

## WHEN CONSIDERING GENETIC TESTING, YOU SHOULD CONSIDER THESE QUESTIONS:

- What information do I hope to get and what would it mean at this point in my life?
- Are the conditions being tested for important to me?
- What is my motivation for testing - informational or medical?
- Will the test results offer peace of mind or increase stress and anxiety?
- Will the test results help me make better health choices? Are there actions I can take to improve my health based on test results?
- Does the vendor provide information I can use to help prevent disease after testing? Will I have access to updates on discoveries and new information?
- Does the vendor provide access to an appropriately trained healthcare professional, such as a certified genetic counselor or a clinical geneticist?
- Does the vendor provide information about the test or laboratory methods and how it develops its test results?
- Are the tests conducted in a CLIA-certified laboratory?
- Will my results be kept private?
- Is the test covered by my insurance policy?

# an introduction to informational genetic testing

### ABOUT MEDCO

Medco is a Fortune 50 US company focused on managing and delivering pharmacy care to more than 60 million lives. Medco promotes personalized medicine through clinical research and programs available to its clients and members.

### ABOUT PERSONALIZED MEDICINE COALITION

Representing a broad spectrum of academic, industrial, patient, provider and payer communities, PMC seeks to advance the understanding and adoption of personalized medicine concepts and products for the benefit of patients.

PMC thanks its member organizations for their support, including 23andMe, DeCode Genetics, DNA Direct, and Navigenics.



## Our genes affect our health

The genetic makeup that we inherit from our parents plays a key role in our lives. Over the last few decades, our understanding of how genetics influence our health has increased significantly. Today, we can test for certain genetic variations that can be used to help guide a person's medical care or to determine the risk for developing certain diseases or health conditions. The goal of this guide is to provide basic information about a particular type of testing known as informational genetic testing.

We each inherit a unique combination of genes from our ancestors. These genes, together with many other factors, such as our environment and lifestyle choices, are responsible for making us individuals. In other words, your genes make up your personal genetic "blueprint." Genes can influence how tall you are, your hair color, aspects of your personality, your potential to develop a variety of diseases and how you would react to certain medications.



## Some Common Conditions Included in Informational Tests

## What is genetic testing?

Genetic testing is an examination of certain genes to answer one or many questions. For example, "Did I inherit a tendency to develop a certain disease?" or "Is this drug right for me?" To date, specific genetic variations have been associated with more than 1,500 health conditions. Tests for these variations may be categorized into two broad groups - medical genetic tests and informational genetic tests - depending on the type of information they provide and the way the test result will be used.

**Medical Genetic Tests** are usually ordered by a healthcare provider. The result of a medical genetic test is used to make a treatment decision. For example, medical genetic tests are ordered to diagnose a disease or to assess the likelihood of treatment success or risk with a certain drug. Most medical genetic tests look at a single gene or a few genes.

**Informational Genetic Tests** can be ordered directly by an individual. They are available to gain a better understanding of general health and disease susceptibility. Other types of information may show you have a certain ancestry or other trait



Age-related Macular Degeneration

Alzheimer's Disease

Asthma

Several types of Cancer

Crohn's Disease

Glaucoma

Heart Attacks

Lupus

Multiple Sclerosis

Obesity

Osteoarthritis

Psoriasis

Rheumatoid Arthritis

Type 1 Diabetes

Type 2 Diabetes



such as eye color. The results of an informational test are returned directly to the individual who ordered it. Informational tests analyze multiple genetic markers to estimate a person's risk of developing a common disease. While informational genetic tests may provide results about medical conditions, more research must be done before the results can be used for medical decision making.

## What will an informational genetic test tell me?

Most test results provide only an estimated risk of developing a disease. You will likely find some uncertainty with your results because the impact of genetic variations and the contribution of environmental influences on health are still not fully understood.

For example, you might receive results stating “your risk of developing colon cancer is 3% greater than that of the average person” (general population risk) or that “your risk of developing Alzheimer’s disease is 6% lower than the general population risk.” It is important to keep in mind that these genetic risk assessments are not guarantees that you will or will not develop a condition.

