

Variant: *NM_000018.4(ACADVL):c.619T>C (p.Ser207Pro)*

Version: 1.0

CA8337769 [↗](#)

551588 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: a8d50bf8-d177-4bc2-b8b3-245d4c748998

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HGVS expressions

NM_000018.4:c.619T>C

NM_000018.4(ACADVL):c.619T>C (p.Ser207Pro)

NC_000017.11:g.7221679T>C

CM000679.2:g.7221679T>C

NC_000017.10:g.7124998T>C

CM000679.1:g.7124998T>C

NC_000017.9:g.7065722T>C

NG_007975.1:g.6846T>C

NG_008391.2:g.3372A>G

ENST00000356839.10:c.619T>C

ENST00000322910.9:c.*574T>C

ENST00000350303.9:c.553T>C

ENST00000356839.9:c.619T>C

ENST00000543245.6:c.688T>C

ENST00000577191.5:n.696T>C

ENST00000577857.5:n.435T>C

ENST00000579286.5:n.800T>C

ENST00000579886.2:c.457T>C

ENST00000580365.1:n.350T>C

ENST00000581378.5:c.337T>C

ENST00000581562.5:n.525-273T>C

ENST00000583312.5:c.619T>C

ENST00000583760.1:n.401T>C

NM_000018.3:c.619T>C

NM_001033859.2:c.553T>C

NM_001270447.1:c.688T>C

NM_001270448.1:c.391T>C

NM_001033859.3:c.553T>C

NM_001270447.2:c.688T>C

NM_001270448.2:c.391T>C

Likely Pathogenic

Met criteria codes **4**

PP4_Moderate

PM3_Strong

PP3

PM2

Expert Panel

ACADVL VCEP [↗](#)

Criteria Specification Information **!**

Evidence submitted by expert panel

ACADVL VCEP

The NM_000018.4(ACADVL): c.619T>C (p.Ser207Pro) variant is a missense variant predicted to cause substitution of serine by proline at amino acid 207. At least two individuals with this variant displayed a newborn screen consistent with very long chain acyl-CoA dehydrogenase (VLCAD) deficiency as well as follow-up acylcarnitine testing showing increased C14:1 acylcarnitines, which is highly specific for VLCAD deficiency (PP4_Moderate, PMID: 26927351). This individual was compound heterozygous for this variant and a distinct pathogenic or likely pathogenic variant confirmed in trans by parental testing (PM3_Strong). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). The computational predictor REVEL gives a score of 0.79, which is above the threshold of 0.75, evidence that correlates with impact to ACADVL function (PP3). In summary, this variant meets the criteria to be classified as likely pathogenic for autosomal recessive VLCAD deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen ACADVL Variant Curation Expert Panel: PM2_Supporting, PM3_Strong, PP3, PP4_Moderate (ACADVL VCEP specifications version 1; approved November 9, 2021).

Met criteria codes

PP4_Moderate			At least one patient with this variant displayed newborn screen consistent with very long chain acyl-CoA dehydrogenase (VLCAD) deficiency as well as follow-up acylcarnitine testing showing increased C14:1 acylcarnitines, which is highly specific for VLCAD deficiency (PP4_Moderate, PMID: 26927351).
PM3_Strong			Confirmed in trans by parental testing to c.1770_1773del (curated by VCEP as pathogenic; cdot nomenclature confirmed by Sanger sequencing in figure 1)
PP3			The computational predictor REVEL gives a score of 0.79, which is above the threshold of 0.75, evidence that correlates with impact to ACADVL function (PP3).
PM2			This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Curation History [↗](#)



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