

Variant: *NM_001323289.2(CDKL5):c.2151A>G (p.Arg717=)*

Version: 1.0

CA10605540 [↗](#)

286706 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 3f21778e-081f-4a6b-887b-b8284481e825

Approved on: 2023/4/14

Published on: 2023/6/16

HGVS expressions

NM_001323289.2(CDKL5):c.2151A>G (p.Arg717=)

NM_001323289.2:c.2151A>G

NC_000023.11:g.18609569A>G

CM000685.2:g.18609569A>G

NC_000023.10:g.18627689A>G

CM000685.1:g.18627689A>G

NC_000023.9:g.18537610A>G

NG_008475.1:g.188965A>G

ENST00000623535.2:c.2151A>G

ENST00000635828.1:c.2151A>G

ENST00000674046.1:c.2151A>G

ENST00000379989.6:c.2151A>G

ENST00000379996.7:c.2151A>G

ENST00000463994.4:c.2151A>G

ENST00000623535.1:c.2151A>G

NM_001037343.1:c.2151A>G

NM_003159.2:c.2151A>G

NM_001323289.1:c.2151A>G

NM_001037343.2:c.2151A>G

NM_003159.3:c.2151A>G

Likely Pathogenic

Met criteria codes **3**

PS2

PM2_Supporting

PS4_Supporting

Not Met criteria codes **1**

PM6

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 p.Arg717= variant in CDKL5 occurs in the de novo state (biological parentage confirmed) in an individual (Invitae internal database) (PS2). The p.Arg717= variant in CDKL5 occurs in the de novo state (biological parentage unconfirmed) in an individual (GeneDx internal database). The p.Arg717= variant has been observed in at least 2 individuals with neurological disease (Invitae and GeneDx internal database). The p.Arg717= variant in CDKL5 is absent from gnomAD (PM2_supporting). In summary, the p.Arg717= variant in CDKL5 is classified as likely pathogenic for a CDKL5-related disorder based on the ACMG/AMP criteria (PS2, PS4_supporting, PM2_supporting).

Met criteria codes

PS2			The p.Arg717= variant in CDKL5 occurs in the de novo state (biological parentage confirmed) in this individual (Invitae internal database)(PS2).
PM2_Supporting			The p.Arg717= variant in CDKL5 is absent from gnomAD
PS4_Supporting			The p.Arg717= variant has been observed in at least 2 individuals with neurological disease (Invitae and GeneDx internal database)

Not Met criteria codes

PM6			The p.Arg717= variant in CDKL5 occurs in the de novo state (biological parentage unconfirmed) in this individual (GeneDx internal database)(PM6).
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Curation History



 

 

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