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Variant: NM_001110792.2(MECP2):c.488A>G (p.Asp163Gly)

Version: 2.0

CA270421

143578 (ClinVar)

Gene: MECP2 (HGNC:4204)
Condition: Rett syndrome (MONDO:0010726)
Inheritance Mode: X-linked inheritance
UUID: b6d47811-23c9-458c-b9ae-f33311d4bb97
Approved on: 2023/10/13
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HGVS expressions

NM_001110792.2(MECP2):c.488A>G (p.Asp163Gly)
NM_001110792.2:c.488A>G
NC_000023.11:g.154031376T>C
CM000685.2:g.154031376T>C
NC_000023.10:g.153296827T>C
CM000685.1:g.153296827T>C
NC_000023.9:g.152950021T>C
NG_007107.2:g.110752A>G
NG_007107.3:g.110728A>G
ENST00000303391.11:c.452A>G
ENST00000453960.7:c.488A>G
ENST00000637917.1:c.65+20A>G
ENST00000303391.10:c.452A>G
ENST00000369957.5:c.*506A>G
ENST00000407218.5:c.468+20A>G
ENST00000453960.6:c.488A>G
ENST00000486506.5:n.2800A>G
ENST00000611468.1:c.440A>G
ENST00000619732.4:c.452A>G
ENST00000622433.4:c.440A>G
ENST00000628176.2:c.432+20A>G
NM_001110792.1:c.488A>G
NM_001316337.1:c.173A>G
NM_004992.3:c.452A>G
NM_001316337.2:c.173A>G
NM_001369391.2:c.173A>G
NM_001369392.2:c.173A>G
NM_001369393.2:c.173A>G
NM_001369394.1:c.173A>G
NM_001369394.2:c.173A>G
NM_001386137.1:c.-129+20A>G
NM_001386138.1:c.-129+20A>G
NM_001386139.1:c.-129+20A>G
NM_004992.4:c.452A>G

Likely Pathogenic

Met criteria codes 5

PM2_Supporting | PS4_Moderate | PP3 | PP4 | PM1

Evidence Links 1

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.Asp151Gly variant in MECP2 (NM_004992.3) has been reported in an individual with a clinical phenotype suggestive of Rett syndrome (PMID 15173251) (PP4). The p.Asp151Gly variant has been observed in at least 3 other individuals with atypical Rett syndrome or clinical features of Rett syndrome (PMID 15173251, 32472557, internal database - University of Chicago, internal database - Invitae) (PS4_Moderate). The p.Asp151Gly variant occurs in the well-characterized Methyl-DNA binding (MDB) functional domain of MECP2 (PM1). The p.Asp151Gly 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.Asp151Gly variant in MECP2 is classified as Likely Pathogenic based on the ACMG/AMP criteria (PS4_moderate, PM1, PM2_supporting, PP3, PP4).

Met criteria codes

PM2_Supporting	✔	The p.Asp151Gly variant in MECP2 is absent from gnomAD
PS4_Moderate	✔	The p.Asp151Gly variant has been observed in at least 3 other individuals with either atypical Rett syndrome or clinical features of Rett syndrome (PMID 15173251, 32472557, internal database - University of Chicago, internal database - Invitae) (PS4_moderate). PS4_supporting PubMed:12180070
PP3	✔	Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own
PP4	✔	The p.Asp151Gly variant in MECP2 (NM_004992.3) has been reported in an individual with a clinical phenotype suggestive of Rett syndrome (PMID 15173251) (PP4).
PM1	✔	The p.Asp151Gly variant occurs in the well-characterized Methyl-DNA binding (MDB) functional domain of MECP2

Curation History

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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.