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CARING FOR PEOPLE WITH

ATAXIA-TELANGIECTASIA

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ATAXIA – TELANGIECTASIA (A-T)

A-T is a rare, neurodegenerative, recessive genetic disease that causes:

- Progressive loss of muscle control
- Immune system problems
- A high rate of cancer

WHAT CAUSES A-T?

A-T occurs when a person carries defects in both copies of the ATM gene. The ATM gene manages the cell's response to multiple forms of stress including double-strand breaks in DNA. When functioning properly, the protein produced by the ATM gene recognizes that there is a break in DNA, recruits other proteins to repair the break, and stops the cell from making new DNA until the repair is complete. Without this protein, particular areas of the brain (in the cerebellum) that control coordinated movement cease to work normally. The lack of ATM also interferes with the development of lymphocytes, a type of white blood cell that helps to fight infections and produce antibodies.

SYMPTOMS

The severity of A-T symptoms varies among individuals and at different ages. The following are common symptoms of A-T:

- Poor coordination (ataxia) that is apparent in young children and typically starts to worsen during primary school years (between 5-12 years of age)
- Difficulty in coordinating eye movements with head movements (oculomotor apraxia)
- Involuntary movements
- Small dilated blood vessels (telangiectases) over the white (sclera) of the eyes, making them appear bloodshot. Telangiectasia also occurs on sun-exposed areas of the skin. Telangiectasia is not apparent in infancy but may first appear by age 5-8 years.
- Infections, especially of the sinuses and lungs
- Cancer (primarily, but not exclusively, lymphomas and leukemias)
- Delayed onset or incomplete pubertal development, and very early menopause
- Slowed rate of growth (weight and/or height)
- Drooling, particularly in young children when they are tired or concentrating on activities
- Slurred, slow, or distorted speech sounds (dysarthria)
- Diabetes
- Premature age-related changes in hair and skin

NEUROLOGIC PROBLEMS

The first indications of ataxia in A-T usually occur during the toddler years. Children start walking at the usual age (about 12 months of age), but may not improve much from their initial wobbly gait. Sometimes they have problems standing or sitting still and tend to sway backward or from side to side. During the primary school years, walking becomes more difficult, and children will use doorways and walls for support. Children with A-T often appear to have better balance when they run or walk quickly, in comparison to when they walk slowly or stand in one place. At approximately 10 years old, children with typical forms of A-T start using a wheelchair, at first for long distances but eventually in place of all walking.

During school years, children have increasing difficulty with reading because of impaired coordination of eye movement. At the same time, problems with fine motor functions (writing, coloring, using utensils to eat) and slurring of speech (dysarthria) develop. Most of these neurologic problems stop progressing after the age of about 12-16 years.

Involuntary movements may start at any age and may worsen over time. These extra movements can take many forms, including:

- Chorea (small jerks of the hands and feet that look like fidgeting)
- Athetosis (slower twisting movements of the upper body)
- Dystonia (adoption of stiff and twisted postures)
- Myoclonic jerks (occasional uncontrolled jerks)
- Tremors (various rhythmic movements with attempts at coordinated action)

IMMUNODEFICIENCY AND PREDISPOSITION TO INFECTION

About two-thirds of people with A-T have immune system abnormalities. The most common abnormalities are:

- Low levels of one or more classes of immunoglobulins (IgG, IgA, IgM, or IgG subclasses)
- Impaired antibody responses to vaccines or infections
- Low numbers of lymphocytes (especially T-lymphocytes) in the blood
- Frequent infections of the upper (colds, sinus and ear infections) and lower (bronchitis and pneumonia) respiratory tract

MANAGING NEUROLOGIC PROBLEMS

There is no treatment known to slow or stop the progression of the neurologic problems. The treatment of A-T is symptomatic and supportive.

Physical, occupational and speech therapies and exercise may help maintain function but will not slow the course of neurodegeneration. Therapeutic exercises should not be used to the point of fatigue and should not interfere with activities of daily life.

Certain anti-Parkinson and anti-epileptic drugs may be useful to manage symptoms, but should be prescribed only in consultation with a neurologist. These drugs have varying degrees of success in people with A-T.

