A-T CHILDREN'S® PROJECT

CARING FOR PEOPLE WITH ATAXIA-TELANGIECTASIA

Ataxia-telangiectasia (A-T) is a genetic disease that causes loss of muscle control and balance, cancer, lung disease and immune system problems in children and young adults, shortening their lives.

The nonprofit A-T Children's Project partners with academic and industry investigators worldwide – organizing and supporting innovative research, conferences, clinical teams, data platforms and biomarker development – to optimize disease management strategies, develop new treatments and find a cure.

RESOURCES FOR CAREGIVERS

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A-T Cancer Consultations
For up-to-date information on cancer consultations, please visit **atcp.org/cancer-consultations**

International A-T Clinics
Please visit **atcp.org/international**

To find the information from this booklet online, please go to **atcp.org/Caring**
For more in-depth information, please go to **atcp.org/Handbook**

This guide aims to provide a clear understanding of A-T. Remember, each individual's experience with A-T can be different, and it's important to consult healthcare professionals for personal medical advice.

THANK YOU

Thank you to these clinicians for their dedication to the evaluation and treatment of people with A-T and for their work on this handbook:

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- Children's Hospital of Philadelphia: Sharon McGrath-Morrow, MD
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ATAXIA-TELANGIECTASIA (A-T)

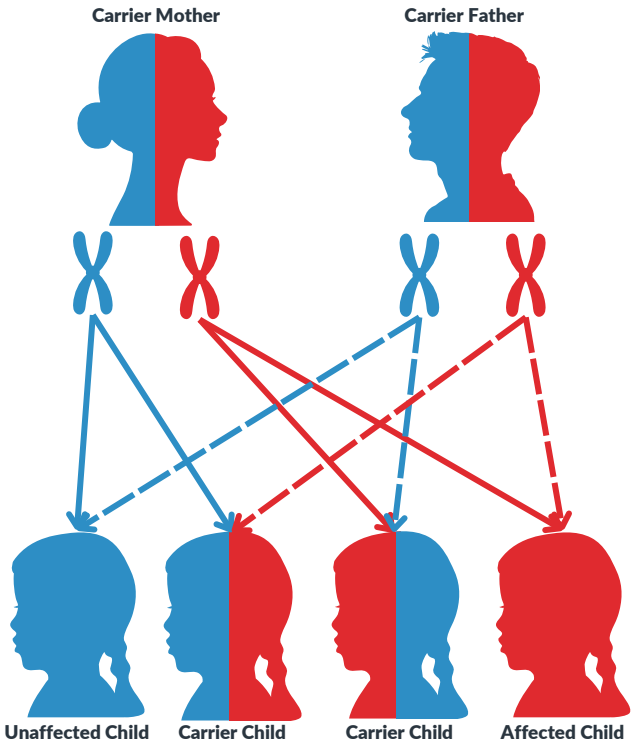
Ataxia-telangiectasia, **often abbreviated as A-T**, is a rare disease that is passed down in families. It affects various parts of the body. The main issues that people with A-T face include:

- **Motor Function Challenges:** This means difficulties with movements and coordination. It affects activities like walking, speaking, and movements of the eyes. Both large movements (like walking) and small movements (like writing) can be impaired.
- **Weakened Immune System:** People with A-T often have problems with immunity that can make them more likely to get infections or more likely to have serious infections.
- **Lung Disease:** There can be problems due to infection, difficulty clearing mucus from the airways by coughing, and swallowing incoordination that leads to inhalation of food and saliva into the lungs.
- **Impaired Feeding and Swallowing:** This may interfere with the safety and adequacy of oral intake, cause lung problems, interfere with growth and negatively affect overall well-being.
- **Increased Risk of Cancer:** There is a higher chance of developing certain types of cancer, especially but not limited to blood cancers (lymphomas and leukemias).
- **Growth Problems:** A-T can affect a person's growth in height and weight.
- **Delayed or Impaired Puberty:** Puberty might start late or progress differently for someone with A-T.
- **Diabetes:** There is an increased risk of developing diabetes compared to the general population.

WHAT CAUSES A-T?

INHERITANCE AND GENETIC FACTORS

Ataxia-telangiectasia (A-T) is an inherited disorder, similar to traits like eye color, height, and the risk of developing high blood pressure that are passed down in families. Our cells function based on instructions contained in genes, and each person has two copies of almost every gene: one from their mother and one from their father. A-T is an “autosomal recessive disorder” which means that the disease only occurs if both copies of a person’s ATM gene is defective (mutated). In almost all cases, that occurs because both parents carry one mutated copy of the ATM gene, and their child has inherited the mutated copy from each of the parents. People with one normal copy and one mutated copy of the ATM gene are carriers and are generally healthy themselves. It is important to know that siblings and other family members can have A-T or be carriers, and that the chances of that happening are equal in males and females.



AUTOSOMAL RECESSIVE INHERITANCE PATTERN

Because A-T requires both copies of the ATM gene to be mutated, its inheritance pattern is referred to as autosomal recessive. Carrier parents of a child with A-T have a 25% chance with each conception of having another child with A-T, a 50% chance of having a child who is a carrier of one ATM mutation, and a 25% chance of having a child without any ATM mutations.

Cellular Function and the Role of ATM Protein

Cells, which make up our bodies, require various proteins to perform their functions. The ATM protein is one of these essential proteins. Our DNA, which is like an instruction manual or recipe book, contains the instructions to create proteins, organized into individual recipes known as genes.

Genetic Variants and Mutations

Everyone has slight differences in their genes, known as “variants.” Most of these variants are harmless and contribute to our unique characteristics. However, some variants can be damaging, leading to errors in the genetic recipe and potentially causing health issues. These damaging variants, also referred to as mutations or pathogenic variants, can be inherited or occur spontaneously.

Impact of ATM Gene Mutations

The ATM protein is critical for managing cellular responses to various stressors, including DNA damage and metabolic stress. If a person inherits disease-causing mutations in both of their ATM genes, one from each parent, they will develop A-T. This results in low or absent ATM protein levels, affecting brain cells in the cerebellum responsible for coordinated movement, immune cells (lymphocytes), and many other cells in the body leading to the symptoms of A-T.

Risk of Cancer

Individuals with only one mutated ATM gene (A-T carriers) do not have A-T but have an increased risk of certain cancers, notably breast cancer in women. Refer to “A-T Carriers and Cancer Risk” on page 14.

HOW IS A-T DIAGNOSED?

Diagnosing A-T can be a challenging process due to its rarity. Often, children are initially thought to have conditions such as immunodeficiency, cerebral palsy, or other neurological disorders like Friedreich's ataxia before they are diagnosed correctly with A-T. The diagnostic process involves a combination of physical examination and laboratory tests. While an elevated alpha-fetoprotein level in the blood may raise suspicion, the definitive diagnosis of A-T should be confirmed through genetic testing.

FORMS OF A-T: CLASSIC AND MILD

Ataxia-telangiectasia manifests in two forms: classic and mild. The distinction between these forms lies in the severity of the mutations and the resulting function of the ATM protein.

CLASSIC A-T

- In this form, mutations in the ATM gene are severe. They typically lead to either the complete absence of ATM protein or the production of a non-functional version of the protein.
- Classic A-T is characterized by more pronounced symptoms due to the complete deficit in ATM protein function.

MILD A-T

- In a small minority of cases, mutations in the ATM gene allow for the production of a limited amount of functional ATM protein.
- Even a small percentage of normal ATM protein function, ranging from 5 to 10%, can lead to milder symptoms in A-T compared to the classic form.
- This reduced severity is due to the presence of some functional ATM protein, albeit at lower levels than normal.

The type and severity of the mutations in the ATM gene thus play a crucial role in determining the form of A-T and the extent of its symptoms.

A-T IS A MULTI-SYSTEM DISEASE THAT CHANGES WITH AGE.

