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Models for Cancer Genetic Risk Assessment Programs

by Cara Egan

In the near future cancer risk assessment that includes counseling, support, and education may become standard criteria for establishing a comprehensive cancer program. Already a new cancer susceptibility gene is identified almost monthly. Publicity about these findings is prompting patients to ask questions about their risk of cancer. They will come to expect their health care providers to provide answers. Initially, cancer centers may be reluctant to invest in a hereditary screening program that is time consuming and requires highly trained staff. Yet the overall value of the program to patients, their families, and the community may eventually convince hospital leadership to incorporate hereditary cancer screening as an essential cancer program component.

Many community hospitals have already developed, staffed, and launched hereditary screening programs. This article provides a close-up look at the operations of three model programs:

- *The North Shore Medical Center* is a 346-bed acute care hospital in Salem, Mass. About 1,300 new analytic cancer patients pass through its cancer center each year. The Bernadette Giuffrida Hereditary Cancer Screening Service opened in 1995.
- *Roswell Park Cancer Institute* in Buffalo, N.Y., is a 420-bed freestanding, NCI-designated comprehensive cancer center that provides services to about 1,800 new analytic cancer patients each year. The Institute offers the Breast Cancer Risk Evaluation Program and risk assessment for other hereditary cancers through the Clinical Genetics Service and the respective specialty clinics.
- *The Cancer Institute of Health Midwest* represents and coordinates the cancer programs and services of eleven hospitals within Health Midwest, an integrated health care delivery system of fourteen community hospitals in the Kansas City, Mo., area. These hospitals have access to the Cancer Prevention Clinic, which specifically targets high-risk families. Health Midwest sees about 2,800 new analytic cancer patients each year.

Staff from these hereditary cancer screening programs agree that ensuring patient confidentiality is a major program requirement. Many individuals fear that disclosure of genetic information could eventually lead to discrimination by insurance companies, employers, and other entities with access to their medical records. To maintain confidentiality, these programs provide participants the option of sharing information with their primary care physicians. Fear of disclosure prevents many individuals from deciding to have genetic testing or participating in the available clinical research. As a result, persons at risk do not necessarily receive information or options relevant to risk of developing cancer, nor do the programs have access to the wealth of data important to further understanding.

NORTH SHORE MEDICAL CENTER

The Bernadette Giuffrida Hereditary Cancer Screening Service at North Shore Medical Center was funded in part from a donation by the husband of a former cancer patient. As the endowment grows, it will allow hereditary cancer risk assessments to be provided free of charge to individuals and families with a history of cancer. The program was instituted to provide in a comprehensive community setting answers to the general public's questions about cancer genetic risk. During its first year of operation, forty-seven people participated in the program.

Staffing. A nurse practitioner coordinates the program, which includes conducting the initial genetic risk interviews, discussing results of the completed risk assessments, referring high-risk individuals to genetic counseling and testing, and coordinating community education symposia on hereditary cancers. Ricki Preston, R.N.C., N.P., program coordinator, has twenty years of oncology nursing experience during which she has developed educational seminars in nursing, including a three-day course for nurses in genetics and cancer care at Fox Chase Cancer Center in Philadelphia, Pa.

Operation. Participants in the screening program are usually referred by oncologists, nurses, primary care physicians, or gynecologists practicing at North Shore's cancer center and throughout the community. Twenty-five to 30 percent of participants are self-referred. First contact with a participant is usually by telephone. At this time, the program coordinator explains the purpose of the program and listens to the participant's concerns and expectations. Individuals with a family history that warrants further investigation are mailed a form and requested to list birthdates, dates of death, any cancer diagnoses, and dates of cancer diagnosis of all relatives, spanning three generations.

Within two to three weeks the participant brings the completed form to an appointment scheduled with the program coordinator, who conducts an education session about genetics and hereditary illnesses. The program coordinator then performs an initial analysis, using computerized software to help identify people at high risk of developing cancer—those with

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early onset of a cancer diagnosis, multiple family members with cancer, multiple cancers in one person, or bilateral cancers. Depending on the outcome of the initial analysis, the program coordinator may also briefly discuss the possibility of genetic testing.

