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Testing genes for future risks

High-tech procedure can help determine likelihood of disease

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Last year, Penny Jankovich was diagnosed with cancer in both breasts.

A month after undergoing a lumpectomy, the Acworth woman, who was adopted and unaware of her biological family's medical history, received a genetic risk assessment to determine whether she carried the gene that would give her a greater than 70 percent chance of recurrence of breast and ovarian cancer. The results were positive.

"I thought I'd be on my way, but this little genetic test has changed everything, and thank God for it," said Jankovich, 46, who will undergo a double mastectomy with reconstruction and a hysterectomy. "Without it, I would've been a ticking time bomb."

Genetic testing, performed on a sample of blood, hair, skin or other tissue, identifies changes in chromosomes, genes or proteins associated with inherited disorders. The results can confirm or rule out a condition or your chance of developing or passing a genetic disorder to other family members who share some common genetic material.

Genetic testing is available for more than 1,500 disorders and diseases, including neurological and cardiovascular diseases, according to the National Society of Genetic Counselors. Cancer, for instance, is genetic —- linked to a change or mutation in a gene —- but less than 10 percent of cancer is hereditary.

"We're kind of looking for a needle in a haystack," said Kimberly King-Spohn, a genetics counselor with WellStar Kennestone Hospital, which began conducting genetic risk assessment in December.

A lot of cancers are part of syndromes, so if you test positive for a gene-causing cancer, you might be at a risk for other forms. If there are several family members who have the same type of cancer or developed cancer at a younger age, there may be an increased risk of a hereditary cancer syndrome link. So testing primarily targets people who have such a strong family history.

Carol Belcastro is one such case. Her father died of pancreatic cancer and two of her first cousins were diagnosed with breast, ovarian and kidney cancer. Belcastro's genetic testing confirmed that she and one of her sisters carried the gene, making them predisposed to breast and ovarian cancer.

The Lilburn woman underwent a partial mastectomy and a total hysterectomy. Her sister opted to monitor her health with mammograms.

"My decision was a no-brainer after seeing what both of my cousins and Dad went through," said Belcastro, 47, who has two daughters with a 50 percent chance of inheriting the gene. "I had the gift of prevention, and I felt I was extremely fortunate to be proactive."

Genetic testing equips people with the information to better manage their health and make decisions about health risks, but it cannot detect all genetic changes that can cause disease. Other genetic and environmental factors, lifestyle choices and family medical history also affect a person's risk of developing disorders.

"It's not about telling people what to do," said Angela Trepanier, president of the National Society of Genetic Counselors. "It's to provide the information to help people make informed decisions based on their own values and beliefs."

Still, genetic testing is helping doctors learn more about disease and treatment, said Andrew Faucett, director of the Genomics & Public Health Program at Emory School of Medicine, which has conducted genetic risk assessment for about 30 years, primarily for research and on infants. Instead of treating people after they get sick, genetic testing will allow doctors to identify a gene, better understand the illness and then target treatment. Eventually, doctors might be able to slow the onset or progression of a disease or even stop it from happening, he said.

"We've just opened the door for genetic testing to change the way medicine works," said Faucett. "Genetics will give us the tools to catch things before you're ill."

Clip 'n' keep!

WHY ASK WHY?

Genetic testing helps identify genes that may increase your likelihood of developing certain disorders. Here are some of the most common conditions people test for:

> Cystic fibrosis

> Fragile X syndrome (mental impairment)

> Array-based genome hybridization (mental retardation)

> BRCA1/BRCA2 (breast cancer)

> Thrombophilia (blood clotting)

Source: National Society of Genetic Counselors

TALK BEFORE YOU TEST

Before you have genetic testing, talk to a genetics counselor to help you understand your risk and possible outcomes of the test. Talking to a counselor may also eliminate the need for genetic testing as counselors are trained in medical genetics and can evaluate family history to calculate your risk factors.

"Genetic counseling can lessen a person's heightened sense of risk," said Claudia Tinkle, genetics testing manager at DeKalb Medical, one of the first community hospitals in Atlanta to offer the service. "It's peace of mind."

To search for a genetic counselor, visit www.nsgc.org and click on the Find a Counselor link.

OTHER QUESTIONS

If you might go for genetic testing, here are some questions to consider:

> What decisions will you need or want to make once you know the results? Consider the implications of having the test and the impact it could have on you and your family, medically, emotionally and in terms of life planning.

> What are the credentials of the person who will give you the test results? It is important that someone who has expertise in genetics helps you understand and interpret your test results. For example, genetic counselors have specialized training in medical genetics and counseling and earn a master's degree in their field.

> Is the correct test being ordered? Discuss with your genetic counselor which test(s) specifically address your medical concerns. There are many genetic tests, each of which is looking for a different medical condition. If the wrong test is ordered, it may not give you accurate information about your medical concerns.

> How valid are the test results? Some results will be clear while others will be inconclusive or require additional testing.

> What are your next steps once you have the test results? Take time to learn more about what these results mean for your health and who else in your family may be at risk and may need this information.