Some information might be especially sensitive and you may wish to consider “opting out” of receiving results for certain diseases. For example, you may choose not to receive test results that are associated with a risk for Alzheimer’s or other diseases for which there are limited or no medical disease prevention interventions currently available.



## What can I do with the results of an informational test?

Informational genetic testing is performed mainly to provide information about an individual's risk of disease. This may encourage people to become more proactive in their healthcare. For example:

- If you have an increased risk of developing type 2 diabetes, you might choose to make changes to your diet and exercise habits and ask your healthcare provider to help you develop a plan to watch for early symptoms of disease.
- If you have an increased risk for developing glaucoma, you might be more conscientious about scheduling an annual eye exam.

If you find that you are at equal or lower risk of developing a specific illness than the general population, it does not mean that you should forgo the usual precautions. Remember that environment and other factors also affect your health. Regular checkups and a healthy lifestyle are very important regardless of your genetic risk factors. Keep in mind that as with any health-related decision, you should work with your healthcare provider to develop a health management strategy that suits your personal needs.

## How can I order a test?

Informational genetic tests are available from a variety of vendors and can often be ordered online or over the phone.

The Clinical Laboratory Improvement Amendments passed by the U.S. Congress in 1988 (CLIA) require inspections of all clinical laboratories to ensure they meet quality standards. Not all genetic testing is conducted in CLIA-certified laboratories, so it is important to know whether the vendor you are considering uses such a laboratory.



It is important to carefully review all of the information available about the test. You should be required to sign a consent form that states you understand the benefits, limitations and potential risks of undergoing genetic testing. If you have any questions, you should not send off your sample until you fully understand the answers.

### What is a “sample” and how do I submit it?

Most genetic tests require a “sample” from a cheek swab or saliva, which is easily shipped to the vendor. Some tests may require a blood sample, which will need to be drawn by a trained healthcare provider.

### Should I speak with a genetic counselor before ordering a test?

Genetic counselors are health professionals specially trained to interpret genetic information and explain its significance to you and your family. They can work with you to understand the risks and benefits of genetic testing, discuss options for genetic testing, interpret test results, and review actions you may want to take based on the test result. If you are uncertain whether or not a genetic test is appropriate for you, seeking the advice of a genetic counselor is recommended.

### What about protecting my privacy?

Your genetic information belongs to you. You should only disclose this information to people who have your best interests in mind. If your healthcare provider recommends a medical genetic test, the results usually become part of your private medical record at that office. You can ask your healthcare provider about the federal, state, and local policies that are in place to protect the privacy of your medical record and genetic testing results.



If you decide to pursue informational genetic testing, it is important to carefully review the vendor’s privacy policy. Typically, the policy will state that the vendor will not share any of your personal data or results with any third parties unless you specifically allow it. Third parties may include insurance companies, health management organizations, hospitals, doctors, businesses and government agencies, among others.

You may decide to share your informational genetic test results with your healthcare providers. Any informational genetic test result or other information you share with your healthcare provider may become part of your medical record.

Informational genetic testing is a new field and regulations are evolving. You should make sure the genetic testing company complies with the existing federal and state requirements, and that it adheres to professional best practices (see questions on the back cover). Currently, results of genetic tests that take place outside of regular medical practice may not be protected by federal privacy protections for medical information, like the Health Insurance Portability and Accountability Act of 1996 (HIPAA).

The Genetic Information Nondiscrimination Act of 2008 (GINA) protects consumers from discrimination by health insurers and employers on the basis of genetic information.

### Will my insurance cover genetic testing?

In some cases, insurance will cover the cost of genetic tests, especially medical genetic tests that are ordered by your healthcare provider. However, most informational genetic tests are not covered by insurance. If you are interested in submitting the costs of your genetic test to your insurance provider, you should contact your provider beforehand to ask about coverage.