

Variant: *NM_001323289.2(CDKL5):c.1311dup (p.Ser438fs)*

Version: 1.0

CA170444 [↗](#)

143774 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 12905ea2-5ad9-4efb-a505-4fac9bc22028

Approved on: 2024/4/18

Published on: 2024/7/31

HGVS expressions

NM_001323289.2:c.1311dup

NM_001323289.2(CDKL5):c.1311dup (p.Ser438fs)

NC_000023.11:g.18604235dup

CM000685.2:g.18604235dup

NC_000023.10:g.18622355dup

CM000685.1:g.18622355dup

NC_000023.9:g.18532276dup

NG_008475.1:g.183631dup

ENST00000623535.2:c.1311dup

ENST00000635828.1:c.1311dup

ENST00000637881.1:c.1311dup

ENST00000674046.1:c.1311dup

ENST00000379989.6:c.1311dup

ENST00000379996.7:c.1311dup

ENST00000463994.4:c.1311dup

ENST00000623535.1:c.1311dup

NM_001037343.1:c.1311dup

NM_003159.2:c.1311dup

NM_001323289.1:c.1311dup

NM_001037343.2:c.1311dup

NM_003159.3:c.1311dup

Pathogenic

Met criteria codes **3**

PVS1

PM2_Supporting

PM6

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 3.0.0*

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

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

Rett and Angelman-like Disorders VCEP

The c.1311dup variant in CDKL5 is predicted to cause a premature stop codon that leads to a truncated or absent protein in a gene where loss-of-function is an established mechanism. There is significant evidence that loss of this region of the gene is pathogenic (PVS1). The c.1311dup variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with early onset seizures and infantile spasms (PMID 19793311) (PM6). The c.1311dup variant in CDKL5 is absent from gnomAD v2.1.1 (PM2_supporting). In summary, the c.1311dup variant in CDKL5 is classified as pathogenic for a CDKL5-related disorder based on the ACMG/AMP criteria (PVS1, PM6, PM2_supporting).

Met criteria codes

PVS1			The c.1311dup variant in CDKL5 is predicted to cause a premature stop codon that leads to a truncated or absent protein in a gene where loss-of-function is an established mechanism. There is significant evidence that loss of this region of the gene is pathogenic (PVS1).
PM2_Supporting			The c.1311dup variant in CDKL5 is absent from gnomAD v2.1.1.
PM6			The c.1311dup variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with early onset seizures and infantile spasms (PMID 19793311) (PM6).

Curation History



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