Primary Hyperoxaluria Type 2

PH2 is a rare genetic disorder. It is characterized by frequent kidney stones that can lead to kidney failure if left untreated. A person must have two variants in the GRHPR gene in order to have this condition.

Erin, you **do not have the variant** we tested.

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

How To Use This Test

This test does not diagnose any 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 Carrier Status tutorial

See Scientific Details

Intended Uses

- To test for the 103delG variant in the GRHPR gene.
- To identify carrier status for PH2.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.

Important Ethnicities

- This test is expected to identify the majority of carriers in people of **European** descent.
- This test does **not** include the most common variant found in people of Asian descent.

You are not a carrier of the variant covered by this test.

However, this test does not cover variants that may be more common in people of your ethnicity.

We ruled out the tested variant for PH2.

This variant is very rare in all ethnicities.

You still have a chance of being a carrier for PH2.

We cannot estimate your chances because sufficient data is not available for your ethnicity.

About Primary Hyperoxaluria Type 2

Also known as: L-Glyceric Aciduria, D-Glycerate Dehydrogenase Deficiency, GR/HPR Deficiency

When symptoms develop

Symptoms typically develop during childhood.

How it's treated

There is currently no known cure. Treatment focuses on managing oxalate levels and hydration in order to slow the development of kidney disease. Kidney transplantation is considered in some cases.

Typical signs and symptoms

- Frequent kidney stones
- Kidney failure if untreated

Ethnicities most affected

This condition is most common in people of European and Asian descent.

Read more at

Genetics Home Reference

GeneReviews

National Organization for Rare Disorders

Consider talking to a healthcare professional if you are thinking about having children.

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Connect with a GC

Share your results with a healthcare professional.

Print report

Learn more about this condition and connect with support groups.

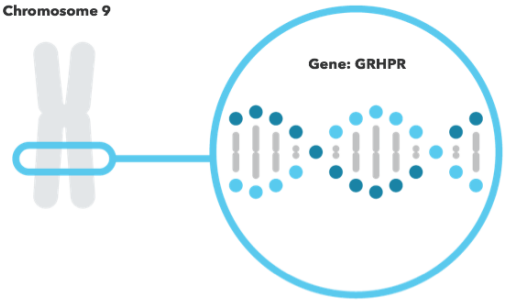
Learn more

PH2 is caused by variants in the GRHPR gene.

GRHPR

The GRHPR gene contains instructions for making an enzyme called enzyme glyoxylate reductase/hydroxypyruvate reductase (GR/HPR). One function of this enzyme is to convert a molecule called glyoxylate into another molecule called glycolate. Certain variants in GRHPR disrupt this function, resulting in the harmful buildup of glyoxylate in the body.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected	View All Tested Markers
0	1

Marker Tested	Your Genotype*		Additional Information
103delG Gene: GRHPR Marker: i5012628	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [1 , 2 , 4 , 6] | ClinVar

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

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

Post-test carrier risk for PH2 is the chance of still being a carrier for the condition if you do not have the variant tested. This chance depends on how common it is to be a carrier for PH2 and whether the variants we tested tend to be found in people of your ethnicity.

Because you do not have the variant we tested, your chances of still being a carrier are lower than for someone who has not been tested. However, we cannot provide an exact estimate because the information needed to calculate post-test carrier risk is not available for your ethnicity.

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Primary Hyperoxaluria Type 2 is indicated for the detection of the 103delG variant in the GRHPR gene. This test is intended to be used to determine carrier status for PH2 in adults, but cannot determine if a person has two copies of a tested variant.

Special Considerations

- This test does not include a large fraction of GRHPR variants that cause PH2.
- There are currently no professional guidelines in the U.S. for carrier testing for this condition.

Test Performance Summary

Carrier Detection Rate & Relevant Ethnicities

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be identified by this test. Carrier detection rate differs by ethnicity and is provided only where sufficient data is available.

European	68%	[5]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 49 samples with known variant status. 49 out of 49 genotype results were correct. About 1 in 1,300 samples may receive a **Not Determined** result. This can be caused by random test error or unexpected DNA sequences that interfere with the test. It can also be caused by having two copies of a variant tested.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Positive results in individuals whose ethnicities are not commonly associated with this condition may be incorrect. Individuals in this situation should consider genetic counseling and follow-up testing.
- 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. Cramer SD et al. (1999). "The gene encoding hydroxypyruvate reductase (GRHPR) is mutated in patients with primary hyperoxaluria type II." Hum Mol Genet. 8(11):2063-9.
2. Cregeen DP et al. (2003). "Molecular analysis of the glyoxylate reductase (GRHPR) gene and description of mutations underlying primary hyperoxaluria type 2." Hum Mutat. 22(6):497.
3. Rumsby G et al. (1993). "Primary Hyperoxaluria Type 2"
4. Rumsby G et al. (2004). "Evaluation of mutation screening as a first line test for the diagnosis of the primary hyperoxalurias." Kidney Int. 66(3):959-63.
5. Takayama T et al. (2014). "Ethnic differences in GRHPR mutations in patients with primary hyperoxaluria type 2." Clin Genet. 86(4):342-8.
6. Webster KE et al. (2000). "Identification of missense, nonsense, and deletion mutations in the GRHPR gene in patients with primary hyperoxaluria type II (PH2)." Hum Genet. 107(2):176-85.