

Variant: NM_000033.4(ABCD1):c.615G>A (p.Ala205=)

Version: 1.0

CA10549983 [↗](#)

774266 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: a928787f-f83a-496d-b6d2-18131bf1e0e3

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HGVS expressions

NM_000033.4:c.615G>A

NM_000033.4(ABCD1):c.615G>A (p.Ala205=)

NC_000023.11:g.153725881G>A

CM000685.2:g.153725881G>A

NC_000023.10:g.152991336G>A

CM000685.1:g.152991336G>A

NC_000023.9:g.152644530G>A

NG_009022.2:g.6014G>A

NG_023231.1:g.3866C>T

ENST00000218104.6:c.615G>A

ENST00000218104.5:c.615G>A

ENST00000370129.4:c.60G>A

NM_000033.3:c.615G>A

Benign

Met criteria codes **3**

BA1 BP7 BP4

Not Met criteria codes **6**

BS4 BS3 BS2 BP5 BP2 PS3

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 NM_000033.4(ABCD1):c.615G>A (p.Ala205=) variant is a synonymous variant that is outside of the splice region and not predicted by SpliceAI to impact splicing (BP4, BP7). The GrpMax filtering allele frequency of the c.615G>A variant in ABCD1 is 0.002175, gnomAD v4.1, which is higher than the ClinGen Peroxisomal Disorders VCEP threshold of ≥ 0.00017 for BA1, and therefore meets this criterion (BA1). 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 Disorders VCEP: BA1, BP4, BP7 (ClinGen Peroxisomal Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ABCD1 Version 1.0.0)

Met criteria codes

BA1			The GrpMax filtering allele frequency of the c.615G>A variant in ABCD1 is 0.002175, gnomAD v4.1, which is higher than the ClinGen Peroxisomal Disorders VCEP threshold of ≥ 0.00017 for BA1, and therefore meets this criterion (BA1).
BP7			The NM_000033.4(ABCD1):c.615G>A (p.Ala205=) variant is a synonymous variant that is outside of the splice region and not predicted by SpliceAI to impact splicing (BP4, BP7).
BP4			The computational splicing predictor SpliceAI gives a score of 0 (threshold ≤ 0.1) suggesting that the variant has no impact on splicing (BP4).

Not Met criteria codes

BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			more than 10 hemizygous healthy adult males per gnomAD but affected status can not be confirmed
BP5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 

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