Agenesis of the Corpus Callosum with Peripheral Neuropathy

ACCPN is a rare genetic disorder. It is characterized by an incomplete connection between the two sides of the brain. This causes developmental disability, weakness, and loss of sensation. A person must have two variants in the SLC12A6 gene in order to have this condition.

You do not have the variant we tested.

0 variants detected in the SLC12A6 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 T813fsX813 variant in the SLC12A6 gene.

• To identify carrier status for ACCPN.

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 French Canadian descent.

About Agenesis of the Corpus Callosum with Peripheral Neuropathy

Also known as:

Andermann Syndrome

Symptoms typically develop during infancy.

Typical signs and symptoms

• Weakness and sensory loss that worsens over time

• Poor or absent reflexes

• Tremors

• Developmental disability

• Shortened lifespan

Read more at:

Genetics Home Reference' GeneReviews'

Ethnicities most affected

This condition is most common in people of French Canadian descent, particularly from the Charlevoix and Saguenay-Lac-Saint-Jean regions of Quebec.

How it's treated

There is currently no known cure. Treatment focuses on physical and occupational therapy as well as other forms of supportive care as symptoms worsen, often into adulthood.

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.

Share your results with a healthcare professional

Learn more about this condition and connect with support groups.
ACPN is caused by variants in the SLC12A6 gene.

The SLC12A6 gene encodes a protein known as the potassium-chloride (K-CI) cotransporter. This protein controls the levels of water, potassium, and chloride inside of cells, which is important for proper brain development and activity. A person must have two copies of a tested variant. The test is most relevant for people of French Canadian descent, particularly from the Charlevoix and Saguenay-Lac-Saint-Jean regions of Quebec French Canadian, particularly from the Charlevoix and Saguenay-Lac-Saint-Jean regions of Quebec. This test does not cover all variants that could cause this condition.

Post-Test Carrier Risk

For people of partial French Canadian descent, post-test carrier risk is less than that for those who are fully French Canadian. The post-test carrier risk depends on how much French Canadian ancestry a person has.

Text Interpretation

This report provides an estimate of the chance of being a carrier for a given ethnicity for people who do not have the variant tested. This is known as the pre-test carrier risk.

Post-test carrier risk is based on the average chance of being a carrier for a given ethnicity. The test may give you the best chance of detecting a carrier if done prior to conception. If you have no other information, this is the best estimate of carrier risk.

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Pre-test carrier risk for relevant ethnicities

<table>
<thead>
<tr>
<th>Ethnicity</th>
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</tr>
</thead>
<tbody>
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<td>French Canadian</td>
<td>8.8%</td>
</tr>
<tr>
<td>Partial French Canadian</td>
<td>1.7%</td>
</tr>
</tbody>
</table>

Test Details

Test Performance

The Centers for Disease Control and Prevention (CDC) does not have a recommendation for the SLC12A6 gene. You are advised to consult with a healthcare professional to determine whether this test is appropriate for you.

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Warnings and Limitations

This test does not diagnose any health conditions.

Positive results in individuals whose detailed medical history is not consistently associated with this condition may be incorrect.

If you are concerned about your results, consult with a healthcare professional.

Learn more about the testing on the "Overview" page.

References


Change Log

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