CONDITION

Ataxia - telangiectasia

Learn how ataxia-telangiectasia affects movement, immunity, breathing, and cancer risk, and how coordinated care can manage symptoms and complications.

WHAT IT IS
Rare inherited multisystem condition

BODY SYSTEM
Nervous, immune and respiratory systems

MAIN SYMPTOMS
Poor balance and coordination, involuntary movements, telangiectasias, repeated infections

WHAT CAUSES IT
Inherited ATM gene changes

HOW IT IS DIAGNOSED
ATM genetic testing of both copies

MAIN TREATMENT
Supportive multidisciplinary care

CAN IT BE CURED?
No, genetic change cannot be reversed

IS IT AN EMERGENCY?
Sometimes, severe breathing trouble or uncleared choking

LONG-TERM OUTLOOK
Usually progressive, with lung damage and cancer risk

Overview

Ataxia-telangiectasia, often shortened to A-T, is a rare inherited condition that affects several body systems. It mainly causes worsening problems with balance and coordination, changes in immune function, and a higher risk of certain cancers.

"Ataxia" means difficulty with balance and coordinated movement. "Telangiectasias" are small, widened blood vessels that can be seen near the surface of the skin or in the whites of the eyes. These blood vessels are usually harmless and do not generally bleed.

A-T is caused by changes in the ATM gene. This gene helps cells respond to damaged DNA. When the gene does not work properly, DNA damage can build up and affect the nervous system, immune system, lungs, and other tissues.

Most people with classic A-T begin showing movement problems in early childhood. The condition usually progresses over time, but its course differs from person to person. Some people have a milder form, sometimes called variant A-T, with later onset and slower progression.

A-T matters because it requires care across several specialties. Movement and swallowing difficulties may affect daily activities and nutrition. Immune problems can increase the risk of infection. Lung disease and cancer risk also need ongoing attention. There is no cure at present, but coordinated treatment can reduce complications, support independence, and improve quality of life.

Symptoms

Early symptoms often appear when a child is learning to walk. A child may sway while standing, fall often, or have difficulty walking in a straight line. These signs can initially look like ordinary clumsiness.

Movement symptoms become more noticeable as A-T progresses. They may include:

Telangiectasias often develop after movement symptoms begin, although timing varies. They commonly appear in the whites of the eyes and may also occur on the ears, face, neck, or other areas exposed to sunlight. Their absence does not rule out A-T, especially in a young child or someone with a milder form.

Immune system problems vary widely. Some people have few infections, while others develop repeated sinus, ear, or lung infections. Infections may last longer than expected or return after treatment. Chronic cough, noisy breathing, or shortness of breath can also occur.

Other possible features include slowed growth, delayed puberty, fatigue, drooling, and difficulty maintaining weight. Some people develop insulin resistance or diabetes. Learning may be affected by physical limitations, fatigue, speech difficulty, or slower information processing. Overall intellectual ability may remain relatively preserved, so communication and school supports should not be based only on speech or movement limitations.

People with variant A-T may first develop symptoms during adolescence or adulthood. They may have tremor, abnormal muscle contractions, balance problems, or nerve-related weakness without the full pattern seen in classic childhood A-T.

- Unsteady walking and poor balance
- Difficulty coordinating the hands and arms
- Involuntary jerking, twisting, or writhing movements

- Tremor or muscle stiffness
- Slurred or slow speech
- Difficulty controlling eye movements
- Trouble chewing or swallowing

Causes and risk factors

A-T results from disease-causing variants in both copies of the ATM gene. It follows an autosomal recessive inheritance pattern. This means an affected person usually receives one altered copy from each biological parent.

Parents of a child with A-T are usually carriers. Carriers generally do not develop A-T because their other ATM copy is working. When both parents are carriers, each pregnancy has a chance of resulting in an affected child, a carrier child, or a child who inherited neither familial variant. A genetic counselor can explain the exact chances without making assumptions about an individual family.

ATM carrier status also has health implications. In particular, women with certain disease-causing ATM variants have an increased risk of breast cancer. Cancer screening decisions for carriers should be based on the specific genetic result, family history, age, and current professional guidance.

A-T is not caused by something a parent did during pregnancy. It is not contagious. Having relatives from the same ancestral or geographic community may increase the chance that both parents carry the same rare variant, but A-T can occur in any population.

Diagnosis

A clinician may suspect A-T from the pattern of progressive balance problems, abnormal eye movements, telangiectasias, frequent infections, or a family history of the condition. Because these features may not appear at the same time, diagnosis can be delayed.

Blood tests may support the evaluation. Many people with A-T have a high level of alpha-fetoprotein , or AFP, after early childhood. AFP is a protein that is normally higher before birth and usually falls during infancy. A raised AFP level is not specific to A-T and cannot confirm the diagnosis by itself.

Immune testing may measure infection-fighting blood cells and antibodies called immunoglobulins. Results differ among people with A-T. Some have low levels of certain

immunoglobulins or fewer lymphocytes, which are white blood cells involved in immune defense.

The diagnosis is usually confirmed by genetic testing that finds disease-causing variants in both copies of the ATM gene. In uncertain cases, specialized laboratories may test for ATM protein or its activity.

Other examinations may include neurological assessment, lung function testing, swallowing evaluation, nutrition review, and hearing or vision assessment when indicated. Doctors may also evaluate for other conditions that can cause ataxia.

Cells in people with A-T are unusually sensitive to ionizing radiation, such as X-rays and computed tomography, or CT, scans. This does not mean that all medical imaging must be avoided. Clinicians generally limit unnecessary exposure and use options such as ultrasound or magnetic resonance imaging, or MRI, when these can answer the same question. A medically necessary X-ray or CT scan may still be appropriate after the benefits and risks are considered.

