The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_001323289.2(CDKL5):c.863C>G (p.Thr288Arg)

Version: 1.0

CA412355847

1879751 (ClinVar)

Gene: CDKL5 (HGNC:6792)
Condition: CDKL5 disorder (MONDO:0100039)
Inheritance Mode: X-linked inheritance
UUID: 18e3e3c3-2b62-4a8c-9bb7-eed9a840b51a
Approved on: 2025/10/28
Published on: 2025/12/23

HGVS expressions

NM_001323289.2(CDKL5):c.863C>G (p.Thr288Arg)
NM_001323289.2:c.863C>G
NC_000023.11:g.18598499C>G
CM000685.2:g.18598499C>G
NC_000023.10:g.18616619C>G
CM000685.1:g.18616619C>G
NC_000023.9:g.18526540C>G
NG_008475.1:g.177895C>G
ENST00000623535.2:c.863C>G
ENST00000635828.1:c.863C>G
ENST00000637881.1:c.863C>G
ENST00000674046.1:c.863C>G
ENST00000379989.6:c.863C>G
ENST00000379996.7:c.863C>G
ENST00000463994.4:c.863C>G
ENST00000623535.1:c.863C>G
NM_001037343.1:c.863C>G
NM_003159.2:c.863C>G
NM_001323289.1:c.863C>G
NM_001037343.2:c.863C>G
NM_003159.3:c.863C>G

Likely Pathogenic

Met criteria codes 4

PS2 PP3 PM2_Supporting PS4_Supporting

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 p.Thr288Arg variant in CDKL5 has been reported as a de novo occurrence (biological parentage confirmed) in an individual with early onset epilepsy (internal database - CeGaT Center for Human Genetics Tuebingen) (PS2). The p.Thr288Arg variant has been observed in at least 1 other individual with features of CDKL5 disorder (internal database - LabCorp (formerly Invitae)) (PS4_Supporting). The p.Thr288Arg 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.Thr288Arg variant in CDKL5 is classified as Likely Pathogenic for CDKL5 disorder based on the ACMG/AMP criteria (PS2, PS4_Supporting, PM2_Supporting, PP3). (CDKL5 Specifications v5.0; curation approved on 10/28/2025)

Met criteria codes

PS2	✔	The p.Thr288Arg variant in CDKL5 has been reported as a de novo occurrence (biological parentage confirmed) in an individual with early onset epilepsy (internal database - CeGaT Center for Human Genetics Tuebingen) (PS2).
PP3	✔	Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3).
PM2_Supporting	✔	The p.Thr288Arg variant in CDKL5 is absent from gnomAD v4.1 (PM2_Supporting).
PS4_Supporting	✔	The p.Thr288Arg variant has been observed in at least 1 other individual with features of CDKL5 disorder (internal database - LabCorp (formerly Invitae)) (PS4_Supporting).

Curation History

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