



What is Ataxia-Telangiectasia?

Ataxia-telangiectasia (A-T) is an inherited disease that affects the nervous system, the immune system, and leads to an increased risk of cancer. It involves the absence or reduction of the ATM (ataxia-telangiectasia mutated) protein that is involved in the control of cell division, DNA repair, natural cell death, and other important cell functions. Symptoms associated with ataxia-telangiectasia are due to lack of control in cell growth and DNA repair, which leads to premature cell death or the formation of cancer cells. Ataxia-telangiectasia is also known as Louis-Bar syndrome.¹

What are the symptoms of Ataxia-Telangiectasia and what treatment is available?

Ataxia-telangiectasia is a progressive disease, typically presenting before five years of age. The symptoms of A-T may include:²

- Ataxia (difficulty coordinating movement)
- Chorea (involuntary muscle movements)
- Oculomotor apraxia (difficulty moving the eyes)
- Slurred speech and swallowing difficulties
- Oculocutaneous telangiectasias (tiny, red "spider" veins, appearing in the eyes and skin)
- Recurrent respiratory infections (due to decreased lung function)
- Cancer (particularly leukemia and lymphoma)
- Premature aging

Currently there is no cure for A-T, and life expectancy is shortened. For most individuals with A-T the life expectancy is more than 25 years of age, although some individuals have lived into their 50s. Treatment for A-T is supportive and can include intravenous medications and aggressive lung therapies to prevent and treat infections. Cancer treatment is challenging due to the increased toxicity experienced by individual with A-T that is associated with chemotherapy and radiation.²

How is Ataxia-Telangiectasia inherited?

A-T is an autosomal recessive disease caused by mutations in the *ATM* gene.¹ An individual who inherits one copy of an *ATM* gene mutation is a "carrier." Carriers are not affected with A-T, but may be at an increased risk for coronary heart disease and malignancies, particularly breast cancer in females.¹ An individual who inherits two mutations in this gene, one from each parent, is expected to be affected with A-T.

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Ataxia-Telangiectasia?

Ataxia-telangiectasia can occur in individuals of all races and ethnicities, and it is estimated to affect approximately 1 in 100,000 to 1 in 40,000 live births worldwide.¹



Estimated Carrier Frequencies for Select Ethnic Groups

Ethnicity	Carrier Rate
North African Jewish	1 in 81 ³
Norwegian	1 in 197 ⁴
General	1 in 100 ²

Having a relative who is a carrier or who is affected can also increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

- A-T Children's Project: <http://www.atcp.org>
- A-T Society: <http://www.atsociety.org.uk>

References

1. Ataxia-telangiectasia. *Genetics Home Reference* Available at <http://ghr.nlm.nih.gov/condition/ataxia-telangiectasia> Accessed on: February 17, 2012
2. Gatti, R. Ataxia-Telangiectasia. *GeneReviews*. Available at <http://www.ncbi.nlm.nih.gov/books/NBK26468/> Accessed: February 17, 2012
3. Gilad S *et al.* Ataxia-telangiectasia: founder effect among North African Jews *Hum Mol Genet* 1996; 5(12):2033-2037.
4. Olsen J *et al.* Cancer in patients with ataxia-telangiectasia and in their relatives in the Nordic countries *J Nat Can Inst* 2001 93(2): 121-127.