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Variant: NM_001110792.2(MECP2):c.535C>T (p.Arg179Trp)

Version: 1.0

CA170308

143603 (ClinVar)

Gene: MECP2 (HGNC:4204)
Condition: Rett syndrome (MONDO:0010726)
Inheritance Mode: X-linked inheritance
UUID: f50c6e2c-0485-4079-a589-cb0a7c521757
Approved on: 2021/12/13
Published on: 2023/2/24

HGVS expressions

NM_001110792.2(MECP2):c.535C>T (p.Arg179Trp)
NM_001110792.2:c.535C>T
NC_000023.11:g.154031329G>A
CM000685.2:g.154031329G>A
NC_000023.10:g.153296780G>A
CM000685.1:g.153296780G>A
NC_000023.9:g.152949974G>A
NG_007107.2:g.110799C>T
NG_007107.3:g.110775C>T
ENST00000303391.11:c.499C>T
ENST00000453960.7:c.535C>T
ENST00000637917.1:c.65+67C>T
ENST00000303391.10:c.499C>T
ENST00000407218.5:c.469-43C>T
ENST00000453960.6:c.535C>T
ENST00000486506.5:n.2847C>T
ENST00000611468.1:c.485C>T
ENST00000619732.4:c.499C>T
ENST00000622433.4:c.487C>T
ENST00000628176.2:c.433-43C>T
NM_001110792.1:c.535C>T
NM_001316337.1:c.220C>T
NM_004992.3:c.499C>T
NM_001316337.2:c.220C>T
NM_001369391.2:c.220C>T
NM_001369392.2:c.220C>T
NM_001369393.2:c.220C>T
NM_001369394.1:c.220C>T
NM_001369394.2:c.220C>T
NM_001386137.1:c.-128-43C>T
NM_001386138.1:c.-128-43C>T
NM_001386139.1:c.-128-43C>T
NM_004992.4:c.499C>T

Met criteria codes 3

Expert Panel

Pathogenic

PM2_Supporting PS2_Very Strong PP1_Strong

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 p.Arg167Trp variant (NM_004992.3) in MECP2 has been reported as a de novo occurrence (biological parentage confirmed) in at least 2 individuals with MECP2-related disorders through testing completed at GeneDx (PS2_very strong). The variant has been reported to segregate in at least five informative meioses in published literature (Couvert et al., 2001) (PP1_strong). The p.Arg167Trp variant in MECP2 is absent from gnomAD (PM2_supporting). In summary, the p.Arg167Trp variant in MECP2 is classified as pathogenic for Rett syndrome based on the ACMG/AMP criteria (PS2_very strong, PP1_strong, PM2_supporting).

Met criteria codes

PM2_Supporting	✔	The p.Arg179Trp variant in MECP2 is absent from gnomAD
PS2_Very Strong	✔	The p.Arg179Trp variant in MECP2 has been reported as a de novo occurrence (biological parentage confirmed) in at least 2 individuals with MECP2-related disorders through testing completed at GeneDx
PP1_Strong	✔	The variant has been reported to segregate in at least five informative meioses in published literature (Couvert et al., 2001)

Curation History

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