Family members. Moreover, although the DNA sample based on feelings of responsibility and commitment to other Rather, it is a socially situated decision, one that is often the needs and preferences of the individual being tested. The decision to undergo genetic testing is anchored in the individual’s explanatory model or beliefs about the onset, cause, predictability, seriousness, course, and treatment of the genetic condition being tested for. The decision to be tested is rarely an autonomous decision based solely on the needs and preferences of the individual being tested. Rather, it is a socially situated decision, one that is often based on feelings of responsibility and commitment to other family members. Moreover, although the DNA sample used to conduct a genetic test is obtained from an individual, the individual’s decision to undergo testing, as well as the individual’s test results, can have profound and enduring implications for other family members and the family as a system.

According to Sobel and Cowan, the decision whether to undergo genetic testing can be a litmus test, by which family members perceive and judge each others’ loyalty to the family—to its beliefs, values, and rules. To date, few researchers have used a family perspective to examine how families respond to genetic testing. Researchers interested in the genetic testing experience have typically used an individual perspective to examine topics such as attitudes and knowledge about genetic testing, rates of genetic test acceptance, psychological and behavioral consequences of genetic testing, and the disclosure of genetic information to others. Most of the published studies on genetics and the family are epidemiologic studies that focus on molecular biology and the collection of biologic data; they typically do not focus on the family as a social unit that influences and is influenced by the genetic testing experience. In addition, they seldom focus on the complex ethical and social issues that individuals and families encounter during the genetic testing experience.

The purpose of this article is to explore the family experience of genetic testing. Two family stories, or cases, are presented to illustrate how families define and manage the ethical and social issues that emerge during 2 types of genetic testing: mutation analysis for Huntington’s disease (HD) and genetic testing for breast and ovarian cancer susceptibility (BRCA1/BRCA2 testing). Mutation analysis for HD is an example of presymptomatic testing. If the gene mutation for HD is present, symptoms of this progressive, neurodegenerative disorder are certain to appear if the individual lives long enough. BRCA1/BRCA2 testing is an example of predispositional testing. Predispositional testing differs from presymptomatic testing in that a positive result does not indicate a 100% risk of developing the condition being tested for. A meta-analysis of data from 22 studies found that the average cumulative risks in BRCA1-mutation carriers by age 70 years were 65% for breast cancer and 39% for ovarian cancer. Corresponding estimates for BRCA2-mutation carriers were 45% for breast cancer and 11% for ovarian cancer.

The gene mutations associated with HD and hereditary breast and ovarian cancer are inherited in an autosomal dominant pattern; thus, each offspring of an individual found to carry one of these mutations has a 50% chance of inheriting the same mutation. Currently, there are a variety of management options for individuals found to carry a BRCA mutation, such as increased surveillance, risk-reducing surgery, and chemoprevention. Management options for individuals who test positive for the HD gene

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**INTRODUCTION**

Genetic testing is increasingly being recognized as both a family and an individual experience. There is growing recognition among clinicians and researchers that families influence and are influenced by how individual family members make sense of, respond to, and use the information they receive during the genetic testing experience. The decision to undergo genetic testing is anchored in the individual’s explanatory model or beliefs about the onset, cause, predictability, seriousness, course, and treatment of the genetic condition being tested for. The decision to be tested is rarely an autonomous decision based solely on the needs and preferences of the individual being tested. Rather, it is a socially situated decision, one that is often based on feelings of responsibility and commitment to other family members. Moreover, although the DNA sample used to conduct a genetic test is obtained from an individual, the individual’s decision to undergo testing, as well as the individual’s test results, can have profound and enduring implications for other family members and the family as a system.

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The gene mutations associated with HD and hereditary breast and ovarian cancer are inherited in an autosomal dominant pattern; thus, each offspring of an individual found to carry one of these mutations has a 50% chance of inheriting the same mutation. Currently, there are a variety of management options for individuals found to carry a BRCA mutation, such as increased surveillance, risk-reducing surgery, and chemoprevention. Management options for individuals who test positive for the HD gene
mutation are much more limited; treatment that will alter the natural history of HD has yet to be found.10

The 2 families presented in this article participated in an ongoing study, Family Experience of Genetic Testing: Ethical Dimensions,28 in which 118 family members from 67 families have participated. These 2 families were purposefully selected because their stories exemplify the complexity of the genetic testing experience. In addition, the story of the family living with HD shows how negative consequences can occur for the individual tested, other family members, the marital relationship, and the family system, even when the test results indicate that the individual does not carry a deleterious gene mutation.

