G6PD Deficiency

G6PD deficiency is a genetic disorder that affects the production of a protein called glucose-6-phosphate dehydrogenase (G6PD). This enzyme is important for the production of red blood cells. People with G6PD deficiency may have a higher risk of developing certain health conditions, including hemolytic anemia, which can cause the breakdown of red blood cells.

How to Use This Test

Physical and Medical Information

Please read the physical information that follows to understand what this test is and how it may affect you.

1 variant detected

You have one of the two genetic variants we tested linked to G6PD deficiency.

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

Consider sharing this result with a healthcare professional.

About G6PD Deficiency

Alleviation of G6PD Deficiency Symptoms, G6PD Deficiency

You may want to discuss your options with a healthcare professional to help manage any symptoms you may be experiencing.

Consider sharing this result with a healthcare professional.

If you have a family history of G6PD deficiency, your healthcare provider may recommend testing.

If you or your family have questions about this test, please contact your healthcare provider.
G4PD Deficiency

G4PD deficiency is a common hereditary disorder that can lead to hemolytic anemia. It results from deficiencies of G4PD, an enzyme that catalyzes the production of NADPH, a coenzyme that is essential for maintaining red blood cell membrane stability.

Signs and Symptoms

- Hemolytic anemia
- Fatigue
- Pallor
- Jaundice
- Pale nails
- Cyanosis

You have one of the genetic variants tested.

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<th>Gene</th>
<th>Name</th>
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<th>Other</th>
<th>Conditions</th>
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<tbody>
<tr>
<td>NADH</td>
<td>1.10.1.1</td>
<td>E</td>
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<td>T</td>
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<tr>
<td>GTPS1</td>
<td>GTPase</td>
<td>E</td>
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<td>GTPase deficiency</td>
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Test Interpretation

The test results indicate that you have G4PD deficiency with a single variant in the GTPS1 gene. This variant is associated with increased risk for hemolytic anemia. However, the severity of the condition may vary depending on additional genetic factors and environmental factors.

Other Traits

- G4PD deficiency is a hereditary condition that is passed down through the family. Inheritance patterns include autosomal recessive or autosomal dominant.
- Individuals with G4PD deficiency may require blood transfusions or splenectomy (removal of the spleen) to manage hemolytic anemia.
- G4PD deficiency can cause complications such as liver disease and gallstones.

Test Details

- Indicators for Use: Hemolytic anemia management, G4PD deficiency diagnosis
- Test Type: Molecular genetic test
- Test Method: Next-generation sequencing
- Test Panel: GTPS1 gene
- Test Sensitivity: 100%
- Test Specificity: 100%
- Test Accuracy: 100%
- Test Duration: 14 days
- Test Cost: $500
- Reporting: Online
- Additional Information: The test results are available online.

Warnings and Limitations

- Always consult with a healthcare professional before making any medical decisions.
- The test results should not be used as a sole basis for diagnosing or treating medical conditions.
- The test results may not be applicable to all individuals.

References

G6PD Deficiency

What does this text say?  

The report says my genetic result is based on the sex I indicated in my account settings. What does that mean?  

Where can I learn more about G6PD deficiency, support groups, and other resources?  

My report says one variant was detected. What does this mean?  

This means you have one of the two genetic variants we tested. Most females with just one copy of a variant are not expected to develop symptoms of G6PD deficiency. However, certain factors can affect the chances of developing symptoms in females with your genetic result, including the percentage of G6PD-deficient red blood cells in the body, which varies from female to female and can change over time. In addition, it is possible to have another genetic variant not included in this test.

Is this answer helpful? Yes No

Why do males only need one variant to have an increased risk of developing symptoms of G6PD deficiency, whereas most females need two variants or two copies of a variant?  

The variants we tested are in the G6PD gene, which is located on the X chromosome. This means the G6PD gene is inherited differently than most genes, in a manner called X-linked recessive inheritance.

Is this answer helpful? Yes No

My report says one variant was detected. What are some things I could do?  

Your genetic result means you have one of the two genetic variants we tested. Most females need to have two copies of a variant, or two different variants, to develop symptoms of G6PD deficiency. However, it is still possible to have another genetic variant not included in this test.

Consider talking to a healthcare professional if:
- You have a family history of G6PD deficiency or hemolytic anemia.
- You think you might have symptoms of G6PD deficiency.
- You have concerns about your results.

Is this answer helpful? Yes No

How could my result affect my family?  

Since you share G6PD with your family members, they may also be interested in your result. If you are thinking about talking to family members about your results, see this article for a discussion of things to consider before having the conversation.

Because you have one copy of a variant we tested, it is expected that:
- Each of your children has a 50% chance of inheriting this variant from you. Any male children who inherit this variant are at risk of developing symptoms of G6PD deficiency.
- At least one of your parents has the variant.
- Your siblings may also have this variant.

In most cases, female children who inherit this variant from you are not expected to develop symptoms of G6PD deficiency unless they also inherit a variant from your partner. This is because of the way this variant is inherited, called X-linked recessive inheritance.

Is this answer helpful? Yes No

Have more questions? Check out our Customer Care Help Center.