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FAMILY DISCLOSURE IN GENETIC TESTING FOR CANCER SUSCEPTIBILITY: DETERMINANTS AND CONSEQUENCES†

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I. INTRODUCTION AND OVERVIEW

The isolation of the BRCA1 and BRCA2 genes has made it possible to identify women at increased risk for breast and ovarian cancer, thereby facilitating informed decisions about surveillance and cancer prevention options.[1] Despite these potential medical benefits, the identification of carriers of deleterious mutations raises numerous psychological and social challenges for those being tested and for their family members.[2] One of the more pressing and least studied issues involves the process and outcomes of disclosure of genetic information within families. The present article addresses family disclosure of information about genetic testing for cancer susceptibility. Following an overview of the clinical aspects of family disclosure and the empirical literature on this topic, we present our preliminary data on the determinants and outcomes of disclosure of BRCA1 and BRCA2 ("BRCA1/2") genetic information within hereditary breast cancer families. These data are supplemented with case studies of patients, highlighting the motivations for and against disclosure and il-

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1. *See* Douglas F. Easton et al., *Breast and Ovarian Cancer Incidence in BRCA1-Mutation Carriers*, 56 AM. J. HUM. GENETICS 265 (1995); Deborah Ford et al., *Risks of Cancer in BRCA1-Mutation Carriers*, 343 LANCET 692 (1994); Richard Wooster et al., *Identification of the Breast Cancer Susceptibility Gene BRCA2*, 378 NATURE 789, 790 (1995). These studies, the first two of which are from the Breast Cancer Linkage Consortium, established that the lifetime risks of breast and ovarian cancer associated with BRCA1 mutations are about 85% and 63%, respectively, with onset often at a younger age than observed in the general population. *See* Easton et al., *supra*, at 270; Ford et al., at 270. Risks for breast cancer in women with BRCA2 mutations were found to be comparable to BRCA1, but the ovarian cancer risks were lower. *See* Wooster, *supra*, at 790. Prostate cancer risks appear to be elevated in male BRCA1 carriers. In addition, colon cancer risks may be elevated in men and women with a BRCA1 or BRCA2 mutation, and other more rare cancers have been associated with BRCA2 alterations. *See id.* Another study found lower risks of breast and ovarian cancer associated with three common mutations in Ashkenazi Jewish individuals who did not necessarily have a family history of cancer. Jeffery P. Struewing et al., *The Risk of Cancer Associated with Specific Mutations of BRCA1 and BRCA2 Among Ashkenazi Jews*, 336 NEW ENG. J. MED. 1401, 1401 (1997). The risks were still markedly elevated over the general population. *See id.* In addition, prostate cancer risks were elevated, though colon cancer risks were not. *See id; see also generally* Wylie Burke et al., *Recommendations for Follow-up Care of Individuals with an Inherited Predisposition to Cancer: II. BRCA1 and BRCA2*, 277 JAMA 997 (1997) (discussing provisional recommendations for early detection and cancer prevention in individuals with a BRCA1 or BRCA2 mutation, including heightened surveillance often commencing at an early age, and reviewing the data regarding the options for prophylactic surgery).

2. *See* Caryn Lerman et al., *BRCA1 Testing in Families with Hereditary Breast-Ovarian Cancer: A Prospective Study of Patient Decision Making and Outcomes*, 275 JAMA 1885, 1889 (1996) (discussing patients' perception of the benefits, limitations, and risks of testing, which included social concerns such as fears about insurance discrimination, and concerns about emotional adaptation and response of relatives to test results).

lustrating key counseling issues. Finally, we summarize these data and discuss the health-related and legal implications.

II. FAMILY DISCLOSURE OF GENETIC INFORMATION IN THE BRCA1 AND BRCA2 GENETIC COUNSELING SETTING

Disclosure of genetic information about cancer susceptibility has numerous implications for patients, family members, health care providers, and researchers. In the clinical and research settings, disclosure of one's mutation status provides a gateway for other family members to have access to genetic testing research protocols. Typically, BRCA1/2 testing within a family begins with a woman who has been diagnosed with breast or ovarian cancer, often at a young age (referred to as the proband). If a known disease-conferring mutation is identified, other first-degree relatives such as siblings and children have a 50% likelihood of also carrying the mutation and having an increased cancer risk.[3] In some families, it is also possible to identify more distant relatives who are at increased risk such as nieces, nephews, and cousins. With knowledge of the particular mutation carried by the proband, it becomes possible to offer testing to other family members for that same mutation.[4] However, in the interest of protecting the confidentiality of the participant, researchers or clinicians should not approach other family members about their risk status or about testing. A common process, employed in most clinical research settings, is to discuss with the proband the implications of her test result for other family members as well as the attendant personal and social risks.[5] Probands are then given the option to contact their relatives directly, to have the health care provider contact their relatives,

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3. *See generally* Barbara B. Biesecker et al., *Genetic Counseling for Families with Inherited Susceptibility to Breast and Ovarian Cancer*, 269 JAMA 1970 (1993). BRCA1 and BRCA2 alterations are inherited in an autosomal dominant fashion, which means that each child of a parent with an alteration has a 50% chance of having the same alteration. *See generally id.* Male and female offspring are at equal risk of inheriting BRCA1 and BRCA2 mutations. *See generally id.*

4. Within high-risk families, the advantage to first testing a woman with breast or ovarian cancer diagnosed at an early age is that she is most likely to carry an alteration if one is present within the family. *See* Maggie Ponder & Josephine M. Green, *BRCA1 Testing: Some Issues in Moving from Research to Service*, 5 PSYCHO-ONCOLOGY 223, 223 (1996). It is possible to test individuals without knowledge of whether there is a BRCA1 or BRCA2 mutation present in their family (*e.g.*, if all relatives with breast or ovarian cancer are deceased). *Id.* at 228. In such scenarios, a positive result will still yield useful information. However, a negative test result is not considered to be informative because it is not possible to distinguish whether the patient did not inherit a mutation present in her family or whether there is no detectable BRCA1 or BRCA2 mutation in the family. *Id.* at 227.

