Clinical Genetic Service leaflet reference: 019

Southeast of Scotland Genetic Service
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ATM c.7271T>G Predictive Testing

Information for patients

A gene variant has been identified in your family that is thought to have contributed to the cancer diagnoses in your family. This means that a test is available for you, to help further clarify your cancer risk.

If you do not carry the gene variant, your risk of developing cancer may be similar to other people in the general population and your children would not be at risk of inheriting the gene variant.

If you carry the gene variant, your lifetime risk of breast cancer is significantly increased and your children or future children are at a 1 in 2 (50%) risk of inheriting the gene variant.

The following management options are available if you have inherited the ATM gene variant. Some of these **may** also be appropriate to consider should you decide not to proceed with genetic testing.

Cancer risks and management options for ATM c.7271T>G carriers

Female breast cancer: Studies estimate the lifetime risk of developing breast cancer with the above variant may be around 60%. However, an individual's actual risk is thought to be dependent on how many breast cancers there are in their immediate family.

Screening - screening for women at a significantly increased risk of breast cancer is by a combination of mammography and MRI scanning. Current recommendations are:

- 30-39 years: Annual breast MRI
- 40-50 years: Annual breast MRI and mammogram
- 51 years +: Mammogram every year

Risk reducing breast surgery: At present there is not enough long term data to know whether preventative mastectomy should be considered. It may be appropriate, particularly if there is a strong family history of breast cancer.

Risk reducing therapies: Certain medications, known as endocrine risk reducing therapies, have been shown to decrease breast cancer risk in women with a family history of breast cancer. However, there have been no specific studies of these medications in ATM carriers. Endocrine risk reducing therapies can be considered by ATM carriers in the context of a family history of breast cancer.

Be breast aware: We would encourage you to be breast aware and seek advice from your GP if you notice any changes in your breasts. The risk of male cancer is very low (similar to population risk). We would encourage male ATM carriers to be chest aware and report any changes or concerns to their GP for further assessment.

Pancreatic Cancer: Studies estimate an increased risk of pancreatic cancer in both men and women with an ATM variant. There is no effective screening or preventative therapies for pancreatic cancer at present although research is ongoing. ATM carriers who have a family history of pancreatic cancer may be eligible to take part in such research.

Healthy living: We would encourage you to maintain a healthy lifestyle to reduce your overall risk of developing cancer. Some lifestyle factors are thought to reduce the risk of breast cancer:

- Giving up smoking
- Maintaining a healthy weight and take regular exercise
- Limiting how much alcohol you drink.

At present there is not enough evidence to support an increased risk of additional cancers in association with ATM. Therefore, no other additional screening is currently advised.

Additional information

Family Planning: If you have inherited the variant, each of your children has a 50:50 (1 in 2) chance of inheriting the gene variant from you. This chance is the same for each pregnancy. There are several reproductive options available. This is a personal decision and there is no right or wrong decision. More information can be provided following your result.

Insurance and genetic test results: In 2018 the Association of British Insurers agreed to an open-ended moratorium which means that you will not have to disclose the results of your predictive genetic test to them when you are taking out insurance, unless their position changes. For further information please see: www.abi.org.uk/data-and-resources/tools-and-resources/genetics/genetics-faqs/

Ataxia-Telangiectasia (AT): In some cases, a baby may inherit an ATM gene variant from both their mother and their father resulting in a rare genetic condition called Ataxia-Telangiectasia (AT). This is mainly relevant if both parents are related to each other. Please speak to your Genetic Clinician if you have concerns about this.

For further information and support please see:

www.atsociety.org.uk/about-a-t/genetic-aspects-of-a-t/carrier-testing/

www.atsociety.org.uk/

Plan of action going forward

Please tick as appropriate		Enter details
	You have decided to proceed with testing and we have made the following arrangements for you to receive the results:	
	A follow up appointment has been arranged on:	
	You have decided to get back in touch when you feel ready to proceed with testing.	

Following testing, results are usually available within 6 weeks. Results are confirmed in writing to you and copied to your referring clinician/GP.

You were seen by:	Genetic Counsellor	Patient Label
Date:		