

Variant: *NM_003159.2(CDKL5):c.180G>A (p.Glu60=)*

Version: 1.0

CA200253 [↗](#)

94107 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 366fe5fb-49b5-47f1-8b83-008f62074465

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HGVS expressions

NM_003159.2:c.180G>A

NM_003159.2(CDKL5):c.180G>A (p.Glu60=)

NC_000023.11:g.18575388G>A

CM000685.2:g.18575388G>A

NC_000023.10:g.18593508G>A

CM000685.1:g.18593508G>A

NC_000023.9:g.18503429G>A

NG_008475.1:g.154784G>A

ENST00000623535.2:c.180G>A

ENST00000635828.1:c.180G>A

ENST00000637881.1:c.180G>A

ENST00000674046.1:c.180G>A

ENST00000379989.6:c.180G>A

ENST00000379996.7:c.180G>A

ENST00000463994.4:c.180G>A

ENST00000623364.3:c.180G>A

ENST00000623535.1:c.180G>A

ENST00000624700.3:c.180G>A

NM_001037343.1:c.180G>A

NM_001323289.1:c.180G>A

NM_001323289.2:c.180G>A

NM_001037343.2:c.180G>A

NM_003159.3:c.180G>A

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.Glu60= variant in CDKL5 is 0.9% in East 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.Glu60= variant is observed in at least 1 unaffected individual (internal database) (BS2_supporting). The silent p.Glu60= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Glu60= variant in CDKL5 is classified as benign based on the ACMG/AMP criteria (BA1, BS2_supporting, BP7).

Met criteria codes

BA1			The allele frequency of the p.Glu60= variant in CDKL5 is 0.9% in East 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.
BS2_Supporting			The p.Glu60= variant is observed in at least 1 unaffected individual (internal database)
BP7			The silent p.Glu60= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide

Curation History



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