

# Questions and answers for patients

## Why should I have a genetic test if I have been diagnosed with, or am suspected of having, a heritable condition?

Heritable conditions are caused by mutations, or changes in the DNA of certain genes in your body. Genetic testing is a diagnostic tool that can help identify the disease-causing change in your DNA, and is becoming a standard diagnostic procedure in several fields of medicine. The reasons for choosing genetic testing have become more important in individual health management over the years, and several international medical organizations now recommend it. Here are some reasons to choose genetic testing:

- **Confirmation of diagnosis**

Genetic conditions often have observable traits – changes to your body that may cause symptoms. Many medical conditions may share identical or similar traits/symptoms. In these cases, a genetic diagnosis can help confirm if a condition is caused by a specific gene change. It could even alter your current clinical diagnosis. An accurate diagnosis with the help of genetic testing allows your physician to find the best care and management plan for you.

- **Testing other family members at risk**

If a condition has a genetic cause, there is a higher chance of passing on the genetic change to your children. Your siblings and parents could also have the same change and be affected by the condition. Knowing that the cause of your condition is genetic allows your physician to identify risks to other family members, and provides a solid basis for clinical follow-up and preventative actions for affected family members. Genetic diagnosis may also help increase the lifespan and improve quality of life for other family members who were unaware they were at risk for a genetic condition.

- **Personalized treatment and management**

With a genetic diagnosis, your physician can identify and evaluate your risks. It can help in fine-tuning your treatment and follow-up care.

- A genetic diagnosis means accurate genetic counseling for you and allows your physician to estimate recurrence in your family.
- Genetic testing can also tell you that you do not have the genetic change/ mutation causing the condition in your family. This information may determine that no further clinical follow up is needed for you

## What will my genetic test results tell me?

We provide your physician with a clinical statement from the genetic test, complete with all the relevant findings, and evidence from medical publications and literature used in the interpretation of the test results. We also give your physician recommendations for further care, such as genetic counseling and family member testing. Your test results will reveal one of three possible outcomes:

1. **Positive result**

If you have a positive result, it means that a disease-causing genetic change has been identified that can explain your symptoms. If you do not currently have symptoms, it can tell you that you have an increased chance of developing symptoms.

2. **Negative result**

If you have a negative result, it means you do not have a genetic change that can be identified. This may mean that you do not need to worry about developing symptoms associated with the condition you are being tested for. However, if you do have symptoms, and your result is negative, it could mean that today's scientific methods cannot identify the cause or that a different genetic test may be more suitable for you. This should be discussed with your ordering physician and/or genetic counselor.

3. **Variant of Unknown Significance (VUS)**

If you have a variant of unknown significance, it means that there is not enough collected evidence in the scientific field of medicine that can tell us if your genetic change is definitely disease-causing or not. Future research and more experience with patients who have the same genetic findings may alter the classification of the identified VUS.

## Should I have genetic counseling before the test?

We recommend that you receive genetic counseling alongside genetic testing. You should attend counseling both before the test and after the results are available.

## How long will it take to get my results?

Our standard diagnostics procedure will take about 21 days from the day we receive your test sample. For an additional charge, your physician can get results within 7-10 days through our Express Service.

## How will I receive the test results?

We will send your results directly to your physician or other ordering health care professional. They will then share them with you and discuss how they affect your future health care, and if the results have significance for your family.

## Genetic knowledge gives you answers

Blueprint Genetics offers genetic testing for a wide variety of heritable conditions. We can provide you with the answers you and your physician are searching for to help make decisions about your health and future. Our genetic testing services are among the most accurate, scientifically advanced, and highest quality available. We also have the fastest results delivery time – your physician will get your genetic test results within 21 days, or just 7–10 days through our Express Service.

Our clinical genetic tests must be ordered through a certified health care professional. If you are interested in our services, please consult your physician, or get in touch with our customer support team for more information.



## Genetic testing can provide you with the answers you and your physician are searching for to help make decisions about your health and future.

Find out how genetic diagnostics from Blueprint Genetics can help you and your family. Or contact our customer support with any questions about our services or billing inquiries.

Learn more about us and find more resources at:  
[www.blueprintgenetics.com](http://www.blueprintgenetics.com)

We're here to help!

Guide

## A patient's guide to understanding genetic testing

