How to Develop and Implement a **Cancer Genetics Risk Assessment Program**

Clinical and economic considerations

by Christopher Ho, MS

Between 4 to 11 percent of the most common cancers (e.g., breast, ovarian, pancreas, colon) are inherited,[1-4] so the discovery of hereditary cancer genes, such as BRCA1, BRCA2, hMLH1, hMSH2, has caused a paradigm shift in how the healthcare industry thinks about and treats cancer. Instead of waiting to treat a patient after the onset of a disease, we now have the ability to identify and manage individuals at high risk *prior* to the onset of symptoms.

As recently as the early 1990s the field of cancer genetics was limited primarily to university-based institutions where large linkage studies were performed in the hope of discovering a genetic cause to any of the variety of documented hereditary cancer syndromes. Today the practice of cancer genetics risk assessment is no longer limited to large university-based hospitals, but can be provided in community cancer centers as well. In fact, community-based cancer programs and even some larger physician practices face increased demand from well-informed and Internet-savvy patients to offer cancer genetics services, specifically, risk assessment education and testing for inherited susceptibility cancer genes.

CLINICAL BENEFITS OF CANCER RISK ASSESSMENT AND GENETIC TESTING

Organizations like the Association of Community Cancer Centers (ACCC), the American Society of Clinical Oncology (ASCO), the American Gastroenterological Association (AGA), and many others have established guidelines and recommendations advocating genetic counseling and testing at the community level.[5-7]

Two of the most well-documented inherited cancer syndromes are hereditary breast and ovarian cancer (HBOC) associated with mutations in BRCA1 and BRCA2 and hereditary non-polyposis colorectal cancer (HNPCC) associated with mutations in hMLH1 and hMSH2.[1,8]

The National Comprehensive Cancer Network (NCCN) and others recommend earlier and more frequent surveillance for individuals identified as having HBOC or HNPCC because of the significantly higher risk of a second cancer in these patients.[9-11] For example, patients with breast cancer due to BRCA mutations have a 52 to 64 percent chance to develop a second breast cancer in their lifetime and at least a 16 percent chance to develop ovarian cancer after breast cancer.[10,11] Patients with colon cancer due to HNPCC have a 50 percent chance to develop a second cancer within 15 years.[12]

Surveillance can include semi-annual mammograms and/or breast MRI, transvaginal ultrasound examination, semi-annual blood work for carcinogenic markers, and annual colonoscopies.[13-16] Risk-reduction surgery, such as oophorectomy or prophylactic mastectomy, is another option presented to individuals with HBOC.

A cancer risk assessment and genetic testing program allows these patients to be identified and placed on the NCCN-recommended intensive management program. This program has been shown in several studies to have numerous patient benefits such as:

- Increased compliance with recommended screening[17,18]
- Reduced incidence of cancer[17,19,20]
- Increased detection of early-stage cancer.[17,19,20]

Perhaps most importantly, the NCCN-recommended intensive management program can lengthen the lives of this high-risk patient population.[21] One recent study that looked at the gain in life from prophylactic oophorectomy and mastectomy for a BRCA mutation carrier found an estimated 4.9 year gain in life expectancy for a 30-year-old BRCA mutation carrier undergoing these surgeries.[21] This study compares favorably to the estimated 0.9 year gain in life expectancy from adjuvant chemotherapy in a 45-year-old woman with breast cancer.[22]

The benefits of a cancer risk assessment program and genetic testing extend well beyond those patients already

diagnosed with cancer to the patient's family members. Once an individual with cancer is found to carry a harmful mutation, his or her relatives can be tested for that specific family mutation. Genetic testing can determine which family members are truly at high risk based on whether they inherited the family mutation that causes the cancer in this family. While those family members who have inherited the mutation will need intensive surveillance and maybe even prophylactic surgeries, family members who did *not* inherit the mutation can follow normal population risk-screening guidelines—despite the family history.

