

Variant: *NM_003159.2(CDKL5):c.578A>G (p.Asp193Gly)*

Version: 1.0

CA171648 [↗](#)

143826 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance (dominant (HP:0001423))

UUID: ca9cd59d-d8f3-466f-a2e9-fa39541401c8

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HGVS expressions

NM_003159.2(CDKL5):c.578A>G (p.Asp193Gly)

NM_003159.2:c.578A>G

NC_000023.11:g.18587977A>G

CM000685.2:g.18587977A>G

NC_000023.10:g.18606097A>G

CM000685.1:g.18606097A>G

NC_000023.9:g.18516018A>G

NG_008475.1:g.167373A>G

ENST00000623535.2:c.578A>G

ENST00000635828.1:c.578A>G

ENST00000637881.1:c.578A>G

ENST00000674046.1:c.578A>G

ENST00000379989.6:c.578A>G

ENST00000379996.7:c.578A>G

ENST00000463994.4:c.578A>G

ENST00000623535.1:c.578A>G

ENST00000623610.1:n.292A>G

NM_001037343.1:c.578A>G

NM_001323289.1:c.578A>G

NM_001323289.2:c.578A>G

NM_001037343.2:c.578A>G

NM_003159.3:c.578A>G

Likely Pathogenic

Met criteria codes 5

PP1

PP3

PP4

PM6_Strong

PM2_Supporting

Not Met criteria codes 1

PM5

Evidence Links 1

Expert Panel

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

Criteria Specification Information **!**

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

Rett and Angelman-like Disorders VCEP

The p.Asp193Gly variant in CDKL5 has been reported in at least 2 de novo occurrences (biological parentage unconfirmed) in affected individuals (PMID 23583054) (PM6_Strong). The p.Asp193Gly variant in CDKL5 is absent from gnomAD (PM2_Supporting). The variant has been reported to segregate in two informative meioses (PMID: 23583054) (PP1). Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3). The p.Asp193Gly variant in CDKL5 has been reported in an individual with a clinical phenotype suggestive of a CDKL5-associated disorder (PP4). In summary, the p.Asp193Gly variant in CDKL5 is classified as likely pathogenic for CDKL5-associated disorder based on the ACMG/AMP criteria (PM6_strong, PM2_supporting, PP1, PP3, PP4).

Met criteria codes

PP1			The variant has been reported to segregate in two informative meioses (PMID: 23583054)
			The p.Asp193Gly variant has been reported to segregate in two affected family members with CDKL5 disorder. PubMed:23583054 
PP3			Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own
PP4			The p.Asp193Gly variant in CDKL5 has been reported in an individual with a clinical phenotype suggestive of a CDKL5-associated disorder
PM6_Strong			The p.Asp193Gly variant in CDKL5 has been reported in at least 2 de novo occurrences (biological parentage unconfirmed) in affected individuals (PMID 23583054)
PM2_Supporting			The p.Asp193Gly variant in CDKL5 is absent from gnomAD.

Not Met criteria codes

PM5			Multiple missense variants have been previously identified within this codon (p.Asp193His and p.Asp193Asn); however these sequence variants do not meet the classification of pathogenic (PMID 27781031, 29655203).
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Curation History 

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