

Variant: *NM_000051.4(ATM):c.6047A>G (p.Asp2016Gly)*

Version: 1.0

CA163663 [↗](#)

140823 (ClinVar) [↗](#)

Gene: ATM ([HGNC:472](#))

Condition: ATM-related cancer predisposition ([MONDO:0700270](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: e04f89d0-ae09-4376-9083-3c1d3af96ff1

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HGVS expressions

NM_000051.4(ATM):c.6047A>G (p.Asp2016Gly)

NM_000051.4:c.6047A>G

NC_000011.10:g.108315863A>G

CM000673.2:g.108315863A>G

NC_000011.9:g.108186590A>G

CM000673.1:g.108186590A>G

NC_000011.8:g.107691800A>G

NG_009830.1:g.98032A>G

NG_054724.1:g.158970T>C

ENST00000452508.7:c.6047A>G

ENST00000713593.1:c.*5518A>G

ENST00000278616.9:c.6047A>G

ENST00000525056.2:n.466A>G

ENST00000682286.1:n.804A>G

ENST00000682302.1:n.465A>G

ENST00000683174.1:n.7531A>G

ENST00000683524.1:n.1271A>G

ENST00000684152.1:n.1761A>G

ENST00000527805.6:c.*1111A>G

ENST00000675595.1:c.*1111A>G

ENST00000675843.1:c.6047A>G

ENST00000278616.8:c.6047A>G

ENST00000452508.6:c.6047A>G

ENST00000524792.5:n.2262A>G

ENST00000525729.5:c.641-6792T>C

ENST00000529588.5:c.471A>G

ENST00000532765.1:n.364A>G

ENST00000533690.5:n.1451A>G

NM_000051.3:c.6047A>G

NM_001330368.1:c.641-6792T>C

NM_001351110.1:c.*39-6792T>C

NM_001351834.1:c.6047A>G

NM_001330368.2:c.641-6792T>C

NM_001351110.2:c.*39-6792T>C

NM_001351834.2:c.6047A>G

Likely Pathogenic

Met criteria codes 3

PM3_StrongPP3PM2_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 1.4.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Hereditary Breast, Ovarian and Pancreatic Cancer VCEP

The c.6047A>G variant in ATM is a missense variant predicted to cause substitution of aspartic acid by glycine at amino acid 2016 (p.Asp2016Gly). This variant has been detected in at least three unrelated individuals with Ataxia-Telangiectasia (PMID: 21965147, 26896183, 31741144). This variant is absent from gnomAD v.4.1.0. The computational predictor REVEL gives a score of 0.797, which is above the threshold of 0.7333, evidence that correlates with impact to ATM function. In summary, this variant meets the criteria to be classified as a 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

PM3_Strong	i ✓	This variant has been detected in 3 unrelated individuals with Ataxia-Telangiectasia (PMID: 21965147, 31741144, 26896183, PM3_strong).
PP3	i ✓	The computational predictor REVEL gives a score of 0.797, which is above the threshold of 0.733, evidence that correlates with impact to ATM function (PP3).
PM2_Supporting	i ✓	This variant is absent from gnomAD v.2.1.1 (PM2_Supporting).

Curation History

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