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Case Report

Complicated Pneumonia in a Case of Ataxia Telangiectasia

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ABSTRACT

Ataxia Telangiectasia (AT) is a rare autosomal recessive disease with multisystemic disorder. It is characterized by progressive neurological impairment, cerebellar ataxia, variable immunodeficiency, impaired organ maturation, x-ray hypersensitivity, ocular and cutaneous telangiectasia and predisposition to malignancy.

Here we report a case of 11 year male boy who was operated for cleft palate at an age of 2 years and diagnosed with at the age of 5 years now presented with complicated pneumonia.

Key words: Ataxia telangiectasia, cleft palate, complicated pneumonia, malignancy.

INTRODUCTION

AT is a rare inherited disorder that affects the nervous, immune and other body system. [1] It is seen in all regions of the world. Its incidence is 1 in 100,000 births. [2] The male and female are equally affected. The two diagnostic hallmarks are 1st being ataxia and 2nd is occulo cutaneous telangiectasia. It is relentlessly progressive. [3] 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. [4] It also plays an important role in normal development and activity of several body systems and immune system.

CASE REPORT

11 year old male boy born out of 3rd degree consanguineous marriage was admitted in emergency department of paediatrics MKCG Medical College with complains of difficulty in breathing for last 10 days which now became severe. The child had ataxic gait and bulbar telangiectasia (Fig 1 and 2). On taking history, it was found that the child was operated for cleft palate at the age of 2 years and diagnosed as a case of AT at the age of 5 years. The AFP level was found to be high. Since the time of diagnosis there have been repeated chest infections which have been treated appropriately. The child has been immunized with all the vaccines as per ACVIP schedule. This time the respiratory rate was 52/min and breath sounds were grossly reduced more on the left side of the chest. It was suspected as a case of pulmonary TB. Mantoux and sputum AFB and CBNAAT came negative and on doing a chest x-ray, multiple pneumatocele were found (fig3).Hence a diagnosis of complicated pneumonia was made and appropriate antibiotics (ceftriaxone and ampiclox) were given for 14 days following which the child improved. The serum immunoglobulin levels were tested and found to be low (IgA<.10 mg/dL and IgG-1939mg/dL)

Fig 1 abnormal stature

Fig 2 bulbar telangiectasia

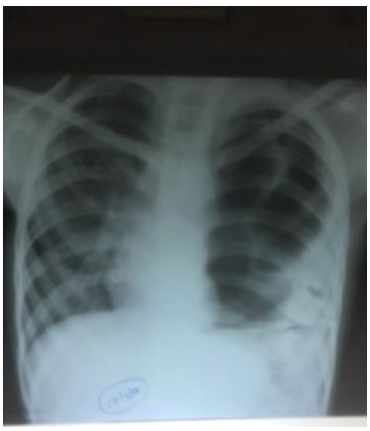

Fig 3 chest x-ray showing pneumatocele

DISCUSSION

A case of AT is a host for many diseases because of weakened immune system. They may develop chronic lung infections and are also at a risk of developing cancer like leukemia and lymphoma. Beyond 5 years of age, the progression of ataxia becomes apparent and by the age of 11 years the child is wheel chair bound. Death typically occurs in early adolescence from bronchopulmonary infection (46%), malignancy (21%) and from other causes by (5%). [5] The median age of death is 20 years.

CONCLUSION

There are no reports on association of cleft palate and AT till now. All cases of ataxia should have ophthalmologic consultation to look for telangiectasia (bulbar and retinal).Initial 5 years of life has recurrent acute respiratory infections and failure to thrive. Bulbar telangiectasia will clinch the diagnosis of AT and to confirm Alfa foeto-protein estimation should be done. [6] Progressive ataxia develops after 5 years. Early presentation, pes caves, UMN signs and telangiectasia, in view of ataxia will give a clue towards Friedreich's ataxia. Neuroimaging though can diagnose ICSOL, but in these two conditions, neuroimaging will not be of much help. 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.

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