

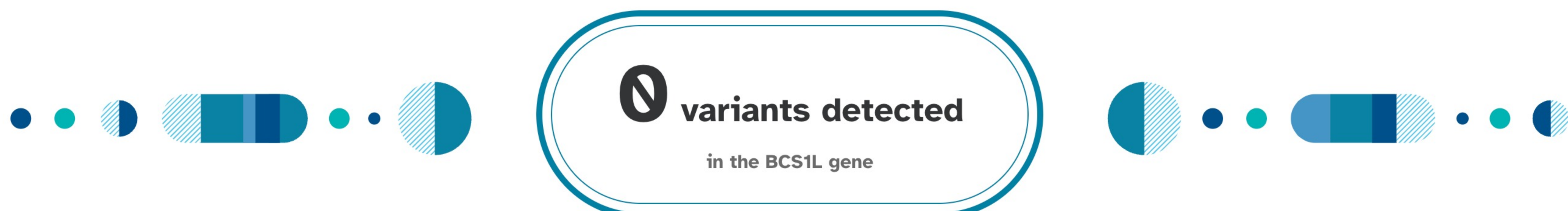
# GRACILE Syndrome

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.

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 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 may be less relevant for you because the variants that cause GRACILE syndrome are rarely found in people of your ethnicity.

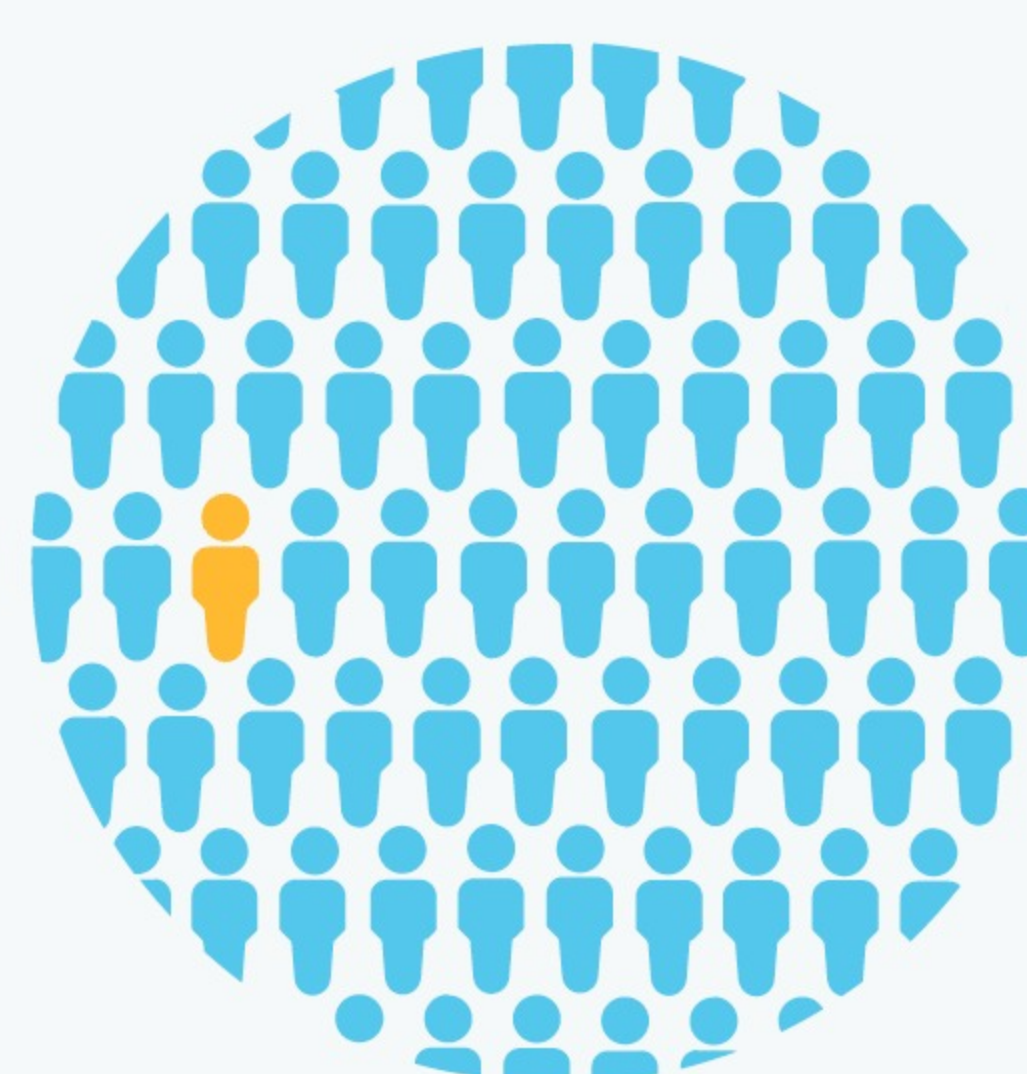


We ruled out the tested variant for GRACILE syndrome.

This variant is most common in people of **Finnish** descent.

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

We cannot estimate your chances because this condition is rare and not well studied in your ethnicity.



