

Variant: *NM_001110792.2(MECP2):c.1458T>C (p.Pro486=)*

Version: 1.0

CA519704534 [↗](#)

516510 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 4b9145aa-2ae7-4e22-93bb-533ed5e9a32d

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HGVS expressions

NM_001110792.2(MECP2):c.1458T>C (p.Pro486=)

NM_001110792.2:c.1458T>C

NC_000023.11:g.154030406A>G

CM000685.2:g.154030406A>G

NC_000023.10:g.153295857A>G

CM000685.1:g.153295857A>G

NC_000023.9:g.152949051A>G

NG_007107.2:g.111722T>C

NG_007107.3:g.111698T>C

ENST00000303391.11:c.1422T>C

ENST00000453960.7:c.1458T>C

ENST00000303391.10:c.1422T>C

ENST00000453960.6:c.1458T>C

ENST00000619732.4:c.1422T>C

ENST00000628176.2:c.*794T>C

NM_001110792.1:c.1458T>C

NM_001316337.1:c.1143T>C

NM_004992.3:c.1422T>C

NM_001316337.2:c.1143T>C

NM_001369391.2:c.1143T>C

NM_001369392.2:c.1143T>C

NM_001369393.2:c.1143T>C

NM_001369394.1:c.1143T>C

NM_001369394.2:c.1143T>C

NM_001386137.1:c.753T>C

NM_001386138.1:c.753T>C

NM_001386139.1:c.753T>C

NM_004992.4:c.1422T>C

Likely Benign

Met criteria codes **3**

BP5

BP4

BS2_Supporting

Not Met criteria codes **2**

BS1

PM2

Expert Panel

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

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The highest population minor allele frequency of the p.Pro474= variant in MECP2 (NM_004992.3) in gnomAD v4.1 is 0.000008935 in European (Non-Finnish) population (not sufficient to meet BS1 criteria). The p.Pro474= variant is observed in at least 1 unaffected individual (GeneDx internal database) (BS2_supporting). The p.Pro474= variant is found in a patient with an alternate molecular basis of disease (GeneDx internal database) (BP5). Splice prediction analysis does not suggest an impact to splicing (BP4). In summary, the p.Pro474= variant in MECP2 is classified as likely benign based on the ACMG/AMP criteria (BS2_supporting, BP5, BP4).

Met criteria codes

BP5			The p.Pro474= variant in MECP2 (NM_004992.3) is found in a patient with an alternate molecular basis of disease (GeneDx internal database).
BP4			Splice prediction analysis, using multiple computational tools does not suggest an impact to splicing (BP4).
BS2_Supporting			The p.Pro474= variant in MECP2 (NM_004992.3) is observed in at least 1 unaffected individual (GeneDx internal database).

Not Met criteria codes

BS1			The highest population minor allele frequency of the p.Pro474= variant in MECP2 (NM_004992.3) in gnomAD v4.1 is 0.000008935 in European (Non-Finnish) population (not sufficient to meet BS1 criteria).
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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