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Case Report

Ataxia Telangiectasia: A Case Report

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ABSTRACT

Ataxia Telangiectasia (AT) is a rare autosomal recessive neurodegenerative disease. It is characterized by progressive cerebellar ataxia, oculocutaneous telangiectasia, recurrent respiratory and sinus infections. AT is caused by a defect in the ATM gene, the only gene known to be associated with AT. I am reporting on an eight-year-old Saudi girl who presented with an unsteady gait, frequent falls, and telangiectasia of the eyes. She has also had frequent episodes of respiratory tract infections.

Keywords: Ataxia telangiectasia, ATM gene, Cerebellar atrophy.

INTRODUCTION

AT is a rare inherited disorder that affects the nervous, immune and other body systems. [1] Its incidence is 1 in 100,000 births. [2] Males and females are equally affected. The two diagnostic hallmarks are having ataxia and oculocutaneous telangiectasia, but it is relentless in its progression. [3] The mutation of the ATM gene mapped on 11q22-23is 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 the immune system.

CASE REPORT

An eight-year-old girl, the first child of her first-degree cousin parents, without a family history of significant diseases. She was seen in The Pediatric Clinic of the National Guard Comprehensive Specialized Clinic in Riyadh, Saudi Arabia with complaints of repeated falls while walking for the last 2 years. She also developed problems in daily activities, such as eating, carrying, gripping or holding, as well as increased difficulty with writing and coloring. Her speech became slurred and she developed drooling of saliva. Since she was three years of age, she had been suffering from repeated episodes of otitis media. She attained her milestones of development at appropriate ages. She has healthy five- and three-year-old younger brothers without any familial history of this illness or malignancy. Physical examination revealed mild pallor with bilateral bulbar telangiectasia, Figure 1. Her body build and nutritional status were below the third centile. Cranial nerves were intact with normal fundoscopic findings. There was marked hypotonia in the lower limbs with diminished deep tendon reflexes. Her gait was ataxic with an impaired tandem gait. Sensory function was intact. Cerebellar function was abnormal, showed past-pointing, dysdiadochokinesia, and nystagmus.

Investigations revealed a normal CBC, renal profile, blood sugar, vitamin D, TSH, and CPK. IGA and IGE were low. Serum α- Fetoprotein was 244 ng/L. Flow cytometry: reduced CD4 and CD8 T-cell count, reduced B-cell counts and normal NK-cell. MRI of the brain showed mild

cerebellar and vermian atrophy, Figure 2. After counseling, symptomatic treatments were ensured. Physical and speech therapy were also advised.

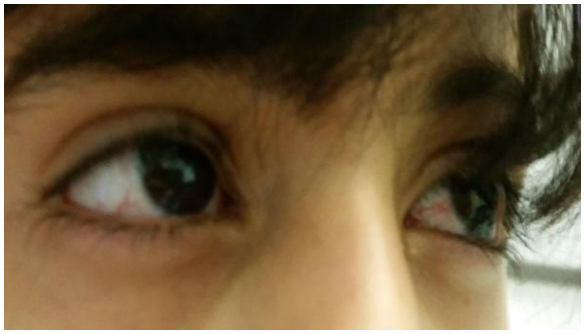

Figure 1: Bilateral bulbar conjunctival telangiectasia

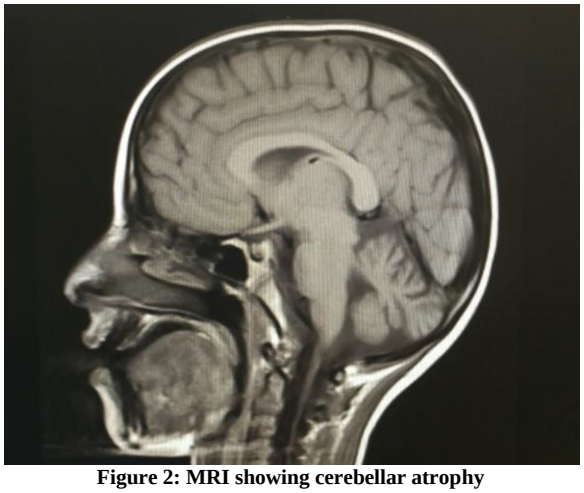

Figure 2: MRI showing cerebellar atrophy

DISCUSSION

In my index case, there was progressive cerebellar ataxia, bulbar telangiectasia, raised serum alpha-fetoprotein, decreased IGA and IGE levels, and characteristic MRI findings. These issues finalized our diagnosis of AT.

In general, AT is a multisystem disorder characterized by progressive neurologic impairment, cerebellar ataxia, progressive immunodeficiency, ocular and cutaneous telangiectasia, increased risk of lymphoreticular malignancy, and hypersensitivity to ionizing radiation. Telangiectasia can also appear on sun-exposed areas of skin such as the face and ears. Both humoral and cellular immunity show impairment in patients with AT. The most common immunologic abnormality is the absence or a decreased level of serum and salivary level of IgA. This deficiency has been found in 50% to 80% of AT patients. [5] These patients also have an increased susceptibility to sinopulmonary infection, x-ray hypersensitivity, and predisposition to malignancy. [6] Chromosomal abnormalities in the specific cell population are a characteristic finding in AT patients. The specific defect has been shown as an increased tendency for spontaneous breakage and rearrangement of chromosomes 2,6,7,8,14,22, and X. [7,8] Clinical characteristics include progressive ataxia (100%), telangiectasia of skin or conjunctivae (83.8%) and the ears (70.2%), eye movement disorder (apraxia of horizontal and vertical saccadic eye movements) (80.6%), choreoathetosis (87.1%), and dysarthria in almost all cases. The index case had progressive ataxia, telangiectasia over bulbar conjunctiva and dysarthria.

Mental retardation occurs in nearly 10% of patients, which was shown in a recent study. [9] It is not a common feature of the disease, but arrested cognitive development for new skills appears in some patients as they grow. Serum α-fetoprotein level, a useful screening test is usually elevated in AT, which is evident in this case. My index case also has mild cerebellar atrophy on MRI, further suggesting the possibility of AT. [9]

CONCLUSION

AT is a rare, multisystemic disease. Clinicians are not familiar with many cases, so it is commonly misdiagnosed; however, progressive ataxia, bulbar telangiectasia, and recurrent sinopulmonary infections can finalize the AT diagnosis. Management must be multidisciplinary. Prognosis is poor, and depends primarily on respiratory infections and the susceptibility to cancer.

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