



# GENETIC TESTING

WHAT IT MEANS  
for  
YOUR HEALTH  
&  
for  
YOUR FAMILY'S  
HEALTH



## What is genetic testing?

Genetic testing uses laboratory methods to look at your genes, which are the DNA instructions you inherit from your mother and your father. Genetic tests may be used to identify increased risks of health problems, to choose treatments or to assess response to treatments.

## Why might I consider a genetic test?

There are many reasons to consider getting a genetic test. For example, you might decide to undergo a genetic test if you:

- have signs of a disease;
- think you may have an increased risk to get a disease in the future;
- think you might pass a disease on to your children; or
- are pregnant and want your fetus to be tested for a disease.



## What can I learn about my health from genetic testing?

The results of your genetic testing may help to:

- diagnose a disease;
- find gene changes responsible for an already diagnosed disease;
- assess how severe a disease might be;
- guide selection of medicines and other treatments;
- find gene changes that increase risk of developing a disease; or
- find gene changes that could be passed on to children.



## How is genetic testing done?

Genetic tests are done on a small sample of tissue from your body. These tissues may include:

- blood,
- cells swabbed from inside your mouth,
- saliva,
- hair,
- skin,
- tumors, or
- the fluid that surrounds a fetus during pregnancy.

The sample is collected by your health care provider and sent to a laboratory that tests it for certain changes in your DNA. The lab usually gives the test results in writing to your health care provider, who then discusses them with you.

## What are the different types of genetic tests?

Diagnostic testing is used to precisely identify the disease that is making a person ill. The results of a diagnostic test may help you make choices about how to treat or manage your health problems.

Predictive and presymptomatic genetic tests are used to find gene changes that increase a person's likelihood of developing diseases. The results of these tests provide you with information about your risk of developing a specific disease. Such information may be useful in decisions about your lifestyle and health care.

Carrier testing is used to find people who “carry” a change in a gene that is linked to disease. Carriers may show no signs of the disease. However, carriers can pass on the gene change to their children, who may develop the disease or become carriers

themselves. Some diseases require a gene change to be inherited from both parents for the disease to occur.

This type of testing usually is offered to people who have a family history of a specific inherited disease or who belong to certain ethnic groups that have a higher risk of specific inherited diseases.

Prenatal testing is offered during pregnancy to help identify fetuses that have certain diseases.

Newborn screening is used to test babies one or two days after birth to find out if they have certain diseases known to cause problems with health and development.

Pharmacogenomic testing gives information about how certain medicines are processed by an individual's body. This type of testing can help your health care provider choose the medicines that work best with your genetic makeup.

Research genetic testing is used to learn more about the contributions of genes to health and to disease. Sometimes the results may not be directly helpful to you, but they may benefit others by helping researchers expand their understanding of the human body.

### **What are some questions to ask when considering a genetic test?**

If you are trying to decide whether or not to get a genetic test, some questions you may want to ask are:

- Are there any ways to prevent or treat the disease for which I'm being tested?
- Will my health insurance cover the cost of test?
- What safeguards are there to protect the privacy of my test results, especially from insurers and employers?

## **What are some diseases or situations in which genetic tests are currently used?**

- Rare types of breast and ovarian cancers that run in families (BRCA1, BRCA2)
- A rare type of colon cancer that runs in families (HNPCC)
- Iron overload disease
- Cystic fibrosis
- Sickle cell anemia
- Down syndrome in fetuses
- Selection of treatments for breast cancer

## **Should I discuss genetic testing with my health care provider?**

You may want to ask your health care provider about genetic testing if you:

- have a family history of a rare, inherited disease, such as cystic fibrosis or sickle cell anemia;
- have a history of more common diseases, such as cancer, heart disease or stroke, that affects multiple relatives in several generations of your family;
- have parents, siblings or other relatives who have developed before the age of 50 common diseases that typically affect older individuals, such as colon cancer, breast cancer or heart disease; or
- are thinking about having a baby and you or your partner have a family history of inherited disease, or belong to an ethnic group with a higher risk of a specific inherited disease.

## **Who can help me understand genetic testing?**

Your health care provider can help you understand genetic testing or may suggest that you see a genetic specialist to get more information. To find genetic specialists near where you live, go to:

GeneTests

<http://www.genetests.org/>

This site gives information about how to find genetic clinics in your state. Click on “Clinic Directory.”

National Cancer Institute’s Cancer Genetic Services

[www.cancer.gov/search/genetics\\_services/](http://www.cancer.gov/search/genetics_services/)

This site gives you a list of professionals who provide services related to cancer genetics. These services include cancer risk assessment, genetic counseling and genetic susceptibility testing.

## **How can I learn more about genetic testing?**

You can learn more about genetic testing from these Web sites provided by the National Institutes of Health:

**National Human Genome Research Institute**

[www.genome.gov/health](http://www.genome.gov/health)

**National Cancer Institute**

[www.cancer.gov/cancertopics/](http://www.cancer.gov/cancertopics/)

**UnderstandingCancer/genetesting**

**Genetics Home Reference**

<http://ghr.nlm.nih.gov/handbook/testing>

**MedlinePlus**

[www.nlm.nih.gov/medlineplus/genetictesting.html](http://www.nlm.nih.gov/medlineplus/genetictesting.html)

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