

Variant: *NM_001323289.2(CDKL5):c.2265A>T (p.Ala755=)*

Version: 1.0

CA233655 [↗](#)

166820 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: ad487149-5605-45f6-b664-92b49a06f2f7

Approved on: 2026/4/23

Published on: 2026/6/26

HGVS expressions

NM_001323289.2(CDKL5):c.2265A>T (p.Ala755=)

NM_001323289.2:c.2265A>T

NC_000023.11:g.18613264A>T

CM000685.2:g.18613264A>T

NC_000023.10:g.18631384A>T

CM000685.1:g.18631384A>T

NC_000023.9:g.18541305A>T

NG_008475.1:g.192660A>T

ENST00000623535.2:c.2265A>T

ENST00000635828.1:c.2265A>T

ENST00000674046.1:c.2265A>T

ENST00000379989.6:c.2265A>T

ENST00000379996.7:c.2265A>T

ENST00000463994.4:c.2265A>T

ENST00000623535.1:c.2265A>T

NM_001037343.1:c.2265A>T

NM_003159.2:c.2265A>T

NM_001323289.1:c.2265A>T

NM_001037343.2:c.2265A>T

NM_003159.3:c.2265A>T

Likely Benign

Met criteria codes **3**

BP7

BP4

BS2

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 5.0.0*

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

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

Rett and Angelman-like Disorders VCEP

The p.Ala755= variant is observed in at least 2 unaffected individuals (internal database - GeneDx) (BS2). The silent p.Ala755= variant is not predicted to affect splicing using Splice AI and does not affect a highly conserved nucleotide (BP7, BP4). In summary, the p.Ala755= variant in CDKL5 is classified as likely benign based on the ACMG/AMP criteria (BS2, BP4, BP7). (CDKL5 Specifications v5.0; curation approved on 4/23/2026)

Met criteria codes

BP7			The silent p.Ala755= variant is not predicted to affect splicing using Splice AI and does not affect a highly conserved nucleotide (BP7, BP4).
BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			The p.Ala755= variant is observed in at least 2 unaffected individuals (internal database - GeneDx) (BS2).

Not Met criteria codes

BS1			The highest population minor allele frequency of the p.Ala755= variant in CDKL5 in gnomAD v4.1 is 0.000002 in the Non-Finnish European population (not sufficient to meet BS1 criteria).
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Curation History



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