

Variant: *NM_000018.4(ACADVL):c.1839G>A (p.Arg613=)*

Version: 1.0

CA8338296 [↗](#)

254698 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 83a8c832-a9c6-499c-9556-dc51ea0866e5

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HGVS expressions

NM_000018.4(ACADVL):c.1839G>A (p.Arg613=)

NM_000018.4:c.1839G>A

NC_000017.11:g.7224968G>A

CM000679.2:g.7224968G>A

NC_000017.10:g.7128287G>A

CM000679.1:g.7128287G>A

NC_000017.9:g.7069011G>A

NG_007975.1:g.10135G>A

NG_008391.2:g.83C>T

NG_033038.1:g.14577C>T

ENST00000356839.10:c.1839G>A

ENST00000322910.9:c.*1794G>A

ENST00000350303.9:c.1773G>A

ENST00000356839.9:c.1839G>A

ENST00000542255.6:c.718G>A

ENST00000543245.6:c.1908G>A

ENST00000578033.1:n.264G>A

ENST00000578319.5:n.420G>A

ENST00000578711.1:n.1464G>A

ENST00000578809.5:n.411G>A

ENST00000579425.5:n.955G>A

ENST00000579546.1:c.574G>A

ENST00000583848.5:c.205G>A

ENST00000583850.5:n.610G>A

ENST00000583858.5:c.770G>A

NM_000018.3:c.1839G>A

NM_001033859.2:c.1773G>A

NM_001270447.1:c.1908G>A

NM_001270448.1:c.1611G>A

NM_001033859.3:c.1773G>A

NM_001270447.2:c.1908G>A

NM_001270448.2:c.1611G>A

Likely Benign

Met criteria codes 3

PM2

BP7

BP4

Evidence Links 0

Expert Panel

ACADVL VCEP

Criteria Specification Information

[Criteria Specification:](#) ClinGen ACADVL Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 1

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

ACADVL VCEP

The c.1839G>A p.(Arg613=) variant is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by PhyloP100way (conservation score: 0.95) (BP4, BP7). The highest population minor allele frequency in gnomAD v2.1.1 is 0.008134 in African/African American population, which is lower than the ClinGen ACADVL Variant Curation Expert Panel threshold (<0.001) for PM2_Supporting; however, this is not considered conflicting evidence with BP4 and BP7. To our knowledge, this variant has not been reported in the literature in any individuals with autosomal recessive very long chain acyl-CoA dehydrogenase (VLCAD) deficiency. In summary, this variant meets the criteria to be classified as likely benign 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: BP4, BP7, PM2_Supporting (ACADVL VCEP specifications version 1; approved November 9, 2021).

Met criteria codes

PM2		The highest population minor allele frequency in gnomAD v2.1.1 is 0.008134 in African/African American population, which is lower than the ClinGen ACADVL Variant Curation Expert Panel threshold (<0.001) for PM2_Supporting, meeting this criterion (PM2_Supporting).
BP7		The c.1839G>A p.(Arg613=) variant is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by PhyloP100way (conservation score: 1.046) (BP4, BP7).
BP4		The c.1839G>A p.(Arg613=) variant is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by PhyloP100way (conservation score: 1.046) (BP4, BP7).

Curation History

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