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Variant: NM_001110792.1(MECP2):c.1200_1243del44 (p.Pro401Terfs)

Version: 1.0

CA198846

143406 (ClinVar)

Gene: MECP2 (HGNC:4204)
Condition: Rett syndrome (MONDO:0010726)
Inheritance Mode: X-linked inheritance
UUID: 234c54c2-8825-49ba-9156-a14d379d903e
Approved on: 2021/3/24
Published on: 2021/5/10

HGVS expressions

NM_001110792.1(MECP2):c.1200_1243del44 (p.Pro401Terfs)
NM_001110792.1:c.1200_1243del44
NM_001110792.1:c.1200_1243delACCTCCACCTGAGCCCGAGAGCTCCGAGGACCCCACCAGCC
NC_000023.11:g.154030627_154030670del
CM000685.2:g.154030627_154030670del
NC_000023.10:g.153296078_153296121del
CM000685.1:g.153296078_153296121del
NC_000023.9:g.152949272_152949315del
NG_007107.2:g.111464_111507del
NG_007107.3:g.111440_111483del
ENST00000303391.11:c.1164_1207del
ENST00000453960.7:c.1200_1243del
ENST00000303391.10:c.1164_1207del
ENST00000407218.5:c.*536_*579del
ENST00000453960.6:c.1200_1243del
ENST00000619732.4:c.1164_1207del
ENST00000628176.2:c.*536_*579del
NM_001110792.1:c.1200_1243del
NM_001316337.1:c.885_928del
NM_004992.3:c.1164_1207del
NM_001110792.2:c.1200_1243del
NM_001316337.2:c.885_928del
NM_001369391.2:c.885_928del
NM_001369392.2:c.885_928del
NM_001369393.2:c.885_928del
NM_001369394.1:c.885_928del
NM_001369394.2:c.885_928del
NM_001386137.1:c.495_538del
NM_001386138.1:c.495_538del
NM_001386139.1:c.495_538del
NM_004992.4:c.1164_1207del

Pathogenic

Met criteria codes 4

PS4 PM2_Supporting PS2_Very Strong PVS1

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 p.Pro389* variant in MECP2 is predicted to cause a premature stop codon that leads to a truncated or absent protein where loss-of-function is an established mechanism. There is significant evidence that loss of this region of the gene is pathogenic (PVS1). The p.Pro389* variant in MECP2 has been reported as a de novo occurrence (biological parentage confirmed) in at least 2 individuals with Rett syndrome (internal database, GeneDx) (PS2_very strong). This variant has been observed in at least 5 other individuals with Rett syndrome (PMID 26984561, 21982064, 20151026, 19652677, RettBASE) (PS4). The p.Pro389* variant in MECP2 is absent from gnomAD (PM2_supporting). In summary, the p.Pro389* variant in MECP2 is classified as Pathogenic for Rett syndrome based on the ACMG/AMP criteria (PVS1, PS2_very strong, PS4, PM2_supporting).

Met criteria codes

PS4	✔	The p.Pro389* variant has been observed in at least 5 other individuals with RettSyndrome (PMID 26984561,21982064, 20151026,19652677,RettBASE)
PM2_Supporting	✔	The variant p.Pro389* is absent from controls in Exome Sequencing Project, 1000 Genomes Project, or Exome Aggregation Consortium
PS2_Very Strong	✔	The p.Pro389* variant in MECP2 has been reported as a confirmed de novo occurrence in at least 2 individuals with Rett Syndrome (internal database, GeneDx)
PVS1	✔	The variant p.Pro389* in MECP2 is predicted to cause a premature stop codon that leads to a truncated or absent protein in a gene where loss-of-function is an established mechanism.

Curation History

Showing 1 to 1 of 1 rows

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_001110792.1(MECP2):c.1200_1243...	Pathogenic	Rett Syndrome	2021-05-10	1.0	-	MECP2

Showing 1 to 1 of 1 rows

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