

Variant: *NM_001323289.2(CDKL5):c.2555C>T (p.Pro852Leu)*

Version: 1.0

CA294764 [↗](#)

156690 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 8068e478-0128-4e9e-ba26-c670045b3859

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HGVS expressions

NM_001323289.2:c.2555C>T

NM_001323289.2(CDKL5):c.2555C>T (p.Pro852Leu)

NC_000023.11:g.18628429C>T

CM000685.2:g.18628429C>T

NC_000023.10:g.18646549C>T

CM000685.1:g.18646549C>T

NC_000023.9:g.18556470C>T

NG_008475.1:g.207825C>T

ENST00000623535.2:c.2555C>T

ENST00000674046.1:c.2678C>T

ENST00000379989.6:c.2555C>T

ENST00000379996.7:c.2555C>T

ENST00000623535.1:c.2555C>T

NM_001037343.1:c.2555C>T

NM_003159.2:c.2555C>T

NM_001323289.1:c.2555C>T

NM_001037343.2:c.2555C>T

NM_003159.3:c.2555C>T

Benign

Met criteria codes **1**

BA1

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.Pro852Leu variant in CDKL5 is 0.031% in African sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BA1). In summary, the p.Pro852Leu variant in CDKL5 is classified as benign based on the ACMG/AMP criteria (BA1).

Met criteria codes

BA1			The allele frequency of the p.Pro852Leu variant in CDKL5 is 0.031% in African sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions. (BA1)
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Curation History



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