

Variant: NM_000033.4(ABCD1):c.54G>A (p.Thr18=)

Version: 1.0

CA10549902 [↗](#)

444828 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: f60baeb0-e0a6-4c3b-96aa-5a7d6929c12c

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HGVS expressions

NM_000033.4(ABCD1):c.54G>A (p.Thr18=)

NM_000033.4:c.54G>A

NC_000023.11:g.153725320G>A

CM000685.2:g.153725320G>A

NC_000023.10:g.152990775G>A

CM000685.1:g.152990775G>A

NC_000023.9:g.152643969G>A

NG_009022.2:g.5453G>A

NG_023231.1:g.4427C>T

ENST00000218104.6:c.54G>A

ENST00000218104.5:c.54G>A

NM_000033.3:c.54G>A

Likely Benign

Met criteria codes **3**

BS1

BP7

BP4

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.54G>A variant (NM_000033.4) is a synonymous (silent) variant (p.Thr18=) that is not predicted by SpliceAI to impact splicing. The filtering allele frequency (the lower threshold of the 95% CI of 41/1036386) of the c.55G>T (p.Ala19Ser) variant in ABCD1 is 0.00002617 for non-Finnish European exomes by gnomAD v4.1, which is higher than the ClinGen Peroxisomal Disorders VCEP threshold (≥ 0.000017) for BS1, and therefore meets this criterion (BS1). 10 hemizygotes are noted in gnomAD v4.1 for this variant. The computational splicing predictor SpliceAI gives a score of 0 for donor and acceptor loss and gain, suggesting that the variant has no impact on splicing. The exonic variant in exon 1 is not located in the first or last three bases of the exon. (BP4, BP7). In summary, this variant meets the criteria to

be classified as likely benign for X-linked ABCD1 deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen Peroxisomal Disorders VCEP: BS1, BP4, BP7. (ClinGen Peroxisomal Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ABCD1 Version 1.0.0)

Met criteria codes

BS1			The filtering allele frequency (the lower threshold of the 95% CI of 41/1036386) of the c.55G>T (p.Ala19Ser) variant in ABCD1 is 0.00002617 for non-Finnish European exomes by gnomAD v4.1, which is higher than the ClinGen Peroxisomal Disorders VCEP threshold (≥ 0.000017) for BS1, and therefore meets this criterion (BS1). 10 hemizygotes are noted in gnomAD v4.1 for this variant.
BP7			The computational splicing predictor SpliceAI gives a score of 0 for donor and acceptor loss and gain, suggesting that the variant has no impact on splicing. The exonic variant in exon 1 is not located in the first or last three bases of the exon. (BP7)
BP4			The computational splicing predictor SpliceAI gives a score of 0 for donor and acceptor loss and gain, suggesting that the variant has no impact on splicing (BP4)

Curation History



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