Ataxia-telangiectasia (A-T) is characterized by a range of symptoms that evolve and change throughout an individual's life. The type, severity, and timing of these symptoms can vary significantly from person to person. Neurological problems often worsen during primary school years, typically between ages 5 to 14. However, individuals with A-T may experience a variety of health issues at different life stages.

FEATURES OF A-T DURING DIFFERENT STAGES OF LIFE

This table outlines the presence and progression of various symptoms and health challenges associated with A-T at different ages. It's important to note that not every person with A-T develops every symptom, and that the onset and severity of symptoms differ from person to person.

Features	< 5 years	5-15 years	15-20 years	> 20 years
Ataxia (Poor Coordination)	✓	✓	✓	✓
Deterioration of Neurologic Function		✓	✓*	✓*
Immunodeficiency	✓	✓	✓	✓
Infections (Nose, Sinuses, Lungs)	✓	✓	✓	✓
Pulmonary (Lung) Symptoms	✓	✓	✓	✓
Dysarthria (Speech Issues)		✓	✓	✓
Dysphagia (Chewing and Swallowing Problems)		✓	✓	✓
Telangiectasia		✓	✓	✓
Growth Failure		✓	✓	
Nutritional Compromise		✓	✓	✓
Lymphoid Cancers	✓	✓	✓	✓
Solid Tumor Cancers				✓
Liver Disease			✓	✓
Diabetes			✓	✓
Elevated Lipid Levels			✓	✓

*may have progression of extra uncontrolled movements or neuropathy

NEUROLOGY IN A-T

NEUROLOGIC PROBLEMS

The initial signs of neurologic issues in A-T typically emerge during the toddler years. While most children with A-T begin walking around the age of 12 months, they do not progress in motor coordination at a standard rate. An early prominent feature is difficulty standing still or sitting in a stationary position. Children with A-T tend to take staggered steps forwards and backwards, sway or lean almost precariously to one side or backward when trying to maintain position. Affected individuals tend to display better balance when running or walking quickly compared to walking slowly or standing still.

During the primary school years (approximately ages 5 to 12), children with typical forms of A-T have a progressive decline in walking and hand coordination. As the condition advances, a wheelchair becomes necessary for improved mobility, especially for longer distances, by about age 10. Although the loss of walking is a difficult transition, the use of motorized wheelchairs provides an effective way for independent mobility without fatigue.

Difficulty with controlling breath to make sounds and shaping them into clear words (speech articulation and phonation) may become noticeable during preschool years and may or may not deteriorate over time. Affected individuals often prefer communicating within their family and with friends rather than speaking in public or unfamiliar settings.

Throughout the primary school years, children frequently encounter increasing challenges with reading due to impaired coordination of eye movements and keeping them stable. Simultaneously, fine motor skills such as writing, coloring, using utensils, and personal grooming (brushing teeth and hair) may deteriorate. Generally, most of these neurologic problems stop progressing after approximately 12-15 years of age.

While instability and imprecision in both fine and gross motor movements, referred to as "ataxia," are common in all individuals with A-T, some may also experience additional movement control abnormalities superimposed on the ataxia. These movements include:

- Dystonia (characterized by a tendency toward a sustained twisted position, often affecting the head)
- Task-specific dystonic tremor (involving repetitive jerking triggered by attempting specific tasks)
- Chorea (manifesting as distal "piano playing movements" of the fingers)
- Athetosis (involving slower twisting movements of the hands and arms)

Some of these abnormal movements may worsen or not become apparent until after the age of 15. They may respond partially to medications used in the treatment of other movement disorders. While "tremor" is a commonly used term to describe general ataxia, specific tremor characterized by rhythmic oscillation is less typical. In certain cases, this form of tremor may also be amenable to medication.

ONGOING CLINICAL TRIALS HOLD PROMISE FOR POTENTIAL THERAPIES THAT MAY OFFER ASSISTANCE.

Overall, the primary neurological symptoms of A-T include:

- Poor coordination (ataxia) affecting fine motor tasks (involving upper extremities) and gross motor tasks (resulting in wobbliness while walking), which progressively worsens during the first 12-15 years of life.
- Difficulty in fine control of eye movement, leading to challenges in visual tasks such as reading.
- Involuntary movements, which may intensify during the later teenage and young adult years
- Slurred, slowed, or soft speech (dysarthria) and delayed responses to questions. This symptom often arises early and may not show clear progression compared to other neurological problems.
- Problems with chewing, swallowing, and self-feeding.
- Drooling.

Most of the prominent neurologic problems in A-T stem from damage to cells located in the cerebellum. This region of the brain plays a crucial role in coordinating the functions of other nerves involved in complex repetitive activities like walking. Under normal circumstances, healthy cerebellum function allows individuals to perform these activities smoothly without conscious thought.

WHAT A-T DOES *NOT* DO

Unlike other better known neurologic disorders like Alzheimer's or Parkinson's disease, A-T does not progress relentlessly to claim all neurologic functions. Most of the progressive decline slows or stalls out in early teen years.

A-T neurodegeneration does not impair:

- Awareness or judgement
- Memory
- Hearing
- Vision
- Personality
- Sense of humor

It rarely impairs control of bowel or bladder functions.

MANAGING NEUROLOGIC PROBLEMS

Currently, there is no known treatment to halt or slow the progression of the neurologic challenges associated with A-T. However, ongoing clinical trials hold promise for potential therapies that may offer assistance. At present, the approach to managing the neurologic problems of A-T focuses on alleviating symptoms and providing support. This involves various interventions, including physical, occupational, and speech therapies, along with appropriate exercise regimens. While these measures can help people with A-T manage day-to-day activities and tasks, they do not slow the overall course of neurodegeneration.

It is essential to emphasize that therapeutic exercise should be pursued with the goal of enhancing overall health. However, it should not be pursued to the point of inducing fatigue that interferes with daily activities. It is important to recognize that enhancing general coordination through training is not feasible in A-T. Instead, these therapies may offer valuable insights into potential "workarounds" or alternative strategies to improve the ability to complete specific tasks.

For individuals with A-T, consulting neurologists who specialize in movement disorders is advisable. These specialists are well-equipped to evaluate the condition and recommend appropriate treatments. Some medications used to manage symptoms of movement disorders, such as those related to Parkinson's syndrome or epilepsy, may prove beneficial in mitigating A-T symptoms. However, it is essential to note that the effectiveness of these drugs can vary among individuals with A-T.

It is also important to know that addressing the non-neurologic issues associated with A-T, such as undernutrition, appears to have a positive impact on neurologic function. Comprehensive care that includes addressing these aspects of the condition can contribute to a more holistic approach to managing A-T.

IMMUNOLOGY IN A-T

IMMUNODEFICIENCY AND PREDISPOSITION TO INFECTION IN A-T

Approximately two-thirds of people with A-T show immune system abnormalities in lab tests. However, not everyone with these abnormalities experiences symptoms. The most frequent lab findings include:

- Lower than normal levels of immunoglobulins (IgG, IgA, IgG subclasses and IgE).
- Decreased numbers of lymphocytes (particularly T-lymphocytes) in the blood.
- Reduced antibody responses to vaccines or infections.

These immune system issues can make individuals with A-T more likely to have frequent infections, both in the upper respiratory tract (colds, sinusitis, and ear infections) and the lower respiratory tract (bronchitis and pneumonia). That said, almost half of people with classic A-T do not have significant problems with infections, and people with mild A-T do even better. People with both forms of A-T may have more problems with infections, especially in the chest, as they get older.

MANAGING IMMUNODEFICIENCY AND INFECTION RISKS

It's important for everyone with A-T to undergo a thorough immunologic assessment at regular intervals, but at least once every two to five years even if prior assessments have been normal and the individual is not having problems with infections. This assessment should include:

- Measuring serum immunoglobulin levels (IgG, IgA, and IgM).
- Evaluating antibody responses to vaccines (such as pneumococcal conjugate, Hemophilus influenzae type b, and influenza vaccines).
- Counting the different types of lymphocytes in the blood (including CD4 and CD8 T-lymphocytes, and B-lymphocytes).

