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.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
Approved on: 2022-03-09
Published on: 2025-09-15

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

BS1 BP2_Strong

Not Met criteria codes 3

BP4 PM2 BA1

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.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

BS1	✔	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).
BP2_Strong	✔	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).

Not Met criteria codes

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

Curation History

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