

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.

Overview Scientific Details

Jamie, you **have one of the variants** we tested.

You could pass this variant on to your children.

1 variant detected

in the ACADM 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.

[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 **European** descent.

You are a carrier.

You could pass this variant on to your children.



We detected one variant for MCAD deficiency.

People with only one variant are not expected to have MCAD deficiency.

Your results may be relevant for you if you're thinking about starting a family.

If you and your partner are both carriers, each child may have a **25% chance** of having this condition. Your relatives may also wish to consider testing if they plan to have children.



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.

🌡️ 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.

🏥 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.

Read more at: [MedlinePlus](#) [GeneReviews](#) [National Organization for Rare Disorders](#)

Consider talking to a healthcare professional if you are thinking about having children.



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 your family.

Share your report



If you have other concerns about your results, consult 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](#)

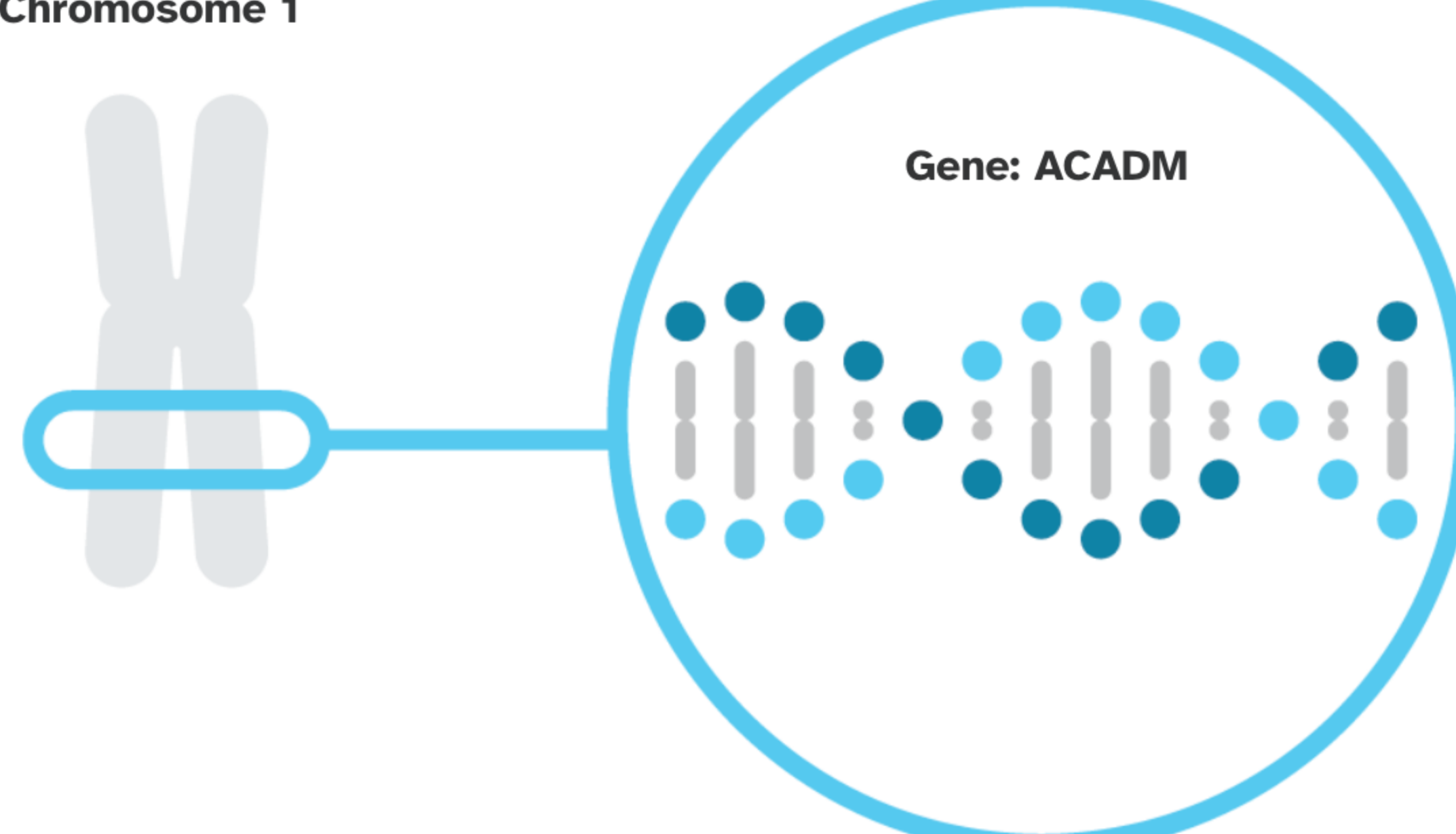
MCAD deficiency is caused by variants in the ACADM gene.

ACADM

The ACADM gene contains instructions for making an enzyme called medium-chain acyl-CoA dehydrogenase (MCAD). This enzyme functions in the mitochondria and 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 [MedlinePlus](#)*

Chromosome 1



You have one variant detected by this test.

Variants Detected		View All Tested Markers	
Marker Tested	Genotype*	Additional Information	
c.199T>C Gene: ACADM Marker: i5012758	C Variant copy from one of your parents	T 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 [2, 9, 12, 20] 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 Details

Indications for Use

The 23andMe PGS Carrier Status Test for MCAD Deficiency is indicated for the detection of four 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

- ACMG recommends that people of all ethnicities who are considering having children should be offered carrier screening for MCAD deficiency.

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	80%	[3, 7, 14]
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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

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- Andresen BS et al. (2001). "Medium-chain acyl-CoA dehydrogenase (MCAD) mutations identified by MS/MS-based prospective screening of newborns differ from those observed in patients with clinical symptoms: identification and characterization of a new, prevalent mutation that results in mild MCAD deficiency." *Am J Hum Genet*. 68(6):1408-18. ^
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- Matsubara Y et al. (1990). "Identification of a common mutation in patients with medium-chain acyl-CoA dehydrogenase deficiency." *Biochem Biophys Res Commun*. 171(1):498-505. ^

See all references ▾

Change Log

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

Date	Change
Dec. 7, 2022	<p>The carrier detection rate was updated for customers who self-report having European ancestry. The chances of still being a carrier were also updated for customers with no variants detected who self-report having European ancestry.</p> <p>The names of the genetic variants included in the report were updated to align with the naming conventions used in the scientific literature.</p>
March 2, 2018	<p>The variant Y42H (i5012758) was added to the report. Customers who have this variant will see this variant detected in their result, and see updated content in their report.</p> <p>The carrier frequency and carrier detection rate were updated for customers who self-report having European ancestry. The chances of still being a carrier were also updated for customers with no variants detected who self-report having European ancestry.</p>
Feb. 18, 2016	<p>Due to improvements in data analysis, some customers who previously reported a "Not Determined" result for one or more of the following genetic markers may see a genotype at these markers: i5003117, i5012759. This may also update the overall report result for these customers.</p>
Oct. 21, 2015	MCAD Deficiency report created.



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