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Variant: NM_001323289.2(CDKL5):c.1564T>G (p.Leu522Val)

Version: 1.0

CA10360394

434664 (ClinVar)

Gene: CDKL5 (HGNC:6792)
Condition: CDKL5 disorder (MONDO:0100039)
Inheritance Mode: X-linked inheritance
UUID: 32174cab-38f2-4ca9-816b-e98c24809005
Approved on: 2023/2/20
Published on: 2023/3/31

HGVS expressions

NM_001323289.2:c.1564T>G
NM_001323289.2(CDKL5):c.1564T>G (p.Leu522Val)
NC_000023.11:g.18604488T>G
CM000685.2:g.18604488T>G
NC_000023.10:g.18622608T>G
CM000685.1:g.18622608T>G
NC_000023.9:g.18532529T>G
NG_008475.1:g.183884T>G
ENST00000623535.2:c.1564T>G
ENST00000635828.1:c.1564T>G
ENST00000674046.1:c.1564T>G
ENST00000379989.6:c.1564T>G
ENST00000379996.7:c.1564T>G
ENST00000463994.4:c.1564T>G
ENST00000623535.1:c.1564T>G
NM_001037343.1:c.1564T>G
NM_003159.2:c.1564T>G
NM_001323289.1:c.1564T>G
NM_001037343.2:c.1564T>G
NM_003159.3:c.1564T>G

Benign

Met criteria codes 3
BS1 BS2 BP5

Not Met criteria codes 5
PM2 PM1 BP4 PS4 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 allele frequency of the p.Leu522Val variant in CDKL5 is 0.011% in European Non-Finnish sub population in gnomAD, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1). Additionally, the p.Leu522Val variant is observed in at least 2 unaffected individuals (internal database - GeneDx, internal database - Ambry Genetics) (BS2) and is found in a patient with an alternate molecular basis of disease (internal database - GeneDx) (BP5). In summary, the p.Leu522Val variant in CDKL5 is classified as Benign for CDKL5-associated disorder according to ACMG/AMP criteria (BS1, BS2, BP5).

Met criteria codes

BS1	✔	The allele frequency of the p.Leu522Val variant in CDKL5 is 0.011% in European Non-Finnish sub population in gnomAD, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1).
BS2	✔	The p.Leu522Val variant is observed in at least 2 unaffected individuals (internal database - GeneDx, internal database - Ambry Genetics).
BP5	✔	The p.Leu522Val variant is found in a patient with an alternate molecular basis of disease (internal database - GeneDx).

Not Met criteria codes

PM2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_001323289.2(CDKL5):c.1564T>G (...	Benign	CDKL5 Disorder	2023-03-31	1.0	-	CDKL5

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