

Variant: *NM_001110792.2(MECP2):c.553C>G (p.Pro185Ala)*

Version: 1.0

CA270458 [↗](#)

143608 (ClinVar) [↗](#)

Gene: MECP2 ([HGNC:4204](#))

Condition: Rett syndrome ([MONDO:0010726](#))

Inheritance Mode: X-linked inheritance

UUID: 08f6e1b8-034e-4d11-b2f0-d9e0543e6161

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HGVS expressions

NM_001110792.2(MECP2):c.553C>G (p.Pro185Ala)

NM_001110792.2:c.553C>G

NC_000023.11:g.154031311G>C

CM000685.2:g.154031311G>C

NC_000023.10:g.153296762G>C

CM000685.1:g.153296762G>C

NC_000023.9:g.152949956G>C

NG_007107.2:g.110817C>G

NG_007107.3:g.110793C>G

ENST00000303391.11:c.517C>G

ENST00000453960.7:c.553C>G

ENST00000637917.1:c.65+85C>G

ENST00000303391.10:c.517C>G

ENST00000407218.5:c.469-25C>G

ENST00000453960.6:c.553C>G

ENST00000486506.5:n.2865C>G

ENST00000611468.1:c.503C>G

ENST00000619732.4:c.517C>G

ENST00000622433.4:c.505C>G

ENST00000628176.2:c.433-25C>G

NM_001110792.1:c.553C>G

NM_001316337.1:c.238C>G

NM_004992.3:c.517C>G

NM_001316337.2:c.238C>G

NM_001369391.2:c.238C>G

NM_001369392.2:c.238C>G

NM_001369393.2:c.238C>G

NM_001369394.1:c.238C>G

NM_001369394.2:c.238C>G

NM_001386137.1:c.-128-25C>G

NM_001386138.1:c.-128-25C>G

NM_001386139.1:c.-128-25C>G

NM_004992.4:c.517C>G

Likely Benign

BP5

BS2

Not Met criteria codes 5

BP4

BS1

PP4

PM2

PM1

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.Pro173Ala variant in MECP2 (NM_004992.3) has been reported in an individual with a clinical phenotype suggestive of Rett syndrome; this individual was reported to have a second variant in MECP2 that was classified as Pathogenic (PMID 11241840). The p.Pro173Ala variant in MECP2 (NM_004992.3) is observed in at least 2 unaffected individuals (internal database - GeneDx; internal database - Invitae) (BS2). The p.Pro173Ala variant is found in at least 2 patients with an alternate molecular basis of disease (internal database - GeneDx; internal database - Invitae) (BP5). The p.Pro173Ala variant in MECP2 is present in 3 female and 2 male individual(s) in gnomAD (0.0024%) (not sufficient to meet BS1 criteria). In summary, the p.Pro173Ala variant in MECP2 is classified as Likely Benign based on the ACMG/AMP criteria (BS2, BP5).

Met criteria codes

BP5			The p.Pro173Ala variant is found in at least 2 patients with an alternate molecular basis of disease (internal database - GeneDx; internal database - Invitae) (BP5).
BS2			The p.Pro173Ala variant in MECP2 (NM_004992.3) is observed in at least 2 unaffected individuals (internal database - GeneDx; internal database - Invitae) (BS2).

Not Met criteria codes

BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			The p.Pro173Ala variant in MECP2 is present in 3 female and 2 male individual(s) in gnomAD (0.0024%) (not sufficient to meet BS1 criteria).
PP4			The p.Pro173Ala variant in MECP2 has been reported in an individual with a clinical phenotype suggestive of Rett syndrome; this individual was reported to have a second variant in MECP2 that was classified as Pathogenic (PMID 11241840).
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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