

Variant: *NM_000033.4(ABCD1):c.750G>C (p.Val250=)*

Version: 1.0

CA238543 [↗](#)

193032 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 596104d5-6f6e-4053-9e5e-70a0ae0b1f08

Approved on: 2026/4/16

Published on: 2026/4/16

HGVS expressions

NM_000033.4(ABCD1):c.750G>C (p.Val250=)

NM_000033.4:c.750G>C

NC_000023.11:g.153726016G>C

CM000685.2:g.153726016G>C

NC_000023.10:g.152991471G>C

CM000685.1:g.152991471G>C

NC_000023.9:g.152644665G>C

NG_009022.2:g.6149G>C

NG_023231.1:g.3731C>G

ENST00000218104.6:c.750G>C

ENST00000218104.5:c.750G>C

ENST00000370129.4:c.195G>C

NM_000033.3:c.750G>C

Likely Benign

Met criteria codes **3**

BP7

BP4

PM2_Supporting

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.750G>C (NM_000033.4) variant in ABCD1 is a variant occurring in exon 1. This variant is absent from gnomAD v2.1.1 and v3.1.2. It is reported in gnomAD v4.1 at a frequency of 0.000001120 (1/892612 alleles) in the non-Finnish European population with no hemizygotes, which is below the threshold of 0.0000017 set by the Peroxisomal Disorders VCEP (PM2_Supporting). The results from SpliceAI in silico predictor give a score of 0, which suggests that the variant does not impact ABCD1 function (BP4, BP7). Although the Bayesian points system suggests that a total of -1 pt can yield a likely benign classification, it is conservative to assume a minimum score of -2 pts for

likely benign classifications. In summary, the Peroxisomal Disorders VCEP classified the variant as likely benign using the ACMG/AMP criteria applied, as specified by the ClinGen Peroxisomal Disorders Variant Curation Expert Panel: PM2_supporting, BP4, BP7 (ClinGen Peroxisomal Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ABCD1 Version 1.0.0)

Met criteria codes

BP7			The ABCD1 c.750G>C (p.Val250=) variant is a synonymous (silent) variant that is not predicted by SpliceAI to impact splicing (SpliceAI score: 0.0). Conservation is not considered in the BP7 code per the Walker et al guidance.
BP4			Results from in the silico predictor SpliceAI suggest that the variant does not impact ABCD1 function (SpliceAI score: 0.0), which is below the threshold of 0.2 set by the ClinGen Peroxisomal Disorders VCEP.
PM2_Supporting			This variant is absent from gnomAD v2.1.1 and v3.1.2. It is reported in gnomAD v4.1 at a frequency of 0.000001120 (1/892612 alleles) in the non-Finnish European population with no hemizygotes, which is below the threshold of 0.0000017 set by the Peroxisomal Disorders VCEP (PM2_Supporting).

Curation History



 ▾

 ▾

owing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.