Ataxia-telangiectasia (A-T)

Ataxia-telangiectasia (A-T) is a rare hereditary condition that affects the nervous system, immune system, and other body systems. Other names for A-T include **Louis-Bar syndrome**, **cerebello-oculocutaneous telangiectasia**, and **immunodeficiency with ataxia telangiectasia**.

- What are the effects of ataxia-telangiectasia (A-T)?
- How common is ataxia-telangiectasia (A-T)?
- What causes ataxia-telangiectasia (A-T)?
- How is ataxia-telangiectasia (A-T) diagnosed?
- What types of cancer are linked with ataxia-telangiectasia (A-T)?
- What are the cancer risks for people who carry an ATM gene mutation?
- How often should people with ataxia-telangiectasia (A-T) (or an ATM gene mutation) be screened for cancer?
- Questions to ask the health care team

What are the effects of ataxia-telangiectasia (A-T)?

People with A-T have trouble with movement and an increased risk of some types of cancer (see below).

Children with A-T may begin staggering and appear unsteady (called **ataxia**) shortly after learning to walk. Most people with A-T eventually will need to use a wheelchair. Over time, people with A-T tend to develop slurred speech and have trouble with writing and other tasks.

People with A-T also have weakened immune systems, are prone to infections, and appear to be particularly sensitive to ionizing radiation, such as x-rays.

How common is ataxia-telangiectasia (A-T)?

A-T is rare. It affects about 1 out of every 40,000 to 100,000 people. The chance that a person is a carrier of a single *ATM* gene mutation is about 1%, or 1 in 100.

What causes ataxia-telangiectasia (A-T)?

Changes (mutations) in the *ATM* gene on chromosome 11 cause A-T.

Both parents must have and pass on the mutated *ATM* gene for their child to have the condition. People with only one *ATM* mutation gene are **carriers** of the disease, but they do not have any symptoms.

Options exist for people who carry an *ATM* gene mutation and might want to have children. For more information, talk with an assisted reproduction specialist at a fertility clinic.

How is ataxia-telangiectasia (A-T) diagnosed?

A-T is initially suspected when a child develops signs of ataxia, meaning unsteady walking. To confirm if the child has A-T, imaging and blood tests are done, such as:

Genetic testing: This is a blood test to see if the child has a mutation in both of their *ATM*

types, including breast cancer[3], ovarian cancer[4], stomach cancer[5], melanoma[6], and sarcoma[7].

What are the cancer risks for people who carry an *ATM* gene mutation?

People with one *ATM* gene mutation (carriers) also have an increased risk of developing breast cancer. Studies have shown that carriers have about twice the lifetime risk of developing female breast cancer compared to the general population. We do not yet know whether there is an increased risk of male breast cancer among *ATM* carriers.

Some data suggests that *ATM* mutation carriers may also have an increased risk for ovarian, pancreas, skin and prostate cancers, but this isn't entirely clear.

More research is needed to clarify the risk of cancer and other conditions among *ATM* mutation carriers.

How often should people with ataxia-telangiectasia (A-T) (or an ATM gene mutation) be screened for cancer?

Cancer screening for people with A-T

Children and adults with A-T should see their doctor regularly and be monitored for signs of cancer. People with A-T who have frequent infections are encouraged to have their immune status checked regularly.

Cancer screening for *ATM* gene mutation carriers

Females with only 1 *ATM* gene mutation (carriers) also have an increased risk of developing breast cancer. Because of this, the National Comprehensive Cancer Network (NCCN) recommends that these women have yearly breast cancer screening with a mammogram[8] and possibly a breast MRI[9], often starting at an earlier age than normal.

Screening options may change over time as new technologies are developed and more is learned about A-T. It is important to talk with your health care team about which screening tests[10] to have and when to have them.

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