

Variant: *NM_001323289.2(CDKL5):c.290T>C (p.Leu97Pro)*

Version: 1.0

CA412349776 [↗](#)

870171 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 417c9128-ca8a-4527-b13a-9798da802bd0

Approved on: 2023/4/14

Published on: 2023/6/16

HGVS expressions

NM_001323289.2(CDKL5):c.290T>C (p.Leu97Pro)

NM_001323289.2:c.290T>C

NC_000023.11:g.18579855T>C

CM000685.2:g.18579855T>C

NC_000023.10:g.18597975T>C

CM000685.1:g.18597975T>C

NC_000023.9:g.18507896T>C

NG_008475.1:g.159251T>C

ENST00000623535.2:c.290T>C

ENST00000635828.1:c.290T>C

ENST00000637881.1:c.290T>C

ENST00000674046.1:c.290T>C

ENST00000379989.6:c.290T>C

ENST00000379996.7:c.290T>C

ENST00000463994.4:c.290T>C

ENST00000623535.1:c.290T>C

NM_001037343.1:c.290T>C

NM_003159.2:c.290T>C

NM_001323289.1:c.290T>C

NM_001037343.2:c.290T>C

NM_003159.3:c.290T>C

Likely Pathogenic

Met criteria codes **3**

PS2

PM2_Supporting

PP3

Evidence Links **0**

Expert Panel

Rett and Angelman-like Disorders VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Leu97Pro variant in CDKL5 occurs in the de novo state (biological parentage confirmed) in an individual (GeneDx Internal Database) (PS2). The p.Leu97Pro variant in CDKL5 is absent from gnomAD (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.Leu97Pro variant in CDKL5 is classified as likely pathogenic for a CDKL5-related disorder based on the ACMG/AMP criteria (PS2, PM2_supporting, PP3).

Met criteria codes

PS2			The p.Leu97Pro variant in CDKL5 occurs in the de novo state (biological parentage confirmed) in an individual (GeneDx Internal Database)
PM2_Supporting			The p.Leu97Pro variant in CDKL5 is absent from gnomAD
PP3			Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own

Curation History



 

 

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