G6PD Deficiency

G6PD deficiency is an enzyme disorder that prevents blood from converting glucose to energy properly. It can lead to a life-threatening condition called hemolytic anemia, where red blood cells break down too quickly. This is especially dangerous for men and boys, as it can cause severe symptoms and requires medical attention.

How To Use This Test

This test does not diagnose G6PD deficiency or any other health condition. It is meant to be a tool that provides you with information about your condition, if you have any questions about your health.

See Frequently Asked Questions

4 Intended Uses:

You can use this test to:

- Assess your risk for developing hemolytic anemia.
- Monitor your condition.

5 Limitations:

- This test does not diagnose other conditions.
- It is not intended for use by non-professionals.

Important Considerations:

- G6PD deficiency is a rare condition that affects males and females. It is more common in people of African, Mediterranean, and South Asian descent.
- If you have a family history of the condition, you may be at increased risk.

There is a chance of having another variant linked to G6PD deficiency. However, people with variants linked to G6PD deficiency are not at increased risk for developing the condition if they are not at increased risk for the variant.

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

- Certain medications
- Certain infections
- Certain foods

If you have any of the above factors, consult your healthcare provider.

About G6PD deficiency

G6PD deficiency is a rare inherited disorder that affects males and females of all ethnic backgrounds. It is caused by a deficiency in the enzyme glucose-6-phosphate dehydrogenase (G6PD), which leads to an increased risk of hemolytic anemia.

Who is at risk for G6PD deficiency?

- Males are more likely to have the condition.
- Females with a family history of the condition are also at risk.

How does G6PD deficiency affect the body?

- It affects the production of red blood cells.
- Red blood cells that are affected by the condition may break down too quickly.

Important Considerations:

- G6PD deficiency is a rare condition that affects males and females. It is more common in people of African, Mediterranean, and South Asian descent.
- If you have a family history of the condition, you may be at increased risk.

Learn more about G6PD deficiency.

See Frequently Asked Questions for more information.

You do not have the two variants we tested linked to G6PD deficiency.

You could still have variants not included in this test.

You do not have the two variants we tested. These variants are not currently found in people of African, Mediterranean, or South Asian descent. For more information, consult your healthcare provider.

Cabinet medications

- Certain medications can trigger G6PD deficiency, including some antibiotics, aspirin, and certain pain medications.

Cabinet foods

- Certain foods can also trigger G6PD deficiency, including certain fruits and vegetables.

If you have any of the above factors, consult your healthcare provider.
**GeneDx Deficiency**

GFAP deficiency occurs in approximately 10% of patients with unexplained growth failure or mental deficiency. This is a rare, progressive, multifaceted disorder that can have significant developmental consequences. It is characterized by reduced growth, intellectual disability, and seizures. The disorder is caused by mutations in the GFAP gene, which encodes a protein that plays a critical role in the structural integrity of neurons.

**Genetic Causes**

- **GFAP Gene Mutations**
  - The gene is located on chromosome 17q21.1.
  - The mutations are typically inherited in an autosomal recessive manner.

**Diagnosis**

- **Molecular Testing**
  - DNA sequencing is the primary diagnostic test for GFAP deficiency.
  - It identifies specific mutations in the GFAP gene.

**Clinical Features**

- **Neurological**
  - Growth failure
  - Intellectual disability
  - Seizures
  - Retinal degeneration

- **Diagnostic Tests**
  - Brain MRI
  - EEG
  - Retinal examination

**Treatment**

- **Supportive Care**
  - Nutritional support
  - Physical therapy
  - Occupational therapy

**Prevention**

- **Carrier Testing**
  - For unaffected parents, carrier testing is available.

**References**

G6PD Deficiency

G6PD deficiency is a common genetic condition caused by a deficiency of an enzyme called glucose-6-phosphate dehydrogenase (G6PD). This enzyme helps protect red blood cells from damage. In people with G6PD deficiency, red blood cells are more likely to become damaged in certain environments or triggers, which can lead to episodes of anaemia. This test includes two common variants linked to G6PD deficiency.

Overview  Scientific Details  Frequently Asked Questions

What does this test do? 
This test looks for the two most common variants linked to G6PD deficiency.

What do these tests do? 
These tests do not indicate which variant of G6PD deficiency is present or how severe the condition is.

The report says the variant included in this test may occur most common and be studied in certain ethnicities, what does this mean? 
The MMIV variant is most common and best studied in people of African descent. It is also found in people with all racial ancestry, including people of Hispanic or Latin descent. The SIVMV variant is more common and best studied in people of Southern European, Kurdish, Jewish, Middle Eastern, Central Asian, and South Asian descent. However, if a male has a variant detected on his female has had two copies of a variant of a son, he is still expected to be at risk for G6PD deficiency, regardless of ethnic background. See Scientific Details for more information.

What can the genetic test result be based on the sex of the individual? 
The sex-specific results will be based on the sex of the individual.

Can you learn more about the G6PD deficiency genetic counseling? 
For more information, please contact a genetic counselor or health professional.

Can you learn more about the G6PD deficiency support groups and other resources? 
You can learn more about G6PD deficiency from the following resources:

- G6PD Deficiency Association
- g6pd deficiency foundation
- National Organization for Rare Disorders (NORD)
- Genetic and Rare Disease Information Center

If you have questions about your results or how they might affect you or your family, a genetic counselor may be able to help. Learn more about genetic counseling.

My report shows a variant was detected, what does this mean? 
You have not been identified with the genetic variant detected in the test. People without any genetic variants in the G6PD gene are not at risk of developing symptoms of G6PD deficiency. However, you could still have another genetic variant not included in this test.

My report shows a variant was detected, what does this mean? 
You have been identified with the genetic variant detected in the test. People with G6PD deficiency may experience symptoms of the condition, which can be managed with lifestyle changes and medication.

Can I order this test again? 
Yes, you can order this test again.

Can I get a copy of my results? 
Yes, you can get a copy of your results.

More questions? Check out our Customer Care Help Center.