National Ataxia Foundation

Ataxia-Telangiectasia (A-T)

Ataxia-telangiectasia (A-T) is a rare genetic disorder caused by mutations in the ATM gene. A-T is the second most common childhood recessive ataxia, with Friedreich Ataxia being the most common. The prevalence of A-T is estimated to be between 1 in 40,000 and 1 in 100,000 people worldwide. It is estimated that 1 in 100 people in the United States are carriers of the mutated ATM gene.

Symptoms

Like many other forms of Ataxia, A-T is marked by poor balance and coordination. In fact, the word Ataxia means incoordination. There can also be problems coordinating muscles that control speech, swallowing, and eye movement. Other common A-T symptoms include immune system problems, increased cancer risk, and chronic lung infections. Less common symptoms may include Telangiectasia (red veins in the corners of the eyes or on the surface of the ears or the face), muscle twitches, and neuropathy. Intelligence is not typically impacted by A-T.

Prognosis

A-T symptoms usually begin in early childhood. The first symptom is usually cerebellar ataxia between the ages of 2 and 5 years old. This condition is progressive, meaning that symptoms will continue to worsen over time. People with A-T will eventually need to use a wheelchair. Lifespan is significantly shortened, with children with A-T living until adolescence or early adulthood. The three most common causes of death for people with A-T are cancer, infection, and progressive lung failure. As the treatment and early detection of cancer and infection have improved, so too has the prognosis for children with A-T. Challenges remain regarding the treatment of chronic lung failure.

Some individuals have Mild Variant A-T, also known as Variant A-T or adult-onset A-T. People with Variant A-T begin to develop symptoms in late adolescence or early adulthood. Their symptoms tend to be milder than Classic or childhood-onset A-T. However, this condition is also progressive. Due to the rarity of Variant A-T, there is still much that we don't know about the prognosis for these patients.

Treatments such as physiotherapy, occupational therapy, and speech-language therapy can also significantly improve the lives of people with A-T. Medications to treat specific symptoms may also be helpful.

Genetics

A-T is an inherited genetic disorder. It is caused by an abnormality of a single gene called the ATM gene. The abnormality can be passed from generation to generation by family members who carry it. Males and females are equally likely to inherit the genes that cause A-T.

Inherited diseases like A-T occur when one pair of the body's 20,000 genes does not work properly. Genes are microscopic structures within the cells of our bodies that contain instructions for every feature we inherit from our parents. Two copies of each gene are inherited; one copy from the mother and one from the father. A-T is autosomal recessive, which means that an individual only develops symptoms of the disease if both copies of his/her ATM gene are not working properly.

An individual who has one copy of an altered or nonfunctioning ATM gene does not develop any neurological symptoms and is called a carrier. For people who are carriers, the normal ATM gene mostly compensates for the nonfunctioning copy of the gene. However, a child whose parents are both carriers can inherit a "double dose" of the altered ATM gene and will therefore develop A-T.

Most of the time, carriers have no idea that they have an abnormal ATM gene because they do not have neurological symptoms. It is often only when a child is diagnosed with A-T that the parents learn that they are both carriers. When both parents are carriers, each of their children has a 25 percent chance of having A-T and a 50 percent chance of being a carrier. Carriers of the mutated ATM gene are at increased risk of developing cancer and heart disease.

Gene tests can be performed for diagnostic purposes to determine what kind of Ataxia is within a person or family. Genetic testing can also be done, in some circumstances, even before there are symptoms, to determine whether a person carries the abnormal gene or genes that cause Ataxia. This is called predictive or presymptomatic testing. A gene test can also be used to determine whether a fetus has an abnormal Ataxia gene. This is called prenatal testing. Anyone who is considering a predictive or prenatal test should consult with a genetic counselor to discuss the reasons for the test, the possible outcomes, and how those outcomes might affect the person emotionally, medically, or socially.

Diagnosis

Early diagnosis is critical for A-T symptom management. Depending on when or in what order symptoms begin to show, different clinical specialists will be involved in the diagnosis process. Clinical examinations from a doctor can help assess balance, coordination, vision, and speech impairments. MRI scans are often used to confirm cerebellar atrophy or degeneration. Blood tests can be used to detect elevated levels of molecules that suggest someone has A-T. However, a specific diagnosis of A-T can only be made by a genetic test. Genetic testing can identify the specific mutations present in the ATM gene.