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Variant: NM_001323289.2(CDKL5):c.2785C>T (p.Gln929Ter)

Version: 1.0

CA412368963

648838 (ClinVar)

Gene: CDKL5 (HGNC:6792)
Condition: CDKL5 disorder (MONDO:0100039)
Inheritance Mode: X-linked inheritance
UUID: d555f471-2ea7-407c-87e3-a580486d75c9
Approved on: Wed, 01 Jul 2026 00:00:00 +0000
Published on: Thu, 24 Sep 2026 00:00:00 +0000

HGVS expressions

NM_001323289.2(CDKL5):c.2785C>T (p.Gln929Ter)
NM_001323289.2:c.2785C>T
NC_000023.11:g.18628659C>T
CM000685.2:g.18628659C>T
NC_000023.10:g.18646779C>T
CM000685.1:g.18646779C>T
NC_000023.9:g.18556700C>T
NG_008475.1:g.208055C>T
ENST00000623535.2:c.2785C>T
ENST00000674046.1:c.2908C>T
ENST00000379989.6:c.2713+72C>T
ENST00000379996.7:c.2713+72C>T
ENST00000623535.1:c.2785C>T
NM_001037343.1:c.2713+72C>T
NM_003159.2:c.2713+72C>T
NM_001323289.1:c.2785C>T
NM_001037343.2:c.2713+72C>T
NM_003159.3:c.2713+72C>T

Pathogenic

Met criteria codes 4

PS2 PM2_Supporting PVS1 PS4_Moderate

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**

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Gln929Ter variant in CDKL5 is predicted to cause a premature stop codon that leads to a truncated or absent protein in a gene where loss-of-function is an established mechanism. There is significant evidence that loss of this region of the gene is pathogenic (PVS1). The p.Gln929Ter variant has been observed in 3 individuals with features of CDKL5 disorder (PMID 35934918, PMID 31440721, GeneDx internal database) (PS4_Moderate). The p.Gln929Ter variant in CDKL5 has been reported as a de novo occurrence (biological parentage confirmed) in an individual with CDKL5 disorder (GeneDx internal database) (PS2). The p.Gln929Ter variant in CDKL5 is absent from gnomAD v4 (PM2_Supporting). In summary, the p.Gln929Ter variant in CDKL5 is classified as pathogenic for CDKL5 disorder based on the ACMG/AMP criteria (PVS1, PS2, PS4_Moderate, PM2_Supporting). (CDKL5 Specifications v6.0.0; curation approved on 7/1/2026.)

Met criteria codes

PS2	✔	The p.Gln929Ter variant in CDKL5 has been reported as a de novo occurrence (biological parentage confirmed) in an individual with CDKL5 disorder (GeneDx internal database) (PS2).
PM2_Supporting	✔	The p.Gln929Ter variant in CDKL5 is absent from gnomAD v4 (PM2_Supporting).
PVS1	✔	The p.Gln929Ter variant in CDKL5 is predicted to cause a premature stop codon that leads to a truncated or absent protein in a gene where loss-of-function is an established mechanism. There is significant evidence that loss of this region of the gene is pathogenic (PVS1).
PS4_Moderate	✔	The p.Gln929Ter variant has been observed in 3 individuals with features of CDKL5 disorder (PMID 35934918, PMID 31440721, GeneDx internal database) (PS4_Moderate).

Curation History

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