## 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.

### 🧪 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

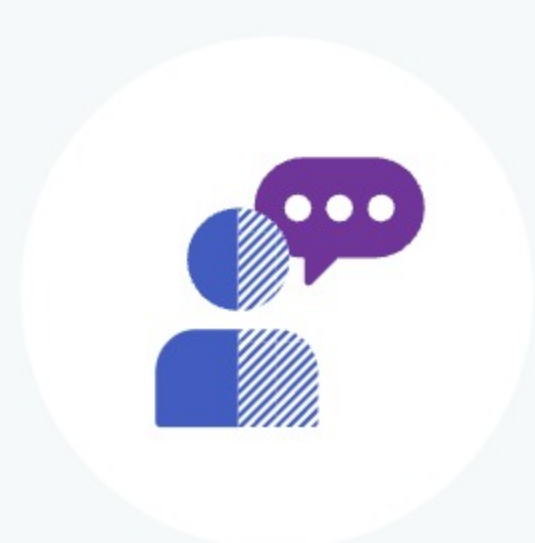
This syndrome is most common in people of Finnish descent.

### 🏥 How it's treated

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

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



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Gift a kit

Refer friends, earn rewards.

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- Research Overview
- Surveys and Studies
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# GRACILE Syndrome

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Overview **Scientific Details**

