

Variant: *NM_001110792.2(MECP2):c.1440G>A (p.Arg480=)*

Version: 1.0

CA199303 [↗](#)

138196 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 090f55c0-5f35-4dde-8462-b853758a24c0

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HGVS expressions

NM_001110792.2:c.1440G>A

NM_001110792.2(MECP2):c.1440G>A (p.Arg480=)

NC_000023.11:g.154030424C>T

CM000685.2:g.154030424C>T

NC_000023.10:g.153295875C>T

CM000685.1:g.153295875C>T

NC_000023.9:g.152949069C>T

NG_007107.2:g.111704G>A

NG_007107.3:g.111680G>A

ENST00000303391.11:c.1404G>A

ENST00000453960.7:c.1440G>A

ENST00000303391.10:c.1404G>A

ENST00000453960.6:c.1440G>A

ENST00000619732.4:c.1404G>A

ENST00000628176.2:c.*776G>A

NM_001110792.1:c.1440G>A

NM_001316337.1:c.1125G>A

NM_004992.3:c.1404G>A

NM_001316337.2:c.1125G>A

NM_001369391.2:c.1125G>A

NM_001369392.2:c.1125G>A

NM_001369393.2:c.1125G>A

NM_001369394.1:c.1125G>A

NM_001369394.2:c.1125G>A

NM_001386137.1:c.735G>A

NM_001386138.1:c.735G>A

NM_001386139.1:c.735G>A

NM_004992.4:c.1404G>A

Likely Benign

Met criteria codes **3**

BP4 BP5 BP7

Evidence Links **0**

Expert Panel

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

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Rett and Angelman-like Disorders VCEP

The silent p.Arg468= variant in MECP2 (NM_004992) is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). The p.Arg468= variant is found in two patients with an alternate molecular basis of disease (INVITAE internal cases) (BP5). Splice prediction analysis, using multiple computational tools does not suggest an impact to splicing (BP4). In summary the p.Arg468= variant in MECP2 is classified as Likely Benign for Rett syndrome disorder based on the ACMG/AMP criteria (BP7, BP4, BP5).

Met criteria codes

BP4			Splice prediction analysis, using multiple computational tools does not suggest an impact to splicing BP4
BP5			The p.(Arg468=) variant is found in two patients with an alternate molecular basis of disease (Invitae internal cases) (BP5)
BP7			The silent p.(Arg468=) variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide BP7

Curation History



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Showing 1 to 1 of 1 rows

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