

*Variant: NM_000018.4(ACADVL):c.1730_1740del
(p.Ala577fs)*

Version: 1.0

CA16043045 [↗](#)

373427 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 15da60a7-a687-4f0f-a077-919299116b68

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HGVS expressions

NM_000018.4:c.1730_1740del

NM_000018.4(ACADVL):c.1730_1740del (p.Ala577fs)

NC_000017.11:g.7224693_7224703del

CM000679.2:g.7224693_7224703del

NC_000017.10:g.7128012_7128022del

CM000679.1:g.7128012_7128022del

NC_000017.9:g.7068736_7068746del

NG_007975.1:g.9860_9870del

NG_008391.2:g.350_360del

NG_033038.1:g.14844_14854del

ENST00000356839.10:c.1730_1740del

ENST00000322910.9:c.*1685_*1695del

ENST00000350303.9:c.1664_1674del

ENST00000356839.9:c.1730_1740del

ENST00000542255.6:c.537-22_537-12del

ENST00000543245.6:c.1799_1809del

ENST00000578033.1:n.61_71del

ENST00000578319.5:n.311_321del

ENST00000578711.1:n.1189_1199del

ENST00000578809.5:n.302_312del

ENST00000579425.5:n.846_856del

ENST00000579546.1:c.465_475del

ENST00000583074.5:n.300-22_300-12del

ENST00000583848.5:c.96_106del

ENST00000583850.5:n.501_511del

ENST00000583858.5:c.661_671del

ENST00000585203.6:n.921_931del

NM_000018.3:c.1730_1740del

NM_001033859.2:c.1664_1674del

NM_001270447.1:c.1799_1809del

NM_001270448.1:c.1502_1512del

NM_001033859.3:c.1664_1674del

NM_001270447.2:c.1799_1809del

NM_001270448.2:c.1502_1512del

Likely Pathogenic

Met criteria codes 2

PVS1PM2_Supporting

Evidence Links 0

Expert Panel

ACADVL VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

ACADVL VCEP

The NM_000018.4(ACADVL): c.1730_1740del (p.Ala577Glyfs*11) variant in ACADVL is a frameshift variant predicted to cause a premature stop codon in biologically-relevant-exon 18 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1; PMIDs 9973285, 11590124). 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 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: PVS1, PM2_Supporting.

Met criteria codes

PVS1

The NM_000018.4(ACADVL): c.1730_1740del (p.Ala577Glyfs*11) variant in ACADVL is a frameshift variant predicted to cause a premature stop codon in biologically-relevant-exon 18 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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