The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000051.4(ATM):c.3137T>C (p.Leu1046Pro)

Version: 2.0

CA195169

186558 (ClinVar)

Gene: ATM (HGNC:472)
Condition: ATM-related cancer predisposition (MONDO:0700270)
Inheritance Mode: Autosomal dominant inheritance
UUID: 3eed550b-ae19-4435-acea-e76457ec9345
Approved on: 2022/3/16
Published on: 2025/9/15

HGVS expressions

NM_000051.4(ATM):c.3137T>C (p.Leu1046Pro)
NM_000051.4:c.3137T>C
NC_000011.10:g.108272591T>C
CM000673.2:g.108272591T>C
NC_000011.9:g.108143318T>C
CM000673.1:g.108143318T>C
NC_000011.8:g.107648528T>C
NG_009830.1:g.54760T>C
ENST00000452508.7:c.3137T>C
ENST00000713593.1:c.*2608T>C
ENST00000278616.9:c.3137T>C
ENST00000683174.1:n.3287T>C
ENST00000527805.6:c.3137T>C
ENST00000675595.1:c.2972T>C
ENST00000675843.1:c.3137T>C
ENST00000278616.8:c.3137T>C
ENST00000452508.6:c.3137T>C
ENST00000527805.5:c.3137T>C
NM_000051.3:c.3137T>C
NM_001351834.1:c.3137T>C
NM_001351834.2:c.3137T>C

Likely Pathogenic

Met criteria codes 3

PM2_Supporting PP3 PM3_Strong

Evidence Links 0

Expert Panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

Criteria Specification Information

- **Criteria Specification:** *ClinGen Hereditary Breast, Ovarian and Pancreatic Cancer Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ATM Version 1.1*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

The ATM c.3137T>C (p.Leu1046Pro) variant is absent in GnomAD v2.1.1 (PM2_Supporting). In silico protein predictors (REVEL Impact: 0.854; Align-GVGD: C65) predict that this alteration is deleterious (PP3). This variant has also been observed in a homozygous and compound heterozygous state (presumed) in multiple individuals with ataxia-telangiectasia (PMIDs: 27664052, 31429931) (PM3_Strong). In summary, this variant meets criteria to be classified as likely pathogenic based on the ACMG/AMP criteria applied as specified by the HBOP Variant Curation Expert Panel.

Met criteria codes

PM2_Supporting	✔	This variant is absent in GnomAD v2.1.1 (PM2_Supporting).
PP3	✔	In silico protein predictors (REVEL Impact: 0.854; Align-GVGD: C65) predict that this alteration is deleterious (PP3).
PM3_Strong	✔	This variant has been observed in a homozygous and compound heterozygous state (presumed) in multiple individuals with Ataxia-Telangiectasia (27664052, 31429931: PM3_Strong) (4)

Curation History

Showing 1 to 5 of 5 rows

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