

Variant: *NM_001110792.2(MECP2):c.631C>T (p.Pro211Ser)*

Version: 1.0

CA415173156 [↗](#)

1604076 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 225454eb-ef99-4096-8e55-7a563085f9c2

Approved on: 2025/2/28

Published on: 2025/3/26

HGVS expressions

NM_001110792.2:c.631C>T

NM_001110792.2(MECP2):c.631C>T (p.Pro211Ser)

NC_000023.11:g.154031233G>A

CM000685.2:g.154031233G>A

NC_000023.10:g.153296684G>A

CM000685.1:g.153296684G>A

NC_000023.9:g.152949878G>A

NG_007107.2:g.110895C>T

NG_007107.3:g.110871C>T

ENST00000303391.11:c.595C>T

ENST00000453960.7:c.631C>T

ENST00000637917.1:c.65+163C>T

ENST00000303391.10:c.595C>T

ENST00000407218.5:c.522C>T

ENST00000453960.6:c.631C>T

ENST00000619732.4:c.595C>T

ENST00000622433.4:c.583C>T

ENST00000628176.2:c.486C>T

NM_001110792.1:c.631C>T

NM_001316337.1:c.316C>T

NM_004992.3:c.595C>T

NM_001316337.2:c.316C>T

NM_001369391.2:c.316C>T

NM_001369392.2:c.316C>T

NM_001369393.2:c.316C>T

NM_001369394.1:c.316C>T

NM_001369394.2:c.316C>T

NM_001386137.1:c.-75C>T

NM_001386138.1:c.-75C>T

NM_001386139.1:c.-75C>T

NM_004992.4:c.595C>T

Benign

Met criteria codes **2**

BS2

BS1

Expert Panel

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

Not Met criteria codes 4

PS4BP4PM1BA1

Evidence Links 0

Criteria Specification Information

[Criteria Specification:](#) ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MECP2 Version 3.0.0

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The highest population minor allele frequency of the NM_004992.4: c.595C>T (p.Pro199Ser) variant in MECP2 in gnomAD v4.1 is 0.0002 in the East Asian population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥ 0.00008) for BS1, and therefore meets this criterion (BS1). The p.Pro199Ser variant is observed in at least 2 unaffected individuals (internal data, LabCorp Genetics Inc.) (BS2). The p.Pro199Ser variant is not currently published and is not present in additional databases (internal and publicly available), therefore, no additional criteria are applicable at this time. In summary, the p.Pro199Ser variant in MECP2 is classified as Benign based on the ACMG/AMP criteria (BS1, BS2) (MECP2 Specifications v3.0; curation approved on 02/28/2025).

Met criteria codes

BS2

i

✓

The p.Pro199Ser variant (NM_004992.4) is observed in at least 2 unaffected individuals (internal data, LabCorp Genetics Inc.) (BS2).

BS1

i

✓

The highest population minor allele frequency of the p.Pro199Ser variant in MECP2 (NM_004992.4) in gnomAD v4.1 is 0.0002 in the East Asian population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥ 0.00008) for BS1, and therefore meets this criterion (BS1).

Not Met criteria codes

PS4

i

✗

The p.Pro199Ser variant (NM_004992.4) is not currently published and is not present in additional databases (internal and publicly available), therefore, no additional criteria are applicable at this time.

BP4

i

✗

Computational prediction analysis tools are inconclusive for this variant.

PM1

i

✗

Outside PM1 region.

BA1

i

✗

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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