

Variant: *NM_000018.4(ACADVL):c.663C>T (p.Ser221=)*

Version: 1.0

CA8337806 [↗](#)

383052 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 8dc05298-3c59-48f9-8cad-699747cf0849

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HGVS expressions

NM_000018.4(ACADVL):c.663C>T (p.Ser221=)

NM_000018.4:c.663C>T

NC_000017.11:g.7221992C>T

CM000679.2:g.7221992C>T

NC_000017.10:g.7125311C>T

CM000679.1:g.7125311C>T

NC_000017.9:g.7066035C>T

NG_007975.1:g.7159C>T

NG_008391.2:g.3059G>A

ENST00000356839.10:c.663C>T

ENST00000322910.9:c.*618C>T

ENST00000350303.9:c.597C>T

ENST00000356839.9:c.663C>T

ENST00000543245.6:c.732C>T

ENST00000577191.5:n.740C>T

ENST00000577857.5:n.479C>T

ENST00000579286.5:n.844C>T

ENST00000580365.1:n.394C>T

ENST00000581378.5:c.381C>T

ENST00000581562.5:n.565C>T

ENST00000582379.1:n.47C>T

ENST00000583760.1:n.445C>T

NM_000018.3:c.663C>T

NM_001033859.2:c.597C>T

NM_001270447.1:c.732C>T

NM_001270448.1:c.435C>T

NM_001033859.3:c.597C>T

NM_001270447.2:c.732C>T

NM_001270448.2:c.435C>T

Likely Benign

Met criteria codes **3**

BP7

BP4

BS1

Evidence Links **0**

Expert Panel

ACADVL VCEP [↗](#)

Criteria Specification Information **!**

Evidence submitted by expert panel

ACADVL VCEP

The NM_000018.4(ACADVL):c.663C>T (p.Ser221=) variant is a synonymous (silent) variant that is not predicted by SpliceAI, MaxEntScn and NNSplice to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by phyloP (BP4, BP7). The highest population minor allele frequency in gnomAD v2.1.1 is 0.00353 in African/African American population, which is higher than the ClinGen ACADVL Variant Curation Expert Panel threshold (≥ 0.0035) for BS1, and therefore meets this criterion (BS1). In summary, this variant meets the criteria to be classified as likely 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: BS1, BP4, BP7.

Met criteria codes

BP7			The NM_000018.4(ACADVL):c.663C>T (p.Ser221=) variant is a synonymous (silent) variant that is not predicted by SpliceAI, MaxEntScn and NNSplice to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by phyloP (BP4, BP7).
BP4			The NM_000018.4(ACADVL):c.663C>T (p.Ser221=) variant is a synonymous (silent) variant that is not predicted by SpliceAI, MaxEntScn and NNSplice to impact splicing. In addition, it occurs at a nucleotide that is not conserved as shown by phyloP (BP4, BP7).
BS1			The highest population minor allele frequency in gnomAD v2.1.1 is 0.00353 in African/African American population, which is higher than the ClinGen ACADVL Variant Curation Expert Panel threshold (≥ 0.0035) for BS1, and therefore meets this criterion (BS1).

Curation History [↗](#)



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Showing 1 to 1 of 1 rows

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