

Variant: *NM_001110792.2(MECP2):c.872C>T (p.Ala291Val)*

Version: 1.0

CA270535 [↗](#)

143710 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 073e9a0d-f2cc-49d2-bf45-62947b3c21a3

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HGVS expressions

NM_001110792.2:c.872C>T

NM_001110792.2(MECP2):c.872C>T (p.Ala291Val)

NC_000023.11:g.154030992G>A

CM000685.2:g.154030992G>A

NC_000023.10:g.153296443G>A

CM000685.1:g.153296443G>A

NC_000023.9:g.152949637G>A

NG_007107.2:g.1111136C>T

NG_007107.3:g.111112C>T

ENST00000303391.11:c.836C>T

ENST00000453960.7:c.872C>T

ENST00000637917.1:c.66-56C>T

ENST00000303391.10:c.836C>T

ENST00000407218.5:c.*208C>T

ENST00000453960.6:c.872C>T

ENST00000619732.4:c.836C>T

ENST00000622433.4:c.824C>T

ENST00000628176.2:c.*208C>T

NM_001110792.1:c.872C>T

NM_001316337.1:c.557C>T

NM_004992.3:c.836C>T

NM_001316337.2:c.557C>T

NM_001369391.2:c.557C>T

NM_001369392.2:c.557C>T

NM_001369393.2:c.557C>T

NM_001369394.1:c.557C>T

NM_001369394.2:c.557C>T

NM_001386137.1:c.167C>T

NM_001386138.1:c.167C>T

NM_001386139.1:c.167C>T

NM_004992.4:c.836C>T

Likely Pathogenic

Met criteria codes **3**

PS4

PP4

PM2_Supporting

Expert Panel

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

Not Met criteria codes 3

PP3PM1BP4

Evidence Links 0

Criteria Specification Information

[Criteria Specification:](#) ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MECP2 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 p.Ala279Val variant in MECP2 (NM_004992.4) has been observed in at least 5 individuals with a clinical phenotype consistent with Rett syndrome (PMID: 15526954, ClinVar, internal database - Invitae) (PS4, PP4). The p.Ala279Val variant in MECP2 is absent from gnomAD v4.1 (PM2_Supporting). In summary, the p.Ala279Val variant in MECP2 is classified as Likely Pathogenic for Rett syndrome based on the ACMG/AMP criteria (PS4, PM2_supporting, PP4).

Met criteria codes

PS4			The p.Ala279Val variant has been observed in at least 5 individuals with a clinical phenotype consistent with Rett syndrome (PMID: 15526954, ClinVar, internal database - Invitae) (PS4).
PP4			The p.Ala279Val variant in MECP2 (NM_004992.4) has been reported in an individual with a clinical phenotype suggestive of Rett syndrome (PMID: 15526954) (PP4).
PM2_Supporting			The p.Ala279Val variant in MECP2 (NM_004992.4) is absent from gnomAD v4.1 (PM2_Supporting).

Not Met criteria codes

PP3			REVEL: 0.492
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4			REVEL: 0.492 Computational prediction analysis tools are inconclusive for this variant.

Curation History

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