

Variant: *NM_001323289.2(CDKL5):c.609G>C (p.Glu203Asp)*

Version: 1.0

CA412353244 [↗](#)

2925658 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 07560285-835d-4a37-8835-129c671fe8ab

Approved on: 2025/6/25

Published on: 2025/12/23

HGVS expressions

NM_001323289.2:c.609G>C

NM_001323289.2(CDKL5):c.609G>C (p.Glu203Asp)

NC_000023.11:g.18588008G>C

CM000685.2:g.18588008G>C

NC_000023.10:g.18606128G>C

CM000685.1:g.18606128G>C

NC_000023.9:g.18516049G>C

NG_008475.1:g.167404G>C

ENST00000623535.2:c.609G>C

ENST00000635828.1:c.609G>C

ENST00000637881.1:c.609G>C

ENST00000674046.1:c.609G>C

ENST00000379989.6:c.609G>C

ENST00000379996.7:c.609G>C

ENST00000463994.4:c.609G>C

ENST00000623535.1:c.609G>C

ENST00000623610.1:n.323G>C

NM_001037343.1:c.609G>C

NM_003159.2:c.609G>C

NM_001323289.1:c.609G>C

NM_001037343.2:c.609G>C

NM_003159.3:c.609G>C

Uncertain Significance

Met criteria codes **3**

PM2_Supporting

PM6

PM5

Not Met criteria codes **4**

BP4

PP3

PS4

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 4.1.0*

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

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

Rett and Angelman-like Disorders VCEP

The p.Glu203Asp variant in CDKL5 is absent from gnomAD v4.1 (PM2_Supporting). The p.Glu203Asp variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with CDKL5 disorder (PMID 19362436) (PM6). A pathogenic missense variant (p.Glu203Gly) has been previously identified within this codon which indicates that this residue is critical to the function of the protein (ClinVar, internal database - Labcorp Genetics) (PM5). Computational prediction analysis tools are inconclusive for this variant. In summary, the p.Glu203Asp variant in CDKL5 is classified as VUS based on the ACMG/AMP criteria (PM2_Supporting, PM6, PM5). (CDKL5 Specifications v.4.1; curation approved on 06/25/2025)

Met criteria codes

PM2_Supporting			The p.Glu203Asp variant in CDKL5 is absent from gnomAD v4.1 (PM2_Supporting).
PM6			The p.Glu203Asp variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with CDKL5 disorder (PMID 19362436).
PM5			A pathogenic missense variant (p.Glu203Gly) has been previously identified within this codon which indicates that this residue is critical to the function of the protein (ClinVar, internal database - Labcorp Genetics) (PM5). Two other variants reported at codon, but neither classified as pathogenic: 1) p.E203G c.608A>G is VLP by Invitae w/de novo occurrence and 2) p.E203Q c.607G>C is reported in PMID 33106377 in adult proband meeting PS4

Not Met criteria codes

BP4			Computational prediction analysis tools are inconclusive for this variant.
PP3			Computational prediction analysis tools are inconclusive for this variant.
PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

▾

▾

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.