

Complexities in Cancer Risk Counseling: Presentation of Three Cases

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Complexities abound in the identification and management of families at increased risk for inherited forms of cancer. One of the ways to learn as a profession how best to provide cancer risk counseling (CRC) is to share counseling experiences. Such cases can provide insight into the issues raised by families and ways in which genetic counselors have handled complex situations. Here we describe three CRC cases initially presented at the 1995 American College of Medical Genetics meeting. The first case involves balancing the importance of informing a family of the presence of an inherited cancer syndrome with the family's right "not to know." The second case illustrates the difficulties in assisting an individual to make medical management decisions in the face of uncertain risk information. The third case describes the complex interactions with a woman before and after her decision to have prophylactic surgery.

KEY WORDS: breast cancer; cancer risk counseling; cancer genetics; prophylactic mastectomy.

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INTRODUCTION

Cancer risk counseling (CRC) is a communication process concerning an individual's possible increased risk of developing specific forms of cancer. The increased risk may stem from inherited susceptibilities, lifestyle choices, or exposures to carcinogens. CRC includes: obtaining detailed family, medical, and lifestyle histories, documentation of cancer-related diagnoses, pedigree analysis, risk assessment and counseling, and discussion of options for early detection and prevention (Schneider, 1994; Kelly, 1991).

The collection of a detailed cancer history involves obtaining information about family members who have developed cancer, including site of cancer and age of onset. Unaffected family members should also be included in the pedigree, with either current age or age and cause of death. Following the collection of a cancer history, information about cancer diagnoses are confirmed whenever possible through retrieval of pathology reports. The discussion then turns to risk assessment. Individuals are given estimates of their risk for developing specific forms of cancer and risk for carrying a germline mutation in a cancer susceptibility gene. Certain families fit the classic pattern of hereditary cancer syndrome, e.g., multiple family members with similar or related cancers occurring at earlier than average ages in two or more generations. Some families have a very low likelihood of carrying a dominant cancer susceptibility gene; e.g., pedigree includes a few cases of cancer in relatives who are over age 65. Families who have some, but not all of the features consistent with a hereditary cancer syndrome pose the greatest challenges to genetic counselors in terms of risk assessment. For example, breast cancer is one of the cancers that can occur as an isolated case, in familial clusters, or primarily but not solely due to an inherited dominant gene mutation.

One goal of providing risk information is to assist the individual in making more informed and thoughtful decisions about cancer surveillance or prevention. Counselors may be called upon to discuss the pros and cons of prophylactic surgery with women in high risk families. Psychological ramifications of the risk estimate information and medical management options and the possible need for further support services must also be considered.

Consultands enter into CRC with a variety of medical, psychological, and genetic counseling needs. The genetic counselor has some role in the assessment and management of all three domains (Peters and Stopfer, 1996). We present three breast cancer cases which deal with specific counseling challenges. Each case is presented in the following manner: indication for referral, cancer history, major counseling issues, and case

resolution. The names used in the cases have been changed to protect patient confidentiality.

**CASE 1: COUNSELING FOR LI-FRAUMENI SYNDROME:
THE RIGHT NOT TO KNOW**

Indication for Referral

Barbara was referred to our Cancer Risk Evaluation Program when her sister Marsha was diagnosed with breast cancer. Marsha's medical oncologist became concerned when he learned that Marsha's brother had developed a brain tumor and that other relatives had also had cancer. Marsha relayed this information to her sister, who was interested in learning her own risk for developing breast cancer. Barbara had also read about the discovery of the *BRCA1* gene, and wondered if she could be tested for a *BRCA1* mutation in order to clarify her own risk for breast cancer. She also indicated that she was interested in learning exactly what steps she could take to prevent herself from developing breast cancer.

Cancer History

During the initial telephone conversation, Barbara asked about the details of the Cancer Risk Evaluation Program and what kind of information it would give her. She was informed that a detailed cancer history would be collected, followed by a discussion of the history's implications for the family. At our request, she set out to document each occurrence of cancer in her family prior to her appointment. As shown in Fig. 1, two of Barbara's sons had developed cancer; one had leukemia and one had a soft tissue sarcoma. In addition, she had an uncle with stomach cancer, who had a daughter who died at age 6 of an osteosarcoma. She had only recently learned about the specific cancer diagnoses in her uncle and cousin.

Major Counseling Issues

At the session, Barbara said that she had often wondered why both her sons had developed cancer. The family physician had told her repeatedly in the past that it was just "an unfortunate coincidence" that both of her sons had developed cancer. She had never worried about her own risk of developing cancer, until her sister was diagnosed with breast cancer.

