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# *Variant: NM_001110792.2(MECP2):c.1097G>T (p.Arg366Leu)*

Version: 1.0

*CA170157*

*143317 (ClinVar)*

**Gene**: MECP2 (HGNC:4204)
**Condition**: Rett syndrome (MONDO:0010726)
**Inheritance Mode**: X-linked inheritance
**UUID:** 36c5a87e-0e4c-45fe-a7b8-8c1788eef672
**Approved on**: 2025/5/7
**Published on**: 2025/6/30

## *HGVS expressions*

**NM_001110792.2(MECP2):c.1097G>T (p.Arg366Leu)**
**NM_001110792.2:c.1097G>T**
NC_000023.11:g.154030767C>A
CM000685.2:g.154030767C>A
NC_000023.10:g.153296218C>A
CM000685.1:g.153296218C>A
NC_000023.9:g.152949412C>A
NG_007107.2:g.111361G>T
NG_007107.3:g.111337G>T
ENST00000303391.11:c.1061G>T
ENST00000453960.7:c.1097G>T
ENST00000303391.10:c.1061G>T
ENST00000407218.5:c.*433G>T
ENST00000453960.6:c.1097G>T
ENST00000619732.4:c.1061G>T
ENST00000628176.2:c.*433G>T
NM_001110792.1:c.1097G>T
NM_001316337.1:c.782G>T
NM_004992.3:c.1061G>T
NM_001316337.2:c.782G>T
NM_001369391.2:c.782G>T
NM_001369392.2:c.782G>T
NM_001369393.2:c.782G>T
NM_001369394.1:c.782G>T
NM_001369394.2:c.782G>T
NM_001386137.1:c.392G>T
NM_001386138.1:c.392G>T
NM_001386139.1:c.392G>T
NM_004992.4:c.1061G>T

**Benign**

**Met criteria codes** 2

BS2 BS1

Evidence Links 0

Expert Panel

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 4.1.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

## Evidence submitted by expert panel

***Rett and Angelman-like Disorders VCEP***

**The p.Arg354Leu variant in MECP2 (NM_004992.4) is observed in at least 2 unaffected individuals (GeneDx internal database) (BS2). The highest population minor allele frequency of the p.Arg354Leu variant in MECP2 in gnomAD v4.1 is 0.0001252 in the European (non-Finnish) population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥0.00008) for BS1 and therefore meets this criterion (BS1). In summary, the p.Arg354Leu variant in MECP2 is classified as benign based on the ACMG/AMP criteria (BS1, BS2). (MECP2 Specifications v.4.1; curation approved on [5/7/2025])**

**Met criteria codes**

| | | |
|---|---|---|
| **BS2** | ✔ | The p.Arg354Leu variant in MECP2 (NM_004992.4) is observed in at least 2 unaffected individuals (GeneDx internal database) (BS2). |
| **BS1** | ✔ | The highest population minor allele frequency of the p.Arg354Leu variant in MECP2 (NM_004992.4) in gnomAD v4.1 is 0.0001252 in the European (non-Finnish) population, which is higher than the ClinGen Rett and Angelman-like Disorders VCEP threshold (≥0.00008) for BS1, and therefore meets this criterion (BS1). |

## Curation History

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