of nondisclosure is greater than the harms attendant upon disclosure (American Society of Human Genetics, 1998). The Institute of Medicine's Committee on Assessing Genetic Risks supports this position but adds another restriction: the disclosure is limited to the information necessary for diagnosis or treatment of the relative (Institute of Medicine Committee, 1994).

Opinions in the most relevant judicial cases also differ, with *Safer v. Estate of Pack* holding that a physician should have warned a daughter about the hereditary nature of her father's

colorectal cancer (Safer v. Estate of Pack, 1996) and with Pate v. Threkel placing the responsibility to warn on the patient (Pate v. Threkel, 1995).

People generally express positive views about informing family members about inherited cancer risk information (Hughes *et al.*, 2002; Peterson *et al.*, 2003), particularly those in families with hereditary cancer syndromes (Hallowell *et al.*, 2003; Kenen *et al.*, 2004; Wilson *et al.*, 2004). In fact, testing decisions are often strongly motivated by a desire to provide information for one's family (Julian-Reynier *et al.*, 2000). However, not all persons who are at risk for inheriting a mutation are informed of this possibility (Ayme *et al.*, 1993; Falk *et al.*, 2003). Studies have identified barriers to genetic risk communication (Wilson *et al.*, 2004) including a lack of preventive strategies available (Lehmann *et al.*, 2000); personal factors such as discomfort discussing this type of information (Daly *et al.*, 1999; Hallowell *et al.*, 2003) and concern for upsetting relatives (Hughes *et al.*, 2002; Kenen *et al.*, 2004); family-based factors such as estrangement (Falk *et al.*, 2003) and distance of family relationship (Claes *et al.*, 2003; Green *et al.*, 1997); and sociocultural factors such as concerns about discrimination and the need for consent (Benkendorf *et al.*, 1997).

Approximately half of these studies queried families with known or suspected hereditary cancer syndromes, and all focused on breast and ovarian cancer risk (Benkendorf *et al.*, 1997; Claes *et al.*, 2003; Daly *et al.*, 1999; Forrest *et al.*, 2003; Green *et al.*, 1997; Hallowell *et al.*, 2003; Hughes *et al.*, 2002; Julian-Reynier *et al.*, 2000). Only one of these studies directly questioned study participants about the duty to warn at-risk relatives and the role of the health professional in information dissemination (Forrest *et al.*, 2003), although an additional three studies, whose subjects did not have family histories suggestive of a cancer syndrome, queried the duty to warn (Durfy *et al.*, 1999; Julian-Reynier *et al.*, 1996; Lehmann *et al.*, 2000). The study reported here is responsive to the research need to study at-risk family members (Lehmann *et al.*, 2000; Gollust *et al.*, 2005) by being the first study to query members of hereditary nonpolyposis colorectal cancer (HNPCC) families.

The duty to warn is arguably particularly relevant in the context of HNPCC. Genetic testing can identify deleterious mutations that predispose to HNPCC, and HNPCC also may be diagnosed clinically using the Amsterdam criteria (Vasen *et al.*, 1999). The lifetime risk of colorectal cancer in individuals with HNPCC is estimated to be up to 82%, with substantially increased risks for endometrial, ovarian, and other associated cancers (Aarnio *et al.*, 1999; Dunlop *et al.*, 1997; Vasen *et al.*, 2001). Because of young age of onset, recommended cancer surveillance for persons with a clinical or genetic diagnosis of HNPCC includes colonoscopy every 1 to 2 years beginning by age 25, well below the average population age 50, and annual transvaginal ultrasonography and endometrial biopsy beginning at age 30 (Burke *et al.*, 1997; Lynch and de la Chapelle, 2003). Surveillance can reduce the incidence of colorectal cancer by more than 50% and overall mortality by 65% (Jarvinen *et al.*, 2000). At the present time, genetic counseling and testing for this adult-onset condition is advised for persons who are 18 years old or older.

