

Variant: *NM_001323289.2(CDKL5):c.211A>G (p.Asn71Asp)*

Version: 1.0

CA199365 [↗](#)

156591 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 9c047601-c1b5-474b-b4ff-ce324421c5d7

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HGVS expressions

NM_001323289.2(CDKL5):c.211A>G (p.Asn71Asp)

NM_001323289.2:c.211A>G

NC_000023.11:g.18575419A>G

CM000685.2:g.18575419A>G

NC_000023.10:g.18593539A>G

CM000685.1:g.18593539A>G

NC_000023.9:g.18503460A>G

NG_008475.1:g.154815A>G

ENST00000623535.2:c.211A>G

ENST00000635828.1:c.211A>G

ENST00000637881.1:c.211A>G

ENST00000674046.1:c.211A>G

ENST00000379989.6:c.211A>G

ENST00000379996.7:c.211A>G

ENST00000463994.4:c.211A>G

ENST00000623535.1:c.211A>G

NM_001037343.1:c.211A>G

NM_003159.2:c.211A>G

NM_001323289.1:c.211A>G

NM_001037343.2:c.211A>G

NM_003159.3:c.211A>G

Likely Pathogenic

Met criteria codes 4

PS4

PP4

PM6

PM2_Supporting

Not Met criteria codes 4

PP3

PM1

PM5

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 6.0*

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

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

Rett and Angelman-like Disorders VCEP

The p.Asn71Asp variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with CDKL5 disorder (PMID: 21482751) (PM6, PP4). It has been observed in at least 5 additional individuals with CDKL5 disorder (PMID: 37583270, 19362436, 22872100, internal database - GeneDx) (PS4). The p.Asn71Asp variant is also absent in gnomAD v4.1.1 (PM2_Supporting). In summary, the p.Asn71Asp variant in CDKL5 is classified as Likely Pathogenic for CDKL5 disorder based on the ACMG/AMP criteria (PM6, PS4, PM2_Supporting, PP4). CDKL5 Specifications v6.0; curation approved on 25-Aug-2026.

Met criteria codes

PS4			The p.Asn71Asp variant has been observed in at least 5 individuals with CDKL5 disorder (PMID: 37583270, 19362436, 22872100, 21482751, ClinVar) (PS4).
PP4			The p.Asn71Asp variant in CDKL5 has been reported in an individual with a clinical phenotype suggestive of CDKL5 disorder (PMID: 21482751) (PP4).
PM6			The p.Asn71Asp variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with CDKL5 disorder (PMID: 21482751) (PM6).
PM2_Supporting			The p.Asn71Asp variant in CDKL5 is absent from gnomAD v4.1 (PM2_Supporting).

Not Met criteria codes

PP3			Computational prediction analysis tools are inconclusive for this variant.
PM1			Not located in ATP binding region: aa 19-43; TEY phosphorylation site: aa 169-171
PM5			Alternative changes p.Asn71Lys and p.Asn71Ser do not currently meet the criteria to be classified as LP/P.
BP4			Computational prediction analysis tools are inconclusive for this variant.

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