

MCAD Deficiency

MCAD deficiency is a rare genetic disorder characterized by episodes of very low blood sugar while fasting or under stress. A person must have two variants in the ACADM gene in order to have this condition.

Erin, you **do not have the variants** 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

- Tests for **multiple variants** in the ACADM gene.
- To identify carrier status for MCAD deficiency.

- 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 **Northern European** descent.

You are likely not a carrier.

This result is relevant for you because you have **European** ancestry.

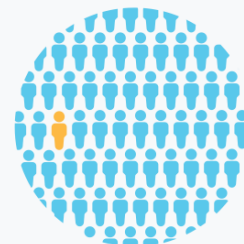


We ruled out the most common variants for MCAD deficiency in people of European descent.

You still have a chance of being a carrier for MCAD deficiency.

You may still have up to a **1 in 280 chance** of carrying a variant not covered by this test.

[See Scientific Details](#)



About MCAD Deficiency

Also known as: Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency, ACADM Deficiency



When symptoms develop

Symptoms typically develop during infancy or early childhood.

How it's treated

There is currently no known cure. Early diagnosis, avoiding fasting, and making certain diet modifications can help limit symptoms and prevent complications.



Typical signs and symptoms

- Severely low blood sugar
- Fatigue
- Vomiting
- Seizures
- Liver problems



Ethnicities most affected

This condition is most common in people of Northern European descent.

Read more at

[Genetics Home Reference](#)

[GeneReviews](#)

[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](#)

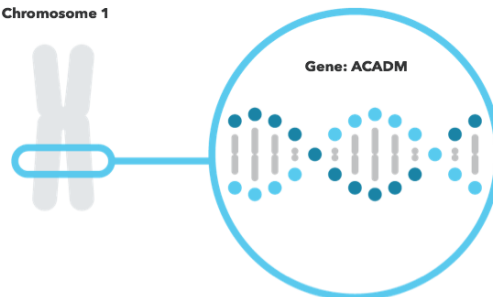
MCAD deficiency is caused by variants in the ACADM gene.

ACADM




The ACADM gene contains instructions for making an enzyme called medium-chain acyl-CoA dehydrogenase (MCAD). This enzyme is needed to make energy for the body from fats called medium-chain fatty acids. Certain variants in the ACADM gene disrupt this function, resulting in low energy and a buildup of fatty acids.

Read more at [Genetics Home Reference](#)

Chromosome 1



You have no variants detected by this test.

Variants Detected		View All Tested Markers	
0		3	
Marker Tested	Your Genotype*	Additional Information	
K304E Gene: ACADM Marker: IS012759	A Typical copy from one of your parents 	A Typical copy from your other parent	<ul style="list-style-type: none">> Biological explanation> Typical vs. variant DNA sequence(s)> Percent of 23andMe customers with variant> References [1, 7, 11, 12] ClinVar ↗
R181C Gene: ACADM Marker: IS012760	C Typical copy from one of your parents 	C Typical copy from your other parent	<ul style="list-style-type: none">> Biological explanation> Typical vs. variant DNA sequence(s)> Percent of 23andMe customers with variant> References [4, 5, 10] ClinVar ↗
S220L Gene: ACADM Marker: IS003117	C Typical copy from one of your parents 	C Typical copy from your other parent	<ul style="list-style-type: none">> Biological explanation> Typical vs. variant DNA sequence(s)> Percent of 23andMe customers with variant> References [1, 8, 13] 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 European descent only.

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

European	1 in 280	[3, 9]
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Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for MCAD Deficiency is indicated for the detection of three variants in the ACADM gene. This test is intended to be used to determine carrier status for MCAD deficiency in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of European 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.

European	64%	[2 , 10]
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Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 150 samples with known variant status. 150 out of 150 genotype results were correct. Fewer than 1 in 100,000 samples may receive a **Not Determined** result for one or more variants included in this test. 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

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6. Matern D et al. (1993). "Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency" [↗](#)
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12. Yokota I et al. (1992). "Impaired tetramer assembly of variant medium-chain acyl-coenzyme A dehydrogenase with a glutamate or aspartate substitution for lysine 304 causing instability of the protein." *J Biol Chem.* 267(36):26004-10. [↗](#)
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