ATAXIA TELANGIECTASIA: A RARE CASE REPORT

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ABSTRACT

Ataxia-Telangiectasia (A-T) is a rare hereditary neurodegenerative disorder. Ataxia and telangiectasia are the hallmarks of the disease. Individuals with classic A-T have mutations in ATM, the only gene known to be associated with ataxia telangiectasia. We report a 13-year-old girl who presented with respiratory tract infection. She also had slurred speech, unsteady gait and telangiectasias in the eyes.

KEYWORDS

Ataxia, Telangiectasia, Neurodegenerative Disease.

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INTRODUCTION

Ataxia Telangiectasia (AT) is an autosomal recessive syndrome characterised by progressive cerebellar ataxia, immune-deficiency, which usually takes the form of sinopulmonary infections, oculocutaneous telangiectasia, X-ray hypersensitivity and predisposition to lymphoid malignancies. The responsible gene, ATM gene has been mapped to band 11q22-23.

The first report of a family with ataxia telangiectasia is attributed to Syllaba and Henner (1926).¹ They observed progressive choreoathetosis and ocular telangiectasia in 3 members of a single family. The next report was published by Louis-Bar (1941), who described the disease in 9-year-old Belgian child.² The disorder was further elaborated and delineated as a distinct clinico-pathological entity by Biemond (1957) and Boder and Sedgwick (1957); these latter authors suggested the name Ataxia-telangiectasia and Centerwall and Meller (1958) proposed the eponym Louis-Bar Syndrome.

CASE REPORT

A 13-year-old girl, 3rd issue of a third degree consanguineous marriage was admitted to the Paediatric Department of Krishna Institute of Medical Sciences, Deemed University, Karad, Maharashtra, on 18/06/2016 with respiratory tract infection.

The patient had history of frequent admissions in view of lower respiratory tract infection. She attended school till 9 years of age and was academically good, then she developed progressive gait deterioration.

Her birth history was uneventful and she attained her milestones of development at appropriate ages.

She has a healthy 17-year elder brother and there is no family history of such kind of illness. The 1st child died at the age of one and a half years due to aspiration following convulsion.

On physical examination, pallor and bilateral conjunctival telangiectatic lesions were noted. She was undernourished. Her sexual maturity was stage 2 according to Tanner's staging. There were evidences of collapse-consolidation of left lung in the form of decreased chest expansion and tactile vocal fremitus on the left side. Her speech was slurred and she was unable to stand without support. Cranial nerves were intact with normal fundoscopic findings. There was marked hypotonia in both the lower limbs with absent deep tendon reflexes. Sensory function was preserved. Cerebellar function was abnormal and showed nystagmus, ocular apraxia, intention tremor, past-pointing and dysidiadochokinesis.

Lab investigations revealed mild anaemia and mild leukocytosis. Serum alpha-fetoprotein was 145 ng/mL. Mantoux test was negative. Chest X-ray showed collapse-consolidation of left middle segment. MRI Brain showed diffuse cerebellar atrophy and dilatation of fourth ventricle. USG abdomen – pelvis revealed normal findings.

The child was treated for respiratory tract infection symptomatically and supportively. Speech therapy and physiotherapy was advised.

Fig. 1: Telangiectasias of the Eyes

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Fig. 2: Inability of the Patient to Stand without Support

DISCUSSION

Our patient is a fairly typical case of ataxia-telangiectasia presenting with most of the features of this disorder. A-T is caused by a defect in the ATM gene, which is responsible for managing the cell's response to multiple forms of stress including double-strand breaks in DNA. ATM signalling is required to sense and initiate repair of DNA double-strand breaks. Therefore, nuclear genomic instability resulting from loss of this function is regarded as a major mechanism underlying the pathology of A-T.³

The incidence of ataxia-telangiectasia is about 1 case in 100,000 births.⁴ It occurs equally among males and females. No characteristic features are detectable during early childhood. Ataxia is the first diagnostic hallmark having its onset in the first years of life. Beyond the age of 5 years, the progression of ataxia becomes increasingly apparent and the child requires a wheelchair by age 10 or 11 years.

Oculocutaneous telangiectasia, the second diagnostic hallmark of ataxia-telangiectasia,⁵ usually has a later onset

than the ataxia, typically at age 3-6 years. The progression of the disease is apparent in subsequent years. The clinical diagnosis becomes most apparent after age 10 years.

A-T patients are often very thin. This may be due to a poor appetite or the inherent characteristics of the disorder. Patients tend to have delayed puberty. Mental retardation occurs in nearly 10% of patients. It is not a common feature of the disease, but an arrested cognitive development for new skills appears in some patients as they grow.

People with A-T have a highly increased incidence (approximately 25% lifetime risk) of cancers, particularly lymphomas and leukaemia, but other cancers can occur. Chronic lung disease develops in most of these patients.

Immunodeficiency is a cardinal feature of this disorder and it is an important factor in causing recurrent sinopulmonary infections. The defect in the immunological system is a dysgammaglobulinaemia, characterised by selective deficiency of IgA in the serum, saliva, tears, nasal and respiratory mucosa and IgE deficiency in the serum.

Diagnosis is usually achieved clinically by examination and identification of both ataxia and oculoteliangiectasia or skin telangiectasia. Laboratory tests often show elevated serum AFP level, low lymphocyte count and other immunological abnormalities. MRI and Computed Tomography (CT) scans may show cerebellar atrophy. Cytogenetic and molecular testing will confirm the diagnosis.

CONCLUSION

Ataxia Telangiectasia (AT) follows progressive course. Many patients are confined to a wheelchair in their teens.

The life expectancy of people with A-T is highly variable. The average is approximately 25 years, but continues to improve with advances in care. Survivals up to 40 years have been reported.

The two most common causes of death are chronic lung disease and cancer.

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