MANAGING IMMUNOLOGIC PROBLEMS

All individuals with A-T should have at least one comprehensive immunologic evaluation that measures:

- Number and type of lymphocytes in the blood (T-lymphocytes and B-lymphocytes)
- Levels of serum immunoglobulins (IgG, IgA, and IgM)
- Antibody responses to T-dependent (e.g., tetanus, Haemophilus influenzae b) and T-independent (23-valent pneumococcal polysaccharide) vaccines

If the tests show significant abnormalities of the immune system, an allergist/immunologist or infectious disease specialist will be able to discuss various treatment options, which might include the use of prophylactic antibiotics and minimized exposure to infection.

Sometimes vaccines can overcome immunity problems. Vaccines against common bacterial respiratory pathogens such as Hemophilus influenzae, pneumococci, and influenza virus (the "flu") are commercially available and often help to boost antibody responses, even in individuals with low immunoglobulin levels. If vaccines do not work and the person with A-T continues to have problems with infections, gamma globulin therapy (IV or subcutaneous infusions of antibodies collected from typical individuals) may help. (See **Vaccine Schedules**.)

Typically, the pattern of immunodeficiency seen in a person with A-T by age five will be the same pattern seen throughout that person's life. However, 10-20% of people with A-T will have immunologic function deteriorate as they get older.

If the person with A-T begins to get more infections, it is important to reassess immune function. If immune function deteriorates, additional therapy may be needed. Additionally, if infections are in the lungs, testing the person's swallowing is important. A poor swallow reflex may cause aspiration into the lungs leading to infections. (See **Feeding, Swallowing, and Nutrition**.)

A small number of people with A-T have levels of one or more types of immunoglobulin elevated far beyond the normal range. In a few cases, the immunoglobulin levels can be so high that the blood becomes thick and does not flow properly. Therapy for this problem must be tailored to the specific abnormality found and its severity.

Most people with A-T have low lymphocyte counts in the blood, and often the T-lymphocyte counts are particularly low. Though low lymphocyte counts can cause susceptibility to germs that do not cause symptoms in healthy people (opportunistic infections), this is generally not a problem for people with A-T. The one exception is that chronic or recurrent warts are common.

The number and function of T-lymphocytes should be re-evaluated if a person with A-T is treated with corticosteroid drugs such as prednisone for longer than a few weeks or is treated with chemotherapy for cancer. If lymphocyte counts are low in people taking those types of drugs, prophylactic antibiotics may be recommended to prevent specific types of opportunistic infections.

In people with A-T who have low levels of IgA, further testing should be performed to determine if the IgA level is low or completely absent. If it is absent, there is a slightly increased risk of a transfusion reaction. “Medical Alert” bracelets are not necessary, but the family and primary physician should be aware that if there is elective surgery requiring red cell transfusion, the cells should be washed to decrease the risk of an allergic reaction.

People with A-T also have an increased risk of developing autoimmune or chronic inflammatory diseases. This risk is probably a secondary effect of their immunodeficiency and not a direct effect of the lack of ATM protein. The most common examples of such disorders in A-T include immune thrombocytopenia (ITP), arthritis, and vitiligo.

LUNG DISEASE

Respiratory symptoms and lung complications are common. Recurrent or chronic respiratory symptoms during the first few years of life can adversely affect lung function later life. Children and adults with A-T are at increased risk for decline in lung function during respiratory illnesses, after anesthesia for a surgical procedure, and during treatment for malignancies.

Chronic lung disease develops in more than 25 percent of people with A-T. Three major types of lung disease can develop:

- Recurrent/chronic sinopulmonary infections and bronchiectasis, a condition in which bronchial tubes are permanently damaged, resulting in recurrent lower airway infections. These problems may be caused or made worse by immunodeficiency and aspiration.

- Inability to clear mucus, because of ineffective cough and swallowing dysfunction. Difficulty taking deep breaths and an ineffective cough can make it difficult to clear oral and bronchial secretions. This can lead to prolonged respiratory symptoms following common viral respiratory illnesses.

- Restrictive interstitial lung disease or pulmonary fibrosis causes a small number of individuals to have decreased pulmonary reserve, trouble breathing, a need for supplemental oxygen and chronic cough in the absence of lung infections. Recurrent injury to the lungs caused by chronic infections or aspiration may cause lung fibrosis and scarring. The damage may be worsened because of inadequate tissue repair in ATM-deficient cells.

