Genetics 101

BY LINDSEY BYRNE, MS, CGC

As a genetic counselor, I am often asked about the numerous terms associated with hereditary cancer genetic testing—from patients and providers alike. Below are answers to questions I am commonly asked.

How Often is Cancer Hereditary?

Most cancer occurs by random chance and is not genetic. Sometimes, cancer is caused by a combination of different genetic and environmental risk factors like smoking or exposure to certain chemicals. Only rarely (5%-10% of cases) is cancer hereditary. This means that a single change in a gene puts a person or family at high risk of developing cancer. The specific genetic change is related to the level of risk and types of cancer that a person may develop.

What Is Genetic Testing?

Genetic testing involves analysis of gene makeup and sequences in cells. Genes are the instructions in cells that act as recipes for different proteins needed by the body to stay healthy. A small number of these genes help prevent cancer. Some people inherit a mutation, a harmful change in 1 of these genes that puts them at higher risk of developing a certain kind of cancer.

Hereditary *or* ***germline genetic testing*** looks for risks for cancer that are inherited and that could be passed on to other family members. If you have a positive germline test result, it does not mean you have or will get cancer—it just means that your chances are higher. It will explain why some individuals are diagnosed with cancer. Other relatives (eg, parents, children, and siblings) who may also be at risk for cancer could then make decisions about ordering genetic testing for themselves.

Somatic genetic testing examines a person's cancer, tumor, or blood. This testing usually looks for genetic changes that are associated with the tumor (somatic mutations) and that may be a target for treatment or can explain features of a cancer.

Research genetic testing can be performed to identify new genes that may predispose people to cancer. Researchers continuously search for additional genes that can be associated with cancer in individuals and their families.

The Role of the Genetic Counselor

Genetic counselors are an important part of the health care team. They guide and support patients seeking more information about how inherited cancer may affect them or their families and interpret genetic testing results based on personal and family histories of cancer. There is currently a high demand for hereditary genetic testing and, thus, genetic counselors.

Genetic counselors work closely and communicate frequently with other members of the multidisciplinary cancer care team that includes physicians, advanced practice providers, and nurses. Some genetic counselors are embedded within smaller teams that work in a single department (eg, those who work with patients having breast or gastrointestinal cancer), but most are not limited to 1 type of cancer.

Genetic counselors often provide an informed perspective on the benefits and limitations of germline testing and collaborate with physicians on the results of testing and what they mean for their patients.

A Day in the Life of a Genetic Counselor

For genetic counselors, tasks vary from day to day. I balance work among clinical, research, and education responsibilities, but not all genetic counselors have these same responsibilities. Two days each week, I complete clinical tasks such as counseling patients in person, over the phone, or over video and offering and ordering genetic testing. Many genetic counselors have the flexibility to adjust the format of appointments based upon patient need and their organization's policies. When I am not counseling patients, I am involved in the Ohio State University genetic counseling training program as a course director, clinical student supervisor, and research thesis committee advisor or member.

Additionally, I work on numerous research studies; specifically, I work on the *BAP1* gene research study. The purpose of this study is to identify and better understand the cancer and skin tumor risks associated with having a *BAP1* mutation. At present, the information we have on these risks was obtained from a fairly small number of selected patients; these data may not accurately reflect the true risks for people with *BAP1* mutations. Thus, we are enrolling individuals and their families who are *BAP1* positive to continue to better understand this syndrome.

Reimbursement Outlook

Some genetic counselors bill for their genetic counseling services by using a facility fee or through joint appointments with a physician.

Many, however, do not, because genetic counselors are not recognized nationally by the Centers for Medicare & Medicaid Services as providers. Unfortunately, as a result, many patients are unable to access genetic counseling services.

Many medical professionals and societies, including the National Society of Genetic Counselors, are advocating for the passage of the Access to Genetic Counselor Services Act, which would, among other changes, provide status for genetic counselors under Medicare Part B. This policy change would allow direct beneficiary access to genetic counseling services and provide direct reimbursement to genetic counselors for services furnished under Medicare Part B.

There are some ways that genetic counselors can bill within their clinics. For example, genetic counselors who run a clinic independently can bill a facility fee. But administrators of smaller facilities find it difficult to pay for a provider who is not able to get directly reimbursed for services. In other words, smaller cancer programs and practices find it challenging to hire genetic counselors because they cannot bill for their services.

Every cancer program has a different policy regarding phone, video, and in-person billing. Some grant flexibility in appointment format, which is critically important to patients who live in rural locations, underserved areas, or regions of the country with limited access to genetic counselors, while other cancer programs only offer in-person appointments.

A Growing Need

As more national guidelines recognize the importance of genetic testing in cancer, the demand for genetic counseling and testing will increase. National Comprehensive Cancer Network guidelines now include more individuals with cancer than they did in years past. Genetic testing should be considered at any age if results can aid in medical management such as:

- Supporting treatment decision-making for a current cancer diagnosis
- Determining eligibility for specific clinical trials or research studies focusing on cancer prevention strategies
- Improving cancer screening plans for individuals and their families for multiple cancer types.

At The Ohio State University Medical Center, we are constantly evaluating the needs of our patients and families. Our main location is in Columbus, Ohio, but we also currently support 2 hospitals that are over an hour away. We plan to continue to add additional locations to expand genetic counseling services to our greater communities. Overall, genetic counselors tend to be accessed in larger cities, and we are working to reduce location as a barrier to care by allowing individuals to access genetic counseling and testing closer to home.

Recently, 2 additional genetic counselors were hired. One of these genetic counselors will work specifically with our affiliate sites via telehealth. The second genetic counselor was hired as referrals for cancer genetics continued to increase year after year. Much of this greater demand has involved referrals for individuals with breast cancer. As the guidelines continue to expand for patients who should be recommended for cancer genetic testing, leaders of our cancer program will monitor the need to hire additional genetic counselors. OI

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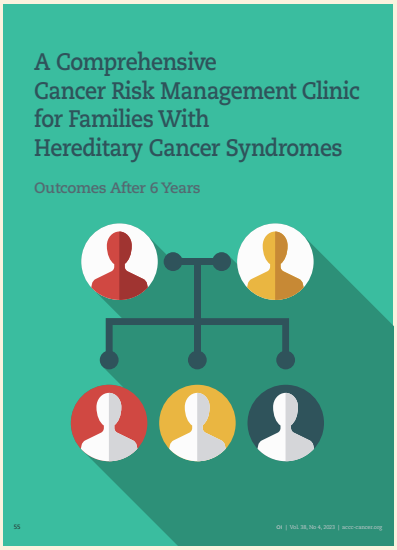

ICYMI: A Comprehensive Cancer Risk Management Clinic for Families with Hereditary Cancer Syndromes

With more individuals and families needing long-term cancer risk management, Aurora Health Care in Milwaukee, Wisconsin, developed a hereditary cancer center staffed by a medical oncologist, cancer nurse coordinator, genetic counselors, and support staff. Over a 6-year period, 889 patients and relatives established care at this hereditary cancer center. Each patient learned of their cancer risk(s) based on their genetic testing results, cancer risk management plan, and recommendations for lifestyle modification. In this 2023 *Oncology Issues* article, read how this clinic provides longitudinal care and collects data to track several outcomes.