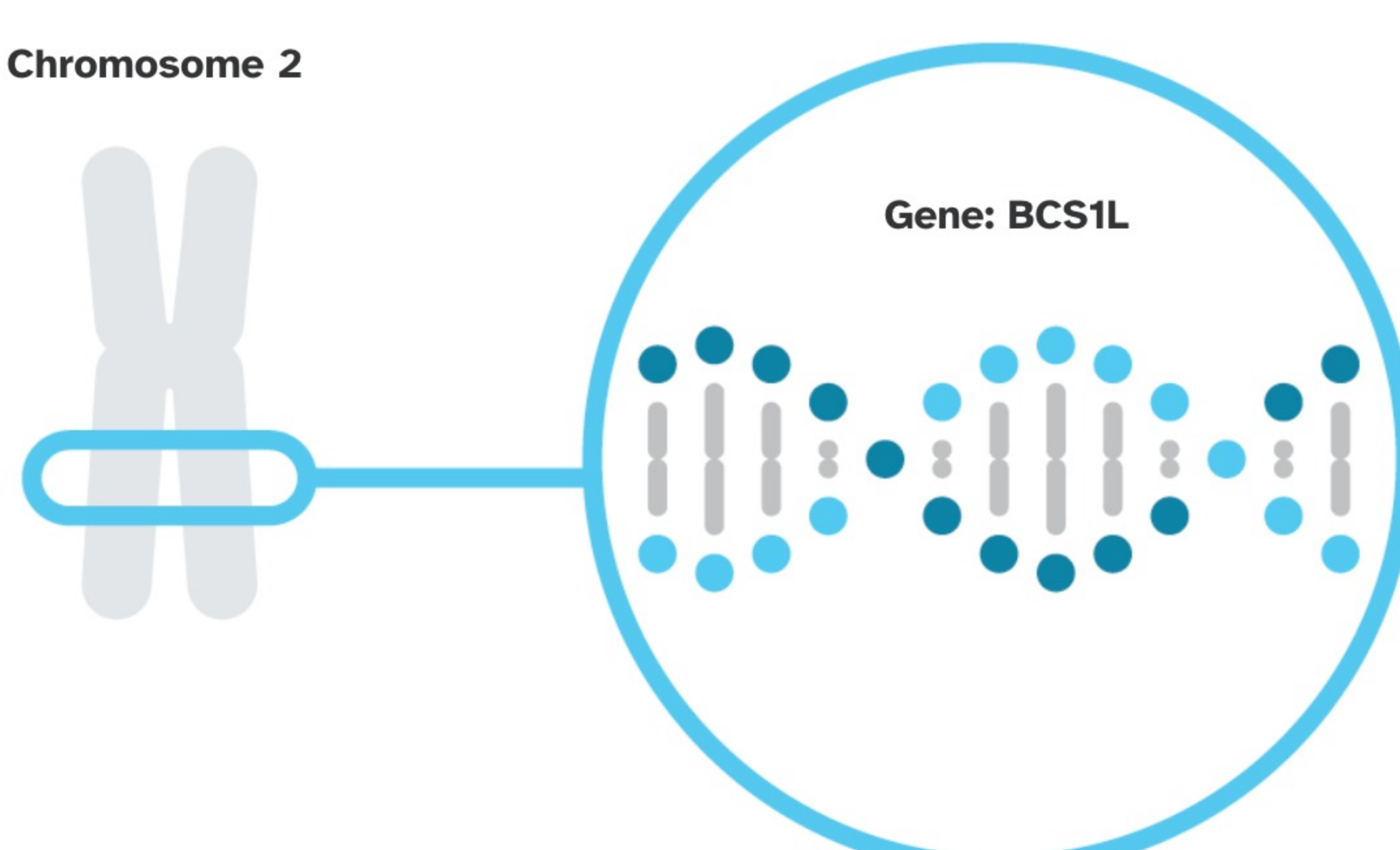
GRACILE syndrome is caused by variants in the BCS1L gene.

## 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](#)

### Chromosome 2



You have no variants detected by this test.

Variants Detected		View All Tested Markers	
Marker Tested	Your Genotype*	Additional Information	
<b>S78G</b> Gene: BCS1L Marker: <b>i5012660</b>	<b>A</b> Typical copy from one of your parents	 <b>A</b> Typical copy from your other parent	<ul style="list-style-type: none"> <li>Biological explanation</li> <li>Typical vs. variant DNA sequence(s)</li> <li>Percent of 23andMe customers with variant</li> <li>References [ 1, 2, 3, 4, 5, 6 ]   ClinVar</li> </ul>

\*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 ]
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## 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 ]
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#### 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.
- 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

- Fellman V et al. (2008). "Screening of BCS1L mutations in severe neonatal disorders suspicious for mitochondrial cause." J Hum Genet. 53(6):554-8. ^
- Fellman V. (2002). "The GRACILE syndrome, a neonatal lethal metabolic disorder with iron overload." Blood Cells Mol Dis. 29(3):444-50. ^
- 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. ^
- 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. ^
- 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. ^
- 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. ^

## Change Log

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

Date	Change
Feb. 18, 2016	Due to improvements in data analysis, some customers who previously received a "Not Determined" result for this report may see an updated result.
Oct. 21, 2015	GRACILE Syndrome report created.



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