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.217_218del (p.Glu73fs)

Version: 2.0

CA6264537

569567 (ClinVar)

Gene: ATM (HGNC:472)
Condition: ATM-related cancer predisposition (MONDO:0700270)
Inheritance Mode: Autosomal dominant inheritance
UUID: 673ac80a-e147-4045-9c13-2d6372ba5558
Approved on: 2023/9/5
Published on: 2025/9/15

HGVS expressions

NM_000051.4(ATM):c.217_218del (p.Glu73fs)
NM_000051.4:c.217_218del
NC_000011.10:g.108229209_108229210del
CM000673.2:g.108229209_108229210del
NC_000011.9:g.108099936_108099937del
CM000673.1:g.108099936_108099937del
NC_000011.8:g.107605146_107605147del
NG_009830.1:g.11378_11379del
ENST00000452508.7:c.217_218del
ENST00000683914.2:c.217_218del
ENST00000713593.1:c.217_218del
ENST00000278616.9:c.217_218del
ENST00000682147.1:n.347_348del
ENST00000682430.1:n.316_317del
ENST00000682465.1:c.217_218del
ENST00000682516.1:n.351_352del
ENST00000682956.1:n.351_352del
ENST00000683150.1:c.217_218del
ENST00000683174.1:n.367_368del
ENST00000683468.1:c.217_218del
ENST00000683488.1:n.4877_4878del
ENST00000683914.1:c.217_218del
ENST00000684029.1:c.217_218del
ENST00000684037.1:c.217_218del
ENST00000684061.1:n.351_352del
ENST00000684179.1:n.351_352del
ENST00000527805.6:c.217_218del
ENST00000638443.1:c.217_218del
ENST00000639240.1:c.217_218del
ENST00000639953.1:c.217_218del
ENST00000640388.1:c.217_218del
ENST00000675595.1:c.217_218del
ENST00000675843.1:c.217_218del
ENST00000278616.8:c.217_218del
ENST00000452508.6:c.217_218del
ENST00000526567.5:c.217_218del
ENST00000527805.5:c.217_218del

ENST00000527891.5:c.217_218del
ENST00000530958.5:c.217_218del
ENST00000532931.5:c.217_218del
ENST00000601453.2:c.217_218del
NM_000051.3:c.217_218del
NM_001351834.1:c.217_218del
NM_001351835.1:c.217_218del
NM_001351836.1:c.217_218del
NM_001351834.2:c.217_218del
NM_001351835.2:c.217_218del
NM_001351836.2:c.217_218del

Pathogenic

Met criteria codes 4

PM3_Very Strong | PM2_Supporting | PVS1 | PM5_Supporting

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 c.217_218del (p.Glu73Metfs*26) variant in ATM is a frameshift variant in a biologically-relevant-exon predicted to cause a premature stop codon leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism. This alteration results in a termination codon upstream of the most C-terminus pathogenic alteration (ATM p.Arg3047*), as classified by the HBOP VCEP, and is expected to be more deleterious. This variant has been detected in at least 5 individuals with Ataxia-Telangiectasia (PMIDs 26677768; 26896183; 22649200; 10817650). This variant is absent from gnomAD v2.1. In summary, this variant meets criteria to be classified as pathogenic for autosomal dominant hereditary breast cancer and autosomal recessive Ataxia-Telangiectasia based on the ACMG/AMP criteria applied, as specified by the HBOP VCEP. (PVS1, PM5_Supporting, PM3_VeryStrong, PM2_Supporting)

Met criteria codes

PM3_Very Strong	✔	This variant has been observed in at least four homozygous and compound heterozygous (confirmed and presumed) in multiple individuals with Ataxia-Telangiectasia (PMIDS 26677768; 26896183; 22649200; 10817650: 8 POINTS-PM3_VeryStrong).
PM2_Supporting	✔	This variant is absent from gnomAD v2.1 (PM2_Supporting).
PVS1	✔	The c.217_218del p.Glu73Metfs*26 variant in ATM is a frameshift variant in biologically-relevant-exon predicted to cause a premature stop codon leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism.

PM5_Supporting	✔	This alteration results in a termination codon upstream of the most C-terminus pathogenic alteration (ATM p.Arg3047*), as classified by the HBOP VCEP, and is expected to be more deleterious.

Curation History

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