Leigh Syndrome, French Canadian Type

LSFC is a rare genetic disorder. It is characterized by life-threatening periods of lactic acid buildup and brain injury as well as failure to gain weight. A person must have two variants in the LRPPRC gene in order to have this condition.

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 LRPPRC gene

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**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**

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**Intended Uses**

- To test for the A354V variant in the LRPPRC gene.
- To identify carrier status for LSFC.

**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** other subtypes of Leigh syndrome.

**Important Ethnicities**

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

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You are likely not a carrier.

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

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We ruled out the tested variant for LSFC.

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

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

We cannot estimate your chances because this condition is rare and not well studied in your ethnicity.
About Leigh Syndrome, French Canadian Type

When symptoms develop
Symptoms typically develop during infancy.

How it’s treated
There is currently no known cure. Treatment focuses on providing nutritional support, managing symptoms, and preventing complications.

Typical signs and symptoms
- Buildup of lactic acid in the body
- Episodes of brain injury
- Failure to gain weight
- Poor muscle control and muscle spasms
- Distinctive facial features
- Early death

Ethnicities most affected
This condition is most common in people of French Canadian descent, particularly from the Saguenay-Lac-Saint-Jean region of Quebec.

Read more at
- Genetics Home Reference
- National Organization for Rare Disorders

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

LSFC is caused by variants in the LRPPRC gene.

The LRPPRC gene contains instructions for making a protein called leucine-rich PPR motif-containing protein. This protein controls the levels of an enzyme called complex IV (COX) that is necessary for the cell to generate energy. Certain variants in LRPPRC result in a form of the protein that cannot properly regulate COX levels.

Read more at Genetics Home Reference
References