If the tests reveal significant immune system abnormalities, consulting an allergist/immunologist or infectious disease specialist is recommended. They can discuss treatment options that may include gamma globulin therapy, preventive antibiotics, and strategies to reduce exposure to infections.

Vaccinations can sometimes help mitigate the predisposition to infection. Standard childhood vaccines, particularly against common bacterial respiratory pathogens like Hemophilus influenzae, pneumococci, and the influenza virus, can enhance antibody responses, sometimes even in those with low immunoglobulin levels. Vaccines can also enhance other parts of the immune response, even in people who are not able to make antibody to the same vaccines.

VACCINE RECOMMENDATIONS FOR INDIVIDUALS WITH A-T

The vaccination schedule for those with A-T may vary based on their antibody function.

FOR ALL PERSONS WITH A-T

- **Influenza Vaccine:** An injected influenza vaccine (killed virus) should be administered annually, typically in the fall. This recommendation applies whether or not the individual is receiving gamma globulin treatment.
- **COVID Vaccination:** Follow the CDC's guidance for COVID mRNA vaccines.

IF ANTIBODY FUNCTION IS NORMAL

- **Routine Childhood Immunizations:** Except for the measles/mumps/rubella (MMR) vaccine, all standard childhood vaccinations, including those for other live viruses such as the varicella (chickenpox) vaccine, are recommended. *Note: The MMR vaccine might be linked to the development of chronic skin sores (granulomas) in a very small number of people with A-T, and therefore this vaccine should be avoided in all people with A-T.*
- **Pneumococcal Vaccination:** Those who have previously received the Prevnar-7 or Prevnar-13 vaccine should get the Prevnar-20 pneumococcal vaccine.
- **Prevnar-20 Vaccine Schedule:** The Prevnar-20 vaccine should be administered approximately every five years.

IF ANTIBODY FUNCTION IS ABNORMAL AND GAMMA GLOBULIN THERAPY IS RECEIVED

- Generally, routine childhood immunizations may not be necessary for these individuals, except for the influenza and COVID vaccines. Those are recommended because influenza and COVID rapidly mutate or change their cell surface proteins, so that gamma globulin does not always contain protective antibody to those viruses. Even though antibody responses may not be optimal, the vaccines may stimulate some antibody production and also stimulate the immune function of T-lymphocytes. A specialist in managing immunodeficiency can provide recommendations about the need for specific vaccines.

GAMMA GLOBULIN THERAPY FOR PEOPLE WHO DO NOT HAVE NORMAL RESPONSES TO VACCINES

If vaccinations are not effective and individuals with A-T continue to struggle with infections, gamma globulin therapy may be beneficial. This treatment involves intravenous (in a vein) or subcutaneous (under the skin) infusions of antibodies collected from people with healthy immune systems.

CONSISTENCY AND CHANGES IN IMMUNODEFICIENCY PATTERNS

Typically, the pattern of immunodeficiency in a person with A-T is established by the age of five and remains stable throughout their life. However, about 10-20% of individuals with A-T experience a decline in immune function over time. Worsening of infection-related issues at any age warrants a reassessment of immune function.

SWALLOWING FUNCTION AND LUNG INFECTIONS

When lung infections occur, evaluating the person's swallowing function is crucial. Difficulty coordinating swallowing can lead to aspiration (inhaling food or liquid into the lungs), increasing the risk of infections. See also “Feeding, Chewing, and Swallowing” on page 10.

LOW LYMPHOCYTE COUNTS AND VIRAL INFECTIONS

Most people with A-T have low lymphocyte counts, often specifically affecting CD4 T-cells. While this usually doesn't cause problems, it may result in a tendency to have chronic or recurrent viral skin infections, such as warts and molluscum contagiosum. Treatment to compensate for low CD4 cell counts is generally not necessary, except when someone with A-T is undergoing long-term treatment with corticosteroids (such as prednisone) or chemotherapy for cancer. In these cases, antibiotics might be recommended to prevent infections from certain types of germs, also called opportunistic pathogens.

RISK OF AUTOIMMUNE AND CHRONIC INFLAMMATORY DISEASES

Individuals with A-T have an increased risk of developing autoimmune or chronic inflammatory diseases. This risk is more likely a secondary effect of their immunodeficiency rather than a direct result of the lack of ATM protein. Common conditions seen in individuals with A-T include immune thrombocytopenia (ITP), arthritis and vitiligo, among others.

VACCINE RECOMMENDATIONS FOR HOUSEHOLD MEMBERS OF INDIVIDUALS WITH A-T

It's crucial for the health and safety of individuals with A-T that every household member, as well as those in close contact (such as grandparents and aides), have all standard age-appropriate vaccines. It is particularly important that they get annual vaccines for the main respiratory viruses.

- **Annual Influenza Vaccine:** Each fall, all household and close contact individuals should receive an injected influenza vaccine (killed virus type).
- **COVID Vaccination:** Adherence to the CDC's guidance for COVID vaccines is recommended for all household members and close contacts.
- **Measles/Mumps/Rubella (MMR) Vaccine**: This should be given to all previously unimmunized household members and close contacts. There is no risk to a person with A-T if another family member gets the MMR vaccine.

PULMONOLOGY IN A-T

LUNG DISEASE

Individuals with A-T may experience chronic lung disease in several ways:

- **Recurrent/Chronic Sinopulmonary Infections:** These frequent infections can lead to bronchiectasis, especially when compounded by immunodeficiency, poor airway clearance, swallowing dysfunction, and aspiration (inhaling liquid, food, or saliva into the lungs). Bronchiectasis is a condition where the bronchial tubes are permanently damaged, causing repeated lower airway infections.

- **Difficulty Clearing Airway Mucus:** Due to challenges in taking deep breaths and effectively coughing, individuals with A-T might struggle to clear airway mucus. This can lead to lung damage, exacerbate existing lung issues, and increase the severity of respiratory illnesses during common viral respiratory infections. Poor clearance of airway secretions may also cause airway inflammation and bacterial colonization.

- **Interstitial Lung Disease and Pulmonary Fibrosis:** While less common, some individuals with A-T develop interstitial lung disease or inflammation in the lung tissue that may or may not cause irreversible scarring (pulmonary fibrosis). This condition can reduce lung capacity, cause breathing difficulties, necessitate supplemental oxygen, and lead to chronic cough even in the absence of lung infections. Recurrent lung injuries from chronic infections, aspiration, or cancer treatments can contribute to lung fibrosis and scarring.

- **Restrictive Lung Disease Without Pulmonary Fibrosis:** This form of lung disease, characterized by the inability to take deep breaths during exercise or stress, can occur in A-T without pulmonary fibrosis. Severe scoliosis and related structural chest wall abnormalities can also lead to restrictive lung disease. All these problems are caused by reduced cerebellar function that ordinarily coordinates the muscles of the chest and diaphragm to facilitate an effective cough.

- **Swallowing Problems:** As many individuals with A-T age, they often develop swallowing problems, increasing their risk of coughing, wheezing, aspiration, and other lung injuries.

Importance of Regular Respiratory Assessments and Interventions

Regular respiratory assessments and appropriate interventions are crucial for managing the effects of chronic lung disease in A-T. Proper care and monitoring can significantly improve the quality of life for those affected. People with A-T should undergo lung function testing at least once a year starting at the age of five or when they can follow directions. Chest physiotherapy and/or cough assist devices can help with airway clearance and decrease the likelihood of pneumonias or airway disease caused by respiratory viruses or a weak cough. Some people will respond to bronchodilators which can further help with clearing mucus from the airways. Some people who have bronchiectasis will benefit from chronic antibiotic use. As people with A-T age, lung function may decrease significantly and non-invasive ventilation during sleep may help with chronic gas exchange abnormalities and quality of life during the day.

