Know Your Genes

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Between 5 and 10 percent of cancers are hereditary, meaning they are passed on genetically from parents to children. Genetic testing looks for specific changes, or mutations, in a person's genes that can increase their risk of developing certain cancers. Although only a small percentage of cancers have a known genetic cause, identifying individuals with these mutations—and their family members—can lead to early awareness, treatment and, in some cases, prevention.

What is a genetic counselor?

Cancer genetic counselors are a lot like detectives. Most cancers occur because of random changes in your body over your lifetime, but some are hereditary. People are referred to us when it's suspected that cancer could be running through the genes in a family.

Our role is to assess the family history to see whether genetic testing might be appropriate, educate a person about which tests are needed, discuss what their results mean and provide psychosocial support. If the test is positive, we give them information about what they need to do next and help coordinate their care going forward.

How do you accomplish this?

We take a detailed family history on both the mom's and the dad's side going back three generations. This helps us assess the likelihood that a mutated gene is causing cancer in the family. Red flags include cancers developing at an earlier age than expected, the same or related types of cancer in multiple relatives, certain rare cancers or a family member with a known mutation.

We share the results of the history and whether criteria for genetic testing are met. If so, we suggest appropriate testing, discuss how each of the possible results would impact the patient and family members and determine what the next steps would be. If the person consents, blood is drawn and sent to the laboratory.

What happens next?

Depending on the results, we discuss what they mean to the patient and his or her family. If

positive, the talk is usually lengthier. We discuss cancer risks in detail, according to the mutations identified.

One important issue is the options available to reduce future risk. For most cancers, screening more frequently is recommended for early detection. But for some cancers that are considered hard to screen for or are more aggressive, surgery to remove the organ before malignancy develops could be a possibility.

Anxiety and depression are concerns for many people. Counselors provide support and help patients cope during the genetic testing process. If long-term support is needed, we can link patients with mental health professionals.

One thing that sets genetic testing apart is that the results also have implications for the patient's family. We help the patient communicate that information to relatives so they can also benefit.

These test results can also impact future generations. We discuss the chance of cancer susceptibility being passed on to children. For most inherited cancers, the probability of the mutation being present in offspring is 50 percent.

How does this genetic testing differ from testing genes in a tumor?

Unlike what I do, genetic testing of a tumor is done on someone who already has cancer. It looks at genetic mutations within the malignant cells to help determine the most effective treatment for that particular tumor in that particular person.

What about over-the-counter genetic testing?

The test that the Food and Drug Administration has approved analyzes only three of the thousands of variations in the BRCA1 and BRCA2 breast cancer susceptibility genes (Click here to read "Understanding BRCA"). And these three variations are rarely seen outside of Ashkenazi Jewish communities. So the utility of this test is limited to a very small number of mutations within a very small and specific population group.

What do you find most inspiring or hopeful about your work?

With the significant cancer histories some families have, many people have thought it was a question of not if but when they would develop cancer. Using genetic testing, we can now determine not only who is at risk for hereditary cancer—and therefore who needs enhanced screening and management—but also who is not. We can offer these people reassurance that their risk is similar to the general population's. Genetic testing allows people to be proactive about their own health.
