

Variant: *NM_003159.2(CDKL5):c.2445C>T (p.Ser815=)*

Version: 1.0

CA10360539 [↗](#)

258978 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 6d433d86-063b-4149-9c07-abb30a1f5bc2

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HGVS expressions

NM_003159.2:c.2445C>T

NM_003159.2(CDKL5):c.2445C>T (p.Ser815=)

NC_000023.11:g.18625196C>T

CM000685.2:g.18625196C>T

NC_000023.10:g.18643316C>T

CM000685.1:g.18643316C>T

NC_000023.9:g.18553237C>T

NG_008475.1:g.204592C>T

ENST00000623535.2:c.2445C>T

ENST00000635828.1:c.2445C>T

ENST00000674046.1:c.2568C>T

ENST00000379989.6:c.2445C>T

ENST00000379996.7:c.2445C>T

ENST00000623535.1:c.2445C>T

NM_001037343.1:c.2445C>T

NM_001323289.1:c.2445C>T

NM_001323289.2:c.2445C>T

NM_001037343.2:c.2445C>T

NM_003159.3:c.2445C>T

Benign

Met criteria codes **2**

BP7

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.Ser815= variant in CDKL5 is 0.1% 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). The silent

p.Ser815= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Ser815= variant in CDKL5 is classified as benign based on the ACMG/AMP criteria (BA1, BP7).

Met criteria codes

BP7			The silent p.Ser815= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide
BA1			The allele frequency of the p.Ser815= variant in CDKL5 is 0.1% 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.

Curation History



 

 

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