Some people will develop swallowing problems as they age, which increases their risk of coughing, wheezing, or aspiration and other types of lung injury. (See **Feeding, Swallowing and Nutrition**.)

Lung disease may impair sleep efficiency in people with A-T. This can cause fatigue with detrimental effects on many aspects of health. Sleep studies with a polysomnography test can help detect breathing problems such as sleep apnea, often treated with oxygen during the night or pressurized flow of air (CPAP).

VACCINE SCHEDULES

- If antibody function is normal, all routine childhood immunizations including live viral vaccines (measles, mumps, rubella, and varicella) should be given. We recommend special attention to vaccines that can prevent influenza (the “flu”) and some forms of pneumonia.

- People with A-T and all household members should receive the killed (injected) influenza (flu) vaccine every fall.

- People older than two years who have not previously been immunized with Prevnar should receive two (2) doses of Prevnar. At least six months after the last Prevnar has been given and after the child is at least two years old, a 23-valent pneumococcal vaccine (Pneumovax) should be administered. Immunization with the 23-valent pneumococcal vaccine should be repeated approximately every five years for children and adults.

MANAGING LUNG HEALTH

All people with A-T should be seen annually by a pulmonary specialist starting at 2 years of age.

Lung function should be measured by spirometry, and maximum inspiratory and expiratory pressure measurements.

If tolerated, frequent activity and exercise should be encouraged to help improve respiratory health and maintain pulmonary function. Activity should also be encouraged in those who are wheelchair-bound to enhance deep breathing and maintain respiratory muscle strength. No activity or exercise should be performed to the point of exhaustion.

Consider airway clearance techniques (such as chest physiotherapy or cough-assist devices) in persons with acute or chronic chest congestion, or wet cough.

Treat upper (sinusitis) and lower (bronchitis, pneumonia) respiratory tract infections aggressively to limit development of chronic lung disease.

If respiratory symptoms (such as a persistent cough, chest congestion, and/or nasal congestion) persist for more than 7 days after an acute illness, evaluation by a doctor is recommended.

Identify and treat immunodeficiency.

Consider use of chronic antibiotic treatment for people with bronchiectasis.

Maintain nutrition and minimize aspiration. Feeding through a gastrostromy tube may be recommended. (See **Feeding, Swallowing, and Nutrition**.)

History of shortness of breath, with or without exertion may indicate interstitial lung disease and should be evaluated. Treatment with oral steroids and other drugs to reduce inflammation may be indicated.

Get a yearly influenza vaccination (if not contraindicated) for both the individual with A-T and all members of the household.

Get a pneumococcal vaccine every 5 years.

Avoid contact with people who smoke, and limit exposure to air pollutants and respiratory irritants.

FEEDING, SWALLOWING, AND NUTRITION

Feeding and swallowing may become difficult for people with A-T as they get older. Primary goals for feeding and swallowing are to have safe, adequate, and enjoyable mealtimes.

Involuntary movements may make feeding difficult or messy, and may excessively prolong mealtimes. It may be easier to feed with fingers than to use utensils. Drinking from a closed container with a straw may be easier than drinking from an open cup.

Caregivers may need to prepare foods into bite-sized pieces to facilitate self-feeding. Caregivers may need to assist with feeding, when self-feeding is difficult or unduly increases the length of meals. In general, meals should be completed within approximately 30 minutes. Longer meals may be stressful, interfere with other daily activities, and limit the intake of necessary liquids and nutrients.

DYSPHAGIA AND ASPIRATION

- Swallowing problems (dysphagia) are common and typically become apparent after the age of 10 years. Dysphagia is common because neurological changes can interfere with the coordination of mouth and throat (pharynx) movements that is needed for safe and efficient swallowing.
- Coordination problems involving the mouth may make chewing difficult and make meals take longer.
- Coordination problems involving the pharynx may cause liquid, food, and saliva to be inhaled into the airway (aspiration).
- People with dysphagia may not cough when they aspirate (silent aspiration). In turn, silent aspiration may cause lung problems due to inability to cough and clear aspirated food and liquids from the airway.

