**ATM Gene Information**

The *ATM* gene plays a role in preventing people from developing tumors or cancer. Everyone has two copies of *ATM* – one copy of the gene inherited from each parent. If you inherit one copy of an *ATM* gene with an alteration in the genetic code that prevents the gene from working properly, this leads to increased chances to develop cancer. Medically significant alterations in the genetic code are called “mutations” or “pathogenic variants.” Not everyone who is born with an *ATM* mutation will develop cancer – many will not.

### Cancer Risks

The table below shows the risks of cancer for people with *ATM* mutations compared to people in the general population. These are cancer risk estimates over a lifetime and do not reflect yearly risks. Certain interventions may be able to lower these risks.

<table>
<thead>
<tr>
<th>Cancer Type*</th>
<th>ATM Risk</th>
<th>General Population Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Female Breast</td>
<td>20-40%</td>
<td>12-13%</td>
</tr>
<tr>
<td>Pancreatic</td>
<td>5-10% **</td>
<td>1-2%</td>
</tr>
<tr>
<td>Ovarian</td>
<td>2-3%</td>
<td>1-2%</td>
</tr>
<tr>
<td>Prostate</td>
<td>Emerging information about possible increased risk</td>
<td>13%</td>
</tr>
</tbody>
</table>

*Female and male refer to sex assigned at birth, and these risks are based on data from mostly cisgender populations. In addition, general population cancer risks may vary by ancestry and other factors.

**Risk of pancreatic cancer may be higher in those who also have a history of: family history of pancreatic cancer, smoking, heavy drinking, and/or other risk factors.

- **Other cancers**: There may be other cancer risks associated with having an *ATM* mutation, such as colorectal and gastric cancer. Over time, we expect to have more information about cancers associated with this altered gene.

It is possible that the information about cancer risks associated with *ATM* could change over time as our knowledge about this gene expands.
Medical Recommendations

The recommendations below are meant as general guidelines and may change based on other personal risk factors or family history. To make an appointment to discuss personal recommendations with a Cancer Genetics Physician at Dana-Farber, please call 617-632-2178.

- **Breast cancer** screening with annual breast MRI beginning at age 30-35 years and annual mammograms beginning at age 40 are recommended. There is insufficient evidence for removal of both breasts (risk-reducing bilateral mastectomy) based on having an ATM mutation alone.

- **Pancreatic cancer** screening is recommended for individuals with a family history of pancreatic cancer. We recommend surveillance at a specialized center with annual endoscopic ultrasounds and/or MRI/MRCP starting at age 50, or 10 years earlier than the earliest diagnosis of pancreatic cancer in the family. At age 50, those without a family history of pancreatic cancer can consider the benefits and limitations of screening with their provider.

- **Ovarian cancer.** While available screening tests are not reliable, there is insufficient evidence for removal of the ovaries and fallopian tubes (risk-reducing salpingo-oophorectomy) based on having an ATM mutation alone. Recommendations for ovarian cancer risk-reduction may be personalized based on family history and other considerations.

- **Prostate cancer** screening is recommended with annual prostate specific antigen (PSA) beginning at age 40.

These recommendations are based on our current understanding of the ATM gene and may change over time. Recommendations are also influenced by other factors, including but not limited to: cancer treatments, family history, personal preference, and gender affirming care. We recommend that you discuss personalized recommendations with your physician.

Inheritance and Meaning for Your Family

It is important that you share your test results with your biological (blood-related) relatives. Your close relatives, including parents, siblings, and children, have a 50%, or 1 in 2 chance, to have inherited the ATM mutation as well. All individuals in the family can inherit and pass along ATM mutations to their children. Mutations in ATM do not skip a generation. If someone in your family did not inherit the ATM mutation, they cannot pass it along to children.

We recommend that young adults wait until they are at least 18 years old before seeking genetic testing. However, people may wait to do testing until the age their medical care would change if they carried the ATM mutation, and/or they feel emotionally ready to learn this information.

Family Planning

Rarely, a child can inherit two mutations of the ATM gene, one from each parent. Children who are born with two non-working ATM genes will develop a condition called ataxia telangiectasia. Ataxia telangiectasia can be a
serious childhood condition that affects the nervous system, immune system, and other body systems. We recommend that the reproductive partners of individuals with an ATM mutation consider comprehensive testing of the ATM gene to assess whether their children or future children are at risk for ataxia telangiectasia. A child can only inherit two mutations of ATM if both parents carry a mutation of this gene.

Further information about this, including options for lowering the chance of having a child with ataxia telangiectasia, can be discussed with a genetic counselor.

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Glossary of Cancer Genetics Terms

- **Early detection**: The process of finding cancer when it is just starting to develop.
- **Carrier**: A person who has a single mutation in a gene.
- **Gene**: A small piece of DNA that gives instructions for a specific trait.
- **Gender diverse individual**:
- **Heterozygous**: Having a single mutation in a gene.
- **Inherited**: Passed from biological parent to child (through egg and/or sperm).
- **Lifetime cancer risk**: The chance that a person will develop cancer in their life. This is sometimes defined as the chance of developing cancer by the age of 80 or 85.
- **Mutation**: A change in a gene that prevents it from working correctly. Also called likely pathogenic/pathogenic variant.
- **Risk-reducing surgery**: Surgery to remove healthy tissue or organs before cancer develops. Also called prophylactic surgery.
- **Surveillance**: Screening tests or procedures to look for early signs of cancer development or cancer returning (recurrence).
- **Female**: Refers to sex assigned at birth.
- **Male**: Refers to sex assigned at birth.
- **Cisgender**: A person whose sex assigned at birth matches their gender.