Respiratory Symptoms and Long-Term Consequences

Recurrent or chronic respiratory symptoms early in life can adversely affect long-term lung function. Both children and adults with A-T are at a higher risk of pulmonary exacerbations and declining lung function due to respiratory illnesses, and cancer treatments. When undergoing anesthesia or any surgical procedure, people with A-T should have an anesthesiologist experienced in caring for people with neuromuscular issues, and those procedures should generally take place in facilities with specialists in critical care and pulmonology in case they are needed.

PREVALENCE OF CHRONIC LUNG DISEASE IN A-T

It's important to note that **over 25% of individuals with A-T develop chronic lung disease**, and that lung disease is one of the leading causes of death in A-T. This emphasizes the importance of ongoing monitoring and management of respiratory health in this group.

IMPACT OF LUNG DISEASE ON SLEEP AND OVERALL HEALTH IN A-T

Sleep Quality and Health Concerns

Lung disease in individuals with A-T can significantly affect sleep quality, which in turn impacts overall health. Poor sleep efficiency can lead to fatigue and adversely affect various aspects of well-being. To evaluate and address these issues, sleep studies, also known as polysomnograms, are often recommended.

Identifying Breathing Problems Through Sleep Studies

These studies can detect several types of breathing problems that occur during sleep:

- **Upper Airway Obstruction:** Disruptions in sleep due to obstructions in the upper airway, often from low muscle tone.
- **Hypoxia:** Low oxygen levels in the blood during sleep.
- **Hypercarbia:** Elevated carbon dioxide levels in the blood.

If any of these conditions are identified, targeted treatments can be initiated.

INTERVENTIONS FOR BREATHING PROBLEMS DURING SLEEP

- **Supplemental Oxygen:** Providing extra oxygen to ensure adequate blood oxygen levels during sleep. This can improve oxygenation and alleviate sleep-related breathing issues.

- **Non-Invasive Ventilation:** Devices like CPAP (Continuous Positive Airway Pressure) or BIPAP (Bilevel Positive Airway Pressure) can assist with breathing and enhance sleep quality. They deliver a continuous or variable flow of air to help keep the airways open, reducing the risk of airway obstruction and hypoxia.

LINK BETWEEN INFLAMMATION AND LUNG FUNCTION

Recent research has indicated a potential connection between higher levels of inflammatory markers in the blood and reduced lung function in children and young adults with A-T. This association suggests that inflammation might contribute to the decline in lung function in A-T individuals. Addressing inflammation as part of the comprehensive management of A-T-related lung disease is important for optimizing health outcomes and preserving lung function.

COMPREHENSIVE CARE FOR MANAGING LUNG DISEASE IN INDIVIDUALS WITH A-T

Effectively managing lung disease in individuals with A-T requires a multifaceted approach. Here are the key considerations:

- **Annual Pulmonary Specialist Visits:** Starting from the age of two, it's important for individuals with A-T to have yearly appointments with a pulmonary specialist to monitor and manage lung health.
- **Lung Function Assessment:** Regular spirometry tests, including measurements of maximum inspiratory and expiratory pressures, are essential for assessing lung function.
- **Physical Activity:** Engaging in regular physical activity and exercise can help improve respiratory health and maintain pulmonary function. Activities should be moderate and not cause exhaustion, including for those using wheelchairs.
- **Upright Posture:** Maintaining an upright midline posture, especially during growth spurts, can improve chest wall development and gas exchange.
- **Airway Clearance Techniques:** Techniques like chest physiotherapy or cough-assist devices should be considered for individuals with acute or chronic chest congestion or a productive cough.
- **Aggressive Treatment of Infections:** Prompt and appropriate treatment of respiratory infections is crucial to prevent chronic lung disease.
- **Persistent Respiratory Symptoms:** If respiratory symptoms persist for more than seven days after an acute illness, a healthcare professional should evaluate the individual.
- **Immunodeficiency Management:** Timely identification and treatment of immunodeficiency can prevent bronchiectasis.
- **Chronic Antibiotics:** Chronic antibiotic treatment may be necessary for individuals with bronchiectasis.
- **Nutrition and Aspiration:** Proper nutrition and minimizing the risk of aspiration are important. In some cases, gastrostomy tube feeding might be recommended.
- **Interstitial Lung Disease Evaluation:** Symptoms like shortness of breath and hypoxia warrant evaluation for interstitial lung disease, with treatment options like oral steroids and other anti-inflammatory drugs being considered.
- **Reducing Environmental Risks:** Avoiding contact with smokers and limiting exposure to air pollutants and irritants is crucial.
- **Vaccination:** Adhering to recommended vaccine schedules as outlined in the "Vaccine Recommendations for Individuals with A-T" section on page 6 is vital.

Regular monitoring and comprehensive care are key to managing lung disease in individuals with A-T, ensuring their overall well-being.

FEEDING, CHEWING, AND SWALLOWING

FEEDING AND SWALLOWING CHALLENGES IN A-T

As individuals with A-T grow older, they often encounter difficulties with feeding and swallowing, which can significantly impact their health and quality of life. Here are some important aspects to consider:

Feeding Difficulties

- Involuntary movements can make feeding challenging, leading to messiness and longer mealtimes.
- Some may find it easier to self-feed using their fingers instead of utensils.

Swallowing Problems and Aspiration Risks

- Swallowing difficulties, or dysphagia, usually become more apparent after age 10 due to neurological changes associated with A-T.
- Dysphagia occurs due to the problems in coordinating movements within the mouth and throat that may make chewing and efficiency difficult. Foods that are difficult to chew include tough meats (e.g., steak, pork chops), raw fruit (e.g., apples), and raw vegetables (e.g., carrots).
- Difficulties involving the pharynx can lead to aspiration, where liquids, food, or saliva are breathed into the airways and can enter the lungs.
- Silent aspiration (aspiration without a cough) is a common concern in people with A-T age 10 and older. The lack of a cough often delays recognition of this problem.
- Aspiration poses a risk to lung health, underlining the importance of vigilant monitoring and intervention.
- Concerns about aspiration might lead to recommendations for a swallowing evaluation.
- If concerns about swallowing problems and aspiration persist, further assessments like a Videofluoroscopic Swallow Study (VFSS) or Flexible Endoscopic Evaluation of Swallowing (FEES) might be necessary. VFSS involves x-rays and should generally be avoided, if possible. The need for these tests should be discussed with healthcare providers and experienced speech-language pathologists. For children without signs of swallowing difficulties or those under 10 years, a baseline diagnostic swallow study is usually not recommended.

Importance of Addressing Feeding and Swallowing Problems

Properly managing feeding and swallowing challenges is crucial for the health and safety of individuals with A-T. It's essential to make mealtimes enjoyable and nourishing while minimizing risks related to dysphagia and aspiration.

WARNING SIGNS OF SWALLOWING PROBLEMS IN A-T

Individuals with A-T may show various signs that indicate difficulties with feeding and swallowing. Recognizing these signs is key for early intervention and effective management. Here are some common indicators:

- **Coughing or Choking:** Occurring during meals or while drinking.
- **Weight Concerns:** Inadequate weight gain, particularly during growth phases, or unexplained weight loss at any age.
- **Persistent Drooling:** Excessive drooling that continues beyond the typical teething stage.
- **Extended Mealtimes:** Mealtimes typically longer than 40 minutes.
- **Change in Food Preferences:** Reluctance to eat previously enjoyed foods and beverages, or difficulty in consuming them.
- **Chewing Difficulties:** Chewing takes too long or food may be swallowed before it is completely chewed.
- **Unexplained Lung Infections:** An increase in lung infections, particularly in individuals who also show other signs of swallowing difficulties.

Timely recognition of these problems is crucial for providing proper care. Addressing these challenges ensures that individuals with A-T receive adequate nutrition and helps reduce the risk of complications related to swallowing problems.

