

Variant: *NM_001323289.2(CDKL5):c.578A>T (p.Asp193Val)*

Version: 1.0

CA412353107 [↗](#)

2419189 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 1c42d16f-8699-4014-b2e0-f8c8832dcd61

Approved on: 2025/10/28

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HGVS expressions

NM_001323289.2:c.578A>T

NM_001323289.2(CDKL5):c.578A>T (p.Asp193Val)

NC_000023.11:g.18587977A>T

CM000685.2:g.18587977A>T

NC_000023.10:g.18606097A>T

CM000685.1:g.18606097A>T

NC_000023.9:g.18516018A>T

NG_008475.1:g.167373A>T

ENST00000623535.2:c.578A>T

ENST00000635828.1:c.578A>T

ENST00000637881.1:c.578A>T

ENST00000674046.1:c.578A>T

ENST00000379989.6:c.578A>T

ENST00000379996.7:c.578A>T

ENST00000463994.4:c.578A>T

ENST00000623535.1:c.578A>T

ENST00000623610.1:n.292A>T

NM_001037343.1:c.578A>T

NM_003159.2:c.578A>T

NM_001323289.1:c.578A>T

NM_001037343.2:c.578A>T

NM_003159.3:c.578A>T

Likely Pathogenic

Met criteria codes **3**

PM5_Strong

PP3

PM2_Supporting

Not Met criteria codes **2**

PP4

PM1

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 5.0.0*

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

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

Rett and Angelman-like Disorders VCEP

The p.Asp193Val variant in CDKL5 is absent from gnomAD v4.1.0 (PM2_Supporting). At least three different pathogenic missense variants have been previously identified within this codon which indicates that this residue is critical to the function of the protein (PMID: 29655203, 23583054; internal database - Invitae; internal database - GeneDx) (PM5_Strong). Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3). The p.Asp193Val variant in CDKL5 has been reported in an individual with epileptic encephalopathy (PMID: 30898514) (PP4 not met). In summary, the p.Asp193Val variant in CDKL5 is classified as Likely Pathogenic for CDKL5 disorder based on the ACMG/AMP criteria (PM5_Strong, PM2_Supporting, PP3). (CDKL5 Specifications v5.0; curation approved on 10/28/2025)

Met criteria codes

PM5_Strong			Multiple pathogenic missense variants have been previously identified within this codon which indicates that this residue is critical to the function of the protein (PMID: 29655203, 23583054; internal database - Invitae; internal database - GeneDx) (PM5_Strong).
PP3			REVEL 0.839 Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3).
PM2_Supporting			The p.Asp193Val variant in CDKL5 is absent from gnomAD v4.1.0 (PM2_Supporting).

Not Met criteria codes

PP4			The p.Asp193Val variant in CDKL5 has been reported in an individual with epileptic encephalopathy (PMID: 30898514) (PP4 not met).
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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