A cancer genetic program will also reach a sizeable number of people who have not yet been diagnosed with cancer. These individuals may be referred by a healthcare provider or may self-refer due to a family history of risk. Of those cancer-free patients who choose to go ahead with genetic testing, a small percentage will be found to carry an inherited cancer susceptibility mutation. Depending on the inherited cancer syndrome identified, these individuals may also benefit from the appropriate NCCN-recommended surveillance protocol. Even high-risk patients who choose not to undergo testing or patients who are negative for an inherited susceptibility mutation may be candidates for earlier or more frequent surveillance.[15, 16]

Beyond the clinical benefits, a cancer risk assessment program can:

- Help differentiate a community cancer center from its competition
- Enhance the cancer center's reputation within the physician and at-large community
- Assist patients in making more informed healthcare decisions
- Help the cancer program comply with the latest American College of Surgeons (ACS) Commission on Cancer 2004 Program Standards, which have added genetic counseling and testing as a supportive service.[23]

DEVELOPING A CANCER RISK ASSESSMENT PROGRAM

A hereditary cancer risk assessment program is largely a patient education service. For the individual deciding whether to undergo testing, a cancer risk assessment program can provide genetic education, offer an assessment of the individual's own cancer risk, provide information on the benefits and limitations of genetic testing, and facilitate testing when appropriate.

As a first step in program development, typically, an oncology healthcare provider will identify patients that may benefit from cancer risk assessment. The practitioner's decision is based on multiple factors, including the patient's history of early-onset cancer or a family history of cancer. At this time, the patient being referred is asked to gather family history information and fill out forms to assist in insurance verification.

Next, the patient meets with a physician, nurse educator, genetic counselor, or other appropriate healthcare provider to understand his or her risk, the benefits and limitations of genetic testing, and other essential information that makes up informed consent, as defined by ASCO.[6] If the patient goes through with the decision to be tested, the patient then has blood drawn for genetic testing. Finally, the patient meets with his or her provider to receive the results of the testing and plan further management.

Establishing a cancer genetic risk program requires an investment of time and resources; internal and external physician support is critical to success of the program.

The single most important characteristic in developing a strong risk assessment program seems to be the selection of an appropriate patient educator. Traditionally, a genetic counselor (i.e., an individual with a master's degree in genetic counseling from an accredited genetic counseling training program) has been identified as the provider ideally suited and trained to fill this role. The challenge is that the current number of genetic counselors available to provide education and risk assessment is too small to meet the needs of the large number of individuals at risk of having an inherited cancer. A professional status survey conducted by the National Society of Genetic Counselors found that only 140 genetic counselors reported spending more than 50 percent of their time working in oncology.[24]

Because of the shortage of genetic counselors, many community cancer programs are looking towards other practitioners, such as oncology nurses, physician assistants, nurse practitioners, breast health specialists, and/or oncology social workers to act as patient educators for cancer risk assessment and genetic testing. In these instances, patient educators should receive additional

The global genetic "catalog" encoding all of life's amazingly diverse capabilities is astonishing, yet very few details are known.

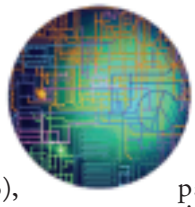

ILLUSTRATION AND CAPTION/U.S. DEPARTMENT OF ENERGY GENOMICS: GTL PROGRAM

training and education specific to genetics and cancer genetics. Such training may be found in course offerings from organizations such as ASCO, the Oncology Nursing Society (ONS), and the American Medical Association (AMA); monographs on genetic testing presented at professional meetings; university-based courses; and industry-sponsored opportunities.

Once physician support has been established and an appropriate patient educator is in place, a few logistical questions must be answered. For example, a decision must be made as to where the patient education will take place. You must also put in place a method for identifying cancer patients that may benefit from the genetic risk assessment program. How will such patients access the program? Finally, some thought and planning must be put into deciding how the program will be presented and mar-

Revenue Impact Case Study

Aurora Healthcare, Milwaukee, Wisc.

