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Variant: NM_001110792.2(MECP2):c.251C>T (p.Pro84Leu)

Version: 1.0

CA170269

143501 (ClinVar)

Gene: MECP2 (HGNC:4204)
Condition: Rett syndrome (MONDO:0010726)
Inheritance Mode: X-linked inheritance
UUID: 0867b6a6-269b-41ae-9f93-b57e0942ba44
Approved on: 2021/10/26
Published on: 2021/12/27

HGVS expressions

NM_001110792.2:c.251C>T
NM_001110792.2(MECP2):c.251C>T (p.Pro84Leu)
NC_000023.11:g.154032369G>A
CM000685.2:g.154032369G>A
NC_000023.10:g.153297820G>A
CM000685.1:g.153297820G>A
NC_000023.9:g.152951014G>A
NG_007107.2:g.109759C>T
NG_007107.3:g.109735C>T
ENST00000303391.11:c.215C>T
ENST00000453960.7:c.251C>T
ENST00000303391.10:c.215C>T
ENST00000369957.5:c.*269C>T
ENST00000407218.5:c.251C>T
ENST00000453960.6:c.251C>T
ENST00000486506.5:n.2563C>T
ENST00000611468.1:c.203C>T
ENST00000619732.4:c.215C>T
ENST00000622433.4:c.203C>T
ENST00000628176.2:c.215C>T
NM_001110792.1:c.251C>T
NM_001316337.1:c.-65C>T
NM_004992.3:c.215C>T
NM_001316337.2:c.-65C>T
NM_001369391.2:c.-65C>T
NM_001369392.2:c.-65C>T
NM_001369393.2:c.-65C>T
NM_001369394.1:c.-65C>T
NM_001369394.2:c.-65C>T
NM_001386137.1:c.-346C>T
NM_001386138.1:c.-346C>T
NM_001386139.1:c.-346C>T
NM_004992.4:c.215C>T

Met criteria codes 2

Expert Panel

Likely Benign

BS2_Supporting BS1

Not Met criteria codes 1

BP4

Evidence Links 0

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.Pro72Leu variant in MECP2 (NM_004992.3) is 0.01338% in the Ashkenazi Jewish 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.Pro72Leu variant is observed in at least 1 unaffected individual (internal database - Invitae) (BS2_supporting). In summary, the p.Pro72Leu variant in MECP2 is classified as Likely Benign based on the ACMG/AMP criteria (BS1, BS2_supporting).

Met criteria codes

BS2_Supporting	✔	The p.Pro72Leu variant is observed in at least 1 unaffected individual (internal database - Invitae).
BS1	✔	The allele frequency of the p.Pro72Leu variant in MECP2 is 0.01338% in the Ashkenazi Jewish 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.

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

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