

Variant: *NM_001110792.2(MECP2):c.1198C>T (p.Pro400Ser)*

Version: 1.0

CA274535 [↗](#)

143393 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 788bd61d-e448-4487-aa28-328c5b73f0d1

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HGVS expressions

NM_001110792.2(MECP2):c.1198C>T (p.Pro400Ser)

NM_001110792.2:c.1198C>T

NC_000023.11:g.154030666G>A

CM000685.2:g.154030666G>A

NC_000023.10:g.153296117G>A

CM000685.1:g.153296117G>A

NC_000023.9:g.152949311G>A

NG_007107.2:g.111462C>T

NG_007107.3:g.111438C>T

ENST00000303391.11:c.1162C>T

ENST00000453960.7:c.1198C>T

ENST00000303391.10:c.1162C>T

ENST00000407218.5:c.*534C>T

ENST00000453960.6:c.1198C>T

ENST00000619732.4:c.1162C>T

ENST00000628176.2:c.*534C>T

NM_001110792.1:c.1198C>T

NM_001316337.1:c.883C>T

NM_004992.3:c.1162C>T

NM_001316337.2:c.883C>T

NM_001369391.2:c.883C>T

NM_001369392.2:c.883C>T

NM_001369393.2:c.883C>T

NM_001369394.1:c.883C>T

NM_001369394.2:c.883C>T

NM_001386137.1:c.493C>T

NM_001386138.1:c.493C>T

NM_001386139.1:c.493C>T

NM_004992.4:c.1162C>T

Benign

Met criteria codes **1**

BA1

Evidence Links **0**

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.Pro388Ser (NM_004992.3) variant in MECP2 is 0.039% in a sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BA1). In summary, the p.Pro388Ser variant in MECP2 is classified as benign based on the ACMG/AMP criteria (BA1).

Met criteria codes

BA1	 	The allele frequency of the p.Pro400Ser variant in MECP2 is 0.039% in a sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions.
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Curation History [↗](#)



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