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Case Report

Ataxia Telengiectasia: A Case Report

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ABSTRACT

Ataxia telangiectasia is a genetically inherited multisystem disorder with predominant feature being telangiectasia, sinopulmonary infection and cerebellar ataxia. In this report, a 10 year boy is described who presented to us with difficulty in walking and congestion in both eyes. He was diagnosed as a case of Ataxia Telangiectasia.

Keyword: Ataxia, Telangiectasia.

INTRODUCTION

Ataxia telangiectasia is a rare inherited disorder that affects the nervous system, immune system, and many parts of the body that leads to severe disability and is characterized by progressive cerebellar ataxia, oculocutaneous telangiectasia and recurrent respiratory and sinus infections.

Ataxia telangiectasia was first described in 1941 in a 9 year old child with progressive cerebellar ataxia and bilateral oculocutaneous telangiectasia are ported by Madame Louis Bar. [1] Earlier known as L Bar Syndrome, the term Ataxia Telangiectasia was introduced in 1958 by Border et al, who recognized the familial incidence proposing an autosomal recessive mode of inheritance for the disease. [2] The disease is sometimes referred to as Border Sedgwick Syndrome.

The defect in DNA repair gene is responsible for the Ataxia telangiectasia. Both allele mutation of ataxiatelangiectasia mutated (ATM) kinase gene results in arrest of cell cycle, DNA repair or apoptosis which leads to predisposition to cancer, ataxia due to progressive cerebellar degeneration, immunodeficiency and telangiectasia. [3,4]

Estimated incidence is in the range of one per 40,000 to 3, 00,000 live births with equal involvement of both sex and there is no racial or geographical preference. [5]

CASE REPORT

A 10 year old boy who is the second issue of non consanguineous parents completely immunized according to EPI schedule was admitted in our hospital with complaints of generalized weakness, cough, fever on and off, and redness in both eyes and unable to walk properly since 5 years.

Initially patient could walk but gradually imbalance became severe enough

to limit his standing ability. Congestion of the conjunctiva developed in both eyes simultaneously and was static. His birth history was uneventful and milestones were achieved on time. There was no significant family history.

On general examination, his vitals were stable, pallor was present, telangiectasia was present on bulbar conjunctiva of both eyes. Horizontal nystagmus was also present. On systemic examination, patient was conscious and oriented. He had scanning speech, ataxic and wide based gait. Cranial nerves were intact. Bilateral symmetrical loss of bulk of muscles was in both upper and lower limb. Tone was normal and power was 4+ in all four limbs. Deep tendon reflexes were absent and plantar were flexors. Sensory examination was within normal limits. Cerebellar involvement signs were positive and showed finger nose test, finger finger nose test, dysdiadochokinesia, heel knee test, tandem walking and Romberg sign. Intentional tremor and past pointing was also present.

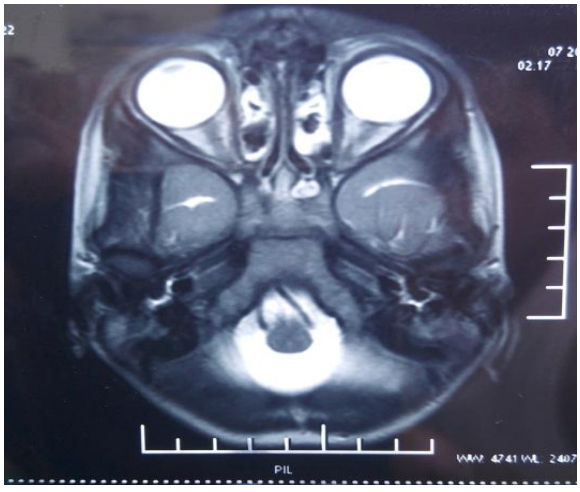

Fig 1: MRI showing cerebellar atrophy

On investigation, complete blood count, liver function test, renal function test, electrolytes, widal, HIV, urine routine microscopy, montoux, sputum for AFB, ultra sonography of abdomen and EEG was normal. X ray chest revealed patchy consolidation in right lung. Alfa feto protein level was 74.2 IU/L (N- 0.5 to 5.5 IU/ml) and Ig A levels was less than 0.26 gm/L (N - 0.7 to 4.0 gm/L). MRI revealed cerebellar atrophy.

Patient was managed conservatively and till date he is on regular follow up in our hospital.

DISCUSSION

In this case report, we have described a 10 year old boy who was diagnosed as a case of Ataxia Telangiectasia. In India AT was first described by Dogra and Manchanda since then various typical and atypical presentations of AT have been reported. [6] Ataxia, ocular telangiectasia, immunodeficiency, sinopulmonary infection and involuntary movements remain as the main presenting symptom in almost all patients of ataxia telangiectasia, all of which were present in our case. [7] Normal tone and absent DTR had made us bit hesitant initially to label this case Ataxia Telangiectasia as both are against the cerebellar lesion but detailed examination and results of investigation suit none other but this clinical profile. [8] Later, with further literature review we found that variant with normal tone and absent DTR are not so uncommon. Also, dystonia may present as late manifestation of the disease. Patient with ataxia telangiectasia have an increased susceptibility to respiratory infection, radiation and predisposition to malignancy. [3,4] Low IgA and high AFP levels are useful screening test with sensitivity of more than 95%. [9,10] M Moin et al in 2007 studied clinical and laboratory feature of ataxia telangiectasia in 104 Iranian patients with the age range of 1.6-23.5 years. Median range of presentation of disease was 9.5

years almost similar to our case. [11] Cranial MRI showing cerebellar atrophy in our case was further suggestive of possibility of ataxia telangiectasia. [12]

CONCLUSION

Ataxia telangiectasia is a rare disease and still is a clinical diagnosis. Clinicians are not familiar with the cases therefore very commonly misdiagnosed. On the other hand, there is an absolute need for further studies which can discover the early diagnostic measures and effective treatment.

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