

Variant: *NM_001323289.2(CDKL5):c.2469C>T (p.Pro823=)*

Version: 1.0

CA10360543 [↗](#)

287145 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 93938cfa-243a-499d-9319-93bef61ed679

Approved on: 2023/12/6

Published on: 2023/12/8

HGVS expressions

NM_001323289.2(CDKL5):c.2469C>T (p.Pro823=)

NM_001323289.2:c.2469C>T

NC_000023.11:g.18625220C>T

CM000685.2:g.18625220C>T

NC_000023.10:g.18643340C>T

CM000685.1:g.18643340C>T

NC_000023.9:g.18553261C>T

NG_008475.1:g.204616C>T

ENST00000623535.2:c.2469C>T

ENST00000635828.1:c.2469C>T

ENST00000674046.1:c.2592C>T

ENST00000379989.6:c.2469C>T

ENST00000379996.7:c.2469C>T

ENST00000623535.1:c.2469C>T

NM_001037343.1:c.2469C>T

NM_003159.2:c.2469C>T

NM_001323289.1:c.2469C>T

NM_001037343.2:c.2469C>T

NM_003159.3:c.2469C>T

Benign

Met criteria codes **3**

BS2 **BS1** **BP7**

Evidence Links **0**

Expert Panel

[Rett and Angelman-like Disorders VCEP](#) [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** *ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for CDKL5 Version 3.0.0*

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The allele frequency of the p.Pro823= variant in CDKL5 is 0.0088% in African/African American sub population in gnomAD v4, 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.Pro823= variant is observed in at least 2 unaffected individuals (GeneDX internal database) (BS2). The silent p.Pro823= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Pro823= variant in CDKL5 is classified as benign based on the ACMG/AMP criteria (BS1, BS2, BP7).

Met criteria codes

BS2			The p.Pro823= variant is observed in at least 2 unaffected individuals (GeneDX internal database)
BS1			The allele frequency of the p.Pro823= variant in CDKL5 is 0.0088% in African/African American 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).
BP7			The silent p.Pro823= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7).

Curation History



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