Breast cancer remains a leading health concern for women. Genetic medical research is beginning to alter breast cancer screening recommendations and breast cancer treatment. This article discusses the use of the Family Systems Genetic Illness (FSGI) Model to provide greater understanding of some of the more common personal and family issues related to breast cancer. After a brief presentation of a case study of a woman who chose to undergo bilateral mastectomy to avoid breast cancer and a critique of the FSGI Model, application of the FSGI Model to breast cancer will be considered. The FSGI Model categorizes genetic illness according to the degree of the genetic risk the illness has, potential severity, age of clinical onset, and whether treatment can alter the onset or course. The article seeks to help nurses assist women in balancing their personal and family responsibilities and choices in light of new knowledge and medical care of breast cancer today.

Mrs. R, an employed 54-year-old Italian American woman, was seen by a homecare nurse the day after her discharge from the hospital, where she had bilateral mastectomy for the prevention of breast cancer. Her husband was home but did not participate in the home visit. About six weeks before the surgery, she had learned that she had a benign breast mass, but she opted for prophylactic bilateral mastectomies because of a strong family history of breast cancer. Five days after surgery, she had returned home, the surgical sites were healing well, and she had a date for reconstructive surgery. She expressed dismay over the development of a breast mass despite exercising regularly, abstaining from cigarettes and alcohol, and keeping her weight down. Mrs. R also believed that where she lived, Long Island, NY, was associated with a much higher rate of breast cancer than other geographical areas, something she had read about regularly in the local press. She said she had not under- gone genetic testing for breast cancer. Whether she had ever been recommended to do so and had declined was unclear.

Accurate, up-to-date information and frank family discussions at various times in the course of illnesses are important to decision making and family relationships. Mrs. R did not have a referral to a cancer genetics specialist, what is now the standard of care for women with a family history that strongly suggests inherited susceptibility to breast cancer (Korde, Calzone, & Zujewski, 2004). In terms of medical care, early detection and timely intervention remain critical. The FSGI Model (Rolland & Williams, 2005) is a longitudinal model that helps to better manage and predict illnesses such as breast cancer.

Decisions about screening and breast cancer management now are guided not only by family and personal history and clinical status but also by genetic testing information. Genetic testing promises to provide more accurate and personalized risk assessment. It may help to reduce the need for biopsies

At a Glance
✦ Genetic medical research is increasingly altering healthcare decisions in cancer care.
✦ The Family Systems Genetic Illness Model provides a useful tool for nurses working with women at high risk for breast cancer and their families.
✦ All women with a family history of breast cancer should see a genetics counselor.

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because genetic testing will more accurately determine which masses are benign without the need for biopsy, and it will help guide treatment decisions (Fuqua et al., 2004; Struwing, 2004). Genetic testing is not without expense or risk, including the potential for being denied health insurance coverage of illnesses identified as “preexisting.” It also challenges young adults with the task of having to integrate information about their genetic health risks while still in the process of making important life decisions. Ideally, the increase in anxiety that is likely to follow learning of a positive genetic risk for breast cancer would help individuals and their families remain alert for early evidence of breast cancer by having regular mammography and clinical breast examinations.

**Family Issues**

Mrs. R’s experience of the unfairness of cancer is understandable, even though healthy lifestyle choices can never provide a guarantee of health or longevity. What also may be an issue for her is her husband’s somewhat limited involvement as a caregiver. At a time when emotional support, acceptance, and practical assistance from him would be particularly welcome, they seemed to be somewhat limited. As the American population ages, the number of people with cancer is increasing, and healthcare expenditures may be limited by government and insurance companies. This will force families themselves to provide more and more care, including nursing services, to their ill family members. Families will need to be more flexible and creative, and nurses will need to help.

In their study of later-life couples coping with breast cancer, Hodgson, Shields, and Rousseau (2003) found that disengaging communication in marriages with women with breast cancer was associated with lower marital satisfaction. Patterns of family communication, characterized by withdrawal and avoiding open communications about cancer and its associated feelings, reduced the quality of the marriage and family life. Whether the absence of Mr. R from the discussion with Mrs. R about her postoperative needs and restoration of her normal life and health is a part of a pattern of disengaging is unclear, but it is a missed opportunity for him to become better informed about his wife’s condition and possibly deepen their relationship. Women who do not have family members to assist them when they are patients and help make decisions are at a disadvantage. Homecare nursing provides a unique opportunity to assess families and help them minimize the limitations imposed by illness and live full lives, but their involvement in the trajectory of illness usually is fairly late. Nurses and advanced practice nurses in primary care sites are more likely to see their patients throughout the course of illnesses such as breast cancer and use the FSGI Model more effectively.