BACKGROUND

Recent advances in genomic research have dramatically altered the landscape of genetic testing. Genetic testing is currently available for more than 1000 conditions, and it is increasingly considered an integral component of mainstream health care.29 Furthermore, many genetic tests are now being marketed directly to consumers.30 The increased availability of genetic testing may result in a wider distribution of the benefits of testing, such as decreased uncertainty, the chance to avoid passing on a gene mutation to future generations, increased psychological well-being, and greater awareness of available treatments or risk-reducing strategies, but it may also contribute to an increase in the risks associated with genetic testing, such as decreased individual and family well-being; increased depression, anxiety, guilt, stigmatization, discrimination, and family conflict; and unnecessary or inappropriate use of risk-reducing options.10

Although few researchers have used a family perspective to study the family response to genetic testing,18,20–24 there has been a notable increase in research concerning families and genetic testing during the past decade. Genetic testing and the family was the focus of a special issue of Families Systems & Health in 1999. At that time, McDaniel and Campbell7 described families and genetic testing as “an area yet to be studied, one that cries out for clinical innovation and scientific inquiry.” In the same issue, Rolland8 noted that with the rapid integration of genetic advances into clinical practice, the need for a family-based systemic approach has never been more urgent. Green4 indicated that clinicians will have to understand the familial impacts of genetic information and the ways in which disputes over genetic information may reflect or intensify other, deeper tensions in families and ethnic communities. Feetham2 argued, “the paucity of attention to family systems and family relationships in the collection and dissemination of genetic information places family members and families at increased risk.”

More recently, in a special issue of the American Journal

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METHODS

Guiding Framework

The guiding framework for this research was the family management style framework (FMSF) developed and refined by Knafl and colleagues.31–33 The 3 major components of the FMSF are definition of the situation, management behaviors, and perceived consequences. A family member’s definition of the situation is the subjective meaning they attribute to important elements of their situation. Definitions of the genetic testing situation change over time and are based on multiple factors, including past experiences with health care providers, family rules and boundaries, family explanatory models of genetic conditions, and culturally based beliefs about being tested for and living with genetic conditions. Management behaviors are defined as discrete behavioral accommodations that family members use to manage their situation on a daily basis. Like definitions of the situation, management behaviors can change over time. Perceived consequences are the actual or expected individual, family, and illness outcomes that shape management behaviors and affect the subsequent definition of the situation. FAMILY MANAGEMENT STYLE is viewed as the configuration formed by individual family members’ definition of their situation, the management behaviors individual family members engage in, and perceived consequences of the situation.

The FMSF takes into account the definitions of the situation, the management behaviors, and perceived consequences for all or a subset of individual family members.31–33 The FMSF can be used to focus broadly on all aspects of a health-related challenge or more narrowly on a circumscribed aspect of the challenge, such as the ethical dimensions. Either way, the framework directs clinicians and researchers to consider both how individual family members and the family unit as a whole actively manage health-related challenges.

Design

A case study approach was used for this research. According to Stake,34 “a case study is both a process of inquiry about the cases and the product of that inquiry.” Case studies are of interest to clinicians and researchers both for their uniqueness and their commonality. Case study methodology fits well with the application of the FMSF, because it allows for the in-depth descriptions of the major compo-
nents of the FMSF. By exploring the genetic testing experiences of these 2 families through semistructured interviews with multiple family members, it was possible to gain a more holistic picture of how individuals and families respond to genetic testing.

Procedure
Institutional review board approvals of the study were obtained before its initiation. Families were recruited from a variety of sites in the Eastern and Mid-Atlantic United States. Families were informed that the investigator for the study would like to interview at least 2 individuals from each family. Families who expressed interest in participating in the study were given an opportunity to discuss the purpose of the study and requirements for study participation before they signed the consent form. Interviews were conducted at a time that was convenient for the family members being interviewed. Families were given the option of being interviewed individually, or together. Families were also given the option of being interviewed in person or by telephone. Both of the families presented in this article chose to be interviewed individually, and 5 of the 6 family members interviewed chose to be interviewed by telephone.

Sample
The majority of the 67 families in the larger study are white, 2-parent, middle-to-upper income families. Names and other aspects of the 2 families presented in this article have been altered slightly or omitted to protect confidentiality (see box at right).