The Giuffreda Hereditary Cancer Screening Service operates in conjunction with OncorMed, Inc., a Maryland-based company that offers genetic services. OncorMed provides the computerized risk assessment software first developed by Henry T. Lynch, M.D., of the Hereditary Cancer Institute at Creighton University in Omaha, Nebr. Participants in North Shore's screening program have access to OncorMed's genetic pedigree analysis, genetic testing services, and on-line consultation with Dr. Lynch on a case-by-case basis. OncorMed's IRB-approved protocols include specific criteria to assure that only high-risk individuals are tested. In addition, no tests are performed on children or individuals who are unable to provide informed consent.

The program coordinator sends the pedigree to Dr. Lynch. Within four to six weeks Dr. Lynch or a member of his staff completes the evaluation and makes recommendations for follow-up. The participant returns for another consultation with the program coordinator.

At this point, there are three possible scenarios:

1. The cancer occurs sporadically, i.e., there is no pattern to the cancer occurrences.

2. The cancer is familial, i.e., there is an increased history of cancer in a family that does not travel from one generation to the next and thus is not necessarily hereditary. However, these patients are still at high risk for developing cancer.

3. The cancer appears to be hereditary. In this instance, the program coordinator helps the individual obtain more complete medical documentation and initiates indepth counseling about the benefits, risks, and limitations of genetic testing, as well as individual education on cancer prevention and lifestyle changes.

Counseling is an essential program component, since many individuals come to the program equipped with a variety of agendas, myths, and fears. The coordinator must help the individual explore all possible scenarios:

- What would the participant do if the test were positive? Negative?
- Would prophylactic treatment be considered?
- How would family members be informed?
- How will this knowledge affect daily life?

Genetic counselors are available through North Shore Medical Center's affiliations with the Dana-Farber Cancer Institute and Massachusetts General Hospital to help individuals deal with the complex ramifications of genetic screening. Individuals believed to be at risk for genetic inheritance of cancer are often referred to these programs where they have access to clinical research.

Individuals with a family history of breast cancer tend to access the hereditary screening program most frequently. During the program's first year of operation, fifty-eight calls were received. Forty-seven individuals were seen by the program coordinator, of which thirteen were determined to be at familial risk and sixteen were determined to be at potential hereditary risk for developing cancer. Participants who were at potential risk of carrying a genetic mutation and who wished to pursue genetic testing were referred to the Dana-Farber Cancer Institute. Preston cites cost of the test and the desire to be involved in clinical research as the primary reasons for participant referral.

"We are not to the point where we can tell someone with a positive test, 'Do X, Y, and Z and you will not develop cancer,' because even prophylactic treatment can offer limited guarantees," said Preston, program coordinator. "At best we can individualize a person's screening process to help identify an early diagnosis."

ROSWELL PARK CANCER INSTITUTE

The Breast Cancer Risk Evaluation Program at Roswell Park Cancer Institute in Buffalo, N.Y., provides education and counseling services to women at increased risk of breast and/or ovarian cancer and those women who perceive themselves to be at high risk. Roswell Park charges a fee for physician consultation, risk assessment, and counseling services, and, if eligible and requested, provides gene testing through laboratory companies that test for mutations in cancer-associated genes. Presently, local HMOs are willing to cover patient expenses for education and risk assessment. However, the extensive time required to accurately assess an individual's risk may cause payers to restrict reimbursement and affect the financial viability of the program.

Staffing. The program is co-directed by Stephen B. Edge, M.D., a medical oncologist, and Carolyn Farrell, M.S., C.N.P., C.G.C., a board-certified genetic counselor. Farrell, who is also a nurse practitioner, meets one-on-one with individuals to provide genetic assessment, education, and counseling

Genetic Testing and Medical Care Management

The decision to test for mutations in cancer-associated genes such as BRCA1 and BRCA2 is an intensely private issue for patients and their families but one that cannot be made alone, according to Tom Frank, M.D., medical director for Myriad Genetic Laboratories. Patients and their families must thoroughly explore the benefits and limitations of testing in consultation with their physician and genetic counseling services. The stakes are high, and sometimes the answers are not black and white. Patients must ask themselves how a positive or negative test result would affect their lives; physicians must consider how a positive or negative test result would affect the medical care management of their patients. While testing is not for everyone, Frank stated, medical oncologists and other cancer-related specialists have an obligation to provide all patients at high risk of developing cancer, especially those considering prophylactic treatment, with as much information on genetic testing as possible.

sessions, which last between one to two hours. Farrell and her colleague, Mary-Jo Rosenblatt, also a genetics counselor, spend a considerable number of hours speaking with prospective participants over the telephone and retrieving and reviewing participants' medical charts. Additional medical, nursing, and allied health professionals assist with this multifaceted program.

