

Variant: *NM_001323289.2(CDKL5):c.2388C>T (p.Ser796=)*

Version: 1.0

CA233657 [↗](#)

166821 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 64f0a058-01b0-4b3f-8cbc-0957e3e5ea8d

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HGVS expressions

NM_001323289.2:c.2388C>T

NM_001323289.2(CDKL5):c.2388C>T (p.Ser796=)

NC_000023.11:g.18625139C>T

CM000685.2:g.18625139C>T

NC_000023.10:g.18643259C>T

CM000685.1:g.18643259C>T

NC_000023.9:g.18553180C>T

NG_008475.1:g.204535C>T

ENST00000623535.2:c.2388C>T

ENST00000635828.1:c.2388C>T

ENST00000674046.1:c.2511C>T

ENST00000379989.6:c.2388C>T

ENST00000379996.7:c.2388C>T

ENST00000623535.1:c.2388C>T

NM_001037343.1:c.2388C>T

NM_003159.2:c.2388C>T

NM_001323289.1:c.2388C>T

NM_001037343.2:c.2388C>T

NM_003159.3:c.2388C>T

Likely Benign

Met criteria codes **2**

BP5 BS2

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.Ser796= variant is observed in at least 2 unaffected individuals (GeneDx internal database) (BS2). The p.Ser796= variant is found in a patient with an alternate molecular basis of disease (GeneDx internal database)(BP5). In summary, the p.Ser796= variant in CDKL5 is classified as likely benign based on the ACMG/AMP criteria (BS2, BP5).

Met criteria codes

BP5			The p.Ser796= variant is found in a patient with an alternate molecular basis of disease (GeneDx internal database) (BP5).
BS2			The p.Ser796= variant is observed in at least 2 unaffected individuals (GeneDx internal database) (BS2).

Curation History

  

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