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.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

PM2_Supporting PS2 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

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

Curation History

Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.