

Remember...

This brochure has some general information about gene changes (mutations) that may or may not cause disease. You might not fall exactly into one of the groups covered in this brochure.

When we write your result, we might be able to use more information that is specific to you. This might be your family history, your symptoms, or the results of other lab tests.

Be sure to talk about your result with your doctor, nurse, or counselor.



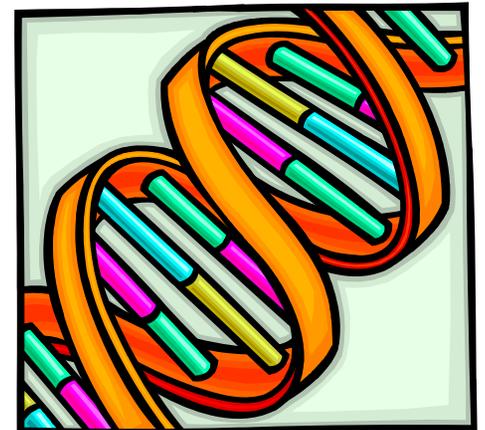
DNA Diagnostic Lab

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The DNA Diagnostic Lab at Johns Hopkins

Understanding your Gene Sequencing Test Result



**Quality, Service,
Excellence**

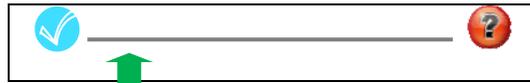
Understanding the Results of Gene Sequence Tests

If a gene is a paragraph in a book, gene sequencing is a test that looks at every single word and letter in that paragraph to find errors that might cause a genetic disease. Sometimes we find clear spelling mistakes, and we are certain that they change the meaning of the paragraph (cause a genetic disease). Sometimes we find words that are spelled in a different way, and we are not sure if they change the meaning of the paragraph or not.

When we write our reports, we gather all the evidence we can find. We make our best judgment about whether that gene change causes disease. We may recommend other tests that might help increase our certainty.

This brochure walks you through the different certainty levels we might report in a gene sequence test. The picture under each group will give you an idea of how certain we are that the type of change we found causes disease. If the arrow is closer to the check mark, we are more certain. If the arrow is closer to the question mark, we are less certain. Your doctor can check off the group that best describes your result and review your report with you.

Disease Causing



We call these changes in DNA 'disease-associated mutations' in the report. Other patients with the disease have these changes. Scientists or doctors have reported these changes in scientific papers. Sometimes many patients have a certain change. Sometimes only one or two patients have it. We usually recommend genetic counseling to help you understand what results mean for diagnosing or managing disease and for your relatives' risks.

Likely Disease Causing



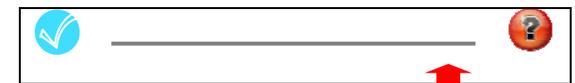
These are DNA changes that are not reported in scientific papers in any patients. However, after looking at the effect they have on the gene (or the protein it makes), we are very certain that they are harmful changes. If you have a clinical diagnosis of a disease, this is very likely to be the gene change that causes disease. We will recommend genetic counseling to help you understand what results mean for diagnosing or managing disease and for your relatives' risks.

Possibly Disease Causing



In researching these DNA changes, we only found *some* evidence that they cause disease. Sometimes, the evidence wasn't very strong. Sometimes we also found some evidence that they may be harmless. We list the evidence in your report for your doctor to review. We may recommend that family members have testing to help us figure out if your gene change is causing your disease. Your doctor may call us in the future to see if we have learned more.

Variant of Uncertain Clinical Significance



We call these changes sequence variations because we do not know if they cause disease or not. After extensive research, we are not able to find any evidence that they are harmful. But we are not able to find any evidence that they are harmless, either. We may recommend that family members have testing to help us figure out if your gene change is causing disease. Your doctor may call us in the future to see if we have learned more.