

Variant: NM_001323289.2(CDKL5):c.2194G>A
(p.Val732Met)

Version: 1.0

CA326989717 [↗](#)

1648546 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: e60ab412-2e39-4495-b15b-87b8eb896679

Approved on: 2025/5/7

Published on: 2025/6/30

HGVS expressions

NM_001323289.2:c.2194G>A

NM_001323289.2(CDKL5):c.2194G>A (p.Val732Met)

NC_000023.11:g.18613193G>A

CM000685.2:g.18613193G>A

NC_000023.10:g.18631313G>A

CM000685.1:g.18631313G>A

NC_000023.9:g.18541234G>A

NG_008475.1:g.192589G>A

ENST00000623535.2:c.2194G>A

ENST00000635828.1:c.2194G>A

ENST00000674046.1:c.2194G>A

ENST00000379989.6:c.2194G>A

ENST00000379996.7:c.2194G>A

ENST00000463994.4:c.2194G>A

ENST00000623535.1:c.2194G>A

NM_001037343.1:c.2194G>A

NM_003159.2:c.2194G>A

NM_001323289.1:c.2194G>A

NM_001037343.2:c.2194G>A

NM_003159.3:c.2194G>A

Uncertain Significance

Met criteria codes **1**

BP4

Not Met criteria codes **1**

BS1

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 highest population minor allele frequency of the p.Val732Met variant in CDKL5 in gnomAD v4.1 is 0.00002163 in the "remaining" population (not sufficient to meet BS1 criteria). The computational predictor REVEL gives a 0.276 of p.Val732Met, which is below the threshold of 0.29. The splice site predictor, SpliceAI, indicated that the variant has no impact on splicing. Based on this evidence, p.Val732Met does not predict a damaging effect on CDKL5 function (BP4). The p.Val732Met variant is not currently published and is not present in additional databases (internal and publicly available), therefore, no additional criteria are applicable at this time. In summary, the p.Val732Met variant in CDKL5 is classified as a variant of uncertain significance based on the ACMG/AMP criteria (BP4). (CDKL5 Specifications v.4.1; curation approved on [5/7/2025])

Met criteria codes

BP4			The computational predictor REVEL gives a 0.276 of p.Val732Met, which is below the threshold of 0.29. The splice site predictor, SpliceAI, indicated that the variant has no impact on splicing. Based on this evidence, p.Val732Met does not predict a damaging effect on CDKL5 function (BP4).
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Not Met criteria codes

BS1			The highest population minor allele frequency of the p.Val732Met variant in CDKL5 in gnomAD v4.1 is 0.00002163 in the "remaining" population (not sufficient to meet BS1 criteria).
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Curation History



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