

Variant: *NM_001323289.2(CDKL5):c.2556G>A (p.Pro852=)*

Version: 1.0

CA10360565 [↗](#)

1046637 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: e984a0f5-e529-4176-8c80-806fae84527e

Approved on: 2023/6/15

Published on: 2023/12/11

HGVS expressions

NM_001323289.2:c.2556G>A

NM_001323289.2(CDKL5):c.2556G>A (p.Pro852=)

NC_000023.11:g.18628430G>A

CM000685.2:g.18628430G>A

NC_000023.10:g.18646550G>A

CM000685.1:g.18646550G>A

NC_000023.9:g.18556471G>A

NG_008475.1:g.207826G>A

ENST00000623535.2:c.2556G>A

ENST00000674046.1:c.2679G>A

ENST00000379989.6:c.2556G>A

ENST00000379996.7:c.2556G>A

ENST00000623535.1:c.2556G>A

NM_001037343.1:c.2556G>A

NM_003159.2:c.2556G>A

NM_001323289.1:c.2556G>A

NM_001037343.2:c.2556G>A

NM_003159.3:c.2556G>A

Likely Benign

Met criteria codes **2**

BP7

BP4

Not Met criteria codes **1**

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 for CDKL5 Version 3.0.0*

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

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

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The c.2556G>A p.Pro852= variant in CDKL5 (NM_001323289.2) is present in 2 female individuals in gnomAD (0.0072%) (not sufficient to meet BS1 criteria). The silent p.Pro852= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP4, BP7). In summary, the p.Pro852= variant in CDKL5 is classified as Likely Benign based on the ACMG/AMP criteria (BP4, BP7).

Met criteria codes

BP7			The silent p.(Pro852=) variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide.
BP4			The silent p.(Pro852=) variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide.

Not Met criteria codes

BS1			The c.2556G>A p.(Pro852=) variant in CDKL5 (NM_001323289.2) is present in 2 female individuals in gnomAD (0.0072%) (not sufficient to meet BS1 criteria).
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Curation History



 

 

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