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.2929T>C (p.Cys977Arg)

Version: 1.0

CA382547092

1027443 (ClinVar)

Gene: ATM (HGNC:472)
Condition: ATM-related cancer predisposition (MONDO:0700270)
Inheritance Mode: Autosomal dominant inheritance
UUID: e96a90b3-7e2e-4b54-8b56-c28de27cff37
Approved on: 2026/6/19
Published on: 2026/8/5

HGVS expressions

NM_000051.4:c.2929T>C
NM_000051.4(ATM):c.2929T>C (p.Cys977Arg)
NC_000011.10:g.108271258T>C
CM000673.2:g.108271258T>C
NC_000011.9:g.108141985T>C
CM000673.1:g.108141985T>C
NC_000011.8:g.107647195T>C
NG_009830.1:g.53427T>C
ENST00000452508.7:c.2929T>C
ENST00000713593.1:c.*2400T>C
ENST00000278616.9:c.2929T>C
ENST00000682516.1:n.2863T>C
ENST00000683174.1:n.3079T>C
ENST00000684037.1:c.*1664T>C
ENST00000527805.6:c.2929T>C
ENST00000675595.1:c.2764T>C
ENST00000675843.1:c.2929T>C
ENST00000278616.8:c.2929T>C
ENST00000419286.2:n.208T>C
ENST00000452508.6:c.2929T>C
ENST00000527805.5:c.2929T>C
NM_000051.3:c.2929T>C
NM_001351834.1:c.2929T>C
NM_001351834.2:c.2929T>C

Likely Pathogenic

Met criteria codes 3

PM2_Supporting PP3 PM3_Strong

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 1.5.0*

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

The c.2929T>C variant in ATM is missense variant predicted to cause substitution of cystine by arginine at amino acid 977 (p.Cys977Arg). This variant has been detected in individual(s) with Ataxia-Telangiectasia (PMID: 34445196). This variant is absent from gnomAD v4.1.0. The computational predictor REVEL gives a score of 0.82, 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, PM2_Supporting, PP3)

Met criteria codes

PM2_Supporting	✔	This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PP3	✔	The computational predictor REVEL gives a score of 0.82, which is above the threshold of 0.733, evidence that correlates with impact to ATM function (PP3).
PM3_Strong	✔	This variant has been detected in at least 1 individual with Ataxia-Telangiectasia (PMID: 34445196, PM3_strong).

Curation History

Showing 1 to 1 of 1 rows

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