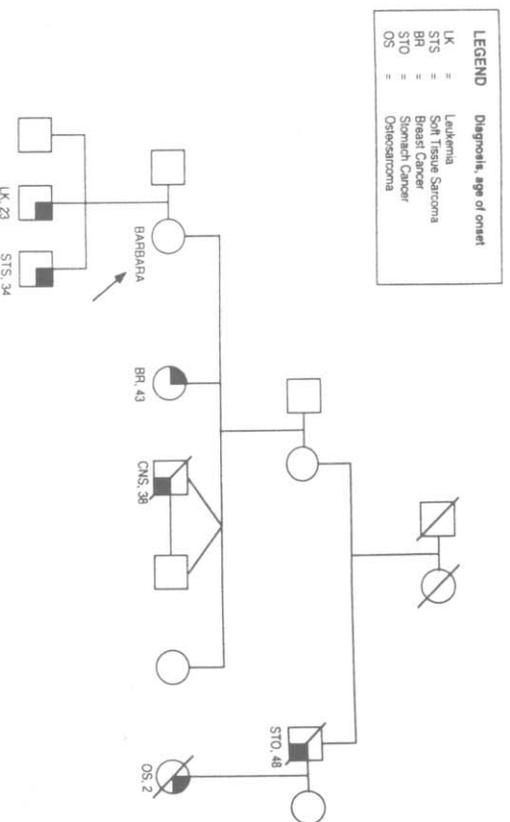


Fig. 1. Barbara's history which is suggestive of Li-Fraumeni syndrome.

The pattern of cancer in this family is consistent with Li-Fraumeni Syndrome (LFS) and Barbara was counseled that there could be a common underlying cause for all the different types of cancer in her family. She seemed surprised to hear this. In response to her query about whether she could be tested for an alteration in the *BRCA1* gene, she was informed that the pattern of cancer in her family seemed more consistent with a different gene. She did not ask for the name of this gene or for an explanation as to why our team believed it to be consistent with her cancer history. We discussed with Barbara the fact that some individuals want to learn about inherited factors associated with cancer susceptibility, while others do not. We also explained that, regardless of whether she wanted this information or not, we could suggest a tailored cancer risk management program for her. Barbara was encouraged to let us know how much information she wanted. At no time in the discussion did Barbara ask what her risks were for carrying a gene mutation and our team did not volunteer that, given her placement in the pedigree, she had a high likelihood of being an obligate carrier.

Instead, Barbara focused her questions on the testing process and how the genetic test results might be beneficial to her. We explained to her that the testing process would begin by testing one of her relatives who had developed cancer, and that if a mutation were identified, then she and other relatives could be offered predisposition testing.

We also reviewed her current breast cancer surveillance, which included annual mammograms and biannual clinical breast exam and concluded that this was a sensible plan to follow. She was also being seen annually by her family physician. We did mention that her current surveillance pattern would remain unchanged. There are no special screening recommendations for individuals at risk for LFS except for increased breast surveillance (Li et al., 1992). She stated emphatically that prophylactic mastectomy was not something she would ever consider.

Counseling concluded with a long discussion about whether she felt genetic testing could benefit her sons. Barbara revealed that one of her sons, who had been treated 8 years ago for a sarcoma, still worried about the possibility of another cancer diagnosis. She also expressed her hesitation to involve her sons in the testing process, since she did not want to burden them with worrisome, and perhaps unwanted, information.

Case Resolution

Barbara had been under the impression that her genetic counseling session would focus solely on her risks of developing breast cancer and the option of predisposition testing for a breast cancer susceptibility gene. When it became clear to her that she could be at increased risk for other forms of cancer, she became noticeably less interested in hearing more information. If Barbara were found to carry a *p53* germline mutation, she would be at 90% risk of developing breast cancer or another LFS related cancer by age 70 (Li et al., 1992). Although Barbara was told of the possible increased risks of other malignancies, she was not given any specific risk figures.

Our multidisciplinary team, consisting of a genetic counselor, oncologist, and oncology nurse discussed and concluded that, since her surveillance would not change, Barbara had the right “not to know” that her family could have LFS.

About 1 week after the clinic visit, the genetic counselor called Barbara to inquire whether she had any questions or would like to schedule a follow-up appointment. At this time, Barbara explained that she had decided in not to proceed further with the testing process and was not interested in learning more exactly about the unifying link that was causing her family members to be susceptible to cancer. Ultimately, Barbara felt that the anxiety associated with learning she could be at increased risk for several forms of cancer was greater than the potential medical benefits.

