

Variant: *NM_001110792.2(MECP2):c.1161C>T (p.Ser387=)*

Version: 1.0

CA10558501 [↗](#)

500106 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 573c7d9d-0d39-416b-a905-bd1d44b2b1a9

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HGVS expressions

NM_001110792.2(MECP2):c.1161C>T (p.Ser387=)

NM_001110792.2:c.1161C>T

NC_000023.11:g.154030703G>A

CM000685.2:g.154030703G>A

NC_000023.10:g.153296154G>A

CM000685.1:g.153296154G>A

NC_000023.9:g.152949348G>A

NG_007107.2:g.111425C>T

NG_007107.3:g.111401C>T

ENST00000303391.11:c.1125C>T

ENST00000453960.7:c.1161C>T

ENST00000303391.10:c.1125C>T

ENST00000407218.5:c.*497C>T

ENST00000453960.6:c.1161C>T

ENST00000619732.4:c.1125C>T

ENST00000628176.2:c.*497C>T

NM_001110792.1:c.1161C>T

NM_001316337.1:c.846C>T

NM_004992.3:c.1125C>T

NM_001316337.2:c.846C>T

NM_001369391.2:c.846C>T

NM_001369392.2:c.846C>T

NM_001369393.2:c.846C>T

NM_001369394.1:c.846C>T

NM_001369394.2:c.846C>T

NM_001386137.1:c.456C>T

NM_001386138.1:c.456C>T

NM_001386139.1:c.456C>T

NM_004992.4:c.1125C>T

Likely Benign

Met criteria codes **2**

BP4 BS2

Not Met criteria codes **2**

BP7 BS1

Expert Panel

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

Criteria Specification Information **!**

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The c.1125C>T (p.Ser375=) variant in MECP2 (NM_004992.3) is present in 2 XY individuals in gnomAD (0.001%) (not sufficient to meet BS1 criteria). The p.Ser375= variant is observed in at least 3 unaffected individuals (internal database - Invitae) (BS2). Splice prediction analysis, using multiple computational tools does not suggest an impact to splicing (BP4). In summary, the p.Ser375= variant in MECP2 is classified as Likely Benign based on the ACMG/AMP criteria (BS2, BP4).

Met criteria codes

BP4			Splice prediction analysis, using multiple computational tools does not suggest an impact to splicing (BP4).
BS2			The c.1125C>T (p.Ser375=) variant is observed in at least 6 unaffected individuals (internal database - Invitae) (BS2).

Not Met criteria codes

BP7			PhastCons 1.00, PhyloP 1.24
BS1			The c.1125C>T (p.Ser375=) variant in MECP2 is present in 2 male individuals in gnomAD (0.001%) (not sufficient to meet BS1 criteria).

Curation History [↗](#)



 

 

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