ATAXIA TELANGIECTASIA: A CASE REPORT

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ABSTRACT
Ataxia Telangiectasia (AT) is a rare, neurodegenerative disease that affects many parts of the body and causes severe disability is characterized by progressive cerebellar ataxia, oculocutaneous telangiectasia, and recurrent respiratory and sinus infections. AT is caused by a defect in the ATM gene, which is responsible for recognizing and correcting errors induplicating DNA when cells divide, and in destroying the cells when the errors can not be corrected. An 11 years old girl who is the first issue of non consanguineous parents completely immunized according to EPI schedule was admitted to lahore general hospital, Lahore, on 02.06.2014 with the complaints of generalized weakness and difficulty to walk from the 8th year. The patient has one younger brother, 8 years old who also has such complaints. On examination the patient was found mildly anemic with congestions in both eyeballs that is radiating from the both corners to limbus. The patient was conscious and cooperative. Cranial nerves were intact. There were hypotonia which was more marked in lower limbs with diminished jerks and flexor plantar reflexes. Gait was wide based and ataxic. She was diagnosed as a case of AT. This case is presented as academic interest.

Key words: Ataxia Telangiectasia (AT), neurodegenerative disease.

INTRODUCTION
The first case described in the literature was a 9-year-old child with progressive cerebellar ataxia and bilateral oculocutaneous telangiectasia reported in 1941 by Madame Louis-Bar. Initially known as the Louis-Barsyndrome, the term ataxia-telangiectasia was introduced in 1958 by Boder et al, who recorded the clinical features and recognized the familial incidence proposing an autosomal recessive mode of inheritance for the disease¹. The disease is sometimes referred to as Boder-Sedgwick syndrome. The ataxia-telangiectasia mutated (ATM) kinase initiates a well-characterized response to DNA damage, resulting in arrest of cell-cycle, DNA repair, or apoptosis2–5. Mutations in the ATM gene, though tolerated, result in the fatal childhood disorder ataxia-telangiectasia (AT), characterized by symptoms including predisposition to cancer, ataxia due toprogressive cerebellar degeneration, immunodeficiency, and telangiectasias (spider veins)²–⁵. ATM signaling is required to sense and initiate repair of DNA double-strand breaks. Therefore, nuclear genomic instability resulting from loss of this function is regarded as a major mechanism underlying the pathology of A-T³–⁶. However, this disease presents with a wide-array of symptoms, not all of which are readily explained by nuclear genomic instability, and study of cell and animal models of A-T has led to much speculation about additional thogenic mechanisms⁷. One such mechanism for which there is substantial experimental evidence is oxidative stress⁸–¹². How ATM is involved in oxidative stress management remains unclear as do other potential roles in Cellular homeostasis in the absence of DNA damage.

CASE REPORT
An 11 years old Najma who is the first issue of non consanguineous parents completely immunized according to EPI schedule was admitted to LAHORE GENERAL HOSPITAL, on 02.06.2014 with the complaints of generalized weakness and difficulty to walk from the 8th year. The patient initially could try to walk but later on she fell on the ground as she developed imbalance in walking that gradually manifested also in standing capacity within one month. The patient has no other significant past illness except frequent respiratory illness since birth. Her birth history is uneventful but she achieved her developmental milestones delayed. She attained her sitting at the age of 2 years and walking and standing at the age of 3 and 4 years respectively. The patient has one younger brother of 8 years old who also has such complaints. On examination, the patient was found mildly anemic with prominent blood vessels over bulbar conjunctiva which were radiating away from the limbus (figure 4). The patient was conscious and cooperative. Cranial nerves were intact.
There was hypotonia which was more marked in lower limbs with diminished jerks and flexor plantar reflexes. Gait was wide based and ataxic. Sensory function was intact. Cerebellar function was abnormal showed nystagmas, slurred speech, intention tremor, past pointing and dysdiadochokinesia. On investigation, complete blood count, lipid profile, LFT, chest x-rays, serum electrolytes, ultrasonogram of whole abdomen and ECG showed normal results. MRI showed cerebellar atrophy (figure 2). Serum alpha-fetoprotein was 950 ng/L. Her Ig level is IgA 40 (normal 60-300), IgG 1200, IgM 360. In the hospital she was offered physiotherapy with consultation of physical rehabilitation department: counselling of the family regarding course of the disease and inherited nature of the illness, explained to the parents. Only the conservative treatment was advised to avoid respiratory tract infection. Physical and speech therapy was advised.

**DISCUSSION**

In general, Ataxia-telangiectasia (AT) is a multisystem disorder characterized by progressive neurologic impairment. Cerebellar ataxia, variable immunodeficiency, and ocular and cutaneous telangiectasia. Patients with AT have an increased susceptibility to sinopulmonary infection, x-ray hypersensitivity, and predisposition to malignancy. These patients with AT who had been diagnosed and treated at Children’s Medical Center during a 20-year period (1985-2005) were analyzed. The study population comprised 104 patients (54 male). The median age of patients at the time of the study was approximately nine years. Clinical characteristics included progressive ataxia (100% of patients) that was present in our case, telangiectasia of skin or conjunctivae (83.8%) and of ears (70.2%), eye movement disorder (apraxia of horizontal and vertical saccadic eye movements (80.6%), choreoathetosis (87.1%), speech dysarthria in almost all and that is also presentation. Our patient, acute and recurrent sino pulmonary infections affected 75% of patients where our patient has been suffering from recurrent respiratory tract infections since birth. Our patient has cerebellar atrophy on MRI that was appearing like trifoliate it was suggesting to the point of possibility of ataxia telangiectasia. The average serum alpha-fetoprotein level in study patients was elevated at 149 ng/L and in our case it is more than 1350 ng/L. Elevated serum α-fetoprotein level was described as a useful screening test for AT.

Ataxia Telangiectasia (AT) 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. Many patients are confined 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.

REFERENCES: