

Variant: *NM_000033.4(ABCD1):c.55G>T (p.Ala19Ser)*

Version: 1.0

CA337233489 [↗](#)

1329074 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 9f04e4c4-f0d8-4e61-a41e-e9ee7032453b

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HGVS expressions

NM_000033.4:c.55G>T

NM_000033.4(ABCD1):c.55G>T (p.Ala19Ser)

NC_000023.11:g.153725321G>T

CM000685.2:g.153725321G>T

NC_000023.10:g.152990776G>T

CM000685.1:g.152990776G>T

NC_000023.9:g.152643970G>T

NG_009022.2:g.5454G>T

NG_023231.1:g.4426C>A

ENST00000218104.6:c.55G>T

ENST00000218104.5:c.55G>T

NM_000033.3:c.55G>T

Likely Benign

Met criteria codes **2**

BS3_Supporting

BS1

Not Met criteria codes **1**

BP4

Evidence Links **1**

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.55G>T (NM_000033.4) variant in ABCD1 is a missense variant predicted to cause substitution of alanine by serine at amino acid position 19 (p.Ala19Ser). The filtering allele frequency (the lower threshold of the 95% CI of 8/1038015) of the c.55G>T (p.Ala19Ser) variant in ABCD1 is 0.00007991 for African/African American 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). 4 hemizygotes are noted in gnomAD v4.1 for this variant. Beta-oxidation activity assay in X-ALD fibroblasts showed 100% of wild-type levels indicating that this variant does not impact protein function (PMID: 23300730)(BS3_Supporting). The computational predictor REVEL gives a score of 0.612, which is neither above nor

below the thresholds predicting a damaging (≥ 0.85) or benign (≤ 0.5) impact on ABCD1 function. In summary, this variant meets the criteria to be classified as likely benign for X-linked adrenoleukodystrophy based on the ACMG/AMP criteria applied, as specified by the ClinGen Peroxisomal Disorders VCEP: BS1. BS3_Supporting. (ClinGen Peroxisomal Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ABCD1 Version 1.0.0)

Met criteria codes

BS3_Supporting			Beta-oxidation activity assay in X-ALD fibroblasts showed 100% of wild-type levels indicating that this variant does not impact protein function (PMID: 23300730)(BS3_Supporting).
Authors generated a constructed carrying the Ala19Ser substitution and transfected X-ALD fibroblasts, after which ALDP was detected by Western blotting and shown to be similar to WT (Figure 1) and beta-oxidation was restored to 100% of WT levels (inferred from Figure 2) PubMed:23300730 			
BS1			The filtering allele frequency (the lower threshold of the 95% CI of 8/1038015) of the c.55G>T (p.Ala19Ser) variant in ABCD1 is 0.00007991 for African/African American 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). 4 hemizygotes are noted in gnomAD v4.1 for this variant.

Not Met criteria codes

BP4			The computational predictor REVEL gives a score of 0.612, which is neither above nor below the thresholds predicting a damaging (≥ 0.85) or benign (≤ 0.5) impact on ABCD1 function.
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Curation History



 

 

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