

Variant: *NM_000018.4(ACADVL):c.797_798del (p.Pro266fs)*

Version: 1.0

CA1139665143 [↗](#)

932845 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: b0b0b1e6-86e9-4a5c-b8b9-d5a64083b3fb

Approved on: 2023/1/10

Published on: 2023/1/10

HGVS expressions

NM_000018.4(ACADVL):c.797_798del (p.Pro266fs)

NM_000018.4:c.797_798del

NC_000017.11:g.7222221_7222222del

CM000679.2:g.7222221_7222222del

NC_000017.10:g.7125540_7125541del

CM000679.1:g.7125540_7125541del

NC_000017.9:g.7066264_7066265del

NG_007975.1:g.7388_7389del

NG_008391.2:g.2829_2830del

ENST00000356839.10:c.797_798del

ENST00000322910.9:c.*752_*753del

ENST00000350303.9:c.731_732del

ENST00000356839.9:c.797_798del

ENST00000543245.6:c.866_867del

ENST00000577191.5:n.969_970del

ENST00000581378.5:c.515_516del

ENST00000582379.1:n.181_182del

NM_000018.3:c.797_798del

NM_001033859.2:c.731_732del

NM_001270447.1:c.866_867del

NM_001270448.1:c.569_570del

NM_001033859.3:c.731_732del

NM_001270447.2:c.866_867del

NM_001270448.2:c.569_570del

Likely Pathogenic

Met criteria codes 2

PVS1

PM2_Supporting

Not Met criteria codes 1

PP4

Evidence Links 0

Expert Panel

ACADVL VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

ACADVL VCEP

The NM_000018.4: c.797_798del (p.Pro266ArgfsTer31) 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). At least one individual with this variant was identified by newborn screen, but this information is insufficient to use toward classification (PMID: 26385305). 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 (ACADVL VCEP specifications version 1; approved November 8, 2021).

Met criteria codes

PVS1			The NM_000018.4: c.797_798del (p.Pro266ArgfsTer31) 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).
PM2_Supporting			This variant is absent from gnomAD v2.1.1 (PM2_supporting).

Not Met criteria codes

PP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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Curation History



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