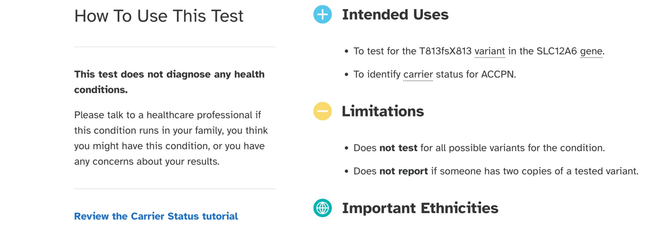
Agenesis of the Corpus Callosum with Peripheral Neuropathy

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.

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 T813fsX813 variant in the SLC12A6 gene.
- To identify carrier status for ACCPN.

Limitations

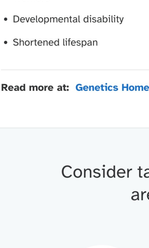
- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.

Important Ethnicities

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

You are likely not a carrier.

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

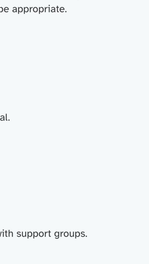


We ruled out the tested variant for ACCPN.

This variant is most common in people of **French Canadian** descent.

You still have a chance of being a carrier for ACCPN.

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



About Agenesis of the Corpus Callosum with Peripheral Neuropathy

Also known as: Andermann Syndrome

When symptoms develop

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

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.

Read more at: Genetics Home Reference GeneReviews

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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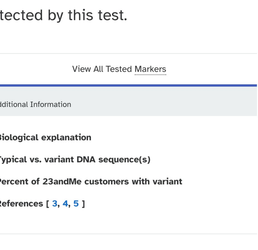
Overview Scientific Details

ACCPN is caused by variants in the SLC12A6 gene.

SLC12A6

The SLC12A6 gene contains instructions for making a protein called a potassium-chloride (K-Cl) cotransporter. This protein controls the levels of water, potassium, and chloride inside of cells, which is important for proper nerve function. Certain variants in SLC12A6 disrupt this protein's function, leading to altered brain development and activity.

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
T813fsX813 **Gene:** SLC12A6 **Marker:** i5012573	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [3, 4, 5]

*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 French Canadian descent only.

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

French Canadian, particularly from the Charlevoix and Saguenay-Lac-Saint-Jean regions of Quebec	1 in 22,999,999	[1]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Agenesis of the Corpus Callosum with Peripheral Neuropathy is indicated for the detection of the T813fsX813 variant in the SLC12A6 gene. This test is intended to be used to determine carrier status for ACCPN in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of French Canadian 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.

French Canadian, particularly from the Charlevoix and Saguenay-Lac-Saint-Jean regions of Quebec	>99%	[3]

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. **Dupré N et al. (2003). "Hereditary motor and sensory neuropathy with agenesis of the corpus callosum." Ann Neurol. 54(1):9-18.**
2. **Dupré N et al. (2006). "Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum". [Updated 2014 June 12].**
3. **Howard HC et al. (2002). "The K-Cl cotransporter KCC3 is mutant in a severe peripheral neuropathy associated with agenesis of the corpus callosum." Nat Genet. 32(3):384-92.**
4. **Salin-Cantegrel A et al. (2011). "Transit defect of potassium-chloride Co-transporter 3 is a major pathogenic mechanism in hereditary motor and sensory neuropathy with agenesis of the corpus callosum." J Biol Chem. 286(32):28456-65.**
5. **Shekarabi M et al. (2012). "Loss of neuronal potassium/chloride cotransporter 3 (KCC3) is responsible for the degenerative phenotype in a conditional mouse model of hereditary motor and sensory neuropathy associated with agenesis of the corpus callosum." J Neurosci. 32(11):3865-76.**

Change Log

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

Date	Change
Oct. 21, 2015	Agenesis of the Corpus Callosum with Peripheral Neuropathy report created.