

Variant: NM_001323289.2(CDKL5):c.1930C>T
(p.Leu644Phe)

Version: 1.0

CA16608795 [↗](#)

393097 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 5b63fcee-8ee0-427e-977d-e2106ba134a9

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Published on: 2023/3/31

HGVS expressions

NM_001323289.2(CDKL5):c.1930C>T (p.Leu644Phe)

NM_001323289.2:c.1930C>T

NC_000023.11:g.18604854C>T

CM000685.2:g.18604854C>T

NC_000023.10:g.18622974C>T

CM000685.1:g.18622974C>T

NC_000023.9:g.18532895C>T

NG_008475.1:g.184250C>T

ENST00000623535.2:c.1930C>T

ENST00000635828.1:c.1930C>T

ENST00000674046.1:c.1930C>T

ENST00000379989.6:c.1930C>T

ENST00000379996.7:c.1930C>T

ENST00000463994.4:c.1930C>T

ENST00000623535.1:c.1930C>T

NM_001037343.1:c.1930C>T

NM_003159.2:c.1930C>T

NM_001323289.1:c.1930C>T

NM_001037343.2:c.1930C>T

NM_003159.3:c.1930C>T

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Not Met criteria codes **4**

BP4

PP3

BA1

BS1

Evidence Links **0**

Expert Panel

Rett and Angelman-like Disorders VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Rett and Angelman-like Disorders VCEP

The p.Leu644Phe variant in CDKL5 is absent from gnomAD (PM2_Supporting). In summary, the p.Leu644Phe variant in CDKL5 is classified as Uncertain significance based on the ACMG/AMP criteria (VUS).

Met criteria codes

PM2_Supporting   The p.Leu644Phe variant in CDKL5 is absent from gnomAD.

Not Met criteria codes

BP4			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
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



 

 

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