G4PD Deficiency

G4PD deficiency is a genetic condition characterized by a hereditary enzyme deficiency. This enzyme, encoded by a gene on chromosome 4, plays a critical role in the breakdown of vitamin B6, a nutrient essential for the production of red blood cells. Individuals with G4PD deficiency may experience a variety of symptoms, including fatigue, muscle weakness, and anemia.

You may be at risk of developing symptoms of G4PD deficiency if your gene test results indicate a variant in the G4PD gene. This rare condition is often overlooked, and symptoms may not appear until later in life.

For people with G4PD deficiency, symptoms can be triggered by various factors, including

- Certain medications or supplements
- Infections
- Stress or illness
- Blood transfusions
- Certain foods or beverages

Important: If you have symptoms of G4PD deficiency, consult a healthcare professional for proper diagnosis and management.

Consider sharing your results with a healthcare professional to discuss any potential treatments or lifestyle changes.

Learn more at: [Gene by Gene](https://www.genebygene.com) or [Authority4D](https://www.authority4d.com)
**Scientific Details**

G6PD deficiency is a common genetic disorder that can be caused by a small number of point mutations in the G6PD gene. This disorder leads to a deficiency in the enzyme glucose-6-phosphate dehydrogenase (G6PD), which is important for the production of red blood cells. As a result, individuals with G6PD deficiency may experience hemolytic anemia, a condition in which red blood cells break down prematurely.

**You have one copy of the genetic variant we tested.**

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<th>Gene</th>
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**Test Interpretation**

- **Medical significance:**
  - Familiar with the most common genetic disorder involving a deficiency in G6PD, this is important for the production of red blood cells. As a result, individuals with G6PD deficiency may experience hemolytic anemia, a condition in which red blood cells break down prematurely.

**Other Factors**

- **Medical history:**
  - Family members who may have a history of hemolytic anemia or other blood disorders.

**Test Details**

- **Indications for test:**
  - To understand the risk of hemolytic anemia in individuals with G6PD deficiency.

- **Test Performance Summary:**
  - This test is highly specific and can be used to identify individuals with G6PD deficiency.

**References**

Frequently Asked Questions

**G6PD Deficiency**

**What does this text do?**

- The report says the variant included in this test is best studied in people of African descent. If you’re of African descent, or you have any known ancestry from Africa, this variant may be more relevant.
- Where can I learn more about G6PD deficiency, report groups, and other resources?
- My sample was one copy of the Widal test result was detected. What does this mean?

This means you have one copy of the variant, but it is not detected in the Widal test. The variant is inherited as an autosomal recessive trait. Most people need two copies of the variant to develop symptoms of G6PD deficiency. It is possible to have another rare genetic variant not included in this test.

**Why did my report list one variant to have an increased risk for developing symptoms of G6PD deficiency, whereas another report lists the variant as not detected at all?**

The variant we tested for is in the G6PD gene, and is located on the X chromosome. This means that it is inherited differently than most genes, as a carrier can have a risk of inheriting the trait. More about X-linked recessive inheritance.

**What does not hold at risk for developing symptoms of G6PD deficiency mean?**

The G6PD enzyme is important in protecting red blood cells against damage. Certain variants in the G6PD gene, including the Widal test result in this report, make the G6PD enzyme less stable. This results in reduced levels of the G6PD enzyme, which can lead to the degradation of red blood cells 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, fatigue, and jaundice. Some women with one copy of the Widal test result in this report may have slightly reduced levels of the G6PD enzyme in their red blood cells. However, research shows that women with one copy of this variant are not at an increased risk for developing anemia related to G6PD deficiency.

**My report was one copy of a variant was detected. What does this mean?**

Most women with this genetic variant do not have an increased risk for developing symptoms of G6PD deficiency. However, it is possible to have another variant not included in this test.

Consider telling a health-care 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.

**How could my result affect my family?**

Since you shared your test with your family, we may also be interested in your result. If you are thinking about telling family members about your results, read this article for a discussion of things to consider before having the conversation.

Because you have one copy of the 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 may be at risk for developing symptoms of G6PD deficiency.
- At least one of your parents has this variant.
- Your siblings may also have this variant.

Because other ways this variant is inherited, any female children who inherit this variant from you are unlikely to have symptoms of G6PD deficiency unless they also inherit a variant from your partner. Learn more about how this variant is inherited, and a linked presence calculator.

**More questions? Check out our Customer Care Help Center.**