The NATION'S HEALTH
May 2022

When's it best to get a test? The facts on genetic testing

By Teddi Nicolaus

Have you ever wondered why health care providers ask so many questions about your family? They might ask if any of your relatives had heart disease or diabetes, for example. They aren't being nosy. Your genes — which you share with family — hold secrets to your health.

Genes are the DNA instructions you inherit from your mother and father. It's estimated that every human has as many as 25,000 genes. Our genes can play a role in many things about us, such as if our hair is curly or straight, our food preferences and even our ability to carry a tune. Your genes also contain clues that can provide information about your risk for developing a range of health problems over your lifetime. Those can include high blood pressure, Parkinson's disease, and some types of cancer, including breast, ovarian and colon cancer. Of course, your environment and lifestyle play a big role too.

If you have a family history of certain diseases or conditions, you should chat with your health care team to see if genetic testing makes sense for you. Genetic tests can show if you have changes — called variants — in your genes that can put you at higher risk for certain health problems.

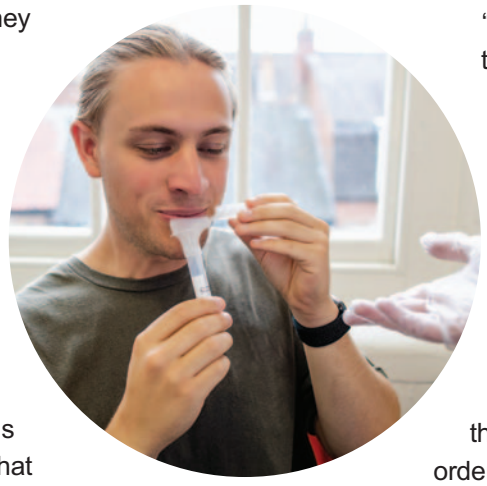
Genetic tests might look at just one, many or all of your genes. The results can help establish a specific diagnosis as well as help physicians tailor a prevention or treatment plan specific to your unique needs. The tests are usually done in a lab using a sample of your saliva or blood.

Over the past decade, the ability to do genetic testing has made it easier for doctors to find out what's making someone sick and to help them, says Marc S. Williams, MD, FACMG, president of the American College of Medical Genetics and Genomics.

"A diagnosis doesn't change the fact that an individual has a condition, but it allows us to treat the condition better," says Williams, professor and director emeritus of the Genomic Medicine Institute at Geisinger.

Knowledge is power

If you're thinking about genetic testing for yourself or a family member, keep in mind there are many different kinds of genetic tests. Start the journey by making an appointment with your physician to see if you should consider genetic testing, and if so, which test is the most appropriate.



"Because of the complexity, we generally recommend that a genetics professional be involved with those decisions as well, to help make sure that the proper test gets ordered," Williams says.

Genetics professionals are key players on your health care team. They can diagnose genetic conditions, select the most appropriate genetic tests for your specific situation, provide counseling, and recommend a personalized treatment plan if a risk is identified.

Keep in mind that the results of genetic tests can greatly impact your life, so the tests won't be administered until you've been counseled about the risks, benefits and limits of testing.

A fear of learning that they or someone they love is at increased risk for a genetic condition can lead many people to avoid important tests that can potentially improve their long-term health. But tests can't always determine if you will develop a disease, or how serious it'll be.

Learning that you carry one of the gene variants associated with a disease or condition doesn't mean you will necessarily go on to develop the disease or condition. It simply means that your risk, compared to the general population, is higher than someone who doesn't have one of those variants.

>> For more on genetic diseases and testing, via www.genome.gov

It is also important to keep in mind that the science is changing rapidly, and "a negative test today will not necessarily be a negative test tomorrow," Williams says.

"The advantage of genetic testing as opposed to almost any other type of testing that's done in medicine is that once we do the test, we can go back to that information and we can reinterpret it using the newest knowledge."

While some genetic tests can be bought online, the best ones are those ordered by your doctor. Be aware, though, that thorough, high-quality genetic testing can cost up to several thousand dollars. Health insurance plans may cover part or all of the cost of genetic testing if it is recommended by your doctor. However, the types of genetic tests covered through private health insurance vary from company to company, so always check with your insurance provider to ask about coverage before you get tested.

Health insurance discrimination concerns have led some people to skip genetic testing. Fortunately, a 2008 federal law made it illegal for health insurers to use testing results to determine eligibility for coverage.

Art and photos courtesy iStockphoto: Illustrations by Bortonia; man with saliva sample, SolStock; happy woman, Julia Amaral; counselor, FatCamera