WARNING SIGNS OF A SWALLOWING PROBLEM

- Choking or coughing when eating or drinking
- Poor weight gain (during ages of expected growth) or weight loss at any age
- Excessive drooling
- Mealtimes longer than 40 minutes on a regular basis
- Foods or drinks previously enjoyed are now refused or difficult
- Chewing problems
- Increased number of otherwise unexplained lung infections, particularly in people with other signs of swallowing problems

MANAGING FEEDING, SWALLOWING, AND NUTRITION PROBLEMS

Oral intake may be aided by teaching persons with A-T how to drink, chew and swallow more safely. Treatments for swallowing problems should be determined following evaluation by a speech-language pathologist. Dieticians may help treat nutrition problems by recommending dietary modifications, including high calorie foods or food supplements.

A feeding (gastrostomy) tube is recommended when any of the following occur:

- A child cannot eat enough to grow or a person of any age cannot eat enough to maintain weight
- Aspiration is problematic
- Mealtimes are stressful or too long, or interfere with other activities

Feeding tubes can decrease the risk of aspiration by enabling persons to avoid liquids or foods that are difficult to swallow and providing adequate calories without the stress and time commitment of prolonged meals. Gastrostomy tubes do not prevent people from eating by mouth. Once a tube is in place, the general goal should be to maintain weight at the 10-25th percentile. Re-feeding immediately after gastrostomy tube placement should be done gradually to optimize acceptance and to minimize the risk of aspiration from gastroesophageal reflux.

CANCER

People with A-T have an increased incidence (approximately 25% lifetime risk) of cancers, particularly lymphomas and leukemia, but other cancers can occur. When possible, treatment should avoid the use of radiation therapy, and chemotherapy drugs that work in a way that is similar to radiation therapy (radiomimetic drugs) as these are particularly toxic for people with A-T. The special problems of managing cancer are sufficiently complicated that treatment should be managed only in academic oncology centers after consultation with physicians who have specific expertise in A-T. (See **A-T Cancer Consultations**.)

There is no way to predict which individuals will develop cancer. Routine screening blood tests for leukemia and lymphoma are not helpful. It is worthwhile, however, to consider cancer as a diagnostic possibility whenever possible symptoms of cancer arise.

COMMON WARNING SIGNS OF LEUKEMIA AND LYMPHOMA

Recurrent or persistent fever without explanation	Bruising
Pale appearance	Body aches and bone pain
Swollen lymph nodes in neck, armpits, groin, and abdomen	

A-T CARRIERS AND CANCER RISK

Women who are A-T carriers (who have one mutated copy of the ATM gene) have a 2.5 fold increased risk for the development of breast cancer compared to the general population. This includes all mothers of A-T children and some female relatives.

If an A-T carrier, male or female, develops cancer, they can be treated with standard doses of radiation and chemotherapy.

EYE AND VISION

Most people with A-T develop prominent blood vessels (telangiectasia) in the membrane that covers the white part (sclera) of the eye. These often do not occur until 5-8 years of age and do not affect vision.

Vision (ability to see objects in focus) is normal, but visual functioning is often impaired by difficulty controlling the movement of the eyes. This most affects visual functions that require fast, accurate eye movements from point to point (e.g. reading).

Eye misalignments (strabismus) are common, but may be treatable with surgery. There may be difficulty in seeing objects up close, and some people may benefit from the use of inexpensive reading glasses.

TELANGIECTASIA

Telangiectasia over the white (sclera) of the eye usually occurs by the age of 5-8 years, but can occur later or not at all. Although potentially a cosmetic problem, these ocular telangiectases do not bleed or itch though they are sometimes misdiagnosed as chronic conjunctivitis. It is their constant nature, not changing with time, weather or emotion that marks them as different from other visible blood vessels.

Telangiectases can also appear on the sun-exposed areas of skin, especially the face and ears. Telangiectases on the eyes and skin pose no medical problems.

Rarely, telangiectasia occurs in other tissues and may cause complications. For example, it can occur in the bladder as a late complication of chemotherapy with cyclophosphamide, has been seen deep inside the brain of older people with A-T, and occasionally arises in the liver and lungs.