The objectives of this study were to describe HNPCC family members' perceptions about who has a right to know about a genetic mutation in the family and who should disclose this information to family members, with a focus on the role that should be played by health professionals in disseminating this information.

Materials and Methods

Study design

A cross-sectional, qualitative study to evaluate family communication regarding HNPCC (Koehly *et al.*, 2003; Peterson *et al.*, 2003), which included queries analyzed for this report about the duty to warn and the role of the health professional in information dissemination, was conducted as part of a multiphase longitudinal study of psychosocial aspects of HNPCC genetic counseling and testing (Gritz *et al.*, 2005; Vernon *et al.*, 1999). The family communication study was reviewed and approved by The University of Texas M.D. Anderson Cancer Center Institutional Review Board, and was conducted concurrently with a clinical research protocol that offered genetic counseling and testing for HNPCC at no cost first to colorectal cancer patients, and then to their biologic relatives. Each proband, the first person tested in a family, who was HNPCC mutation-positive, was asked to provide relatives' names and addresses and written permission for the researchers to contact them regarding the communication study and the opportunity to receive genetic counseling and testing.

Study population

The eligibility criteria for families were: (1) at least one family member had participated in the longitudinal study and was currently undergoing genetic testing and counseling for HNPCC or had completed the same and had received positive genetic test results and (2) the family included at least five members estimated to be at 50% risk of carrying a mutation who were at least 18 years of age and who could speak and read English. We included families with at least five at-risk members to assure that we had an adequate number of persons within the family network to evaluate communication patterns in a meaningful way. Family members who were not biologically related to mutation carriers (e.g., spouses) were included, as were biologic relatives of the proband regardless of whether they had undergone genetic counseling or testing.

Recruitment

Recruitment and data collection procedures have been described in detail elsewhere (Gritz *et al.*, 1999; Koehly *et al.*, 2003). Letters of invitation to participate in the family communication study were mailed to the probands, their at-risk relatives, and the spouses of both whom we had permission to contact. Study staff subsequently telephoned these potential participants to provide additional details about the study and to answer questions. Family members who consented to participate in the study made an appointment to complete a semistructured interview, which was conducted by telephone. All interviews were tape-recorded and transcribed verbatim.

Interview guide

A semistructured interview guide was developed to evaluate families' communication about and responses to genetic counseling, testing and surveillance for HNPCC. Semistructured questions were based on ethics literature discussions of the duty to warn (Campbell and Lustig, 1994), models of family functioning (Sawom and Harrigan, 1994), and on the

investigators' prior research findings with these families. For this paper, we analyzed responses to questions regarding family members' right to know about genetic information and the duty to warn others about genetic risk. The questions analyzed are included in Table 2.

Qualitative analysis

Transcripts were coded and analyzed using qualitative methods to identify emergent themes related to the research questions (Miles and Huberman, 1994). One investigator (R.P.) reviewed the transcripts and developed a code list and coding rules. Two coders independently coded respondents' answers. There was a high level of concordance between the coders, with 10 disputes resolved by R.P. Findings from the analysis also underwent expert review by other study investigators. The transcripts were supplemented by data obtained from genetic counseling charts and other study records, including a family history questionnaire, demographics and genetic testing status. Quotations were included verbatim, and names were eliminated to protect confidentiality.

Quantitative analysis

SPSS version 10.1 (SPSS Inc., Chicago, IL) was used for descriptive statistics to explore bivariate relationships between sociodemographic and medical variables for study participants and nonparticipants using χ^2 tests.

Results

Characteristics of the study population

Of the 53 families participating in the longitudinal study, 16 met study criteria and each family contributed from 1 to 9 participants. We contacted 155 individuals from these families and 52% (80/155) completed an interview. An HNPCC-predisposing mutation had been identified in most families (12/16) at the time of the interview, and about half of at-risk family members had undergone genetic counseling, donated blood for genetic testing and received their test results before the interview. Table 1 summarizes study participants' characteristics. Nonparticipants were more likely to be male, spouses, to have no personal history of cancer, and to have declined genetic counseling and testing. Participants and nonparticipants did not differ on age, race, marital status, having children, and the presence of an identified mutation in the family. Nor did they differ on the proportion of mutation carriers compared to noncarriers among those who completed genetic counseling and testing.

