

Variant: *NM_001110792.2(MECP2):c.972C>T (p.Val324=)*

Version: 1.0

CA222807 [↗](#)

95207 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: e99c40e8-47d2-488a-a7e4-ef2ad06e21bd

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HGVS expressions

NM_001110792.2:c.972C>T

NM_001110792.2(MECP2):c.972C>T (p.Val324=)

NC_000023.11:g.154030892G>A

CM000685.2:g.154030892G>A

NC_000023.10:g.153296343G>A

CM000685.1:g.153296343G>A

NC_000023.9:g.152949537G>A

NG_007107.2:g.111236C>T

NG_007107.3:g.111212C>T

ENST00000303391.11:c.936C>T

ENST00000453960.7:c.972C>T

ENST00000637917.1:c.110C>T

ENST00000303391.10:c.936C>T

ENST00000407218.5:c.*308C>T

ENST00000453960.6:c.972C>T

ENST00000619732.4:c.936C>T

ENST00000622433.4:c.922C>T

ENST00000628176.2:c.*308C>T

NM_001110792.1:c.972C>T

NM_001316337.1:c.657C>T

NM_004992.3:c.936C>T

NM_001316337.2:c.657C>T

NM_001369391.2:c.657C>T

NM_001369392.2:c.657C>T

NM_001369393.2:c.657C>T

NM_001369394.1:c.657C>T

NM_001369394.2:c.657C>T

NM_001386137.1:c.267C>T

NM_001386138.1:c.267C>T

NM_001386139.1:c.267C>T

NM_004992.4:c.936C>T

Likely Benign

Met criteria codes **4**

PM2_Supporting

BP4

BS2_Supporting

BP7

Expert Panel

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

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

Computational analysis prediction tools suggest that the p.Val312= variant in MECP2 (NM_004992) does not have a deleterious impact; however this information does not predict clinical significance on its own (BP4). The silent p.Val312= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). The p.Val312= variant is observed in at least 1 unaffected individual (GeneDx-internal database) (BS2_Supporting). The p.Val312= variant in MECP2 is absent from gnomAD (PM2_Supporting). In summary, the p.Val312= variant meets the criteria to be classified as Likely Benign. Although there are both pathogenic and benign types of evidence for this variant, the pathogenic evidence is not considered inconsistent with the final classification. ACMG/AMP criteria applied, as specified by the ClinGen Rett/Angelman-like expert panel: (BP4, BP7, BS2_Supporting, PM2_Supporting).

Met criteria codes

PM2_Supporting			The (p.Val312=) variant in MECP2 is absent from gnomAD (PM2_Supporting)
BP4			Computational analysis prediction tools suggest that the p.(Val312=) variant does not have a deleterious impact; however this information does not predict clinical significance on its own
BS2_Supporting			The p.Val312= variant is observed in at least 1unaffected individuals (GeneDx internal case)
BP7			The silent p.(Val312=) variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide

Curation History [↗](#)

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