



Pathogenic (or harmful) variants in the ATM gene have been associated with an increased lifetime risk of breast cancer (up to 38-69% lifetime risk). Some studies have also suggested associated risk for other cancers, such as [pancreatic](#) or [prostate](#) cancer, although these are not as well established.

ATM gene variants related to cancer risk are inherited in an [autosomal dominant](#) pattern, meaning that children of someone who carries a variant each have a 50% risk to inherit the variant and associated cancer risks. Notably, women and men both have the ATM gene and have the same chances to inherit and pass down variants in these genes. Therefore, both sides of the family are important when assessing inherited risk.

<b>Cancer type</b>	<b>Risk in the general population</b>	<b>Risk in ATM pathogenic variant carriers</b>
Female breast	12%	38% - 69%

### **Genetic testing for ATM**

Genetic testing for pathogenic variants in ATM are currently available, but there are a few different ways to approach testing:

- [Single site analysis](#): Testing specific to a known mutation in the family
- Full gene [sequencing](#) and [rearrangement analysis](#): Comprehensive testing to search for all currently detectable mutations in the gene
- [Gene panels](#): Newer, more broadly based gene tests that would include not only the ATM gene, but other genes known or suspected to be associated with increased cancer risks

### **Cancer Screening & Management for ATM**

If you are tested and found to have a mutation in the ATM gene, it is recommended to discuss your management plan with your healthcare team, and if available, to seek consultation through a specialized high-risk clinic. General recommendations are included here based on updated guidelines of the [NCCN](#), but may be tailored to your specific medical and family history.

For women, breast cancer screening recommendations include starting annual mammograms at age 40 with consideration of breast MRI starting at age 40, or 5-10 years prior to the earliest diagnosis of breast cancer in the family, whichever is earlier.



If there is a history of other cancers in the family, your doctor or healthcare provider may recommend additional, earlier, or more frequent screening for those other cancers based on the family history.

### **Ataxia Telangiectasia**

The above information about cancer risks is relevant for people who have a single mutation in the ATM gene. Importantly, if a child inherits mutations in both copies the ATM gene (one mutation from mom and one mutation from dad), this causes a different genetic condition called ataxia telangiectasia. Ataxia telangiectasia is a rare [autosomal recessive](#) condition that causes progressive problems with movement and coordination (ataxia), enlarged blood vessels in the eyes and skin (telangiectasias), and increased risk for infection and cancer in children (e.g. leukemia, lymphoma). Therefore, people with a mutation in the ATM gene who are pregnant or planning to have children are recommended to seek genetic counseling to clarify the risk to their children.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about hereditary cancer predisposition.