

Variant: NM_000018.4(ACADVL):c.1808del (p.Cys603fs)

Version: 1.0

CA2499224908 [↗](#)

1075156 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 58415c85-72ce-40b5-a9cf-d3fe0437986c

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HGVS expressions

NM_000018.4(ACADVL):c.1808del (p.Cys603fs)

NM_000018.4:c.1808del

NC_000017.11:g.7224865del

CM000679.2:g.7224865del

NC_000017.10:g.7128184del

CM000679.1:g.7128184del

NC_000017.9:g.7068908del

NG_007975.1:g.10032del

NG_008391.2:g.186del

NG_033038.1:g.14680del

ENST00000356839.10:c.1808del

ENST00000322910.9:c.*1763del

ENST00000350303.9:c.1742del

ENST00000356839.9:c.1808del

ENST00000542255.6:c.687del

ENST00000543245.6:c.1877del

ENST00000578033.1:n.233del

ENST00000578319.5:n.389del

ENST00000578711.1:n.1361del

ENST00000578809.5:n.380del

ENST00000579425.5:n.924del

ENST00000579546.1:c.543del

ENST00000583848.5:c.174del

ENST00000583850.5:n.579del

ENST00000583858.5:c.739del

NM_000018.3:c.1808del

NM_001033859.2:c.1742del

NM_001270447.1:c.1877del

NM_001270448.1:c.1580del

NM_001033859.3:c.1742del

NM_001270447.2:c.1877del

NM_001270448.2:c.1580del

Uncertain Significance

Met criteria codes 2

PVS1_Moderate

PM2_Supporting

Evidence Links 0

Expert Panel

ACADVL VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

ACADVL VCEP

The c.1808del (p.Cys603fs) variant in ACADVL, also known as p.Cys603LeufsTer77, is a frameshift variant that may cause loss of function of the protein; however it is predicted to escape nonsense mediated decay and remove <10% of the protein (PVS1_Moderate). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). Due to limited evidence, this variant is classified as a variant of uncertain significance 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_Moderate, PM2_Supporting (ACADVL VCEP specifications version 1; approved November 8, 2021).

Met criteria codes

PVS1_Moderate

The c.1808del (p.Cys603fs) variant in ACADVL, also known as p.Cys603LeufsTer77, is a frameshift variant that may cause loss of function of the protein; however it is predicted to escape nonsense mediated decay and remove <10% of the protein (PVS1_Moderate).

PM2_Supporting

This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Curation History

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