Ataxia Telangiectasia: A Case Report

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Abstract
Ataxia telangiectasia (AT) is a complex multisystem disorder characterized by progressive neurological impairment, variable immunodeficiency and oculo-cutaneous telangiectasia. Ataxia telangiectasia is a member of chromosomal breakage syndromes and it is caused by a mutation in the ataxia-telangiectasia mutated (ATM) gene. We are reporting an eight year old girl of AT presented with difficulty in walking, frequent fall, trembling of the whole body, difficulty in speech. [J Shaheed Suhrawardy Med Coll, 2014;6(1):41-43]

Key Words: Ataxia telangiectasia, neurological impairment, chromosomal breakage syndromes, mutation

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Introduction
Ataxia telangiectasia (AT) is a genetically determined autosomal recessive, multisystem disorder causing neurodegeneration and immunological abnormalities resulting in increased susceptibility to infection and malignancies, endocrine deficiencies, increased sensitivity to ionizing radiation and an anomaly of DNA repair. The ataxia manifests as the child starts walking while ocular telangiectasia is usually appreciated around 4-6 years of age. Vessels over exposed bulbar conjunctiva become prominent and appear fan shaped. Neurological manifestations consist of ataxia, choreoathetosis, ocular movement abnormalities, mental retardation and dystonia. Syllaba and Henner1 first described this condition in 1929. Individuals of all races and ethnicities are affected equally. The incidence world-wide is estimated to be between 1 in 40,000 and 1 in 100,000 people2,3. Both males and females are equally affected. AT results from mutations of a single gene, ataxia-telangiectasia mutated (ATM), located on chromosome 11q22-23 encoding a large basic protein involved in cell cycle control and DNA damaging repair4,5.

Case Report
An eight years old girl came with the complaints of progressive difficulty in walking with frequent fall, trembling of the whole body, difficulty in speech for last six years. The patient was the second issue of consanguineous parents. The child was developmentally alright up to two years of age. Subsequently the parents noticed that the child had unsteadiness during walking with frequent fall which was progressively deteriorating day by day. Later the patient can't walk without support. The
Ataxia telangiectasia (AT) (also referred to as Louis-Bar syndrome) is a rare, neurodegenerative, inherited disease causing severe disability. Ataxia refers to poor coordination and telangiectasia to small dilated blood vessels, both of which are hallmarks of the disease.

Symptoms most often first appear in early childhood (the toddler stage) when children begin to walk. Though they usually start walking at a normal age, they wobble or sway when walking, standing still or sitting, and may appear almost as if they are drunk. In late pre-school and early school age they develop difficulty moving the eyes in a natural manner from one place to the next (oculomotor apraxia). They develop slurred or distorted speech, and swallowing problems. Some have an increased number of respiratory tract infections (ear infections, sinusitis, bronchitis, and pneumonia). Because not all children develop in the same manner or at the same rate, it may be some years before AT is properly diagnosed. Most children with AT have stable neurologic symptoms for the first 4-5 years of life, but begin to show increasing problems in early school years.

AT is caused by a defect in the ATM gene. In simple terms, the protein produced by the ATM gene recognizes that there is a break in DNA, recruits other proteins to fix the break, and stops the cell from making new DNA until the repair is complete. Prominent blood vessels (telangiectasia) over the white (sclera) of the eyes usually occur by the age of 5-8 years, but sometimes later or not at all. The absence of telangiectasia does not exclude the diagnosis of A-T.

About two-thirds of people with A-T have abnormalities of the immune system. The most common abnormalities are low levels of one or more classes of immunoglobulins.
(IgG, IgA, IgM or IgG subclasses), having low numbers of lymphocytes (especially T-lymphocytes) in the blood. Some people have frequent infections of the upper (colds, sinus and ear infections) and lower (bronchitis and pneumonia) respiratory tract.

People with A-T have a highly 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. Women who are A-T carriers (who have one mutated copy of the ATM gene), development of breast cancer compared to the general population.

A small number of people develop a chronic inflammatory skin disease (granulomas). Chronic lung disease develops in more than 25% of people with A-T. Feeding and swallowing can become difficult for people with A-T as they get older.

A variety of laboratory abnormalities occur in most people with A-T including: elevated and slowly increasing alpha-immunodeficiency with low levels of immunoglobulins (especially IgA, IgG subclasses, and IgE) and low number of lymphocytes in the blood; chromosomal instability (broken pieces of chromosomes); increased sensitivity of cells to x-ray exposure (cells die or develop even more breaks and other damage to chromosomes); cerebellar atrophy on MRI scan.

The diagnosis can be confirmed in the laboratory by finding an absence or deficiency of the ATM protein in cultured blood cells, an absence or deficiency of ATM function (kinase assay), or mutations in both copies of the cell's ATM gene. These more specialized tests are not always needed, but are particularly helpful if a child's symptoms are atypical.

The life expectancy of people with A-T is highly variable. The average is approximately 25 years, but continues to a wheelchair in their teens. Some patients have been able to attend college and live independently, and some have lived into the fifth decade of life. The two most common causes of death are chronic lung disease (about 1/3 of cases) and cancer (about 1/3 of cases).

**Conclusion**

Ataxia Telangiectasia (A-T) should be considered in children with ataxia, telangiectasia and evidence of combined humoral and cellular immunodeficiency. A-T follows progressive course. It must be stressed that the course of the disease can be quite variable and it is difficult to predict the course in any given individual. Even within families, where the specific genetic defect is the same, there can be great variability in the type and severity of different neurologic problems and immunodeficiency.

**References**