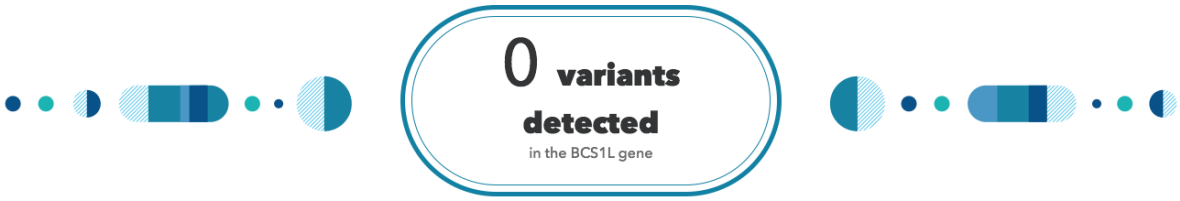
GRACILE Syndrome

GRACILE syndrome is a rare genetic disorder. It is characterized by impaired growth before birth, iron buildup, liver damage, and death in infancy. A person must have two variants in the BCS1L 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 or you have any concerns about your results.

Review the Carrier Status tutorial

See Scientific Details

Intended Uses

- To test for the S78G variant in the BCS1L gene.
- To identify carrier status for GRACILE syndrome.

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 most relevant for people of **Finnish** descent.

You are likely not a carrier.

This result is relevant for you because you have **Finnish** ancestry.

We ruled out the most common variant for GRACILE syndrome in people of Finnish descent.

You still have a chance of being a carrier for GRACILE syndrome.

You may still have up to a **1 in 1,100,000 chance** of carrying a variant not covered by this test.

See Scientific Details

About GRACILE Syndrome

Also known as: Fellman Syndrome. GRACILE is short for: growth retardation, aminoaciduria, cholestasis, iron overload, lactic acidosis, and early death

When symptoms develop

Symptoms typically develop before birth.

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms and ultimately providing end-of-life supportive care.

Typical signs and symptoms

- Small size at birth
- Poor growth and weight gain
- Iron buildup in the liver
- Buildup of lactic acid in the body
- Kidney and liver problems
- Death in infancy

Ethnicities most affected

This syndrome is most common in people of Finnish descent.

Read more at

Genetics Home Reference

Genetic and Rare Diseases Information Center

Orphanet

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.

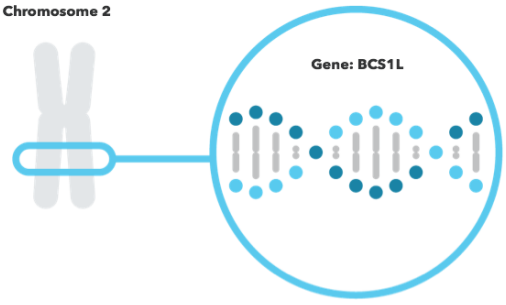
Print report

GRACILE syndrome is caused by variants in the BCS1L gene.

BCS1L

The BCS1L gene contains instructions for making part of a protein called complex III that helps cells turn food into energy. This protein is found in mitochondria, small structures inside of cells that produce energy for the body. Certain variants in BCS1L prevent cells from making enough complex III.

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
S78G Gene: BCS1L Marker: i5012660	**A** Typical copy from one of your parents	**A** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [1 , 2 , 3 , 4 , 5 , 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

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 Finnish descent only.

- For people of partial Finnish descent, post-test carrier risk is less than that for those who are fully Finnish. The exact post-test risk depends on how much Finnish 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

Finnish	1 in 1,100,000	[2]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for GRACILE Syndrome is indicated for the detection of the S78G variant in the BCS1L gene. This test is intended to be used to determine carrier status for GRACILE syndrome in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of Finnish descent.

Special Considerations

- 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.

Finnish	>99%	[1 , 6]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 46 samples with known variant status. 46 out of 46 genotype results were correct. Fewer than 1 in 100,000 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. Fellman V et al. (2008). "Screening of BCS1L mutations in severe neonatal disorders suspicious for mitochondrial cause." J Hum Genet. 53(6):554-8.
2. Fellman V. (2002). "The GRACILE syndrome, a neonatal lethal metabolic disorder with iron overload." Blood Cells Mol Dis. 29(3):444-50.
3. Kotarsky H et al. (2010). "Characterization of complex III deficiency and liver dysfunction in GRACILE syndrome caused by a BCS1L mutation." Mitochondrion. 10(5):497-509.
4. Kotarsky H et al. (2012). "Metabolite profiles reveal energy failure and impaired beta-oxidation in liver of mice with complex III deficiency due to a BCS1L mutation." PLoS One. 7(7):e41156.
5. Levéen P et al. (2011). "The GRACILE mutation introduced into Bcs1l causes postnatal complex III deficiency: a viable mouse model for mitochondrial hepatopathy." Hepatology. 53(2):437-47.
6. Visapää I et al. (2002). "GRACILE syndrome, a lethal metabolic disorder with iron overload, is caused by a point mutation in BCS1L." Am J Hum Genet. 71(4):863-76.