

Variant: *NM_001110792.2(MECP2):c.244T>C (p.Ser82Pro)*

Version: 1.0

CA415177504 [↗](#)

560190 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: c40905b6-de17-46c9-9639-e7f133ca5bda

Approved on: 2026/1/28

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HGVS expressions

NM_001110792.2(MECP2):c.244T>C (p.Ser82Pro)

NM_001110792.2:c.244T>C

NC_000023.11:g.154032376A>G

CM000685.2:g.154032376A>G

NC_000023.10:g.153297827A>G

CM000685.1:g.153297827A>G

NC_000023.9:g.152951021A>G

NG_007107.2:g.109752T>C

NG_007107.3:g.109728T>C

ENST00000303391.11:c.208T>C

ENST00000453960.7:c.244T>C

ENST00000303391.10:c.208T>C

ENST00000369957.5:c.*262T>C

ENST00000407218.5:c.244T>C

ENST00000453960.6:c.244T>C

ENST00000486506.5:n.2556T>C

ENST00000496908.5:n.339T>C

ENST00000611468.1:c.196T>C

ENST00000619732.4:c.208T>C

ENST00000622433.4:c.196T>C

ENST00000628176.2:c.208T>C

ENST00000631210.1:n.487T>C

NM_001110792.1:c.244T>C

NM_001316337.1:c.-72T>C

NM_004992.3:c.208T>C

NM_001316337.2:c.-72T>C

NM_001369391.2:c.-72T>C

NM_001369392.2:c.-72T>C

NM_001369393.2:c.-72T>C

NM_001369394.1:c.-72T>C

NM_001369394.2:c.-72T>C

NM_001386137.1:c.-353T>C

NM_001386138.1:c.-353T>C

NM_001386139.1:c.-353T>C

NM_004992.4:c.208T>C

Uncertain Significance

Met criteria codes 2

PM2_SupportingBS2

Not Met criteria codes 3

PS4PP3PM1

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 5.0.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Ser70Pro variant in MECP2 (NM_004992.4) is observed in at least 2 unaffected individuals (Labcorp (formerly Invitae)- internal database, GeneDx-internal database) and has not been previously published as associated with disease (BS2). The p.Ser70Pro variant in MECP2 is absent from gnomAD v4.1 (PM2_Supporting). Computational prediction analysis tools are inconclusive for this variant. In summary, the p.Ser70Pro variant in MECP2 is classified as a variant of uncertain significance based on the ACMG/AMP criteria (BS2, PM2_Supporting). (MECP2 Specifications v.5.0; curation approved on 01/28/2026).

Met criteria codes

PM2_Supporting

The p.Ser70Pro variant in MECP2 is absent from gnomAD v4.1 (PM2_Supporting).

BS2

The p.Ser70Pro variant is observed in at least 2 unaffected individuals (Labcorp (formerly Invitae) - internal database) (GeneDx - internal database).

Not Met criteria codes

PS4

Identified in one proband from GeneDx with developmental delay and behavioral issues, inherited from a parent without a MECP2-related phenotype.

PP3

REVEL = 0.536. Computational prediction analysis tools are inconclusive for this variant.

PM1

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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