

Variant: *NM_000018.4(ACADVL):c.1022G>C (p.Arg341Thr)*

Version: 1.0

CA16620600 [↗](#)

418711 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 8c1e2ecf-8b0b-4a6f-a1f3-c4aa0c798e8f

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HGVS expressions

NM_000018.4(ACADVL):c.1022G>C (p.Arg341Thr)

NM_000018.4:c.1022G>C

NC_000017.11:g.7222810G>C

CM000679.2:g.7222810G>C

NC_000017.10:g.7126129G>C

CM000679.1:g.7126129G>C

NC_000017.9:g.7066853G>C

NG_007975.1:g.7977G>C

NG_008391.2:g.2241C>G

ENST00000356839.10:c.1022G>C

ENST00000322910.9:c.*977G>C

ENST00000350303.9:c.956G>C

ENST00000356839.9:c.1022G>C

ENST00000543245.6:c.1091G>C

ENST00000578824.5:n.171G>C

ENST00000581378.5:c.740G>C

ENST00000582379.1:n.406G>C

ENST00000583858.5:c.51G>C

NM_000018.3:c.1022G>C

NM_001033859.2:c.956G>C

NM_001270447.1:c.1091G>C

NM_001270448.1:c.794G>C

NM_001033859.3:c.956G>C

NM_001270447.2:c.1091G>C

NM_001270448.2:c.794G>C

Uncertain Significance

Met criteria codes **2**

PP3

PM2_Supporting

Evidence Links **0**

Expert Panel

ACADVL VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

ACADVL VCEP

The c.1022G>C (NM_000018.4) variant in ACADVL is a missense variant predicted to cause substitution of arginine by threonine at amino acid 341 (p.Arg341Thr). To our knowledge, this variant is not reported in the medical literature. This variant is absent from gnomAD v2.1.1 (PM2_Supporting). The computational predictor REVEL gives a score of 0.971, 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 variant of uncertain significance 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: PM2_Supporting, PP3 (ACADVL VCEP specifications version 1; approved November 9, 2021).

Met criteria codes

PP3			The computational predictor REVEL gives a score of 0.971, which is above the threshold of 0.75, evidence that correlates with impact to ACADVL function (PP3).
PM2_Supporting			This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Curation History



 

 

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