

Variant: *NM_001323289.2(CDKL5):c.469G>C (p.Ala157Pro)*

Version: 1.0

CA412352120 [↗](#)

1387019 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 66d37eeb-e461-41c1-b2d6-4c2179e29b0a

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HGVS expressions

NM_001323289.2:c.469G>C

NM_001323289.2(CDKL5):c.469G>C (p.Ala157Pro)

NC_000023.11:g.18584268G>C

CM000685.2:g.18584268G>C

NC_000023.10:g.18602388G>C

CM000685.1:g.18602388G>C

NC_000023.9:g.18512309G>C

NG_008475.1:g.163664G>C

ENST00000623535.2:c.469G>C

ENST00000635828.1:c.469G>C

ENST00000637881.1:c.469G>C

ENST00000674046.1:c.469G>C

ENST00000379989.6:c.469G>C

ENST00000379996.7:c.469G>C

ENST00000463994.4:c.469G>C

ENST00000623535.1:c.469G>C

NM_001037343.1:c.469G>C

NM_003159.2:c.469G>C

NM_001323289.1:c.469G>C

NM_001037343.2:c.469G>C

NM_003159.3:c.469G>C

Likely Pathogenic

Met criteria codes **4**

PM2_Supporting PS2 PP3 PM5

Not Met criteria codes **1**

PM1

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.Ala157Pro variant in CDKL5 has been reported as a de novo occurrence (biological parentage confirmed) in an individual with clinical features of CDKL5 disorder (internal database - Invitae) (PS2). A pathogenic missense variant (p.Ala157Val) has been previously identified within this codon which indicates that this residue is critical to the function of the protein (ClinVar - ClinGen Rett and Angelman-like Disorders Variant Curation Expert Panel) (PM5). The p.Ala157Pro variant in CDKL5 is absent from gnomAD (PM2_Supporting). Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3). In summary, the p.Ala157Pro variant in CDKL5 is classified as Likely Pathogenic for CDKL5 disorder based on the ACMG/AMP criteria (PS2, PM5, PM2_Supporting, PP3).

Met criteria codes

PM2_Supporting			The p.Ala157Pro variant in CDKL5 is absent from gnomAD (PM2_Supporting).
PS2			The p.Ala157Pro variant in CDKL5 has been reported as a de novo occurrence (biological parentage confirmed) in an individual with clinical features of CDKL5 disorder (internal database - Invitae) (PS2_moderate).
PP3			Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3).
PM5			A pathogenic missense variant (p.Ala157Val) has been previously identified within this codon which indicates that this residue is critical to the function of the protein (ClinVar - ClinGen Rett and Angelman-like Disorders Variant Curation Expert Panel) (PM5).

Not Met criteria codes

PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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Curation History



 

 

Showing 1 to 1 of 1 rows

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