

Variant: NM_000018.4(ACADVL):c.1533G>A (p.Arg511=)

Version: 1.0

CA10587278 [↗](#)

254695 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 09cb4f93-9073-4758-b6c9-1d4517c0e238

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HGVS expressions

NM_000018.4:c.1533G>A

NM_000018.4(ACADVL):c.1533G>A (p.Arg511=)

NC_000017.11:g.7224321G>A

CM000679.2:g.7224321G>A

NC_000017.10:g.7127640G>A

CM000679.1:g.7127640G>A

NC_000017.9:g.7068364G>A

NG_007975.1:g.9488G>A

NG_008391.2:g.730C>T

NG_033038.1:g.15224C>T

ENST00000356839.10:c.1533G>A

ENST00000322910.9:c.*1488G>A

ENST00000350303.9:c.1467G>A

ENST00000356839.9:c.1533G>A

ENST00000542255.6:c.391G>A

ENST00000543245.6:c.1602G>A

ENST00000578319.5:n.28G>A

ENST00000578711.1:n.817G>A

ENST00000578809.5:n.105G>A

ENST00000579391.1:n.141G>A

ENST00000579425.5:n.649G>A

ENST00000579546.1:c.272G>A

ENST00000579894.5:n.320G>A

ENST00000582450.1:n.41G>A

ENST00000583074.5:n.154G>A

ENST00000583850.5:n.308G>A

ENST00000583858.5:c.464G>A

ENST00000585203.6:n.724G>A

NM_000018.3:c.1533G>A

NM_001033859.2:c.1467G>A

NM_001270447.1:c.1602G>A

NM_001270448.1:c.1305G>A

NM_001033859.3:c.1467G>A

NM_001270447.2:c.1602G>A

NM_001270448.2:c.1305G>A

Likely Benign

Met criteria codes 2

BP4BP7

Evidence Links 0

Expert Panel

ACADVL VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

ACADVL VCEP

The c.1533G>A (p.Arg511=) variant is a synonymous (silent) variant that is not predicted by (SpliceAI, MaxEntScn, NNSplice) to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by the 100 vertebrate Basewise Conservation by PhyloP track in the UCSC genome browser (BP4, BP7). This variant is absent from gnomAD v2.1.1; however, this is not considered conflicting evidence with BP4 and BP7. 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 (ACADVL VCEP specifications version 1; approved November 8, 2021)

Met criteria codes

BP4

The results from 3 in silico splicing predictors (SpliceAI, MaxEntScn, NNSplice) support that this variant does not affect splicing (BP4).

BP7

In addition, it occurs at a nucleotide that is not conserved as shown by the 100 vertebrate Basewise Conservation by PhyloP track in the UCSC genome browser (BP4, BP7).

Curation History

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