

Variant: *NM_001110792.2(MECP2):c.564C>G (p.Pro188=)*

Version: 1.0

CA232982 [↗](#)

143612 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 70f1aa13-fba0-4de2-a067-4a5181b1c1d3

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HGVS expressions

NM_001110792.2:c.564C>G

NM_001110792.2(MECP2):c.564C>G (p.Pro188=)

NC_000023.11:g.154031300G>C

CM000685.2:g.154031300G>C

NC_000023.10:g.153296751G>C

CM000685.1:g.153296751G>C

NC_000023.9:g.152949945G>C

NG_007107.2:g.110828C>G

NG_007107.3:g.110804C>G

ENST00000303391.11:c.528C>G

ENST00000453960.7:c.564C>G

ENST00000637917.1:c.65+96C>G

ENST00000303391.10:c.528C>G

ENST00000407218.5:c.469-14C>G

ENST00000453960.6:c.564C>G

ENST00000486506.5:n.2876C>G

ENST00000619732.4:c.528C>G

ENST00000622433.4:c.516C>G

ENST00000628176.2:c.433-14C>G

NM_001110792.1:c.564C>G

NM_001316337.1:c.249C>G

NM_004992.3:c.528C>G

NM_001316337.2:c.249C>G

NM_001369391.2:c.249C>G

NM_001369392.2:c.249C>G

NM_001369393.2:c.249C>G

NM_001369394.1:c.249C>G

NM_001369394.2:c.249C>G

NM_001386137.1:c.-128-14C>G

NM_001386138.1:c.-128-14C>G

NM_001386139.1:c.-128-14C>G

NM_004992.4:c.528C>G

Likely Benign

Met criteria codes 3

BS2BP7BP4

Not Met criteria codes 1

PP4

Evidence Links 0

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 c.528C>G (p.Pro176=) variant in MECP2 (NM_004992.3) is observed in at least 2 unaffected individuals (internal database - Invitae) (BS2). The silent p.Pro176= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP4, BP7). The p.Pro176= variant in MECP2 is present in gnomAD v2.1.1 at a frequency of 0.003275% (no criteria met). In summary, the p.Pro176= variant in MECP2 is classified as Likely Benign based on the ACMG/AMP criteria (BS2, BP4, BP7).

Met criteria codes

BS2

i

✓

The c.528C>G (p.Pro176=) variant is observed in at least 2 unaffected individuals (internal database - Invitae) (BS2).

BP7

i

✓

The silent p.Pro176= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7).

BP4

i

✓

Splice prediction analysis, using multiple computational tools does not suggest an impact to splicing (BP4).

Not Met criteria codes

PP4

i

✗

The p.Pro176= variant in MECP2 was identified in a male with non-specific intellectual disability (PMID: 12111644).

Curation History

Showing 1 to 1 of 1 rows

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