During her counseling session, Barbara had expressed interest in having her brother seen in our program. He was reportedly a monozygotic

twin of the brother previously diagnosed with a brain tumor—a second possible obligate carrier. Barbara was told that we were happy to meet with him, but that he would need to make the initial contact with our center. When the brother called, we discussed the reasons supporting learning and not learning about the possible cancer syndrome in the family. During this lengthy phone conversation, the words “Li-Fraumeni Syndrome” were never used and he, like his sister, never asked about the specific name of the hereditary cancer syndrome. The brother stated that he would think about participating in our genetic testing research study, but has so far not initiated any further contact.

CASE 2: MAKING MEDICAL MANAGEMENT DECISIONS WHEN CANCER RISK ASSESSMENT IS UNCERTAIN

Indication for Referral

At age 45, near the age when her paternal aunt was first diagnosed with breast cancer, Sandy requested prophylactic surgery. Based on the family history (see Fig. 2), her request was denied and Sandy was reassured by medical personnel that her risk of breast cancer was not substantially

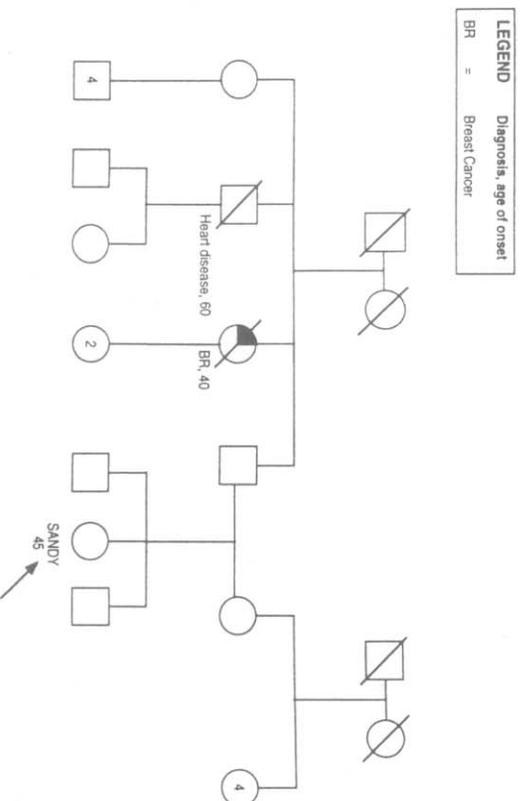


Fig. 2. Sandy's initial history when prophylactic mastectomy was requested and denied.

increased. Five years later, Sandy was diagnosed with breast cancer and had a unilateral mastectomy. She was then referred for genetic counseling to consider prophylactic removal of the remaining breast. She was interested in having a prophylactic mastectomy on the remaining breast because of her concerns about developing another breast cancer and the failure of standard surveillance measures to detect her initial breast cancer at an early stage. The oncologic surgeon was reluctant to perform the surgery, stating that it was not routinely done for patients with sporadic, unilateral cases of ductal carcinoma. For this reason, Sandy was referred for a genetic consultation to discuss issues related to prophylactic mastectomy.

Cancer History

The initial contact with Sandy was by telephone and the genetic counselor asked Sandy to explore her family history for any types of cancer and obtain verification of the diagnoses where possible. When she came for her counseling appointment, a pedigree was constructed which included a mother, four sisters and a maternal grandmother who were all cancer-free. However, Sandy related that her paternal aunt was diagnosed with breast cancer at age 40. As a result of her inquiries, Sandy learned that her paternal grandmother also developed breast cancer at age 45, which was new and disturbing information to Sandy (see Fig. 3).

Major Counseling Issues

The central focus of our discussion was whether, based on the family history, a prophylactic mastectomy was an appropriate choice given her level of risk and whether Sandy understood the medical, social, and personal ramifications of this procedure.

The risk assessment for this family included a discussion of three possibilities:

1. *There was a dominant cancer susceptibility gene in this family:* The three diagnoses of pre- or perimenopausal breast cancer spanning three generations were suggestive of hereditary breast cancer. Histories of breast cancer on the paternal side of the family remain underreported and the significance of such a history may go unrecognized by other health care professionals. In this family, the lack of breast cancer in Sandy's mother or four sisters was taken as evidence that Sandy's cancer was sporadic. Additionally, kindreds that are small or consist of more male than female relatives can mask a dominant inheritance pattern of cancer.

mastectomy. Since efforts to obtain documentation of the cancer diagnoses for other relatives were unsuccessful, the risk counseling was compromised and would make a poor basis for offering prophylactic surgery. Sandy was also informed that prophylactic mastectomy would not eliminate her risk and that there was very limited data available to tell her what her remaining breast cancer risk would be after surgery (Wapnir, 1990). Despite this lack of information Sandy continued to want the surgery and so our discussion turned to other issues.

