

Variant: *NM_000033.4(ABCD1):c.601G>A (p.Val201Met)*

Version: 1.0

CA10549981 [↗](#)

585357 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 4408a239-4756-42b8-a60b-720440715873

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HGVS expressions

NM_000033.4:c.601G>A

NM_000033.4(ABCD1):c.601G>A (p.Val201Met)

NC_000023.11:g.153725867G>A

CM000685.2:g.153725867G>A

NC_000023.10:g.152991322G>A

CM000685.1:g.152991322G>A

NC_000023.9:g.152644516G>A

NG_009022.2:g.6000G>A

NG_023231.1:g.3880C>T

ENST00000218104.6:c.601G>A

ENST00000218104.5:c.601G>A

ENST00000370129.4:c.46G>A

NM_000033.3:c.601G>A

Benign

Met criteria codes **2**

BP4

BA1

Not Met criteria codes **1**

PP3

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.601G>A (NM_000033.4) variant in ABCD1 is a missense variant predicted to cause substitution of Valine by Methionine at amino acid 201 (p.Val201Met). The GrpMax filtering allele frequency is 0.001360, gnomAD v4.1, which is higher than the ClinGen Peroxisomal Disorders VCEP threshold of ≥ 0.00017 for BA1, and therefore meets this criterion (BA1). The computational predictor REVEL gives a score of 0.423, (threshold ≤ 0.5) and the splice site predictor SpliceAI indicated that the variant has no impact on splicing with a score of 0.00 for donor/acceptor and loss/gain (threshold ≤ 0.1), evidence that does not predict a damaging effect on ABCD1 function (BP4). In summary,

this variant meets the criteria to be classified as Benign for X-linked adrenoleukodystrophy based on the ACMG/AMP criteria applied, as specified by the ClinGen Peroxisomal VCEP: BA1, BP4. (ClinGen Peroxisomal Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ABCD1 Version 1.0.0)

Met criteria codes

BP4			The computational predictor REVEL gives a score of 0.423, (threshold ≤ 0.5) and the splice site predictor SpliceAI indicated that the variant has no impact on splicing with a score of 0.00 for donor/acceptor and loss/gain (threshold ≤ 0.1), evidence that does not predict a damaging effect on ABCD1 function (BP4).
BA1			The GrpMax filtering allele frequency is 0.001360, gnomAD v4.1, which is higher than the ClinGen Peroxisomal Disorders VCEP threshold of ≥ 0.00017 for BA1, and therefore meets this criterion (BA1). p.Val201Met is present in 13 and 9 hemizygous individuals from gnomAD v2.1 and v3.1 respectively. However, of these individuals, only 3 and 1 hemizygous variant carriers are over 40y.

Not Met criteria codes

PP3			REVEL score of 0.423 is consistent with a non pathogenic or neutral variant effect since it is below the cut-off 0.85
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Curation History

  

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