

Variant: *NM_001323289.2(CDKL5):c.93A>G (p.Arg31=)*

Version: 1.0

CA171658 [↗](#)

94115 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 3a17ecd4-9313-4127-93cc-85046b3f66ae

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HGVS expressions

NM_001323289.2(CDKL5):c.93A>G (p.Arg31=)

NM_001323289.2:c.93A>G

NC_000023.11:g.18510848A>G

CM000685.2:g.18510848A>G

NC_000023.10:g.18528968A>G

CM000685.1:g.18528968A>G

NC_000023.9:g.18438889A>G

NG_008475.1:g.90244A>G

ENST00000623535.2:c.93A>G

ENST00000635828.1:c.93A>G

ENST00000637881.1:c.93A>G

ENST00000674046.1:c.93A>G

ENST00000379989.6:c.93A>G

ENST00000379996.7:c.93A>G

ENST00000463994.4:c.93A>G

ENST00000623364.3:c.93A>G

ENST00000623535.1:c.93A>G

ENST00000624700.3:c.93A>G

NM_001037343.1:c.93A>G

NM_003159.2:c.93A>G

NM_001323289.1:c.93A>G

NM_001037343.2:c.93A>G

NM_003159.3:c.93A>G

Likely Benign

Met criteria codes **2**

BP7

BS2

Not Met criteria codes **3**

BA1

BS1

PM2

Evidence Links **0**

Expert Panel

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

Criteria Specification Information **!**

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

Rett and Angelman-like Disorders VCEP

The p.Arg31= variant in CDKL5 is present in 3 female and 2 male individuals in gnomAD (0.002%) (not sufficient to meet BS1 criteria). The p.Arg31= variant is observed in at least 2 unaffected individuals (internal database) (BS2). The silent p.Arg31= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Arg31= variant in CDKL5 is classified as likely benign based on the ACMG/AMP criteria (BS2, BP7).

Met criteria codes

BP7			The silent p.Arg31= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide
BS2			The p.Arg31= variant is observed in at least 2 unaffected individuals (internal database)

Not Met criteria codes

BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			The p.Arg31= variant in CDKL5 is present in 5 female and male individuals in gnomAD (0.002%) (not sufficient to meet BS1 criteria).
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



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