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# A rare case of Ataxia Telangiectasia with cerebellar atrophy: A case report

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## ABSTRACT

Ataxia Telangiectasia (A-T) is an uncommon genetic disorder that affects multiple systems in the body, encompassing the nervous and immune systems. The diagnosis of A-T does not have a definitive diagnostic test and it primarily relies on a clinical examination, elimination of other similar conditions and supportive laboratory tests. The majority of individuals diagnosed with A-T exhibit identified mutations that arise in ATM gene, which plays a crucial role in the repair of damaged DNA. The high level of alpha-fetoprotein on blood test is also consistent with A-T. We report a 10-year-old male child who presented with difficulty in maintaining postural control, unsteady gait, slight bulbar telangiectasia and neurological signs such as dysidiadochokinesia, past pointing and intentional tremors. A-T has no conclusive cure, only supportive treatment such as vitamin E supplementation and balance exercises can be helpful in managing symptoms and maintaining quality of life.

**Keywords:** Cerebellar atrophy, Immunodeficiency, alpha-fetoprotein, Ataxia - Telangiectasia

## 1. INTRODUCTION

Ataxia Telangiectasia is a rare disease of autosomal recessive inheritance pattern, involving multiple systems resulting in immunological abnormalities and increased susceptibility to infections and malignancies. Due to the mutations that arise in ataxia telangiectasia mutated (ATM) gene, located on chromosome 11q22-23, which is involved in regulating the cell cycle and repairing DNA, causes this condition (Sonia et al., 2017). Ataxia onset is a typical hallmark of the disease and generally appears when the child starts walking. Ocular Telangiectasia is another characteristic feature, usually present between the ages of 4 to 6 years. The slight prominence of blood vessels over the exposed bulbar conjunctiva gives a fan shaped appearance.

In addition to Ataxia and ocular telangiectasia, people also experience range of neurological manifestations including choreoathetosis, abnormal eye movements, mental retardation and dystonia. These symptoms tend to worsen over time and there is no cure for Ataxia Telangiectasia. The disease is associated with an increased susceptibility to infection, malignancies and

endocrine deficiencies. Therapeutic interventions targeting the control of reactive oxygen species levels may offer potential benefits for A-T patients (Reichenbach et al., 2002).

Counselling and education play an important role in management of A-T, as there is no cure for this disease. It is important for the child's caregivers to work closely with health care providers to develop a comprehensive treatment plan that addresses the child's individual needs and helps to maintain their quality of life (Kamat, 2023).

## 2. CASE DESCRIPTION

A 10-year-old male child, first birth order of twins born out of non-consanguineous marriage presented with complaints of unsteadiness while sitting without support since 10 months of age, unsteady gait since 15 months of age, abnormality of speech associated with breakage of words and unclear pronunciation but could be able to communicate meaning since 3 years of age, redness in eyes with prominent blood vessels aggravated on anger since 6 years of age (Figure 1), progressive increase in swaying while walking and multiple episodes of fall while playing since 7 years of age.



**Figure 1** Bulbar telangiectasia

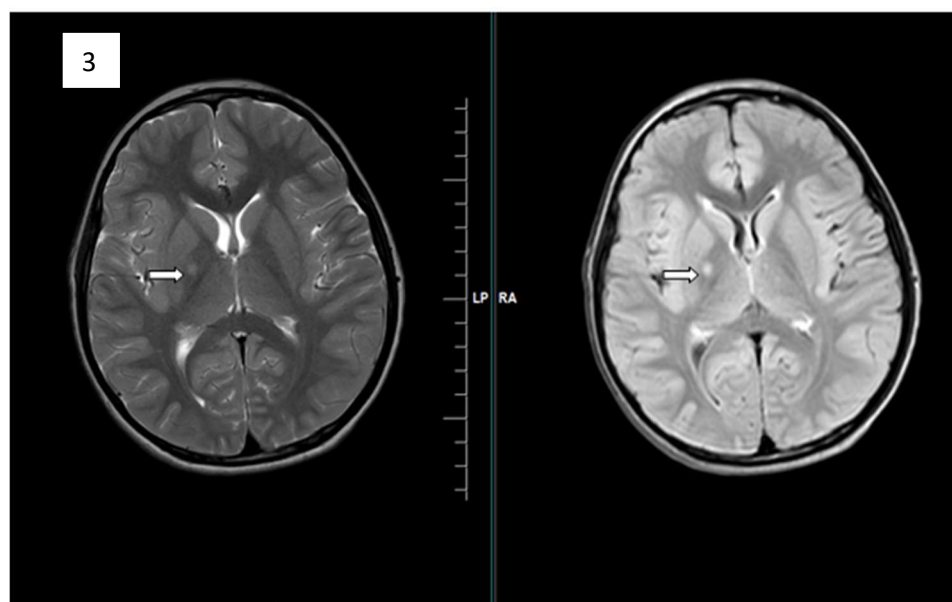
Since then, child was being facing challenges in carrying out routine activities which he was previously capable of accomplishing. According to the information provided by parents, child achieved developmental milestones within the expected range up until the age of 10 months, but later the progress of child was subsequently disrupted. Also, there is no significant family and birth history. On examination the child was oriented to time, place and person and interactive with the surroundings. And, was able to lift arm above head, comb hair and there was no difficulty in taking objects from height but had apraxia on horizontal gaze. Patient was vitally stable.