Five years earlier, Sandy had requested prophylactic surgery from her breast surgeon. The surgery had not been performed, because her risks of breast cancer due to her family history were felt to be at population levels. Thus, the idea of having prophylactic surgery was not a new one for Sandy, but rather one that she had considered even before her breast cancer diagnosis. Not surprisingly, she also expressed bitterness that the procedure had not been done when initially requested and frustration that her physicians were, in her opinion, still not taking her breast cancer risks seriously enough. A second issue that arose and needed to be handled diplomatically was the discrepancy between the surgeon and genetic counselor about the potential importance of a positive breast cancer history in the paternal lineage.

Furthermore, Sandy's breast cancer, diagnosed at Stage 2, was discovered by herself—1 month after a mammogram which had been read as normal. This had left her feeling “let down” by the standard surveillance methods and more eager to minimize her risks by having surgery. In addition, she had cystic breasts and had already undergone two biopsies in the year since her diagnosis, which she indicated were highly stressful.

The counselor and patient also explored options other than prophylactic surgery including one-on-one breast self exam counseling, chemoprevention (e.g., Tamoxifen), and modifying lifestyle (e.g., changes in diet and exercise). At the current time, none of the risk reduction strategies have been proven effective in a woman with a germline mutation in a breast cancer susceptibility gene. Some effort was spent reviewing the pros and cons of the various risk reduction strategies and exploring how comfortable Sandy would feel if prophylactic surgery were not performed. Sandy continued to favor surgery, despite understanding the real, but unquantified remaining risk of breast cancer.

The counseling discussion included information about possible increased risks of ovarian cancer, surveillance options, and the pros and cons of prophylactic oophorectomy. Sandy was interested in being monitored for ovarian cancer, but not in having her ovaries surgically removed. It is not uncommon for women in high risk families to focus on their risks for developing one specific form of cancer. Although Sandy understood her potential increased risk for ovarian cancer, she was clearly much more con-

cerned about her breast cancer risks, because of her personal and family history of breast cancer.

Because Sandy had undergone a mastectomy already, it was possible to explore her adjustment to the loss of one breast. She indicated that she was coping very well post-surgery and that her feelings about herself had not diminished in any way. She felt that she and her husband had dealt successfully with the crisis of her breast cancer diagnosis and that he was supportive of her decision to have prophylactic surgery.

Case Resolution

At the time Sandy was counseled, *BRCA1* or *BRCA2* testing was not available except under research protocols with strict eligibility criteria. Although we discussed that genetic testing would be possible in the future, Sandy was not interested in delaying her decision about prophylactic surgery until such testing became available. Sandy felt that her decisions about wanting surgery would not change, regardless of her results. Thus, she elected to have a prophylactic mastectomy, which was performed by the referring surgeon.

In follow-up with Sandy, she has expressed satisfaction with her decision. Sandy indicated that she felt “more balanced and normal” post-surgery. She also expressed feelings of relief and satisfaction that her concerns about her family history had been heard. She seemed to have less anxiety about developing another cancer, even though she was counseled that, if she had a genetic predisposition, she could have higher risks of other malignancies, especially ovarian cancer. It is unclear whether she will at some point decide to pursue genetic testing, because she feels that she has already dealt with her own risks and does not have any children. Sandy was told to recontact the genetic counselor if she becomes interested in pursuing genetic testing at some point in the future.

CASE 3: COUNSELING A WOMAN ABOUT PROPHYLACTIC MASTECTOMY

Indication for Referral

Jane was self-referred to a multidisciplinary breast center for CRC 1 year after her mother had died from recurrent breast cancer. She was interested in determining her chance for getting breast cancer and learning about the risks and benefits of risk reduction and early detection options.

In her words, her main motivation was to “avoid dying of breast cancer like my mother.”

Cancer History

At the time of the session, Jane was 33 years old, healthy, and married, with no plans to have children. She had married her 52-year-old husband, shortly before her mother died. Her father had died at age 52 when she was an adolescent. Jane’s family history was deceptively simple on intake with one first-degree relative with breast cancer; however, further inquiry provided a more complex family history of cancer (Fig. 4). Jane has three sisters, none of whom have developed cancer. Their mother initially had breast cancer at 44 years and almost 20 years later developed metastases and a second breast cancer primary. She died several years thereafter. Jane had been intensively involved in the caretaking of her mother. She also had three distant relatives in two generations who reportedly had breast cancer; at least one case was premenopausal. While there were voluminous medical records available on the mother, none were available on the other affected relatives.