Operation. At the initial interview the genetic counselor, the nurse practitioner, and/or physician/oncologist compile a detailed medical and family history, as well as a woman's cancer screening and mammography history. The physician then performs a physical examination, including a breast examination, and offers recommendations for future cancer screening based on the woman's particular risk and other factors. Women who have not had a recent mammogram can receive one at this time.

Often that same day the woman meets individually with a genetic counselor to discuss her family history, genetic risks, and the benefits and limitations of gene testing. While there are no absolute criteria for defining people at high risk, for the most part individuals considered to be at high risk are those with a positive family history of cancer, with several relatives having either breast or ovarian cancer, and at least one incidence of early onset of disease. The oncologist/physician and/or genetic counselor will address recommendations to modify screening guidelines based on the individual's risk and family history. For example, a woman with a family history of breast cancer at early onset may be recommended to receive mammography screening at an earlier age than the general population.

The primary focus of the Roswell Park program is on risk assessment and counseling. Genetic testing is secondary. Only about 15 to 20 percent of program participants are eligible and choose genetic testing. Roswell Park provides gene testing through a number of labs, including Myriad Genetic Laboratories. If genetic testing is appropriate, participants return for additional consultations to learn about the potential benefits, risks, and limitations of testing, and to discuss their test results.

As laboratory companies that test for cancer-associated genes become increasingly available, more cancer testing programs will develop. Edge cautions hospitals to remember what their primary purpose should be: cancer risk assessment and counseling.

In addition to physician and counseling components, program participants must have access to local, regional, and national clinical research, a necessary component of any hereditary cancer risk assessment program.

Having top quality cancer genetics services enhances a hospital's reputation in the community, according to Farrell. "Hopefully third-party payers, physicians, and patients will find that the indirect benefits of these programs will outweigh costs that may at first glance seem prohibitive or unnecessary."

THE CANCER INSTITUTE OF HEALTH MIDWEST

The Cancer Prevention Clinic at Trinity Lutheran Hospital in Kansas City, Mo., provides education, risk assessment, and counseling free of charge to eligible participants. Funding is provided through a community philanthropic grant. The program's primary focus is to help individuals reduce their cancer risk, diagnose cancer at the earliest possible stage, and increase their understanding and decrease anxiety related to developing cancer. Criteria based on American Society of Human Genetics recommendations are used to determine participant eligibility. Participants may include anyone who has a higher risk of cancer based on family history, personal history of previous malignancy, or personal or family history of atypical malignancy (atypical age of diagnosis or site).

Staffing. The program is staffed by Sukumar Ethirajan, M.D., a medical oncologist and medical director of the program, and Amy Strauss Tranin, R.N., M.S., O.C.N., a full-time genetic cancer risk counselor with a background in oncology nursing. Tranin has supplemented her experience in oncology with course work in basic science and genetics and plans to earn a degree in genetic counseling. Tranin is a member of the National Society of Genetics Counseling.

Operations. Eligible participants who are referred to the program are mailed an extensive questionnaire that includes questions about family history, lifestyle, and environmental factors. Referrals originate from primary care physicians, gynecologists, oncologists, or are self-referred.

Over the course of possibly several visits, the genetic cancer risk counselor spends time working with the participant to develop the most accurate risk information possible. From information in the questionnaire, she develops a three-generation pedigree and then reviews the participant's risk factors and determines any inheritance patterns in the pedigree. Family life experiences, particularly related to cancer, are also explored.

The Real-Life

I had just turned 40 when I was diagnosed with breast cancer, the disease that had killed my mother at age 47. My doctors thought it likely that my grandmother, a nonsmoker who died of lung cancer at 56, was misdiagnosed and had died of breast cancer too. So in September 1994, as I was approaching the three-year anniversary of my diagnosis, one of my physicians phoned to tell me that Memorial-Sloan Kettering Cancer Center in New York City, where I was treated, was going to be involved in a research study of BRCA1 and needed volunteers to give blood and have their genes studied. With a premenopausal diagnosis and a family background like mine, my doctor thought I would be a perfect candidate.