Treatment and management

There is currently no treatment that reverses the underlying genetic change. Management focuses on preserving function, preventing infection and lung damage, supporting nutrition, and finding complications early.

Physical and occupational therapy can help maintain safe movement and participation in daily activities. Treatment may include stretching, positioning, energy conservation, and recommendations for walkers, wheelchairs, seating systems, or home adaptations. The goal is not to force normal movement but to improve comfort, safety, and independence.

Speech-language therapists can help with speech clarity and communication tools. They also assess swallowing. If food, drink, or saliva enters the airway, known as aspiration, changes in food texture, posture, pacing, or feeding methods may reduce risk. A feeding tube may be discussed when eating takes too long, weight cannot be maintained, or aspiration remains a concern. This decision should reflect the person's medical needs and preferences.

Respiratory care may include airway-clearance techniques, treatment of infections, and support for an effective cough. Lung specialists may monitor breathing and decide whether inhaled treatments, cough-assist devices, or other therapies are appropriate.

Immune treatment depends on test results and infection history. Some people need prompt or preventive antibiotics like amoxicillin or doxycycline . Immunoglobulin replacement may be considered when antibody deficiency is clinically important. Vaccination plans should be reviewed with an immunology team because immune function differs among people with A-T.

Whether a live vaccine is appropriate depends on the person's specific immune status.

A dietitian can help address low weight, constipation, diabetes risk, and the extra effort needed for eating. Care should also include dental health, sleep, mental health, education, and social participation.

Medicines are sometimes used for tremor, stiffness, involuntary movements, drooling, or other symptoms, but benefits vary. These treatments should be chosen and monitored by clinicians familiar with A-T.

Cancer treatment requires specialists who understand the condition's radiation sensitivity and possible medication toxicity. Standard radiation therapy is generally avoided when possible. Chemotherapy may need careful selection or adjustment, but treatment decisions depend on the cancer and the individual situation.

Complications

Repeated respiratory infections, weak cough, and aspiration can gradually damage the lungs. Possible complications include bronchiectasis, in which airways become permanently widened and collect mucus, and interstitial lung disease, which causes inflammation or scarring in lung tissue.

Swallowing difficulty may lead to poor nutrition, dehydration, or aspiration pneumonia. Reduced mobility can contribute to muscle tightness, abnormal spinal curvature, pain, and loss of bone strength.

A-T increases the risk of cancer, especially cancers of blood-forming and immune cells such as leukemia and lymphoma. Solid tumors can also occur, particularly as people grow older. Many symptoms that resemble cancer have other causes, but persistent swollen lymph nodes, unexplained bruising, unusual bleeding, ongoing fever, night sweats, marked fatigue, or unexplained weight loss should be assessed promptly.

Immune deficiency can make some infections more frequent or severe. Diabetes, liver abnormalities, and delayed sexual development may also occur.

Seek emergency medical care for severe trouble breathing, blue or gray lips, choking that does not clear, loss of consciousness, or another sudden life-threatening change. A new one-sided weakness, facial droop, or sudden major change in speech also needs emergency assessment and should not be assumed to be part of A-T.

Contact the clinical team promptly for fever when an immune or cancer specialist has provided a fever plan, worsening breathing, repeated choking, dehydration, inability to take needed fluids, or signs of a serious infection. Stable changes in balance, equipment needs, nutrition, or

school function can usually be addressed through routine follow-up.

Follow up

A-T benefits from regular, coordinated care. The exact schedule depends on age, symptoms, immune test results, lung health, and access to specialists.

Follow-up commonly involves neurology, clinical genetics, immunology, respiratory medicine, rehabilitation, nutrition, and speech-language therapy. Other specialists may include gastroenterology, endocrinology, oncology, orthopedics, and palliative care. Palliative care can support comfort and quality of life at any stage and is not limited to end-of-life care.

Appointments may review:

There is no single cancer screening plan that fits everyone with A-T. Screening and investigation must balance cancer risk with sensitivity to radiation. New or persistent symptoms should be evaluated rather than waiting for the next routine visit.

Genetic counseling can help relatives understand testing options, reproductive choices, and health considerations for ATM carriers. Families may also benefit from care coordination, disability services, school planning, and support organizations familiar with rare neurological conditions.

- Infections, cough, breathing, and airway clearance
- Swallowing safety, eating time, weight, and hydration
- Mobility, falls, pain, posture, and equipment needs
- Immune function and vaccination planning
- New lumps, bruising, persistent fever, or other possible cancer signs
- School, communication, emotional health, and caregiver needs

Sources

- GeneReviews. Ataxia-Telangiectasia. University of Washington, Seattle; updated January 19, 2023.
- Rothblum-Oviatt C, Wright J, Lefton-Greif MA, et al. Ataxia telangiectasia: a review. Orphanet Journal of Rare Diseases. 2016;11:159.

- Bhatt JM, Bush A, van Gerven M, et al. ERS statement on the multidisciplinary respiratory management of ataxia telangiectasia. European Respiratory Review. 2015;24(138):565-581.
- National Library of Medicine. Ataxia-telangiectasia. MedlinePlus Genetics.
- National Cancer Institute. Ataxia-telangiectasia and cancer risk. Cancer Genetics Overview.
- van Os NJH, Jansen AFM, van Deuren M, et al. Ataxia-telangiectasia: immunodeficiency and survival. Clinical Immunology. 2017;178:45-55.