5. *See* Biesecker et al., *supra* note 3, at 1972-73. The authors concluded that a protocol to test for presymptomatic BRCA1 gene mutations should include:

or not to have any further contact with relatives.[6] Probands are also provided with written materials to share with their relatives to facilitate the discussion.

The genetic counselor is perhaps best situated to facilitate informed decisions about family disclosure by reviewing the potential benefits and risks with the patient. In deciding whether to disclose a positive test result, one may consider the potential medical benefits for other relatives. For example, disclosure of one's own test result may be required to provide a relative with the opportunity to be tested for the specific mutation in the family, should she or he decide to do so.[7] As mentioned above, such information may have medical value, particularly to female family members who may have a significantly elevated breast and ovarian cancer risk.[8] A potential benefit to the proband is that disclosure of a positive test result may also elicit both emotional support and instrumental assistance in seeking and obtaining information and medical care.[9] However, disclosure of genetic test results has potential risks, including loss of privacy, employment and insurance discrimination, and stigmatization.[10] Individual distress and family conflict may also be generated by disclosure of genetic information.[11] Despite the importance of family disclosure, there are limited empirical data available on this topic.

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(1) precounseling education and assessment; (2) a multidisciplinary team with expertise in the screening and management of breast and ovarian cancer, inheritance, DNA testing, and psychosocial counseling issues of late-onset disorders; and (3) follow-up services for the management of the increased risk for cancer as well as the residual emotional reactions on behalf of family members.

Id. at 1974; *see also* Lerman et al., *supra* note 2, at 1886-87 (BRCA1 counseling protocol).

6. *See* Biesecker et al., *supra* note 3, at 1972.

7. *See* Ponder & Green, *supra* note 4, at 227.

8. *See* Easton et al., *supra* note 1, at 265; Ford et al., *supra* note 1, at 692; Wooster et al., *supra* note 1, at 789; Struewing et al., *supra* note 1, at 1401.

9. *See* Biesecker et al., *supra* note 3, at 1972 (noting that a majority of family members opted to share the results of BRCA1 testing with family members in an effort to receive their support).

10. *See* Mark A. Rothstein, *Genetic Testing: Employability, Insurability, and Health Reform*, 17 J. NAT'L CANCER INST. MONOGRAPHS 87 (1995); Paul R. Billings et al., *Discrimination as a Consequence of Genetic Testing*, 50 AM. J. HUM. GENETICS 476 (1992).

11. *See* Robert T. Croyle et al., *Psychological Responses to BRCA1 Mutation Testing: Preliminary Findings*, 16 HEALTH PSYCHOL. 63, 67-69 (1997) (demonstrating that female carriers with no history of cancer or prophylactic surgery had high levels of test-related distress as measured by standard psychological assessments, but that overall, levels of general distress were not increased in this group); Henry T. Lynch et al., *A Descriptive Study of BRCA1 Testing and Reactions to Disclosure of Test Results*, 79 CANCER 2219, 2223, 2225-26 (1997) (containing anecdotal, qualitative descriptions of patient responses to testing, including sadness and survivor guilt). *But see* Lerman et al., *supra* note 2, at 1890 (finding that a subset of the BRCA1 carriers described in the Lynch et al. paper did not exhibit increases in depression and functional impairment when evaluated using standardized quantitative measures).

The following section provides an overview of published data about the processes and outcomes of family disclosure in the genetic testing context.

III. LITERATURE REVIEW ON FAMILY COMMUNICATION REGARDING GENETIC TESTING

Initial research on family communication about genetic testing suggests that most individuals will contact family members to obtain information about their family's medical history before counseling. Researcher Josephine Green and colleagues found that 78% of women who were scheduled for a genetic counseling session for inherited breast-ovarian cancer susceptibility communicated with a family member before their appointment to obtain family history information.[12] Specifically, probands were most likely to contact female relatives (i.e., mothers or sisters) for information about their family history.[13] Reasons for not contacting relatives who could have provided medical information about the family included not wanting to upset the relative with discussions about cancer.[14] Other reasons for not contacting relatives included lost communication with relatives and large age differences between siblings.[15] This study also found that 88% of respondents shared their post-counseling summary letter with at least one relative.[16]

Studies of family communication about other genetic disorders (e.g., cystic fibrosis) suggest that feedback provided by relatives through verbal and/or nonverbal communication may motivate or discourage individuals from undergoing genetic testing.[17] A study of cystic fibrosis testing found that a person's perceptions of their siblings' reactions to abortion was a significant predictor of usage of prenatal testing for this disorder.[18] Specifically, respondents who perceived that their siblings would approve of aborting an affected

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12. *See* Josephine Green et al., *Family Communication and Genetic Counseling: The Case of Hereditary Breast and Ovarian Cancer*, 6 J. GENETIC COUNSELING 45, 51 (1997).

