

### What is ataxia-telangiectasia?

Ataxia-telangiectasia is an inherited disease that affects the nervous system and the immune system and is associated with an increased risk of cancer. In individuals with ataxia-telangiectasia, the ATM (ataxia-telangiectasia mutated) protein is absent or reduced, affecting 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.<sup>1</sup>

### 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:<sup>2</sup>

- 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 ataxia-telangiectasia, and life expectancy is shortened. For most individuals life expectancy is more than 25 years of age, and some individuals have lived into their 50s. Treatment is supportive and can include intravenous medications and aggressive lung therapies to prevent and treat infections. Cancer treatment is challenging because individuals with ataxia-telangiectasia experience increased toxicity associated with chemotherapy and radiation treatment.<sup>2</sup>

### How is ataxia-telangiectasia inherited?

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

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.<sup>1</sup>

**Estimated carrier frequencies for select populations**

Population	Carrier frequency
North African Jewish	1 in 81 <sup>3</sup>
Norwegian	1 in 197 <sup>4</sup>
Worldwide	1 in 100 <sup>1</sup>

Having a relative who is a carrier or who is affected can 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>

AT 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.