

Variant: NM_001110792.2(MECP2):c.189C>G (p.His63Gln)

Version: 1.0

CA170260 [↗](#)

143493 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 9df8c8dd-156f-4007-98f1-db9a11397fc3

Approved on: 2025/5/7

Published on: 2025/6/30

HGVS expressions

NM_001110792.2:c.189C>G

NM_001110792.2(MECP2):c.189C>G (p.His63Gln)

NC_000023.11:g.154032431G>C

CM000685.2:g.154032431G>C

NC_000023.10:g.153297882G>C

CM000685.1:g.153297882G>C

NC_000023.9:g.152951076G>C

NG_007107.2:g.109697C>G

NG_007107.3:g.109673C>G

ENST00000303391.11:c.153C>G

ENST00000453960.7:c.189C>G

ENST00000303391.10:c.153C>G

ENST00000369957.5:c.*207C>G

ENST00000407218.5:c.189C>G

ENST00000453960.6:c.189C>G

ENST00000486506.5:n.2501C>G

ENST00000496908.5:n.284C>G

ENST00000611468.1:c.141C>G

ENST00000619732.4:c.153C>G

ENST00000622433.4:c.141C>G

ENST00000628176.2:c.153C>G

ENST00000631210.1:n.432C>G

NM_001110792.1:c.189C>G

NM_001316337.1:c.-127C>G

NM_004992.3:c.153C>G

NM_001316337.2:c.-127C>G

NM_001369391.2:c.-127C>G

NM_001369392.2:c.-127C>G

NM_001369393.2:c.-127C>G

NM_001369394.1:c.-127C>G

NM_001369394.2:c.-127C>G

NM_001386137.1:c.-408C>G

NM_001386138.1:c.-408C>G

NM_001386139.1:c.-408C>G

NM_004992.4:c.153C>G

Uncertain Significance

Met criteria codes 2

BS2_SupportingPM2_Supporting

Not Met criteria codes 1

BP4

Evidence Links 0

Expert Panel

Rett and Angelman-like Disorders VCEP

Criteria Specification Information

Criteria Specification: ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MECP2 Version 4.1.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.His51Gln variant in MECP2 (NM_004992.4) is absent from gnomAD v4.1 (PM2_Supporting). However, the p.His51Gln variant is observed in at least 1 unaffected individual (PMID 21300488) (BS2_supporting). Computational prediction analysis tools are inconclusive for this variant. In summary, the p.His51Gln variant in MECP2 is classified as a variant of uncertain significance based on the ACMG/AMP criteria (PM2_supporting, BS2_supporting). (MECP2 Specifications v.4.1; curation approved on [5/7/2025])

Met criteria codes

BS2_Supporting

The p.His51Gln variant is observed in at least 1 unaffected individual (PMID 21300488).

PM2_Supporting

The p.His51Gln variant in MECP2 is absent from gnomAD v4.1 (PM2_Supporting).

Not Met criteria codes

BP4

Computational prediction analysis tools are inconclusive for this variant.

Curation History

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