# 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 cerebellartaxia, oculocutaneous telangiectasia, and recurrent respiratory and sinus infections. AT is caused bya 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 notbecorrected. 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 the8th year. The patient has one younger brother,8 years old who also has such complaints. Onexamination the patient was found mildly anemic with congestions in both eyeballs that isradiating from the both corners to limbus. The patient was conscious and cooperative. Cranialnerves 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 wasintroduced in 1958 by Boder et al, who recordedthe clinical features and recognized thefamilial incidence proposing an autosomalrecessive mode of inheritance for the disease[1].The disease is sometimes referred to as Boder-Sedgwick syndrome. The ataxia-telangiectasia mutated (ATM) kinase initiates a wellcharacterizedresponse to DNA damage,resulting in arrest of cell-cycle, DNA repair, orapoptosis2–5. Mutations in the ATM gene,though tolerated, result in the fatal childhooddisorder ataxia-telangiectasia (AT),characterized by symptoms includingpredisposition to cancer, ataxia due toprogressive cerebellar degeneration,immunodeficiency, and telangiectasias (spiderveins)[2–5]. ATM signaling is required to senseand initiate repair of DNA double-strandbreaks. Therefore, nuclear genomic instabilityresulting from loss of this function is regardedas a major mechanism underlying thepathology of A-T [3–6]. However, this diseasepresents with a wide-array of symptoms, notall of which are readily explained by nucleargenomic instability, and study of cell andanimal models of A-T has led to muchspeculation about additional thogenicmechanisms[7] . One such mechanism for whichthere is substantial experimental evidence isoxidative tress[8–12] . How ATM is involved inoxidative stress management remains unclearas do other potential roles in Cellularhomeostasis in the absence of DNA damage.

## CASE REPORT
An 11 years old Najma who is the first issue of nonconsanguineous parents completelyimmunized according to EPI schedule wasadmitted to LAHORE GENERAL HOSPITAL, on 02.06.2014 with the complaints ofgeneralized weakness and difficulty to walkfrom the 8th year. The patient initially couldtry to walk but later on she fell on the groundas she developed imbalance in walking that gradually menifested also in standing capacitywithin one month The patient has no other significantpast illness except frequent respiratory illnesssince birth.Her birth history is uneventful but sheachieved her developmental milestonesdelayed. She attained her sitting at the age of2 years and walking and standing at the age of3 and 4 years respectively. The patient has oneyounger brother of 8 years old who also has suchcomplaints. On examination, the patient wasfound mildly anemic he had prominent blood vessels over bulbar conjuctiva which were radiating away from the limbus(figure.4). The patient was conscious andcooperative. Cranial nerves were intact.

There washypotonia which was more marked inlower limbs with diminished jerks and flexorplantar reflexes. Gait was wide based andataxic.Sensory function was intact. Cerebellarfunction was abnormal showed asnystagmas,slurred speech, intention tremor, past pointingand dysdiadochokinesia.Oninvestigation,complete blood count, lipid profile, LFT, chest xrays,serum electrolytes, ultrasonogram of wholeabdomen and ECG showed normal results. MRIshowed cerebellar atrophy(figure.2). Serum alpha-fetoprotein was 950 ng/L.HerIg level is IgA40(normal 60-300),IgG(1200),IgM(360).in the hospital she was offered physiotherapy with consultation of physical rehabilitation department:couselling of the family regarding course of the disease and inherited nature of the illness,explained to the parents. Onlythe conservative treatment was advised to avoidrespiratory tract infection. Physical and speechtherapy was advised.

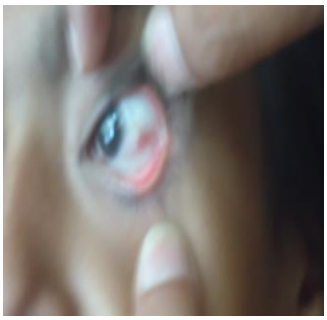

**Fig. 1:** Telangiectasias in eye

**Fig. 2:** MRI Brain showing Ceebellar Atrophy

**Fig. 3:** Dull Expressionless Face of patient.

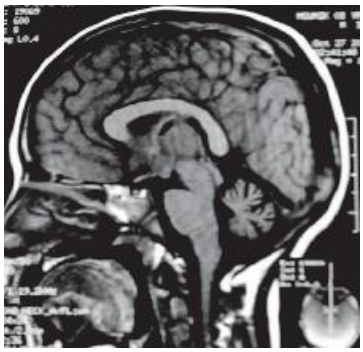

**Fig. 4:** 11 year old Najma

## DISCUSSION

In general, Ataxia-telangiectasia (AT) is amultisystem disorder characterized byprogressive neurologic impairment, Cerebellarataxia, variable immunodeficiency, and ocularand cutaneous telangiectasia. Patients with AThave an increased susceptibility to sinopulmonary infection, x-ray hypersensitivity,and predisposition to malignancy[13].These patients with AT who had beendiagnosed and treated at Children's MedicalCenter during a 20-year period (1985-2005)were analyzed. The study population comprised104 patients (54 male). The median age ofpatients at the time of the study wasapproximately nine years. Clinicalcharacteristics 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%),speechdysarthria in almost all and that is also presentation. Our patient .Acute and recurrent sino pulmonary infectionsaffected 75% of patients where our patient hasbeen suffering from recurrent respiratory tractinfections since birth. Our patient hasCerebellar atrophy on MRI that was appearinglike trifoliate it was suggesting to the point ofpossibility of ataxia Telangiectasia[15].Averageserum alpha-fetoprotein level in study patients waselevated at 149 ng/L and in our case it is morethan 1350 ng/L. Elevated serum □-fetoproteinlevel was described as a useful screening test for AT.

Ataxia Telangiectasia (AT) follows progressivecourse. It must be stressed that the course ofthe disease can be quite variable and it isdifficult to predict the course in any givenindividual. Even within families, where thespecific genetic defect is the same, there canbe great variability in the type and severity ofdifferent neurologic problems andimmunodeficiency. Many patients are confinedto a wheelchair in their teens. Some patientshave been able to attend college

and liveindependently, and some have lived into thefifth decade of life.

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