MANAGING FEEDING, CHEWING, AND SWALLOWING PROBLEMS

Optimizing Mealtime Efficiency

- **Duration of Meals:** Meals should ideally not exceed 30-40 minutes, as prolonged mealtimes can be stressful and disruptive, potentially hindering the intake of essential nutrients.
- **Caregiver Assistance:** If self-feeding extends meal durations, caregivers may need to assist. This can include helping with utensils or offering bite-sized food pieces. Directly placing food and liquids into the individual's mouth might also be necessary.

Enhancing Oral Intake with Safer Techniques

- **Chewing and Drinking Strategies:** Teaching safer methods for chewing, drinking, eating, and swallowing is beneficial. For instance, using a straw and maintaining a neutral chin position (parallel to the floor) can minimize the risk of aspiration.
- **Avoiding Certain Positions:** Tilting the head back, especially when drinking from an open cup, increases aspiration risk.

Professional Guidance and Dietary Modifications

- **Speech-Language Pathologist (SLP) Evaluation:** A SLP can offer tailored strategies to manage swallowing difficulties.
- **Dietician Consultation:** Dieticians can recommend dietary changes to increase caloric intake per bite, addressing nutritional concerns.

When to Consider a Feeding Tube

- **Inadequate Oral Intake:** Recommended when oral intake is insufficient for growth or maintaining weight.
- **Aspiration and Respiratory Concerns:** Necessary if aspiration is causing respiratory problems.
- **Challenging Mealtimes:** Considered when mealtimes are too long, stressful, or interfere with other activities.
- **Benefits of Feeding Tubes:** Feeding tubes (such as a “g-tube”) ensure adequate nutrition and hydration, reduce aspiration risks, and can improve the overall quality of life by simplifying feeding processes.

Feeding tubes are a supportive measure and do not prevent eating by mouth; they are an important tool in maintaining adequate nutrition.

NUTRITION AND GROWTH IN A-T

Maintaining a balanced and nutritious diet is critical to many aspects of A-T. For example, inadequate caloric intake can lead to decreased muscle mass, weakness, and fatigue, all of which will worsen neurologic function. Severe protein or caloric deficiencies can also suppress immune function. On the other hand, excessive caloric or carbohydrate intake can increase the underlying risk of developing diabetes, and diets with too many unhealthy lipids can increase the risk for elevated cholesterol and triglyceride levels.

GROWTH IN A-T

The absence of the ATM protein can interfere with the growth of many cells in the body and the rate of childhood growth. Most children with classic A-T grow normally until roughly two years of age, when their weight and height growth fall relative to the growth of children in the general population. This is seen when their growth percentiles drop on the standard growth charts used in a doctor’s office, called growth faltering. There are now specific growth charts that make it easier to recognize serious growth faltering in children with A-T.

PROBLEMS THAT MAY CAUSE GROWTH FALTERING IN A-T

- **Consuming too few calories:** Chewing and swallowing can be slow, difficult and tiresome. People with A-T may have difficulty finishing meals within customary timeframes such as school lunch breaks or family meals. They may become self-conscious about their eating and thus try to avoid eating much around others. The good news is that inadequate caloric intake is a problem that may be relatively easy to solve.

- **Consuming too few nutrients:** Some of the foods that are tastiest and easiest to chew are also the least nutritious. Avoiding fresh fruits and vegetables because they are difficult to chew can lead to vitamin deficiencies. The preference to eat less nutritious foods can be reinforced by caregiver concerns about getting their children to eat enough. In moderation, snack foods can increase calories consumed, but when too much of the diet is "junk food," the body doesn't get the high-quality nutrients that it needs. A nutritionist should be able to assist with suggestions and recommendations for your child to eat a better-balanced diet.

- **Medical-related causes:** Serious diseases such as recurrent infections and cancer, along with some of the medicines used to treat specific illnesses may affect growth and appetite.

- **Depression:** Chronic low mood can suppress appetite.

- **Neurologic problems:** Losing the ability to walk independently can lead to a sedentary lifestyle. The lack of exercise can hinder muscle development. Exercise can be an important way to increase appetite, as well as maintain heart/lung function and general muscle strength. Healthcare professionals, such as a physical therapist, should be consulted about the safest and most efficient exercise program.

It is important to consult with healthcare professionals, including dieticians, about how to best manage nutrition or growth problems, and for advice about a diet that supports a healthy lifestyle.

USING FEEDING TUBES FOR NUTRITION

When to Consider a Feeding Tube

A gastrostomy tube (G-tube) is the most common feeding tube. It is recommended when oral intake fails to provide enough nutrition or support overall health. It's a tube inserted through the abdominal wall into the stomach, allowing direct delivery of nutrition and water. Having the tube in place does not prevent eating by mouth, bathing, swimming, or other activities. It can be used as needed, and there is no necessity to use it every day. Consultation with a gastroenterologist or surgeon will help determine the best feeding tube for your child.

Benefits of Feeding Tubes

This method allows for safe and adequate calorie and liquid intake without the effort or problems that can occur with eating and drinking. Supplemental calories can even be administered during sleep. The aim is to ensure steady growth. A-T specific growth charts are very useful for assessing impaired growth.

CANCER IN A-T

CANCER RISKS

Individuals with A-T have about a 25% lifetime risk of developing cancer.
Here's what is known about cancer in people with A-T:

- **Cancer in Children (under 20 years old):** In children and adolescents, cancers of blood cells such as lymphomas and leukemias are the most common.
- **Cancer in Adults (over 20 years old):** Adults with A-T are not only at risk for blood cancers but also a variety of solid organ cancers. These types of cancers, including those of the gastrointestinal tract, skin, and breast, are typically seen in much older individuals in the general population. The key challenge in diagnosing these cancers in A-T patients lies in considering such a possibility in a young adult. Early diagnosis is very important for a better prognosis in solid organ cancers.
- **Risk Prediction:** Currently, there is no established method to predict which individuals with A-T are at the highest risk of developing cancer.

Understanding these features is vital for the early detection and management of cancer in individuals with A-T, contributing to improved outcomes.

SCREENING FOR BLOOD CELL CANCERS

Importance of Symptom-Based Screening: Routine screening for lymphoid cancers in the absence of symptoms has not been shown to be effective in early diagnosis. The treatment and prognosis for these cancers depend more on the characteristics of the affected lymphocytes rather than the extent of the disease at diagnosis. Therefore, we do not recommend routine screening for blood cancers at any age in the absence of symptoms.

Common Warning Signs of Lymphoma and Leukemia: The symptoms of blood cell cancers can be like those of mononucleosis, and include:

- **Recurrent or Persistent Fevers:** Fevers that occur frequently or persist without an apparent cause such as a cough or other obvious infection.
- **Easy Bleeding or Bruising:** Unusual susceptibility to bleeding or bruising.
- **Pale Appearance:** Noticeably paler skin than usual.
- **Swollen Lymph Nodes:** Enlargement of lymph nodes in areas like the neck, armpits, or groin.
- **Body Aches and Bone Pain:** Persistent or unexplained pain in the body or bones.
- **Unexplained Weight Loss:** Significant weight loss that can't be explained by changes in diet or exercise.

When to Seek Medical Attention: If any of these symptoms are present, it's important to consult a doctor. A physical examination, complete blood count, and possibly other tests will be necessary for a proper assessment.

SCREENING FOR SOLID TUMORS

General Recommendations for Screening

Clinicians and researchers are still trying to determine guidelines for when routine screenings such as breast MRIs and colonoscopies should be performed in the absence of symptoms. For now, without supporting data, we recommend the following screening tests:

- Annual physical exam for people with A-T at all ages. A complete head-to-toe exam means that people need to get out of their chairs, get undressed and lie down on an examination table. There are no shortcuts to this requirement. The exam should include checks relevant to the individual's gender (such as a breast examination, a Pap smear, and a prostate exam) as appropriate.
- Annual MRI scan of the chest, abdomen, and pelvis for people with A-T over the age of 20 years (although this may not be covered by all insurance policies because it is not an official recommendation from the expert panels on cancer screening).
- Staggered Scheduling. To ensure continuous monitoring, it's recommended to stagger the physical exam and MRI so that one or the other is done every six months.
- Guidance for Doctors. While more data are being collected, every doctor caring for a person with A-T needs to be aware that all kinds of cancers can occur in people with A-T, and that their differential diagnosis for many types of symptoms needs to include cancer even in people of relatively young age for cancer.

