

Variant: *NM_001110792.2(MECP2):c.1186C>T (p.Pro396Ser)*

Version: 1.0

CA10558495 [↗](#)

589521 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: e4bdbb3c-2c00-449e-a92f-3e252d0979be

Approved on: 2025/10/28

Published on: 2025/12/23

HGVS expressions

NM_001110792.2:c.1186C>T

NM_001110792.2(MECP2):c.1186C>T (p.Pro396Ser)

NC_000023.11:g.154030678G>A

CM000685.2:g.154030678G>A

NC_000023.10:g.153296129G>A

CM000685.1:g.153296129G>A

NC_000023.9:g.152949323G>A

NG_007107.2:g.111450C>T

NG_007107.3:g.111426C>T

ENST00000303391.11:c.1150C>T

ENST00000453960.7:c.1186C>T

ENST00000303391.10:c.1150C>T

ENST00000407218.5:c.*522C>T

ENST00000453960.6:c.1186C>T

ENST00000619732.4:c.1150C>T

ENST00000628176.2:c.*522C>T

NM_001110792.1:c.1186C>T

NM_001316337.1:c.871C>T

NM_004992.3:c.1150C>T

NM_001316337.2:c.871C>T

NM_001369391.2:c.871C>T

NM_001369392.2:c.871C>T

NM_001369393.2:c.871C>T

NM_001369394.1:c.871C>T

NM_001369394.2:c.871C>T

NM_001386137.1:c.481C>T

NM_001386138.1:c.481C>T

NM_001386139.1:c.481C>T

NM_004992.4:c.1150C>T

Benign

Met criteria codes **2**

BS2

BS1

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 highest population minor allele frequency of the p.Pro384Ser variant in MECP2 (NM_004992.4) in gnomAD v4.1.0 is 0.00001762 in the South Asian population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥ 0.0000083) for BS1, and therefore meets this criterion (BS1). The p.Pro384Ser variant is observed in at least 2 unaffected individuals (internal database - Labcorp (formerly Invitae); internal database - Ambry) (BS2). In summary, the p.Pro384Ser variant in MECP2 is classified as benign based on the ACMG/AMP criteria (BS1, BS2). (MECP2 Specifications v5.0.0; curation approved on 10/28/2025)

Met criteria codes

BS2			The p.Pro384Ser variant is observed in at least 2 unaffected individuals (internal database - Labcorp (formerly Invitae); internal database - Ambry) (BS2).
BS1			The highest population minor allele frequency of the p.Pro384Ser variant in MECP2 (NM_004992.4) in gnomAD v4.1.0 is 0.00001762 in the South Asian population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥ 0.0000083) for BS1, and therefore meets this criterion (BS1).

Not Met criteria codes

BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			REVEL 0.424

Curation History [↗](#)



 

 

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