

Variant: *NM_001323289.2(CDKL5):c.2465G>A (p.Arg822His)*

Version: 2.0

CA10360541 [↗](#)

377094 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 32cab4fb-2e61-45c0-bb5a-c29787eeeb9a

Approved on: 2023/2/20

Published on: 2023/12/8

HGVS expressions

NM_001323289.2(CDKL5):c.2465G>A (p.Arg822His)

NM_001323289.2:c.2465G>A

NC_000023.11:g.18625216G>A

CM000685.2:g.18625216G>A

NC_000023.10:g.18643336G>A

CM000685.1:g.18643336G>A

NC_000023.9:g.18553257G>A

NG_008475.1:g.204612G>A

ENST00000623535.2:c.2465G>A

ENST00000635828.1:c.2465G>A

ENST00000674046.1:c.2588G>A

ENST00000379989.6:c.2465G>A

ENST00000379996.7:c.2465G>A

ENST00000623535.1:c.2465G>A

NM_001037343.1:c.2465G>A

NM_003159.2:c.2465G>A

NM_001323289.1:c.2465G>A

NM_001037343.2:c.2465G>A

NM_003159.3:c.2465G>A

Likely Benign

Met criteria codes **2**

BP4

BS2

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 Version 2*

[↗](#) **PDF**

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

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

Rett and Angelman-like Disorders VCEP

The p.Arg822His variant in CDKL5 is present in 3 XX and 1 XY individuals in gnomAD v3.1.2 (0.0076%) (not sufficient to meet BS1 criteria). Computational analysis prediction tools suggest that the p.Arg822His variant does not have a deleterious impact; however this information does not predict clinical significance on its own (BP4). The p.Arg822His variant is observed in at least 2 unaffected individuals (internal database-GeneDX) (BS2). In summary, the p.Arg822His variant in CDKL5 is classified as Likely Benign based on the ACMG/AMP criteria (BS2, BP4).

Met criteria codes

BP4			Computational analysis prediction tools suggest that the p.Arg822His variant does not have a deleterious impact; however this information does not predict clinical significance on its own.
BS2			The p.Arg822His variant is observed in at least 2 unaffected individuals (internal database-GeneDX).

Not Met criteria codes

BS1			The p.Arg822His variant in CDKL5 is present in 3 XX and 1 XY individuals in gnomAD v3.1.2 (0.0076%) (not sufficient to meet BS1 criteria).
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Curation History



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