

Variant: *NM_001110792.2(MECP2):c.1231C>T (p.Pro411Ser)*

Version: 1.0

CA206184 [↗](#)

211454 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: a428a427-c02e-4e6c-bc18-ef901cf5795f

Approved on: 2022/12/9

Published on: 2022/12/23

HGVS expressions

NM_001110792.2(MECP2):c.1231C>T (p.Pro411Ser)

NM_001110792.2:c.1231C>T

NC_000023.11:g.154030633G>A

CM000685.2:g.154030633G>A

NC_000023.10:g.153296084G>A

CM000685.1:g.153296084G>A

NC_000023.9:g.152949278G>A

NG_007107.2:g.111495C>T

NG_007107.3:g.111471C>T

ENST00000303391.11:c.1195C>T

ENST00000453960.7:c.1231C>T

ENST00000303391.10:c.1195C>T

ENST00000407218.5:c.*567C>T

ENST00000453960.6:c.1231C>T

ENST00000619732.4:c.1195C>T

ENST00000628176.2:c.*567C>T

NM_001110792.1:c.1231C>T

NM_001316337.1:c.916C>T

NM_004992.3:c.1195C>T

NM_001316337.2:c.916C>T

NM_001369391.2:c.916C>T

NM_001369392.2:c.916C>T

NM_001369393.2:c.916C>T

NM_001369394.1:c.916C>T

NM_001369394.2:c.916C>T

NM_001386137.1:c.526C>T

NM_001386138.1:c.526C>T

NM_001386139.1:c.526C>T

NM_004992.4:c.1195C>T

Likely Benign

Met criteria codes **2**

BS1

BS2_Supporting

Not Met criteria codes **1**

BP4

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.Pro399Ser variant in MECP2 (NM_004992.3) is 0.01% in Latino/Admixed American sub population in gnomAD, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1). The p.Pro399Ser variant is observed in 1 unaffected individual (internal database) (BS2_supporting). In summary, the p.Pro399Ser variant in MECP2 is classified as a Likely Benign based on the ACMG/AMP criteria (BS1, BS2_supporting).

Met criteria codes

BS1			The allele frequency of the p.Pro399Ser variant in MECP2 (NM_004992.3) is 0.01% in Latino/Admixed American sub population in gnomAD, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1).
BS2_Supporting			The p.Pro399Ser variant in MECP2 (NM_004992.3) is observed in 1 unaffected individual (internal database).

Not Met criteria codes

BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
------------	---	---	--

Curation History [↗](#)



 

 

Showing 1 to 1 of 1 rows

--

