Liverpool Women's NHS Foundation Trust

ATM Gene alteration identified: Patient Information

Why have I been given this information sheet?

You have been given this information sheet because you, or someone in your family has been found to have an alteration in a gene called *ATM.* This sheet should accompany a letter from our Clinical Genomics Service which will have additional information and recommendations specific for your family.

What is *ATM*?

ATM is a gene that is known to be connected with breast cancer risk.

What cancer risks are there if you have an *ATM* gene alteration?

We estimate that someone who has an ATM gene alteration has approximately a 17-30% chance of developing breast cancer over their lifetime, falling into the moderate risk group. An exception to this is a specific ATM alteration, c.7271T>G, which confers a risk of breast cancer over 30% which is characterised as high risk.[1] These risks are compared to the general population risk of around 15% (1 in 7) which is considered a low risk of developing breast cancer. It is possible that *ATM* alterations also increase the risks of developing other types of cancer and these risks should hopefully become clearer in the future.

If you have had breast cancer, the *ATM* gene alteration is likely to be the main explanation for this. However, there may also have been other factors involved, both genetic and environmental, as well as an element of chance.

How is the *ATM* gene alteration inherited?

We have two copies of every gene including *ATM*. We give one copy of each gene to our children in the sperm and one copy in the egg. Someone with an *ATM* gene alteration usually has this alteration in only one copy of *ATM*. They also have a normal copy of the *ATM* gene. Their children can inherit either the normal or the altered copy. This means they have a 50% (1 in 2) chance of having the same gene alteration.

If someone with an *ATM* alteration has a partner who also carries an alteration in one of their *ATM* genes then there is a 25% (1 in 4) chance of both parents passing on an *ATM* alteration which leads to a rare condition known as ataxia telangiectasia. This is a rare condition that causes enlarged blood vessels under the skin and uncoordinated movements among other neurological symptoms. However, the chance of someone in the general population carrying an *ATM* alteration is low.

What cancer surveillance is available for individuals with an *ATM* alteration?

We recommend increased surveillance using mammograms from the age of 40 years. For those individuals why carry the high risk *ATM* alteration we recommend using a combination of breast MRI and mammograms from the age of 30 years. The aim is to detect breast cancers at an earlier stage, when the cancer has a better prognosis and is easier to treat. However, surveillance is not perfect and may miss cancers. In addition, sometimes changes are seen on the scans that later turns out not to be a cancer, but

[1]Goldgar, D.E., Healey, S., Dowty, J.G. *et al*. Rare variants in the ATM gene and risk of breast cancer (2011). *Breast Cancer Res*;13(4):R73. doi: 10.1186/bcr2919.

do cause worry and can lead to unnecessary extra tests. We are not currently recommending surveillance for other cancers.

What preventative surgery options are available for individuals with an *ATM* alteration?

At the moment we do not have enough long-term data on the breast cancer risks in *ATM* mutation carriers to know whether preventative removal of the breasts (also called risk reducing mastectomy) should be considered. Please talk to your Genetic Counsellor or Clinical Geneticist if you would like to look into this further.

Who should be offered a test for the *ATM* gene alteration?

Genetic testing for the *ATM* alteration may be helpful for your relatives. Genetic testing is only offered through a Clinical Genomics Service. Your Genetic Counsellor or Clinical Geneticist can provide a letter for your relative to take to their GP to request a referral to Clinical Genomics.

What can I do to help my risk if I have an *ATM* gene alteration?

We recommend all women minimise risk of breast cancer by maintaining a healthy diet and having plenty of exercise. You should also report any changes in your breasts to your GP. If you have a personal or family history of breast cancer you should discuss this with your GP before taking hormone replacement therapy.

Where can I get further information?

The AT society (ataxia telangiectasia) is a registered charity that support people who have two *ATM* alterations resulting in ataxia telangiectasia. Their website provides information and resources for people with this condition. Within the genetic aspects of A-T section on their website you can also find information about being a carrier of one *ATM* alteration.

Are there any research studies for *ATM*?

There are many new and exciting research studies looking at the impact of genomics on cancer development. One such study is the Epidemiological Study Of Familial Breast Cancer (Embrace) which aims to identify risk factors contributing to the development of cancer in those with an inherited cancer gene alteration. If you are interested in discussing the possibility of participating in any research studies please get in contact with the Clinical Genomics team at Liverpool Women's Hospital.

If you need more advice about any aspect of ATM, you are welcome to contact:

Liverpool Centre for Genomic Medicine
Liverpool Women's Hospital NHS Foundation Trust
Crown Street
Liverpool
L8 7SS
Telephone: 0151 802 5001 or 5008 Email: lwft.clingen@nhs.net

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This leaflet can be made available in different formats on request. If you would like to make any suggestions or comments about the content of this leaflet, then please contact the Patient Experience Team on 0151 702 4353 or by email at pals@lwh.nhs.uk

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