Major Counseling Issues

Extensive CRC was provided as described elsewhere (Kelly, 1991, 1992a; Peters, 1994a,b; Schneider, 1994). In Jane’s case, multiple sessions were spent discussing the genetic, epidemiological, statistical, and medical aspects of breast cancer, in large part because intellectual mastery contributed to Jane’s sense of control over her breast cancer risk. Genetic susceptibility testing for *BRCA1* or *BRCA2* mutations was not available at the time Jane was counseled. If DNA testing had been possible, then discussions would have also included the appropriateness of testing as well as the perceived risks and benefits.

During the counseling session Jane was noted to have a cognitive approach to problem-solving and was focused solely on her risk of breast cancer. Her overall mood was anxious, as suggested by her very rapid, pressured speech and attention to detail. Jane’s anxiety about having breast cancer led her to strongly consider having prophylactic mastectomies.

When Jane was seen at the community breast center where the genetic counselor was working at the time, individuals interested in prophylactic surgery were asked to wait 6 months before proceeding with surgery. This waiting period allowed Jane to fully consider the ramifications of her decision as well as explore other options for risk reduction and early detec-

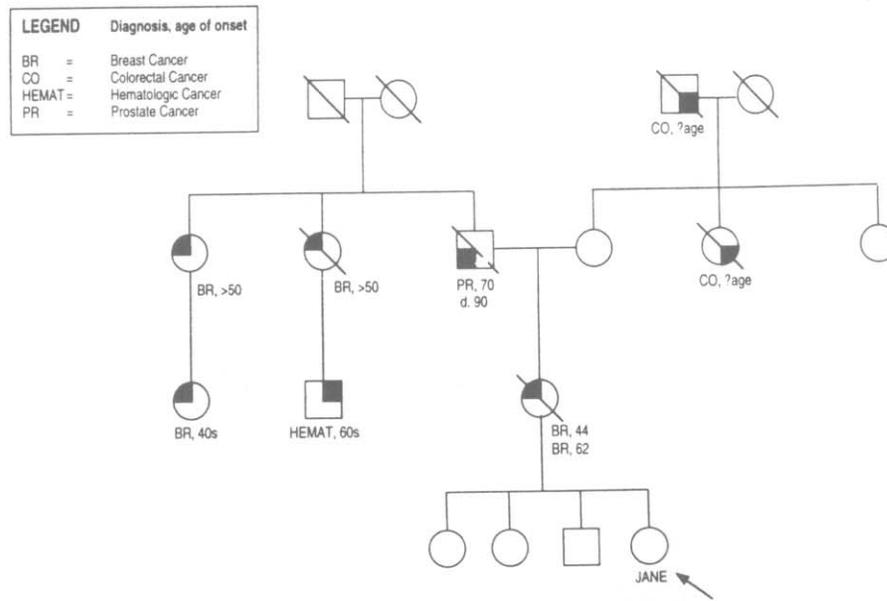


Fig. 4. Maternal history of breast cancer leading Jane to consider prophylactic mastectomy.

tion. Table I describes the many issues that can be included in a discussion about prophylactic surgery. Concurrent with the ongoing risk analysis and genetic counseling, other medical, psychological, and nutritional evaluations and interventions were occurring. For example, the coordinator of the Breast Cancer Prevention Trial met with Jane to discuss the possibilities of chemoprevention using Tamoxifen. In this case, Jane's age precluded her from being eligible in this national study. A nutritionist was available for consultation regarding lifestyle and dietary changes. Additionally, Jane received instruction and coaching in breast self-examination (BSE) by an oncology nurse. She also chose to meet with three surgeons before deciding about having surgery, what type of surgery to have, whether to have reconstruction and if so, what type of procedure.

The Breast Center staff psychologist met with Jane for six months to deal with possible residual grief over the loss of her mother, to resolve any underlying emotional issues which might be affecting her decision about surgery, and to help her make a decision about prophylactic surgery. In her case, Jane had a strong belief that her cancer risk was confined only to her breast, and that by removing her breast, she removed her risk. Rather than feeling psychologically threatened by the thought of having her breasts removed, Jane firmly believed that mastectomy would bring her longed-for relief. Although there is no published evidence that prophylactic surgery definitely decreases cancer morbidity and mortality in high risk women, Jane was convinced that the procedure would protect her. Psy-

Table I. Issues that Genetic Counselors Should Include in Prophylactic Surgery Evaluation

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- Collect family history of cancer
 - Document cancer diagnoses
 - Provide risk assessment and genetic counseling
 - Consider DNA testing, when appropriate
 - Provide options for risk reduction and surveillance
 - Discuss risk, benefits, and limitations of options
 - Emphasize that surgery does not eliminate cancer risk
 - Mention need for surveillance of other organs at risk
 - Facilitate peer support via network or support group
 - Provide psychosocial support and interventions as necessary
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chosocial support also included several sessions of couples therapy with Jane and her husband, which was felt to be helpful in dealing with relationship issues such as sexuality and self-esteem. Jane's husband remained very supportive throughout the entire process.

