The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

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
Approved on: 2023/4/14
Published on: 2023/6/16

HGVS expressions

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

PM5 PM6 PM2_Supporting PP3

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

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)
PM6	✔	The p.Cys152Arg variant in CDKL5 occurs in the de novo state (biological parentage unconfirmed) in an individual (GeneDx Internal Database)
PM2_Supporting	✔	The p.Cys152Arg variant in CDKL5 is absent from gnomAD
PP3	✔	Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own.

Curation History

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