On central nervous system examination, patient had signs of cerebellar dysfunction evidenced by ataxic gait, intentional tremors, dysdiadochokinesia, rebound phenomenon and slurred speech. And, on motor nerve examination, child had hypotonia with diminished deep tendon reflexes. However, Cranial nerves examination, fundus examination, sensory examination and other systemic examinations were normal. Routine basic investigations like Complete Blood Count (CBC), Kidney function test (KFT), Liver function test (LFT), Serum electrolytes were normal. IQ testing showed 71 indicative of borderline intellectual functioning. EEG showed prolonged, generalized 2-3 HZ spike wave complex that have frontal prominence.

MRI brain was conducted and report revealed prominent cerebellar spaces and folia (Figure 2) indicative of cerebellar atrophy. Axial T2 and flair sequences showed focal hyperintensity in right globus pallidus (Figure 3). And, Alpha-fetoprotein report was suggestive of 640 ng/ml. The results of the tandem mass spectrometry suggested that everything was normal. Based on the findings, patient's diagnosis indicated presence of ataxia telangiectasia. Patient began receiving occupational therapy, speech therapy, physiotherapy and supplemented with Multivitamins. He was discharged and asked for follow up. To date, patient has not reported with the same symptoms or deterioration of symptoms.



**Figure 2** MRI Brain axial T2 sequence showing prominent cerebellar folia suggesting cerebellar atrophy



**Figure 3** MRI brain axial T2WI sequences showing focal hyperintensity in right globus pallidus

### 3. DISCUSSION

Ataxia Telangiectasia is characterized by the progressive onset of cerebellar ataxia during infancy, which is a prominent feature of the disorder. Additionally, affected individuals often develop progressive telangiectasia on the bulbar conjunctiva, as well as occasionally exhibiting a butterfly rash on the face (Border and Sedwik, 1958). Frequent sinopulmonary infections, including bronchiectasis are also common in people with Ataxia Telangiectasia and the peculiarity of eye movements can resemble ophthalmoplegia.

Additionally, retardation of stature growth and significant dwarfing are often observed in individuals and intelligence is typical borderline or normal. As the ataxia progresses, individuals may encounter challenges with tests that rely upon visual-motor coordination, leading to decline in IQ scores below the normal range. A vital role of the ATM protein is to regulate cell cycle checkpoints, which halt cell division to facilitate DNA repair in response to detected damage. This involves activating signalling pathways that prompt cell cycle arrest and DNA repair. This emphasizes the crucial role of ATM in maintaining genome integrity and preventing cancer development (Kastan and Lim, 2000).

ATM has shown to activate the antioxidant response pathway, which helps to scavenge excess ROS and maintain cellular homeostasis. This suggests that enhancing the activity of ATM could be potential therapeutic strategy for conditions associated with oxidative stress (Reichenbach et al., 2002). The use of routine serum alpha-fetoprotein testing for all children with persistent ataxia will help with early detection. Diagnosis was aided by the alpha-fetoprotein level being 640 ng/ml (Cabana et al., 1998; Huang et al., 2001).

Alpha-fetoprotein values are used as diagnostic index for neurological illness. Elevated levels of AFP were observed and found to be proportional to the severity of neurological manifestations in patients with the illness (Ayache et al., 1994). As cerebellar degeneration advances, it can lead to loss of granule cells in cerebellum which is important for fine motor control. This can cause variety of symptoms including difficulty with coordination, balance and speech. In addition, it can also affect the muscles involving in feeding and swallowing, making these tasks increasingly difficult over times (Kamsler et al., 2001).

Symptomatic treatment of infections and neurologic symptoms, immunoglobulin replacement therapy and regular monitoring for the development of malignancies can help in treating A-T (Martínez-Córdoba and Espinosa-García, 2020). In the above-mentioned case, the history, neurological examination and investigations were consistent with findings of Ataxia - Telangiectasia. Early diagnosis and management can help to improve outcomes and quality of life of the affected child. It is important for the health care providers to recognize the key features of Ataxia - Telangiectasia in order to make an accurate diagnosis and provide appropriate management and support for affected individuals and their families.

### 4. CONCLUSION

If a family member is diagnosed with Ataxia - Telangiectasia, carrier testing can be offered to other at-risk relatives to determine if they carry mutated ATM gene. Prenatal testing can also be offered to couples who are at risk of having a child with A-T to ascertain whether the fetus has inherited the mutated gene. Although, currently there is no known cure for Ataxia - Telangiectasia, symptomatic treatment is crucial and should involve an interdisciplinary team of health care professionals. Treatment options may include physical therapy, occupational therapy, speech therapy, paediatric immunology, paediatric haematologist- oncologist, nutritionist, child psychiatrist and paediatric neurology services. Early intervention and appropriate management can improve quality of life and outcomes for patients.

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#### Author's contribution

All the authors contributed equally to the case report.

#### Informed Consent

Informed Consent was obtained from the patient.

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**Conflict of interest**

The authors declare that there is no conflict of interests.

**Data and materials availability**

All data sets collected during this study are available upon reasonable request from the corresponding author.

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