

Variant: NM_000051.4(ATM):c.8486C>T (p.Pro2829Leu)

Version: 1.0

CA16619259 [↗](#)

418777 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 13918561-f552-46bc-a22c-f090b5a93152

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HGVS expressions

NM_000051.4:c.8486C>T
NM_000051.4(ATM):c.8486C>T (p.Pro2829Leu)
NC_000011.10:g.108345810C>T
CM000673.2:g.108345810C>T
NC_000011.9:g.108216537C>T
CM000673.1:g.108216537C>T
NC_000011.8:g.107721747C>T
NG_009830.1:g.127979C>T
NG_054724.1:g.129023G>A
ENST00000452508.7:c.8486C>T
ENST00000713593.1:c.*7957C>T
ENST00000278616.9:c.8486C>T
ENST00000638786.2:n.1184C>T
ENST00000682286.1:n.3243C>T
ENST00000682302.1:n.2904C>T
ENST00000683174.1:n.9970C>T
ENST00000683524.1:n.3710C>T
ENST00000684152.1:n.3902C>T
ENST00000684180.1:n.960C>T
ENST00000684447.1:n.4979C>T
ENST00000527805.6:c.*3550C>T
ENST00000675595.1:c.*3621C>T
ENST00000675843.1:c.8486C>T
ENST00000278616.8:c.8486C>T
ENST00000452508.6:c.8486C>T
ENST00000524755.5:c.227-10518G>A
ENST00000524792.5:n.4701C>T
ENST00000525729.5:c.641-36739G>A
ENST00000526725.1:n.272-5446G>A
ENST00000527531.5:c.*1196+9105G>A
ENST00000615746.4:c.*1196+9105G>A
NM_000051.3:c.8486C>T
NM_001330368.1:c.641-36739G>A
NM_001351110.1:c.695-10518G>A
NM_001351834.1:c.8486C>T
NR_147053.2:n.2301+9105G>A
NM_001330368.2:c.641-36739G>A

NM_001351110.2:c.695-10518G>A
NM_001351834.2:c.8486C>T
NR_147053.3:n.2299+9105G>A

Likely Pathogenic

Met criteria codes 3

PM3_Strong PP1 PP3

Not Met criteria codes 1

PM2

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 1.5*

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

The c.8486C>T variant in ATM is a missense variant predicted to cause substitution of proline by leucine at amino acid 2829 (p.Pro2829Leu). This variant has been detected in individual(s) with Ataxia-Telangiectasia (personal communication; PMID: 9711876; PMID: 37438524). This variant has been reported to segregate with Ataxia-Telangiectasia in two affected individuals from one family (personal communication). The highest population minor allele frequency in gnomAD v4.1.0 is 0.00001667 in the Admixed American population (PM2_Supporting, BS1, and BA1 are not met). The computational predictor REVEL gives a score of 0.952, 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, PP1, PP3)

Met criteria codes

PM3_Strong



This variant has been detected in 3 individuals with Ataxia-Telangiectasia. Of those individuals, two were confirmed in trans by family testing (personal communication). One individual was homozygous for the variant (PMID: 9711876; PMID: 37438524)(PM3_Strong).

PP1



This variant has been reported to segregate with Ataxia-Telangiectasia in two affected individuals from one family (personal communication)

PP3



The computational predictor REVEL gives a score of 0.952, which is above the threshold of 0.733, evidence that correlates with impact to ATM function (PP3). SpliceAI does not predict any aberrant splicing associated with this variant.

Not Met criteria codes

PM2



The highest population minor allele frequency in gnomAD v4 is 0.000016 (1/60002 alleles) in Admixed American population which is higher than the ClinGen HBOP threshold 0.001% for PM2_Supporting

Curation History 

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Showing 1 to 1 of 1 rows

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