Results

The frequencies of coded answers to the questions are reported in Table 2. Respondents offered more than one response to open-ended questions, and some respondents did not answer all questions. An analysis of the transcripts with more than one response to a question suggested that second or third mentions were just as important to the responder. We therefore report all responses and tally the percentage of responders who offered each response, in order to fully describe our respondents' views.

Right to know—The overwhelming majority of family members indicated that all persons in the family should be informed about the identification of an HNPCC-predisposing mutation in the family. A smaller percentage suggested that this information should only be shared among biologic relatives.

Duty to inform—Most indicated that the most appropriate person to share the information would be a member of the family, and specifically mentioned that such persons included the proband, family members with the most experience in genetic testing or cancer, or anyone in the family. Interestingly, health professionals were mentioned by approximately one quarter of participants as being appropriate informants.

Health professional—Asked specifically if it was permissible for a health professional to inform family members about the genetic mutation, most agreed, and only a few agreed with qualifiers such as “with permission.” A few reported that it was a health professional's duty to inform family members. As one stated,

I think it comes back to the Hippocratic Oath. The first law is to do no harm. If to say nothing is to do harm, then I think you have an obligation to pass the information on.

Study participants suggested several advantages to involving health professionals in the dissemination of the genetic risk information. First, health professionals can be trusted to understand the information. As one person explained:

Let's say something happens to me and {my daughter} doesn't get the word or whatever. I would rather she find out from a place like {name of hospital} than not at all or from my husband who really doesn't understand.

Second, participants also believed that when health professionals take the lead in informing family members, family-based barriers might be overcome. When asked about the most appropriate informant, one participant explained.

... probably more appropriate would be staff maybe to call and say there is this mutation... . When family members do it then, you know, you just get in a big uproar, nothing gets done.

Another explained that health professionals could overcome such barriers as geographical or emotional/social distance and death or refusal of the proband to share:

[Y]ou've got families that are sometimes so scattered and people that are not blood related can be closer than people that are blood related and true you have a proband that either died or chose, you know, not to tell somebody else, that needed to know.

Some specifically mentioned family communication road-blocks due to fragile family relations. As one said:

You don't want things [dissemination of information throughout the family] to stop just depending on someone's fragile family relations.

Third, respondents were also attuned to privacy issues and some thought involving health professionals would enhance privacy, since professionals could be counted on not to reveal individual results:

I think that [a health professional serving as informant] is even better, actually, because I can't imagine somebody in the family saying, "We have a genetic alteration," and then not telling who has it. A health professional has confidentiality.

Another referenced the individual's right to know:

I know there are these, you know, privacy things and concern about liabilities and law suits and everything that everybody has to worry about these days too much, but I think one person's privacy should not override and infringe upon another person's right to know—that's where you get into the duty to inform.

Still another gave a utilitarian analysis, justifying the breach of one person's confidentiality by the good it would bring to many:

I think it would be appropriate for the hospital to let us know... . Yeah, because here you are talking about more people and just breaking the confidentiality with one person and this is a matter of maybe 30 or more people.

Right to know another's genetic test results—Some participants distinguished sharing news that a mutation had been found in the family from sharing individual test results. They believed that confidentiality would be adequately protected if information about the identification of an HNPCC mutation in the family was shared without sharing individual genetic test results.

I think knowing someone's test results is no one's business... . But {the family has} the right to know that there is a genetic alteration within the family but not the need to know who it is, because if there is a probability that you may have {the mutation} I think is important to know.

