

Variant: *NM_000018.4:c.632_633del*

Version: 1.0

CA624860663 [↗](#)

934787 (ClinVar) [↗](#)

Gene: [ACADVL](#)

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

Inheritance Mode: Autosomal recessive inheritance

UUID: fe32806f-b34b-4751-b60e-7b95e8f83b26

Approved on: 2022/6/4

Published on: 2022/6/4

HGVS expressions

NM_000018.4:c.632_633del

NC_000017.11:g.7221961_7221962del
CM000679.2:g.7221961_7221962del
NC_000017.10:g.7125280_7125281del
CM000679.1:g.7125280_7125281del
NC_000017.9:g.7066004_7066005del
NG_007975.1:g.7128_7129del
NG_008391.2:g.3091_3092del
ENST00000356839.10:c.632_633del
ENST00000322910.9:c.*587_*588del
ENST00000350303.9:c.566_567del
ENST00000356839.9:c.632_633del
ENST00000543245.6:c.701_702del
ENST00000577191.5:n.709_710del
ENST00000577857.5:n.448_449del
ENST00000579286.5:n.813_814del
ENST00000580365.1:n.363_364del
ENST00000581378.5:c.350_351del
ENST00000581562.5:n.534_535del
ENST00000582379.1:n.16_17del
ENST00000583312.5:c.647_648del
ENST00000583760.1:n.414_415del
NM_000018.3:c.632_633del
NM_001033859.2:c.566_567del
NM_001270447.1:c.701_702del
NM_001270448.1:c.404_405del
NM_001033859.3:c.566_567del
NM_001270447.2:c.701_702del
NM_001270448.2:c.404_405del

Likely Pathogenic

Met criteria codes **2**

PM2_Supporting PVS1

Evidence Links **0**

Expert Panel

ACADVL VCEP [↗](#)

Criteria Specification Information **!**

Evidence submitted by expert panel

ACADVL VCEP

The c.632_633del (p.Val211GlyfsTer41)variant in ACADVL is a frameshift variant predicted to cause a premature stop codon in biologically-relevant-exon 9/20 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). To our knowledge, this variant has not been reported in the literature in patients with ACADVL related disease or in functional studies. The ACADVL Variant Curation Expert Panel VCEP classified the variant as likely pathogenic based on PVS1+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: PMS2_Supporting, PVS1. (Required: VCEP specifications version 2.0; 1/14/22.)

Met criteria codes

PM2_Supporting			This variant is absent from gnomAD v2.1.1 (PM2_Supporting).
PVS1			The c.632_633del (p.Val211GlyfsTer41)variant in ACADVL is a frameshift variant predicted to cause a premature stop codon in biologically-relevant-exon 9/20 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1; PMIDs 9973285, 11590124).

Curation History [↗](#)

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