13. *See id.* at 51-52.

14. *See id.* at 52.

15. *See id.*

16. *See id.* at 53.

17. *See* Dorothy C. Wertz et al., *Attitudes Toward the Prenatal Diagnosis of Cystic Fibrosis: Factors in Decision Making Among Affected Families*, 50 AM. J. HUM. GENETICS 1077, 1083 (1992). Cystic fibrosis is a potentially lethal genetic disease which results in the production of abnormally thick mucus which can clog the lungs and cause severe infections. *See generally* Francis S. Collins, *Cystic Fibrosis: Molecular Biology and Therapeutic Implications*, 256 SCIENCE 774 (1992). Carriers of the disease have no symptoms, but carrier parents have a 25% chance of having an affected child. *See id.*

18. *See* Wertz et al., *supra* note 17, at 1082-83.

fetus were three times more likely to use prenatal diagnosis.[19] In the BRCA1/2 testing context, probands who had strong positive beliefs about the benefits of genetic testing were likely to also encourage other family members to participate in genetic testing.[20] These studies underscore the influence of family disclosure and communication on decision making about genetic testing.

Although most individuals may disclose their genetic test results to family members, many are reluctant to provide clinicians and researchers with direct access to these family members. In a survey of attitudes about BRCA1/2 testing among high-risk women, a majority (>80%) felt that health care providers should not disclose their test results to immediate family members without their written consent.[21] In a cystic fibrosis screening program, only 54% of probands provided the research team with contact information for their at-risk relatives.[22] Thus, most genetic testing participants desire to maintain control over the diffusion of genetic information to relatives. Further, these decisions are typically made without consulting with family members.

Willingness to communicate with family members about genetic testing and genetic disorders may be influenced by factors such as gender[23] and cultural background.[24] For example, women appear to be more likely to discuss genetic testing with their female relatives (i.e., daughters) than with male relatives (i.e., brothers).[25] This may be attributable to perceptions that only mothers, sisters, and daughters are at-risk for cancer.[26] Our own data on BRCA1/2 testing, presented in the next section, provide further support for gender differences in family communication about BRCA1/2 testing.

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19. *See id.* at 1081-82.

20. *See* Andrea Farkas Patenaude et al., *Acceptance of Invitations for p53 and BRCA1 Predisposition Testing: Factors Influencing Potential Utilization of Cancer Genetic Testing*, 5 PSYCHO-ONCOLOGY 241, 245 (1996).

21. *See* Judith L. Benkendorf et al., *Patients' Attitudes About Autonomy and Confidentiality in Genetic Testing for Breast-Ovarian Cancer Susceptibility*, 73 AM. J. MED. GENETICS 296, 298 (1997).

22. *See* J.R. Sorenson et al., *Proband and Parent Assistance in Identifying Relatives for Cystic Fibrosis Carrier Testing*, 63 AM. J. MED. GENETICS 419, 421 (1996).

23. *See* Martin Richards, *Families, Kinship, and Genetics*, *in* THE TROUBLED HELIX: SOCIAL AND PSYCHOLOGICAL IMPLICATIONS OF THE NEW HUMAN GENETICS 249, 251 (Theresa Marteau & Martin Richards eds., 1996).

24. *See* James C. McCroskey & Virginia P. Richmond, *Willingness to Communicate: A Cognitive View*, *in* COMMUNICATION, COGNITION, AND ANXIETY 19, 31-32 (Melanie Booth-Butterfield ed., 1990).

25. *See* Ponder & Green, *supra* note 4, at 229-30.

26. *See id.* at 230.

Family communication may also differ among individuals with different ethnic or cultural backgrounds.[27] Culture has been described as a system that influences behavior and perceptions.[28] For example, the culture of many African Americans may generally be characterized as emphasizing the principle of spirituality and valuing interconnectedness, uniqueness, positivity, and sharing.[29] The culture of many European Americans is generally based on individualism and values the right to choose, honesty, sharing, and communication.[30] Research has shown that patterns of family communication about BRCA1 testing differ between African American and Caucasian women.[31] In a recent study, Caucasian women at increased risk for breast cancer were significantly more likely than African American women to communicate about genetic testing with a spouse and a parent.[32] Specifically, 66% of Caucasian women discussed genetic testing for hereditary breast cancer with their spouse, and 40% discussed it with a parent versus about 27% of African American women who discussed this issue with a spouse or parent.[33]

IV. PRELIMINARY DATA ON THE DETERMINANTS AND OUTCOMES OF FAMILY COMMUNICATION ABOUT BRCA1 AND BRCA2 TESTING

A. Research Questions

The published literature described previously provides some initial insights into the processes and determinants of communication of genetic information within families. However, it is important to assess communication processes and outcomes in a systematic manner and to address several key questions about family communication which are unanswered at present. Our research on BRCA1/2 testing in hereditary breast cancer seeks to fill some gaps in our knowledge about family communication by addressing the following research questions:

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27. *See* McCroskey & Richmond, *supra* note 24, at 31.

28. *See* COLLINS O. AIRHIHENBUWA, HEALTH AND CULTURE: BEYOND THE WESTERN PARADIGM 3 (1995).

29. *See* Anita P. Jackson & Susan J. Sears, *Implications of an Africentric Worldview in Reducing Stress for African American Women*, 71 J. COUNSELING & DEV. 184, 186 (1992).

