

Variant: *NM\_000051.4(ATM):c.1066-6T>G*

Version: 2.0

CA151456 [↗](#)

3038 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UUID:** bccd9f16-4086-4c14-8d78-04c52ad58630

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### *HGVS expressions*

**NM\_000051.4:c.1066-6T>G**

NM\_000051.4(ATM):c.1066-6T>G

NC\_000011.10:g.108248927T>G

CM000673.2:g.108248927T>G

NC\_000011.9:g.108119654T>G

CM000673.1:g.108119654T>G

NC\_000011.8:g.107624864T>G

NG\_009830.1:g.31096T>G

ENST00000452508.7:c.1066-6T>G

ENST00000713593.1:c.\*537-6T>G

ENST00000278616.9:c.1066-6T>G

ENST00000682516.1:n.1200-6T>G

ENST00000682956.1:n.1200-6T>G

ENST00000683174.1:n.1216-6T>G

ENST00000683605.1:n.561-6T>G

ENST00000684037.1:c.\*1-6T>G

ENST00000684061.1:n.1200-6T>G

ENST00000684179.1:n.1035-6T>G

ENST00000527805.6:c.1066-6T>G

ENST00000675595.1:c.901-6T>G

ENST00000675843.1:c.1066-6T>G

ENST00000278616.8:c.1066-6T>G

ENST00000452508.6:c.1066-6T>G

ENST00000527805.5:c.1066-6T>G

NM\_000051.3:c.1066-6T>G

NM\_001351834.1:c.1066-6T>G

NM\_001351834.2:c.1066-6T>G

**Benign**

**Met criteria codes** **2**

**BP2\_Strong** **BS1**

**Not Met criteria codes** **3**

**PM2** **BA1** **BP4**

**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.1066-6T>G variant has a GnomAD (v2.1.1) filtering allele frequency of 0.2081% (NFE) which is above the ATM BS1 threshold of .05% (BS1). In silico predictors (SpliceAI Acceptor Loss 0.62; MaxEntScan -2.49) are indeterminate about whether this variant affects splicing. This variant has been observed in a homozygous and compound heterozygous state (presumed) in multiple individuals without Ataxia-Telangiectasia (BP2\_Strong, GTR Lab IDs: 61756, 26957, 500031). In summary, this variant meets criteria to be classified as benign based on the ACMG/AMP criteria applied as specified by the HBOP Variant Curation Expert Panel

#### Met criteria codes

<b>BP2_Strong</b>			This variant has been observed in a homozygous and compound heterozygous state (presumed) in multiple individuals without Ataxia-Telangiectasia (BP2_Strong, GTR Lab IDs: 61756, 26957, 500031).
<b>BS1</b>			This variant has a GnomAD (v2.1.1) filtering allele frequency of 0.2% (NFE) which is above the ATM BS1 threshold of 0.05% (BS1).

#### Not Met criteria codes

<b>PM2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BA1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP4</b>			In silico predictors (SpliceAI Acceptor Loss 0.62; MaxEntScan -2.49) are indeterminate about whether this variant affects splicing.

### Curation History [↗](#)

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