Another used this distinction between family and personal information as the basis for suggesting how the health professional should approach the dissemination of information:

I do feel like people do need to know but I also believe in confidentiality, wholeheartedly. [The health professional could say], for instance, a genetic alteration has been found in your family's history, maybe not to even state the person ... and that if you would like to know more about this, please contact us. See, then that person has the right of choice... . It's just not blatantly coming through the mail saying we found an alteration in Uncle Joe. That's still maintaining that confidentiality for that person, but its making available the medical information to that family.

Informing children of nontesters—In an effort to further probe the boundaries of individual privacy, over two thirds of participants were further asked whether children, who were not informed about the genetic mutation by parents who refused genetic counseling and testing, should be informed. All but one stated that children should be told.

As one respondent remarked:

Oh boy, that's stepping on toes, isn't it? ... to be really honest and, of course, this is close to me, but I probably feel that they really did need to know.

Approximately one fourth of participants stipulated that a child must be over 18, and one stipulated that a child should be informed of the family mutation but not of individual results. Some respondents focused on the need for screening colonoscopies as their justification for telling children:

I was worried about them... . They're going to be lax. They're not going to take care. They're not going to go to get a colonoscopy. So they should have it [genetic testing]. And if they don't have the gene, then they should get it [a colonoscopy] every once in while. But if they have the gene, they should take it [a colonoscopy] every 2 years... .

Although less than half of the respondents were further probed about their views regarding who should inform a child about the existence of a familial mutation, most of those queried thought this responsibility lay with the children's other relatives. Approximately one third placed this responsibility on a health professional. When asked, "Should another family member or a health professional approach the child?" one respondent replied,

Health professional. You'd get into too much family of origin trash. Getting someone else mad. That's why a professional would be better.

Discussion

To the best of our knowledge, this is the first study to evaluate HNPCC family members' perceptions about the duty to warn relatives about genetic risk information, and the role that health professionals should play in the disclosure process. Unlike studies of hereditary breast and ovarian cancer (HBOC) families, this population included both men and women who are at risk for HNPCC cancers as well as their spouses with whom they share children at risk, providing gender diversity usually unavailable in studies of HBOC families. Still, our results with HNPCC families support those found in HBOC families (Green *et al.*, 1997; Hallowell *et al.*, 2003) that family members are generally willing to and do share with each other information about the existence of a cancer-predisposing gene mutation, in spite of concerns and barriers. The responsibility to inform children and the focus on the importance of surveillance were major themes for HNPCC families as they were for HBOC families (D'Agincourt-Canning, 2001; Forrest *et al.*, 2003). We were able to delve more deeply into the contentious issue of the role of health professionals in information dissemination, particularly in light of concern for confidentiality of health care information. Although our participants identified family members as the first-line informants, they, like the family members interviewed by Forrest (2003), thought health professionals were an important resource, particularly in overcoming barriers to dissemination. These barriers included those identified by other studies, such as incomplete information or lack of understanding of genetics and cancer syndromes, family dynamics, and individual coping styles. Some recommended that a health professional be the first-line informant, particularly if the proband consents, so that barriers could be overcome. As has been reported in other studies,

people trust their own health providers (Kass *et al.*, 1996), and these HNPCC family members judged these trusted advisors to be important resources in their attempt to disseminate genetic risk information within their families.

A repeated theme was the concern for confidentiality of information. Many were attuned to this issue but judged that the importance of knowing about the existence of the familial mutation was enough to outweigh concerns of confidentiality, particularly if individual results were not revealed. The strength of the conviction that information about the cancer-predisposing gene mutation in the family should be at least offered to all family members was demonstrated by the answers to our question about the correct action if a parent refused testing resulting in a child not being informed about the familial mutation. All but one respondent thought the child should be informed, a few even reporting that a health professional should tell the child. In general, Health Insurance Portability and Accountability Act (HIPAA) regulations would not be a barrier to the supportive role for health professionals envisioned by these respondents, because these probands would gladly consent to information dissemination. Informing the child of a parent who did not test may be an exception, but as others have argued, HIPAA, though well intentioned, may, in certain circumstances, compromise patient care (Blackburn, 2004; Salen and Parker, 2002).

