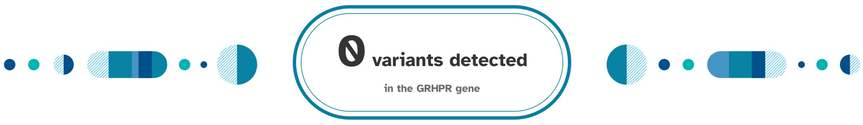
Primary Hyperoxaluria Type 2

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.

Overview | Scientific Details

Jamie, 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.
- Does **not cover** other types of primary hyperoxaluria.

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 likely not a carrier.

We ruled out the tested variant for PH2.

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

You may still have up to a **1 in 880 chance** of carrying a variant not covered by this test.

See Scientific Details

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.

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.

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.

Read more at: Genetics Home Reference | GeneReviews | National Organization for Rare Disorders

Consider talking to a healthcare professional if you are concerned about your results.

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

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.

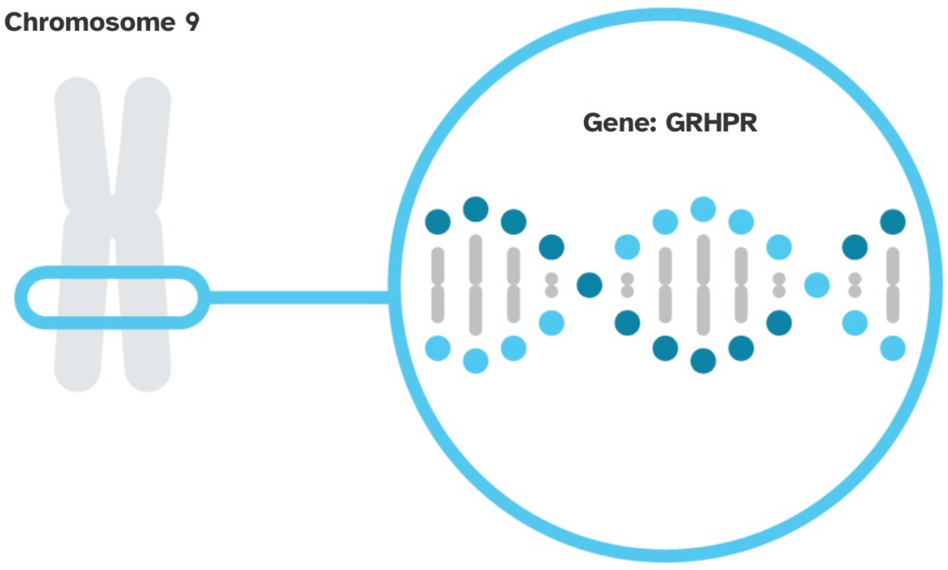
Overview Scientific Details

PH2 is caused by variants in the GRHPR gene.

GRHPR

The GRHPR gene contains instructions for making an enzyme called 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

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, 7] | 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

This report provides an estimate of the chances of still being a carrier for people who do not have the variant(s) tested. This is known as the **post-test carrier risk**.

Post-test carrier risk is based on the average chance of being a carrier for a given ethnicity and the carrier detection rate of the test for a given ethnicity.

View technical article on estimating post-test carrier risk.

Post-Test Carrier Risk

This report provides an estimate of the post-test carrier risk for people of European descent only.

- For people of partial European descent, post-test carrier risk is less than that for those who are fully European. The exact post-test risk depends on how much European ancestry a person has.
- Post-test risk for other ethnicities cannot be provided because sufficient data is not available.

Post-test carrier risk for relevant ethnicities

European	1 in 880	[3]

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

Warnings and Limitations

- This test does not cover all variants that could