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.

Erin, you do not have the variant we tested.

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

0 variants detected in the CLN5 gene

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.

Intended Uses

- To test for the Y992X 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.

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

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

Ethnicity most affected
This condition is most common in people of Finnish descent.

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

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

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

<table>
<thead>
<tr>
<th>Marker Tested</th>
<th>Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Y392X</td>
<td>AT</td>
<td>- Biological explanation</td>
</tr>
<tr>
<td>Gene: CLNS</td>
<td>AT</td>
<td>- Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: IS012678</td>
<td></td>
<td>- Percent of 23andMe customers with variant</td>
</tr>
</tbody>
</table>

*Typical copy from one of your parents

Additional Information:
- Biological explanation
- Typical vs. variant DNA sequence(s)
- Percent of 23andMe customers with variant

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**

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**

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Finnish 1</td>
<td>1 in 1,800</td>
</tr>
</tbody>
</table>

### Test Details

**Indications for Use**

The 23andMe PGx Carrier Status Test for Neuronal Ceroid Lipofuscinosis (CLNS-Related) is indicated for the detection of the Y392X variant in the CLNS gene. This test is intended to be used to determine carrier status for CLNS-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.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>Finnish 1</td>
<td>94%</td>
</tr>
</tbody>
</table>

**Analytical Performance**

Accuracy was determined by comparing results from this test with results from sequencing for 48 samples with known variant status. 48 out of 48 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