I have always thought it essential to be as informed a participant as possible in the health care decisions that have so

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Implications of Genetic Testing for Breast Cancer

by Gayle Feldman

affected my life, and therefore set out to understand the basics of breast cancer genetics and testing and its implications for me and my family. At first, though, I didn't realize how complicated it would turn out to be.

I thought at the time I was tested—and so did the doctor who talked with me—that the results would come within six months, and I would be given the choice of knowing or not knowing if I were a mutation carrier. Well, two years went by before I was to hear anything further. And in the interim, the whole issue of breast cancer genetics exploded onto the news.

In autumn 1996, I received a phone call inviting me to come to the hospital to talk about the pros and cons of knowing the results of my test. I accepted, and met with a genetic counselor and a physician to discuss the following scenarios: If the results were positive, what would that mean medically? Psychologically? Practically? How would it affect my relationship with my family? What would be the pros and cons of testing for them? And if the results were negative, what would that mean?

Every woman who goes through genetic testing will need to think long and hard about questions like those. Unfortunately, many women who opt for commercial testing won't have the professional counseling they need. Most general practitioners aren't educated to deal with this, nor are most surgeons or oncologists. Testing negative for BRCA1 does not put you in the clear for BRCA2 or for the as-yet unidentified third BRCA mutation that researchers have postulated. If you test positive, the bottom line is, of how much use is the information that you are a mutation carrier and at high risk?

Prophylactic mastectomy is one route that is sometimes advised. The idea of removing a healthy breast because it might develop cancer down the line is a horrific idea to most women, as I know from personal experience. Because of a combination of factors involving the type of cancer cells I had and my family history, when I was facing the initial mastectomy back in 1991, my doctors recommended that I follow up that surgery with a prophylactic mastectomy of the other side.

I said "no." A year later, in 1992, when I was told that I had a 50 to 60 percent chance of developing a new primary cancer in the other breast, I did choose to have a prophylactic mastectomy. Even with such statistics staring me in the face, it was not an easy decision. But for me, it was the right decision—one that enabled me to concentrate on getting on with my life.

Yet faced with the possibility of testing positive for BRCA1, I asked myself, what then? I already had an ultrasound once a year to check my ovaries. Was that not enough? Should they too be removed? If I tested positive, my doctors would recommend that my sisters be tested. But would they want to be?

And what about the implications for the next generation, for my son and my nieces? The guilt of knowing that you have passed on the mutation to your children can be very difficult to handle. While some people take charge and want to know, others are in denial and want to run away.

As for this particular breast cancer survivor, I tested negative for BRCA1. I look back on two generations of my family who did not survive and know that I am here today because of the advances made in the thirty years since my mother died. I gave my blood, did my small bit to help further the study of breast cancer genetics so that the next generation of my family will not have to talk in terms of survival, so they will know a cure.

Purported instances of cancer in a family are verified with medical records whenever possible. If the participant has not had a recent physical, the medical oncologist performs one.

Retrieving medical records is a laborious but essential part of the process. Participants are encouraged to bring as much information as they can during their first visit. Information traced back from even two generations may often be inaccurate. Family members are asked to release their personal medical records. In the case of the deceased, the closest living relative must release the records.

Once the necessary medical records are collected and examined, the pedigree is developed. The medical oncologist and genetic cancer risk counselor collaborate to identify those individuals at high risk and develop a management plan for each participant. The counselor and oncologist then meet again with the participant to review the findings and make recommendations.

Ten percent of the program's participants go on to have cancer genetic testing. While the program uses several labs that test for cancer-associated genes, OncorMed, Inc., is the primary lab provider. Through OncorMed, the Cancer Prevention Clinic has the option of receiving an additional consultation with Dr. Henry T. Lynch.

Major emphasis is placed on providing accurate genetic risk assessment and counseling; genetic testing is a secondary goal. "We are committed to establishing clinical relevance before pursuing genetic testing," said Ethirajan. For hospitals in the process of developing hereditary cancer screening programs, Ethirajan recommends incorporating the three major components of physician consultation, genetic counseling/risk assessment, and genetic testing. Ethirajan believes that staff with combined oncology and genetic experience can best understand the complexities of inherited cancer.