# *ATM* gene

## Associated Syndrome Name: *ATM*-associated cancer risk

### *ATM* Summary Cancer Risk Table

| CANCER | GENETIC CANCER RISK |
|---|---|
| Breast | High Risk |
| Pancreatic | High Risk |
| Prostate | High Risk |

### *ATM* gene Overview

*ATM*-associated cancer risk [1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15]

- Women with *ATM* mutations have a risk for breast cancer that is significantly increased over the 12.5% lifetime risk for women in the general population of the United States. The increase in risk may be especially significant at young ages.
- Men and women with *ATM* mutations have an increased risk for pancreatic cancer. The exact risk has not yet been established, but the available data suggests that the risk is approximately 5% to age 80. The risk may be higher in individuals with a family history of pancreatic cancer.
- Men with mutations in *ATM* have an increased risk for prostate cancer. There is also evidence that prostate cancers in men with *ATM* mutations are more likely to be aggressive.
- There is some evidence that individuals with *ATM* mutations have an increased risk for other cancers, including melanoma, gastric, colorectal, and ovarian cancer. However, the increase in risk may be small, and there are currently no medical management guidelines related to these cancers.
- Although there are increased risks for cancer in men and women with mutations in *ATM*, there are interventions that may reduce these risks. Guidelines from the National Comprehensive Cancer Network (NCCN) that may apply are listed below. Since information about the cancer risks associated with *ATM* mutations is relatively new, and there is still some uncertainty about the best ways to reduce these risks, it may be appropriate to interpret these results in consultation with cancer genetics experts in this emerging area of knowledge.

### *ATM* gene Cancer Risk Table

| CANCER TYPE | AGE RANGE | CANCER RISK | RISK FOR GENERAL POPULATION |
|---|---|---|---|
| Female Breast | To age 50 [9, 16] | Up to 9% | 2.1% |
| | To age 80 [7, 8, 9, 16] | 17%-52% | 10.8% |
| Pancreatic | To age 80 [2, 3, 10, 16] | 5% | 1.1% |
| Prostate | To age 80 [1, 3, 16, 17, 18] | 26%-44% | 10.6% |

### *ATM* Cancer Risk Management Table

The overview of medical management options provided is a summary of professional society guidelines. The most recent version of each guideline should be consulted for more detailed and up-to-date information before developing a treatment plan for a particular patient.

This overview is provided for informational purposes only and does not constitute a recommendation. While the medical society guidelines summarized herein provide important and useful information, medical management decisions for any particular patient should be made in consultation between that patient and his or her healthcare provider and may differ from society guidelines based on a complete understanding of the patient's personal medical history, surgeries and other treatments.

| CANCER TYPE | PROCEDURE | AGE TO BEGIN | FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS) |
|---|---|---|---|
| Female Breast | Breast awareness - Women should be familiar with their breasts and promptly report changes to their healthcare provider. Periodic, consistent breast self-examination (BSE) may facilitate breast awareness.[19] | 18 years | NA |

| CANCER TYPE | PROCEDURE | AGE TO BEGIN | FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS) |
|---|---|---|---|
| | Clinical encounter, including clinical breast exam, ongoing risk assessment and risk-reduction counseling[19] | 25 years, or 5 to 10 years younger than the earliest age of breast cancer diagnosis in the family | Every 6 to 12 months |
| | Mammography[19] | Age 40, or modified to a younger age based on the family history of breast cancer | Annually |
| | Consider breast MRI with and without contrast[19] | 30 to 35 years, or modified to a younger age based on the family history of breast cancer | Annually |
| | Consider additional risk-reduction strategies.[19, 20] | Individualized | NA |
| Pancreatic | Consider endoscopic ultrasound (EUS) and/or contrast-enhanced MRI/magnetic resonance cholangiopancreatography (MRCP). It is recommended that patients who are candidates for pancreatic cancer screening be managed by a multidisciplinary team with experience in screening for pancreatic cancer, preferably within a study setting.[19] | Age 50, or 10 years younger than the earliest age of pancreatic cancer diagnosis in the family | Annually |
| | Provide education about ways to reduce pancreatic cancer risk, such as not smoking and losing weight.[21] | Individualized | Individualized |
| Prostate | Incorporating information about increased risk due to gene mutation, consider prostate cancer screening. Discuss potential benefits and harms of baseline digital rectal examination (DRE) and prostate specific antigen (PSA).[17, 19] | 40 years | Every 1-2 years, or adjusted based on PSA |
| For Patients With A Cancer Diagnosis | For patients with a gene mutation and a diagnosis of cancer, targeted therapies may be available as a treatment option for certain tumor types (e.g., PARP-inhibitors).[22] | NA | NA |

## Information for Family Members

The following information for Family Members will appear as part of the MMT for a patient found to have a mutation in the ATM gene.

This patient's relatives are at risk for carrying the same mutation(s) and associated cancer risks as this patient. Cancer risks for females and males who have this/these mutation(s) are provided below.

Family members should talk to a healthcare provider about genetic testing. Close relatives such as parents, children, brothers and sisters have the highest chance of having the same mutation(s) as this patient. Other more distant relatives such as cousins, aunts, uncles, and grandparents also have a chance of carrying the same mutation(s). Testing of at-risk relatives can identify those family members with the same mutation(s) who may benefit from surveillance and early intervention.

In rare instances, an individual may inherit mutations in both copies of the *ATM* gene, leading to the condition ataxia-telangiectasia (A-T). Most individuals with A-T will have symptoms in childhood, including neuronal degeneration, radiosensitivity and immunological deficiency. There is also a high risk of cancer, primarily leukemias and lymphomas. The children of this patient are at risk of inheriting A-T only if the other parent is also a carrier of an *ATM* mutation. Screening the other biological parent of any children for *ATM* mutations may be appropriate.[23]

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