



Fact Sheet

ATM Mutations

Brought to you by the *National Society of Genetic Counselors, Cancer Special Interest Group*

What you should know about *ATM* Mutations

People with a single *ATM* mutation are at an increased risk to develop certain cancers, such as female breast cancer, pancreatic cancer, and prostate cancer. Individuals with two *ATM* mutations (one from each parent) have Ataxia Telangiectasia (AT). AT is a childhood onset genetic condition that is characterized by ataxia (impaired balance or coordination), telangiectases (enlarged blood vessels) which occur in eyes and on the skin, immune deficiency, and an increased risk for some cancers, such as leukemia and lymphoma.

Cancer Risks and Features Associated with *ATM* Mutations

Cancer Type	Lifetime Risk	General Population risk
Female Breast Cancer	17-52%	12%
Pancreatic Cancer	Increased	1.6%
Prostate Cancer	Elevated*	11%

*There is evidence that men with *ATM* mutations are more likely to develop aggressive or lethal prostate cancer

Genetics and Inheritance of *ATM* Mutations

Genes are our body's instructions. They provide our body with information about how to grow and develop. When there is a mutation in a gene it can cause the gene to no longer function correctly. Each person has two copies of every gene. One copy is inherited from their mother and the other copy is inherited from their father.

Cancer risks associated with mutations in the *ATM* gene are inherited in an autosomal dominant fashion. This means that children, siblings and parents of individuals with an *ATM* mutation have a 50% (1 in 2) chance of having the mutation as well. Both males and females can inherit an *ATM* mutation and can pass it on to their children.

If two individuals with *ATM* mutations have a child together, there is a 25% (1 in 4) chance that the child will inherit both *ATM* mutations. Individuals who inherit two mutations in the *ATM* gene (one from each parent) have a different condition called Ataxia Telangiectasia (AT).

Managing Cancer Risks

Female Breast Cancer

- Starting at 40y
 - Annual mammogram with consideration of tomosynthesis
 - Consider breast MRI with contrast
- Evidence currently insufficient for Risk Reducing Prophylactic Mastectomy
 - Manage based on family history

Managing Cancer Risks (Continued)

Pancreatic Cancer

- If a close blood relative has a history of pancreatic cancer:
 - Annual screening with contrast-enhanced MRI/MRCP and/or endoscopic ultrasound starting at 50y OR 10 years younger than youngest pancreatic cancer case in family
- If no family history of pancreatic cancer:
 - No screening is currently recommended

Prostate Cancer

- Starting at 50y or 5-10 years earlier than the youngest age at diagnosis of prostate cancer in the family
 - Consider Annual prostate specific antigen (PSA) blood test
 - Consider Annual digital rectal exam (DRE)

Genetic Counseling

In many families, the cancer history may be due to a combination of genetic and environmental factors. In addition, other genetic conditions (i.e. other gene mutations) may appear clinically similar to *ATM* mutations. For this reason, a detailed review of the family history by a genetics professional is important before pursuing genetic testing. A genetic counselor can help determine which, if any, genetic tests may be helpful for a family and review the benefits, risks and limitations of genetic testing. Genetic testing is usually performed through a blood or saliva sample.

Genetic test results can be complicated and are most useful when interpreted by a genetics professional in the context of an individual's personal and family history. To locate a genetic counselor near you, please visit www.nsgc.org and click on the 'Find a Genetic Counselor' link.

Genetic Discrimination

The Genetic Information Nondiscrimination Act (GINA) was signed into federal law in 2008. GINA prohibits health insurers and most employers from discriminating against individuals based on genetic information (including the results of genetic tests and family history information). According to GINA, health insurance companies cannot consider genetic information to be a preexisting condition, nor can they use it to make decisions regarding coverage or rates. GINA also makes it illegal for most employers to use genetic information in making decisions about hiring, firing, promotion, or terms of employment. It is important to note that GINA does not offer protections for life insurance, disability insurance, or long-term care insurance. More information about GINA can be found by contacting a local genetic counselor or by visiting www.ginahelp.org.

Resources

- My Support 360: <https://mysupport360.com/associations/genes/atm-gene-mutations/>
- FORCE: <https://www.facingourrisk.org/understanding-brca-and-hboc/information/hereditary-cancer/other-genes/basics/atm.php>

References

- My Support360 <https://mysupport360.com/associations/genes/atm-gene-mutations/>
- Na, R., Zheng, L., Han, M., et al. (2017). Germline mutations in ATM and BRCA1/2 distinguish for lethal and indolent prostate cancer and are associated with early age at death. *European Urology*, 71(5), 740-747. <https://doi.org/10.1016/j.eururo.2016.11.033>
- Genetics Home Reference <https://ghr.nlm.nih.gov/gene/ATM>
- American Cancer Society <https://www.cancer.org/cancer/prostate-cancer/about/key-statistics.html>
- "NCCN Clinical Practice Guidelines in Oncology Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic." NCCN, 4 Dec. 2019.
- Gatti R, Perlman S. Ataxia-Telangiectasia. 1999 Mar 19 [Updated 2016 Oct 27]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK26468/>