

Variant: *NM_001323289.2(CDKL5):c.1722G>A (p.Pro574=)*

Version: 1.0

CA209032 [↗](#)

210643 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: fba2c948-8d64-4381-abb6-5a41773d9c59

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HGVS expressions

NM_001323289.2(CDKL5):c.1722G>A (p.Pro574=)

NM_001323289.2:c.1722G>A

NC_000023.11:g.18604646G>A

CM000685.2:g.18604646G>A

NC_000023.10:g.18622766G>A

CM000685.1:g.18622766G>A

NC_000023.9:g.18532687G>A

NG_008475.1:g.184042G>A

ENST00000623535.2:c.1722G>A

ENST00000635828.1:c.1722G>A

ENST00000674046.1:c.1722G>A

ENST00000379989.6:c.1722G>A

ENST00000379996.7:c.1722G>A

ENST00000463994.4:c.1722G>A

ENST00000623535.1:c.1722G>A

NM_001037343.1:c.1722G>A

NM_003159.2:c.1722G>A

NM_001323289.1:c.1722G>A

NM_001037343.2:c.1722G>A

NM_003159.3:c.1722G>A

Benign

Met criteria codes **2**

BA1 **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.Pro574= variant in CDKL5 in South Asian sub population in gnomAD, which is high enough to be classified as benign based on the thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BA1). The silent

p.Pro574 variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Pro574= variant in CDKL5 is classified as Benign based on the ACMG/AMP criteria (BA1, BP7).

Met criteria codes

BA1			The allele frequency of the p.Pro574= variant in CDKL5 in South Asian sub population in gnomAD, which is high enough to be classified as benign based on the thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions
BP7			The silent p.Pro574 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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