Interview Guide
The interview guide for the semistructured interviews included open-ended questions about 1) how family members define the genetic testing experience in general, and the ethical and social issues that have emerged during genetic testing; 2) the management behaviors family members used during the genetic testing experience; 3) how family members dealt with ethical problems that emerged during the genetic testing experience; and 4) the impact of the genetic testing experience on individual and family functioning. Examples of interview questions include the following: “Tell me what it was like for you (or your family member) to undergo genetic testing.” “How did the results compare to the results you expected?” “What kind of decisions did you have to make after you were informed of the results?” “Tell me about the hardest decision that you had to make.” “What advice did you receive from health care providers?”

Data Management and Analysis
All of the interviews for the larger study were recorded on a digital recorder, transcribed verbatim, and processed further by using Atlas.ti, a software program designed for the management, coding, and retrieval of narrative data. By using well-established qualitative analysis techniques, coding categories and themes were developed through a process of constant comparative analysis, a technique in which the investigator simultaneously collects information through interviews, reads interviews as individual cases, disassembles interviews through coding, rearranges coded categories into patterns, and reintegrates the patterned categories into a conceptualization that encompasses the experiences of all participants. Consistent with the guiding framework and the aims of the larger study, coding categories focus on how family members define the genetic testing experience, how family members manage ethical issues associated with genetic testing, and the consequences of the genetic testing experience.

By following guidelines developed by Miles and Huberman, a data matrix was constructed to summarize the responses of the individual family members across the defining, managing, and consequence themes. The majority of these themes were previously identified by Knafl and colleagues. However, a number of the themes developed by Knafl and colleagues were modified to reflect the fact that in families affected by adult-onset disorders, such as HD and hereditary breast or ovarian cancer, it is common for multiple family members from more than one genera-

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**FAMILY A**
Family A has a strong family history of HD; multiple family members from at least 4 generations have been diagnosed with HD. In one generation, 8 of the 11 siblings had HD. The 3 family members who were interviewed include Joe, the husband of a woman who was beginning to show symptoms of HD prior to her unexpected death at the age of 58, and 2 of their 3 adult daughters. The 2 daughters who were interviewed, Renee and Susan, underwent mutation analysis for HD when they were in their late 30s or early 40s, and both tested negative for the deleterious HD mutation. The third daughter, Julie, has chosen not to undergo testing. All 3 daughters were married and had children prior to becoming aware that their mother had HD. They knew that their grandmother had HD, but their mother had been told by their family physician that HD was “nothing she needed to be concerned with—it was something that mothers could pass onto their sons, but not their daughters.”

**FAMILY B**
Family B had no family history of breast or ovarian cancer prior to 1 of the 6 adult siblings, Brenda, being diagnosed with breast cancer at the age of 30. Within 2 years after Brenda’s diagnosis, Mary was diagnosed with breast cancer at the age of 41. Six years later, Brenda was diagnosed with breast cancer for the second time. Shortly after this, the 4 sisters decided to go as a group to a session with a genetic counselor. Brenda and Mary chose to undergo BRCA1/BRCA2 testing, and they both tested positive for a BRCA1 mutation. The other 2 sisters, Linda and Arlene, chose to undergo intensive surveillance rather than be tested. All 4 of the sisters are married or partnered; the brothers are not married. None of 6 siblings have children. The family members who were interviewed include Brenda, Mary, and Linda.
tion to be tested for and diagnosed with the condition. Because of this, members of the family of origin may have a stronger influence on how family members define and manage the challenges associated with being tested for and living with a genetic condition than do members of the immediate family. For example, siblings may play a bigger role in the genetic testing experience than do spouses.

**FINDINGS**

**Family A: Negative Consequences Can Occur Even When the Test Results Are “Good”**

Finding out that Renee and Susan did not carry the HD mutation was great news, especially for Joe, the father. According to Renee, the first daughter to be tested, when she told her father that she had tested negative, “He just started crying and hugging me. I mean he cried for probably half an hour. He was just so happy.” When Joe was asked what it was like to hear that 2 of his daughters had tested negative, his response was, “I am not sure of the full meaning of euphoria but I am going to use it because it has been like living on cloud nine.” Although the consequences of genetic testing were primarily positive for Family A, there were negative consequences. Unexpected negative consequences are the focus for this family’s story.

**Definition of the Situation**

Both Renee and Susan were certain that their test results would be positive, so they planned their lives accordingly. Renee noted that since becoming aware of her risk for developing HD, she had planned her life in 5-year segments. Prior to undergoing genetic testing, she had decided that when she tested positive for HD, she was going to start talking to people in the legislature about HD. She planned to be the “Michael Fox of HD.” Finding out she was negative for the HD mutation “burst her bubble.”