30. *See* Judith N. Martin et al., *Conversational Improvement Strategies for Interethnic Communication: African American and European American Perspectives*, 61 COMM. MONOGRAPHS 236, 237 (1994).

31. *See* Chanita Ann Hughes, *Genetic Testing for Inherited Breast-Ovarian Cancer Susceptibility: The Role of Communication and Personality Characteristics*, 62, 64-65 (1997) (unpublished Ph.D. dissertation, Howard University) (on file with the Department of Psychology, Howard University).

32. *See id.* at 65.

33. *See id.*

(1) Among carriers and noncarriers of BRCA1/2 mutations, what are the rates of self-reported disclosure of BRCA1/2 test results to different family members?; (2) Are women more likely to disclose their BRCA1/2 test results than are males?; and (3) What are the psychological consequences to the proband of disclosing BRCA1/2 test results to family members? The first two of these questions are addressed in a family-based study of BRCA1/2 testing, conducted in collaboration with Dr. Henry Lynch at Creighton University. The third question is addressed in a clinic-based study conducted at the Lombardi Cancer Center at Georgetown University Medical Center.

B. Study #1: A Family-Based Study of BRCA1 and BRCA2 Testing

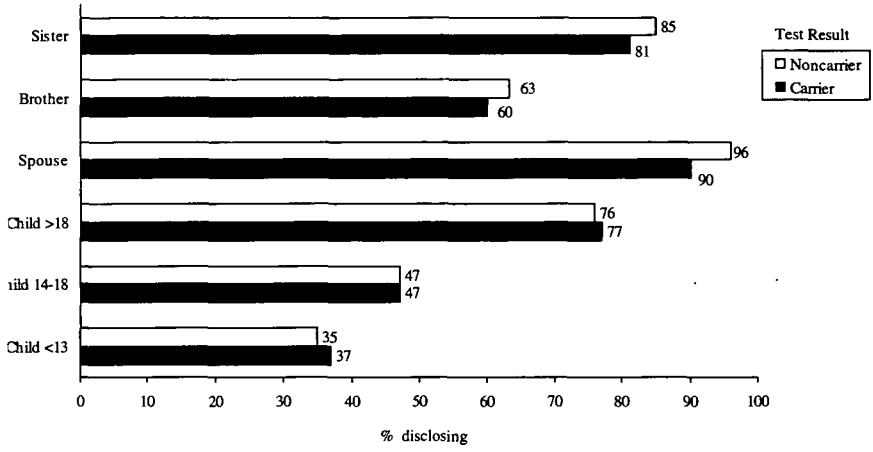
In this prospective cohort study, eligible participants are male and female members of hereditary breast cancer families who participated in earlier genetic linkage studies contributing to the isolation of the BRCA1/2 genes. Consequently, the pedigrees had been completed as part of the earlier research and the contact information on all family members was available. Thus, in contrast to most clinic-based studies, the proband is not placed in the position of providing contact information for other relatives at the time of study entry.

The current study was conducted on a family by family basis. First, letters of introduction were mailed to family members to inform them that the breast cancer susceptibility gene in their family had been identified and that genetic counseling and testing are now available. Consenting family members were asked to participate in a baseline telephone interview to assess demographic characteristics, risk factors, and psychosocial well-being. Individuals interested in genetic counseling and testing had the opportunity to participate in a pre-test education session; most of these sessions were conducted with the extended family. Those who elected to receive their BRCA1/2 test results did so after completing additional written consent forms and participating in individual genetic counseling. In this study, we are following mutation carriers, noncarriers, and decliners of BRCA1/2 testing for a one-year period to evaluate the psychosocial and medical impact of testing. The data on family communication presented here are based on the one-month follow-up assessment.

The frequencies for self-reported disclosure of BRCA1/2 test results among 201 carriers and noncarriers of BRCA1/2 mutations are shown in Figure 1. Overall, rates of disclosure within the first month following testing were quite high. For example, 81% of carriers disclosed their results to a sister and 60% disclosed to a brother. The rates of disclosure to minor children were surprisingly high, consider-

ing the fact that there are no immediate medical implications for young children.[34] Seventy-seven percent of carriers disclosed to an adult child, 47% disclosed to a child age fourteen to eighteen and 37% disclosed to a child under age thirteen.

FIGURE 1. FAMILY DISCLOSURE OF BRCA1/2 TEST RESULTS BY CARRIER STATUS

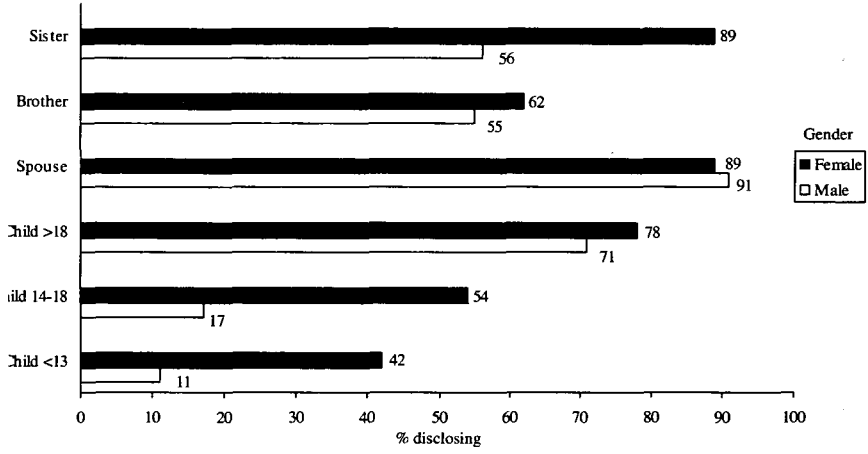


With respect to gender differences, self-reported rates of disclosure of test results among eighty-nine male and female mutation carriers are shown in Figure 2. Female carriers were more likely than males to disclose to a variety of family members. This was especially true for disclosure to sisters (89% of females versus 56% of males) and disclosure to children ages fourteen to eighteen (54% of females and

