

Variant: *NM_001110792.2(MECP2):c.1244C>G*
(*p.Pro415Arg*)

Version: 1.0

CA415166876 [↗](#)

1212000 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 569b7d06-0464-4678-af06-a301f3670b83

Approved on: 2023/12/6

Published on: 2024/10/25

HGVS expressions

NM_001110792.2(MECP2):c.1244C>G (p.Pro415Arg)

NM_001110792.2:c.1244C>G

NC_000023.11:g.154030620G>C

CM000685.2:g.154030620G>C

NC_000023.10:g.153296071G>C

CM000685.1:g.153296071G>C

NC_000023.9:g.152949265G>C

NG_007107.2:g.111508C>G

NG_007107.3:g.111484C>G

ENST00000303391.11:c.1208C>G

ENST00000453960.7:c.1244C>G

ENST00000303391.10:c.1208C>G

ENST00000407218.5:c.*580C>G

ENST00000453960.6:c.1244C>G

ENST00000619732.4:c.1208C>G

ENST00000628176.2:c.*580C>G

NM_001110792.1:c.1244C>G

NM_001316337.1:c.929C>G

NM_004992.3:c.1208C>G

NM_001316337.2:c.929C>G

NM_001369391.2:c.929C>G

NM_001369392.2:c.929C>G

NM_001369393.2:c.929C>G

NM_001369394.1:c.929C>G

NM_001369394.2:c.929C>G

NM_001386137.1:c.539C>G

NM_001386138.1:c.539C>G

NM_001386139.1:c.539C>G

NM_004992.4:c.1208C>G

Likely Benign

Met criteria codes **1**

BS2

Not Met criteria codes **3**

Expert Panel

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

BA1 BS1 PM2

BS1

PM2

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 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.Pro403Arg variant in MECP2 (NM_004992.4) is present in 3 XX and 2 XY individual(s) in gnomAD (0.0003564%) (not sufficient to meet BS1 criteria). The p.Pro403Arg variant is observed in 3 unaffected individuals (internal database - GeneDx) (BS2). 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.Pro403Arg variant in MECP2 (NM_004992.4) is classified as likely benign based on the ACMG/AMP criteria (BS2).

Met criteria codes

BS2



The p.Pro415Arg variant is observed in at least 2 unaffected individuals (internal database) (BS2).

Not Met criteria codes

BA1



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

BS1



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

PM2



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

Curation History



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