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Variant: NM_001110792.2(MECP2):c.1250C>T (p.Pro417Leu)

Version: 2.0

CA199322

143438 (ClinVar)

Gene: MECP2 (HGNC:4204)
Condition: Rett syndrome (MONDO:0010726)
Inheritance Mode: X-linked inheritance
UUID: 88dbea59-2fc0-49e6-9200-9a675d39f17a
Approved on: 2021/12/22
Published on: 2024/2/23

HGVS expressions

NM_001110792.2:c.1250C>T
NM_001110792.2(MECP2):c.1250C>T (p.Pro417Leu)
NC_000023.11:g.154030614G>A
CM000685.2:g.154030614G>A
NC_000023.10:g.153296065G>A
CM000685.1:g.153296065G>A
NC_000023.9:g.152949259G>A
NG_007107.2:g.111514C>T
NG_007107.3:g.111490C>T
ENST00000303391.11:c.1214C>T
ENST00000453960.7:c.1250C>T
ENST00000303391.10:c.1214C>T
ENST00000407218.5:c.*586C>T
ENST00000453960.6:c.1250C>T
ENST00000619732.4:c.1214C>T
ENST00000628176.2:c.*586C>T
NM_001110792.1:c.1250C>T
NM_001316337.1:c.935C>T
NM_004992.3:c.1214C>T
NM_001316337.2:c.935C>T
NM_001369391.2:c.935C>T
NM_001369392.2:c.935C>T
NM_001369393.2:c.935C>T
NM_001369394.1:c.935C>T
NM_001369394.2:c.935C>T
NM_001386137.1:c.545C>T
NM_001386138.1:c.545C>T
NM_001386139.1:c.545C>T
NM_004992.4:c.1214C>T

Benign

Met criteria codes 2

BS1 BS2_Stand Alone

Not Met criteria codes 1

Expert Panel

Rett and Angelman-like Disorders VCEP

BA1

Evidence Links 0

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.Pro405Leu (NM_004992.3) variant in MECP2 is 0.029% in Latino 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.Pro405Leu variant is observed in at least 2 unaffected individuals (GeneDx internal database)(BS2). In summary, the p.Pro405Leu 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.Pro405Leu variant in MECP2 is 0.029% in Latino 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.
BS2_Stand Alone	✔	The p.Pro405Leu variant is observed in at least 2 unaffected individuals (GeneDx internal database)

Not Met criteria codes

BA1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

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.1250C>T (...	Benign	Rett Syndrome	2024-02-23	2.0	-	MECP2
View	NM_001110792.2(MECP2):c.1250C>T (...	Benign	Rett Syndrome	2021-12-27	1.0	-	MECP2

Showing 1 to 2 of 2 rows

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