“It totally messed up my whole life because, all of a sudden, I had to figure out what I was going to do with the rest of my life. I didn’t go to college because of HD. I didn’t do this because of HD. I didn’t do that because of HD. Now I had my whole life ahead of me and I might live to be 100 . . . I had to figure out what I was going to do. My husband, on the other hand, he had prepared himself for taking care of me. Now, I didn’t need to be taken care of anymore . . . His bubble burst too; he wanted to take care of me. I think the negative test result impacted us worse than what a positive test result would have.”

Although getting a negative test result did not seem as difficult for Susan and her immediate family as it was for Renee and her family, it did force Susan to come up with a new plan for how she was going to help individuals and families affected by HD. Like Renee, Susan was sure her tests results would be positive. She had decided that once she tested positive for HD, she would get involved in drug trials for HD. When she tested negative, this was no longer an option.

**Management Behaviors**

Family members differed in how they responded to the negative test results. Renee reported having difficulty telling her sisters.

“Telling my sisters was absolutely the worst because they were still at risk. I just didn’t want to tell them because I knew they were still at risk. I was afraid that they would not be okay. I didn’t want to tell them. I didn’t want to be okay without them being okay.”

Renee also reported experiencing marital problems after finding out her results.

“My husband was acting real strange and he just wouldn’t talk at all. And that’s kind of how he is when he doesn’t want to deal with something, he doesn’t talk. And he was getting real withdrawn . . . I mean it got to that point where we were going two separate ways. So I said, ‘okay I want a divorce, get out of here. Go on. Move on.’ He refused. He slept on the couch for about 3 weeks. He just refused to leave. Finally, I said, ‘we are going to counseling.’ So we started going to counseling.”

Susan decided that if she could not help other individuals and families by enrolling in HD drug trials, she could do other things. For example, she got involved in fundraising for HD research, and she started giving presentations about HD. She has also tried to encourage and facilitate open communication about HD in both her immediate and extended family.

Julie, the sister who chose not to be tested, expressed skepticism when she was informed that her sisters had tested negative for the HD mutation. Her response to Renee was, “How do you know they are right . . . they might have made a mistake.” When Julie’s husband asked Joe if he could help persuade Julie to undergo testing, Joe declined. He told Julie’s husband,

“No, I am not going to do that. I think that is a decision she has to make on her own. And now, it is totally up to you. If you think it is going to be a factor in your marriage or if you think you could not handle it, you had better be up front with her. If you are going to accept it in sickness and health, wait and see.”

**Perceived Consequences**

Given that both Renee and Susan tested negative for the HD mutation, none of their children or grandchildren are at risk
for developing HD. Because of this, HD no longer seems to be the dominant focus in either Renee’s or Susan’s immediate family. However, because Julie’s mutation status continues to be unknown, and there are a number of family members in the extended family who are symptomatic for HD, it is likely that HD will continue to be part of Family A for many years to come.

Susan reported that one of the hardest things she had to do after receiving her test results was go to her uncle’s funeral. He died on the same day she found out her results. He had been suffering from HD for many years, and in Susan’s opinion, “He wasn’t really living—he was just existing.” Seeing family members who were symptomatic for HD but had not been tested yet made Susan feel guilty. It also made her question why they had HD and she did not. Her uncle’s wife as well as her cousins reassured her that everyone was thrilled she had tested negative.

Family B: One Family, Many Different Responses

Genetic testing for breast and ovarian cancer susceptibility was defined as a family experience in Family B. Yet, the right of each member of the family to make his or her own decisions about genetic testing and available management options was clearly respected. The 3 sisters who were interviewed described both their immediate and their extended family as emotionally close and very supportive. Spouses were described as supportive but not that involved in decision making about genetic testing and risk-reducing options.

Definition of Situation

When asked about the genetic counseling session, Linda said, “We all went with Brenda and Mary to show our support. We went as a family.” Brenda noted,

“We learned in that session and shortly thereafter that even though we are blood sisters, raised together in the same household, in a very close family, with the same parents, we had totally different responses to this. Totally different!”

In terms of their views about whether there might be a BRCA mutation in the family, Mary felt “there probably was one” and Brenda was “on the fence.” Linda, the sister with the strongest background in science, doubted that a mutation would be found because “it didn’t follow the standard pattern.”

Management Behaviors

The 4 sisters differed in how they used the information they received during the genetic counseling session. Brenda and Mary, the 2 sisters with a history of breast cancer, decided to undergo BRCA testing and risk-reducing surgery. The other 2 sisters chose not to undergo BRCA testing. Linda indicated that it really would not change anything if she underwent genetic testing.

