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Variant: NM_000018.4(ACADVL):c.419G>A (p.Gly140Glu)

Version: 2.0

CA397722868

840694 (ClinVar)

Gene: ACADVL (HGNC:37)
Condition: very long chain acyl-CoA dehydrogenase deficiency (MONDO:0008723)
Inheritance Mode: Autosomal recessive inheritance
UUID: 276f014b-c151-4bf6-9719-f4b3c95db493
Approved on: 2023/4/11
Published on: 2023/4/11

HGVS expressions

NM_000018.4(ACADVL):c.419G>A (p.Gly140Glu)
NM_000018.4:c.419G>A
NC_000017.11:g.7221000G>A
CM000679.2:g.7221000G>A
NC_000017.10:g.7124319G>A
CM000679.1:g.7124319G>A
NC_000017.9:g.7065043G>A
NG_007975.1:g.6167G>A
NG_008391.2:g.4051C>T
ENST00000356839.10:c.419G>A
ENST00000322910.9:c.*374G>A
ENST00000350303.9:c.353G>A
ENST00000356839.9:c.419G>A
ENST00000543245.6:c.488G>A
ENST00000577191.5:n.496G>A
ENST00000577433.5:n.627G>A
ENST00000577857.5:n.293+170G>A
ENST00000579286.5:n.600G>A
ENST00000579886.2:c.257G>A
ENST00000580365.1:n.150G>A
ENST00000581378.5:c.118G>A
ENST00000581562.5:n.466G>A
ENST00000582056.5:n.602G>A
ENST00000582166.1:n.400G>A
ENST00000583312.5:c.419G>A
ENST00000584103.5:c.452G>A
NM_000018.3:c.419G>A
NM_001033859.2:c.353G>A
NM_001270447.1:c.488G>A
NM_001270448.1:c.191G>A
NM_001033859.3:c.353G>A
NM_001270447.2:c.488G>A
NM_001270448.2:c.191G>A

Met criteria codes 4

Expert Panel

Likely Pathogenic

PP3 PM3 PP4_Moderate PM2_Supporting

Evidence Links 0

ACADVL VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

ACADVL VCEP

The c.419G>A (p.Gly140Glu) variant in ACADVL is a missense in exon 6. This variant has been described in one reportedly affected individual, but this individual does not meet our guidelines for consideration of phenotypic or genotypic data (PMID: 15210884). However, information provided to the ACADVL VCEP by an external clinical lab shows increased C14:1 level in a pattern consistent with very long chain acyl-CoA dehydrogenase (VLCAD) in at least two individuals (PP4_moderate). These individuals also had an additional likely pathogenic variant detected confirmed in trans (PM3). This variant is absent from gnomAD population database v2.1.1 (PM2_supporting), and the computational predictor REVEL gives a score of 0.96, which is above the threshold of 0.75, evidence that correlates with impact to ACADVL function (PP3). 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: PM2_supporting, PM3, PP3, PP4_moderate (ACADVL VCEP specifications version 1; approved November 8, 2021). This variant was originally curated July 12, 2022 and the recurated classification was approved by the expert panel on April 11, 2023.

Met criteria codes

PP3	✔	REVEL 0.963 >0.75
PM3	✔	Information provided the ACADVL VCEP shows a likely pathogenic variant confirmed in trans to this variant.
PP4_Moderate	✔	Information provided to the ACADVL VCEP provided by an external clinical laboratory shows increased C14:1 level in a pattern consistent with VLCAD deficiency in two patients (PP4_moderate).
PM2_Supporting	✔	Absent from gnomAD

Curation History

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