

Variant: *NM_001110792.2(MECP2):c.686C>T (p.Pro229Leu)*

Version: 2.0

CA10581615 [↗](#)

236302 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: f2c5bce1-28a5-423f-911e-2ae5970fcbb4

Approved on: 2024/10/30

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HGVS expressions

NM_001110792.2:c.686C>T

NM_001110792.2(MECP2):c.686C>T (p.Pro229Leu)

NC_000023.11:g.154031178G>A

CM000685.2:g.154031178G>A

NC_000023.10:g.153296629G>A

CM000685.1:g.153296629G>A

NC_000023.9:g.152949823G>A

NG_007107.2:g.110950C>T

NG_007107.3:g.110926C>T

ENST00000303391.11:c.650C>T

ENST00000453960.7:c.686C>T

ENST00000637917.1:c.65+218C>T

ENST00000303391.10:c.650C>T

ENST00000407218.5:c.*22C>T

ENST00000453960.6:c.686C>T

ENST00000619732.4:c.650C>T

ENST00000622433.4:c.638C>T

ENST00000628176.2:c.*22C>T

NM_001110792.1:c.686C>T

NM_001316337.1:c.371C>T

NM_004992.3:c.650C>T

NM_001316337.2:c.371C>T

NM_001369391.2:c.371C>T

NM_001369392.2:c.371C>T

NM_001369393.2:c.371C>T

NM_001369394.1:c.371C>T

NM_001369394.2:c.371C>T

NM_001386137.1:c.-20C>T

NM_001386138.1:c.-20C>T

NM_001386139.1:c.-20C>T

NM_004992.4:c.650C>T

Likely Pathogenic

Met criteria codes **3**

PS2

PS4_Moderate

PM2_Supporting

Expert Panel

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

- [Criteria Specification: ClinGen Rett and Angelman-like Disorders Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MECP2 Version 3.0.0](#)
- [Criteria Specification Approval History](#)
- [Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Pro217Leu variant in MECP2 (NM_004992.3) has been reported as a de novo occurrence (biological parentage both confirmed and unconfirmed) in 2 individuals with neurodevelopmental disorders (PMID: 27824329, LabCorp) (PS2). The p.Pro217Leu variant has been observed in individuals with neurodevelopmental disorders (PMID: 27824329, LabCorp, Invitae internal database) (PS4_moderate). The p.Pro217Leu in MECP2 is absent from gnomAD (PM2_supporting). In summary, the p.Pro217Leu variant in MECP2 is classified as likely pathogenic for Rett syndrome based on the ACMG/AMP criteria (PS2, PS4_moderate, PM2_supporting).

Met criteria codes

PS2			The p.Pro217Leu variant in MECP2 (NM_004992.3) has been reported as a de novo occurrence (biological parentage both confirmed and unconfirmed) in 2 individuals with neurodevelopmental disorders (PMID: 27824329, LabCorp)
PS4_Moderate			The p.Pro217Leu variant has been observed in individuals with neurodevelopmental disorders (PMID: 27824329, LabCorp, Invitae internal database) (PS4_moderate).
PM2_Supporting			The p.Pro217Leu in MECP2 (NM_004992.4) is absent from gnomAD.

Curation History [↗](#)

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Showing 1 to 2 of 2 rows

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