

Variant: *NM_001323289.2(CDKL5):c.454T>C (p.Cys152Arg)*

Version: 1.0

CA412351039 [↗](#)

658951 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: c46cc2df-17be-4942-8afe-fa2db0c1c722

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HGVS expressions

NM_001323289.2(CDKL5):c.454T>C (p.Cys152Arg)

NM_001323289.2:c.454T>C

NC_000023.11:g.18581941T>C

CM000685.2:g.18581941T>C

NC_000023.10:g.18600061T>C

CM000685.1:g.18600061T>C

NC_000023.9:g.18509982T>C

NG_008475.1:g.161337T>C

ENST00000623535.2:c.454T>C

ENST00000635828.1:c.454T>C

ENST00000637881.1:c.454T>C

ENST00000674046.1:c.454T>C

ENST00000379989.6:c.454T>C

ENST00000379996.7:c.454T>C

ENST00000463994.4:c.454T>C

ENST00000623535.1:c.454T>C

NM_001037343.1:c.454T>C

NM_003159.2:c.454T>C

NM_001323289.1:c.454T>C

NM_001037343.2:c.454T>C

NM_003159.3:c.454T>C

Likely Pathogenic

Met criteria codes 4

PP3

PM6

PM5

PM2_Supporting

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.Cys152Arg variant in CDKL5 occurs in the de novo state (biological parentage unconfirmed) in an individual (GeneDx Internal Database) (PM6). The p.Cys152Arg 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). A pathogenic missense variant (p.Cys152Phe) has been previously identified within this codon which indicates that this residue is critical to the function of the protein (PMID 15499549; 23651931) (PM5). In summary, the p.Cys152Arg variant in CDKL5 is classified as likely pathogenic for CDKL5-related disorders based on the ACMG/AMP criteria (PM6, PM2_supporting, PP3, PM5).

Met criteria codes

PP3			Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own.
PM6			The p.Cys152Arg variant in CDKL5 occurs in the de novo state (biological parentage unconfirmed) in an individual (GeneDx Internal Database)
PM5			A pathogenic missense variant (p.C152F) has been previously identified within this codon which indicates that this residue is critical to the function of the protein (PMID 15499549; 23651931)
PM2_Supporting			The p.Cys152Arg variant in CDKL5 is absent from gnomAD

Curation History



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