

Variant: NM_001110792.2(MECP2):c.377G>C (p.Gly126Ala)

Version: 1.0

CA270359 [↗](#)

143540 (ClinVar) [↗](#)

Gene: MECP2 ([HGNC:4204](#))
Condition: Rett syndrome ([MONDO:0010726](#))
Inheritance Mode: X-linked inheritance
UUID: 9ddf71e8-5a4c-405f-8f52-47ab776f6a69
Approved on: Mon, 24 Aug 2026 00:00:00 +0000
Published on: Thu, 01 Oct 2026 00:00:00 +0000

HGVS expressions

NM_001110792.2(MECP2):c.377G>C (p.Gly126Ala)
NM_001110792.2:c.377G>C
NC_000023.11:g.154032243C>G
CM000685.2:g.154032243C>G
NC_000023.10:g.153297694C>G
CM000685.1:g.153297694C>G
NC_000023.9:g.152950888C>G
NG_007107.2:g.109885G>C
NG_007107.3:g.109861G>C
ENST00000303391.11:c.341G>C
ENST00000453960.7:c.377G>C
ENST00000303391.10:c.341G>C
ENST00000369957.5:c.*395G>C
ENST00000407218.5:c.377G>C
ENST00000453960.6:c.377G>C
ENST00000486506.5:n.2689G>C
ENST00000611468.1:c.329G>C
ENST00000619732.4:c.341G>C
ENST00000622433.4:c.329G>C
ENST00000628176.2:c.341G>C
NM_001110792.1:c.377G>C
NM_001316337.1:c.62G>C
NM_004992.3:c.341G>C
NM_001316337.2:c.62G>C
NM_001369391.2:c.62G>C
NM_001369392.2:c.62G>C
NM_001369393.2:c.62G>C
NM_001369394.1:c.62G>C
NM_001369394.2:c.62G>C
NM_001386137.1:c.-220G>C
NM_001386138.1:c.-220G>C
NM_001386139.1:c.-220G>C
NM_004992.4:c.341G>C

Likely Pathogenic

PM2_SupportingPP3PM1PM6

Rett and Angelman-like Disorders VCEP

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 6.0
- Criteria Specification Approval History
- Criteria Specifications for this VCEP

Evidence submitted by expert panel

Rett and Angelman-like Disorders VCEP

The p.Gly114Ala variant in MECP2 (NM_004992.4) has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with clinical features of Rett syndrome (RettBASE) (PM6). The p.Gly114Ala variant occurs in the well-characterized methyl-DNA binding (MBD) functional domain of the MECP2 gene (PM1). The p.Gly114Ala variant in MECP2 is absent from gnomAD v4.1 (PM2_Supporting). Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3). In summary, the p.Gly114Ala variant in MECP2 is classified as Likely Pathogenic for Rett syndrome based on the ACMG/AMP criteria (PM1, PM6, PM2_Supporting, PP3). (MECP2 Specifications v6.0; curation approved on 8/24/2026)

Met criteria codes

PM2_Supporting			The p.Gly114Ala variant in MECP2 is absent from gnomAD v4.1 (PM2_Supporting).
PP3			REVEL 0.956 Computational prediction analysis tools suggests a deleterious impact; however, this information does not predict clinical significance on its own (PP3).
PM1			The p.Gly114Ala variant occurs in the well-characterized methyl-DNA binding (MBD) functional domain of the MECP2 gene (PM1).
PM6			The p.Gly114Ala variant in MECP2 (NM_004992.4) has been reported as a de novo occurrence (biological parentage unconfirmed) in an individual with clinical features of Rett syndrome (RettBASE) (PM6).

Curation History

Showing 1 to 1 of 1 rows

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