34. *But see* Ann-Marie Codori et al., *Genetic Testing for Cancer in Children: Short-term Psychological Effect*, 150 ARCHIVES PEDIATRIC ADOLESCENT MED. 1131 (1996); F.J.M. Grosfeld et al., *Psychological Risks of Genetically Testing Children for a Hereditary Cancer Syndrome*, 32 PATIENT EDUC. & COUNSELING 63, 64 (1997). These studies address genetic testing for conditions such as familial adenomatous polyposis, which can be associated with colon cancer in adolescents, and multiple endocrine neoplasia type 2A, which is associated with a serious form of thyroid cancer, for which prophylactic surgery in children is a consideration. *See generally* Codori et al., *supra*; Grosfeld et al., *supra*. In general, both studies concluded that there may be significant benefits to offering testing to children for predisposition to these disorders. *See generally* Codori et al., *supra;* Grosfeld et al. *supra*. There are other rare cancer predisposition syndromes for which it may be appropriate to test children, but the major reason for testing children is when there is an immediate medical benefit. *See* The American Society of Human Genetics Board of Directors and The American College of Medical Genetics Board of Directors, *ASHG/ACMG Report: Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents*, 57 AM. J. HUM. GENETICS 1233, 1234-36 (1995). In addition, the potential psychological harm must be weighed against the possible benefits. *See id.*

17% of males). One interpretation of these findings is that women are more comfortable communicating about health issues and dealing with the emotional sequelae of disclosure of a positive result. From a social perspective, it is not uncommon for women to take more of the responsibility for caretaking within the family.[35] It is also possible that the female spouses of the male mutation carriers in this study had disclosed the results to family members. However, these data are not available at the present time.

FIGURE 2. DISCLOSURE OF BRCA1/2 TEST RESULTS BY GENDER: CARRIERS ONLY



The results also indicated that the effects of carrier status (i.e., BRCA1/2 positive or negative) on disclosure varied by gender. For example, among males, noncarriers were more likely than carriers to disclose results to their sisters (78% versus 56%, respectively). By contrast, in females, the rate of disclosure to sisters was uniformly high (88%) and did not differ based on carrier status. The same pattern emerged for disclosure of BRCA1/2 test results to children. Among males, 33% of noncarriers and 17% of carriers disclosed their test results to a child age fourteen to eighteen. Among females, 53% disclosed to such a child, and there was no effect of carrier status on disclosure. Thus, it appears that men may be more comfortable sharing good news than bad news with other family members.

35. *See* Martin Richards, *Families, Kinship, and Genetics, in* THE TROUBLED HELIX: SOCIAL AND PSYCHOLOGICAL IMPLICATIONS OF THE NEW HUMAN GENETICS 249, 258 (Theresa Marteau & Martin Richards eds., 1996).

We also found that the likelihood of disclosing positive results with young children decreased as the education level of the participant increased. For example, 100% of carriers with less than high school education disclosed their results to a child age fourteen to eighteen, compared with 58% of high school and college graduates and 30% of participants with post-graduate education. To the extent that education level correlates with knowledge, we might interpret this to mean that increasing knowledge of the complexities and risks of disclosure (particularly to children) might dissuade some participants from disclosing to young children.

C. Study #2: A Clinic-Based Study of BRCA1 and BRCA2 Testing

As a result of their prior participation in genetic studies, the participants in the family-based study described above were more aware of the issues and complexities involved in genetic testing than most clinical populations. Further, counseling was performed on a family basis, thereby minimizing the disclosure burden to initial probands. Therefore, as a point of comparison, we are conducting a prospective cohort study of the outcomes of BRCA1/2 testing in the clinical setting. The study design is similar to that described above for Study #1, except that the testing process flows through the initial proband who is the gateway for providing access to other family members (after the proband's results are obtained, and if the result is positive). Further, all counseling and testing is conducted on an individual, rather than family, basis.

Despite differences in the method of ascertaining families, the rates of family disclosure in the clinic-based study were very similar to those for the family-based study. For example, about 81% of carriers and noncarriers disclosed to sisters and 45% disclosed to brothers. However, disclosure to children occurred less frequently in this setting and was more common among noncarriers than among carriers. For example, 40% of noncarriers disclosed their test results to a child age fourteen to eighteen as compared to 14% of carriers. Further, 21% of noncarriers disclosed to a child under age thirteen as compared to 9% of carriers. This suggests that some genetic testing participants may be motivated to disclose negative results for the purpose of reassuring their children.

With regard to the psychological impact of disclosure on the proband, the outcome appears to depend on the object of the disclosure. For example, BRCA1/2 carriers (mostly females in this study) who disclosed their result to their sister exhibited a small decrease in psychological distress, while those who elected not to tell exhibited a

small increase. This difference in trend was both statistically and clinically significant. Thus, this finding suggests that sharing a positive test result with a sister may initially have a positive effect on quality of life. This may be attributable to the fact that the proband fulfills a perceived responsibility to share information that could be medically significant to a close relative, and/or the fact that the proband may obtain emotional support from the relative.

