

*Variant: NM_001110792.2(MECP2):c.1096C>T
(p.Arg366Cys)*

Version: 1.0

CA206493 [↗](#)

158880 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 9a0ddb9b-0b82-4d80-9cde-6793c04b5f7e

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HGVS expressions

NM_001110792.2:c.1096C>T

NM_001110792.2(MECP2):c.1096C>T (p.Arg366Cys)

NC_000023.11:g.154030768G>A

CM000685.2:g.154030768G>A

NC_000023.10:g.153296219G>A

CM000685.1:g.153296219G>A

NC_000023.9:g.152949413G>A

NG_007107.2:g.111360C>T

NG_007107.3:g.111336C>T

ENST00000303391.11:c.1060C>T

ENST00000453960.7:c.1096C>T

ENST00000303391.10:c.1060C>T

ENST00000407218.5:c.*432C>T

ENST00000453960.6:c.1096C>T

ENST00000619732.4:c.1060C>T

ENST00000628176.2:c.*432C>T

NM_001110792.1:c.1096C>T

NM_001316337.1:c.781C>T

NM_004992.3:c.1060C>T

NM_001316337.2:c.781C>T

NM_001369391.2:c.781C>T

NM_001369392.2:c.781C>T

NM_001369393.2:c.781C>T

NM_001369394.1:c.781C>T

NM_001369394.2:c.781C>T

NM_001386137.1:c.391C>T

NM_001386138.1:c.391C>T

NM_001386139.1:c.391C>T

NM_004992.4:c.1060C>T

Benign

Met criteria codes **2**

BS1

BS2

Evidence Links **0**

Expert Panel

Rett and Angelman-like Disorders VCEP [↗](#)

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The allele frequency of the p.Arg354Cys variant in MECP2 (NM_004992.3) is 0.028% in East Asian sub population in gnomAD, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1). The p.Arg354Cys variant is observed in at least 2 unaffected individuals (internal database) (BS2). In summary, the p.Arg354Cys variant in MECP2 is classified as Benign based on the ACMG/AMP criteria (BS1, BS2).

Met criteria codes

BS1			The allele frequency of the p.Arg354Cys variant in MECP2 (NM_004992.3) is 0.028% in East Asian sub population in gnomAD, which is high enough to meet the BS1 criteria based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1).
BS2			The p.Arg354Cys variant in MECP2 (NM_004992.3) is observed in at least 2 unaffected individuals (internal database) (BS2).

Curation History



 

 

Showing 1 to 1 of 1 rows

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