

Variant: *NM_001323289.2(CDKL5):c.291C>T (p.Leu97=)*

Version: 1.0

CA10360246 [↗](#)

748686 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 6fb20210-ced8-47f1-ba35-7a2d754e4145

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HGVS expressions

NM_001323289.2:c.291C>T

NM_001323289.2(CDKL5):c.291C>T (p.Leu97=)

NC_000023.11:g.18579856C>T

CM000685.2:g.18579856C>T

NC_000023.10:g.18597976C>T

CM000685.1:g.18597976C>T

NC_000023.9:g.18507897C>T

NG_008475.1:g.159252C>T

ENST00000623535.2:c.291C>T

ENST00000635828.1:c.291C>T

ENST00000637881.1:c.291C>T

ENST00000674046.1:c.291C>T

ENST00000379989.6:c.291C>T

ENST00000379996.7:c.291C>T

ENST00000463994.4:c.291C>T

ENST00000623535.1:c.291C>T

NM_001037343.1:c.291C>T

NM_003159.2:c.291C>T

NM_001323289.1:c.291C>T

NM_001037343.2:c.291C>T

NM_003159.3:c.291C>T

Benign

Met criteria codes **3**

BS2 **BS1** **BP7**

Not Met criteria codes **3**

BA1 **PP4** **PM2**

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 allele frequency of the p.Leu97= variant in CDKL5 is 0.011% in African sub population in gnomAD, which is high enough to be classified as likely benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1). The p.Leu97= variant is observed in at least 2 unaffected individuals (internal database) (BS2). The silent p.Leu97= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). The p.Leu97= variant in CDKL5 has been reported apparently homozygous in an individual with infantile epileptic encephalopathy (PMID 28454995); no criteria were applied for this case. In summary, the p.Leu97= variant in CDKL5 is classified as benign based on the ACMG/AMP criteria (BS1, BS2, BP7).

Met criteria codes

BS2			The p.Leu97= variant is observed in at least 2 unaffected individuals (internal database)
BS1			The allele frequency of the p.Leu97= variant in CDKL5 is 0.011% in African sub population in gnomAD, which is high enough to be classified as likely benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions.
BP7			The silent p.Leu97= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide

Not Met criteria codes

BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			PMID 28454995: Patient ID:21, affected with infantile epileptic encephalopathy, reported homozygous for the p.L97= variant. Parental testing results not mentioned. Not applying any criteria. The p.Leu97= variant in CDKL5 has been reported apparently homozygous in an individual with infantile epileptic encephalopathy (PMID 28454995); no criteria were applied for this case.
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



 

 

Showing 1 to 1 of 1 rows

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