According to its 2001 Annual Report, 1,024 patients were diagnosed and treated for breast or colon cancer at one Aurora Healthcare Hospital. Using a conservative estimate of 7 percent mutation prevalence in breast cancer cases, it can be reasonably assumed that approximately 73 patients diagnosed and treated at this hospital had cancer that was due to an inherited susceptibility (26 with inherited colon cancer and 47 with inherited breast cancer.)[1,30] These patients would usually receive the same management and care as the other 951 patients even though patients with an inherited cancer are diagnosed at significantly younger ages and are at up to a 64 percent risk of a second cancer.[9]

ILLUSTRATION/GETTYIMAGES.COM

Using typical selection criteria to identify all 73 patients diagnosed with inherited cancer, a total of five times the number of patients (365 patients) will need to undergo risk assessment and testing.[25] Assuming $200 of revenue from each patient undergoing genetic testing, the cancer risk assessment program will bring in approximately $73,000 in revenue for those 365 patients. (The $200 is an average reimbursement rate for two office visits based on Medicare's average allowable reimbursement of $79.82 to $132.06.)

Once testing has identified the 26 patients with inherited colon cancer, more revenue will be generated from additional colonoscopies. For our purposes, we can assume $681 in additional revenue for each patient or approximately $17,706 total.[26]

For the 47 patients identified

Playing the Numbers

217,000
The number of people the American Cancer Society estimates will be diagnosed with breast cancer in the United States in 2004.[29]

22 percent
Estimated percentage of individuals presenting with newly diagnosed breast cancer who have personal and family histories that are indicative of hereditary risk.[25]

47,000
The number of breast cancer patients who are candidates for hereditary cancer evaluation, based on the data at left. Similar estimates could be made regarding patients with inherited colon, ovarian, and/or endometrial cancer.

keted to the community and referring physicians.

While these tasks may appear daunting, resources are available to help in the program development process. In addition to materials available through nonprofit organizations like the National Society of Genetic Counselors (NSGC), the International Society of Nurses in Genetics (ISONG), and/or ONS, some genetic testing laboratories offer complimentary educational resources.

In this case study, **the bottom line** *of the cancer genetics risk assessment program shows a net income of* ***approximately $191,600****.*

with breast cancer and BRCA mutations, revenue related to recommended prophylactic oophorectomy may be anticipated to be about $230,000 or $5,000/per patient. [27]

For the cancer risk assessment program in this case study, the overall yearly net revenue from these patients is approximately $320,706.

The next step is to calculate the direct costs to the facility, keeping in mind that most cancer program administrators must at least break even with these services.

In this case study, the cancer risk genetics assessment program is staffed by two genetic counselors with a salary of $47,593 each.[24] Assuming a benefit rate of 23 percent, the total salary of both counselors comes to $117,078. With ancillary supplies (i.e., patient education materials, office supplies, continuing education units) of $6,000 for each counselor, the total directs costs to the facility are approximately $129,078.

In this case study, the bottom line of the cancer genetics risk assessment program shows a net income of approximately $191,600. These revenue statistics are taken from published estimates of net revenue from the various procedures used in the case study. The revenue from education includes both pre- and post-test education.

ECONOMIC IMPACT OF CANCER RISK ASSESSMENT PROGRAMS

While the clinical benefits of providing a cancer risk assessment and genetic testing service are obvious, community cancer programs planning to develop a cancer risk assessment program must also understand the economic impact of providing this service.

The main costs associated with a cancer risk assessment program are personnel related, including the patient educator's salary, which will depend on the percentage of time this staff member dedicates to the program. Typically, no equipment or software purchases are necessary. Other ancillary costs include overhead charges related to the use of office space, telephone and computer charges, business cards, and patient education materials. (See case study on page 24 and 25.)

