

Variant: *NM_000051.3(ATM):c.1442T>G (p.Leu481Ter)*

Version: 2.0

CA382534080 [↗](#)

453367 (ClinVar) [↗](#)

Gene: ATM ([HGNC:472](#))

Condition: ATM-related cancer predisposition ([MONDO:0700270](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: f9cb171c-3f39-4add-9c85-9254d5f10266

Approved on: 2022/3/9

Published on: 2025/9/15

HGVS expressions

NM_000051.3(ATM):c.1442T>G (p.Leu481Ter)

NM_000051.3:c.1442T>G

NC_000011.10:g.108250907T>G

CM000673.2:g.108250907T>G

NC_000011.9:g.108121634T>G

CM000673.1:g.108121634T>G

NC_000011.8:g.107626844T>G

NG_009830.1:g.33076T>G

ENST00000452508.7:c.1442T>G

ENST00000713593.1:c.*913T>G

ENST00000278616.9:c.1442T>G

ENST00000682516.1:n.1576T>G

ENST00000682956.1:n.1576T>G

ENST00000683174.1:n.1592T>G

ENST00000683605.1:n.937T>G

ENST00000684037.1:c.*377T>G

ENST00000684061.1:n.1576T>G

ENST00000684179.1:n.1411T>G

ENST00000527805.6:c.1442T>G

ENST00000675595.1:c.1277T>G

ENST00000675843.1:c.1442T>G

ENST00000278616.8:c.1442T>G

ENST00000452508.6:c.1442T>G

ENST00000527805.5:c.1442T>G

NM_001351834.1:c.1442T>G

NM_001351834.2:c.1442T>G

NM_000051.4:c.1442T>G

Pathogenic

Met criteria codes **4**

PM2_Supporting

PVS1

PM5_Supporting

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

Evidence submitted by expert panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

The ATM c.1442T>G (p.Leu481Ter) variant is expected to produce an NMD-prone transcript due to a nonsense or frameshifting event (PVS1). In the absence of potential splicing rescue mechanisms in ATM, all PVS1-eligible truncating variants are expected to be pathogenic based on the existence of known pathogenic C-terminal truncations in the last exon (PM5_Supporting). This variant has been observed in a compound heterozygous state (confirmed) in one individual with Ataxia-Telangiectasia (PMID 21665257: PM3_Strong). This variant is absent in GnomAD v2.1.1 (PM2_Supporting). In summary, this variant meets criteria to be classified as 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).
PVS1		✓	Expected to produce an NMD-prone transcript due to a nonsense or frameshifting event (PVS1).
PM5_Supporting		✓	In the absence of potential splicing rescue mechanisms in ATM, all PVS1-eligible truncating variants are expected to be pathogenic based on the existence of known pathogenic C-terminal truncations in the last exon (PM5_Supporting)
PM3_Strong		✓	This variant has been observed in a compound heterozygous state (confirmed) in one individual with Ataxia-Telangiectasia (PMID 21665257: PM3_Strong) (4 POINTS)

Curation History [↗](#)

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