

Variant: NM_001110792.2(MECP2):c.1171C>T (p.Pro391Ser)

Version: 1.0

CA10558498 [↗](#)

1170846 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 7d00b5af-dc6e-4fa1-b2c9-abb6ae37ead2

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HGVS expressions

NM_001110792.2:c.1171C>T

NM_001110792.2(MECP2):c.1171C>T (p.Pro391Ser)

NC_000023.11:g.154030693G>A

CM000685.2:g.154030693G>A

NC_000023.10:g.153296144G>A

CM000685.1:g.153296144G>A

NC_000023.9:g.152949338G>A

NG_007107.2:g.111435C>T

NG_007107.3:g.111411C>T

ENST00000303391.11:c.1135C>T

ENST00000453960.7:c.1171C>T

ENST00000303391.10:c.1135C>T

ENST00000407218.5:c.*507C>T

ENST00000453960.6:c.1171C>T

ENST00000619732.4:c.1135C>T

ENST00000628176.2:c.*507C>T

NM_001110792.1:c.1171C>T

NM_001316337.1:c.856C>T

NM_004992.3:c.1135C>T

NM_001316337.2:c.856C>T

NM_001369391.2:c.856C>T

NM_001369392.2:c.856C>T

NM_001369393.2:c.856C>T

NM_001369394.1:c.856C>T

NM_001369394.2:c.856C>T

NM_001386137.1:c.466C>T

NM_001386138.1:c.466C>T

NM_001386139.1:c.466C>T

NM_004992.4:c.1135C>T

Benign

Met criteria codes **3**

BS2 BS1 BP5

Not Met criteria codes **2**

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 allele frequency of the p.Pro379Ser variant in MECP2 (NM_004992.3) is 0.017% in African sub population in gnomAD v2, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1). The p.Pro379Ser variant is observed in the hemizygous state in at least 2 unaffected individuals (internal database - GeneDx, internal database - Invitae) (BS2). The p.Pro379Ser variant is found in at least 3 patients with an alternate molecular basis of disease (internal database - GeneDx, internal database - Invitae) (BP5). In summary, the p.Pro379Ser variant in MECP2 is classified as benign based on the ACMG/AMP criteria (BS1, BS2, BP5).

Met criteria codes

BS2			The p.Pro379Ser variant is observed in the hemizygous state in at least 2 unaffected individuals (internal database - GeneDx, internal database - Invitae) (BS2).
BS1			The allele frequency of the p.Pro379Ser variant in MECP2 (NM_004992.3) is 0.017% in African/African American sub population in gnomAD v2, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1).
BP5			The p.Pro379Ser variant is found in at least 3 patients with an alternate molecular basis of disease (internal database - GeneDx, internal database - Invitae) (BP5).

Not Met criteria codes

BP4			Computational prediction analysis tools are inconclusive for this variant (no criteria met).
PP3			Computational prediction analysis tools are inconclusive for this variant (no criteria met).

Curation History [↗](#)

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