On the other side of the equation is revenue that is generated both through direct patient contact and downstream revenue. The downstream revenue is generated from the different medical management options available to patients that test positive for certain mutations. These medical management options would not typically be recommended to patients who do not have an inherited cause of their cancer.[13-16] As stated earlier, for patients with HBOC or HNPCC, medical management options may include breast MRI, extra colonoscopies, prophylactic oophorectomy, and/or prophylactic mastectomy.[13-16]

To more fully understand the economic impact of a cancer risk assessment program, you must first identify the population of patients at your facility, as shown in the case study. In this example, a cancer genetic risk assessment program with two genetic counselors generated a net income of approximately $191,600.

Whether your cancer center decides to add a cancer genetics risk assessment program or not, many patients are expressing interest in the service and finding that the cost of such testing is well worth the concurrent benefits. As mentioned previously, individuals who test positive for genetic mutations may be able to extend their life expectancy, reduce their cancer risk through chemotherapy and surgery, and/or educate their family members about the likelihood of a family mutation.[28] Even individuals who undergo genetic testing and come up negative for genetic mutations may be reassured about their chance of cancer risk due to heredity and avoid intensive and costly monitoring and prevention strategies, as well as aggressive interventions such as risk-reducing surgery.[28] The bottom line: Offering cancer genetic risk assessment services can help your patients make important healthcare decisions.

Christopher Ho, MS, is a genetic counselor and area

…individuals who **test positive for genetic mutations**
may be able to extend their life expectancy,
reduce their cancer risk through *chemotherapy*,
and *surgery*…and/or educate their family members…

manager for Myriad Genetics Laboratories, Inc., in Oak Creek, Wisc.

REFERENCES

[1]Claus EB, Schildkraut JM, Thompson WD, et al. The genetic attributable risk of breast and ovarian cancer. *Cancer*. 1996; 77:2318-2324.

[2]Risch HA, McLaughlin JR, Cole DE, et al. Prevalence and penetrance of germline BRCA1 and BRCA2 mutations in a population of 649 women with ovarian cancer. *Am J Hum Genet*. 2001;68:700-710.

[3]Cruz-Correa M, Giardiello FM. Diagnosis and management of hereditary colon cancer. *Gastroenterol Clin North Am*. 2002;31: 537-549.

[4]Lynch HT, Smyrk T, Kern SE, et al. Familial pancreatic cancer: A review. *Semin Oncol*. 1996;23(2): 251-275.

[5]Association of Community Cancer Centers: *Standards for Cancer Programs*. Available online at: *www.accc-cancer.org*. Accessed September 9, 2004.

[6]American Society of Clinical Oncology Policy Statement. Update: Genetic testing for cancer susceptibility. *J Clin Oncol*. 2003;21(12):2397-2406.

[7]American Gastroenterological Association. American Gastroenterological Association medical position statement: Hereditary colorectal cancer and genetic testing. *Gastroenterology.* 2001; 121:195-97.

[8]Peltomäki P, Vasen HFA. The ICG on HNPCC. Mutations predisposing to hereditary nonpolyposis colorectal cancer: Database and results of a collaborative study. *Gastroenterology*. 1997;113:1146-1158.

[9]Ford D, Easton DF, Bishop DT, et al. Risks of cancer in BRCA1 mutation carriers. *Lancet*. 1994:343:692-695.

[10]The Breast Cancer Linkage Consortium: Cancer risks in BRCA2 mutations carriers. *JNCI*. 1999; 15: 1310-1316.

[11]Frank TS, Manley SA, Olopade OI, et al. Sequence analysis of BRCA1 and BRCA2: Correlation of mutations with family history and ovarian cancer risk. *J Clin Oncol*. 1998;16(7):2417-2425.

[12]Mecklin JP, Jarvinen HJ. Clinical features of colorectal carcinoma in cancer family syndrome. *Dis Colon Rectum*. 1986; 29:160-164.