By contrast, the reverse pattern was observed in the context of disclosure of positive test results to young children. In this case, probands who did not disclose their positive test results experienced reductions in distress, while those who did disclose experienced significant increases. Although preliminary, it is tempting to speculate that disclosure to young children may generate, rather than alleviate, psychological distress in carriers. Guilt about transmitting risk to one's offspring may be exacerbated by such discussions.

V. CASE STUDIES OF FAMILY DISCLOSURE IN THE CLINICAL RESEARCH SETTING

The concepts and results presented above are elucidated further by three case studies of the processes and outcomes of family disclosure of BRCA1/2 test results within the clinic-based study described above. These vignettes are based on actual cases but have been modified to protect privacy.

A. Case #1: All in Good Time

Ann is a fifty-five year old married Caucasian woman who tested positive for a BRCA1 alteration. Her medical history is significant for bilateral breast cancer diagnosed in her forties, for which she underwent mastectomies. She had her ovaries and uterus removed in her fifties as a preventive measure. Her mother died from ovarian cancer in her forties, and one of Ann's daughters had breast cancer at age thirty. Ann has two other adult daughters and an older brother and sister, none of whom has a history of cancer. Her siblings have adult sons and daughters. She also has several maternal cousins who are at risk for inheriting this alteration.

For Ann, there are few medical implications of this test result. However, there are several relatives who may now be tested. If found to carry this alteration, they would face increased risks for breast and ovarian cancer in women, and prostate cancer in male relatives.[36] During the initial pre-test genetic counseling session, Ann expressed

36. *See* Easton et al., *supra* note 1, at 265; Ford et al., *supra* note 1, at 692.

interest in testing to contribute to breast cancer research and also to gain information for her family, especially her daughters. Prior to obtaining her test results, Ann was concerned about the family's reaction to her results should she test positive, and acknowledged that, as a parent considering implications to her young adult daughters, she would harbor potential feelings of sadness, guilt, and even anger if she tested positive. She had only very limited discussions with her family about her decision to pursue testing. Of particular concern to her were the limitations in available screening and prevention options and how the information might affect her daughters' future childbearing decisions. Although she recognized the difficulty in communicating this information with her family, and the potential for significant emotional distress, she felt strongly about the importance of sharing this information.

When Ann received genetic counseling regarding her positive results, implications to family members were discussed in addition to exploring her own reactions and feelings. Of note, she was counseled that her daughter with breast cancer was very likely to carry this alteration, though Ann was not planning to share the information with her right away. The two individuals with whom Ann shared her results most immediately were her minister and her sister. Her sister was interested in testing and their discussions heightened Ann's concerns about the potential for insurance discrimination, as individuals without a prior history of cancer often have somewhat different worries about how their insurers will handle this type of "pre-existing" condition. She also began to explore with her sister issues related to the dissemination of this information to the rest of the family. Ann's sister had concerns about her own children learning about their aunt's test result.

Ann decided to defer discussion about her results with many relatives. For example, she decided not to disclose to her brother because he was having chronic medical problems. She also decided not to disclose to her daughter with breast cancer because she was undergoing chemotherapy, or to her two other daughters, one of whom was newly married and one of whom was pregnant. Ann clearly perceived the latter two events as happy occasions, and believed that news about her test result could wait until a more appropriate time. Within a year, she shared the information with all her daughters and her brother. Ann also contacted by phone some of her cousins with whom she had a relationship, but was not interested in contacting cousins with whom she had not seen or spoken to in many years. Eventually, her brother and sister were tested, but all of her daughters have declined testing at

the present time. Ann is undergoing counseling now to address her and her family's experiences with cancer and genetic testing, as well as other interpersonal issues.

Analysis: Although some individuals are highly motivated to pursue testing for the sake of family members and to share test results with these relatives, established patterns of communication within the family and the occurrence of other life circumstances are likely to influence how, when, and with whom test results are discussed.

B. Case #2: Don't Ask, Don't Tell

Deborah is a fifty-four year old married Caucasian woman who tested positive for a BRCA2 alteration. Her medical history is significant for unilateral breast cancer diagnosed at age fifty-two for which she underwent breast conserving surgery (lumpectomy), followed by radiation and chemotherapy. Her sister had breast cancer in her mid-fifties, and there is a very strong family history of cancer on their father's side of the family, including breast cancer in two aunts, male breast cancer, pancreatic cancer, and ovarian cancer. With the exception of Deborah, all individuals in the family with a diagnosis of cancer have died. Deborah has three children in their twenties and several nieces in their thirties who she thought would probably be interested in genetic testing. She also has numerous cousins who are also at risk. Prior to learning her test results, Deborah had informed several relatives that she had obtained genetic testing and alerted them to the approximate time in which she would receive her results.

