

Variant: *NM_001110792.2(MECP2):c.1120C>T (p.Pro374Ser)*

Version: 1.0

CA415168068 [↗](#)

429415 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 06cf7507-c288-40ba-bd1e-b620664b794d

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HGVS expressions

NM_001110792.2:c.1120C>T

NM_001110792.2(MECP2):c.1120C>T (p.Pro374Ser)

NC_000023.11:g.154030744G>A

CM000685.2:g.154030744G>A

NC_000023.10:g.153296195G>A

CM000685.1:g.153296195G>A

NC_000023.9:g.152949389G>A

NG_007107.2:g.111384C>T

NG_007107.3:g.111360C>T

ENST00000303391.11:c.1084C>T

ENST00000453960.7:c.1120C>T

ENST00000303391.10:c.1084C>T

ENST00000407218.5:c.*456C>T

ENST00000453960.6:c.1120C>T

ENST00000619732.4:c.1084C>T

ENST00000628176.2:c.*456C>T

NM_001110792.1:c.1120C>T

NM_001316337.1:c.805C>T

NM_004992.3:c.1084C>T

NM_001316337.2:c.805C>T

NM_001369391.2:c.805C>T

NM_001369392.2:c.805C>T

NM_001369393.2:c.805C>T

NM_001369394.1:c.805C>T

NM_001369394.2:c.805C>T

NM_001386137.1:c.415C>T

NM_001386138.1:c.415C>T

NM_001386139.1:c.415C>T

NM_004992.4:c.1084C>T

Likely Benign

Met criteria codes **1**

BS2

Not Met criteria codes **2**

BS1

BP4

Expert Panel

Rett and Angelman-like Disorders VCEP [↗](#)

Criteria Specification Information **!**

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Pro362Ser variant in MECP2 (NM_004992.3) is present in 1 XX and 1 XY individual in gnomAD (0.0011%) (not sufficient to meet BS1 criteria). The p.Pro362Ser variant in MECP2 is observed in at least 3 unaffected individuals (internal database - Invitae) (BS2). In the absence of conflicting evidence, this is sufficient evidence to classify as likely benign based on the specifications defined by the ClinGen Rett and Angelman-like Disorders Variant Curation Expert Panel. In summary, the p.Pro362Ser variant in MECP2 is classified as Likely Benign based on the ACMG/AMP criteria (BS2).

Met criteria codes

BS2			The p.Pro362Ser variant in MECP2 (NM_004992.3) is observed in at least 3 unaffected individuals (internal database - Invitae) (BS2).
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Not Met criteria codes

BS1			The p.Pro362Ser variant in MECP2 is present in 1 XX and 1 XY individual in gnomAD (0.0011%) (not sufficient to meet BS1 criteria).
BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)



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