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Variant: NM_000051.4(ATM):c.2152dup (p.Cys718fs)

Version: 1.0

CA2499220586

1027442 (ClinVar)

Gene: ATM (HGNC:472)
Condition: ATM-related cancer predisposition (MONDO:0700270)
Inheritance Mode: Autosomal dominant inheritance
UUID: 60ff5886-a7dc-40d7-897a-e5d9a96667e1
Approved on: 2026/6/19
Published on: 2026/8/5

HGVS expressions

NM_000051.4(ATM):c.2152dup (p.Cys718fs)
NM_000051.4:c.2152dup
NC_000011.10:g.108256242dup
CM000673.2:g.108256242dup
NC_000011.9:g.108126969dup
CM000673.1:g.108126969dup
NC_000011.8:g.107632179dup
NG_009830.1:g.38411dup
ENST00000452508.7:c.2152dup
ENST00000713593.1:c.*1623dup
ENST00000278616.9:c.2152dup
ENST00000682516.1:n.2286dup
ENST00000683174.1:n.2302dup
ENST00000683605.1:n.1647dup
ENST00000684037.1:c.*1087dup
ENST00000684061.1:n.2286dup
ENST00000527805.6:c.2152dup
ENST00000675595.1:c.1987dup
ENST00000675843.1:c.2152dup
ENST00000278616.8:c.2152dup
ENST00000452508.6:c.2152dup
ENST00000527805.5:c.2152dup
NM_000051.3:c.2152dup
NM_001351834.1:c.2152dup
NM_001351834.2:c.2152dup

Pathogenic

Met criteria codes 3

PM2_Supporting PVS1 PM5_Supporting

Not Met criteria codes 1

PM3

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.2152dup (p.Cys718Leufs*20) variant in ATM is a frameshift variant predicted to cause a premature stop codon in biologically-relevant-exon 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 one individual with Ataxia-Telangiectasia (PMID: 34445196). This variant is absent from gnomAD v4.1.0. In summary, this variant meets the criteria to be classified as 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. (PVS1, PM5_Supporting, PM2_Supporting)

Met criteria codes

PM2_Supporting	✔	This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PVS1	✔	The c.2152dup p.Cys718Leufs*20)(NM_000051.4) variant in ATM is a frameshift variant predicted to cause a premature stop codon leading to nonsense mediated decay (PVS1).
PM5_Supporting	✔	This alteration results in a termination codon upstream of the most C-terminal pathogenic alteration (ATM p.Arg3047*), as classified by the HBOP VCEP, and is expected to be more deleterious (PM5_Supporting).

Not Met criteria codes

PM3	✖	This variant has been detected in at least 1 individual with Ataxia-Telangiectasia (PMID: 34445196, PM3 not included for this variant to avoid double dipping)

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000051.4(ATM):c.2152dup (p.Cys7…	Pathogenic	ATM-Related Cancer Predisposition	2026-08-05	1.0	ClinGen Hereditary Breast, Ovarian and Pancreatic Cancer Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ATM Version 1.5.0	ATM

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