“I mean, really my only recourse other than getting screened every 6 months like I am now is to have a bilateral mastectomy and a hysterectomy. I am just not prepared to do that right now. Never having had the disease, it’s hard to prophylactically remove organs that are perfectly healthy.”

Both Mary and Brenda chose to undergo risk-reducing surgery, but Mary chose to have reconstructive surgery and Brenda did not. When asked how she made her decision, Brenda said,

“I had given each breast a chance. But after the second diagnosis of cancer, they had to go, Playboy or not. I knew from the start, even when I was first diagnosed, a mastectomy would have been a big deal to me because I had Playboy boobs and I loved my boobs and they were the only thing I loved about myself. And that was back in the times when I thought looks really mattered . . . I always thought that’s how I got through it so good the first time, because I didn’t have to face a mastectomy. I always knew that if I had a mastectomy, I wouldn’t have one. I’d have two because I didn’t want to be lopsided and I’d never have a prosthesis or reconstruction. That was my gut of guts. Mary’s gut of guts was ‘I can’t wake up with nothing.’”

As with the decision about whether to undergo testing, Brenda and Mary respected each other’s right to make their own decisions, even if they did not agree with or understand the decision. According to Brenda,

“We respected each other’s opinions, but I will probably never totally understand her decision and she’ll never understand my decision. And we found that’s okay. These decisions are so highly personal that even a sibling can’t relate to what you are going through. And we had to learn that. It was like ‘oh my gosh.’”

Perceived Consequences

When asked if it had been helpful to go to the genetic counseling session as a group, Linda replied, “Yes, for us it was helpful. I mean, we are all pretty close, and for us, this is kind of [a] family thing.” Mary noted that “it wasn’t bad.” Brenda thought differently about their “group approach” to genetic counseling.

“I think it was just hard with the 4 of us because we found that we all had such different responses, and I think it was very hard on the doctor and the genetic counselor because here they were being plummeted by four individuals . . . my little sister didn’t ask a whole lot, she was just kind of there in shock I think.
And then you had the rest of us—two scholastic research people who were just going at them with these questions and you got me trying to lighten up the air, joke about it a little.”

Later in the interview, Brenda noted that when she got her test results she went alone. She had decided it was wonderful and great to have lots of family support, but it was also exhausting.

Brenda and Mary indicated that they were pleased with the decisions they had made about risk-reducing and reconstructive surgery. Linda, on the other hand, seemed to be having second thoughts about her choice of increased surveillance, rather than genetic testing. According to Brenda, “Linda is rethinking her decision. She is kind of sick of the surveillance stuff already.” When Linda was asked if she ever thought she would change her decision about testing, she indicated that once she retired, her medical benefits would not be as good. Because of this, it may become too expensive for her to undergo the type of intensive screening she was currently getting. She went on to acknowledge that getting tested might make it possible for her to stop the intensive screening.

CONCLUSION

Genetic testing is no longer the sole purview of genetic specialists. It is rapidly becoming part of routine health care. Until recently, most individuals and families who underwent genetic testing were tested at tertiary centers by genetic specialists who followed carefully developed protocols. Unfortunately, many of the clinicians who are now offering genetic testing may not be following these protocols. For example, at a recent HD support group meeting, a young man shared his story of HD testing. He knew his grandmother had died of HD, but no one else in his family had been tested. His father, as well as his aunts and uncles, were adamant about not getting tested. He and his wife were wanting to think about having children, so he decided that unlike the rest of his family, he did want to be tested. He called his family physician, told him what he wanted, and the physician ordered testing for HD. There was no counseling before or after the testing. Moreover, there was no discussion of possible ethical and social issues. Fortunately, the test results were negative. The young man and his wife were relieved, and his father’s right not to know had not been violated.

The above situation, as well as the 2 family stories presented in this article, vividly illustrate why it is so critical that midwives and other clinicians become proficient in assessing how families define and manage the ethical and social issues that emerge during genetic testing. Use of a family framework, such as the FMSF, which takes into account both the perspective of the individual and the family as a whole, may help in the development and implementation of tailored, culturally sensitive interventions that not only lead to improved outcomes, but save valuable resources such as time and money. Use of the FMSF can also help to identify individual, family, and sociocultural factors that contribute to optimal individual and family outcomes.

In conclusion, our current understanding of the family experience of genetic testing is limited. Use of family frameworks, such as the FMSF, can greatly enhance our understanding of how individuals and families respond to genetic testing. The key is that we need to use them. We need to shift the focus from an individual to a family perspective, and we need to do it now.

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