

Variant: NM_001323289.2(CDKL5):c.71A>G (p.Tyr24Cys)

Version: 1.0

CA412489792 [↗](#)

976005 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: d8e89261-2759-41e2-98f7-b0266448c3bc

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HGVS expressions

NM_001323289.2:c.71A>G

NM_001323289.2(CDKL5):c.71A>G (p.Tyr24Cys)

NC_000023.11:g.18510826A>G

CM000685.2:g.18510826A>G

NC_000023.10:g.18528946A>G

CM000685.1:g.18528946A>G

NC_000023.9:g.18438867A>G

NG_008475.1:g.90222A>G

ENST00000623535.2:c.71A>G

ENST00000635828.1:c.71A>G

ENST00000637881.1:c.71A>G

ENST00000674046.1:c.71A>G

ENST00000379989.6:c.71A>G

ENST00000379996.7:c.71A>G

ENST00000463994.4:c.71A>G

ENST00000623364.3:c.71A>G

ENST00000623535.1:c.71A>G

ENST00000624700.3:c.71A>G

NM_001037343.1:c.71A>G

NM_003159.2:c.71A>G

NM_001323289.1:c.71A>G

NM_001037343.2:c.71A>G

NM_003159.3:c.71A>G

Pathogenic

Met criteria codes **5**

PM2_Supporting

PS2

PS4

PM1

PP3

Not Met criteria codes **1**

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.Tyr24Cys variant in CDKL5 has been reported as a de novo occurrence (biological parentage both confirmed and unconfirmed) in 2 individuals with clinical features of CDKL5 disorder (internal database - GeneDx; internal database - Variantyx) (PS2). The p.Tyr24Cys variant has been observed in at least 3 additional individuals with clinical features of CDKL5 disorder (PMID 29264392; internal database - Labcorp (formerly Invitae); internal database - 3billion) (PS4). The p.Tyr24Cys variant occurs in the well-characterized ATP binding region functional domain of the CDKL5 gene (PM1). The p.Tyr24Cys 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.Tyr24Cys variant in CDKL5 is classified as Pathogenic for CDKL5 disorder based on the ACMG/AMP criteria (PS2, PS4, PM1, PM2_Supporting, PP3). (CDKL5 Specifications v6.0; curation approved on 8/24/2026)

Met criteria codes

PM2_Supporting			The p.Tyr24Cys variant in CDKL5 is absent from gnomAD v4.1 (PM2_Supporting).
PS2			The p.Tyr24Cys variant in CDKL5 has been reported as a de novo occurrence (biological parentage both confirmed and unconfirmed) in 2 individuals with clinical features of CDKL5 disorder (internal database - GeneDx; internal database - Variantyx) (PS2).
PS4			The p.Tyr24Cys variant has been observed in at least 3 additional individuals with clinical features of CDKL5 disorder (PMID 29264392; internal database - Labcorp (Invitae); internal database - 3billion) (PS4).
PM1			The p.Tyr24Cys variant occurs in the well-characterized ATP binding region functional domain of the CDKL5 gene (PM1).
PP3			Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3).

Not Met criteria codes

PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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