

Variant: *NM_001110792.2(MECP2):c.895G>C (p.Ala299Pro)*

Version: 1.0

CA270538 [↗](#)

143716 (ClinVar) [↗](#)

Gene: MECP2 ([HGNC:4204](#))

Condition: Rett syndrome ([MONDO:0010726](#))

Inheritance Mode: X-linked inheritance

UUID: 0f98a548-1dd4-40b1-8250-58006cc20cfc

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HGVS expressions

NM_001110792.2:c.895G>C

NM_001110792.2(MECP2):c.895G>C (p.Ala299Pro)

NC_000023.11:g.154030969C>G

CM000685.2:g.154030969C>G

NC_000023.10:g.153296420C>G

CM000685.1:g.153296420C>G

NC_000023.9:g.152949614C>G

NG_007107.2:g.1111159G>C

NG_007107.3:g.1111135G>C

ENST00000303391.11:c.859G>C

ENST00000453960.7:c.895G>C

ENST00000637917.1:c.66-33G>C

ENST00000303391.10:c.859G>C

ENST00000407218.5:c.*231G>C

ENST00000453960.6:c.895G>C

ENST00000619732.4:c.859G>C

ENST00000622433.4:c.847G>C

ENST00000628176.2:c.*231G>C

NM_001110792.1:c.895G>C

NM_001316337.1:c.580G>C

NM_004992.3:c.859G>C

NM_001316337.2:c.580G>C

NM_001369391.2:c.580G>C

NM_001369392.2:c.580G>C

NM_001369393.2:c.580G>C

NM_001369394.1:c.580G>C

NM_001369394.2:c.580G>C

NM_001386137.1:c.190G>C

NM_001386138.1:c.190G>C

NM_001386139.1:c.190G>C

NM_004992.4:c.859G>C

Likely Benign

Met criteria codes **3**

BS2

BP5

BP2

Expert Panel

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

Not Met criteria codes 3

BP4 PP4 PM1

Evidence Links 0

Criteria Specification Information !

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Ala287Pro variant in MECP2 (NM_004992.3) is observed in at least 2 unaffected individuals (internal database - GeneDx, internal database - Baylor) (BS2). The p.Ala287Pro variant is observed in the MECP2 gene where a second pathogenic variant in the same gene is present in the patient (internal database - GeneDx) (BP2). The p.Ala287Pro variant is found in a patient with an alternate molecular basis of disease (internal database - Baylor) (BP5). The p.Ala287Pro variant in MECP2 is present in gnomAD v2.1.1 at a frequency of 0.001638% (no criteria met). In summary, the p.Ala287Pro variant in MECP2 is classified as Likely Benign based on the ACMG/AMP criteria (BS2, BP2, BP5).

Met criteria codes

BS2			The p.Ala287Pro variant is observed in at least 2 unaffected individuals (internal database - GeneDx, internal database - Baylor) (BS2).
BP5			The p.Ala287Pro) variant is found in a patient with an alternate molecular basis of disease (internal database - Baylor) (BP5).
BP2			The p.Ala287Pro variant is observed in the MECP2 gene where a second pathogenic variant in the same gene is present in the patient (internal database - GeneDx) (BP2).

Not Met criteria codes

BP4			REVEL score 0.553 does not meet PP3 or BP4.
PP4			The p.Ala287Pro variant in MECP2 was reported in PMID 11055898, a cohort of patients referred for MECP2 genetic testing with no specific clinical information available.
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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