Variant: NM_001110792.2(MECP2):c.1199C>T (p.Pro400Leu)

Version: 1.0

CA270246

143398 (ClinVar)

Gene: MECP2 (HGNC:4204)
Condition: Rett syndrome (MONDO:0010726)
Inheritance Mode: X-linked inheritance
UUID: c9f8a43d-6228-4bee-8416-b26cbcfdb0e4
Approved on: 2021-12-13
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HGVS expressions

NM_001110792.2:c.1199C>T
NM_001110792.2(MECP2):c.1199C>T (p.Pro400Leu)
NC_000023.11:g.154030665G>A
CM000685.2:g.154030665G>A
NC_000023.10:g.153296116G>A
CM000685.1:g.153296116G>A
NC_000023.9:g.152949310G>A
NG_007107.2:g.111463C>T
NG_007107.3:g.111439C>T
ENST00000303391.11:c.1163C>T
ENST00000453960.7:c.1199C>T
ENST00000303391.10:c.1163C>T
ENST00000407218.5:c.*535C>T
ENST00000453960.6:c.1199C>T
ENST00000619732.4:c.1163C>T
ENST00000628176.2:c.*535C>T
NM_001110792.1:c.1199C>T
NM_001316337.1:c.884C>T
NM_004992.3:c.1163C>T
NM_001316337.2:c.884C>T
NM_001369391.2:c.884C>T
NM_001369392.2:c.884C>T
NM_001369393.2:c.884C>T
NM_001369394.1:c.884C>T
NM_001369394.2:c.884C>T
NM_001386137.1:c.494C>T
NM_001386138.1:c.494C>T
NM_001386139.1:c.494C>T
NM_004992.4:c.1163C>T

Benign

Met criteria codes 2

BS1 BS2

Evidence Links 0

Expert Panel

Rett and Angelman-like Disorders VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The allele frequency of the p.Pro388Leu (NM_004992.3) variant in MECP2 is 0.014% in Latino sub population in gnomAD, which is high enough to be classified as likely benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1). Thep.Pro388Leu variant is observed in at least 2 unaffected individuals (GeneDx internal database) (BS2). In summary, the p.Pro388Leu variant in MECP2 is classified as benign based on the ACMG/AMP criteria (BS1, BS2).

Met criteria codes

BS1	✔	The allele frequency of the p.Pro388Leu variant in MECP2 is 0.014% in Latino sub population in gnomAD, which is high enough to be classified as likely benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions.
BS2	✔	The p.Pro388Leu variant is observed in at least 2 unaffected individuals (GeneDx internal database)

Curation History

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