

Variant: *NM_001323289.2(CDKL5):c.514G>A (p.Val172Ile)*

Version: 1.0

CA204612 [↗](#)

208653 (ClinVar) [↗](#)

Gene: CDKL5 ([HGNC:6792](#))

Condition: CDKL5 disorder ([MONDO:0100039](#))

Inheritance Mode: X-linked inheritance

UUID: fe4f6838-f920-4a6c-bd90-89a5725803d0

Approved on: 2025/5/7

Published on: 2025/6/30

HGVS expressions

NM_001323289.2:c.514G>A

NM_001323289.2(CDKL5):c.514G>A (p.Val172Ile)

NC_000023.11:g.18584313G>A

CM000685.2:g.18584313G>A

NC_000023.10:g.18602433G>A

CM000685.1:g.18602433G>A

NC_000023.9:g.18512354G>A

NG_008475.1:g.163709G>A

ENST00000623535.2:c.514G>A

ENST00000635828.1:c.514G>A

ENST00000637881.1:c.514G>A

ENST00000674046.1:c.514G>A

ENST00000379989.6:c.514G>A

ENST00000379996.7:c.514G>A

ENST00000463994.4:c.514G>A

ENST00000623535.1:c.514G>A

NM_001037343.1:c.514G>A

NM_003159.2:c.514G>A

NM_001323289.1:c.514G>A

NM_001037343.2:c.514G>A

NM_003159.3:c.514G>A

Uncertain Significance

Met criteria codes **2**

PM2_Supporting

PM6

Not Met criteria codes **1**

BP4

Evidence Links **0**

Expert Panel

[Rett and Angelman-like Disorders VCEP](#) [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** *ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for CDKL5 Version 4.1.0*

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Rett and Angelman-like Disorders VCEP

The p.Val172Ile variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with early onset epileptic encephalopathy (PMID 25356970) (PM6). The p.Val172Ile variant in CDKL5 is absent from gnomAD v4.1 (PM2_Supporting). Computational prediction analysis tools are inconclusive for this variant. In summary, the p.Val172Ile variant in CDKL5 is classified as a variant of uncertain significance based on the ACMG/AMP criteria (PM6, PM2_supporting). (CDKL5 Specifications v.4.1; curation approved on [5/7/2025])

Met criteria codes

PM2_Supporting			The p.Val172Ile variant in CDKL5 is absent from gnomAD v4.1 (PM2_Supporting).
PM6			The p.Val172Ile variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with early onset epileptic encephalopathy (PMID 25356970) (PM6).

Not Met criteria codes

BP4			Computational prediction analysis tools are inconclusive for this variant.
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Curation History



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