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.1810C>T (p.Pro604Ser)

Version: 1.0

CA157068

127343 (ClinVar)

Gene: ATM (HGNC:472)
Condition: ATM-related cancer predisposition (MONDO:0700270)
Inheritance Mode: Autosomal dominant inheritance
UUID: c43104b1-238b-496b-b4d2-7c03e1318375
Approved on: 2025-11-11
Published on: 2026-04-10

HGVS expressions

NM_000051.4:c.1810C>T
NM_000051.4(ATM):c.1810C>T (p.Pro604Ser)
NC_000011.10:g.108252824C>T
CM000673.2:g.108252824C>T
NC_000011.9:g.108123551C>T
CM000673.1:g.108123551C>T
NC_000011.8:g.107628761C>T
NG_009830.1:g.34993C>T
ENST00000452508.7:c.1810C>T
ENST00000713593.1:c.*1281C>T
ENST00000278616.9:c.1810C>T
ENST00000682516.1:n.1944C>T
ENST00000683174.1:n.1960C>T
ENST00000683605.1:n.1305C>T
ENST00000684037.1:c.*745C>T
ENST00000684061.1:n.1944C>T
ENST00000527805.6:c.1810C>T
ENST00000675595.1:c.1645C>T
ENST00000675843.1:c.1810C>T
ENST00000278616.8:c.1810C>T
ENST00000452508.6:c.1810C>T
ENST00000525012.5:n.4C>T
ENST00000527805.5:c.1810C>T
ENST00000533526.1:n.4C>T
NM_000051.3:c.1810C>T
NM_001351834.1:c.1810C>T
NM_001351834.2:c.1810C>T

Benign

Met criteria codes 1

BA1

Not Met criteria codes 2

BP4 PP3

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 1.4.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

The c.1810C>T variant in ATM is a missense variant predicted to cause substitution of proline by serine at amino acid 604 (p.Pro604Ser). The filtering allele frequency (the lower threshold of the 95% CI of 71/6026) of the c.1810C>T variant in ATM is 0.009580 for the Middle Eastern chromosomes by gnomAD v4.1.0, which is higher than the HBOP VCEP threshold (>0.005) for BA1, and therefore meets this criterion. The computational predictor REVEL gives a score of 0.379, which is neither above nor below the thresholds predicting a damaging or benign impact on ATM function. In summary, this variant meets the criteria to be classified as benign 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. (BA1)

Met criteria codes

BA1	✔	The GnomAD Filtering Allele Frequency is 0.009580, which is higher than the HBOP VCEP threshold (>0.5%) for BA1, meeting this criterion (BA1).

Not Met criteria codes

BP4	✖	The computational predictor REVEL gives a score of 0.379, which is neither above nor below the thresholds predicting a damaging or benign impact on ATM function (PP3, BP4 not met).
PP3	✖	The computational predictor REVEL gives a score of 0.379, which is neither above nor below the thresholds predicting a damaging or benign impact on ATM function (PP3, BP4 not met).

Curation History

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