

Variant: NM_004992.3(MECP2):c.301C>T (p.Pro101Ser)

Version: 1.0

CA270328 [↗](#)

143524 (ClinVar) [↗](#)

Gene: MECP2 ([HGNC:4204](#))
Condition: Rett syndrome ([MONDO:0010726](#))
Inheritance Mode: X-linked inheritance
UUID: 2b8ace0a-d687-49ca-a70c-e5ca323bc4b3
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HGVS expressions

NM_004992.3:c.301C>T
NM_004992.3(MECP2):c.301C>T (p.Pro101Ser)
NC_000023.11:g.154032283G>A
CM000685.2:g.154032283G>A
NC_000023.10:g.153297734G>A
CM000685.1:g.153297734G>A
NC_000023.9:g.152950928G>A
NG_007107.2:g.109845C>T
NG_007107.3:g.109821C>T
ENST00000303391.11:c.301C>T
ENST00000453960.7:c.337C>T
ENST00000303391.10:c.301C>T
ENST00000369957.5:c.*355C>T
ENST00000407218.5:c.337C>T
ENST00000453960.6:c.337C>T
ENST00000486506.5:n.2649C>T
ENST00000611468.1:c.289C>T
ENST00000619732.4:c.301C>T
ENST00000622433.4:c.289C>T
ENST00000628176.2:c.301C>T
NM_001110792.1:c.337C>T
NM_001316337.1:c.22C>T
NM_001110792.2:c.337C>T
NM_001316337.2:c.22C>T
NM_001369391.2:c.22C>T
NM_001369392.2:c.22C>T
NM_001369393.2:c.22C>T
NM_001369394.1:c.22C>T
NM_001369394.2:c.22C>T
NM_001386137.1:c.-260C>T
NM_001386138.1:c.-260C>T
NM_001386139.1:c.-260C>T
NM_004992.4:c.301C>T

Pathogenic

PM5_Strong PM2_Supporting PS4
PP3 PP4 PM1

Rett and Angelman-like Disorders VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence Links 0

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Pro101Ser variant has been observed in at least 4 other individuals with Rett syndrome (RettBASE, internal database) (PS4). Multiple pathogenic missense variants have been previously identified within this codon which indicates that this residue is critical to the function of the protein (PMID 10767337, 31439979) (PM5_Strong). The p.Pro101Ser variant in MECP2 is absent from gnomAD (PM2_Supporting). Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3). The p.Pro101Ser variant in MECP2 has been reported in an individual with a clinical phenotype suggestive of Rett syndrome (PMID 11269512) (PP4). In summary, the p.Pro101Ser variant in MECP2 is classified as pathogenic for Rett syndrome based on the ACMG/AMP criteria (PS4, PM5_strong, PM2_supporting, PP3, PP4).

Met criteria codes

PM5_Strong			Multiple pathogenic missense variants have been previously identified within this codon which indicates that this residue is critical to the function of the protein (PMID 10767337, 31439979)
PM2_Supporting			The p.Pro101Ser variant in MECP2 is absent from gnomAD
PS4			The p.Pro101Ser variant has been observed in at least 4 other individuals with Rett syndrome (RettBASE, internal database)
PP3			Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own
PP4			The p.Pro101Ser variant in MECP2 has been reported in an individual with a clinical phenotype suggestive of Rett syndrome (PMID 11269512)
PM1			The variant p.Pro101Ser in MECP2 is affecting a mutational hotspot (Methyl-DNA binding (MDB) domain: between aa 90-162; PM1

Curation History

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