**GPD Deficiency**

GPD deficiency is a rare genetic disease that occurs when the body cannot properly convert the nutrient galactose into glucose. It is caused by a genetic mutation that affects the production of the enzyme GPD1. In some cases, GPD deficiency can be severe and lead to intellectual disability, developmental delays, and other health problems. In other cases, it may be relatively mild and go unnoticed.

**How To Use This Test**

The test involves two components: 1) GPD1 activity measurement and 2) sequencing of the GPD1 gene. The results are interpreted by a healthcare professional.

- **GPD1 Activity Measurement:** This test measures the activity of GPD1 in a blood sample. It can help determine if GPD deficiency is present.
- **GPD1 Gene Sequencing:** This test looks for mutations in the GPD1 gene. It can help identify genetic causes of GPD deficiency.

**Important Information**

- The test is available for infants and children. It is not suitable for adults.
- The test results are reported to healthcare providers.
- The test is performed by a healthcare professional.

**You have an increased risk of developing symptoms of GPD deficiency based on your genotype:**

For treatment, discuss this result with a healthcare professional.

**For people with GPD deficiency, symptoms can be triggered by certain factors:**

- **Systemic infections:** Infections can weaken the immune system, making it more difficult to fight off infections.
- **Growth and development:** GPD deficiency can affect growth and development, leading to smaller body size and delayed development.

**About GPD Deficiency**

GPD deficiency is a genetic disorder that causes the body to have difficulty breaking down certain sugars. This can lead to symptoms such as developmental delays, intellectual disability, and physical issues. It is usually diagnosed in infancy, and treatment involves managing symptoms and providing support.

**For more information, contact your healthcare provider or visit:**

- **Healthline:** [www.healthline.com](http://www.healthline.com)
- **National Institute of Health:** [www.nih.gov](http://www.nih.gov)
G6PD Deficiency

G6PD deficiency is a hereditary blood disorder that affects the production of an enzyme called glucose-6-phosphate dehydrogenase. Individuals with G6PD deficiency have reduced or absent levels of this enzyme, which can lead to a variety of symptoms, including anemia, jaundice, and hemolytic anemia. The deficiency is inherited in an autosomal recessive pattern, meaning that an individual must inherit two copies of the mutated gene (one from each parent) to have the disorder.

Diagnostic Tests

Several tests are available to diagnose G6PD deficiency, including:

1. Hematologic Tests: These tests measure the number of red blood cells and other blood components. In individuals with G6PD deficiency, the red blood cell count may be low due to hemolysis (destruction of red blood cells).

2. Enzyme Assays: These tests measure the activity of glucose-6-phosphate dehydrogenase in red blood cells. Low enzyme activity indicates G6PD deficiency.

3. Storage Experiments: This test involves incubating red blood cells from an individual with G6PD deficiency in a solution containing oxidants. If the enzyme is deficient, the red blood cells will hemolyze (break down) in response to the oxidants.

4. Genetic Testing: This test is available for individuals suspected of having G6PD deficiency. It can confirm the presence of the G6PD gene mutation responsible for the disorder.

Other Factors

G6PD deficiency can be triggered by certain factors, including:

- Drugs: Certain medications, such as antibiotics, can trigger hemolysis in individuals with G6PD deficiency.
- Viruses: Infections, particularly those caused by certain viruses like hepatitis A and B, can cause hemolysis in those with G6PD deficiency.
- Alcohol: Excessive alcohol consumption can also trigger hemolysis in those with G6PD deficiency.

Test Details

- **Indications for Use:**
  - Individuals with a family history of G6PD deficiency.
  - Those with symptoms suggestive of hemolytic anemia.

- **Test Performance Summary:**
  - Sensitivity: 98%
  - Specificity: 99%
  - Positive Predictive Value: 98%
  - Negative Predictive Value: 99%

- **Warnings and Limitations:**
  - The test may not be accurate in certain conditions, such as during acute infections or during pregnancy.
  - Interpretation of results should be done by a qualified healthcare provider.

References

G6PD Deficiency

G6PD deficiency is a common genetic condition caused by defects in an enzyme called glucose-6-phosphate dehydrogenase, or G6PD. The G6PD enzyme helps protect red blood cells from damage. In people with G6PD deficiency, red blood cells are destroyed upon exposure to certain environmental triggers, which can lead to episodes of anemia. This test includes two common variants linked to G6PD deficiency.

Overview | Scientific Details | Frequently Asked Questions

What does this test do?

The report says the variants included in this test are most common and least studied in certain ethnicities. What does this mean?

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.

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 variant we tested for is 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. Learn more about X-linked recessive inheritance.

What does increased risk mean?

The G6PD enzyme is important for protecting red blood cells against damage. Certain variants in the G6PD gene can make the G6PD enzyme less effective at protecting these cells, which can lead to their destruction in response to certain medications, infections, and foods. When red blood cells are destroyed, people with G6PD deficiency may experience symptoms of anemia, including dark urine, pale skin, a rapid heartbeat, and jaundice.

If your report says you have an "increased risk," it means that your chances of developing anemia due to G6PD deficiency are increased based on your genetic result for the test.

However, many people with G6PD deficiency do not develop symptoms. Exposure to certain environmental triggers - such as certain medications, infections, and foods - is usually required to develop symptoms. Learn more about factors that can trigger symptoms of G6PD deficiency.

The exact percentage of people with your genetic result who develop symptoms of G6PD deficiency is not currently known.

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

How could my result affect my family?

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

Because you have one variant, it is expected that:

• Each of your female children will inherit this variant from you. In most cases, they are not expected to develop symptoms of G6PD deficiency unless they also inherit a variant from your partner.

• Your mother has this variant.

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

Because of the way this variant is inherited, your male children are not expected to inherit this variant from you. Learn more about how this variant is inherited, called X-linked recessive inheritance.

Is this answer helpful? Yes  No

Is this answer helpful? Yes  No

Is this answer helpful? Yes  No

Is this answer helpful? Yes  No

Is this answer helpful? Yes  No

Is this answer helpful? Yes  No

Is this answer helpful? Yes  No

Is this answer helpful? Yes  No

Is this answer helpful? Yes  No

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