**The Family Systems Genetic Illness Model**

The FSGI Model (Rolland & Williams, 2005) provides a framework to inform healthcare providers on the family issues patients experience when living with their genetic personal health risks. Genetic testing has the advantage of providing more personal health information in the predisease stage of illness when prevention can be most effective. The implications of having such self-knowledge as a young adult, before important life decisions related to marriage, family, and work are made, are likely to be significant. Such knowledge is important for individuals with high genetic risk but also for later generations of female family members. Nurses and primary care providers who work with women like Mrs. R earlier in patients’ lives recommend genetic counseling and realistically discuss the advantages and limitations of diet, exercise, and other cancer-prevention efforts. An overwhelming amount of health and medical information is directed at women and their families, and nurses need to help their patients be appropriately skeptical and balance their responsibilities and choices.

The value of risk-reduction efforts can be seen in the diagnosis of heart disease, particularly myocardial infarction, which has been dramatically reduced as a cause of death and disability for middle-age adults since the 1980s in the United States and Europe. Unfortunately, exercise and weight loss, although helpful (McTiernan, 2003), have not been as effective in the prevention of breast cancer as they have for heart disease, and the estrogen-lowering medication tamoxifen lacks the safety profile and ease of use that cholesterol-lowering statin medications have (Korde et al., 2004). Women with a high risk of breast cancer do have other risk-reduction options, including prophylactic surgery. Total mastectomy is considered the most effective (de Carvalho, Jenkins, Nehrebecky, & Lahl, 2003), but it is very invasive and carries considerable physiologic and psychological ramifications.

The FSGI Model is an update of Rolland’s (1984) Family Systems Illness (FSI) Model (see Figure 1), which was developed to help understand the psychosocial aspects of medical illnesses. The FSI Model did not well appreciate the potential of secondary cancers that can follow breast cancer or the importance of comorbidities that are particularly common later in life. Rolland (1994, 1998) broadened the FSI Model to better include the multigenerational family’s patterns, culture, gender, and beliefs system issues.

The updated FSGI Model (see Figure 2) includes genomic medicine and focuses more attention on the preclinical phase of illnesses. The preclinical or presymptomatic phase of illness, according to Rolland and Williams (2005), is the period of time after patients have learned of their genetic risks but before any symptoms become evident. Before discussing the merits of theories as a guide to clinical practice, considerations must be made. The first consideration when applying a model such as

<table>
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<tr>
<td>Acute</td>
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</tr>
<tr>
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<td>Mild</td>
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<td>Progressive</td>
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**Figure 1. The Elements of the Family Systems Illness Model**
the FSGI to families with cancer is to keep in mind the diversity of ways individuals and families deal with information about their health risks. Some individuals and families seem to decide not to attend to or discuss their health risks at all (“disengagement” strategies), whereas others become preoccupied about their risks and talk openly with various people. Coping theory suggests that learning about an illness and its treatments is usually more effective than seeking distractions and avoiding thinking and talking about stressful life events, at least in situations where something can be done about the events (Lazarus & Lazarus, 1994). Nurses need to appreciate and respect each person’s and family’s perspective, values, and style of communicating and making decisions, even when the perspectives may seem unreasonable.

The second consideration when applying the FSGI Model in clinical practice is to encourage families not to see genetics as unalterable fate, but rather as important information for the person-family-environment process. Genetic testing can help people make more responsible, or at least better informed, decisions. Nurses also have to be aware that different people define family in different ways (Pinsof, 2002; Walsh, 2003). Biologically oriented health professionals think primarily of the biologic definitions of family, and more psychosocial oriented disciplines such as social work think of families as potentially instrumental and emotional sources of support for patients. Poston et al. (2003), for example, defined a family as two or more people who think of themselves as family and who, ideally, provide for each other’s care and support on a regular basis.

Family quality of life is defined by Poston et al. as the ability to maintain family conditions in which each family member can meet his or her needs and enjoy time together without putting undue restrictions on each other’s freedom. Maintaining family quality of life—while living with the knowledge that a member is at high risk for a potentially lethal illness and that if the family has children, they are likely to pass that genetic risk on—is not easy, but it also can make families closer and stronger. Facing reminders of the vulnerability and limitations of human life is humbling and helps people have more sensible expectations of close ties. Those reminders can help families review their priorities and become more humane to each other and others. Family relationships are seen in the FSGI Model as potential resources and become more humane to each other and others. Family close ties. Those reminders can help families review their priorities and become more humane to each other and others. Family close ties. Those reminders can help families review their priorities and become more humane to each other and others.

The paradox of human life is such that clearer perception of its limitations can be very enabling (Parse, 1998).

