G6PD Deficiency

G6PD deficiency is a common genetic condition caused by a deficiency or absence of a specific enzyme in the body. The enzyme is responsible for producing energy for cells in the liver, red blood cells, and other organs when oxygen levels are low. The condition can cause severe anemia in affected individuals.

You have the genetic variant tested.

You have an increased risk for developing symptoms of G6PD deficiency. The condition can be triggered by a variety of factors, including:

- Certain medications
- Infections
- Exposure to certain drugs
- Alcohol

Your risk for developing symptoms of G6PD deficiency depends on many factors. Symptoms can range from mild to severe, and may include:

- Anemia
- Jaundice
- Fatigue
- Headache
- Abdominal pain

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

About G6PD Deficiency

Anemia: Commonly caused by low levels of the enzyme G6PD, resulting in reduced energy production in affected individuals.

Your physician may recommend:

- Nutritional supplements
- Medications to manage symptoms

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

Learn more

See the Frequently Asked Questions for more information.
**Scientific Details**

G6PD deficiency is a common genetic condition that occurs in up to 4% of the world’s population, mainly in African, Mediterranean, and Asian populations. The deficiency is caused by a mutation in the G6PD gene, which encodes the enzyme glucose-6-phosphate dehydrogenase. G6PD is involved in the shunt pathway of glucose metabolism and plays a critical role in maintaining the red blood cell’s integrity by converting oxidized glutathione to reduced glutathione. The deficiency can lead to hemolytic anemia and other health issues in the presence of certain medications or infections.

You have the genetic variant we tested.

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<th>Variant Detailed</th>
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<tr>
<td><strong>Variant Name</strong></td>
<td><em>IVS-1-3G&gt; A</em></td>
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**Variant Summary**

- **Gene:** G6PD
- **Location:** 21q22.3
- **Functional Impact:** Missense
- **Clinical Significance:** Moderate

**Test Interpretation**

- **Prognosis:** The G6PD deficiency is mild in most patients, and it can be asymptomatic in some cases. However, it can lead to hemolytic anemia in certain situations, such as after the use of certain medications or infections.
- **Management:** Manage symptoms and avoid triggers. Consult with a healthcare provider for a personalized management plan.

**Other Factors**

- **Environmental:** Avoid triggers like certain medications or infections. Consult with a healthcare provider for a personalized management plan.

**Test Details**

**Indications for Use**

- **Screening:** To identify individuals at risk of G6PD deficiency.
- **Diagnosis:** To confirm the presence of G6PD deficiency in affected individuals.

**Testing Methodology**

- **Next Generation Sequencing (NGS):** High throughput sequencing of the G6PD gene to identify genetic variants.

**Test Performance Summary**

- **Analytic Sensitivity:** 99.99%
- **Analytic Specificity:** 99.99%
- **Interpretive Accuracy:** 99.99%

**References**

GALD Deficiency
Frequently Asked Questions

What does this test do?
This test does the following:

- Measures the level of Galactose-1-phosphate uridylyltransferase (GALT), an enzyme that breaks down galactose.
- Determines if the patient has a deficiency in GALT, which can lead to symptoms of galactosemia.
- Helps assess the risk of developing symptoms of galactosemia.

What does this test cost?
This test costs $100.

The report says the variant included in this test is best studied in people of African descent. What does this mean?
This means that the variant is more common in people of African descent. People of other ethnicities may have different results.

The report says my genetic result is based on the sex assigned at my recent settings. What does this mean?
This means that the result is based on the sex assigned at birth, which may not accurately reflect your sex.

Where can I learn more about GALD deficiency, support groups, and other resources?
You can find more information on the Galactosemia Foundation website.

My report says I am carriers for galactosemia. What does this mean?
This means that you have a genetic variant that can lead to symptoms of galactosemia.

This means you have a genetic variant that is tested.

Note: This test is not developing symptoms of GALD. However, this does not mean you have been developing or will develop symptoms of GALD deficiency. Exposure to a known environmental factor is usually required to trigger symptoms. People with GALD deficiency have no symptoms of GALA deficiency.

Why does one variant need to have an increased risk for developing symptoms of GALD deficiency, whereas another needs no test at all (type of a condition)?
This is because GALD deficiency can be triggered by different factors in different people. The test is designed to identify the most common risk factors for GALD deficiency.

The report says I am carriers for galactosemia. This means that I am a heterozygote or a carrier for the condition. As a carrier, I have one copy of the gene that causes galactosemia, but I do not have symptoms of the condition.

What does increased risk mean?
Increased risk means that you have a variant that is associated with a higher likelihood of developing symptoms of galactosemia. However, it does not mean that you will definitely develop symptoms.

The report says increased risk was detected. What are some things I could do?
If you have an increased risk, you may be advised to undergo testing to identify any potential risk factors for developing symptoms of galactosemia.

How could my result affect my family?
If you have an increased risk, it may be advisable to consult with a healthcare professional to discuss your options.

More questions?
Contact us for more information on how to request a consultation.