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Case Report

Case report of ataxia telangiectasia

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ABSTRACT

Ataxia telangiectasia is rare inherited disorder characterized by progressive, neurodegenerative, variable immunodeficiency, celebral ataxia, ocular and cutaneous telangiectasia. They are prone for sinopulmonary infection also at risk of cancer development. Hallmark of disease is ataxia and telangiectasia. Incidence is 1 in 1,00,000 live births. 10-year-old presented to us with chief complaint of not able to Maintain balance and walk steady since 5 years of age. on examination bulbar telangiectasia was present. Also, dysdiadokinesia, past pointing, intention tremor was present. Magnetic resonance imaging (MRI) of the brain was done. Alpha fetoprotein was more than 1000. Vitamin E with balance exercise as supportive treatment starred. Ataxia begin during infancy when child start to walk, 2nd most hallmark of disease. In bulbar telangiectasia, average life span is 25-year-old. It is a multisystem genetic disorder. Death occurs due to cancer and infection. Counseling plays an important role as there is no cure for disease.

Keywords: Ataxia-telangiectasia, Occulomotor apraxia, ATM gene, Lymphoreticular neoplasia

INTRODUCTION

Ataxia telangiectasia is a rare inherited disorder characterized by progressive neurodegenerative, variable immunodeficiency, cerebral ataxia, ocular and cutaneous telangiectasia. It is an autosomal recessive multisystem involvement disorder.[1]

They are prone for sinopulmonary infection, also at risk of cancer. Ataxia starts at 2 years of age and by adolescence they loss ambulation. Gene involved is ATM gene located at 11q22-q23.[2] ATM mutation results in DNA damage result in arrest in cell cycle, DNA repair or apoptosis.[3-6] Oxidative stress is also implicated in ataxia telangiectasia.[7-11]

There is an immunologic abnormality which involved both cellular and humoral arm of immune system.[7] There is an increased incidence of chromosomal breakdown and rearrangement as there is increase cellular sensitivity to ionizing radiation. Hallmark of disease is ataxia and telangiectasia. Incidence of disease 1 in 100,000 births.[2]

The condition was first described by syllaba and Henner in 1929.[12]

CASE REPORT

A 10-year-old male child was presented to New Civil Hospital with chief complaints of not being able to walk steadily and maintain balance while walking since 5 years of age which was gradually progressive in nature. Patient also gave history of drooling of saliva, slurring of speech. Child was born out of non-consanguineous marriage. Patient was not able to feed self, hold object and also requires support while walking. Past history of repeated upper respiratory tract infection (URTI) was present. Child was immunized for age. On examination bulbar telangiectasia was present. Tone, power, superficial and deep reflexes were normal. Dysdiadockinesia, past pointing, tandem walking, intention tremors were seen. Routine investigations were sent like complete blood count (CBC), C-reactive protein (CRP), renal function test (RFT) with electrolytes, and liver function test (LFT) with enzymes. Neurophysician opinion was taken, and advice for magnetic resonance imaging (MRI) brain and alpha-

fetoprotein was given. MRI brain showed cerebellar atrophy and basal ganglia involvement, and alpha-fetoprotein >1000. Genetic study (whole exome gene sequencing done) which shows ATM gene involvement, which further confirmed the diagnosis. Prognosis was explained to relatives. Since no specific treatment available, balance exercise and vitamin-E was given to patient. Vitamin-E has immunomodulating effect, antioxidant property and helps to slow down disease progression.

Figure 1: Bulbar telangiectasia.

Figure 2:

DISCUSSION

Presenting symptom of ataxia telangiectasia is ataxia, which start when child Begin to walk, develop ataxic gait and truncal sway while walking.[2,13] In our case child presented to us at 10 year of age with ataxia which was progressive in nature.

Ocular manifestation is seen in A-T include oculomotor apraxia of horizontal gaze, strabismus, hypometric saccade pursuit abnormalities and nystagmus. Site of telangiectasia include bulbar conjunctiva, over bridge of nose, on ear, expose surface of extremity.[2] In our case also bulbar telangiectasia was present. Cutaneous changes seen in ataxia telangiectasia are cutaneous telangiectasia, mottled hypopigmentation and hyperpigmentation and poilkilodermatous appearance.

Alfa fetoprotein normal value after 1 year of age is <10 ng/ml.[14] In more than 95% of cases elevated alfa fetoprotein seen.[15] In our case too alfa fetaprotein was elevated which support the diagnosis.

A-T are at risk of lymphoreticular tumor which include lymphoma, leukemia and Hodgkin and brain tumor. In adult solid tumor are more frequent.

Due to immunologic abnormalities, they are prone for sinopulmonary infection as disease progress bronchiectasis develop in these patients. There is decreased level of serum and secretary immunoglobulin IgA, IgG2, IgG4 and IGE level. We have not done IGA or IgG test. In most of studies streptococcal pneumoniae isolated as most frequently encounter infection.[16] As the disease progress feeding and swallowing become difficult due to progressive cerebellar degeneration (loss of granule cell).[17]

Life expectancy is variable average being 25 year most of them die due to infection or tumor.

CONCLUSION

Ataxia telangiectasia is a rare multisystem genetic disorder. Repeated respiratory tract infection, ataxia telangiectasia helps in making diagnosis which is often supported by increased level of alpha-fetoprotein, disease has poor prognosis. Death can occur due to cancer and infection, also increase in morbidity as ambulatory function is affected. Counselling plays a very important role in such cases.

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