

Variant: NM_000033.4(ABCD1):c.692G>A (p.Arg231Gln)

Version: 1.0

CA10549996 [↗](#)

1140271 (ClinVar) [↗](#)

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

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

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

UUID: 71820217-a66e-4118-8cee-bda7afc0e944

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HGVS expressions

NM_000033.4:c.692G>A

NM_000033.4(ABCD1):c.692G>A (p.Arg231Gln)

NC_000023.11:g.153725958G>A

CM000685.2:g.153725958G>A

NC_000023.10:g.152991413G>A

CM000685.1:g.152991413G>A

NC_000023.9:g.152644607G>A

NG_009022.2:g.6091G>A

NG_023231.1:g.3789C>T

ENST00000218104.6:c.692G>A

ENST00000218104.5:c.692G>A

ENST00000370129.4:c.137G>A

NM_000033.3:c.692G>A

Likely Benign

Met criteria codes **2**

BS1 BP4

Not Met criteria codes **2**

BA1 PM2

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:c.692G>A variant in ABCD1 is a missense variant predicted to cause substitution of arginine by glutamine at amino acid 231 (p.Arg231Gln). The GrpMax filtering allele frequency of the c.692G>A variant in ABCD1 is 0.00009439 in the South Asian population in gnomAD v4.1, which is higher than the ClinGen Peroxisomal Disorders VCEP threshold of ≥ 0.000017 for BS1, and therefore meets this criterion (BS1). In addition, the computational predictor REVEL gives a score of 0.357 which is below the threshold of 0.5, and the splice predictor SpliceAI indicated that the variant has no impact on splicing, evidence that does not predict a damaging effect on ABCD1 (BP4).

In summary, this variant meets the criteria to be classified as Likely Benign for adenoleukodystrophy based on the ACMG/AMP criteria applied, as specified by the ClinGen Peroxisomal Disorders VCEP: BS1, BP4. (ClinGen Peroxisomal Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ABCD1 Version 1.0.0)

Met criteria codes

BS1			The GrpMax filtering allele frequency of the c.692G>A variant in ABCD1 is 0.00009439 in the South Asian population in gnomAD v4.1, which is higher than the ClinGen Peroxisomal Disorders VCEP threshold of ≥ 0.000017 for BS1, and therefore meets this criterion (BS1). 9 hemizygotes are listed in total.
BP4			The computational predictor REVEL gives a score of 0.357 which is below the threshold of 0.5, and the splice predictor SpliceAI indicated that the variant has no impact on splicing (score=0), evidence that does not predict a damaging effects on ABCD1 (BP4).

Not Met criteria codes

BA1			The filtering allele frequency of the c.692G>A variant in ABCD1 is 0.00001835 in gnomAD v2.1.1, 0.0001243 in gnomAD v3.1.2, and 0.0001243 (genomes) for all populations which is lower than the ClinGen Peroxisomal Disorders VCEP threshold (0.00017) for BA1, and therefore does not meet this criterion.
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



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