Print

# 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 the most common variant linked to G6PD deficiency in people of African descent.

## play+247b4697b9, you **do not have** the genetic variant we tested.

You could still have a variant not covered by this test.

## 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 **Val68Met** variant 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.

### Important Ethnicities

- The variant included in this test is most common and best studied in people of **African** descent.
- This variant is also found in populations with African ancestry, like Hispanics or Latinos.
- This test does **not** include variants that are more common in people of Mediterranean, Middle Eastern, Asian, or Kurdish Jewish descent.

## You **do not have** the most common variant linked to G6PD deficiency in people of African descent.

However, there are other variants linked to G6PD deficiency that are more common in people of Mediterranean, Middle Eastern, and Asian descent. It is still possible to have a variant not covered by this test.

### You do not have the variant we tested linked to G6PD deficiency.

This variant is most commonly found in people of **African** descent. It is also found in populations with African ancestry, like **Hispanics** or **Latinos**.

See Scientific Details

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

However, even people with variants linked to G6PD deficiency usually do not develop symptoms unless they are exposed to certain environmental triggers.

See Scientific Details

## 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)

### 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 men has G6PD deficiency. Because of the way it is inherited, men are more likely to have this condition than women.

### 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* Genetics Home Reference*

## Learn more about G6PD deficiency.

See our Frequently Asked Questions for more information.

FAQs

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

Print report



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G6PD Deficiency

# Scientific Details

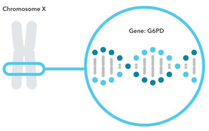
G6PD deficiency is a common genetic condition caused by defects in an enzyme called glucose-6-phosphate dehydrogenase, or G6PD. The G6PD enzyme helps protect red blood cells from damage. In people with G6PD deficiency, red blood cells are destroyed upon exposure to certain environmental triggers, which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

## 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 do not have the genetic variant we tested.

Variants Detected | View All Tested Markers

| Marker Tested | Your Genotype* | | Additional Information |
|---|---|---|---|
| **Val68Met**<br>**Gene:** G6PD<br>**Marker:** rs1050828 | **C**<br>Typical copy from one of your parents | **C**<br>Typical copy from your other parent | **Biological explanation**<br>The variant tested is a change from a C to a T in the DNA sequence of the G6PD gene. This results in an enzyme that is less stable, making it less effective at protecting red blood cells. This variant is also known as c.202G>A, c.292G>A, Val98Met, and G6PD Asahi. This variant is almost always found together with another variant, called Asn126Asp. When these two variants are found together, the combination is called the G6PD A- haplotype.<br>**Typical vs. variant DNA sequence(s)**<br>C Typical Sequence → Substitution → T Variant Sequence<br>**Percent of 23andMe customers with variant**<br>Variant: T<br>European 0.02%<br>African American 14.50%<br>Hispanic or Latino 1.20%<br>South Asian <0.07%<br>**References** [ 4, 6, 8, 9, 11, 13, 17, 18, 19 ] \| 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 abnormalities, such as those observed in 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 variant 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** [ 4, 10, 15, 16 ]

**Risk estimates**

- The variant in this report is most commonly found in people of African descent. Depending on which part of Africa a person's ancestors are from, the chances of having a variant can be more than 20%.
- Not everyone with a variant has G6PD deficiency. Studies suggest that about 70% of males with the variant in this report have the condition, and about 50% of females with two copies of the variant have the condition. Females with just one copy of the variant are not typically at risk for developing symptoms of G6PD deficiency.
- People with G6PD deficiency usually don't have symptoms unless they are exposed to certain medicines, 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**<br>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. | [ 1, 2, 7, 14 ] |
| **Certain infections**<br>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. | [ 1, 2, 7, 14 ] |
| **Certain foods**<br>For some people with G6PD deficiency, eating certain foods such as fava beans (also called broad beans) can trigger symptoms. | [ 1, 2, 7, 14 ] |
| **Certain chemicals**<br>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). | [ 1, 2, 7, 12, 14 ] |

## Test Details

### Indications for Use

The 23andMe PGS Genetic Health Risk Report for G6PD Deficiency is indicated for reporting of the Val68Met variant in the G6PD gene. This report describes if a person has a variant associated with 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 descent.