This case was also emotionally challenging for the genetic counselor. Discussions about prophylactic mastectomy can be very intense, because it is an irrevocable, elective, surgical procedure by which a woman's body is altered in ways that can have profound psychological and metaphorical implications. Additionally, this kind of counseling can elicit a strong degree of counselor empathetic identification. The reactions of the counselor may be unique to this case or may suggest a universal phenomenon for cancer risk counselors. There is currently no data that looks at the emotional impact of cancer risk counseling on counselors themselves, but this would be an important topic to study in the future.

Case Resolution

Jane continued to feel anxious about her chances of developing and dying from breast cancer as she had witnessed in her mother's case. Because of these fears, she stated that even the 11% estimated lifetime risk of breast cancer for the average American woman was intolerably high. At the end of a 6-month evaluation period, Jane chose to have bilateral prophylactic mastectomies.

Three years postsurgery, Jane remains pleased with her decision to use prophylactic mastectomies to reduce her breast cancer risk. She says that she now worries less and is a happier person. She denies experiencing postsurgical depression or grief. Her feelings about her sexuality remain largely unchanged; she states that she misses her breasts, but does not describe this as a significant problem. Breast reconstruction was not done, because she would not want "replacement" breasts that are less than perfect. Interestingly, Jane is now more aware of the 20 year age difference between herself and her husband. Prior to the surgery, she assumed that they would die at approximately the same age. Now Jane recognizes that she will probably outlive him and that she must make provisions for that possibility.

Jane currently volunteers for the American Cancer Society, discussing mastectomy with women having it and has sought to publicize her story for the benefit of other women concerned about their breast cancer risks. She has also made significant career and other substantial lifestyle changes.

DISCUSSION

The three cases presented in this article raise three major areas of discussion:

- Do individuals have the right not to know that their family history is consistent with a hereditary cancer syndrome?
- How can individuals make medical management decisions based on unclear or ambiguous risk assessments?
- What are the psychological issues that go into a decision about prophylactic surgery?

Do Individuals Have the Right Not to Know That Their Family History is Consistent with a Hereditary Cancer Syndrome?

Part of genetic counseling is to allow consultants to control how much information they want. There is a tension created by preserving the patient's right "not to know," while ensuring that his/her medical management is not compromised.

The decision of the team not to reveal the possible LFS diagnosis was influenced by the specific hereditary cancer syndrome that was suspected. It is probable that the presence of another cancer syndrome, such as Familial Adenomatous Polyposis which has clear guidelines for medical management, would have led the team to respond differently. In considering the team's decision, it is important to consider the somewhat unique features of Li-Fraumeni syndrome (LFS).

Li-Fraumeni syndrome (LFS) is a rare autosomal dominant cancer syndrome. The spectrum of tumors in LFS include, but are not limited to soft tissue sarcomas, osteosarcomas, breast carcinoma, brain tumors, leukemia and adrenocortical carcinoma (Li *et al.*, 1988). About 50% of families with LFS have an identifiable germline *p53* mutation. There is a 50% risk of developing cancer by age 30 (Li, 1988), and a 90% risk of cancer by age 70 (Li *et al.*, 1992). Risks of subsequent primary tumors is also increased (Strong *et al.*, 1992). Current medical management of cancer risk associated with LFS is limited. Suggested screening includes an annual visit to a primary care physician and immediate work-up of any suspicious symptoms (Li *et al.*, 1992). Close breast surveillance is suggested (Li *et al.*, 1992) and is the only screening that has been shown to reduce mortality (Shapiro, 1989). Regular blood cell counts and periodic full-body MRIs have not been shown to be of benefit for at-risk family members.

Many individuals who learn of the availability of genetic predisposition testing are interested in participating (Struewing *et al.*, 1995), but need to

be made aware of the possible risks in addition to possible benefits before making the decision to proceed (Lerman, 1993). Individuals may or may not be interested in knowing exactly their calculated lifetime risks of developing cancer. Rather, they may prefer to learn simply whether or not their risk is increased, and if so, what surveillance measures are appropriate. Allowing the consultant to take the lead in determining when, how much, and what information they receive permits them to avoiding specific risk information that might trigger excessive worry.

