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_000051.3(ATM):c.2639-17G>T

Version: 2.0

CA163513

140763 (ClinVar)

Gene: ATM (HGNC:472)
Condition: ATM-related cancer predisposition (MONDO:0700270)
Inheritance Mode: Autosomal dominant inheritance
UUID: 12e51ac3-f131-4b31-8659-d7e9bb1ed185
Approved on: 2022-03-09
Published on: 2025-09-15

HGVS expressions

NM_000051.3:c.2639-17G>T
NM_000051.3(ATM):c.2639-17G>T
NC_000011.10:g.108268393G>T
CM000673.2:g.108268393G>T
NC_000011.9:g.108139120G>T
CM000673.1:g.108139120G>T
NC_000011.8:g.107644330G>T
NG_009830.1:g.50562G>T
ENST00000452508.7:c.2639-17G>T
ENST00000713593.1:c.*2110-17G>T
ENST00000278616.9:c.2639-17G>T
ENST00000682516.1:n.2772+1051G>T
ENST00000683174.1:n.2789-17G>T
ENST00000684037.1:c.*1573+1051G>T
ENST00000527805.6:c.2639-17G>T
ENST00000675595.1:c.2474-17G>T
ENST00000675843.1:c.2639-17G>T
ENST00000278616.8:c.2639-17G>T
ENST00000452508.6:c.2639-17G>T
ENST00000527805.5:c.2639-17G>T
NM_001351834.1:c.2639-17G>T
NM_001351834.2:c.2639-17G>T
NM_000051.4:c.2639-17G>T

Benign

Met criteria codes 3

BA1 BP4 BP2_Strong

Not Met criteria codes 4

BS1 BP7 PP3 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 1.1*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

The ATM c.2639-17G>T variant has a gnomAD v2.1.1 filtering allele frequency of 6.505% (African/African-American; exomes) which exceeds the ATM BA1 threshold of 0.50% (BA1). This variant has been observed in a homozygous state in multiple individuals without biallelic disease (BP2_Strong; GTR Lab ID: 61756). In silico splicing predictors (SpliceAI: AL 0.00/DL 0.00/AG 0.01/DG 0.00; MaxEntScan: +1.92% (wild type = 9.90, variant = 10.09)) find that this variant is unlikely to affect splicing (BP4). 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

BA1	✔	GnomAD v2.1.1 FAF 6.505% (African/African-American; exomes) exceeds ATM BA1 threshold of 0.50%.
BP4	✔	In silico splicing predictors (SpliceAI: AL 0.00/DL 0.00/AG 0.01/DG 0.00; MaxEntScan: +1.92% (wild type = 9.90, variant = 10.09)) find that this variant is unlikely to affect splicing (BP4).
BP2_Strong	✔	This variant has been observed in a homozygous state in multiple individuals without biallelic disease (BP2_Strong; GTR Lab ID: 61756).

Not Met criteria codes

BS1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7	✖	This variant is not considered deep intronic (beyond +20 or beyond -40).
PP3	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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