



## Ataxia Telangiectasia- A Case Report

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### Abstract

*Ataxia Telangiectasia (AT) is a rare autosomal recessive disease with multisystem disorder. A-T is characterized by progressive cerebellar degeneration, telangiectasia, immunodeficiency, recurrent Sino pulmonary infections, radiation sensitivity, premature aging, and a predisposition to cancer development.*

*Here we report a case of 6-year-old female child who presented to our pediatric department with complaints of unsteady gait, feeding difficulty, hand mouth in coordination, slurring of speech diagnosed to be a case of Ataxia Telangiectasia. Our aim is to create a deep insight into the subject and help in adding valuable knowledge to the same.*

**Keywords:** *Ataxia telangiectasia, ATM gene, Cerebellar Atrophy.*

### Introduction

AT is a rare inherited disorder that affects the nervous, immune and other body system<sup>[1]</sup>. Its incidence is 1 in 100,000 births<sup>[2]</sup>. The male and female are equally affected. The two diagnostic hallmarks are 1<sup>st</sup> being ataxia and 2<sup>nd</sup> is oculocutaneous telangiectasia. It is relentlessly progressive<sup>[3]</sup>. The mutation of ATM gene mapped on 11q22-23 is responsible for this disease. The ATM gene is involved in cell division and DNA repair<sup>[4]</sup>. It also plays an important role in normal development and activity of several body systems and immune system.

### Case Report

6-year-old female child born out of non-consanguineous marriage was admitted in emergency department of pediatrics MKCG Medical college with complains of difficulty in

walking steadily and frequently falling while walking since last 6 months. Also, she was unable to feed herself due to in coordination between hand and mouth. She also developed slurring of speech and difficulty in swallowing (deglutition) during the same period. Past history revealed recurrent respiratory tract infections which was treated in outpatient basis and cured. On examination, the child had ataxic gait and bulbar telangiectasia (Fig 1 and 2), oculomotorapraxia, truncal ataxia. The child has been immunized with all the vaccines as per ACVIP schedule.

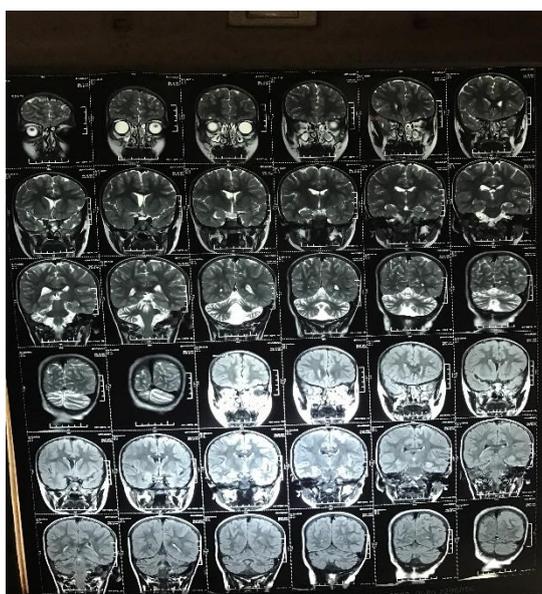
The AFP level was found to be high(194.68 ng/ml). The serum IgA level was done and found to be normal(100 mg/dl).MRI brain showed diffuse cerebellar atrophy [Fig 3].The parents were adequately counselled regarding the complications and prognosis of the disease, and was discharged on day 3.