ADDITIONAL CONSIDERATIONS FOR PEOPLE WITH A FAMILY HISTORY OF CANCER

- If there is a known family history of colon, pancreatic, breast or prostate cancer, additional screening measures may be necessary. An oncologist should be consulted for guidance.

Treatment of Cancer

The special problems of managing cancer are substantially complicated, and treatment should be managed only in academic oncology centers after consulting with physicians who have specific expertise in A-T.

(See **atcp.org/cancer-consultations)**

All kinds of cancers can occur in people with A-T, and their differential diagnosis for many types of symptoms needs to include cancer even in people of relatively young age for cancer.

A-T CARRIERS AND CANCER RISK

Carriers (people with a mutation in only one of their two copies of the ATM gene that will usually include both parents and some family members of people with A-T) are at increased risk for certain types of cancer. Based on that risk, the following recommendations were made by the National Comprehensive Cancer Network (NCCN):

- Women who are A-T carriers have a 2.5-fold increased risk to develop breast cancer compared to the general population. As of 2025, the guidelines for screening include:
  - Yearly screening mammogram (with 3D mammography if available) beginning at age 40, or earlier if there is also a family history of breast cancer. (Note: At this time, there is not enough evidence to suggest that people with an ATM mutation need to avoid mammograms or other screening x-rays as recommended by their doctors.)
  - Consider yearly breast MRI with contrast beginning at age 30-35.

- Men who are A-T carriers have an increased risk to develop prostate cancer. Screening should start at age 40 and include a manual exam of the prostate (requires a rectal exam in the office) and a blood test to measure the PSA level.

- Men and women have a small increased risk for pancreatic and colorectal cancer. However, the increase is so small that enhanced screening is recommended only for A-T carriers who also have a family history.

Please note these recommendations change as new data becomes available through the NCCN. For updates, visit atcp.org/carrier.

- **Risk-Reducing Mastectomy for Women with ATM Mutation**
  - Women with an ATM mutation are advised to have a discussion with their healthcare provider about risk-reducing mastectomy. This decision should be based on their personal and family history of breast cancer.

- **Consultation for Updated Guidelines**
  - Carriers should regularly consult with their doctors to stay informed about any updates in screening guidelines or recommendations.

CANCER TREATMENT FOR A-T CARRIERS

- **Treatment Approach:** If A-T carriers develop cancer, their treatment plan often does not need to differ from standard protocols used for the general population. The treatment strategy should be tailored to the specific type of cancer and the individual's overall health condition.
- **Standard X-rays and CT Scans:** A-T carriers are not at an increased risk of adverse effects from exposure to standard x-rays or CT scans.
- **Cancer Treatment Involving Radiation:** Generally, A-T carriers do not face extra risk from therapeutic x-rays (radiation therapy) or radiomimetic drugs (drugs that work similarly to x-rays). However, the risk may vary depending on the type of ATM mutation present in an individual carrier.

OTHER FEATURES IN A-T

TELANGIECTASIA

- **On the Surface of the Eye:** Visible corkscrew shaped blood vessels on the white (sclera) of the eye typically appear by age 5–8 but can occur later or not at all. They are constant in nature, unlike other visible blood vessels that change over time. These telangiectases are only a cosmetic issue, and not symptomatic of infection or allergy.
- **On the Skin:** Similar blood vessels can occur on sun-exposed skin areas like the face and ears.
- **In Internal Organs:** Rarely, telangiectases develop internally and can cause complications in organs like the bladder, brain, liver, and lungs. This is particularly important in people with A-T who have had certain types of chemotherapy.

PUBERTY

- Development of puberty is often delayed or impaired in both males and females with A-T. Some females experience irregular menstrual periods, incomplete pubertal development, or cessation of periods at a very young age. Low-dose estrogen may be prescribed to regulate periods. A consultation with a gynecologist is advised for these issues.

ENDOCRINE ABNORMALITIES

- **Diabetes:** Commonly occurs due to insulin resistance rather than insulin deficiency. Management by a specialist, weight control, and good nutrition are essential.
- **Early Menopause:** Linked to low estrogen levels, causing symptoms like hot flashes and increased risk of osteoporosis. Supplemental estrogen can be prescribed, but specialist advice is necessary.

SKIN AND HAIR

- This includes immune-related conditions like vitiligo (blotchy loss of pigment in the skin), warts, molluscum contagiosum, and chronic inflammatory skin lesions (granulomas). Granulomas may be linked to MMR vaccination and require the help of a specialist.

ORTHOPEDICS

- **Foot Deformities:** In-toeing, out-toeing and other deformities of the feet are a relatively common result of neuropathy, a malfunction of the long nerves that go from the spinal cord to the feet. If the deformities cause pain or other difficulties with walking or bearing weight with transfers, surgery may be useful.
- **Scoliosis:** Severe cases may require spinal fusion surgery, which is complex and should only be performed at a major medical center that has specialists in spinal surgery as well as neurology, pulmonology, and intensive care.

PREMATURE AGING

- Early graying of the hair and early menopause are features that sometimes appear in A-T.

VISION

- Vision may be normal but visual function may be impaired due to difficulty controlling eye movements. This will affect tasks like reading. Eye misalignments (strabismus) are common and sometimes require surgery.

LIVER

- Liver abnormalities are common in older individuals with A-T. These include chronic inflammation that sometimes leads to cirrhosis, and a metabolic syndrome that includes problems with lipid levels and control of blood sugar levels. Annual liver function tests, lipid level checks, and diabetes screening (hemoglobin A1c) starting in the teenage years are recommended. Specialist referral is advised for persistent abnormalities.

GENERAL MEDICAL CARE IN A-T

SURGERY AND PROCEDURES REQUIRING SEDATION OR ANESTHESIA

- **Hospital Selection:** Surgeries should be conducted at centers equipped with an Intensive Care Unit (ICU). Even dental procedures requiring sedation can be high risk, depending on the individual's age and lung function.
- **Pre-Operative Lung Function Evaluation:** A thorough evaluation should be conducted for all A-T patients before surgery, regardless of age or history of breathing problems. This helps anesthesiologists assess risks and anticipate complications.
- **Post-Anesthesia Considerations:** Individuals with A-T may have difficulty coming off the ventilator after general anesthesia. Alternatives to general anesthesia and strategies for maximizing airway clearance after anesthesia should be explored.

GUIDELINES FOR X-RAYS

- **Increased Sensitivity to Ionizing Radiation:** Individuals with A-T are more sensitive to ionizing radiation, which includes x-rays and gamma rays. Therefore, x-rays should only be used when medically necessary, as exposure can cause cell damage that the body can't repair. Exposure to other forms of radiation, like ultraviolet light, does not require special precautions.
- **When to Perform X-Rays:** X-rays should generally be done only if the results will impact medical management.
- **Diagnosing Pneumonia:** If symptoms like fever, cough, and abnormal breath sounds suggest pneumonia, a clinical diagnosis and antibiotic treatment can be initiated without x-ray confirmation. However, a follow-up chest x-ray or even chest CT scan may be useful if symptoms persist.
- **Dental X-Rays:** Routine screening dental x-rays should be avoided. However, x-rays to evaluate tooth pain or another specific problem, may be reasonable.
- **Minimizing Radiation Exposure:** For necessary x-rays, use frontal view chest radiographs or radiation-sparing techniques for CT scans.
- **MRI and Ultrasound:** There are no contraindications to MRI and ultrasound exams.

