

Variant: *NM_000018.4:c.1966T>A*

Version: 1.0

CA397726273 [↗](#)

1707703 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 0a906e4f-b71a-403e-8791-eb24efee9601

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HGVS expressions

NM_000018.4:c.1966T>A

NC_000017.11:g.7225095T>A

CM000679.2:g.7225095T>A

NC_000017.10:g.7128414T>A

CM000679.1:g.7128414T>A

NC_000017.9:g.7069138T>A

NG_007975.1:g.10262T>A

NG_033038.1:g.14450A>T

ENST00000356839.10:c.1966T>A

ENST00000322910.9:c.*1921T>A

ENST00000350303.9:c.1900T>A

ENST00000356839.9:c.1966T>A

ENST00000542255.6:c.845T>A

ENST00000543245.6:c.2035T>A

ENST00000578033.1:n.391T>A

ENST00000578319.5:n.547T>A

ENST00000578711.1:n.1591T>A

ENST00000578809.5:n.538T>A

ENST00000579425.5:n.1082T>A

ENST00000583848.5:c.332T>A

ENST00000583850.5:n.737T>A

ENST00000583858.5:c.897T>A

NM_000018.3:c.1966T>A

NM_001033859.2:c.1900T>A

NM_001270447.1:c.2035T>A

NM_001270448.1:c.1738T>A

NM_001033859.3:c.1900T>A

NM_001270447.2:c.2035T>A

NM_001270448.2:c.1738T>A

Uncertain Significance

Met criteria codes **2**

PM4

PM2_Supporting

Not Met criteria codes **3**

PP3

PP4

PM5

Expert Panel

ACADVL VCEP [↗](#)

Criteria Specification Information **!**

Evidence submitted by expert panel

ACADVL VCEP

The c.1966T>A variant in ACADVL is a stop-loss predicted to result in a 32-aa peptide extension at the C-terminus (PM4). This variant has been identified in one individual as a heterozygote identified by abnormal newborn screening results suggestive of very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency (PMID: 30194637). This study also showed reduced VLCAD activity in patient lymphocytes, however, this assay does not meet the requirements for use by the ClinGen ACADVL Variant Curation Expert Panel. This variant is absent from gnomAD v2.1.1 (PM2_Supporting). In summary, this variant meets the criteria to be classified as uncertain 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: (PM4, PM2_Supporting)

Met criteria codes

PM4			Additional 32-aa peptide at the C-terminus via stop-loss variant
PM2_Supporting			Not present in gnomAD

Not Met criteria codes

PP3			Available computational evidence does not provide predictions for this variant.
PP4			Reported as a heterozygote with WT. Residual enzyme activity is ~33%, not meeting PP4_moderate.
PM5			c.1968A>C is at the same codon, but is not LP or P

Curation History [↗](#)



 

 

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