

Variant: NM_001110792.2(MECP2):c.338C>G (p.Pro113Arg)

Version: 1.0

CA274626 [↗](#)

143526 (ClinVar) [↗](#)

Gene: MECP2 ([HGNC:4204](#))
Condition: Rett syndrome ([MONDO:0010726](#))
Inheritance Mode: X-linked inheritance
UUID: f3b3ee49-05e1-4b1b-9eac-caaee4fc5588
Approved on: 2022/4/28
Published on: 2023/1/4

HGVS expressions

NM_001110792.2:c.338C>G
NM_001110792.2(MECP2):c.338C>G (p.Pro113Arg)
NC_000023.11:g.154032282G>C
CM000685.2:g.154032282G>C
NC_000023.10:g.153297733G>C
CM000685.1:g.153297733G>C
NC_000023.9:g.152950927G>C
NG_007107.2:g.109846C>G
NG_007107.3:g.109822C>G
ENST00000303391.11:c.302C>G
ENST00000453960.7:c.338C>G
ENST00000303391.10:c.302C>G
ENST00000369957.5:c.*356C>G
ENST00000407218.5:c.338C>G
ENST00000453960.6:c.338C>G
ENST00000486506.5:n.2650C>G
ENST00000611468.1:c.290C>G
ENST00000619732.4:c.302C>G
ENST00000622433.4:c.290C>G
ENST00000628176.2:c.302C>G
NM_001110792.1:c.338C>G
NM_001316337.1:c.23C>G
NM_004992.3:c.302C>G
NM_001316337.2:c.23C>G
NM_001369391.2:c.23C>G
NM_001369392.2:c.23C>G
NM_001369393.2:c.23C>G
NM_001369394.1:c.23C>G
NM_001369394.2:c.23C>G
NM_001386137.1:c.-259C>G
NM_001386138.1:c.-259C>G
NM_001386139.1:c.-259C>G
NM_004992.4:c.302C>G

Pathogenic

PS4PP3PM5_StrongPM2_Supporting

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.Pro101Arg variant in MECP2 (NM_004992.4) has been observed in 4 other individuals with Rett Syndrome (PMID 10991689, RettBASE proband ID: 4426, 6597, 6598) and another individual with Angelman syndrome-like phenotype (PMID 11283202) (PS4). Multiple pathogenic missense variants (p.Pro101His, p.Pro101Leu, p.Pro101Thr, p.Pro101Ser) have been previously identified within this codon which indicates that this residue is critical to the function of the protein (PMID 11269512, RettBASE)(PM5_Strong). The p.Pro101Arg 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). In summary the p.Pro101Arg variant in MECP2 is classified as Pathogenic for Rett Syndrome based on the ACMG/AMP criteria (PS4, PM5_Strong, PM2_supporting, PP3).

Met criteria codes

PS4			PS4: The p.Pro101Arg variant has been observed in 4 other individuals with Rett Syndrome (PMID 10991689,RettBASE proband id 4426,6597,6598) and another individual with Angelman syndrome-like phenotype(PMID 11283202)
PP3			Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own
PM5_Strong			Multiple pathogenic missense variants p.Pro101His,p.Pro101Leu have been previously identified within this codon which indicates that this residue is critical to the function of the protein (PMID 11269512, RettBASE)(PM5_Strong)
PM2_Supporting			The p.Pro101Arg variant in MECP2 is absent from gnomAD

Curation History

▼

▼

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

[ClinGen Terms of Use.](#)

⌘ [Powered by BCM's Genboree.](#)