



## ATM gene

ATM serine/threonine kinase

### Normal Function

The *ATM* gene provides instructions for making a protein that is located primarily in the nucleus of cells, where it helps control the rate at which cells grow and divide. This protein also plays an important role in the normal development and activity of several body systems, including the nervous system and the immune system. Additionally, the ATM protein assists cells in recognizing damaged or broken DNA strands. DNA can be damaged by agents such as toxic chemicals or radiation. Breaks in DNA strands also occur naturally when chromosomes exchange genetic material during cell division. The ATM protein coordinates DNA repair by activating enzymes that fix the broken strands. Efficient repair of damaged DNA strands helps maintain the stability of the cell's genetic information.

Because of its central role in cell division and DNA repair, the ATM protein is of great interest in cancer research.

### Health Conditions Related to Genetic Changes

#### ataxia-telangiectasia

Researchers have identified several hundred mutations in the *ATM* gene that cause ataxia-telangiectasia. People with this disorder have mutations in both copies of the *ATM* gene in each cell. Most of these mutations disrupt protein production, resulting in an abnormally small, nonfunctional version of the ATM protein. Cells without any functional ATM protein are hypersensitive to radiation and do not respond normally to DNA damage. Instead of activating DNA repair, the defective ATM protein allows mutations to accumulate in other genes, which may cause cells to grow and divide in an uncontrolled way. This kind of unregulated cell growth can lead to the formation of cancerous tumors. In addition, *ATM* mutations can allow cells to die inappropriately, particularly affecting cells in a part of the brain involved in coordinating movements (the cerebellum). This loss of brain cells causes the movement problems characteristic of ataxia-telangiectasia.

#### breast cancer

#### other cancers

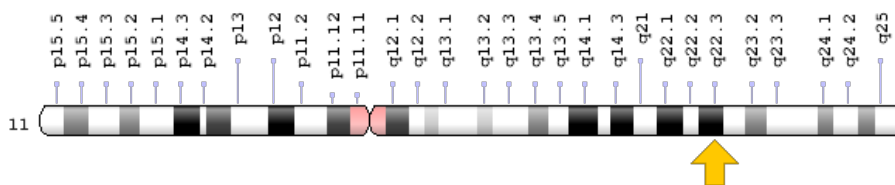
Research suggests that people who carry one mutated copy of the *ATM* gene in each cell may have an increased risk of developing several other types of cancer. In

particular, some studies have shown that cancers of the breast, stomach, bladder, pancreas, lung, and ovaries occur more frequently in *ATM* mutation carriers than in people who do not carry these mutations. The results of similar studies, however, have been conflicting. Additional research is needed to clarify which other types of cancer, if any, are associated with *ATM* mutations.

## Chromosomal Location

Cytogenetic Location: 11q22.3, which is the long (q) arm of chromosome 11 at position 22.3

Molecular Location: base pairs 108,222,484 to 108,369,102 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- AT mutated
- AT protein
- AT1
- ATA
- ataxia telangiectasia mutated
- ataxia telangiectasia mutated (includes complementation groups A, C and D)
- ataxia telangiectasia mutated protein
- ATC
- ATD
- ATDC
- ATE
- ATM\_HUMAN
- human phosphatidylinositol 3-kinase homolog
- serine-protein kinase ATM

- TEL1
- TELO1

## **Additional Information & Resources**

### Educational Resources

- Cancer Medicine (sixth edition, 2003): Ataxia-Telangiectasia  
<https://www.ncbi.nlm.nih.gov/books/NBK13680/#A4590>
- Cancer Medicine (sixth edition, 2003): Cell Cycle Checkpoints, Integration of Signalling, and DNA Repair  
<https://www.ncbi.nlm.nih.gov/books/NBK12902/#A9244>
- Eureka Bioscience Collection: Replication-Independent Double-Strand Breaks (DSBs)  
<https://www.ncbi.nlm.nih.gov/books/NBK6555/#A17952>
- National Cancer Institute: Genetics of Breast and Ovarian Cancer (PDQ)  
<https://www.cancer.gov/types/breast/hp/breast-ovarian-genetics-pdq>

### GeneReviews

- Ataxia-Telangiectasia  
<https://www.ncbi.nlm.nih.gov/books/NBK26468>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ATM%5BTI%5D%29+OR+%28ataxia+telangiectasia+mutated%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

### OMIM

- ATAXIA-TELANGIECTASIA MUTATED GENE  
<http://omim.org/entry/607585>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
<http://atlasgeneticsoncology.org/Genes/ATMID123.html>
- Cancer Genetics Web: ATM  
<http://www.cancerindex.org/geneweb/ATM.htm>
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ATM%5Bgene%5D>

- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=795](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=795)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/472>
- UniProt  
<http://www.uniprot.org/uniprot/Q13315>

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