

Clinical Genetic Testing



Does this test have other names?

Genetic screening, DNA test or testing, chromosomal test, gene testing, DNA-based test

What is this test?

Genetic testing is usually done to screen for or confirm that you have genes that increase the risk for a certain disease. It can be done on newborns, children, or adults. Some parents choose to be tested before or during pregnancy to see whether they are carriers of a certain disease, such as hemophilia.

Your genes are like an instruction book: They hold the blueprint for all your inherited traits, from the color of your eyes, to how tall you will grow.

Your genes can also tell you whether you're at risk of inheriting a disease, such as breast cancer, cystic fibrosis or Huntington disease. They may also help your healthcare provider figure out the best medicines to treat your condition.

A genetic test can find out whether you have damaged, missing, or overactive genes that can cause certain diseases. This test checks your blood or other fluids to look at the pattern (sequence) of the DNA that makes up your genes.

These are different types of genetic tests:

- Gene tests, which look at single genes or fairly short strands of DNA
- Chromosomal tests, which test entire chromosomes or long strands of DNA
- Biochemical tests, which look at protein and enzyme activities

Why do I need this test?

You may need this test, even if you don't have symptoms, if your healthcare provider suspects you have inherited a certain condition. This test can also find out whether your biological child has or might have inherited a disorder from you or the other biological parent.

If you belong to an ethnic or other group in which a certain inherited disease is more common, your healthcare provider may order a test to help determine your risk. Your healthcare provider may also order a test if you have a disease that runs in your family.

Genetic testing is used in the following ways:

- To help your healthcare provider find out the cause of your disease or disorder
- To find out whether you are a carrier of a gene for a disease before or after you have symptoms
- To aid parents in finding out whether an unborn child has a gene for a disease
- To help parents find out whether their newborn baby has a damaged gene that can cause a disease
- To help determine your risk for a certain disease
- To help your healthcare provider find out which medicine or therapy might be best for you

Genetic testing can ease your mind about your or your child's inherited risk for a certain disease if you get a negative result. Finding out you are predisposed to developing a certain disease could lead to your getting more regular screenings or making lifestyle changes to help prevent the disease. Even learning that you have

an inherited disease, painful as the news may be, can help you plan treatment and take other important steps. In some cases, though, positive findings might force you to make some unwelcome choices, so it's your right to choose or turn down genetic testing.

What other tests might I have along with this test?

If you or your child has symptoms of a particular disease, your healthcare provider will likely run specific tests for that disease.

Before you go through any genetic test, your healthcare provider will ask you to get pretest counseling so that you can understand what the test involves, how accurate it is, how you will get the results, and how results will be shared with your healthcare provider. You may also want to ask about emotional and psychological support.

What do my test results mean?

Test results may vary depending on your age, gender, health history, and other things. Your test results may be different depending on the lab used. They may not mean you have a problem. Ask your healthcare provider what your test results mean for you.

If your healthcare provider is looking for a particular gene, your results will be either negative or positive for that gene. Even if you were born with a gene that puts you at risk for a disease, it doesn't necessarily mean you will have it.

The findings from a genetic test may not be simple to understand. Your healthcare provider may refer you to a genetic counselor, who can represent your interests and help translate the results for you.

Genetic counselors can help you understand the science behind genetic testing and the emotional response you may have to the findings. They can also help in researching your family history and looking at your medical records.

How is this test done?

This test can be done on samples of your skin, blood, saliva, hair, urine, or other tissues. You might be asked to give a blood sample or a swab from the inside of your mouth.

Does this test pose any risks?

Having a blood test with a needle carries some risks. These include bleeding, infection, bruising, and feeling lightheaded. When the needle pricks your arm or hand, you may feel a slight sting or pain. Afterward, the site may be sore.

Although this test poses no major physical risks, it can affect your emotions. It should not affect your job status, or your healthcare coverage. The Genetic Information Nondiscrimination Act was passed in 2008 to protect you. The act prevents insurers and employers from using the results of genetic testing to drop your healthcare coverage or to fire you.

Consumers can get genetic testing done with kits that are available on the Internet. Since the results may be complicated and difficult to understand, healthcare providers caution against getting tested without speaking with a genetic counselor first.

Because of the emotional impact of genetic testing, you may want to consider how you will feel if you find out that you or someone you love has a gene that can cause a physical disorder or disease. This may be especially true for a disease such as Alzheimer or Huntington disease, which has no known prevention or cure.

What might affect my test results?

Different kinds of genetic tests yield different information, and results may vary by laboratory. Your results are based on a test of your genes, so lifestyle choices will not affect the findings.

How do I get ready for this test?

You don't need to prepare for this test. Be sure your healthcare provider knows about all medicines, herbs, vitamins, and supplements you are taking. This includes medicines that don't need a prescription and any illegal drugs you may use.

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