

Variant: *NM_000033.4(ABCD1):c.21C>T (p.Pro7=)*

Version: 1.0

CA519210960 [↗](#)

1135384 (ClinVar) [↗](#)

Gene: ABCD1 ([HGNC:215](#))

Condition: adrenoleukodystrophy ([MONDO:0018544](#))

Inheritance Mode: X-linked inheritance (recessive (HP:0001419))

UUID: e0a3a529-4118-4672-a494-cb50bb2c6202

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HGVS expressions

NM_000033.4:c.21C>T

NM_000033.4(ABCD1):c.21C>T (p.Pro7=)

NC_000023.11:g.153725287C>T

CM000685.2:g.153725287C>T

NC_000023.10:g.152990742C>T

CM000685.1:g.152990742C>T

NC_000023.9:g.152643936C>T

NG_009022.2:g.5420C>T

NG_023231.1:g.4460G>A

ENST00000218104.6:c.21C>T

ENST00000218104.5:c.21C>T

NM_000033.3:c.21C>T

Likely Benign

Met criteria codes **3**

BP7

BP4

PM2_Supporting

Evidence Links **0**

Expert Panel

Peroxisomal Disorders VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Peroxisomal Disorders
Expert Panel Specifications to the ACMG/AMP Variant
Interpretation Guidelines for ABCD1 Version 1.0.0

[↗](#) **Criteria Specification Approval History**

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

Evidence submitted by expert panel

Peroxisomal Disorders VCEP

The c.21C>T (p.Pro7=) variant is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing (score of 0.00 for donor/acceptor loss) (BP4, BP7). This variant is absent from gnomAD v2.1.1, v3.1.2, and v4.0.0 (PM2_Supporting). In summary, the Peroxisomal Disorders VCEP classified the variant as likely benign using the ACMG criteria applied, as specified by the ClinGen Peroxisomal Disorders VCEP: PM2_Supporting, BP4, BP7 (ClinGen Peroxisomal Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ABCD1 Version 1.0.0)

Met criteria codes

BP7			The c.21C>T (p.Pro7=) variant is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing (score of 0.00 for donor/acceptor loss). Conservation is not considered in the BP7 code per the Walker et al guidance.
BP4			The computational splicing predictor SpliceAI gives a score of 0.0 for donor/acceptor loss suggesting that the variant has no impact on splicing (BP4).
PM2_Supporting			This variant is absent from gnomAD v2.1.1, v3.1.2, and v4.0.0 (PM2_Supporting).

Curation History



 

 

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