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Variant: NM_001110792.2(MECP2):c.1174_1199del (p.Val392fs)

Version: 1.0

CA232898

143345 (ClinVar)

Gene: MECP2 (HGNC:4204)
Condition: Rett syndrome (MONDO:0010726)
Inheritance Mode: X-linked inheritance
UUID: 670ea38e-226e-4add-8648-64f36bd0bdbb
Approved on: 2022/5/10
Published on: 2022/6/28

HGVS expressions

NM_001110792.2(MECP2):c.1174_1199del (p.Val392fs)
NM_001110792.2:c.1174_1199del
NC_000023.11:g.154030671_154030696del
CM000685.2:g.154030671_154030696del
NC_000023.10:g.153296122_153296147del
CM000685.1:g.153296122_153296147del
NC_000023.9:g.152949316_152949341del
NG_007107.2:g.111438_111463del
NG_007107.3:g.111414_111439del
ENST00000303391.11:c.1138_1163del
ENST00000453960.7:c.1174_1199del
ENST00000303391.10:c.1138_1163del
ENST00000407218.5:c.*510_*535del
ENST00000453960.6:c.1174_1199del
ENST00000619732.4:c.1138_1163del
ENST00000628176.2:c.*510_*535del
NM_001110792.1:c.1174_1199del
NM_001316337.1:c.859_884del
NM_004992.3:c.1138_1163del
NM_001316337.2:c.859_884del
NM_001369391.2:c.859_884del
NM_001369392.2:c.859_884del
NM_001369393.2:c.859_884del
NM_001369394.1:c.859_884del
NM_001369394.2:c.859_884del
NM_001386137.1:c.469_494del
NM_001386138.1:c.469_494del
NM_001386139.1:c.469_494del
NM_004992.4:c.1138_1163del

Pathogenic

Met criteria codes 3

PM2_Supporting PVS1 PS4_Supporting

Expert Panel

Rett and Angelman-like Disorders VCEP

Evidence Links 0

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Val380fs variant 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. There is significant evidence that loss of this region of the gene is pathogenic (PVS1). The p.Val380fs variant has been observed in at least 2 individuals with neurodevelopmental phenotypes (GeneDx internal cases) (PS4_Supporting). The p.Val380fs variant in MECP2 is absent from gnomAD (PM2_Supporting). In summary the p.Val380fs variant in MECP2 is classified as Pathogenic for Rett Syndrome based on the ACMG/AMP criteria (PVS1, PS4_Supporting, PM2_Supporting).

Met criteria codes

PM2_Supporting	✔	PM2_Supporting: The p.Val380fs variant in MECP2 is absent from gnomAD
PVS1	✔	PVS1.The p.Val380fs variant 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. There is significant evidence that loss of this region of the gene is pathogenic
PS4_Supporting	✔	The p.Val380fs variant has been observed in at least 2 individuals with neurodevelopmental phenotypes (GeneDx cases) (PS4_Supporting).

Curation History

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