These respondents' tendency to be more private about individual results than about the existence of a family mutation suggests a distinction between "family information," information about the family's genetic makeup, for example, that an inheritable cancer-predisposing gene has been found in some family member(s) and thus may be found in other family members, and "private health care information," information about an individual's own genetic makeup, for example, an individual's genetic test result. This distinction sheds some light on the debate over whether genetic information is familial or individual, suggesting that the information that a mutation exists in the family belongs to the family, allowing dissemination of this information throughout the family, while individual results are owned by the individual, to be shared as the individual chooses. This distinction could also be helpful in focusing the current emphasis on patient privacy on individual results rather than on the existence of a mutation in a family.

Although this study provides new insight on an important topic in genetic testing for inherited cancer, several limitations must be noted. The family members who were interviewed for this study generally shared genetic information willingly with their families; however, not all family members participated in an interview. Those declining the interview were more likely also to have declined genetic counseling and testing, so this willingness to share may not be characteristic of family members who were not interviewed. In addition, participants may have favorable perceptions about the role of health professionals in disclosing genetic information because of their involvement in genetics research. Because the majority of families interviewed were white and up to 40% had a college or graduate degree, the findings may not be generalizable to more ethnically and socioeconomically diverse populations. Finally, families may be more willing to discuss genetic risk for a condition such as HNPCC, with effective early detection options, compared with other conditions that have limited or unproven screening or treatment recommendations. Although providing support for health professional involvement in dissemination of genetic risk

information, we did not query our participants about preferred methods of family education, such as family meetings or sharing written or media presentations.

Conclusion

Although the current emphasis on privacy may make health professionals cautious in extending their role beyond the person seeking genetic testing, they should remain responsive to individuals who desire their assistance. Many of the individuals interviewed here were concerned about family members who may not be offered information, and they desired professional help in dissemination of information.

Acknowledgments

This research was supported in part by the National Human Genome Research Institute, National Institutes of Health grant number 2R01HG1200-6, and by The Kleberg Human Cancer Genetics Development Fund.

All work reported here is original and the sole work of the authors listed above.

References

- 45 CFR § 160.510(j). Available www.hhs.gov/ocr/regtext.html Last accessed January 27, 2005.
- Aarnio M, Sankila R, Pukkala E, et al. Cancer risk in mutation carriers of DNA-mismatch-repair genes. *Int J Cancer*. 1999; 81:214–218. [PubMed: 10188721]
- American Society of Clinical Oncology. Policy Statement Update: Genetic Testing for Cancer Susceptibility. *J Clin Oncol*. 2003; 21:2397–2406. [PubMed: 12692171]
- American Society of Human Genetics Social Issues Subcommittee on Familial Disclosure. ASHG statement: Professional disclosure of familial genetic information. *Am J Hum Genet*. 1998; 62:474–483. [PubMed: 9537923]
- Ayme S, Macquart-Moulin G, Julian-Reynier C, et al. Diffusion of information about genetic risk within families. *Neuromuscul Disord*. 1993; 3:571–574. [PubMed: 11660032]
- Benkendorf JL, Reutenauer JE, Hughes CA, et al. Patients' attitudes about autonomy and confidentiality in genetic testing for breast-ovarian cancer susceptibility. *Am J Med Genet*. 1997; 73:296–304. [PubMed: 9415688]
- Blackburn, M. HIPAA, heal thyself. Vol. 56. *Johns Hopkins Magazine*; 2004. p. 50-55.
- Burke W, Peterson G, Lynch P, et al. Recommendations for follow-up care of individuals with an inherited predisposition to cancer: I. Hereditary nonpolyposis colon cancer. *JAMA*. 1997; 277:915–919. [PubMed: 9062331]
- Campbell, CS.; Lustig, BA., editors. *Duties to Others*. The Netherlands: Kluwer Academic Publishers; 1994.
- Claes E, Evers-Kiebooms G, Boogaerts A, et al. Communication with close and distant relatives in the context of genetic testing for hereditary breast and ovarian cancer in cancer patients. *Am J Med Genet*. 2003; 116A:11–19. [PubMed: 12476445]
- Council on Ethical and Judicial Affairs of the American Medical Association. [March 18, 2005] Opinion E-2.131: Disclosure of Familial Risk in Genetic Testing. American Medical Association, Code of Medical Ethics: Current Opinion. Available www.ama-assn.org/apps/pf_new/pf_online?f_n=browse&doc=policyfiles/HnE/E-2.131.HTM&&s_t=&st_p=&nth=1&prev_pol=policyfiles/HnE/E-1.02.HTM&nxt_pol=policyfiles/HnE/E-2.01.HTM
- D'Agincourt-Canning L. Experiences of genetic risk: Disclosure and the gendering of responsibility. *Bioethics*. 2001; 15:231–247. [PubMed: 11700677]
- Daly, M.; Itzen, M.; Costalas, JW. The communication of BRCA 1 and BRCA2 test results to at risk relatives. Presented at Breast Cancer Symposium; 1997; San Antonio, TX. In Daly M, Farmer J,

