You have an increased risk for developing symptoms of G6PD deficiency based on your genetic result.

We detected two copies of the c.512+1G>A variant in the G6PD gene.

You are at higher risk for developing symptoms of G6PD deficiency than your unaffected family members.

Some symptoms may occur without exposure to certain triggers. Symptomatic attacks are typically self-limiting and may require treatment with antioxidants and supportive care.

It is important to discuss this result with a healthcare professional.

For more information about G6PD deficiency, see the National Heart, Lung, and Blood Institute (NIH).

For people with G6PD deficiency, symptoms can be triggered by many factors.

Learn more about G6PD deficiency at the National Heart, Lung, and Blood Institute (NIH).

If you have questions about your results on this page, please contact us.
OSFD deficiency is linked to variants in the OSFD1 gene.

You have two copies of the genetic variant we tested.

<table>
<thead>
<tr>
<th>Variant Details</th>
<th>View of Needlebugs</th>
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<tbody>
<tr>
<td><strong>Variant</strong></td>
<td><strong>OSFD1</strong></td>
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<tr>
<td><strong>Heterozygous</strong></td>
<td>F</td>
</tr>
<tr>
<td><strong>Risk</strong></td>
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<tr>
<td><strong>Additional information</strong></td>
<td>- Biological evaluation of genetic susceptibility to needle bugs</td>
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<td><strong>Reference</strong></td>
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</table>

**Test Interpretation**

This report provides information about the genetic condition with respect to needle bugs.

**Other Factors**

OSFD deficiency, a genetic condition, may not fully account for the observed symptoms, which may be influenced by other factors.

**Test Details**

**Indications for use**

For Ludwig N. & Partners’ Needlebug Research OSFD1 deficiency, 20% expressing the risk of needle bugs. The test results support the need for additional genetic evaluation to fully understand the complex relationship between OSFD1 deficiency and needle bugs.

**Test Procedure**

The test involves a blood sample collected from the participant. The sample is then analyzed using advanced genetic technology to determine the presence of OSFD1 genetic variants.

**Test Performance Summary**

The test is highly sensitive and specific, with a very low rate of false positives and negatives. It is designed to provide accurate results within a short timeframe.

**References**


**Change Log**

No recent changes recorded.

Date: June 2019
Frequently Asked Questions

GFRP Deficiency

What does this test do?

The report states the variants included in this test; we have listed studies in patients of African descent. What if I'm of African descent or am only partly of African descent?

The report says my result is based on the one established in my account settings. What does that mean?

Can I learn more about GFRP deficiency, support groups, and other resources?

My report says two copies of the WDR45/16 variant were detected. Does this affect me?

This means you have two copies of the genetic variant we tested.

Why do I need only one variant to have an increased risk for developing symptoms of GFRP deficiency, whereas someone needs two copies or two copies of a variant?

The variant we tested in the GFRP gene, and it is located on the 5 chromosome. This means that it is inherited differently than many genes, in a manner called “recessive” inheritance. Learn more about the 5-first two items at the bottom.

What is increased risk mean?

The GFRP assay is important for preventing retinal bleeds against damage. Certain variants in the GFRP gene, including the one in the test report, make the GFRP enzyme less active. This results in reduced levels of the GFRP enzyme, which can lead to a buildup of compounds that cause glaucoma.

If you report shows you have an increased risk, it means that your chances of developing symptoms of GFRP deficiency are increased based on your genetic result for the test. About 5% of females with this genetic result have GFRP deficiency, which means that one in 20 females has reduced activity of the GFRP enzyme.

How do I know if I have GFRP deficiency?

This genetic result is associated with an increased risk for developing symptoms of GFRP deficiency. Consultation setting up an appointment with a healthcare professional to discuss these results.

How can I find out my results affect my family?

Do you share DNA with your family members, they may also be interested in your result. If you’re thinking about sharing it, talk with your family about your results, see this section for a discussion of how to consider before sharing the information.

Because you have two copies of the variant we tested, it is expected that:

• Both of your children will inherit the variant. This means your male children are at risk for developing symptoms of GFRP deficiency.

• Both of your parents have the variant.

• Both of your female siblings have at least one copy of the variant.

• Each of your male siblings has at least a 50% chance of having the variant.

Because of the risk variant is inherited, your female children are at risk for two copies of GFRP deficiency, unless they do not inherit a variant from their mother. Learn more about this variant in our Healthline Knowledge Base.

I have more questions. Check out our Customer Care Center.