

Variant: NM_001323289.2(CDKL5):c.2572C>T
(p.Arg858Cys)

Version: 1.0

CA10360567 [↗](#)

426292 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 0d082eec-a176-45ad-a6c0-b5ba2d32a963

Approved on: 2026/1/28

Published on: 2026/6/26

HGVS expressions

NM_001323289.2(CDKL5):c.2572C>T (p.Arg858Cys)

NM_001323289.2:c.2572C>T

NC_000023.11:g.18628446C>T

CM000685.2:g.18628446C>T

NC_000023.10:g.18646566C>T

CM000685.1:g.18646566C>T

NC_000023.9:g.18556487C>T

NG_008475.1:g.207842C>T

ENST00000623535.2:c.2572C>T

ENST00000674046.1:c.2695C>T

ENST00000379989.6:c.2572C>T

ENST00000379996.7:c.2572C>T

ENST00000623535.1:c.2572C>T

NM_001037343.1:c.2572C>T

NM_003159.2:c.2572C>T

NM_001323289.1:c.2572C>T

NM_001037343.2:c.2572C>T

NM_003159.3:c.2572C>T

Benign

Met criteria codes **3**

BP5_Strong

BS2

BS1

Not Met criteria codes **1**

PP3

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**

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The highest population minor allele frequency of the p.Arg858Cys variant in CDKL5 in gnomAD v4.1 is 0.00004538 in the Ashkenazi Jewish population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥ 0.0000083) for BS1, and therefore meets this criterion (BS1). The p.Arg858Cys variant is observed in at least 2 unaffected individuals (GeneDx- internal database) (BS2). The p.Arg858Cys variant is found in at least 3 patients with an alternate molecular basis of disease (Labcorp Genetics (formerly Invitae)- internal database; GeneDx- internal database) (BP5_Strong). Computational prediction analysis tools are inconclusive for this variant. In summary, the p.Arg858Cys variant in CDKL5 is classified as benign based on the ACMG/AMP criteria (BS1, BS2, BP5_Strong). (CDKL5 Specifications v.5.0; curation approved on 01/28/2026)

Met criteria codes

BP5_Strong			The p.Arg858Cys variant is found in at least 3 patients with an alternate molecular basis of disease (Labcorp Genetics (formerly Invitae)- internal database; GeneDx- internal database) (BP5_Strong).
BS2			The p.Arg858Cys variant is observed in at least 2 unaffected individuals (GeneDx- internal database) (BS2).
BS1			The highest population minor allele frequency of the p.Arg858Cys variant in CDKL5 in gnomAD v4.1 is 0.00004538 in the Ashkenazi Jewish population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥ 0.0000083) for BS1, and therefore meets this criterion (BS1).

Not Met criteria codes

PP3			Computational prediction analysis tools are inconclusive for this variant.
------------	---	---	--

Curation History



Showing 1 to 1 of 1 rows

--