- Harrop-Stein C, *et al*: Exploring family relationships in cancer risk counseling using the genogram. *Cancer Epidemiol Biomarkers Prev*. 1999; 8:393–398.
- Doukas DJ, Berg JW. The family covenant and genetic testing. *Am J Bioeth*. 2001; 1:2–10.
- Dunlop MG, Farrington SM, Carothers AD, *et al*. Cancer risk associated with germline DNA mismatch repair gene mutations. *Hum Mol Genet*. 1997; 6:105–110. [PubMed: 9002677]
- Durfy SJ, Bowen DJ, McTiernan A, *et al*. Attitudes and interest in genetic testing for breast and ovarian cancer susceptibility in diverse groups of women in western Washington. *Cancer Epidemiol Biomarkers Prev*. 1999; 8:369–375. [PubMed: 10207642]
- Falk MJ, Dugan RB, O'Riordan M, *et al*. Medical geneticists' duty to warn at-risk relatives for genetic disease. *Am J Med Genet*. 2003; 120A:374–380. [PubMed: 12838558]
- Forrest K, Simpson SA, Wilson BJ, *et al*. To tell or not to tell: Barriers and facilitators in family communication about genetic risk. *Clin Genet*. 2003; 64:317–326. [PubMed: 12974737]
- Gollust SE, Apse K, Fuller BP, *et al*. Community involvement in developing policies for genetic testing: Assessing the interests and experiences of individuals affected by genetic conditions. *Am J Pub Health*. 2005; 95:35–41. [PubMed: 15623855]
- Green J, Richards M, Murton F, *et al*. Family communication and genetic counseling: The case of hereditary breast and ovarian cancer. *J Genet Couns*. 1997; 6:45–61.
- Gritz ER, Peterson SK, Vernon SW, *et al*. Psychological impact of genetic testing for hereditary nonpolyposis colorectal cancer. *Clin Oncol*. 2005; 23:1902–1901.
- Gritz ER, Vernon SW, Peterson SK, *et al*. Distress in the cancer patient and its association with genetic testing and counseling for hereditary non-polyposis colon cancer. *Cancer Res Ther Control*. 1999; 8:35–50.
- Hallowell N, Foster C, Eeles R, *et al*. Balancing autonomy and responsibility: The ethics of generating and disclosing genetic information. *J Med Ethics*. 2003; 2:74–79. [PubMed: 12672886]
- Hughes C, Lerman C, Schwartz M, *et al*. All in the family: Evaluation of the process and content of sisters' communication about BRCA1 and BRCA2 genetic test results. *Am J Med Genet*. 2002; 107:143–150. [PubMed: 11807889]
- Institute of Medicine Committee on Assessing Genetic Risks. *Assessing Genetic Risks: Implications for Health and Social Policy*. Washington, D.C.: National Academy Press; 1994.
- Jarvinen HJ, Aarnio M, Mustonen H, *et al*. Controlled 15-year trial on screening for colorectal cancer in families with hereditary nonpolyposis colorectal cancer. *Gastroenterology*. 2000; 118:829–834. [PubMed: 10784581]
- Julian-Reynier C, Eisinger F, Vennin P, *et al*. Attitudes towards cancer predictive testing and transmission of information to the family. *J Med Genet*. 1996; 33:731–736. [PubMed: 8880571]
- Julian-Reynier C, Eisinger F, Chabal F, *et al*. Disclosure to the family of breast/ovarian cancer genetic test results: Patients' willingness and associated factors. *Am J Med Genet*. 2000; 94:13–18. [PubMed: 10982476]
- Kass NE, Sugarman J, Faden R, *et al*. Trust, the fragile foundation of contemporary biomedical research. *Hastings Cent Rep*. 1996; 26:25–29. [PubMed: 8891706]
- Kenen R, Arderm-Jones A, Eeles R. We are talking, but are they listening? Communication patterns in families with a history of breast/ovarian cancer (HBOC). *Psychooncology*. 2004; 13:335–345. [PubMed: 15133774]
- Knoppers BM. Genetic information and the family: Are we our brother's keeper? *Trends Biotechnol*. 2002; 20:85–86. [PubMed: 11814599]
- Koehly LM, Peterson SK, Watts BG, *et al*. A social network analysis of communication about hereditary nonpolyposis colorectal cancer genetic testing and family functioning. *Cancer Epidemiol Biomarkers Prev*. 2003; 12:304–313. [PubMed: 12692104]
- Lehmann LS, Weeks JC, Klar N, *et al*. Disclosure of familial genetic information: Perceptions of the duty to inform. *Am J Med*. 2000; 109:705–711. [PubMed: 11137485]
- Lynch HT, de la Chapelle A. Genomic medicine: Hereditary colorectal cancer. *N Engl J Med*. 2003; 348:919–932. [PubMed: 12621137]
- Mertz JF, Cho MK, Sandar P. Familial disclosure in defiance of nonconsent. *Am J Hum Genet*. 1998; 63:898–899. [PubMed: 9718355]

