

Variant: *NM_001323289.2(CDKL5):c.89G>A (p.Cys30Tyr)*

Version: 2.0

CA412489863 [↗](#)

487576 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 91a503ff-78ef-4eff-989d-4bf3e262f782

Approved on: Mon, 24 Aug 2026 00:00:00 +0000

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HGVS expressions

NM_001323289.2(CDKL5):c.89G>A (p.Cys30Tyr)

NM_001323289.2:c.89G>A

NC_000023.11:g.18510844G>A

CM000685.2:g.18510844G>A

NC_000023.10:g.18528964G>A

CM000685.1:g.18528964G>A

NC_000023.9:g.18438885G>A

NG_008475.1:g.90240G>A

ENST00000623535.2:c.89G>A

ENST00000635828.1:c.89G>A

ENST00000637881.1:c.89G>A

ENST00000674046.1:c.89G>A

ENST00000379989.6:c.89G>A

ENST00000379996.7:c.89G>A

ENST00000463994.4:c.89G>A

ENST00000623364.3:c.89G>A

ENST00000623535.1:c.89G>A

ENST00000624700.3:c.89G>A

NM_001037343.1:c.89G>A

NM_003159.2:c.89G>A

NM_001323289.1:c.89G>A

NM_001037343.2:c.89G>A

NM_003159.3:c.89G>A

Likely Pathogenic

Met criteria codes 5

PP3 PM6 PM1 PS4_Supporting

PM2_Supporting

Not Met criteria codes 7

BP4 BS1 BS3 PS1 PS3 BA1

PM5

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.Cys30Tyr variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with developmental and epileptic encephalopathy (PMID 35701389) (PM6). The p.Cys30Tyr variant has been observed in at least 1 additional individual with clinical features of CDKL5 disorder (internal database - Labcorp (formerly Invitae)) (PS4_Supporting). The p.Cys30Tyr variant occurs in the well-characterized ATP binding region of the CDKL5 gene (PMID: 17993579, 23064044, 28544139, 29264392) (PM1). The p.Cys30Tyr variant in CDKL5 is absent from gnomAD v4.1 (PM2_Supporting). Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3). In summary, the p.Cys30Tyr variant in CDKL5 is classified as Likely Pathogenic for CDKL5 disorder based on the ACMG/AMP criteria (PM1, PM6, PS4_Supporting, PM2_Supporting, PP3). (CDKL5 Specifications v6.0; curation approved on 8/24/2026)

Met criteria codes

PP3			Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3).
PM6			The p.Cys30Tyr variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with developmental and epileptic encephalopathy (PMID 35701389) (PM6).
PM1			The p.Cys30Tyr variant occurs in the well-characterized ATP binding region of the CDKL5 gene (PMID: 17993579, 23064044, 28544139, 29264392) (PM1).
PS4_Supporting			The p.Cys30Tyr variant has been observed in at least 1 additional individual with clinical features of CDKL5 disorder (internal database - Invitae) (PS4_Supporting).
PM2_Supporting			The p.Cys30Tyr variant in CDKL5 is absent from gnomAD v4.1 (PM2_Supporting).

Not Met criteria codes

BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PM5



No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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