### The Psychosocial Impact of Genomic Risk Information

Genetic testing for breast, colon, prostate, and ovarian cancers and Alzheimer disease can help people learn about all of their health risks (Rolland & Williams, 2005) and also can have profound implications for their identities, relationships, commitments, occupations, lifestyles, retirement decisions, and decisions to have children. Nurses often have the opportunity to help people and families put potentially disturbing and overwhelming information into perspective. People and families also will have the opportunity to plan for a range of clinical services, prenatal diagnosis, newborn screening, and carrier testing, as well as the diagnosis and estimation of prognosis of their genetic disorders (Secretary’s Advisory Committee on Genetic Testing, 2000). Nurses’ roles as patient and family educators and advocates will be significant. Because most breast cancers are not linked to a single factor but rather are polygenic in nature, environment plays a much bigger role in illness manifestation (Struewing, 2004). Women have options to favorably alter the illnesses’ expression and course by changing their lifestyles and/or environmental factors as well as taking prophylactic actions. Genetic testing has the potential to help individuals and families attend to the complex genetic risk-environment interaction process. Receiving such information early in life is particularly valuable in terms of modifying illness expression, but the timing also may cause patients to have difficulty accepting and integrating the findings into their previous plans and dreams. Although some faiths and religions may discourage families from having genetic testing, individuals and families ultimately will have to discern what their values and beliefs are and decide for themselves. According to the philosopher Hans-George Gadamer (1996), when important human decisions must be made regarding family and society, “everyone must decide, according their own best knowledge and conscience” (p. 23). Nurses who keep current on the medical and genetic research and explain the information to lay people will be highly valuable to families and members of the healthcare team. Although nurses provide needed information to their patients and correct incomplete or inaccurate information that patients already have, their most important job is to respect patients’ values and decisions.

The FSGI Model considers that illness risk is based, at least in part, on genetic mutations. It also considers how severe an illness is likely to become and when it is likely to be expressed in a person’s and family’s life cycle. It includes consideration of how likely interventions will be able to alter illness onset or progression. The model seeks to highlight the elements of genomic illnesses that are most likely to impact the person and the

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<tr>
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</tr>
<tr>
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<td>Low</td>
<td>Later life (&gt; 60)</td>
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<tr>
<th>Clinical Severity</th>
<th>Treatment Can Alter Onset or Course</th>
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<tbody>
<tr>
<td>Low</td>
<td>Yes</td>
</tr>
<tr>
<td>High</td>
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**Figure 2. The Elements of the Family Systems Genetic Illness Model**

**Genetic Illness Model**
family. Rolland and Williams (2005) separated the preclinical phase of a genomic illness into three subphases: the awareness phase, pretesting phase, and testing/post-testing phase. They described the awareness phase as the period when a person and family first become aware of the genetic risks that they have. In this phase, the illness-related developmental tasks include communicating with the family about the genetic risk and seeking information about what being genetically at risk for breast cancer means.

Families need information to better understand the basics of genetic illnesses and consider the risks and challenges of living with the knowledge that they carry a high genetic risk of breast cancer.

Optimally in the pretesting phase, the family discusses how genetic testing will influence the person and family. Practical, emotional, and short- and long-term issues ideally will be considered. Families need information to better understand the basics of genetic illnesses and consider the risks and challenges of living with the knowledge that they carry a high genetic risk of breast cancer. Families need to be aware that other family members also may be at risk for the illness and need to consider how and when to inform them. Rolland and Williams (2005) suggested that the beliefs and meaning of having genetic tests should be explored during the pretesting phase of the illness. They anticipated that many individuals and families may carry misconceptions and feel that genetic illnesses carry a stigma or are an issue of fate or blame. Lantz and Gregoire (2000) observed that breast cancer can be accompanied by a vicious circle that disrupts some couples’ “will to meaning” (p. 318). From their research on the use of existential couples therapy with families with various illnesses, they found that meaning is something that helps hold couples and families together and helps them deal with everyday challenges. The starting point for existential therapy is people’s or couples’ experiences, and the therapy helps them explore meaning and ways to be better able to deal with their life challenges and enhance the everydayness of their lives and relationships. Anything that disrupts the process of exploring meaning can leave individuals more anxious and depressed and leave couples in conflict. The diagnosis of cancer or the discovery of a high genetic risk for it can lead to an existential-meaning vacuum for individuals and couples. Lantz and Gregoire suggested that such couples would benefit from existential couples therapy to help them fill the vacuum. Although nurses are not expected to have the time or training for the therapy, their interactions with families can appreciate that the diagnosis can have such an effect. Lantz and Gregoire’s approach will help couples ‘notice, actualize, and honor the meaning and meaning potentials in their lives in spite of breast cancer’ (p. 319). Although few people enter relationships and families prepared for such a journey to uncover life-affirming meaning, nurses can be with people at times when questions are most likely to arise.

