Genetic testing could be a lifesaver

Regular mammograms are also needed

PARTNER CONTENT FOR COMPREHENSIVE CANCER CENTERS

Mammograms save lives. When it comes to breast cancer detection, there's another tool worth paying attention to — especially those with a family history of the disease: genetic testing.

According to the Susan G. Komen Foundation, a high-risk gene mutation can increase a woman's lifetime risk for breast cancer from 13% up to 80%. Nationwide, up to 10-15% of breast cancer cases are related to an inherited gene mutation. And that's nothing to scoff at.

"When we're talking about the total number of breast cancer cases in Nevada, 10 percent is a large number of individuals where we can potentially catch their cancer earlier or prevent it altogether," said Ali Khalaf, MS, CGC, oncology genetic counselor with Comprehensive Cancer Centers.

The test itself is relatively simple, performed either by saliva, buccal swab or a blood draw. It has advanced significantly since the 1990s, when only BRCA1/2 mutations could be identified. Back then, it came with a hefty price tag and wasn't largely covered by insurance. Today, most insurance plans cover testing for patients who meet national criteria. And, today's tests can detect many other inherited mutations, such as in ATM, CHEK2 and PALB2 genes.

Genetic testing does not replace mammograms, an essential first step in detection recommended annually for women starting at age 40 who are at general risk. Instead, genetic testing allows us to tailor more personalized screening recommendations for woman who are at increased risk, such as beginning mammograms earlier then 40 as well as whether an individual could benefit from more aggressive forms of screening, such as utilizing both mammograms and breast MRIs at a more frequent interval.

"Genetic testing in the oncology setting allows us to identify individuals with inherited predispositions to cancer before they develop disease," Khalaf said. "That can pretty much save lives."

Who should get tested

The test is most often recommended for women with close relatives who developed breast, ovarian, pancreatic or metastatic prostate cancer, especially if those diagnoses came before the age of 50. People diagnosed with breast cancer before the age of 50 should also consider testing, as well as women with triple-negative breast cancer diagnosed at age 60 or younger, people with multiple primary cancers and families with multiple cancers. Families with cases of male breast cancer also are considered high-risk candidates.

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> ~ Ali Khalaf, MS, CGC, Oncology Genetic Counselor
> Comprehensive Cancer Centers

Certain populations also carry a higher inherited risk. For example, Ashkenazi Jewish women are more likely to have BRCA1/2 mutation that sharply increase the likelihood of breast and ovarian cancers.

Some patients fearing their DNA might be misused have opted out of testing. But the Genetic Information Nondiscrimination Act (GINA) is a federal law that prohibits health insurers and employers from discriminating against people based on their genetic information.

How genetic tests help

Tests help providers move away from one-size-fits-all medicine. For people with a breast cancer diagnosis, Khalaf said genetic testing's value is immense.

"We can make more tailored recommendations for surgery so that they do not develop another breast cancer down the road," he said. For example, a bilateral mastectomy may be recommended rather than a lumpectomy, due to a greater chance of a recurrent or contralateral breast cancer forming.

It also means the patient's close family members can decide if they want to test for gene mutations. This offers individuals empowerment over their health by increasing the likelihood of breast or other cancers being caught early, when it's the most easily treatable and leads to the most favorable outcomes.

"For a young woman who tests positive for BRCA1/2, screening starts much earlier than age 40 for mammograms, and more aggressively — MRIs plus mammograms —and preventative surgery may also be an option," Khalaf said.

The emotional weight

Khalaf explains that processing a positive result can be difficult, especially when there's a recommendation for a preventive surgery to avoid a cancer diagnosis. For some, guilt follows as individuals worry whether they've passed a harmful mutation to children. "That's why genetic counselors are trained in psychosocial support. Anxiety, guilt, fear, they're all normal reactions," Khalaf said. "Some moments demand pausing the cancer conversation altogether and just meeting the patient where they are emotionally," he said.

Local Impact

In Nevada, where the state's breast cancer mortality rates remain among the highest in the nation, the stakes are high. Khalaf said currently the largest group of women undergoing genetic testing is those who have already been diagnosed. However, the real game-changer would be more high-risk women being tested before diagnosis.

"There's no denying that increasing awareness and increasing access to this genetic testing will overall decrease the cancer burden in our society," said Khalaf. "Will this decrease the overall burden of breast cancer in Nevada? 100 percent," he said.

The promise and the limits of access

Access to genetic testing remains uneven in Nevada. Khalaf is currently the only cancer genetic counselor practicing in the state; however, nurse practitioners at Comprehensive Cancer Centers also have been trained.

This shortage of specialists means many women never hear about the option. Awareness among providers has been a challenge. Comprehensive Cancer Centers has made outreach a priority, sending Khalaf to OB/GYNs, primary care physicians, and women's health clinics to explain when genetic testing may be appropriate.

For people who are curious about genetic testing, they can schedule a consultation. During these meetings, Khalaf reviews the patient's history and runs a breast cancer risk model, which considers factors like dense breast tissue that make mammograms less effective. These models help determine if additional screening, such as MRIs, would be beneficial, even in the absence of an identifiable genetic predisposition.

He walks people through their options, including cost and dealing with insurance —and there's no pressure to test. But if they do, he says the goal is not just to know more — but to act on what's learned.

"The last thing we want to do is hand over a positive result and not know what to do with it," he said. "We only test when we know there's a clear path forward".

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