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 variants (also called mutations) in the *ATM* gene that cause ataxia-telangiectasia. This disorder is characterized by progressive difficulty with coordinating movements (ataxia) beginning in early childhood. People with this disorder have variants in both copies of the *ATM* gene in each cell. Most of these variants 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 altered ATM protein allows variants 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* gene variants 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.

Bladder cancer

MedlinePlus Genetics provides information about Bladder cancer

Breast cancer

MedlinePlus Genetics provides information about Breast cancer

Melanoma

MedlinePlus Genetics provides information about Melanoma

Other cancers

Research suggests that people who carry one alterd 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* gene variant carriers than in people who do not carry these variants. 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* gene variants.

Other Names for This Gene

- AT mutated
- AT protein
- AT1
- ATA
- ataxia telangiectasia mutated
- ATM_HUMAN
- human phosphatidylinositol 3-kinase homolog
- serine-protein kinase ATM
- TEL1
- TELO1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ATM (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=472[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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)

Catalog of Genes and Diseases from OMIM

- ATM SERINE/THREONINE KINASE; ATM (https://omim.org/entry/607585)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/472)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ATM[gene])

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Genomic Location

The *ATM* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

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