As increasing numbers of individuals are referred for genetic counseling regarding their family history of cancer, we must not automatically assume that all consultants are interested in learning every detail about their risk. There are benefits and drawbacks to learning potentially worrisome information, especially when risk modifying options are limited. Counseling individuals about cancer risks should include a careful assessment of why they have come for counseling, why they think information about their cancer risk will be helpful for them to know, and what they plan to do with the information. It is also important to clearly describe the available options for surveillance and risk reduction and to be honest if no such options currently exist. For some individuals, the lack of medical benefits will influence their decision about whether to proceed with genetic testing. For example, some individuals elect not to learn their status for Huntington Disease, since there is no way to alter the course of this neurodegenerative disease. Others will choose to learn their risk status despite the fact that this information may not change the course of their disease. These individuals cite reasons such as wanting to be able to plan for the future, and wanting to know if they are at risk or not, because the uncertainty of not knowing would be worse than learning they inherited the disease causing allele (Tibben *et al.*, 1993).

In some situations a genetic counselor may predict that individuals are obligate carriers of a hereditary cancer syndrome, i.e., individuals assumed to carry a mutation in a specific cancer susceptibility gene because they have a child and other relatives with similar forms of cancer. If a susceptibility gene follows autosomal dominant transmission, and a child as well as a grandparent has the susceptibility allele, then the "sandwiched" individual is an obligate carrier. The parent of the affected child must have inherited the disease causing allele if he or she has subsequently passed it along to a child. Counseling obligate carriers must take into consideration the fact that simply explaining the features of the cancer predisposition syndrome and autosomal dominant inheritance will be similar to learning the results of predisposition genetic testing. Without genetic testing, one cannot completely assume that a suspicious pedigree resembling a cancer syndrome is definitely due to the suspected cancer susceptibility gene. However, one must be exceedingly careful in counseling these putative obligate gene carriers, as the information they

may learn during the counseling session may have as dramatic an impact on the individual as disclosure of predisposition testing results.

How Can Individuals Make Medical Management Decisions Based on Unclear or Ambiguous Risk Assessments?

Conveying risk information to patients is challenging. Discussion of risk frequently includes how risk estimates are derived, the multistep process of carcinogenesis and patterns of inheritance associated with hereditary cancers. This creates a framework for discussing personal risk estimates for carrying a gene mutation and for developing specific forms of cancer. As with other rare genetic conditions, families may have received conflicting and inaccurate information from other health care providers. This may make it more difficult for the family to accept the risk estimates given in the session. In addition, there are many situations in which exact risk figures are elusive. Families which include multiple cases of early-onset breast cancer following a vertical transmission for three or more generations are straightforward in terms of risk assessment. Risks can be presented based on placement within the pedigree and the chance of inheriting a putative gene mutation, such as *BRCA1* or *BRCA2*, can be determined. Yet these families are exceptions. Most individuals interested in risk assessment have only a few features consistent with a hereditary breast cancer family and discussions must include the possibility that the cases of cancer can be sporadic, familial, or hereditary (due to a single gene).

When a genetic diagnosis is not obvious, it is customary to approach risk assessment in three ways (Hoskins *et al.*, 1995; Offit and Brown, 1994): (1) considering evidence for and against Mendelian inheritance of a genetic syndrome; (2) offering empirical risk counseling based on published data from similar family cancer constellations; and (3) considering possibilities for genetic susceptibility testing.

The provision of risk information and an individual's perception of these risk estimates is described at length elsewhere (Kelly, 1992a,b; Slovic, 1987; Stefanek, 1990) The Gail Model (Gail *et al.*, 1989) and The Claus Model (Claus *et al.*, 1994) are two empiric risk models which are often utilized with individuals at possible increased risk for breast cancer. Offit and Brown (1994) discuss the usefulness and limitations of these models. Genetic counselors should take care to explain to consultands how empiric risk figures are derived, and that the estimates apply best to the population upon which they were formulated. Risk figures should not be employed in those cases where their sample did not include enough families similar to the one seeking genetic counseling. This is especially true with paternal

inheritance, or families with more than two people affected (Knell, 1993). Caution should be taken when using empiric risk tables or computer programs as a counseling aid with individuals. Individuals need to be told that empiric risk figures only estimate risk based on a sample of families, similar in some ways to theirs, but not identical. It is important to provide the limited usefulness of such estimates in terms of personal risk estimates and to caution that, if a germline mutation is present the empiric risks are not applicable and new risks apply based on the transmission of the gene mutation in the family (Knell, 1993).

