

Variant: *NM_004992.3(MECP2):c.1140G>A (p.Val380=)*

Version: 1.0

CA208360 [↗](#)

138194 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 7277ad9e-b07e-4eeb-a2db-80b1b1fe619f

Approved on: 2021/3/26

Published on: 2021/5/17

HGVS expressions

NM_004992.3(MECP2):c.1140G>A (p.Val380=)

NM_004992.3:c.1140G>A

NC_000023.11:g.154030688C>T

CM000685.2:g.154030688C>T

NC_000023.10:g.153296139C>T

CM000685.1:g.153296139C>T

NC_000023.9:g.152949333C>T

NG_007107.2:g.111440G>A

NG_007107.3:g.111416G>A

ENST00000303391.11:c.1140G>A

ENST00000453960.7:c.1176G>A

ENST00000303391.10:c.1140G>A

ENST00000407218.5:c.*512G>A

ENST00000453960.6:c.1176G>A

ENST00000619732.4:c.1140G>A

ENST00000628176.2:c.*512G>A

NM_001110792.1:c.1176G>A

NM_001316337.1:c.861G>A

NM_001110792.2:c.1176G>A

NM_001316337.2:c.861G>A

NM_001369391.2:c.861G>A

NM_001369392.2:c.861G>A

NM_001369393.2:c.861G>A

NM_001369394.1:c.861G>A

NM_001369394.2:c.861G>A

NM_001386137.1:c.471G>A

NM_001386138.1:c.471G>A

NM_001386139.1:c.471G>A

NM_004992.4:c.1140G>A

Benign

Met criteria codes 3

BS1 BS2 BP7

Not Met criteria codes 1

BA1

Expert Panel

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

Criteria Specification Information **!**

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The allele frequency of the p.Val380= variant in MECP2 is 0.018% in Latino sub population in gnomAD, which is high enough to be classified as likely benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions (BS1). The p.Val380= variant is observed in at least 2 unaffected individuals (internal database) (BS2). The silent p.Val380= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide (BP7). In summary, the p.Val380= variant in MECP2 is classified as benign based on the ACMG/AMP criteria (BS1, BS2, BP7).

Met criteria codes

BS1			The allele frequency of the p.Val380= variant in MECP2 is 0.018% in Latino sub population in gnomAD, which is high enough to be classified as likely benign based on thresholds defined by the ClinGen Rett/Angelman-like Expert Panel for Rett/AS-like conditions
BS2			The p.Val380= variant is observed in at least 2 unaffected individuals (internal database)
BP7			The silent p.Val380= variant is not predicted to affect splicing using multiple computational tools and does not affect a highly conserved nucleotide

Not Met criteria codes

BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
-----	---	---	--

Curation History [↗](#)



 

 

Showing 1 to 1 of 1 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

[ClinGen Terms of Use.](#)

[✕ Powered by BCM's Genboree.](#)