What Genetic Testing Can Reveal about Hereditary Cancer Conditions

Inherited genetic mutations that can cause cancer are rare, but they can greatly increase cancer risk, says a certified genetic counselor.

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Most cancers occur because of changes in our genes during our lifetime, and various factors – such as smoking, carrying excess weight, drinking alcohol, and exposure to the sun – can increase risk. But 5% to 10% of cancers can be linked to something we're born with: inherited genetic mutations.

That's where Michelle Springer, MS, CGC, and her colleagues come in. For 18 years she has been a certified genetic counselor at the University of Colorado Anschutz Cancer Center's Hereditary Cancer Program.

Genetic counselors advise patients on genetic testing that can unlock clues to their cancer risk that may be encoded in their genes since birth. They also help patients understand their options if they test positive for a hereditary cancer condition, usually including more extensive screening and surveillance for cancer.

"Once we know that there is a genetic mutation in the family predisposing someone to cancer, there is so much more that we can do to be proactive and preventative," says Springer, who is also an assistant professor of clinical practice in the CU Anschutz Division of Medical Oncology. "With early detection, we can actually prevent some cancers and change the outcome for patients and their families."

Springer's advice to patients draws on her personal experience. About seven years ago, she tested positive for a mutation in the ATM gene, which increases the lifetime risk of breast and pancreatic cancer.

"I'm so incredibly grateful I have this information and that I'm able to do things differently in my

own life," she says. "The way I approach this with patients now has really changed. It's given me a much deeper understanding and appreciation of what patients and their families might be going through and experiencing as a result of testing positive."

→ Celebrating Genetic Counseling With the CU Cancer Center's Hereditary Cancer Program

'It's not your destiny'

There are hundreds of genetic mutations that can be passed down from parent to child and increase cancer risk. They help explain why certain cancers develop in successive generations of a family, and also why some unusual characteristics of a cancer – such as a cancer that develops at an earlier age than usual, or one person having multiple cancer types – may recur in some families.

Springer emphasizes that having an inherited genetic change like this does not guarantee a person will develop cancer – "it's not your destiny," she says – but it does mean that a person's risk of certain cancers may be higher than average. Most hereditary cancer mutations are dominant in how they are inherited, she says, which means there's a 50% chance the mutation will be passed down to a person's offspring.

Most inherited cancer conditions are uncommon. Typically they are caused by genes that normally suppress tumors.

"We all have these genes, and when they are working properly, they actually help prevent cancer," Springer says. "But when an individual is born with a mutation in one of these genes, it predisposes them to develop certain cancers or tumors."

Mostly rare conditions

Breast, ovarian, colorectal, prostate, and pancreatic cancers are those most often associated with hereditary cancer conditions.

Among the most prevalent of these conditions is hereditary breast and ovarian cancer syndrome

(HBOC). Nearly half of HBOC is caused by mutations in either the BRCA1 or BRCA2 gene, Springer says, and it is estimated that one out of every 400 to 500 people carry a BRCA mutation. Mutations in other genes can predispose a person to hereditary breast cancer as well, she says.

Mutations in the BRCA1 and BRCA2 genes are linked to 5% of breast cancers in women in the United States and about 15% of ovarian cancers. Among men, HBOC can increase the risk of male breast cancer and prostate cancer. The risk for pancreatic cancer is also increased, Springer says.

→ Genetic Counseling Helps Young Woman Take Control After Testing Positive for BRCA2

Another inherited cancer condition is Lynch syndrome, which affects about one in 300 people. It is estimated that 3% of all colorectal cancer is caused by Lynch syndrome, Springer says. Due to the condition being highly underdiagnosed, all colon tumors are now screened for markers related to it. Caused by mutations in genes that repair DNA mismatches, Lynch syndrome significantly increases lifetime risk of developing colorectal, uterine, and ovarian cancers.

Other hereditary cancer conditions are more rare, at least in the U.S. They include Li-Fraumeni syndrome (affecting an estimated one in 5,000 to one in 20,000 people), caused by mutations in the TP53 tumor suppressor gene, leading to a higher risk of early-onset cancers, including breast cancer, brain tumors, sarcomas, and leukemia. And Von Hippel-Lindau syndrome (affecting an estimated one in 36,000 people), caused by mutations in the VHL gene, can cause tumors and cysts to grow in various parts of the body.

→ Elijah Johnson Came to the CU Anschutz Cancer Center to Research the Mutation That Makes Him More Likely to Develop Cancer

‘Hugely powerful’

At the CU Anschutz Cancer Center, Springer is part of a team that offers cancer genetic counseling – known as a personalized cancer risk assessment – either face to face or through a telehealth program that Springer founded in 2019. Genetic counselors advise people on whether they may benefit from genetic testing, what tests may be right for them, how to understand the results, and explain screening options available if someone chooses not to be tested or tests negative. Reviewing non-medical considerations is also an important part of the discussion, she says.

Most people are referred by an oncologist, primary care provider, or other specialist, Springer says. Some individuals self-refer, trying to better understand if the cancer in their family could be due to an inherited predisposition.

Common reasons to consider genetic counseling and testing for hereditary cancer conditions include:

- You or someone in your family was diagnosed with breast cancer or colon cancer before age 50.
- You or a relative had pancreatic cancer, ovarian cancer, or a rare type of cancer at any age.
- Many relatives have had cancer, especially if they were young at diagnosis.
- You or a relative has had more than one type of cancer.
- The same type of cancer is diagnosed in two or more relatives, especially if diagnosed under the age of 50.

Genetic testing is performed by collecting a small biological sample – most commonly blood or saliva – which is sent to a laboratory for analysis. “We can coordinate genetic testing through a saliva kit that we send to your house,” Springer says.

’An incredible team’

Information gathered through a genetic test is “hugely powerful to the patient and their family. It can allow them to do things differently moving forward,” Springer says. “If we know there is a genetic susceptibility or predisposition to cancer, we can increase screening and surveillance. That allows us to detect things at an earlier stage, or in some cases, even prevent some cancers. Knowledge is power, and knowing this information can truly change the outcome in families and save lives.”

Steps following a positive test for hereditary cancer conditions vary widely depending on the

potential cancer involved. They can include earlier or more frequent screenings for cancer, lifestyle adjustments, chemoprevention therapy, and surgery to remove high-risk tissue. There may also be a “lower threshold for response” if a patient develops symptoms suggesting cancer, Springer says.

For those already diagnosed with cancer, genetic testing “can help an individual and their family understand the underlying cause of the cancer,” Springer says. It can also play an important role in treatment for some individuals.

Springer says she’s grateful to be working with “such an incredible team” caring for patients at the cancer center.

“I truly can’t imagine working for a different organization. Patients come from all over to see our providers and take advantage of our clinical trials. At our multidisciplinary clinics where these patients are first seen, we have brilliant minds from different specialties putting their heads together and saying, ‘What is the best treatment plan for this patient?’ And certainly, genetic testing is part of that conversation.”

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