- Miles, M.; Huberman, A. *Qualitative Data Analysis*. Beverly Hills: Sage; 1994.
- Offit K, Groeger E, Turner S, et al. The “duty to warn” a patient's family members about hereditary disease risks. *JAMA*. 2004; 292:1469–1473. [PubMed: 15383518]
- Parker M, Lucassen A. Concern for families and individuals in clinical genetics. *J Med Ethics*. 2003; 29:70–73. [PubMed: 12672885]
- Pate v. Threlkel, 661 So. 2d 278, FL 1995.
- Peterson SK, Watts BG, Koehly LM, et al. How families communication about HNPCC genetic testing: Findings from a qualitative study. *Am J Med Genet*. 2003; 119C:78–86. [PubMed: 12704641]
- Rothstein, MA., editor. *Genetic Secrets: A Policy Framework*. New Haven: Yale University Press; p. 451-495.p. 1997
- Safer v. Estate of Pack, 677 A.2d 1188, NJ Super A.D. 1996.
- Salen DN, Parker SG. The adverse effects of HIPAA on patient care. *N Engl J Med*. 2002; 349:309.
- Sawom KJ, Harrigan M. Measures of family functioning for research and practice. *Sch Inq Nurs Pract*. 1994; 8:5–142. [PubMed: 7938979]
- Sulmasy DP. Duty to warn about hereditary disease risks. *JAMA*. 2005; 293:676. [PubMed: 15701907]
- Vasen HF, Watson P, Mecklin JP, et al. New clinical criteria for hereditary nonpolyposis colorectal cancer (HNPCC, Lynch Syndrome) proposed by the international collaborative group on HNPCC. *Gastroenterology*. 1999; 116:1453–1456. [PubMed: 10348829]
- Vasen HF, Stormorken A, Menko FH, et al. MSH2 mutation carriers are at higher risk of cancer than MLH1 mutation carriers: A study of hereditary nonpolyposis colorectal cancer families. *J Clin Oncol*. 2001; 19:4074–4080. [PubMed: 11600610]
- Vernon SW, Gritz ER, Peterson SK, et al. Design and methodology of psychosocial aspects of genetic testing for hereditary colorectal cancer. *Ann NY Acad Sci*. 1997; 833:190–194. [PubMed: 9616754]
- Vernon SW, Gritz ER, Peterson SK, et al. Intention to learn the results of genetic testing for hereditary colon cancer. *Cancer Epidemiol Biomarkers Prev*. 1999; 8:353–360. [PubMed: 10207640]
- Wachbroit R. Rethinking medical confidentiality: The impact of genetics. *Suffolk University Law Rev*. 1993; 27:1391–1410.
- Wachbroit R. *Disowning Knowledge: Issues in Genetic Testing*. Report from the Institute for Philosophy and Public Policy. 1996; 16:14–16.
- Weijer C. Family duty is more important than rights. *BMJ*. 2000; 321:1466.
- Wertz, DC.; Fletcher, JC.; Berg, K. [online March 18, 2005] Review of Ethical Issues in Medical Genetics: Report of Consultants to WHO. Available whqlibdoc.who.int/hq/2003/WHO_HGN_ETH_00.4.pdf
- Wilson BJ, Forrest K, van Teijlingen ER, et al. Family Communication about Genetic Risk: The little that is know. *Community Genet*. 2004; 7:15–24. [PubMed: 15475667]

