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.

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Jamie, you have **one** of the two genetic variants we tested.

Most females with this genetic result are not expected to develop symptoms of G6PD deficiency. However, you could pass this variant on to your children.

How To Use This Test

This test does not diagnose G6PD deficiency or any other health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Genetic Health Risk tutorial
See Scientific Details
See Frequently Asked Questions

Intended Uses

- Tests for the **V68M** and **S188F** variants in the G6PD gene linked to G6PD deficiency.

Limitations

- Does **not** test for all possible variants linked to G6PD deficiency.
- Your result for this report depends on the sex you indicated in your account settings.

Ethnicity Considerations

- The **V68M** variant included in this test is most common and best studied in people of **African** descent. This variant is also found in people with African ancestry, including people of **Hispanic** or **Latino** descent.
- The **S188F** variant included in this test is most common and best studied in people of **Southern European**, **Kurdish Jewish**, **Middle Eastern**, **Central Asian**, and **South Asian** descent.
- This test does not include variants that are more common in people of East and Southeast Asian descent.

You have **one** of the two variants we tested linked to G6PD deficiency.

We detected one copy of the V68M variant in the G6PD gene.

Most females with just one copy of a variant are not expected to develop symptoms of G6PD deficiency. However, certain factors can affect the chances of developing symptoms in these individuals, including the percentage of G6PD-deficient red blood cells in the body, which varies from female to female and can change over time.

See Scientific Details

There is still a chance of having another variant linked to G6PD deficiency.

However, even females with two variants typically do not develop symptoms of G6PD deficiency unless they are exposed to certain environmental triggers.

See Scientific Details

Your male children may be at risk for G6PD deficiency, even if your partner doesn't have a variant.

Each of your children has a 50% chance of inheriting the V68M variant from you. Males are at risk of developing symptoms of G6PD deficiency even if they only have one copy of a variant. This is because of the way this variant is inherited, called **X-linked recessive inheritance**.

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

Certain medications

A number of medications can trigger symptoms in people with G6PD deficiency. For example, certain medications that are used to treat malaria, cancer, and bacterial infections can trigger symptoms in some people. People with G6PD deficiency should consult with a healthcare professional before starting or stopping any medications.

See Scientific Details for more information

About G6PD Deficiency

Also known as: Glucose-6-Phosphate Dehydrogenase Deficiency, G6PDD

When it develops

Because it is a genetic condition, G6PD deficiency is present at birth. However, people with this condition typically don't develop symptoms unless they are exposed to certain triggering factors. Many people with G6PD deficiency never develop symptoms.

Typical signs and symptoms

- Anemia
- Dark urine
- Fatigue
- Pale skin
- Shortness of breath
- Jaundice (yellowing of the skin and eyes), especially in newborns

How common is the condition?

Approximately 400 million people worldwide have G6PD deficiency. The condition occurs most frequently in people from certain parts of Africa, Asia, the Middle East, and the Mediterranean. About 1 in 10 African American males has G6PD deficiency. Because of the way it is inherited, males are more likely to have this condition than females.

How it's treated

Most people with G6PD deficiency do not require treatment. People with G6PD deficiency often manage their condition by avoiding certain medications and foods that may trigger symptoms. If a person is exposed to a trigger and develops anemia, symptoms usually clear up on their own. However, in some cases patients may require a blood transfusion.

Read more at: Genetic and Rare Diseases Information Center MedlinePlus

Consider sharing this result with a healthcare professional.

If you have a family history of this condition or think you have symptoms, consult with a healthcare professional.

Print report

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

See our Frequently Asked Questions for more information.

FAQs

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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.

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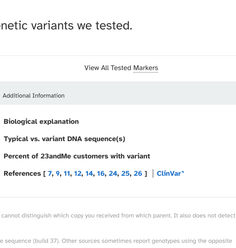
G6PD deficiency is linked to variants in the G6PD gene.

G6PD

The G6PD gene contains instructions for making an enzyme called glucose-6-phosphate dehydrogenase, or G6PD. This enzyme is important for protecting red blood cells against damage. Certain variants in the G6PD gene reduce the levels or activity of the enzyme. This can lead to the destruction of red blood cells in response to certain medications, infections, and foods. The G6PD gene is located on the X chromosome, which means that males have one copy of this gene and females have two.

Read more at Genetics Home Reference*



You have one of the two genetic variants we tested.

Variants Detected | View All Tested Markers

Marker Tested	Genotype*		Additional Information
V68M **Gene:** G6PD **Marker:** rs1050828	**C** Typical copy from one of your parents	**T** Variant copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [7, 9, 11, 12, 14, 16, 24, 25, 26] | ClinVar*

*The genotype shown above is based on the sex you indicated in your account settings. This test cannot distinguish which copy you received from which parent. It also does not detect sex chromosome conditions, such as Turner syndrome.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation

This report provides information about risk for G6PD deficiency in people who have the variants included in this test.

Health Risk Estimates

Risk estimates are based on clinical studies that identify an association between a genotype and a health condition.

Consider talking to a healthcare professional if you have any concerns about your results.

