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.8300T>C (p.Leu2767Pro)

Version: 1.0

CA382516339

1023669 (ClinVar)

Gene: ATM (HGNC:472)
Condition: ATM-related cancer predisposition (MONDO:0700270)
Inheritance Mode: Autosomal dominant inheritance
UUID: bdbeeb61-e719-46cf-a6a4-1675057be466
Approved on: 2025/6/11
Published on: 2025/7/10

HGVS expressions

NM_000051.4(ATM):c.8300T>C (p.Leu2767Pro)
NM_000051.4:c.8300T>C
NC_000011.10:g.108343253T>C
CM000673.2:g.108343253T>C
NC_000011.9:g.108213980T>C
CM000673.1:g.108213980T>C
NC_000011.8:g.107719190T>C
NG_009830.1:g.125422T>C
NG_054724.1:g.131580A>G
ENST00000452508.7:c.8300T>C
ENST00000713593.1:c.*7771T>C
ENST00000278616.9:c.8300T>C
ENST00000638786.2:n.998T>C
ENST00000682286.1:n.3057T>C
ENST00000682302.1:n.2718T>C
ENST00000683174.1:n.9784T>C
ENST00000683524.1:n.3524T>C
ENST00000684152.1:n.3716T>C
ENST00000684180.1:n.774T>C
ENST00000684447.1:n.4793T>C
ENST00000527805.6:c.*3364T>C
ENST00000675595.1:c.*3435T>C
ENST00000675843.1:c.8300T>C
ENST00000278616.8:c.8300T>C
ENST00000452508.6:c.8300T>C
ENST00000524755.5:c.227-7961A>G
ENST00000524792.5:n.4515T>C
ENST00000525729.5:c.641-34182A>G
ENST00000526725.1:n.272-2889A>G
ENST00000527531.5:c.*1197-7961A>G
ENST00000615746.4:c.*1197-7961A>G
NM_000051.3:c.8300T>C
NM_001330368.1:c.641-34182A>G
NM_001351110.1:c.695-7961A>G
NM_001351834.1:c.8300T>C
NR_147053.2:n.2302-7961A>G
NM_001330368.2:c.641-34182A>G

NM_001351110.2:c.695-7961A>G
NM_001351834.2:c.8300T>C
NR_147053.3:n.2300-7961A>G

Likely Pathogenic

Met criteria codes 3

PM3_Strong | PM2_Supporting | PP3

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 1.3.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

The c.8300T>C variant in ATM is a missense variant predicted to cause substitution of leucine by proline at amino acid 2767 (p.Leu2767Pro). This variant has been detected at least in two unrelated individuals with Ataxia-Telangiectasia (PMIDs: 22649200, 26896183). This variant is absent from gnomAD v4.1.0. The computational predictor REVEL gives a score of 0.94, which is above the threshold of 0.733, evidence that correlates with impact to ATM function. In summary, this variant meets the criteria to be classified as likely pathogenic for autosomal dominant ATM-related cancer predisposition and autosomal recessive Ataxia-Telangiectasia based on the ACMG/AMP criteria applied as specified by the HBOP VCEP. (PM3_Strong, PM2_Supporting, PP3)

Met criteria codes

PM3_Strong	✔	This Variant has been detected in at least 2 individuals with Ataxia-Telangiectasia. CASE 1-Heterozygous with the pathogenic variant c.217_218delGA -2 points (PMID:26896183)(Jackson et al.,) CASE 2- PMID: 22649200 –Phenotype confident –c.217_218delGA Phase Unknown –2 points Total-4 points
PM2_Supporting	✔	Absent from controls
PP3	✔	The computational predictor REVEL gives a score of 0.94, which is above the threshold of ≥.733, evidence that correlates with impact to ATM function

Curation History

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