Table 1
Demographic, Medical, and Family Characteristics of Participants ($n = 80$), n (%)

<i>Characteristics</i>		<i>n (%)</i>
Mean age at interview, (median, range)		48 (46, 21–81)
Gender	Female	49 (61)
Race	White	68 (85)
	Hispanic	8 (10)
	African American	4 (5)
Education	High school	14 (18)
	Some college	20 (25)
	College graduate	23 (29)
Household Income	\$50,000	34 (43)
	\$50,000	30 (38)
Married		57 (71)
1 Child		67 (84)
Personal history of cancer		17 (21)
Mutation identified in family		67 (84)
Family position	Proband (1st in family tested)	9 (11)
	Biologic relative	57 (71)
	Spouse	14 (18)
Underwent pretest genetic counseling		40 (50)
Underwent genetic testing	Positive test result	20 (25)
	Negative test result	11 (14)
	Did not receive result	6 (8)
	Declined genetic test	3 (4)

Table 2
Frequency of Responses Regarding Disclosure of HNPCC Genetic Information ($n = 80$)

<i>Questions and coded responses</i>	<i>Responses (responders)</i>	<i>n (% of responders)</i>
1. Who has the right to know there is a genetic mutation causing the cancer that runs in the family?	88 (78)	
Everyone in the family		64 (82)
Biologic relatives		12 (15)
Family doctor		5 (06)
Other		7 (09)
2. Whose duty is it to inform family members about the genetic mutation in the family?	91 (69)	
Anyone in the family or whoever knows		32 (46)
The first person to find out, mutation carrier, cancer patient or the most knowledgeable		30 (43)
Health professional, family physician		20 (29)
Older generation tells the younger generation		7 (10)
Other		2 (03)
3. May a health care professional inform relatives about the genetic mutation?	73 (68)	
Yes		52 (76)
No		7 (10)
Yes, with limitations (e.g., with permission, if individual results not revealed)		6 (08)
The health professional has a duty to tell		4 (06)
Do not know		4 (06)
4. Do family members have the right to know each other's genetic test results?	82 (75)	
No, only the person tested has the right to know, it is up to him/her		42 (56)
Yes, everyone in the family or bloodline has a right to know		22 (29)
The immediate family has a right to know		11 (15)
The person's doctor has a right to know		3 (04)
The person's spouse has a right to know		2 (03)

HNPCC, hereditary nonpolyposis colorectal cancer.