

Ataxia-telangiectasia (AT or A-T), also referred to as **ataxia-telangiectasia syndrome** or **Louis-Bar syndrome** is a rare, neurodegenerative, autosomal recessive disease causing severe disability. Ataxia refers to poor coordination and telangiectasia to small dilated blood vessels, both of which are hallmarks of the disease. A-T affects many parts of the body:

It impairs certain areas of the brain including the cerebellum, causing difficulty with movement and coordination.

It weakens the immune system, causing a predisposition to infection.

It prevents repair of broken DNA, increasing the risk of cancer.

Symptoms most often first appear in early childhood (the toddler stage) when children begin to sit or walk. Though they usually start walking at a normal age, they wobble or sway when walking, standing still or sitting. In late pre-school and early school age, they develop difficulty moving their eyes in a natural manner from one place to the next (oculomotor apraxia). They develop slurred or distorted speech, and swallowing problems.

Some have an increased number of respiratory tract infections (ear infections, sinusitis, bronchitis, and pneumonia). Because not all children develop in the same manner or at the same rate, it may be some years before A-T is properly diagnosed. Most children with A-T have stable neurologic symptoms for the first 4-5 years of life, but begin to show increasing problems in early school years.

A-T is caused by a defect in the ATM gene, which is involved in the recognition and repair of damaged DNA. The prevalence of A-T is estimated to be as high as 1 in 40,000 to as low as 1 in 300,000 people.

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