Neuronal Ceroid Lipofuscinosis (CLN5-Related)

CLN5-related NCL is a rare genetic disorder. It is characterized by seizures, vision loss, and intellectual disability. A person must have two variants in the CLN5 gene in order to have this form of NCL.

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 Y392X variant in the CLN5 gene.
- To identify carrier status for CLN5-related NCL.

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** NCL caused by variants in other genes.

Important Ethnicities

- This test is most relevant for people of **Finnish** descent.

You are likely not a carrier.

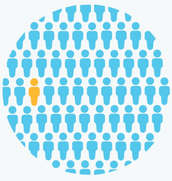
This result may be less relevant for you because the variants that cause CLN5-related NCL are rarely found in people of your ethnicity.

We ruled out the tested variant for CLN5-related NCL.

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

You still have a chance of being a carrier for CLN5-related NCL.

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



About Neuronal Ceroid Lipofuscinosis (CLN5-Related)

Also known as: Late-Infantile NCL, Batten Disease

When symptoms develop

Symptoms typically develop in early childhood.

Ethnicities most affected

This condition is most common in people of Finnish descent.

Typical signs and symptoms

- Intellectual decline
- Seizures
- Loss of ability to control muscles
- Muscle spasms
- Vision loss leading to blindness
- Shortened lifespan

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms, providing physical therapy, and using seizure medications as needed.

Read more at: Genetics Home Reference GeneReviews National Institute of Neurological Disorders and Stroke

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

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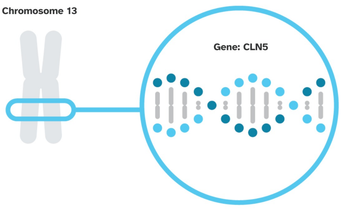
Overview Scientific Details

CLN5-related NCL is caused by variants in the CLN5 gene.

CLN5

The CLN5 gene contains instructions for making a protein called ceroid-lipofuscinosis neuronal protein 5. Although the exact function of this protein is unknown, it is found in compartments within cells called lysosomes that break down and digest material. Certain variants in CLN5 result in defective forms of this protein.

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
Y392X **Gene:** CLN5 **Marker:** i5012678	**AT** Typical copy from one of your parents	**AT** 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, 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 1,899	[6]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Neuronal Ceroid Lipofuscinosis (CLN5-Related) is indicated for the detection of the Y392X variant in the CLN5 gene. This test is intended to be used to determine carrier status for CLN5-related NCL 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	94%	[5]

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

1. Holmberg V et al. (2000). "Phenotype-genotype correlation in eight patients with Finnish variant late infantile NCL (CLN5)." Neurology. 55(4):579-81.
2. Isosomppi J et al. (2002). "Lysosomal localization of the neuronal ceroid lipofuscinosis CLN5 protein." Hum Mol Genet. 11(8):885-91.
3. Lyly A et al. (2009). "Novel interactions of CLN5 support molecular networking between Neuronal Ceroid Lipofuscinosis proteins." BMC Cell Biol. 10:83.
4. Moharir A et al. (2013). "The role of N-glycosylation in folding, trafficking, and functionality of lysosomal protein CLN5." PLoS One. 8(9):e74299.
5. Mole SE et al. (2001). "Neuronal Ceroid-Lipofuscinoses." [Updated 2013 Aug 1].
6. Savukoski M et al. (1998). "CLN5, a novel gene encoding a putative transmembrane protein mutated in Finnish variant late infantile neuronal ceroid lipofuscinosis." Nat Genet. 19(3):286-8.

Change Log

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

Date	Change
Oct. 21, 2015	Neuronal Ceroid Lipofuscinosis (CLN5-Related) report created.