

Variant: *NM_000018.4(ACADVL):c.1251del (p.Ser418fs)*

Version: 1.0

CA2499224903 [↗](#)

1073505 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: ed709b5f-bc3d-4070-a616-dfde5be00520

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HGVS expressions

NM_000018.4:c.1251del

NM_000018.4(ACADVL):c.1251del (p.Ser418fs)

NC_000017.11:g.7223712del

CM000679.2:g.7223712del

NC_000017.10:g.7127031del

CM000679.1:g.7127031del

NC_000017.9:g.7067755del

NG_007975.1:g.8879del

NG_008391.2:g.1339del

NG_033038.1:g.15833del

ENST00000356839.10:c.1251del

ENST00000322910.9:c.*1206del

ENST00000350303.9:c.1185del

ENST00000356839.9:c.1251del

ENST00000542255.6:c.109del

ENST00000543245.6:c.1320del

ENST00000578579.2:n.422del

ENST00000578711.1:n.208del

ENST00000578824.5:n.667del

ENST00000579425.5:n.275del

ENST00000579546.1:c.88del

ENST00000583850.5:n.26del

ENST00000583858.5:c.280del

ENST00000585203.6:n.459del

NM_000018.3:c.1251del

NM_001033859.2:c.1185del

NM_001270447.1:c.1320del

NM_001270448.1:c.1023del

NM_001033859.3:c.1185del

NM_001270447.2:c.1320del

NM_001270448.2:c.1023del

Likely Pathogenic

Met criteria codes **2**

PVS1

PM2_Supporting

Expert Panel

ACADVL VCEP [↗](#)

Evidence submitted by expert panel

ACADVL VCEP

The c.1251del (p.Ser418fs) also known as (p.Ser418AlafsTer12) variant in ACADVL is a frameshift variant predicted to cause a premature stop codon in biologically-relevant-exon 12/20 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1; PMIDs 9973285, 11590124). To our knowledge, this variant has not been reported in the literature in any individuals with very long chain acyl-CoA dehydrogenase (VLCAD) deficiency. Additionally, to our knowledge, functional assays have not been reported for this variant. This variant is absent from gnomAD v2.1.1 (PM2_Supporting). 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: PVS1, PM2_Supporting (VCEP specifications version1; approved November 8, 2021)

Met criteria codes

PVS1			The c.1251del (p.Ser418AlafsTer12) variant in ACADVL is a frameshift variant predicted to cause a premature stop codon in biologically-relevant-exon 12/20 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1; PMIDs 9973285, 11590124).
PM2_Supporting			This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Curation History [↗](#)



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