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Variant: NM_001110792.2(MECP2):c.598C>T (p.Arg200Trp)

Version: 1.0

CA10558590

566728 (ClinVar)

Gene: MECP2 (HGNC:4204)
Condition: Rett syndrome (MONDO:0010726)
Inheritance Mode: X-linked inheritance
UUID: d124c3fa-fce0-4e64-9d18-cbababd9df9d
Approved on: 2026/4/23
Published on: 2026/6/26

HGVS expressions

NM_001110792.2:c.598C>T
NM_001110792.2(MECP2):c.598C>T (p.Arg200Trp)
NC_000023.11:g.154031266G>A
CM000685.2:g.154031266G>A
NC_000023.10:g.153296717G>A
CM000685.1:g.153296717G>A
NC_000023.9:g.152949911G>A
NG_007107.2:g.110862C>T
NG_007107.3:g.110838C>T
ENST00000303391.11:c.562C>T
ENST00000453960.7:c.598C>T
ENST00000637917.1:c.65+130C>T
ENST00000303391.10:c.562C>T
ENST00000407218.5:c.489C>T
ENST00000453960.6:c.598C>T
ENST00000619732.4:c.562C>T
ENST00000622433.4:c.550C>T
ENST00000628176.2:c.453C>T
NM_001110792.1:c.598C>T
NM_001316337.1:c.283C>T
NM_004992.3:c.562C>T
NM_001316337.2:c.283C>T
NM_001369391.2:c.283C>T
NM_001369392.2:c.283C>T
NM_001369393.2:c.283C>T
NM_001369394.1:c.283C>T
NM_001369394.2:c.283C>T
NM_001386137.1:c.-108C>T
NM_001386138.1:c.-108C>T
NM_001386139.1:c.-108C>T
NM_004992.4:c.562C>T

Benign

Met criteria codes 2

BS2 BS1

Expert Panel

Rett and Angelman-like Disorders VCEP

Evidence Links 0

Criteria Specification Information

Criteria Specification: *ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MECP2 Version 5.0.0*

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The highest population minor allele frequency of the p.Arg188Trp variant in MECP2 (NM_004992.4) in gnomAD v4.1 is 0.00001759 in South Asian population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥0.0000083) for BS1, and therefore meets this criterion (BS1). The p.Arg188Trp variant is observed in at least 2 unaffected individuals (internal database) (BS2). In summary, the p.Arg188Trp variant in MECP2 is classified as benign based on the ACMG/AMP criteria (BS1, BS2). (MECP2 Specifications v5.0; curation approved on 4/23/2026)

Met criteria codes

BS2	✔	The p.Arg188Trp variant in MECP2 (NM_004992.4) is observed in at least 2 unaffected individuals (internal database) (BS2).
BS1	✔	The highest population minor allele frequency of the p.Arg188Trp variant in MECP2 (NM_004992.4) in gnomAD v4.1 is 0.00001759 in South Asian population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥0.0000083) for BS1, and therefore meets this criterion (BS1).

Curation History

Showing 1 to 1 of 1 rows

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_001110792.2(MECP2):c.598C>T (p...	Benign	Rett Syndrome	2026-06-26	1.0	ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MECP2 Version 5.0.0	MECP2

Showing 1 to 1 of 1 rows

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