

Variant: *NM_001110792.2(MECP2):c.1117C>T (p.Pro373Ser)*

Version: 1.0

CA415168076 [↗](#)

655951 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 261699fe-f74e-4b38-8e46-427494a03716

Approved on: 2024/4/18

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HGVS expressions

NM_001110792.2(MECP2):c.1117C>T (p.Pro373Ser)

NM_001110792.2:c.1117C>T

NC_000023.11:g.154030747G>A

CM000685.2:g.154030747G>A

NC_000023.10:g.153296198G>A

CM000685.1:g.153296198G>A

NC_000023.9:g.152949392G>A

NG_007107.2:g.111381C>T

NG_007107.3:g.111357C>T

ENST00000303391.11:c.1081C>T

ENST00000453960.7:c.1117C>T

ENST00000303391.10:c.1081C>T

ENST00000407218.5:c.*453C>T

ENST00000453960.6:c.1117C>T

ENST00000619732.4:c.1081C>T

ENST00000628176.2:c.*453C>T

NM_001110792.1:c.1117C>T

NM_001316337.1:c.802C>T

NM_004992.3:c.1081C>T

NM_001316337.2:c.802C>T

NM_001369391.2:c.802C>T

NM_001369392.2:c.802C>T

NM_001369393.2:c.802C>T

NM_001369394.1:c.802C>T

NM_001369394.2:c.802C>T

NM_001386137.1:c.412C>T

NM_001386138.1:c.412C>T

NM_001386139.1:c.412C>T

NM_004992.4:c.1081C>T

Likely Benign

Met criteria codes **2**

BP5 BS2

Not Met criteria codes **1**

BS1

Expert Panel

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

Criteria Specification Information

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Pro361Ser variant in MECP2 (NM_004992.3) is present in 9 XX and 3 XY individual(s) in gnomAD v4.0 (0.001%) (not sufficient to meet BS1 criteria). The p.Pro361Ser variant in the MECP2 gene is observed in at least 2 unaffected individuals (GeneDx internal database) (BS2). The p.Pro361Ser variant is found in a patient with an alternate molecular basis of disease (GeneDx internal database) (BP5). In summary, the p.Pro361Ser variant in MECP2 is classified as likely benign based on the ACMG/AMP criteria (BS2, BP5).

Met criteria codes

BP5			The p.Pro361Ser variant is found in a patient with an alternate molecular basis of disease (GeneDx internal database).
BS2			The p.Pro361Ser variant is observed in at least 2 unaffected individuals (GeneDx internal database).

Not Met criteria codes

BS1			The p.Pro361Ser variant in MECP2 is present in 9 XX and 3 XY individual(s) in gnomAD v4.0 (0.001%) (not sufficient to meet BS1 criteria).
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Curation History [↗](#)

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See Report	Preferred Variant Title	Classification ⓘ	Condition	Published Date	Version ⓘ	Criteria Specification	Gene
View	NM_001110792.2(MECP2):c.1117C>T (...)	Likely Benign	Rett Syndrome ↗	2024-07-01	1.0	ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MECP2 Version 3.0.0 ↗	MECP2 ↗

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