DENTAL CARE GUIDELINES IN A-T

- **Minimize Radiation Exposure:** Due to increased sensitivity to ionizing radiation, routine dental x-rays should be avoided unless necessary, such as for diagnosing tooth pain.
- **Regular Dental Check-Ups:** Regular dental visits are important for maintaining oral health. These check-ups can help in early detection and management of dental issues without relying heavily on x-rays.
- **Communication with the Dentist:** Ensure the dentist is aware of the A-T diagnosis. Discuss any specific concerns, such as difficulties with sitting still for long periods due to motor control issues, or challenges with keeping the mouth open.
- **Sedation and Anesthesia Considerations:** If sedation or anesthesia is required for a dental procedure, discuss the potential risks and necessary precautions with both the dentist and the patient's primary care physician. Individuals with A-T may have specific sensitivities to certain medications or anesthesia. Consultation with a pulmonologist in advance of the dental procedure is recommended in the following situations:
    - Prior to sedation for all children older than 10 and for children younger than 10 with a history of asthma, chronic/recurrent cough or more than one pneumonia per decade of life.
    - Prior to any procedure at any age that will need elective general anesthesia.
- **Accommodating Motor Difficulties:** For patients with motor control challenges, additional support or adaptive equipment might be needed to ensure comfort and safety during dental procedures.
- **Oral Hygiene Education:** Given potential difficulties with fine motor skills, education on adaptive techniques for effective oral hygiene at home is crucial. This might include using electric toothbrushes or modified flossing tools.

CARE THROUGHOUT LIFE FOR INDIVIDUALS WITH A-T

Individualized Long-Term Care:

- A-T presents unique challenges at different life stages. While general issues are outlined in this handbook, it's important to remember that each person with A-T is unique.
- Long-term, individualized follow-up care with healthcare providers familiar with their specific health concerns is essential.
- A primary care provider should conduct a comprehensive physical exam annually for every person with A-T.

COMPONENTS OF AN ANNUAL PHYSICAL EXAM

- **General Health:** Monitoring height and weight, particularly during growth phases and instances of weight loss.
- **Infection Assessment:** Evaluating the frequency and severity of infections.
- **Lung Disease Indicators:** Checking for signs and symptoms of lung disease, and obstructive sleep apnea.
- **Feeding/Swallowing Screening:** Checking for signs and symptoms of mealtime difficulties.
- **Diabetes Screening:** Recommended for individuals over 10 years old or those who are overweight.
- **Liver Function:** Checking liver function in individuals over 10 years old.
- **Lipid Abnormalities:** Screening for cholesterol and triglyceride levels in people over 10 years old.
- **Cancer Screening:** For adults, this includes a full skin and breast exam, and considering the need for breast MRI, colonoscopy, and upper GI endoscopy based on age, history of gastroesophageal reflux, and family history.
- **Mental Health Screening:** Assessing for issues like depression.

LABORATORY TESTS AS PART OF ANNUAL PHYSICAL EXAM FOR A-T

Test	< 10 years	10-20 years	> 20 years
CBC (Complete Blood Count) / Differential	Yes	Yes	Yes
CMP (Comprehensive Metabolic Panel)	-	Yes	Yes
Lipids (Cholesterol and Triglycerides)	-	Yes	Yes
Hgb A1C (Hemoglobin A1C)	-	Yes	Yes
AFP (Alpha-Fetoprotein)	Once every 5-10 years	Once every 5-10 years	Once every 5-10 years
Immunologic Tests	Once every 5 years	Once every 5 years	Once every 5 years
Oncology Screening	-	-	Annual MRI of chest, abdomen, pelvis
Breast Screening (Mammograms or MRI)	-	As Needed	*
Colonoscopy	-	As Needed	*

*Decisions regarding mammograms, MRI for breast screening, and colonoscopy should be made in consultation with healthcare providers, considering factors like family history and individual health status.

NOTES ON LABORATORY TESTS

- **As Needed:** Indicates the need for individualized decision-making based on personal and family medical history.
- **Oncology Screening through MRI:** Recommended for individuals over 20 years of age (although this may be an out-of-pocket cost paid by the family as it is not routinely covered by insurance).
- **AFP:** Periodic AFP measurements can help differentiate classic from mild A-T in the first 10 years. As AFP levels naturally increase with age, tracking these levels over time can establish a trend that may assist in understanding disease progression.

SPECIALIST CONSULTATIONS THROUGHOUT LIFE

Most individuals with A-T will benefit from seeing various specialists throughout their life. The schedule below provides a framework for the healthcare of individuals with A-T, emphasizing the importance of regular monitoring and specialized care at different ages.

Specialist	At A-T diagnosis	1-10 years	10-20 years	> 20 years
Primary Care	Annual	Annual	Annual	Annual
Neurology	Yes	As Needed	As Needed	As Needed
Immunology	Yes	Every 5 years	Every 5 years	Every 5 years
Pulmonology	-	Annual from age 2	Annual	Annual
Speech (Feeding/Swallowing)	As Needed	As Needed	As Needed	As Needed
Genetics	Yes	-	-	-
Oncology	Yes	As Needed	As Needed	As Needed
Ophthalmology	-	As Needed	As Needed	As Needed
Orthopedics	-	As Needed	As Needed	As Needed
Rehabilitation	As Needed	As Needed	As Needed	As Needed
Nutrition	-	As Needed	As Needed	As Needed
Dental Care	-	Routine, but minimize x-rays except for limited diagnostic purposes	Routine, but minimize x-rays except for limited diagnostic purposes	Routine, but minimize x-rays except for limited diagnostic purposes

NOTES ON SPECIALIST CONSULTATIONS

Please note that additional visits should be made with appropriate specialists to help manage specific issues that arise. The frequency and type of specialist visits will vary based on individual health needs, new or changing symptoms, and at times of function changes.

- **As Needed:** Indicates the need for individualized decision-making based on personal and family medical history.
- **Genetics:** Should ideally be consulted at the time of diagnosis. Discussions should cover the risk of having another child with A-T, preventative steps that can decrease the risk of having another affected child (like in-vitro fertilization with testing of fertilized eggs to select those without A-T mutations), detection of carriers in the family, and cancer risks.
- **Neurology:** Should provide counseling on what to expect in the coming years and offer help with specific neurological problems.
- **Oncology**: Should provide counseling on signs and symptoms of cancer in a A-T patient, especially leukemia and lymphoma.
- **Rehabilitation:** Specifically, Physical Medicine & Rehabilitation Specialist; or, Physical or Occupational Therapist.

EDUCATION AND SOCIALIZATION IN A-T

School Experience

- **Adapting to School Challenges:** Children with A-T often enjoy school when proper accommodations are made. Delays in response time, slurred speech, impaired motor control, and multitasking difficulties can impact their learning experience.

- **Educational Placement:** Decisions about educational placement should consider the child's age, needs, and available local resources. These decisions need regular reevaluation.

Social Awareness and Skills

- Most individuals with A-T are socially aware and skilled, benefiting greatly from sustained peer relationships. Schools that allow for long-term friendships are ideal.

Intellectual Abilities and School Performance

- Intellectual disabilities are not typically part of A-T, though school performance may be affected by difficulties in reading, writing, and speech. Understanding and addressing these "input and output" challenges can lead to significant improvements.

Appearance and Fatigue

- Children with A-T are often conscious of their appearance and strive to be like their peers, which can be physically and mentally taxing. A shortened school day may be beneficial for some.

Most individuals with A-T are socially aware and skilled, benefiting greatly from sustained peer relationships. Schools that allow for long-term friendships are ideal.

SCHOOL RECOMMENDATIONS

- **Individualized Education Program (IEP):** Essential for addressing educational barriers. Balancing mainstream class benefits with performance issues is key.

- **Standardized Testing:** Modifications and cautious interpretation are necessary to accurately assess intelligence.

