

Variant: *NM_001323289.2(CDKL5):c.1579C>T (p.Pro527Ser)*

Version: 1.0

CA412361753 [↗](#)

590134 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 195acd73-f998-4ce4-af7b-24705f7e18f6

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HGVS expressions

NM_001323289.2(CDKL5):c.1579C>T (p.Pro527Ser)

NM_001323289.2:c.1579C>T

NC_000023.11:g.18604503C>T

CM000685.2:g.18604503C>T

NC_000023.10:g.18622623C>T

CM000685.1:g.18622623C>T

NC_000023.9:g.18532544C>T

NG_008475.1:g.183899C>T

ENST00000623535.2:c.1579C>T

ENST00000635828.1:c.1579C>T

ENST00000674046.1:c.1579C>T

ENST00000379989.6:c.1579C>T

ENST00000379996.7:c.1579C>T

ENST00000463994.4:c.1579C>T

ENST00000623535.1:c.1579C>T

NM_001037343.1:c.1579C>T

NM_003159.2:c.1579C>T

NM_001323289.1:c.1579C>T

NM_001037343.2:c.1579C>T

NM_003159.3:c.1579C>T

Likely Benign

Met criteria codes **2**

BS1

BS2_Supporting

Not Met criteria codes **4**

BP4

PS4

PP3

PM1

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.Pro527Ser variant in CDKL5 is 0.017% in African sub population in gnomAD, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1). The p.Pro527Ser variant is observed in at least 1 unaffected individual (internal database - Invitae) (BS2_supporting). In summary, the p.Pro527Ser variant in CDKL5 is classified as a Likely Benign based on the ACMG/AMP criteria (BS1, BS2_supporting).

Met criteria codes

BS1			The allele frequency of the p.Pro527Ser variant in CDKL5 is 0.017% in African sub population in gnomAD, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1).
BS2_Supporting			The p.Pro527Ser variant is observed in at least 1 unaffected individual (internal database - Invitae) (BS2_supporting).

Not Met criteria codes

BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			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

Curation History



 

 

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