**Special Considerations**

- This test does not include the Asn126Asp variant in the G6PD gene. The Asn126Asp variant is not linked to G6PD deficiency but is commonly included in genetic tests for this condition.
- 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** [ 3, 10 ]

The variant included in this test is expected to be responsible for up to 90% of cases of G6PD deficiency in people of African descent. This test does not include variants that are more common in people of Mediterranean, Middle Eastern, Asian, and Kurdish Jewish 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

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Hide additional 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 4, 2018** | G6PD Deficiency report created. |



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G6PD Deficiency

# Frequently Asked Questions

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 the most common variant linked to G6PD deficiency in people of African descent.

## G6PD Deficiency

### What does this test do?

This test looks for the Val68Met variant in the G6PD gene linked to G6PD deficiency.

People with the genetic variant included in this test may be at risk for developing symptoms of G6PD deficiency. However, not everyone with this genetic variant will develop symptoms.

This test does not include all possible genetic variants linked to G6PD deficiency.

Is this answer helpful? Yes No

### What does this test **not** do?

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 anemia in people with a variant linked to G6PD deficiency.

Is this answer helpful? Yes No

### The report says the variants included in this test are best studied in people of **African** descent. What if I'm not of African descent or am only partly of African descent?

The tested variant is expected to be less common in people who are only partly of African descent, including people of Hispanic or Latino descent. The variant is expected to be rare in people who are not of African descent.

The effect of this variant on a person's risk for developing symptoms of G6PD deficiency is best studied in people of African descent. However, if a man who is not of African descent has this variant, he is still expected to be at risk for G6PD deficiency. Similarly, if a woman who is not of African descent has two copies of the variant, she is still expected to be at risk for G6PD deficiency. See Scientific Details for more information.

There are other genetic variants linked to G6PD deficiency that can be more common in people of non-African descent. These other variants are not included in this test. It is still possible for a person to have one of these other genetic variants, especially if they are not of African descent.

Is this answer helpful? Yes No

### The report says my genetic result is based on the sex I indicated in my account settings. What does that mean?

For some of our reports, including the G6PD Deficiency report, men and women will receive different versions of the report. The version of the report you receive, and the interpretation of your genetic result, are based on the sex you reported when you first registered your 23andMe kit. For example, if a customer's sex at birth was male but she told us she is female, she will receive the female version of the report.

Your self-reported sex impacts the version of the G6PD Deficiency report you received because of the way this condition is inherited. In this report, the variant we tested is in the G6PD gene, which is located on the X chromosome. Females typically have two X chromosomes, which means that they typically need two copies of a variant, or two different variants, to be at risk for developing symptoms of G6PD deficiency. In contrast, males typically only have one X chromosome, so they only need one copy of a variant in order to be at risk. This inheritance pattern, called X-linked recessive inheritance, also has implications for people who are considering having children. See this help article to learn more.

On the Scientific Details page of the report, your genotype is also based on your self-reported sex, because females and males have different numbers of X chromosomes.

In this report, we use the word "women" to refer to individuals whose genetic sex is female (typically XX). We use the word "men" to refer to individuals whose genetic sex is male (typically XY).

You can update your self-reported sex in your account settings.

Is this answer helpful? Yes No

### 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
- National Organization for Rare Disorders (NORD)
- Genetic and Rare Diseases 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.

Is this answer helpful? Yes No

### My report says **zero variants** were detected. What does this mean?

You do not have the genetic variant we tested. People without any genetic variants in the G6PD gene are not at risk for developing symptoms of G6PD deficiency. However, you could still have another genetic variant not included in this test.

The variant we tested is most common in people of African descent. This test does not include variants that are more common in people of other ethnicities, including people of Mediterranean, Middle Eastern, Asian, and Kurdish Jewish descent.

Is this answer helpful? Yes No

### My report says **zero variants** were detected. What are some things I could do?

Your genetic result means you do not have the most common variant linked to G6PD deficiency in people of African descent. However, it is still possible to have a variant not included in this test.

Consider talking to a healthcare 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.

Is this answer helpful? Yes No

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



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