- **Social Interactions:** Critical for class placement. Establishing peer connections during school years is important.

- **Academic Accommodations:** Allow rest time, shortened days, reduced homework, and modified tests. Encourage children's involvement in problem-solving.

- **Classroom Aides:** Often necessary for assistance with various activities. Monitor the impact on peer relationships.

- **Physical and Academic Scheduling:** Avoid scheduling physically demanding activities before critical academic subjects.

- **Technology Use:** Early use of computers with assistive features and consultations with Assistive Technology specialists are recommended.

- **Communication Skills:** Speech-language pathologists can help improve communication efficiency.

- **Hearing:** Normal throughout life; audiobooks can supplement traditional materials.

- **Physical Education:** Adaptive physical education, including yoga, can be beneficial.

- **Physical Therapy:** Maintains strength and cardiovascular health but will not prevent or delay cerebellar degeneration.

- **Occupational Therapy:** Useful for managing daily living skills.

SPECIAL ISSUES FOR OLDER ADOLESCENTS AND ADULTS WITH A-T

Longer Lifespan

- Advances in managing nutrition, lung disease, immunodeficiency, and cancer have led to more individuals with A-T living beyond the age of 20. Adults with A-T face a distinct set of social and medical challenges.

Transitioning Post-High School

- A critical issue for many is determining life's direction after high school. Options may include attending community college, part-time work, or volunteering, and engaging in adaptive sports or social groups.

- Social isolation and a lack of purpose can significantly impact many adults with A-T. Therefore, planning for life after high school should begin in 9th grade, focusing on curriculum adjustments and life experiences to create and support a long-term plan.

Mental Health and Caregiving

- Counseling and regular screening for depression are essential for adults with A-T.
- As caregivers age, it becomes important to make decisions about long-term care and financial planning.

Newly Recognized Medical Problems in Adults

- Some adults develop issues beyond neurodegeneration, such as weight loss, muscle weakness, and other functional impairments such as rapid, uncontrolled movements of extremities or the head.
- Cancers other than lymphoma and leukemia are increasingly recognized (refer to the “Cancer in A-T” section on page 13).
- Asymptomatic low-grade but chronic inflammation of the liver can progress to clinically significant liver disease and even cirrhosis.

MENTAL HEALTH IN A-T

- Not surprisingly, the multiple problems caused by the lack of ATM, especially the decline in neurologic function that comes at an age when their peers are becoming more independent from adult help, causes many people with A-T to become depressed and anxious.

- Social isolation occurs because of difficulty communicating verbally (delay in initiation of speech and speech that is difficult to understand and very quiet), and because difficulties with gross and fine motor skills make it problematic for people with A-T to participate in the usual social activities in middle and high school.

- Counseling for the affected individual and the rest of the household, and sessions with trained professionals working in the classroom to explain the specific challenges that people with A-T face, may be helpful.

- Medications, such as anti-depressants, may also play a role in treatment.

ISSUES FOR CAREGIVERS

- As children with A-T get older, they become bigger and heavier, and they need more and more physical assistance from caregivers. This is an issue that needs to be considered as the parents and other caregivers become older and perhaps less able to manage all the necessary physical tasks.
    - Consider ways to make the home more accessible to someone with a wheelchair. This may be as simple as removing obstacles in a hallway, widening doorways or replacing outside steps with a ramp.
    - Replace bathtubs and standard showers with ones that have little or no lip so that a wheelchair can be moved as close as possible and the need for physical assistance is minimized.
- Like everyone else, caregivers need an occasional night off and an occasional vacation. If possible, they need to find a way to take a break from their caregiving tasks from time to time. That means finding or hiring someone who is willing to be trained and capable of taking over the care tasks for a night or a weekend occasionally.
- It is equally important for adult caregivers to consider the financial arrangements that may be necessary for the long-term care of a financially dependent child. This requires the help of a financial planner and an attorney.
- Adults with A-T should have an advanced medical directive and medical power of attorney designated in case they face a serious medical condition that renders them unable to take part in medical decision-making.

MENTAL HEALTH AND COUNSELING

- Long-term relationships with mental health professionals are beneficial for individuals with A-T and their families.
- Addressing the challenges of having a family member with a disability, including sibling and marital issues, is important.
- Establishing contact with a trusted counselor soon after diagnosis is recommended to ensure support is available when needed.

CLINICAL TRIALS

We are fortunate that A-T is attracting interest from a variety of medical researchers at universities and at pharmaceutical companies. Advances in the field can come only if people with A-T and their families are willing to take part in clinical trials. Some trials may be testing potential drugs, some may be testing ways to measure the disabilities caused by A-T, and some may be designed to better understand how the loss of ATM affects the function of individual cells or tissues. Some trials may require a substantial investment of time or carry a significant risk of side effects, while others may require only a clinic visit for a set of neurologic tests or collection of a small amount of blood. We urge families affected by A-T to watch for news about clinical trials, and to consider contacting the investigator to get enough information to determine if that trial might be one in which they feel comfortable enrolling.

A-T CHILDREN'S® PROJECT

atcp.org
kids.hope.cure.

QUICK INFORMATION CARD for Urgent Care Providers - Ataxia-Telangiectasia (A-T)

- Designed to provide essential information quickly to healthcare providers in urgent situations
- Available through the A-T Children's Project
- **Email info@atcp.org to request hard copies**
- Ideal for keeping in a wallet, purse, or car's glove compartment for easy access by caregivers
- Useful for sharing vital treatment information with doctors and other healthcare professionals

QUICK INFORMATION

FOR THE CARE OF PATIENTS WITH

ATAXIA-TELANGIECTASIA

NAME

DATE OF BIRTH

Ataxia-telangiectasia (A-T) is an autosomal recessive disease characterized by:

- progressive neurologic degeneration with ataxia, eye movement abnormalities, dysarthia and impaired swallowing,
- immunodeficiency with lymphopenia and hypogammaglobulinemia of varying severity,
  - ☐ I have antibody deficiency and receive gammaglobulin therapy
- predisposition to a wide variety of cancers (lymphoma and leukemia most common, especially in children),
- telangiectasia (often present), especially over the sclerae. In rare instances, telangiectasia may cause complications in bladder, brain and other organs.

Diagnostic X-Rays and CT Scans in A-T

People with A-T have an increased sensitivity to ionizing radiation (x-rays and gamma rays).

- X-rays should be performed only when the result will affect medical management.
- If a patient with A-T has fever, cough, and breath sounds characteristic of pneumonia, a diagnosis of pneumonia can be made clinically and antibiotics can be prescribed without x-ray confirmation. If symptoms persist despite antibiotics, a follow-up chest x-ray may be useful.
- Routine screening dental x-rays should be avoided, but an x-ray to evaluate tooth pain is reasonable.
- In order to keep radiation to a minimum, patients should receive frontal view chest radiographs or radiation-sparing techniques for CT (e.g. fewer CT images per scan).
- There is no contraindication to MRI or ultrasound investigation.

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A-T Cancer Consultations
For up-to-date information, please visit:
atcp.org/cancer-consultations

Management of Acute Infection

A-T patients are predisposed to upper and lower respiratory tract infections because of immunodeficiency, aspiration, and impaired cough.

- Use of antibiotics should be considered for treatment of upper or lower respiratory tract infections that are severe, accompanied by fever, or persist for greater than 7 days.
- Precautions should be taken to reduce the risk of aspiration during respiratory illnesses.

Preoperative Evaluation and Surgery

A preoperative evaluation of lung function should be performed in all A-T patients regardless of age and whether or not they have chronic respiratory symptoms.

- Children and adults with A-T may have difficulty coming off the ventilator after surgery or other procedures requiring general anesthesia.
- Possible alternatives to general anesthesia and strategies that maximize airway clearance following anesthesia should be considered.
- Following placement of a gastrostomy tube, enteral feedings should be advanced with caution.

A-T CHILDREN'S® PROJECT

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