Upon learning her results, Deborah expressed "relief" at finally learning why she developed cancer. Unlike the previous case, these results could have significant medical implications for herself as well as her family. Deborah learned that she was at increased risk for developing another breast cancer (in her affected and opposite breast) and that she also faced an increased risk of ovarian cancer and possibly pancreatic cancer.[37] She was counseled about options for early detection (e.g., frequent screenings for breast cancer, blood tests, and

37. *See* Ford et al., *supra* note 1, at 693 (describing the risks of contralateral breast cancers in BRCA1 carriers estimated at 64% by age 70); Kenneth Offit, *BRCA1: A New Marker in the Management of Patients with Breast Cancer?*, 77 CANCER 599, 600 (1996) (discussing the possibility that women with BRCA1 and BRCA2 alterations may also be at risk for ipsilateral breast cancer and the potential impact on management decisions). It is likely that contralateral breast cancer risks are elevated in BRCA2 carriers as well. *See* Offit, *supra*, at 600; *see also* Wooster et al., *supra* note 1, at 790; Struewing et al., *supra* note 1, at 1401; Catherine M. Phelan et al., *Mutation Analysis of the BRCA2 Gene in 49 Site-Specific Breast Cancer Families*, 13 NATURE GENETICS 120, 121 (1996) (discussing other cancers associated with BRCA2 alterations including pancreatic cancer).

ultrasounds for ovarian cancer) and risk reduction (e.g., use of Tamoxifen, a medication that may reduce the risk of another breast cancer; removal of her breasts and/or ovaries).[38] Although Deborah was concerned about these risks, she opted not to alter her medical management and believed that the other measures she employed to stay healthy, such as having a low fat diet and exercising, were sufficient and provided psychological benefits. She felt healthy and wanted to live with as few reminders of her cancer or her cancer risk as possible.

With respect to communication of her test results, within the first several weeks of learning her results, Deborah shared the information with her husband and a co-worker. She had also dropped hints about having her results to various family members including her children, and some of her nieces and cousins. She reported that none of these individuals inquired further as to what the results were or what the implications to them might be. Her feeling was that if they did not ask her directly for the information, she would not share it. She commented that as her children and nieces were young adults, there was no urgency to share this information, though she was counseled that women who have a BRCA2 alteration may face increased risks for breast and ovarian cancer even in their twenties and thirties. Because her result did not significantly change her medical management, she thought it was likely that it would not significantly impact others. She also feared that if relatives did get testing, they would associate testing positive with a "death sentence." Although she was aware that these relatives have a 50% chance of not having the alteration, and that learning such information could provide a substantial amount of reassurance about their cancer risks, she was more focused on the possibility of their testing positive. Through subsequent discussions with the counselor, Deborah revealed that at times, she felt somewhat guilty about "withholding information" from her family. One strategy for addressing this issue was to role play different language that could be used to disclose the information and to imagine the relatives' reaction along with her response.

It has been over a year since Deborah obtained her results, and no relatives have been notified of this information. Deborah believes that with time, her feelings about communicating her result may change, for example, as her children get older or as they consider having children. If there are changes in Deborah's own history or her

38. *See* Burke et al., *supra* note 1, at 997.

family history of cancer, these events may also affect her feelings about sharing the information.

Analysis: Individuals' beliefs about the impact of test results for themselves may affect their perception of how or whether others will utilize the information, or when they should be notified of the information. The health care providers informed Deborah about who is at risk and offered to facilitate communication with these relatives about the availability of genetic counseling with the option of testing, but were respectful of her wishes not to share the information. In order for individuals to feel comfortable pursuing testing, they must know that researchers and clinicians will handle the information responsibly and respect their autonomy and decision process.

C. Case #3: A Family Affair

Margaret is a sixty-five year old married Caucasian woman who tested positive for a BRCA1 alteration. She had a history of breast cancer at age forty-five, for which she underwent a mastectomy of her affected breast and a preventive mastectomy of her opposite breast. Her family history is notable for two sisters with early onset breast cancer, one of whom also had ovarian cancer and was getting treatment for metastatic ovarian cancer at the time. Margaret also has two sisters and two brothers who have never had cancer. Their mother was diagnosed with breast cancer at age fifty. All of her siblings have adult children, and she has three daughters. Margaret sought genetic testing. She was initially interested in testing to learn about her risk for ovarian cancer and also to gain information for her family. Within six months of learning her results, Margaret opted to have her ovaries removed–a decision influenced by her sister's battle against ovarian cancer.

It was clear from the first meeting with Margaret that she assumed a matriarchal role in this family and that the family was very close. They were also united in family crises, such as the recent death of Margaret's husband and her sister's illness. Within a few months, all of her siblings participated in a group pre-test counseling session (per their request), along with Margaret, and openly shared their hopes and concerns regarding testing. They received their results individually, and all reported that they shared their results, regardless of the outcome, with their children. Some of those children later opted for testing. Margaret's daughters also opted for group counseling, and all received testing. Margaret and her siblings were interested in having the clinical research team assist them in contacting more distant relatives, such as great aunts and uncles and cousins, to invite

them to participate in a free genetic counseling clinical research program. Some of these individuals did participate and were aware of Margaret's experiences, and looked to her for information and support, as did the rest of the family. During follow-up calls, family members often shared their feelings about how relatives were coping with the information. Although subjective, this information allowed the counselor to gain insight into the type of added support or information that could be offered. Margaret's involvement was instrumental in helping the family benefit from genetic counseling, regardless of whether or not they chose to get tested or what their result was if they did get testing.

Analysis: In families that are close-knit, open, and have established lines of communication, the transmission of information about genetic test results may flow with relative ease. Individuals in these families often rely on each other for information, support, and advice about medical decision-making. Furthermore, the individual who initiates testing in such highly motivated families may be central in these activities. These important roles are often beyond the scope of what the counselor is able to provide. However, because there is concern that family members may feel somewhat pressured into getting genetic testing and making certain subsequent decisions, it is incumbent upon the counselor to ensure that individuals are aware of the full spectrum of benefits, limitations, and risks of testing before they decide whether to get tested. The counselor should also be available to help them assimilate and cope with the information.