Source: National Society of Genetic Counselors

COVERAGE AND PROTECTION

Although many insurers, including Medicare, cover genetic testing, many patients fear that a genetic predisposition to disease may lead to elevated premiums or being dropped by their insurer. Others fear that employers will discriminate against them in hiring or promotion decisions.

It is illegal to exclude individuals from a group plan because of their genetic profile, and new legislation has been written to further curtail genetic discrimination. The Genetic Information Nondiscrimination Act prohibits insurers from using genetic information to deny benefits or raise premiums for policyholders. The bill also would bar employers from collecting genetic information or using it to make decisions about hiring, firing or compensation. The bill passed in the House of Representatives, but it has yet to reach the Senate floor.

Source: Staff reports

HOME KITS, RESULTS

The growing market for direct-to-consumer genetic testing suggests that kits may one day hit your local store shelves. Tests include a variety of medical and nonmedical conditions such as baldness, paternity and ancestry, and some are just for entertainment purposes.

There is limited regulatory oversight of these direct-to-consumer testing services, warns the National Society of Genetic Counselors, so the tests may be invalid or unproven, and the results should be interpreted by a genetic counselor. But if you're contemplating using a direct-to-consumer service, the society says to consider the following:

> Are consumer-friendly materials available, developed or reviewed by health care professionals with expertise in genetics (e.g., trained genetic counselors) and suitable for individuals seeking and receiving direct-to-consumer testing services?

> Is information disclosed to potential consumers regarding test purpose, potential limitations, validity and accuracy, using language that is written for consumers?

> Will results be given in a manner understandable to the average consumer, with a clear explanation of their

clinical implications, if any, and including resources providing appropriate follow-up?

> Are patients encouraged to share their medically relevant genetic test results with their health care providers and family members who may also be at risk?

> Are consumer referrals to health care professionals with expertise in genetics available, either on staff or independent of the commercial entity, before and after testing to assure appropriate medical follow-up, including psychological counseling as needed?

> Is there a process for obtaining and documenting informed consent in a manner consistent with accepted medical practices as well as state and local regulations?

> What safeguards are in place to protect the consumer/patient privacy?

> Has the company implemented policies that adhere to testing guidelines and position statements of professional organizations, including the National Society of Genetic Counselors, the American College of Medical Genetics, American Society of Human Genetics and others? These may include relevant guidelines for genetic testing of minors or other potentially vulnerable populations.

> Are the genetic tests performed by appropriately credentialed laboratories ?

NSGC supports consumers' right to access high-quality genetic services, strongly encourages the involvement of appropriately trained clinical genetics professionals in the genetic testing process, and cautions against using direct-to-consumer commercial entities that have not addressed the basic issues outlined above.

Source: National Society of Genetic Counselors

COMMON CANCER MYTHS

The National Society of Genetic Counselors dispels common myths associated with breast and ovarian cancer and genetic testing:

MYTH: Breast and ovarian cancer is always hereditary.

FACT: Only 5 percent to 10 percent of breast and ovarian cancer cases are inherited.

MYTH: Anyone who wants to find out their risk of developing breast and ovarian cancer should get the BRCA1/2 genetic test.

FACT: A BRCA gene test does not test for cancer itself —- only for mutations, or changes, in the BRCA1 or BRCA2 genes that may increase the chance of developing certain cancers over a lifetime. BRCA genetic testing is only for individuals whose personal and/or family history reveals red flags associated with hereditary breast and ovarian cancer. These include:

> onset of breast cancer before age 50

> ovarian cancer at any age

> multiple primary tumors in the same individual

> multiple family members with breast and/or ovarian cancer

> cancer in both breasts or ovaries, or twice in the same one

> presence of male breast cancer

> ethnic background known to have a high frequency of a BRCA1/2 mutation.

MYTH: Breast and ovarian cancer can only be inherited from female family members.

FACT: About half of women who inherit a mutation, or abnormality, in the BRCA1 or BRCA2 genes receive it from their father. Genetic mutations can be inherited from either the mother's or father's side of the family.

MYTH: Men should not undergo genetic testing for breast cancer.

FACT: Men who have a BRCA1 or BRCA2 mutation are at an increased risk to develop cancer, namely breast and prostate, so knowing their genetic status will help them better manage their health. Another reason for men to undergo genetic testing for mutations in BRCA1 and BRCA2 is to determine if they have a mutation that they could pass on to their children.

MYTH: An abnormal (positive) BRCA1/2 genetic test result means breast and/or ovarian cancer will develop in the future.

FACT: A positive test result indicates a heightened risk, but does not mean a person will develop these cancers. Some individuals may inherit the risk factor, but may not develop cancer over his/her lifetime. Individuals with a positive test result should undergo regular screening and consider preventive measures to reduce the risk of developing cancer.

MYTH: A normal (negative) BRCA1/2 genetic test result means a cancer-free future.

FACT: If a person has a strong family history of breast and/or ovarian cancer, they may still have a higher risk of developing these cancers. Not all family histories of breast and/or ovarian cancer will be explained by mutations in the BRCA1 or BRCA2 genes. Other gene mutations may cause cancer and inherited cancer risk.

Source: National Society of Genetic Counselors

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