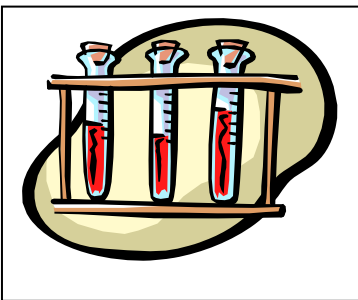


Remember...

This brochure has some general information about why your doctor might choose to offer you genetic testing. Although we use 'doctor' here, doctors, nurse practitioners, counselors, or other providers might discuss genetic testing with you.

Your case may not fit exactly into one of the groups mentioned here. Your doctor can explain the reason for genetic testing in your situation. He or she can also talk with you about the pros and cons of genetic testing and other options for you.



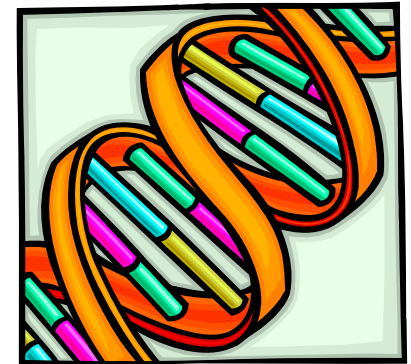
DNA Diagnostic Lab

CMSC 1004
Johns Hopkins Hospital
600 N. Wolfe St.
Baltimore, MD 21287

www.hopkinsmedicine.org/dnadiagnostic

The DNA Diagnostic Lab at Johns Hopkins

Reasons for Genetic Testing



**Quality, Service,
Excellence**

Some Reasons Your Doctor Might Recommend Genetic Testing

Confirmatory / Diagnostic Testing

You doctor may recommend confirmatory or diagnostic testing if he or she is mostly sure you have a genetic condition. Knowing your specific gene change may help you learn more about the symptoms you are at risk for or plan extra tests to monitor your condition. Sometimes you are tested to find out what gene change you have so your relatives (brothers and sisters or children) can have genetic testing.

'Rule Out' Testing

Sometimes your doctor wants to order a genetic test to make sure you *do not* have a genetic condition. You may have one or two symptoms that may or may not be part of the genetic condition. Ruling it out lets your doctor know that you don't have to worry about the extra symptoms that can come with that condition. Usually, genetic tests are good 'rule out' tests if they have a low chance of missing the condition in someone who has it (low false negative rate).

Pre-symptomatic Testing

Your doctor might suggest a pre-symptomatic test if you are healthy but have a family history of a genetic condition that you might develop in the future. If you know the specific gene change that causes the condition in your family, you can have a test that focuses on that change. Sometimes you don't know the specific gene change that causes the disease in your relatives, and they are unable or unwilling to have the test. Your doctor may be able to test you without first testing a relative with the condition.



Carrier Testing

Some genetic conditions require two gene changes (one passed down from each side of the family). A carrier is someone who only has one gene change, not two, and who is healthy. Your doctor might recommend carrier

Carrier Testing (continued)

testing for a condition if it is more common in your ethnic group, if you have other lab work that suggests you are a carrier, or if you have a family history of that condition. Carrier testing may look for the gene changes in your family or a set of common gene changes that cause the condition.

Questions for Your Doctor

Why do I need genetic testing?

What are the pros and cons of testing?

Are there any other options?

How long will results take?

How will I learn my results?

What happens after testing?

How will billing work?

Can you recommend some helpful resources for me?