

Variant: NM_001110792.2(MECP2):c.204C>T (p.Pro68=)

Version: 1.0

CA170266 [↗](#)

143496 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: db56b560-de13-477b-a83b-1a24aa5b5934

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HGVS expressions

NM_001110792.2:c.204C>T

NM_001110792.2(MECP2):c.204C>T (p.Pro68=)

NC_000023.11:g.154032416G>A

CM000685.2:g.154032416G>A

NC_000023.10:g.153297867G>A

CM000685.1:g.153297867G>A

NC_000023.9:g.152951061G>A

NG_007107.2:g.109712C>T

NG_007107.3:g.109688C>T

ENST00000303391.11:c.168C>T

ENST00000453960.7:c.204C>T

ENST00000303391.10:c.168C>T

ENST00000369957.5:c.*222C>T

ENST00000407218.5:c.204C>T

ENST00000453960.6:c.204C>T

ENST00000486506.5:n.2516C>T

ENST00000496908.5:n.299C>T

ENST00000611468.1:c.156C>T

ENST00000619732.4:c.168C>T

ENST00000622433.4:c.156C>T

ENST00000628176.2:c.168C>T

ENST00000631210.1:n.447C>T

NM_001110792.1:c.204C>T

NM_001316337.1:c.-112C>T

NM_004992.3:c.168C>T

NM_001316337.2:c.-112C>T

NM_001369391.2:c.-112C>T

NM_001369392.2:c.-112C>T

NM_001369393.2:c.-112C>T

NM_001369394.1:c.-112C>T

NM_001369394.2:c.-112C>T

NM_001386137.1:c.-393C>T

NM_001386138.1:c.-393C>T

NM_001386139.1:c.-393C>T

NM_004992.4:c.168C>T

Benign

Met criteria codes 1

BA1

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 allele frequency of the p.Pro56= (NM_004992.3) variant in MECP2 is 0.231% in South Asian sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BA1). In summary, the p.Pro56= variant in MECP2 is classified as benign based on the ACMG/AMP criteria (BA1).

Met criteria codes

BA1

The allele frequency of the p.Pro56= variant in MECP2 is 0.231% in South Asian sub population in gnomAD, which is high enough to be classified as benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions.

Curation History

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