

Variant: *NM_001110792.2(MECP2):c.1224C>T (p.Ser408=)*

Version: 1.0

CA207604 [↗](#)

211453 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 2abae5a1-fbd4-4d31-b0d3-dcecef1f42b5

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HGVS expressions

NM_001110792.2(MECP2):c.1224C>T (p.Ser408=)

NM_001110792.2:c.1224C>T

NC_000023.11:g.154030640G>A

CM000685.2:g.154030640G>A

NC_000023.10:g.153296091G>A

CM000685.1:g.153296091G>A

NC_000023.9:g.152949285G>A

NG_007107.2:g.111488C>T

NG_007107.3:g.111464C>T

ENST00000303391.11:c.1188C>T

ENST00000453960.7:c.1224C>T

ENST00000303391.10:c.1188C>T

ENST00000407218.5:c.*560C>T

ENST00000453960.6:c.1224C>T

ENST00000619732.4:c.1188C>T

ENST00000628176.2:c.*560C>T

NM_001110792.1:c.1224C>T

NM_001316337.1:c.909C>T

NM_004992.3:c.1188C>T

NM_001316337.2:c.909C>T

NM_001369391.2:c.909C>T

NM_001369392.2:c.909C>T

NM_001369393.2:c.909C>T

NM_001369394.1:c.909C>T

NM_001369394.2:c.909C>T

NM_001386137.1:c.519C>T

NM_001386138.1:c.519C>T

NM_001386139.1:c.519C>T

NM_004992.4:c.1188C>T

Benign

Met criteria codes **2**

BA1

BS2_Supporting

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.Ser396= variant in MECP2 (NM_004992.3) is 0.15% in European (Finnish) 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). The p.Ser396= variant is observed in 1 unaffected individual (internal database) (BS2_supporting). In summary, the p.Ser396= variant in MECP2 is classified as Benign based on the ACMG/AMP criteria (BA1, BS2_supporting).

Met criteria codes

BA1			The allele frequency of the p.Ser396= variant in MECP2 (NM_004992.3) is 0.15% in European (Finnish) 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)
BS2_Supporting			The p.Ser396= variant in MECP2 (NM_004992.3) is observed in 1 unaffected individual (internal database).

Curation History [↗](#)



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