VI. SUMMARY AND IMPLICATIONS

The quantitative and qualitative (case studies) data presented in this paper have implications, not only in the health care context, but also in the legal arena. The results of both a family-based and clinic-based approach to genetic counseling indicate that the vast majority of genetic counseling participants opted to disclose their test results to immediate adult family members. Consistent with previous research,[39] most of these individuals elected to share the information themselves, rather than have the information disclosed by counselors or other health care providers. Complex psychological and medical issues influenced the decision to disclose, as well as the timing and mode of disclosure. Clinicians and researchers should be sensitive also to cultural influences involved in decisions about family disclosure.

39. *See* Sorensen et al., *supra* note 22, at 421.

Thus, the ability to control the process of disclosure is of great importance to genetic counseling participants. This raises a variety of concerns about the disclosure of genetic information by other sources, such as healthcare providers, insurance companies, or government institutions. From a legal standpoint, the obligations and authority of other sources in disclosure of genetic information is far from clear. For example, two recent legal cases have rendered differing opinions about a physician's responsibility to inform relatives about their risk of developing a genetic disease. The first of these, *Pate v. Threlkel*,[40] concluded that the physician had a duty to warn the patient about the genetic nature of the disease and that the patient could then be expected to warn their family members.[41] It was also stated that disclosure laws would prohibit the physician from warning other family members.[42] The second case, *Safer v. Pack*,[43] reached a differing conclusion. In this case, it was decided that the physician did have a duty to inform the family of their risk of developing a genetic disease.[44] The second case is obviously at odds with both the physician's duty to protect patient confidentiality and with the explicit desires of patients to control the diffusion of their personal genetic information. While this apparent conflict is far from settled, a recent analysis suggests that health care providers have a responsibility to at least inform patients about the implications of their test results to relatives and to encourage (but not advise) patients to share this information.[45] In addition, the American Society of Human Genetics recently published a statement maintaining that "genetic information should be considered as medical information" and further outlining the "exceptional" circumstances under which a health care provider should have a discretionary right to disclose genetic information to at-risk family members.[46] It is not clear from this statement whether disclo-

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40. 661 So.2d 278 (Fla. 1995).

41. *See id.* at 282.

42. *See id.*

43. 677 A.2d 1188 (N.J. Super. Ct. App. Div. 1996), *cert. denied*, 683 A.2d 1163 (N.J. 1996).

44. *See id.* at 1192.

45. *See* Benjamin S. Wilfond et al., *Cancer Genetic Susceptibility Testing: Ethical and Policy Implications for Future Research and Clinical Practice*, 10 J.L. MED. & ETHICS (forthcoming 1998).

46. *See* The American Society of Human Genetics Social Issues Subcommittee on Familial Disclosure, *ASHG Statement: Professional Disclosure of Familial Genetic Information* 62 AM. J. HUM. GENETICS 474, 474 (1998) (discussing that a provider may be permitted to disclose genetic information "where attempts to encourage disclosure on the part of the patient have failed; where the harm is highly likely to occur and is serious and foreseeable; where the at-risk relative(s) is identifiable; and where either the disease is preventable/treatable or medically accepted standards indicate that early monitoring will reduce the genetic risk"

sure of BRCA1/2 test results would fall under this purview.[47] However, even with these considerations, the possibility that government institutions or insurance companies could order and disclose such information poses even greater threats to patient confidentiality and well-being.

The data presented herein also show that females are significantly more likely to disclose genetic information to their relatives, especially when test results are positive and when the relatives are minor children. A particular concern is that such patterns of disclosure may place females at greater risk in the context of family law disputes.[48] For example, it is conceivable that information about a positive mutation status and elevated cancer risk could be used against female mutation carriers in custody disputes or adoption proceedings.[49] This possibility underscores the importance of informing counseling participants about a myriad of potential risks associated with family disclosure beyond the medical and psychosocial risks that are typically addressed.

Although preliminary, other findings from our research suggest that both disclosure and nondisclosure of positive test results to relatives may result in increased psychological distress for the discloser, and possibly for the relatives with whom this information is shared, although data on the latter are not available. Thus, in addition to informing and counseling patients about the medical and legal risks noted above, providers may have an obligation to review the potentially adverse psychological effects of family disclosure. It is arguable that such information should be considered an essential component of the informed consent process which takes place prior to the provision of a blood sample for genetic testing and which is reinforced when results are disclosed.

In the coming years, as genes for several common multiple adult-onset conditions are identified, many more individuals will have the opportunity to learn what their future may hold, and will then have to address the inevitable familial implications of this knowledge. Given the complexities of the medical decision making and psychological adjustment associated with genetic testing, it is hoped that an under-

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or where "[t]he harm that may result from failure to disclose should outweigh the harm that may result from disclosure").

47. *See id.* at 474-83.

48. Telephone Interview with Karen H. Rothenberg, Marjorie Cook Professor of Law and Director, Law and Health Care Program, University of Maryland School of Law (January 7, 1998).

49. *Id.*

standing of the unique determinants and consequences of disclosure to family members can help clinicians provide better counseling to these individuals and will encourage legislators to enact and enforce protections for patient autonomy and confidentiality. This strategy will help ensure that individuals who decide to pursue genetic testing, even in the context of its uncertainties, can obtain maximum benefit while the potential for harm is minimized.