

Variant: *NM_001110792.2(MECP2):c.1162C>T (p.Pro388Ser)*

Version: 1.0

CA148292 [↗](#)

95184 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 42ed0077-9bfb-41a6-9c0b-f3dedb097940

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HGVS expressions

NM_001110792.2(MECP2):c.1162C>T (p.Pro388Ser)

NM_001110792.2:c.1162C>T

NC_000023.11:g.154030702G>A

CM000685.2:g.154030702G>A

NC_000023.10:g.153296153G>A

CM000685.1:g.153296153G>A

NC_000023.9:g.152949347G>A

NG_007107.2:g.111426C>T

NG_007107.3:g.111402C>T

ENST00000303391.11:c.1126C>T

ENST00000453960.7:c.1162C>T

ENST00000303391.10:c.1126C>T

ENST00000407218.5:c.*498C>T

ENST00000453960.6:c.1162C>T

ENST00000619732.4:c.1126C>T

ENST00000628176.2:c.*498C>T

NM_001110792.1:c.1162C>T

NM_001316337.1:c.847C>T

NM_004992.3:c.1126C>T

NM_001316337.2:c.847C>T

NM_001369391.2:c.847C>T

NM_001369392.2:c.847C>T

NM_001369393.2:c.847C>T

NM_001369394.1:c.847C>T

NM_001369394.2:c.847C>T

NM_001386137.1:c.457C>T

NM_001386138.1:c.457C>T

NM_001386139.1:c.457C>T

NM_004992.4:c.1126C>T

Benign

Met criteria codes **2**

BS2 **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 c.1126C>T, p.(Pro376Ser) variant in MECP2 (NM_004992.4) is 0.13% in European (Non-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.(Pro376Ser) variant is observed in at least 2 unaffected individuals (PMID 10944854, PMID 14560307, RettBASE) (BS1). In summary, the p.(Pro376Ser) variant in MECP2 is classified as Benign based on the ACMG/AMP criteria applied (BA1, BS2).

Met criteria codes

BS2			The p.(Pro376Ser) variant is observed in at least 2 unaffected individuals (PMID 10944854, PMID 14560307, RettBASE)
BA1			The allele frequency of the p.(Pro376Ser) variant in MECP2 is 0.13% in European (Non-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)

Curation History [↗](#)



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Showing 1 to 1 of 1 rows

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