

Variant: *NM_001323289.2(CDKL5):c.2107C>G (p.Leu703Val)*

Version: 1.0

CA10360460 [↗](#)

377648 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: ea48b560-cc1c-4ab2-bba8-c4ee39255687

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HGVS expressions

NM_001323289.2:c.2107C>G

NM_001323289.2(CDKL5):c.2107C>G (p.Leu703Val)

NC_000023.11:g.18609525C>G

CM000685.2:g.18609525C>G

NC_000023.10:g.18627645C>G

CM000685.1:g.18627645C>G

NC_000023.9:g.18537566C>G

NG_008475.1:g.188921C>G

ENST00000623535.2:c.2107C>G

ENST00000635828.1:c.2107C>G

ENST00000674046.1:c.2107C>G

ENST00000379989.6:c.2107C>G

ENST00000379996.7:c.2107C>G

ENST00000463994.4:c.2107C>G

ENST00000623535.1:c.2107C>G

NM_001037343.1:c.2107C>G

NM_003159.2:c.2107C>G

NM_001323289.1:c.2107C>G

NM_001037343.2:c.2107C>G

NM_003159.3:c.2107C>G

Likely Benign

Met criteria codes **2**

BS2

BP5

Not Met criteria codes **4**

BS1

BP4

BA1

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 p.Leu703Val variant in CDKL5 is present in 1 female individual in gnomAD (0.0005%) (not sufficient to meet BS1 criteria). The p.Leu703Val variant is observed in at least 2 unaffected individuals (internal database) (BS2). The p.Leu703Val variant is found in a patient with an alternate molecular basis of disease (internal database) (BP5). In summary, the p.Leu703Val variant in CDKL5 is classified as likely benign based on the ACMG/AMP criteria (BS2, BP5).

Met criteria codes

BS2			The p.Leu703Val variant is observed in at least 2 unaffected individuals (internal database) (BS2).
BP5			The p.Leu703Val variant is found in a patient with an alternate molecular basis of disease (internal database) (BP5).

Not Met criteria codes

BS1			The p.Leu703Val variant in CDKL5 is present in 1 female individual in gnomAD (0.0005%) (not sufficient to meet BS1 criteria).
BP4			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
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



 

 

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