

Variant: NM_001110792.2(MECP2):c.553C>A (p.Pro185Thr)

Version: 1.0

CA415173958 [↗](#)

804125 (ClinVar) [↗](#)

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

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

Inheritance Mode: X-linked inheritance

UUID: 620fe876-df5e-4691-833d-2490e0485e3e

Approved on: 2024/8/30

Published on: 2024/12/23

HGVS expressions

NM_001110792.2:c.553C>A

NM_001110792.2(MECP2):c.553C>A (p.Pro185Thr)

NC_000023.11:g.154031311G>T

CM000685.2:g.154031311G>T

NC_000023.10:g.153296762G>T

CM000685.1:g.153296762G>T

NC_000023.9:g.152949956G>T

NG_007107.2:g.110817C>A

NG_007107.3:g.110793C>A

ENST00000303391.11:c.517C>A

ENST00000453960.7:c.553C>A

ENST00000637917.1:c.65+85C>A

ENST00000303391.10:c.517C>A

ENST00000407218.5:c.469-25C>A

ENST00000453960.6:c.553C>A

ENST00000486506.5:n.2865C>A

ENST00000611468.1:c.503C>A

ENST00000619732.4:c.517C>A

ENST00000622433.4:c.505C>A

ENST00000628176.2:c.433-25C>A

NM_001110792.1:c.553C>A

NM_001316337.1:c.238C>A

NM_004992.3:c.517C>A

NM_001316337.2:c.238C>A

NM_001369391.2:c.238C>A

NM_001369392.2:c.238C>A

NM_001369393.2:c.238C>A

NM_001369394.1:c.238C>A

NM_001369394.2:c.238C>A

NM_001386137.1:c.-128-25C>A

NM_001386138.1:c.-128-25C>A

NM_001386139.1:c.-128-25C>A

NM_004992.4:c.517C>A

Uncertain Significance

BS2_Supporting

Not Met criteria codes **6**

PS1

PM2

PM1

PM5

BA1

BS1

Evidence Links 0

Rett and Angelman-like Disorders VCEP

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 highest population minor allele frequency of the p.Pro173Thr variant in MECP2 (NM_004992.4) in gnomAD v4.1 is 0.000002233 in European (non-Finnish) population (not sufficient to meet BS1 criteria). The p.Pro173Thr variant is observed in at least 1 unaffected individual (internal database - GeneDx) (BS2_Supporting). The p.Pro173Thr 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 summary, the p.Pro173Thr variant in MECP2 is classified as a variant of uncertain significance based on the ACMG/AMP criteria (BS2_Supporting). (MECP2 specification v3.0.0, curation approved on 8/30/2024)

Met criteria codes

BS2_Supporting			The p.Pro173Thr variant is observed in at least 1 unaffected individual (internal database - GeneDx) (BS2_Supporting).
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Not Met criteria codes

PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			The highest population minor allele frequency of the p.Pro173Thr variant in MECP2 (NM_004992.4) in gnomAD v4.1 is 0.000002233 in European (non-Finnish) population (not sufficient to meet PM2 criteria).
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			The highest population minor allele frequency of the p.Pro173Thr variant in MECP2 (NM_004992.4) in gnomAD v4.1 is 0.000002233 in European (non-Finnish) population (not sufficient to meet BA1 criteria).
BS1			The highest population minor allele frequency of the p.Pro173Thr variant in MECP2 (NM_004992.4) in gnomAD v4.1 is 0.000002233 in European (non-Finnish) population (not sufficient to meet BS1 criteria).

Curation History 

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