MAYO CLINIC

1-800-533-1710

ACADM Gene Analysis

ACADM

PATIENT NAME				ORDER NUMBER
ACADM, ABNORMAL				**P427000143**
PATIENT ID SA00175839	DATE OF BIRTH 10/30/1990	AGE 34 Y	SEX Male	REQUESTED BY CLIENT CLIENT
COLLECTED 5/26/2025, 7:00 AM	RECEIVED 5/27/2025, 10:43 AM	REPORTED 6/4/2025, 3:07 PM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location. 7028846 MCL RochesterCampus Rochester MN 55901				CLIENT ORDER NUMBER SA00175839 CLIENT MRN SA00175839

TEST DESCRIPTION

Evaluation of the ACADM gene associated with medium chain acyl-CoA dehydrogenase (MCAD) deficiency

SPECIMEN

DNA (ul)

RESULT SUMMARY

Pathogenic Variants Detected

RESULT

Gene (Transcript)	Variant	Zygosity	Classification
ACADM (NM_000016.5)	c.985A>G p.Lys329Glu (p.K329E) Chr1(GRCh37):g.76226846A>G	Homozygous	Pathogenic

The following PATHOGENIC variant was detected:
ACADM (NM_000016.5), chr1(GRCh37):g.76226846A>G, c.985A>G, p.Lys329Glu (p.K329E), homozygous

INTERPRETATION

ACADM c.985A>G (p.Lys329Glu), PATHOGENIC
The homozygous c.985A>G (p.Lys329Glu) missense variant in the ACADM gene (MIM:607008) is an established pathogenic variant. Pathogenic variants in the ACADM gene have been associated with autosomal recessive medium chain acyl-CoA dehydrogenase (MCAD) deficiency (1). This result is supportive of a diagnosis of MCAD deficiency for this individual. Clinical correlation is recommended.

Consultation with a genetics professional is recommended for interpretation of this result and to determine whether reproductive risk assessment and familial testing may be of benefit to this family.

FOLLOW-UP OPTION(S)
Familial Testing:
If relevant, genetic testing for family members is available by ordering FMTT / Familial Mutation, Targeted Testing for the specific variant(s) detected. Please contact the laboratory at 1-800-533-1710 or the online test catalog at www.mayocliniclabs.com for information about FMTT.

REFERENCES:
1) GeneReviews: Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency (https://www.ncbi.nlm.nih.gov/books/NBK1424/) (PMID: 20301597)

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RESOURCES

Disease specific information, including current therapies may be available at:
1. GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1116/
2. https://www.fda.gov/drugs

Information regarding clinical trials, if available, can be found at the following sites:
1. ClinicalTrials.gov: http://clinicaltrials.gov/ct2/search/advanced

ADDITIONAL INFORMATION

A portion of the testing process was performed at Mayo Clinic Laboratories site 189293.

METHOD

Next generation sequencing (NGS) and/or Sanger sequencing was performed to test for the presence of variants in coding regions and intron/exon boundaries of the gene analyzed. The human genome reference GRCh37/hg19 build was used for sequence read alignment. At least 99% of the bases are covered at a read depth >20X. Sensitivity is estimated at >99% for single nucleotide variants, >94% for indels up to 39 base pairs, >95% for deletions up to 75 base pairs and insertions up to 47 base pairs. NGS and/or a PCR-based quantitative method was performed to test for the presence of deletions and duplications in the gene analyzed. See the Genes Analyzed field for a list of gene(s) tested.

There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria. See www.mayocliniclabs.com (TEST ID ACADM) for details regarding genes with regions not routinely covered.

GENES ANALYZED

ACADM

DISCLAIMER

Clinical Correlations
The gene(s) analyzed by this test may have more than one associated phenotype or inheritance pattern. Additionally, the genetic variants detected may have reduced penetrance, variable expressivity, or different classifications based on disease state. Classification and reporting of the variants detected is based on the intended disease state of the test ordered. For more information regarding the phenotypic spectrum which may be involved for a specific gene, see OMIM (www.ncbi.nlm.nih.gov/omim) or GeneReviews (www.genereviews.org).

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the

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recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

If testing was performed because of a clinically significant family history it is often useful to first test an affected family member. Detection of a reportable variant(s) in an affected family member would allow for more informative testing of at risk individuals.

To discuss the availability of further testing options or for assistance in the interpretation of these results, Mayo Clinic Laboratory genetic counselors can be contacted at 1-800-533-1710.

Exome sequencing data is analyzed for the specific gene(s) indicated. If clinically indicated, reflex analysis and interpretation of the whole exome using existing data can be completed by ordering WESPR/Whole Exome Sequencing Panel Reflex, Varies. Note: reflex exome testing cannot be ordered on prenatal specimens.

Technical Limitations
Next generation sequencing may not detect all types of genomic variants. In rare cases, false negative or false positive results may occur. The depth of coverage may be variable for some target regions, but assay performance below the minimum acceptable criteria or for failed regions will be noted. The clinical phenotype observed in this individual and/or family may be due to genetic variants not targeted by this test. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria.

Additionally, low level mosaic variants may not be detected.

This test is validated to detect 95% of deletions up to 75 base pairs (bp) and insertions up to 47 bp. Deletions-insertions (delins) of 40 or more bp, including mobile element insertions, may be less reliably detected than smaller delins.

This analysis targets single and multi-exon deletions/duplications; however, in some instances, single exon resolution cannot be achieved due to isolated reduction in sequence coverage or inherent genomic complexity. Balanced structural rearrangements (such as translocations and inversions) may not be detected.

If the patient has had an allogeneic hematopoietic stem cell transplant or a recent blood transfusion, results may be inaccurate due to the presence of donor DNA. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

This test is not designed to differentiate between somatic and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

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Reclassification of Variants Policy
See www.mayocliniclabs.com (TEST ID ACADM) for information regarding the laboratory's policy for reclassification of variants.

Variant Evaluation
Variant curation is performed using published ACMG-AMP recommendations as a guideline. Other gene-specific guidelines may also be considered. Variants classified as benign or likely benign are not reported.

Results from in silico evaluation tools may change over time and should be interpreted with caution and professional clinical judgment.

TEST CLASSIFICATION
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

RELEASED BY
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