About *ATM* Gene Mutations

About Genes

Genes are in every cell in our bodies. Genes are made of DNA, which gives instructions to cells about how to grow and work together. We have two copies of each gene in each cell—one from our mother and one from our father. When genes work properly, they help stop cancer from developing.

When it works right, the *ATM* gene helps prevent cancer by repairing damage to your DNA. Sometimes changes to the *ATM* gene happen. These changes are called mutations. Mutations can make the *ATM* gene stop working and raise the risk for certain types of cancer.

If you have a mutation in the *ATM* gene, your risk of getting breast and pancreatic cancers is higher than average. The risks for other cancers may also go up with *ATM* mutations. Researchers are studying the *ATM* gene to understand more.

ATM Mutations and Cancer Risk

Breast Cancer

About 1 in 10 women get breast cancer during their lifetime. For women with an *ATM* mutation, about 2-3 in 10 get breast cancer during their life. The risk for women with an *ATM* mutation and a family history of breast cancer is closer to 3-4 in 10. Certain rare mutations, such as c.7271T>G, may raise the risk even more. Women with an *ATM* mutation who already had breast cancer have a higher risk to get a new breast cancer.

Pancreatic Cancer

Pancreatic cancer is rare. About 1 in 100 men and women get this type of cancer in their lifetime. The risk is somewhat higher for people with an *ATM* mutation. The risk is even higher for people with a family history of pancreatic cancer and an *ATM* mutation.

Recommendations

WOMEN

Women should practice breast awareness, which involves being familiar with their breasts. Report any changes to their healthcare provider.

Starting at age 35: Clinical breast exams by a doctor yearly.

Starting at age 40: Yearly mammogram and yearly breast MRI with contrast (scheduled 6 months apart). Depending on family history, this may start earlier in life.

Some medicines can lower the risk of getting breast cancer. Surgery to remove both breasts may be an option for some women who have a strong family history of breast cancer. We do not recommend surgery for most women with *ATM* mutations.

WOMEN AND MEN

Screening for pancreatic cancer has benefits and limitations. We do not recommend this screening for most people with *ATM* mutations. People who have an *ATM* mutation and a family hisotry of pancreatic cancer should be seen in a high risk clinic to discuss pancreatic cancer screening options. Screening may be considered at age 50 or 10 years prior to the youngest relative's age at pancreatic cancer diagnosis (whichever comes first).

KIDS AND SIBLINGS

Children and siblings of people with an *ATM* mutation have a 1 in 2 chance of also having the mutation. We recommend genetic testing and counseling for them after age 18.

If two people with *ATM* mutations have a child together, there is a 1 in 4 chance the child will have a condition called ataxia telangiectasia (AT). Signs of AT include enlarged blood vessels under the skin, uncoordinated movements, and other nervous system problems. If only one parent has a mutation in *ATM*, their children are not at risk for AT.

FAMILY MEMBERS WHO TEST NEGATIVE

Family members without the *ATM* mutation probably do not have a higher risk of getting cancer. The family history of cancer and other risk factors may raise their risk somewhat. Family members who test negative for the *ATM* mutation should talk with their doctor or genetic counselor about the right cancer screenings.

Do you have questions about your risk for cancer?
Our doctors and genetic counselors can help find the cancer screening plan you need.
Call Huntsman Cancer Institute's Family Cancer Assessment Clinic to learn more: 801-587-9555.