

Variant: *NM_001110792.2(MECP2):c.1207C>G (p.Pro403Ala)*

Version: 1.0

CA10558486 [↗](#)

426939 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 3c702aa7-fe97-4b1d-8e66-7943d58146be

Approved on: 2024/8/30

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HGVS expressions

NM_001110792.2(MECP2):c.1207C>G (p.Pro403Ala)

NM_001110792.2:c.1207C>G

NC_000023.11:g.154030657G>C

CM000685.2:g.154030657G>C

NC_000023.10:g.153296108G>C

CM000685.1:g.153296108G>C

NC_000023.9:g.152949302G>C

NG_007107.2:g.111471C>G

NG_007107.3:g.111447C>G

ENST00000303391.11:c.1171C>G

ENST00000453960.7:c.1207C>G

ENST00000303391.10:c.1171C>G

ENST00000407218.5:c.*543C>G

ENST00000453960.6:c.1207C>G

ENST00000619732.4:c.1171C>G

ENST00000628176.2:c.*543C>G

NM_001110792.1:c.1207C>G

NM_001316337.1:c.892C>G

NM_004992.3:c.1171C>G

NM_001316337.2:c.892C>G

NM_001369391.2:c.892C>G

NM_001369392.2:c.892C>G

NM_001369393.2:c.892C>G

NM_001369394.1:c.892C>G

NM_001369394.2:c.892C>G

NM_001386137.1:c.502C>G

NM_001386138.1:c.502C>G

NM_001386139.1:c.502C>G

NM_004992.4:c.1171C>G

Likely Benign

Met criteria codes **1**

BS2

Not Met criteria codes **2**

BP4

PP3

Expert Panel

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

Criteria Specification Information

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Pro391Ala variant in MECP2 (NM_004992.4) is observed in at least 2 unaffected individuals (internal database - Invitae and GeneDx) (BS2). The p.Pro391Ala variant is not currently published and is not present in additional databases (internal and publicly available), therefore, no additional criteria are applicable at this time. In the absence of conflicting evidence, this is sufficient evidence to classify as likely benign based on the specifications defined by the ClinGen Rett and Angelman-like Disorders Variant Curation Expert Panel. In summary, the p.Pro391Ala variant in MECP2 (NM_004992.4) is classified as likely benign for Rett syndrome based on the ACMG/AMP criteria (BS2). (MECP2 specification v.3.0.0, curation approved on 8/30/2024)

Met criteria codes

BS2			The p.Pro391Ala variant in MECP2 (NM_004992.4) is observed in at least 2 unaffected individuals (internal database - Invitae and GeneDx) (BS2).
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Not Met criteria codes

BP4			Computational prediction analysis tools are inconclusive for this variant (no criteria met).
PP3			Computational prediction analysis tools are inconclusive for this variant (no criteria met).

Curation History [↗](#)



 

 

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