

Variant: *NM_001323289.2(CDKL5):c.1002T>C (p.Ala334=)*

Version: 1.0

CA207213 [↗](#)

210642 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 9755cc66-eb01-463d-861d-cab8d7214a19

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HGVS expressions

NM_001323289.2(CDKL5):c.1002T>C (p.Ala334=)

NM_001323289.2:c.1002T>C

NC_000023.11:g.18603926T>C

CM000685.2:g.18603926T>C

NC_000023.10:g.18622046T>C

CM000685.1:g.18622046T>C

NC_000023.9:g.18531967T>C

NG_008475.1:g.183322T>C

ENST00000623535.2:c.1002T>C

ENST00000635828.1:c.1002T>C

ENST00000637881.1:c.1002T>C

ENST00000674046.1:c.1002T>C

ENST00000379989.6:c.1002T>C

ENST00000379996.7:c.1002T>C

ENST00000463994.4:c.1002T>C

ENST00000623535.1:c.1002T>C

NM_001037343.1:c.1002T>C

NM_003159.2:c.1002T>C

NM_001323289.1:c.1002T>C

NM_001037343.2:c.1002T>C

NM_003159.3:c.1002T>C

Benign

Met criteria codes **3**

BA1

BS2_Supporting

BP7

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.Ala334= variant in CDKL5 is 0.07448% in South Asian 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.Ala334= variant is observed in at least 1 unaffected individual (Clinvar 210642) (BS2_supporting). The silent p.Ala334= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Ala334= variant in CDKL5 is classified as Benign based on the ACMG/AMP criteria (BA1, BA2_supporting, BP7).

Met criteria codes

BA1			The allele frequency of the p.Ala334= variant in CDKL5 is 0.07448% in South Asian 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).
BS2_Supporting			The p.Ala334= variant is observed in at least 1 unaffected individual (Clinvar) (BS2_supporting).
BP7			The silent p.Ala334= 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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