

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**

PS2 **PP4** **PM2_Supporting**

Not Met criteria codes **8**

PS1 **BP4** **PP3** **PM1** **PM5**
PM6 **BA1** **BS1**

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**

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

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).
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).
PM2_Supporting			The p.Phe13Ser variant in CDKL5 is absent from gnomAD v4.1.

Not Met criteria codes

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
PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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
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
BA1			

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

Curation History [↗](#)

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