References [6, 7, 13, 19, 20, 23, 28]

Risk estimates

- The World Health Organization (WHO) provides classifications for different G6PD variants based on their residual enzymatic activity. Class III variants have 19–66% of normal enzyme activity, and class II variants have less than 18% of normal enzyme activity. The V68M variant is considered a class III variant, and the S188F variant is considered a class II variant.
- Not everyone with a variant included in this test has G6PD deficiency. For example, studies suggest that about 78% of males with the V68M variant have the condition, and about 59% of females with two copies of this variant have the condition.
- Most females with just one copy of a G6PD variant are not expected to develop symptoms of G6PD deficiency. However, certain factors can affect the chances of developing symptoms in these individuals, including the percentage of G6PD-deficient red blood cells in the body, which varies from female to female and can change over time.
- People with G6PD deficiency usually don't have symptoms unless they are exposed to certain medications, infections, or foods. Many people with the condition never experience symptoms.

Other Factors

G6PD deficiency is a genetic condition. People with this condition are at risk for hemolytic anemia, which occurs when red blood cells are destroyed faster than the body can replace them. Exposure to certain factors is usually required to trigger hemolytic anemia.

This is not a complete list of other factors.

Only people with certain variants in the G6PD gene are at risk for G6PD deficiency and its symptoms.

The factors described here include the most common and well-established risk factors associated with anemia in people with G6PD deficiency. Other factors not listed here may also influence risk for anemia in people with the condition. Specific triggers may vary from person to person, even among people with the same variant.

Consult with a healthcare professional before making any major lifestyle changes.

Other Factors	References
Certain medications A number of medications can trigger symptoms in people with G6PD deficiency. For example, certain medications that are used to treat malaria, cancer, and bacterial infections can trigger symptoms in some people. People with G6PD deficiency should consult with a healthcare professional before starting or stopping any medications.	[4, 5, 10, 17]
Certain infections Certain infections can trigger symptoms in people with G6PD deficiency. These include bacterial infections, such as Salmonella and E. coli, as well as viral infections, such as hepatitis and flu.	[4, 5, 10, 17]
Certain foods For some people with G6PD deficiency, eating certain foods such as fava beans (also called broad beans) can trigger symptoms.	[4, 5, 10, 17]
Certain chemicals For some people with G6PD deficiency, being exposed to certain chemicals can trigger symptoms. These chemicals include naphthalene (which can be found in moth balls) and methylene blue (a dye used in some medical tests and scientific experiments).	[4, 5, 10, 15, 17]

Test Details

Indications for Use

The 23andMe PGS Genetic Health Risk Report for G6PD Deficiency is indicated for reporting of the V68M and S188F variants in the G6PD gene. This report describes if a person has one or more variants linked to G6PD deficiency and a higher risk for episodes of anemia, but it does not describe a person's overall risk of developing symptoms. This report is most relevant for people of African, Southern European, Kurdish Jewish, Middle Eastern, Central Asian, and South Asian descent.

Special Considerations

- This test does not include the N126D variant in the G6PD gene. In genetic testing for G6PD deficiency, the V68M variant and the N126D variant are usually tested together because they are both part of the G6PD A- haplotype. However, the N126D variant itself is not linked to G6PD deficiency.
- Genetic testing for G6PD deficiency in adults in the general population is not currently recommended by any healthcare professional organizations.

Test Performance Summary

Clinical Performance [6, 13, 23]

- The V68M variant included in this test is expected to be responsible for up to 98% of cases of G6PD deficiency in people of African descent.
- The S188F variant included in this test is expected to be responsible for the majority of cases of G6PD deficiency in people of Southern European, Kurdish Jewish, Middle Eastern, and Central Asian descent.
- This test does not include variants that are more common in people of East and Southeast Asian descent.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

1. Al-Ali AK et al. (2002). "Molecular characterization of glucose-6-phosphate dehydrogenase deficiency in the Eastern Province of Saudi Arabia." Clin Chem Lab Med. 40(8):814-6.
2. Al-Musawi BM et al. (2012). "Molecular characterization of glucose-6-phosphate dehydrogenase deficient variants in Baghdad city - Iraq," BMC Blood Disord. 12:4.
3. Alfinito F et al. (1997). "Molecular characterization of G6PD deficiency in Southern Italy: heterogeneity, correlation genotype-phenotype and description of a new variant (G6PD Neapolis)." Br J Haematol. 98(1):41-6.
4. Beutler E. (1994). "G6PD deficiency." Blood. 84(11):3613-36.
5. Beutler E. (2008). "Glucose-6-phosphate dehydrogenase deficiency: a historical perspective." Blood. 111(1):16-24.
6. Cappellini MD et al. (2008). "Glucose-6-phosphate dehydrogenase deficiency," Lancet. 371(9606):64-74.
7. Carter N et al. (2011). "Frequency of glucose-6-phosphate dehydrogenase deficiency in malaria patients from six African countries enrolled in two randomized anti-malarial clinical trials." Malar J. 10:241.
8. Clark TG et al. (2009). "Allelic heterogeneity of G6PD deficiency in West Africa and severe malaria susceptibility." Eur J Hum Genet. 17(8):1060-5.
9. Dombrowski JG et al. (2017). "G6PD deficiency alleles in a malaria-endemic region in the Western Brazilian Amazon." Malar J. 16(1):253.
10. Frank JE. (2005). "Diagnosis and management of G6PD deficiency." Am Fam Physician. 72(7):1277-82.

See all references

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
June 24, 2020	The variant S188F (rs5030868) was added to the report. If this variant was detected, customers will see this reflected in their result.
June 4, 2018	G6PD Deficiency report created.

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G6PD Deficiency

What does this test do?

What does this test **not** do?

The report says the variants included in this test are most common and best 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?

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?

My report says **one variant** was detected. What are some things I could do?

How could my result affect my family?

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