

Variant: *NM_000018.4(ACADVL):c.1077G>A (p.Ala359=)*

Version: 1.0

CA8337934 [↗](#)

541717 (ClinVar) [↗](#)

Gene: ACADVL ([HGNC:37](#))

Condition: very long chain acyl-CoA dehydrogenase deficiency
([MONDO:0008723](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 56d1008c-b09a-4033-91ea-ed20b7c0ed65

Approved on: 2026/1/20

Published on: 2026/1/20

HGVS expressions

NM_000018.4:c.1077G>A

NM_000018.4(ACADVL):c.1077G>A (p.Ala359=)

NC_000017.11:g.7222865G>A

CM000679.2:g.7222865G>A

NC_000017.10:g.7126184G>A

CM000679.1:g.7126184G>A

NC_000017.9:g.7066908G>A

NG_007975.1:g.8032G>A

NG_008391.2:g.2186C>T

ENST00000356839.10:c.1077G>A

ENST00000322910.9:c.*1032G>A

ENST00000350303.9:c.1011G>A

ENST00000356839.9:c.1077G>A

ENST00000543245.6:c.1146G>A

ENST00000578824.5:n.226G>A

ENST00000582379.1:n.461G>A

ENST00000583858.5:c.106G>A

ENST00000585203.6:n.18G>A

NM_000018.3:c.1077G>A

NM_001033859.2:c.1011G>A

NM_001270447.1:c.1146G>A

NM_001270448.1:c.849G>A

NM_001033859.3:c.1011G>A

NM_001270447.2:c.1146G>A

NM_001270448.2:c.849G>A

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Evidence Links **0**

Expert Panel

ACADVL VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen ACADVL Expert Panel
Specifications to the ACMG/AMP Variant Interpretation
Guidelines for ACADVL Version 2.1.0

[↗](#) **Criteria Specification Approval History**

Evidence submitted by expert panel

ACADVL VCEP

The NM_000018.4(ACADVL):c.1077G>A (p.Ala359=) variant is a synonymous (silent) variant is located in the last nucleotide position of exon 10 in ACADVL. Although it is adjacent to the canonical donor site in intron 10 and may potentially generate a cryptic donor gain, it does not meet the ACADVL VCEP’s criteria to be predicted to impact splicing (SpliceAI donor gain 0.45). The highest population minor allele frequency in gnomAD v4.1.0 is 0.000067 in East Asian population, which is lower than the ClinGen ACADVL Variant Curation Expert Panel threshold (<0.001) for PM2_Supporting, meeting this criterion (PM2_Supporting). This variant was detected in at least two individuals during abnormal newborn screening (PMIDs: 26385305, 31737040). In addition, this variant was also identified in homozygous fashion in a patient with abnormal newborn screening (PMID: 27246109). However, without further supporting data, this information is insufficient to be used toward classification. Due to limited evidence, this variant is classified as a variant of uncertain significance for autosomal recessive very long chain acyl-CoA dehydrogenase (VLCAD) deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen ACADVL Variant Curation Expert Panel: PM2_Supporting (ACADVL VCEP specifications version 2; approved May 01, 2025).

Met criteria codes

PM2_Supporting			The highest population minor allele frequency in gnomAD v4.1.0 is 0.000067 in East Asian population, which is lower than the ClinGen ACADVL Variant Curation Expert Panel threshold (<0.001) for PM2_Supporting, meeting this criterion (PM2_Supporting).
----------------	---	---	---

Curation History [↗](#)



 

 

Showing 1 to 1 of 1 rows

--