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Variant: NM_000018.4(ACADVL):c.343G>T (p.Glu115Ter)

Version: 1.1

CA397722699

932735 (ClinVar)

Gene: ACADVL (HGNC:37)
Condition: very long chain acyl-CoA dehydrogenase deficiency (MONDO:0008723)
Inheritance Mode: Autosomal recessive inheritance
UUID: b9dc6fa8-3d91-4c82-9bcd-b5e8f5b06807
Approved on: 2024/2/27
Published on: 2024/2/27

HGVS expressions

NM_000018.4:c.343G>T
NM_000018.4(ACADVL):c.343G>T (p.Glu115Ter)
NC_000017.11:g.7220924G>T
CM000679.2:g.7220924G>T
NC_000017.10:g.7124243G>T
CM000679.1:g.7124243G>T
NC_000017.9:g.7064967G>T
NG_007975.1:g.6091G>T
NG_008391.2:g.4127C>A
ENST00000356839.10:c.343G>T
ENST00000322910.9:c.*298G>T
ENST00000350303.9:c.277G>T
ENST00000356839.9:c.343G>T
ENST00000543245.6:c.412G>T
ENST00000577191.5:n.420G>T
ENST00000577433.5:n.551G>T
ENST00000577857.5:n.293+94G>T
ENST00000579286.5:n.524G>T
ENST00000579886.2:c.202-21G>T
ENST00000580365.1:n.74G>T
ENST00000581378.5:c.42G>T
ENST00000581562.5:n.390G>T
ENST00000582056.5:n.526G>T
ENST00000582166.1:n.324G>T
ENST00000583312.5:c.343G>T
ENST00000584103.5:c.376G>T
NM_000018.3:c.343G>T
NM_001033859.2:c.277G>T
NM_001270447.1:c.412G>T
NM_001270448.1:c.115G>T
NM_001033859.3:c.277G>T
NM_001270447.2:c.412G>T
NM_001270448.2:c.115G>T

Met criteria codes 2

Expert Panel

Likely Pathogenic

PVS1 PM2_Supporting

Not Met criteria codes 1

PP4

Evidence Links 0

ACADVL VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

ACADVL VCEP

The NM_000018.4(ACADVL): c.343G>T (p.Glu115Ter) variant in ACADVL is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 6/20 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 any individuals with VLCADD. The ACADVL Variant Curation Expert Panel VCEP classified the variant as likely pathogenic based on PVS1+PM2_supporting (ACADVL VCEP specifications version 1; approved November 9, 2021). This variant was originally curated November 16, 2021 and the recurated classification was approved by the expert panel on February 27, 2024.

Met criteria codes

PVS1	✔	Truncating exon 6/20
PM2_Supporting	✔	Absent from gnomAD

Not Met criteria codes

PP4	✖	Not found in the literature

Curation History

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