Rolland and Williams (2005) considered the pretesting, testing, and post-testing phases as potential crisis phases. During and after testing, families need to find ways to preserve their identity and hope in the face of new challenges and to continue to uncover meaning and focus while dealing with the potential losses that they face. Rolland and Williams described this as being flexible when life was uncertain. For most people, genetic risk for breast cancer must be considered along with other known risk factors and environmental influences.

In the case of Mrs. R, she was aware of and understood the importance of having a family history of breast cancer and a geographic location associated with the disease. She said that the discovery of a suspicious breast mass motivated her to go ahead with the prophylactic bilateral mastectomy. According to the research evidence that is currently available, breast cancer risk increases with each first-degree relative affected, but the research also points out that most women with breast cancer (88%) have no family members with the illness (Dumitrescu & Cotarla, 2005). Mrs. R did not discuss her current age, age of menarche or menopause, pregnancy and childbirth history, use of oral contraceptives or hormone replacement, or social economic status, even though they are well-established risk factors, nor did she admit to having any genetic testing (Dumitrescu & Cotarla).

Breast Cancer Genetics

Breast cancer genetics research currently highlights the importance of germline mutations. Germline mutations, which represent inheritable changes in DNA that have a high chance of leading to breast cancer, have what is described in genetics as high penetrance. Assuming that Mrs. R’s genetic makeup did put her in a high-risk category for breast cancer, then she could be seen as having high-penetrance genes for the disease. The genes could include BRCA1, BRCA2, p53, ATM, or PTEN, but all of the germline mutations account for only 5%–10% of all breast cancer cases (Dumitrescu & Cotarla, 2005). Women who have the genes are more likely to have breast cancer at an early age (Dumitrescu & Cotarla). Most versions of the mutations involve low-penetrance genes that must interact with other such genes and environmental factors, including lifestyle and medical decisions, diet, hormone use, and pollution, to result in breast cancer. The genetic tests that are better able to determine the expression of gene clusters or patterns are the DNA expression microassays, which can locate tens of thousands of DNA “probes” (Miller & Liu, 2004). The tests help to identify different expression genes, uncover gene clusters, and classify clinically significant samples, all of which have the potential to provide better prognostic certainty and guide treatment decisions. For example, they can help identify which individuals are responsive to the estrogens that contribute to cancer growth. In the absence of such tests, most patients with breast cancer, even those with negative nodes, undergo systemic adjuvant chemotherapy to reduce their risk of recurrence, although as many as 40% will not respond to such treatment (Fuqua et al., 2004).

In the FSGI Model, Rolland and Williams (2005) further categorize genetic risk as having one of three possibilities: (a) a high likelihood based on a high level of penetrance of a
mutation, (b) a variable likelihood reflecting complex multiple genes and gene-environment interactions, and (c) a lower likelihood based on complex, multiple gene and environmental interactions. Variability of likelihood of having the illness and the timing of that illness expression in a person's life cycle provide valuable information to help family members plan for such realities and act in a timely manner. The FSGI Model also factors the severity of health problems along the disease course. Discovering that a patient has the high-penetrance breast cancer susceptible genes (including BRCA1, BRCA2, p53, ATM, and PTEN) provides the clearest genetic information regarding increased risk of developing breast cancer, potentially altering a patient's planning and view of the future. In the FSGI Model, Rolland and Williams grouped onset, course, outcome, and disability typology of the FSI Model into the clinical severity variable, describing it as either high or low severity of clinical symptoms. The item outlines the disease burden of the illness to the patient and family. Patients and families who discover that they have low genetic risk may be distressed if they had been overly restricting their lives, assuming they were at greater risk (Sobel & Cowen, 2003). Having more specific information provides a greater sense of control to individuals and families and helps providers to identify which families have excessive fears and anxiety and may require mental health intervention.

Summary and Conclusion

Mrs. R and others like her are having prophylactic surgery to prevent breast cancer based primarily on their family history, personal history, and environmental risks. Although the clinical application of genetics medical research is in its infancy, its relevance to breast cancer already is making significant advances toward more accurate prognosis and treatment options for women and their families. Illness and death from breast cancer continue to limit human existence, whereas new information and medical treatments increase the complexity of healthcare decisions and family life. Rolland and Williams’ (2005) FSGI Model helps nurses and advanced practice nurses understand how illnesses such as breast cancer likely will affect patients’ lives and families, well before any biologic markers are detectable. Considering the complexity of medical information that currently is available and the risks involved in genetic testing for breast cancer, the best recommendation at this time for people with a family history of breast cancer is to see a genetics counselor. Nurses can help individuals and families prepare for counselors by giving them a list of helpful questions and recommending that they bring a trusted family member or confidant to the meeting. For women who decide to have prophylactic breast surgery, nurses play a significant role in helping them prepare for the emotional and physiologic ramifications. The FSGI Model anticipates the time when families will have an overall genetic risk profile, which can guide them in their prevention efforts and truly give them informed consent.

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