[13]Burke W, Daly M, Garber J, et al. Recommendations for follow-up care of individuals with an inherited predisposition to cancer.II. BRCA1 and BRCA2. *JAMA*.1997;277(12):997-1003.

[14]Burke W, Petersen G, Lynch P, et al. Recommendations for follow-up care of individuals with an inherited predisposition to cancer.I. Hereditary nonpolyposis colon cancer. *JAMA*. 1997; 277(11):915-919.

[15]National Comprehensive Cancer Network. Clinical practice guidelines in oncology – v.1.2003. Genetic/Familial high-risk assessment: Breast and ovarian. Available online at: *www.nccn.org*. Accessed September 9, 2004.

[16]National Comprehensive Cancer Network. Clinical practice guidelines in oncology – v.1.2003. Colorectal screening. Available online at: *www.nccn.org*. Accessed September 9, 2004.

[17]Scheuer L, Kauff ND, Robson ME, et al. Outcome of preventative surgery and screening for breast and ovarian cancer in BRCA mutation carriers. *J Clin Oncol*. 2002;20(5):1260-68.

[18]Botkin JR, Smith KR, Croyle RT, et al. Genetic testing for a BRCA1 mutation: Prophylactic surgery and screening behavior in women 2 years post testing. *Am J Med Genet*. 2003; 118A(3):201-209.

[19]Kauff ND, Satagopan JM, Robson ME, et al. Risk-reducing salpingo-oophorectomy in women with a BRCA1 or BRCA2 mutation. *N Engl J Med*. 2002;346(21):1609-15.

[20]Jarvinen HJ, Aarnio M, Mustonen H, et al. Controlled 15-year trial on screening for colorectal cancer in families with hereditary nonpolyposis colorectal cancer. *Gastroenterology*. 2000;118:829-34.

[21]Schrag D, Kuntz KM, Garber JE, et al. Life expectancy gains from cancer prevention strategies for women with breast cancer and BRCA1 or BRCA2 mutations. *JAMA*. 2000;283(5):617-24.

[22]Malin JL, Keeler E, Wang C, et al. Using cost-effectiveness analysis to define a breast cancer benefits package for the uninsured. *Breast Cancer Res Treat*. 2002;74(2):143-53.

[23]American College of Surgeons. Commission on Cancer Program Standards 2004.

[24]National Society of Genetic Counselors. Professional Status Survey 2002. Available online at: *http://www.nsgc.org/pdf/PSS_2002_2_22.pdf*. Accessed September 9, 2004.

[25]Frank TS, Deffenbaugh AM, Reid JE, et al. Clinical characteristics of individuals with germline mutations in BRCA1 and BRCA2: Analysis of 10,000 individuals. *J Clin Oncol*. 2002;20(6):1480-90.

[26]Sonnenberg A, Delco F, Inadomi JM. Cost-effectiveness of colonoscopy in screening for colorectal cancer. *Ann Intern Med*. 2000:133(8):573-584.

[27]Levine A, Hughes KS. Cost-Effectiveness of the Identification of Women at High Risk for the Development of Breast and Ovarian Cancer. In: Victor G. Vogel (ed.) *Management of Patients at Risk for Breast Cancer*. Oxford: Blackwell Science Inc, 2000.

[28]National Cancer Institute. *Elements of Cancer Genetics Risk Assessment and Counseling*. Available online at: *http://www.nci.nih.gov/cancertopics/pdq/genetics/risk-assessment-and-counseling*. Accessed September 9, 2004.

[29]Ford D, Easton DF, Stratton M, et al. Genetic heterogeneity and penetrance analysis of the BRCA1 and BRCA2 genes in breast cancer families. *Am J Hum Genet.* 1998;62:678-689.

[30]American Cancer Society. Cancer Facts and Figures 2004. Available online at: *www.acs.org*. Accessed June 25, 2004.