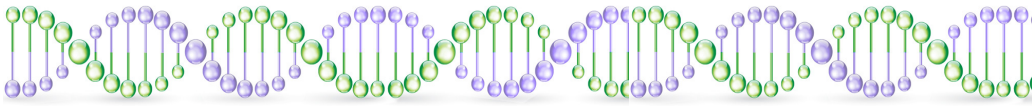
Information Sheet



SYDNEY CANCER GENETICS

Ataxia-Telangiectasia and the ATM gene

Ataxia-Telangiectasia syndrome is rare, affecting 1 in 40,000 to 100,000 people worldwide.

What causes Ataxia-Telangiectasia?

Ataxia-Telangiectasia occurs when an individual inherits mutations in the ATM gene from both their mother and their father (autosomal recessive inheritance). It causes severe disability and is usually diagnosed in early childhood. It is characterised by poor co-ordination and balance ("ataxia") and small clusters of enlarged blood vessels that are red or purple and often spidery in appearance and are seen in the eyes and on the skin ("telangiectasia"). The immune system may also be affected.

What if you inherit one ATM mutation?

A pathogenic ATM mutation is present in 1%-2% of the adult population. When an individual inherits only one copy of the ATM gene with a mutation, the Ataxia Telangiectasia doesn't occur. However, the lifetime risk of breast cancer is increased. The risk is estimated to be moderate (a 15 to 30% lifetime risk for women compared to the average risk of 10 to 15% lifetime).

For women who carry an ATM mutation, annual breast screening from age 40 is recommended.

One specific ATM mutation, written ATM c.7271T>G, is associated with a high lifetime risk of breast cancer. The risk is similar to carrying a germline BRCA2 gene mutation, with studies showing a lifetime risk of 30 to 80% for women. Some women would choose risk reducing mastectomy (removal of the breast tissue) at this level of risk. In the absence of bilateral mastectomies, annual breast screening, including breast MRIs, is recommended from age 30.

Can I have radiotherapy if I carry an ATM mutation?

Individuals with Ataxia Telangiectasia, who have 2 damaged ATM genes, are very sensitive to radiation. This is not true if you have just one ATM mutation. Current guidelines recommend women with one pathogenic ATM mutation have breast screening with mammograms. If a breast cancer is detected, standard treatment should be used, including radiotherapy if needed.

Is Ataxia Telangiectasia inherited?

Yes. Ataxia Telangiectasia is an autosomal recessive hereditary cancer syndrome. There is a 50% chance of a person who carries a single germline ATM mutation, whether male or female, passing the mutation to their son or daughter. If a mutation is identified, then predictive testing is available for blood relatives.

Does this sound like you or your family?

Has an ATM mutation been detected in you or a blood relative?
Make an appointment with Dr Hilda High at Sydney Cancer Genetics. It is a confidential opportunity to discuss your personal and family history of cancer and genetic testing can be organised, if needed.

More information is available on our website, including links to the following:

- BrAsh AT is a support group for individuals and families affected by Ataxia-Telangiectasia. It is based in Queensland but runs activities throughout Australia.
- The Cancer Genetics section of the Cancer Institute's eviQ website provides up-to-date Australian-based management guidelines
- The US National Library of Medicine website has more information about this syndrome.

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