Genetic testing can help identify an inherited condition or disease risk. The test results might help you and your doctor:

- Choose ways to prevent or treat a condition.
- Decide which screening tests you need (to find a disease at an early stage when it might be more treatable).

Genetic testing may also tell you which family members are at risk.

But sometimes a genetic test is not the best way to find an inherited condition or disease risk. A routine blood test or procedure might be just as good. And it might be less costly and more easily available.

**Know what to expect.**
Before you have any genetic test, you should understand its possible benefits, harms, and limitations. And you should think about how it might affect others in your family.

**Talk to your doctor or a medical geneticist.**
You should discuss the reasons for a genetic test with your doctor before your sample for testing is collected. Your doctor may refer you to a medical geneticist. This is a specialist who is trained to:

- Diagnose genetic conditions.
- Select the most appropriate genetic tests.
- Explain test results to patients and their families.
- Recommend personalized treatment and prevention options.
KINDS OF GENETIC TESTS

We have 20,000-25,000 genes. Genetic tests look for changes in genes. These changes are called variants or mutations. There are tests that look at one gene, many genes, or all of your genes at one time.

This article reviews several genetic tests that examine genes in a person’s DNA for changes that can affect health.

EXOME TESTS

An exome test looks at most of your genes. This test can be useful if testing one gene at a time is not an option. And it may be useful if your doctor is not certain which genes should be tested. If you have this test, you might learn about the cause of your condition. You might also learn that you have other genetic conditions.

For example, your doctor might be looking for the cause of an inherited nerve disorder. But the test could also show that you have an increased risk for cancer or heart disease.

The American College of Medical Genetics and Genomics (ACMG) has a list of genes for inherited conditions that can be treated or prevented. When you have an exome test, you can choose to get results from the ACMG list of genes along with other results that you requested.

GENETIC TESTING FOR ALZHEIMER DISEASE

Some tests are better than others.

One genetic test for Alzheimer disease looks for a variant in the APOE gene. The variant can increase the risk of developing Alzheimer disease, usually after age 60. But the APOE test result cannot predict who will develop Alzheimer. Some people have the variant but never get the disease. Others get the disease but do not have the variant.

There are other genetic tests that are better at finding the cause for inherited Alzheimer disease. These tests might be useful if you or a close relative developed the disease before age 60, or if several relatives have the disease. If the disease occurs at an early age, it is more likely to be genetic. In this case, testing might give you or your family useful information.

Keep in mind that there is no known way to prevent Alzheimer disease. One reason for genetic testing is to learn if you have a genetic risk of the disease. This may help you or your family plan for the future. If you have an increased risk, your children may also.
GENETIC TESTING FOR COMMON MTHFR VARIANTS

These tests won’t help predict a risk for blood clots. Certain MTHFR gene variants can cause high levels of an amino acid called homocysteine. This increases the risk of blood clots. A blood clot can block the blood flow in your veins or arteries. In your lungs this can cause a pulmonary embolism, or in your brain this can cause a stroke.

But this genetic test is not a good way to predict your risk for blood clots. Some people have the gene variant but do not get clots. Other people get clots but do not have the variant. Testing your blood homocysteine level will usually provide better information than genetic testing.

GENETIC TESTING FOR HEREDITARY HEMOCROMATOSIS

Consider this test if you have too much iron in your body or a family history of the condition.

Hereditary hemochromatosis is a common genetic condition. It can cause the body to take in and store too much iron. This can lead to arthritis, heart problems, liver disease, type 2 diabetes, and other problems.

These problems can be prevented if the iron overload is discovered early and treated.

Who should get genetic testing for hereditary hemochromatosis?

• People with a family history of hereditary hemochromatosis. The test could help your doctors know if you are at risk for iron overload.
• People with high iron levels. First, you can get a simple blood test to check your iron levels. If you don’t have a family history, start with this simple test. If your iron levels are high, genetic testing may be helpful.
REPEAT TESTING

Usually you don’t need to repeat a genetic test. Your genetic information generally doesn’t change over your lifetime. There’s usually no reason to repeat a genetic test unless:
• Your doctor thinks the lab made an error.
• A new, more accurate test, is available.

Genetic testing may be more costly than another test that is just as useful. Your insurance may not cover genetic testing. Or your insurance may cover each test only once in your lifetime.

Most labs will send you a copy of your genetic test results, if you ask. If you choose to, you can share them with all of your doctors and family members.

It’s important to seek advice before testing. A medical geneticist can help you make sure that:
• You think about your reasons for wanting the test.
• You get the right tests.
• You and your family are prepared for the results.
• You have a personalized plan for dealing with the results.

If your genetic test results show a risk for a condition, ask your doctor if you should get screened for early signs of the condition.