The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000018.4(ACADVL):c.1313G>A (p.Gly438Glu)

Version: 1.0

CA397724846

474882 (ClinVar)

Gene: ACADVL (HGNC:37)
Condition: very long chain acyl-CoA dehydrogenase deficiency (MONDO:0008723)
Inheritance Mode: Autosomal recessive inheritance
UUID: 6a3a4846-0d01-4661-99be-a27b80924de2
Approved on: 2025/4/16
Published on: 2025/4/16

HGVS expressions

NM_000018.4(ACADVL):c.1313G>A (p.Gly438Glu)
NM_000018.4:c.1313G>A
NC_000017.11:g.7223856G>A
CM000679.2:g.7223856G>A
NC_000017.10:g.7127175G>A
CM000679.1:g.7127175G>A
NC_000017.9:g.7067899G>A
NG_007975.1:g.9023G>A
NG_008391.2:g.1195C>T
NG_033038.1:g.15689C>T
ENST00000356839.10:c.1313G>A
ENST00000322910.9:c.*1268G>A
ENST00000350303.9:c.1247G>A
ENST00000356839.9:c.1313G>A
ENST00000542255.6:c.171G>A
ENST00000543245.6:c.1382G>A
ENST00000578711.1:n.352G>A
ENST00000579425.5:n.337G>A
ENST00000579546.1:c.150G>A
ENST00000583074.5:n.32G>A
ENST00000583850.5:n.88G>A
ENST00000583858.5:c.342G>A
ENST00000585203.6:n.521G>A
NM_000018.3:c.1313G>A
NM_001033859.2:c.1247G>A
NM_001270447.1:c.1382G>A
NM_001270448.1:c.1085G>A
NM_001033859.3:c.1247G>A
NM_001270447.2:c.1382G>A
NM_001270448.2:c.1085G>A

Pathogenic

Met criteria codes 6

PP3_Strong PM3_Supporting PM2_Strong PP1_Moderate PM1_Supporting PP2_Moderate

Expert Panel

ACADVL VCEP

Criteria Specification Information

Evidence Links 0

Criteria Specifications for this VCEP

Evidence submitted by expert panel

ACADVL VCEP

Met criteria codes

PP3_Strong	✔	Multiple insilico prediction tools predicts the variant to be pathogenic like Revel, VARITY, AlphaMissense, SIFT, PrimateAI etc
PM3_Supporting	✔	Invitae has reported in ClinVar that this variant has been detected in trans to a pathogenic variant. However, publication details not given
PM2_Strong	✔	Not reported in any population databases (GnomAD, 1000Genomes or any other country specific databases)
PP1_Moderate	✔	This variant has been reported in homozygous state in a Saudi family in a 2 months old female baby with an early-onset severe phenotype with cardiomyopathy and died at 3 months with multi-organ failure. Her brother was further screened and same variant detected in the brother in homozygous state with mild phenotypes like. He is currently 3 years old and have mild phenotype with recurrent habdomyolysis associated mostly with viral infections. He has no hypoglycemia, hepatomegaly or cardiomyopathy.
PM1_Supporting	✔	This variant is located in exon 13 which is in the catalytic domain of the protein. This region is a hotspot for mutations in the gene. In between codon 430-445, there are 5 pathogenic and 6 likely pathogenic variants are reported in ClinVar
PP2_Moderate	✔	Out of 700 missense mutations reported in ClinVar only 8 are reported as benign/likely benign.

Curation History

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