

Variant: *NM_003159.2(CDKL5):c.3003C>T (p.His1001=)*

Version: 1.0

CA147631 [↗](#)

94111 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 0ead7e03-5b27-452b-97a1-207be56ec9d6

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HGVS expressions

NM_003159.2:c.3003C>T

NM_003159.2(CDKL5):c.3003C>T (p.His1001=)

NC_000023.11:g.18653454C>T

CM000685.2:g.18653454C>T

NC_000023.10:g.18671574C>T

CM000685.1:g.18671574C>T

NC_000023.9:g.18581495C>T

NG_008475.1:g.232850C>T

NG_008659.3:g.28995G>A

ENST00000379984.4:c.184+3199G>A

ENST00000673617.1:n.275C>T

ENST00000379984.3:c.184+3199G>A

ENST00000379989.6:c.3003C>T

ENST00000379996.7:c.3003C>T

NM_000330.3:c.184+3199G>A

NM_001037343.1:c.3003C>T

NM_000330.4:c.184+3199G>A

NM_001037343.2:c.3003C>T

NM_003159.3:c.3003C>T

Benign

Met criteria codes **2**

BA1 **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 allele frequency of the p.His1001= variant in CDKL5 is 2.6% in Ashkenazi Jewish 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

p.His1001= variant is observed in at least 2 unaffected individuals (internal database) (BS2). In summary, the p.His1001= variant in CDKL5 is classified as benign based on the ACMG/AMP criteria (BA1, BS2).

Met criteria codes

BA1	✓	The allele frequency of the p.His1001= variant in CDKL5 is 2.6% in Ashkenazi Jewish 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.
BS2	✓	The p.His1001= variant is observed in at least 2 unaffected individuals (internal database).

Curation History [↗](#)

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