

Variant: *NM_001110792.2(MECP2):c.1177C>T (p.Pro393Ser)*

Version: 1.0

CA10558497 [↗](#)

431897 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 3fde14ce-243f-4159-b28d-83b15854bf43

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HGVS expressions

NM_001110792.2:c.1177C>T

NM_001110792.2(MECP2):c.1177C>T (p.Pro393Ser)

NC_000023.11:g.154030687G>A

CM000685.2:g.154030687G>A

NC_000023.10:g.153296138G>A

CM000685.1:g.153296138G>A

NC_000023.9:g.152949332G>A

NG_007107.2:g.111441C>T

NG_007107.3:g.111417C>T

ENST00000303391.11:c.1141C>T

ENST00000453960.7:c.1177C>T

ENST00000303391.10:c.1141C>T

ENST00000407218.5:c.*513C>T

ENST00000453960.6:c.1177C>T

ENST00000619732.4:c.1141C>T

ENST00000628176.2:c.*513C>T

NM_001110792.1:c.1177C>T

NM_001316337.1:c.862C>T

NM_004992.3:c.1141C>T

NM_001316337.2:c.862C>T

NM_001369391.2:c.862C>T

NM_001369392.2:c.862C>T

NM_001369393.2:c.862C>T

NM_001369394.1:c.862C>T

NM_001369394.2:c.862C>T

NM_001386137.1:c.472C>T

NM_001386138.1:c.472C>T

NM_001386139.1:c.472C>T

NM_004992.4:c.1141C>T

Likely Benign

Met criteria codes **2**

BS2 BP5

Not Met criteria codes **4**

PM1 BS1 BP4 PP3

Expert Panel

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

Criteria Specification Information

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Pro381Ser variant in MECP2 (NM_004992.4) is observed in at least 19 unaffected individuals (internal database - GeneDx, internal database - Invitae) (BS2). The p.Pro381Ser variant is found in a patient with an alternate molecular basis of disease (internal database - GeneDx) (BP5). The highest population minor allele frequency of the p.Pro381Ser variant in MECP2 in gnomAD v4.1 is 0.00003574 in the African/African American population (not sufficient to meet BS1 criteria). In summary, the p.Pro381Ser variant in MECP2 is classified as Likely Benign based on the ACMG/AMP criteria (BS2, BP5).

Met criteria codes

BS2			The p.Pro381Ser variant in MECP2 (NM_004992.4) is observed in at least 19 unaffected individuals (internal database - GeneDx, internal database - Invitae) (BS2).
BP5			The p.Pro381Ser variant is found in a patient with an alternate molecular basis of disease (internal database - GeneDx) (BP5).

Not Met criteria codes

PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			The highest population minor allele frequency of the p.Pro381Ser variant in MECP2 in gnomAD v4.1 is 0.00003574 in the African/African American population (not sufficient to meet BS1 criteria).
BP4			The computational predictor REVEL gives a score of 0.451.
PP3			The computational predictor REVEL gives a score of 0.451.

Curation History [↗](#)

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Showing 1 to 1 of 1 rows

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