Women with breast cancer are typically given estimates for recurrence and another breast cancer primary by their oncologist. Factors such as size of the tumor and nodal involvement play heavily in the risks of recurrence (Scanlon EF, 1991). Genetic predisposition may not influence recurrence risks, but significantly alters risks of a second breast cancer. Easton reports that the risk of contralateral breast cancer in a known *BRCA1* mutation carrier may be 64% by age 70 (Ford *et al.*, 1994).

The debate on how to manage individuals with cancer who have *BRCA1* or *BRCA2* mutations continues, as does the debate about how to manage those with mutations who have not yet had cancer and those who *might* have a mutation (King *et al.*, 1993). Clinical guidelines are currently based on expert opinion, and lack data demonstrating the efficacy of cancer risk management strategies. Although the greater availability of genetic testing will allow more families to have more definitive risk information, genetic counselors should not assume that everyone will be interested in having genetic testing in order to assist them in making decisions about prophylactic surgery. A number of individuals will elect to have prophylactic surgery but forego genetic testing. Conversely, others will elect to have genetic testing and choose to continue surveillance practices rather than have prophylactic surgery. These options should be included in the cancer risk counseling discussion.

What Are the Psychological Issues that Go into a Decision About Prophylactic Surgery?

Many psychosocial issues can emerge and should be anticipated in younger women at increased risk for breast cancer (Lerman *et al.*, 1991, 1994). Anxiety and other intense feelings can influence the counseling interaction, leading some consultants to obsess about the numerical risks they face and others to tune out the discussion about risks completely. Genetic counselors need to be aware that the consultant's underlying anxieties and coping mechanisms will influence the understanding of the discussion.

Psychosocial issues have been shown to influence breast cancer screening behaviors and decisions about prophylactic surgery. Kash and colleagues (1992) demonstrated that one group of high risk women showed an inverse relationship between level of anxiety and frequency of breast self exam.

While it may seem from these cases that prophylactic mastectomy is common among women at increased genetic risk for breast cancer, instances of actually having the surgery are quite rare (Stefanek *et al.*, 1995). However, the genetic counselor should be prepared to deal with the woman considering prophylactic mastectomy, since interest in discussing the procedure may be significant in women attending high risk breast surveillance programs (Stefanek *et al.*, 1995). The decision to have prophylactic surgery is a very personal one that is based on varied cognitive and emotional factors. The genetic, medical, and psychological components are therefore best addressed by a multidisciplinary team. A genetic evaluation by a trained genetics professional should always be offered to a woman who is considering prophylactic surgery in order to provide the most accurate risk assessment possible. The medical issues should include not only detailed discussions of surgery, but equally detailed discussion of long-term consequences of surgery as well as other surveillance options that are available.

Stefanek *et al.* (1995) has studied characteristics of high risk women with an interest in prophylactic mastectomy and those who actually underwent the procedure. They found the following characteristics predictive of interest in prophylactic mastectomy: high perceived cancer risk, history of biopsy, underwent frequent breast screening, breast cancer related worry, and cognitive intrusions about breast cancer. Women who indicated a significant level of cognitive intrusion about breast cancer more often opted for surgery than those with fewer intrusive thoughts. While a recent stressful life event prompted women to attend a high risk breast service, it was not found to predict interest in prophylactic surgery. This small survey reported that satisfaction with the decision to undergo surgery was related to the supportiveness of family members and friends as well as the extensive counseling that accompanied the procedure.

Some consultants may be referred for CRC because of an initial interest in prophylactic mastectomy, whereas others may not be interested in prophylactic mastectomy no matter how high the breast cancer risks. While genetic counselors often raise the option of prophylactic surgery, they tend to avoid advising or recommending prophylactic mastectomy for the following reasons:

- It is rare to encounter a person with a cancer risk substantial enough to consider such surgery.

- Risk estimates are fallible estimates and are rarely predictive in particular individuals of specific cancers at predictable ages.
- The decision is often based not only on genetic but also on uniquely personal medical and emotional grounds.
- The efficacy of the procedure has not been established in genetically high risk populations and at least some residual risk of breast cancer remains.
- This is an intensely personal decision about an elective procedure that should be made in consultation with health care providers, but by the consultand herself.

SUMMARY

The cases presented in this article illustrate some of the complexities of cancer risk counseling, even without the added layer of genetic testing. These cases are not meant to exemplify the only way or even the best way to deal with these types of issues. Instead, the hope is that by sharing our cancer risk counseling experiences, we can gain the insight together of how best to provide this specialized area of counseling.

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