

Variant: *NM_000018.4(ACADVL):c.1038G>A (p.Ala346=)*

Version: 1.0

CA145601 [↗](#)

21012 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 13e9aeca-373a-462d-91a4-ae936c164558

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HGVS expressions

NM_000018.4:c.1038G>A

NM_000018.4(ACADVL):c.1038G>A (p.Ala346=)

NC_000017.11:g.7222826G>A

CM000679.2:g.7222826G>A

NC_000017.10:g.7126145G>A

CM000679.1:g.7126145G>A

NC_000017.9:g.7066869G>A

NG_007975.1:g.7993G>A

NG_008391.2:g.2225C>T

ENST00000356839.10:c.1038G>A

ENST00000322910.9:c.*993G>A

ENST00000350303.9:c.972G>A

ENST00000356839.9:c.1038G>A

ENST00000543245.6:c.1107G>A

ENST00000578824.5:n.187G>A

ENST00000582379.1:n.422G>A

ENST00000583858.5:c.67G>A

NM_000018.3:c.1038G>A

NM_001033859.2:c.972G>A

NM_001270447.1:c.1107G>A

NM_001270448.1:c.810G>A

NM_001033859.3:c.972G>A

NM_001270447.2:c.1107G>A

NM_001270448.2:c.810G>A

Benign

Met criteria codes **3**

BP7 BP4 BA1

Evidence Links **0**

Expert Panel

ACADVL VCEP [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

ACADVL VCEP

The c.1038G>A variant in ACADVL is a synonymous variant which occurs in exon 10. The highest population minor allele frequency in gnomAD v2.1.1 is 0.08089 in the African/African American population, which is higher than the ClinGen ACADVL Variant Curation Expert Panel threshold (≥ 0.007) for BA1, and therefore meets this criterion (BA1). The results from 2 in silico splicing predictors (SpliceAI, MutationTaster) support that this variant does not affect splicing. In addition, it occurs at a nucleotide that is not conserved as shown by the 100 vertebrate Basewise Conservation by PhyloP track in the UCSC genome browser (BP4, BP7). In summary, this variant meets the criteria to be classified as benign 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: BA1, BP4, BP7 (VCEP specifications v2.0, approved on 09/16/2021).

Met criteria codes

BP7			Scores for SpliceAI were all below 0.2 (0.00, 0.01, 0.01, 0.06). No change was seen when a 45 bp region centered on the variant was tested with NNSplice. The nucleotide is not conserved as judged by the 100 vertebrate Basewise Conservation by PhyloP track in the UCSC genome browser
BP4			SpliceAI and NNSplice both predict no change in splicing
BA1			Allele frequency in African population 0.08089 in gnomAD

Curation History



 

 

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