G6PD Deficiency

G6PD deficiency is a common genetic condition caused by a deficiency in an enzyme called glucose-6-phosphate dehydrogenase (G6PD). This enzyme is needed to break down harmful byproducts of red blood cell metabolism. When this enzyme is lacking, the body is more likely to produce harmful chemicals that can cause red blood cell destruction (hemolysis), which can lead to severe anemia. This is most commonly seen in men and boys, but can also affect women and girls.

Causes
- G6PD deficiency is inherited in an autosomal recessive pattern.
- A person needs to inherit two copies of the mutant gene (one from each parent) to have the condition.
- There is no known cure for G6PD deficiency, but treatment can help manage symptoms.

How To Use This Test
- This test does not diagnose G6PD deficiency or any other hematologic conditions.
- If you or a family member have a known mutation associated with G6PD deficiency, talk to your doctor about the best way to test for it.

Intended Use
- This test is intended to detect G6PD deficiency and provide a genetic diagnosis of G6PD deficiency.

Limitations
- False-negative results can occur in individuals with low G6PD activity.
- False-positive results can occur in individuals with other hemoglobinopathies.

Important Considerations
- The test excludes G6PD variants associated with anemia due to hemoglobinopathies.
- The test is not recommended for use in populations with a high prevalence of G6PD variants not associated with anemia.

We could not determine if you have either of the two variants tested linked to G6PD deficiency.

We could not rule out either of the two variants we tested linked to G6PD deficiency.

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

About G6PD Deficiency
- G6PD deficiency is a genetic disorder that affects red blood cells.
- It occurs when the body cannot produce enough glucose-6-phosphate dehydrogenase (G6PD), an enzyme needed to break down harmful byproducts of red blood cell metabolism.
- G6PD deficiency is inherited in an autosomal recessive pattern.
- A person needs to inherit two copies of the mutant gene (one from each parent) to have the condition.
- The condition is more common in men and boys than in women and girls.
- G6PD deficiency can be triggered by certain factors, such as medications, infections, and exposure to certain chemicals.
- Symptoms may include fever, weakness, fatigue, and jaundice.

If you are pregnant or planning to become pregnant, talk to your doctor about how G6PD deficiency may affect your health and the health of your baby.

Consider talking to a healthcare professional if this condition runs in your family or you are concerned about your results.
G6PD Deficiency

What is this test for?

This test looks at the G6PD and G6PD* genotypes in the G6PD gene linked to G6PD deficiency. People with the same genetic variants included in this test are at risk of developing symptoms of G6PD deficiency. However, not everyone with these genetic variants will develop symptoms. This test does not include all possible genetic variants linked to G6PD deficiency.

Is this answer helpful? 0 1

What does this test look for?

This test does not diagnose G6PD deficiency. Only a healthcare professional can do that. This test does not include all possible genetic variants linked to G6PD deficiency. This test does not provide a complete assessment of risk for anyone with variants linked to G6PD deficiency.

Is this answer helpful? 0 1

The report app the variants included in the test most common and best studied in certain regions. What does this mean?

The G6PD variant is the most common and best studied in people of African ancestry. It is also found in people with African ancestry, including people of Europe and Asian descent.

The G6PD* variant is a rare common and best studied in people of Southern European, North, Jewish, Middle Eastern, Central Asian, and South Asian descent.

However, if a variant is detected, it is likely that the variant is rare or that it is a new variant, so it is not expected to be at risk for G6PD deficiency, regardless of ethnicity. See Scientific Details for more information.

There are other genetic variants linked to G6PD deficiency that are not included in this test, including variants that are more common in people of East and Southeast Asian descent. It is likely that it will provide a person to have some of these other genetic variants.

Is this answer helpful? 0 1

The report app my genetic results based on the sex it identified in my account settings. What does this mean?

For some of our reports, including the G6PD Deficiency report, results and alleles will vary based on different versions of the report. The version of the report you receive, and the interpretation of your genetic result, will be based on the version you reported when you first enrolled your DNA with us. For example, a control's Y sex variant was marked, but their related female when registering their Y, she likely will receive the female version of the report.

Your self-reported sex is important for the G6PD Deficiency report, as you are expected because of the way this condition is inherited. In this report, if you have self-reported to be the G6PD gender, which is located on the X chromosome. Females have two X chromosomes, whereas males have one X and one Y chromosome. As a result, females have two copies of the G6PD gene, which can lead to symptoms of G6PD deficiency. This test also assesses what results will be if one copy of a variant is not expected to develop symptoms of G6PD deficiency, as it makes results only one copy of a variant in some risk for the condition. This information is provided, called a karyotype, which also have implications for people who are currently being diagnosed with a particular sex variant. This help add to determine what sex it identified in your account settings.

The Scientific Details page of this report, your genotype is also based on your self-reported sex, because females and males have different functions of certain chromosomes. For example, the sex variant "female" refers to individuals whose sex is a female typically XY. We use the word "male" to refer to individuals whose genetic sex is a male typically XY. We use the word "non-binary" to refer to individuals whose genetic sex is unknown this report.

Is this answer helpful? 0 1

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

You can learn more about G6PD deficiency from the following resources:

- G6PD Deficiency Association
- G6PD Deficiency Foundation
- Medical Guidelines for Red Blood Cell Disorders INCREASE
- Genetic and Rare Disease Information Center

If you have questions about your race they might affect your personal, genetic counseling may be able to help. Learn more about genetic counseling.

Is this answer helpful? 0 1

My report says my result could not be determined. What does this mean?

We would expect our result if you are a new blood test to check for genetic variants. This can be caused by either test error or other factors that interfere with the way. It can also occur for individuals whose genetic results were analyzed on a different test system. Additionally, this can happen in people who have a genetic variant in a different than what is expected based on the test they obtained their results.