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.38T>C (p.Phe13Ser)

Version: 2.0

CA412489564

929426 (ClinVar)

Gene: CDKL5 (HGNC:6792)
Condition: CDKL5 disorder (MONDO:0100039)
Inheritance Mode: X-linked inheritance
UUID: 00757f98-adba-4f7e-a510-520b9680a772
Approved on: 2025/6/25
Published on: 2025/12/23

HGVS expressions

NM_001323289.2(CDKL5):c.38T>C (p.Phe13Ser)
NM_001323289.2:c.38T>C
NC_000023.11:g.18507134T>C
CM000685.2:g.18507134T>C
NC_000023.10:g.18525254T>C
CM000685.1:g.18525254T>C
NC_000023.9:g.18435175T>C
NG_008475.1:g.86530T>C
ENST00000623535.2:c.38T>C
ENST00000635828.1:c.38T>C
ENST00000637881.1:c.38T>C
ENST00000674046.1:c.38T>C
ENST00000379989.6:c.38T>C
ENST00000379996.7:c.38T>C
ENST00000463994.4:c.38T>C
ENST00000623364.3:c.38T>C
ENST00000623535.1:c.38T>C
ENST00000624700.3:c.38T>C
ENST00000624953.1:c.38T>C
NM_001037343.1:c.38T>C
NM_003159.2:c.38T>C
NM_001323289.1:c.38T>C
NM_001037343.2:c.38T>C
NM_003159.3:c.38T>C

Likely Pathogenic

Met criteria codes 3

PP4 PS2 PM2_Supporting

Not Met criteria codes 8

PP3 BA1 PM6 PM1 PM5 BS1 PS1 BP4

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 4.1.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Phe13Ser variant in CDKL5 has been reported as a de novo occurrence (biological parentage confirmed) in an individual with CDKL5 disorder (internal database) and as a de novo occurrence (biological parentage unconfirmed) in an individual with CDKL5 disorder (PMID 31313283) (PS2). The p.Phe13Ser variant in CDKL5 is present in an individual with a clinical phenotype suggestive of CDKL5 disorder (internal database - Labcorp Genetics (formerly Invitae)) (PP4). The p.Phe13Ser variant in CDKL5 is absent from gnomAD v4.1 (PM2_Supporting). In summary, the p.Phe13Ser variant in CDKL5 is classified as likely pathogenic for CDKL5 disorder based on the ACMG/AMP criteria (PS2, PP4, PM2_Supporting). (CDKL5 Specifications v.4.1; curation approved on 06/25/2025)

Met criteria codes

PP4	✔	The p.Phe13Ser variant in CDKL5 is present in an individual with a clinical phenotype suggestive of CDKL5 disorder (internal database - Labcorp Genetics (formerly Invitae)) (PP4).
PS2	✔	The p.Phe13Ser variant in CDKL5 has been reported as a de novo occurrence (biological parentage confirmed) in an individual with CDKL5 disorder (internal database) and as a de novo occurrence (biological parentage unconfirmed) in an individual with CDKL5 disorder (PMID 31313283) (PS2).
PM2_Supporting	✔	The p.Phe13Ser variant in CDKL5 is absent from gnomAD v4.1.

Not Met criteria codes

PP3	✖	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
PM6	✖	The p.Phe13Ser variant in CDKL5 has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with CDKL5 disorder (PMID 31313283) - combined with PS2
PM1	✖	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
BS1	✖	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

BP4		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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