SKIN

A-T can cause features of early aging such as premature graying of the hair. It can also cause vitiligo, an auto-immune disease causing loss of skin pigment resulting in a blotchy “bleach-splashed” look. Warts and molluscum contagiosum, which are viral infections of the skin, can be extensive and resistant to treatment.

A small number of people develop chronic inflammatory skin lesions (granulomas). A physician with experience in A-T may need to be consulted for help with treatment.

ORTHOPEDICS

Many individuals with A-T develop deformities of the feet that compound the difficulty they have with walking or standing transfers. If there is a tendency for "twisting of the ankles" (instability at the ankle) with standing or walking, surgery or bracing may be helpful. In a few cases, severe scoliosis occurs. Spinal fusion is only rarely indicated, and corrective surgery is complex and is associated with substantial risk for complications. (See **Surgery and Procedures….**)

PUBERTY

Development of normal puberty is usually delayed or impaired for boys and girls. Some females with A-T have irregular menstrual periods, others stop having periods at an early age, and a few never complete pubertal development to the point of starting menstrual periods. A gynecologist should be consulted for help with these problems.

SURGERY AND PROCEDURES REQUIRING SEDATION OR ANESTHESIA

Surgery should be performed only at centers with an Intensive Care Unit (ICU). Even a dental procedure requiring sedation could be considered high risk for some people with A-T depending on age and lung function. A pre-operative evaluation of lung function should be performed in all people with A-T regardless of age and whether or not they have a history of breathing problems. This evaluation will help the anesthesiologist understand the risks and better anticipate the complications of sedation or general anesthesia.

People with A-T may have difficulty coming off the ventilator after general anesthesia. Possible alternatives to general anesthesia and strategies that maximize airway clearance following anesthesia should be considered.

EXPOSURE TO RADIATION

People with A-T have an increased sensitivity to ionizing radiation (x-rays and gamma rays). Therefore, x-rays should be done only when they are medically necessary, because exposing a person with A-T to ionizing radiation can damage cells in such a way that the body cannot repair them. The body can cope normally with other forms of radiation, such as ultraviolet light, so there is no need for special precautions from exposure to sunlight.

GUIDELINES FOR USE OF DIAGNOSTIC X-RAYS IN A-T

- X-rays should be performed only when the result will affect medical management.
- If a person with A-T has fever, cough, and abnormal 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, a person with A-T 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 exams.

EDUCATION AND SOCIALIZATION

Children with A-T often enjoy school if proper accommodations can be made. Difficulties in school can be caused by a delay in response time to visual, verbal or other cues; slurred and quiet speech; and impaired fine motor control. Multi-tasking is frequently difficult, and busy classrooms can present special challenges. If a child needs to spend time and energy to maintain balance in a chair, it taxes mental resources. Fatigue can be a major factor in daily functioning. Decisions about proper educational placement (extra help in regular classes, special education placement, or the need for a dedicated assistant) are influenced by the age and needs of the child as well as the local resources available. These decisions should be revisited as often as circumstances warrant.

Despite their many neurologic impairments and sometimes their appearance, most individuals with A-T are very socially aware and socially skilled, and thus benefit from sustained peer relationships. Schools and classrooms where A-T students can maintain friendships throughout elementary, middle, and high school are ideal. Some individuals function quite well despite their disabilities, and a few have graduated from college.

It is important to recognize that intellectual disabilities are not regularly a part of the clinical picture of A-T although school performance may be suboptimal because of the many difficulties in reading, writing, and speech. Many of the problems caused by A-T can be compensated for with special attention, as problems are often related to “input and output” issues. Problems with eye movement control make it difficult for people with A-T to read, and yet most fully understand the meaning and nuances of text that is read to them. Delays in speech initiation and lack of facial expression make it seem that they do not know the answers to questions. Reducing the skilled effort needed to answer questions, and increasing the time available to respond, is often rewarded by real accomplishment.

Children with A-T are often very conscious of their appearance, and strive to appear normal to their peers and teachers. Life within the ataxic body can be tiring. The enhanced effort needed to maintain appearances and increased energy expended in abnormal tone and extra movements all contribute to physical and mental fatigue. As a result, for some a shortened school day yields real benefits.