**Fig 1** abnormal stature



**Fig 2** bulbar telangiectasia



**Fig 3** MRI showing diffuse cerebellar atrophy

### Discussion

A-T is characterized by progressive cerebellar degeneration, telangiectasia, immunodeficiency, recurrent Sino pulmonary infections, radiation sensitivity, premature aging, and a predisposition to cancer development, especially of lymphoid origin. Other abnormalities include poor growth, gonadal atrophy, delayed pubertal development and insulin resistant diabetes<sup>[5]</sup>. Most children with classic A-T have stable neurologic symptoms for the first 4–5 years of life, they initially may be labeled as having “ataxic cerebral palsy”<sup>[6]</sup>. During school years children often have increasing difficulty with reading because of impaired coordination of eye movement. At the same time, other problems with fine motor functions (writing, coloring, and using utensils to eat) and with dysarthria may arise<sup>[7,8]</sup>. Sino pulmonary infections are common in people with A-T. About two-thirds of people with A-T have abnormalities of the immune system<sup>[9,10]</sup>. The most common abnormalities are low levels of one or more classes of immunoglobulin (IgG, IgA, IgM or IgG subclasses), failure to make antibodies in response to vaccines or infections, and lymphopenia, especially affecting T-lymphocytes. Feeding and swallowing (deglutition) may become difficult for people with A-T as they age<sup>[11]</sup>. Most devastating symptoms of A-T are a result of progressive cerebellar degeneration, characterized by the gradual loss and/or aberrant location of PCs and, to a lesser extent, the gradual loss of granule cells<sup>[12,13]</sup>. Approximately 95% of people with A-T have elevated serum AFP levels after the age of two, and measured levels of AFP appear to increase slowly over time<sup>[14]</sup>.

### Conclusion

Recurrent Sino pulmonary infections, progressive ataxia and bulbar/retinal telangiectasia can easily clinch the diagnosis of A-T. In the initial 5 years of life they have recurrent acute respiratory infections and failure to thrive and to confirm Alfa fetoprotein estimation should be done<sup>[15]</sup>. Progressive ataxia develops after 5 years. High

degree of suspicion of the rare entity AT will give a path towards clinical diagnosis. There should be good parental counselling and explanation of the disease.

## References

- Huang KY, Shyur SD, Wang CY, Shen EY, Liang DC. Ataxia telangiectasia: report of two cases. *Journal of microbiology, immunology, and infection* Wei mian yu gan ran za zhi. 2001 Mar;34(1):71-5.
- Majumder B, Rahman MJ, Ahmed S. Ataxia Telangiectasia-A Case Report.
- Smith LL, Conerly SL. Ataxia-telangiectasia or Louis-Bar syndrome. *Journal of the American Academy of Dermatology*. 1985 Apr 1;12(4):681-96.
- Savitsky K, Bar-Shira A, Gilad S, Rotman G. A single ataxia telangiectasia gene with a product similar to PI-3 kinase. *Science*. 1995 Jun 23;268(5218):1749.
- Nissenkorn A, et al. Endocrine abnormalities in ataxia telangiectasia: findings from a national cohort. *Pediatr Res*. 2016;79(6):889–94
- Boder E. Ataxia-telangiectasia: an overview. *Kroc Found Ser*. 1985;19:1–63.
- Crawford TO. Ataxia telangiectasia. *Semin Pediatr Neurol*. 1998;5(4):287–94.
- Boder E, Sedgwick RP. Ataxia-telangiectasia; a familial syndrome of progressive cerebellar ataxia, oculocutaneous telangiectasia and frequent pulmonary infection. *Pediatrics*. 1958;21(4):526–54.
- Nowak-Wegrzyn A, et al. Immunodeficiency and infections in ataxia-telangiectasia. *J Pediatr*. 2004;144(4):505–11.
- Driessen GJ, et al. Antibody deficiency in patients with ataxia telangiectasia is caused by disturbed B- and T-cell homeostasis and reduced immune repertoire diversity. *J Allergy Clin Immunol*. 2013;131(5):1367–75. e9
- Lefton-Greif MA, et al. Oropharyngeal dysphagia and aspiration in patients with ataxia-telangiectasia. *J Pediatr*. 2000;136(2):225–31
- Vinters HV, Gatti RA, Rakic P. Sequence of cellular events in cerebellar ontogeny relevant to expression of neuronal abnormalities in ataxia-telangiectasia. *Kroc Found Ser*. 1985;19:233–55.
- Gatti RA, Vinters HV. Cerebellar pathology in ataxia-telangiectasia: the significance of basket cells. *Kroc Found Ser*. 1985;19:225–32
- Stray-Pedersen A, et al. Alpha fetoprotein is increasing with age in ataxia-telangiectasia. *Eur J Paediatr Neurol*. 2007;11(6):375–80.
- Stray-Pedersen A, Borresen-Dale AL, Paus E, Lindman CR, Burgers T, Abrahamsen TG. Alpha fetoprotein is increasing with age in ataxia-telangiectasia. *European journal of paediatric neurology*. 2007 Nov 30;11(6):375-80.