

*Variant: NM_000033.4(ABCD1):c.904_905delinsAT
(p.Glu302Met)*

Version: 1.0

CA1139667828 [↗](#)

872971 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: ce4f7cbe-a04a-4b90-afc1-9da75124cc9c

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HGVS expressions

NM_000033.4(ABCD1):c.904_905delinsAT (p.Glu302Met)

NM_000033.4:c.904_905delinsAT

NC_000023.11:g.153729235_153729236delinsAT

CM000685.2:g.153729235_153729236delinsAT

NC_000023.10:g.152994690_152994691delinsAT

CM000685.1:g.152994690_152994691delinsAT

NC_000023.9:g.152647884_152647885delinsAT

NG_009022.2:g.9368_9369delinsAT

NG_023231.1:g.511_512delinsAT

ENST00000218104.6:c.904_905delinsAT

ENST00000218104.5:c.904_905delinsAT

ENST00000370129.4:c.349_350delinsAT

NM_000033.3:c.904_905delinsAT

Uncertain Significance

Met criteria codes **2**

PM2_Supporting

PP4

Not Met criteria codes **2**

PS4

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.904_905delinsAT variant in ABCD1 is predicted to cause an in-frame substitution of glutamic acid by methionine at amino acid 302 (p.Glu302Met). At least one patient with this variant displayed rapid progression of inflammatory white matter demyelination in childhood and abnormal very long chain fatty acid testing , which is highly specific for adrenoleukodystrophy (PP4, PMID:33547378). This variant is absent from gnomAD v3.1.2, v2.1.1 and v4.1.0 (PM2_Supporting). In summary, this variant meets the criteria to be classified as a variant

of uncertain significance due to insufficient evidence for X-linked adrenoleukodystrophy based on the ACMG/AMP criteria applied, as specified by the ClinGen Peroxisomal Disorders VCEP: PP4, PM2_Supporting (ClinGen Peroxisomal Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ABCD1 Version 1.0.0)

Met criteria codes

PM2_Supporting			This variant is absent from gnomAD v3.1.2, v2.1.1 and v4.1.0 (PM2_Supporting).
PP4			At least one patient with this variant displayed rapid progression of inflammatory white matter demyelination in childhood and abnormal very long chain fatty acid testing, which is highly specific for adrenoleukodystrophy (PP4, PMID:33547378).

Not Met criteria codes

PS4			Only a single case found in a literature search
PP3			The computational splicing predictor SpliceAI gives a score of 0.01 which is below the ClinGen Peroxisomal Disorders VCEP threshold of ≥ 0.5 for acceptor gain, predicting that the variant does not impact splicing.

Curation History



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