SCHOOL RECOMMENDATIONS

- All children with A-T need special attention to the barriers they experience in school. In the United States, this takes the form of an IEP (Individualized Education Program).
- Children with A-T tend to be excellent problems solvers. Their involvement in how to best perform tasks should be encouraged.
- Speech-language pathologists may facilitate communication skills that enable persons with A-T to get their messages across (using key words versus complete sentences) and teach strategies to decrease frustration associated with the increased time needed to respond to questions (e.g., holding up a hand and educating others about the need to allow more time for responses). Traditional speech therapies that focus on producing specific sounds and strengthening lip and tongue muscles are rarely helpful.
- Many children with A-T have full-time classroom aides, especially to help with scribing, transportation through the school, mealtimes and toileting. The impact of an aide on peer relationships should be monitored carefully.
- Physical therapy is useful to maintain strength and general cardiovascular health. However, no amount of practice will slow the cerebellar degeneration or improve neurologic function. Exercise to the point of exhaustion should be avoided.
- Hearing is normal throughout life. Audio books may be a useful adjunct to traditional school materials.
- Early use of computers (starting in preschool) with word completion software should be encouraged. Regular consultations with specialists in Assistive Technology are recommended.
- Adaptive Physical Education can be helpful. Some students find yoga, with assistance, to be very beneficial both physically and mentally.
- Practicing coordination (e.g. balance beam or cursive writing exercises) is not helpful.
- Occupational therapy is helpful for managing daily living skills.
- Allow rest time, shortened days, reduced class schedule, reduced homework, and modified tests as necessary.
- Like all children, those with A-T need to have goals to experience the satisfaction of making progress.
- Social interactions with peers are important and should be taken into consideration for class placement. For everyone, long-term peer relationships can be the most rewarding part of life; for those with A-T, establishing these connections in school years is critical.

ANNUAL PHYSICAL EXAMS

People with A-T should have a complete physical exam once a year. Specific attention should be paid to the following issues:

- Monitoring height and weight gain during ages of expected growth and weight loss at any age
- Assessing frequency and severity of infections
- Looking for signs and symptoms of lung disease
- Screening for feeding or swallowing problems
- Screening for diabetes
- Screening for lipid abnormalities (cholesterol and triglycerides)
- Screening for mental health problems (depression)

MENTAL HEALTH AND COUNSELING

Having a child with a disability can be difficult for everyone in the family. People with A-T and family members often benefit from counseling. Sibling and marital issues should be monitored in addition to the well-being of the affected individual. It is recommended that families seek out a professional soon after diagnosis to establish contact with a trusted counselor should services be needed at a future time.

Thank you to the clinicians at the A-T Clinical Center at The Johns Hopkins Hospital for their dedication to the evaluation and treatment of people with A-T and for their work on this handbook: Howard Lederman, MD, PhD; Tom Crawford, MD; Sharon McGrath-Morrow, MD; Maureen Lefton-Greif, PhD, CCC/SLP; Pavan Vaswani; Jennifer Wright, RN.

Thank you to families and friends of the A-T Children's Project whose grassroots fundraising efforts established and continue to fund the work of the A-T Clinical Center.

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QUICK INFORMATION CARD FOR URGENT CARE PROVIDERS

Please call or email the A-T Children's Project to get copies of these cards mailed to you. Keep the card handy in your wallet or purse so that your child's care provider can have quick access to important treatment information.

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, dysarthria and impaired chewing and swallowing,
- immunodeficiency with lymphopenia and hypogammaglobulinemia of varying severity,
  - ☐ I have antibody deficiency and receive gammaglobulin therapy
- cancer predisposition (non-Hodgkin's lymphoma and leukemia most common),
- telangiectasia (often present), especially over the sclerae. In rare instances, telangiectasia can cause internal complications of bladder, brain, liver and lung.

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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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

For more information, physicians can log in at **UpToDate.com**

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RESOURCES FOR CAREGIVERS

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ATCP.ORG

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

The A-T Children's Project is a nonprofit organization that raises funds to support and coordinate biomedical research projects, scientific conferences and a clinical center aimed at finding life-improving therapies and a cure for ataxia-telangiectasia.