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Ataxia Telangiectasia with Hodgkin's Lymphoma

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SUMMARY

Ataxia Telangiectasia is an autosomal recessive disorder characterized by cerebellar degeneration, occulocutaneous telangiectasias, progressive immunodeficiency, hypersensitivity to ionizing radiations and associated increased risk of variable cancers especially lymphomas and lymphocyte leukemias. It is a rare disease with prevalence of less than 1 per 100,000(1). We report here a case of Ataxia Telangiectasia in seven years old girl complicated with Hodgkin's lymphoma. All her three sisters had similar complaints of ataxia with out lymphoma.

INTRODUCTION

Ataxia Telangiectasia is an autosomal recessive disorder. It is a rare disease characterized by cerebellar degeneration, occulocutaneous telangiectasias, progressive immunodeficiency, hypersensitivity to ionizing radiations and associated increased risk of various cancers specially lymphomas and lymphocytic leukemias[1]. However primary carcinoma of stomach, liver, ovary, salivary glands, oral cavity, breast and pancreas have also been reported. The relative risk that cancer would develops in an adult heterozygote for ataxia telangiectasia, is estimated to be 2-3% for men and 3% for women[3]. Histopathological changes in CNS include Purkinje cell loss with empty baskets and numerous axonal spheroids, dorsal column demyelination with astrocytic proliferation and severe anterior horn cell degeneration[2,4].

Immunodeficiency is characterized by lymphopenia, hypogammaglobulinemias and decreased T-cell response[1]. Elevated alpha-fetoprotein levels readily distinguish Ataxia Telangiectasia from other ataxias and immunodeficiency syndromes. Cytogenetic studies to look for chromosomal aberrations using amniotic cell culture can be used for prenatal diagnosis[5,6].

CASE REPORT

A 7 year old girl presented in OPD with 2 years history of redness of eyes followed by 2 month history of progressively worsening gait and dysarthria. She also developed progressively enlarging non-tender swelling in right cervical area for the last 4 months. She had normal birth events and insignificant past history and was vaccinated. She attained developmental milestones normally till the age of 3 years, when she developed ataxia and dysarthria. Her parents are first cousins. She has 3 brothers and 3 sisters. All her sisters (12 years, 10 years and 9 years old respectively) had similar complaints of ataxia, slurred speech and red eyes with the onset at 5 years of age. Their problems have worsened gradually to such an extent that now they are non bed ridden. On examination, the patient was alert, co-operative with dull facies, slurred (dysarthric) speech and was ataxic. She had conjunctival telangiectasias, nystagmus but normal fundus. Truncal ataxia, tremors, diminished reflexes but normal power and tone was seen. An enlarged lymph node 7x6 cm, firm, non-tender in right anterior cervical region was found. There was no other significant lymph node enlargement or hepatosplenomegaly. Alpha-feto protein was markedly raised. Lymph node biopsy showed Hodgkin's lymphoma. Except for microcytic hypochromic anemia, other hematological or biochemical indices were normal. On the basis of history, examination and laboratory reports, the final diagnosis was Ataxia-Telangiectasia complicated by Hodgkin's lymphoma.

She was diagnosed as a case of Ataxia Telangiectasia on the basis of cerebellar

dysfunction, ocular telangiectasias, raised alpha-fetoprotein and associated lymphoma. Her sisters were diagnosed to have similar disorder without lymphoma.

DISCUSSION

Ataxia Telangiectasia is an autosomal recessive condition. The usual presentation is with the signs of cerebellar dysfunction i.e., dysarthria, ataxia, nystagmus etc. closely followed by appearance of telangiectasias especially occular[2]. There may be associated history of repeated chest or sinus infections. CNS symptoms tend to progress over the years and can make the patient bed ridden. Mortality is usually due to overwhelming infections or carcinomas[6,7]. Other important causes of ataxia include infectious causes like cerebellar abscess, brain tumors including tumors of cerebellum and frontal lobe as well as neuroblastoma, several metabolic disorders including abetalipoproteinemia, arginosuccinicacidurea & Hartnup disease, Friedreich's ataxia is another important cause of ataxia. In the absence of telangiectasia, Ataxia Telangiectasia is mostly confused with a cerebral palsy but the diagnosis is made once telangiectasia appears. The patients as well as first-degree relatives (heterozygotes) have usually a high incidence of cancer. Most common are the lymphomas and lymphocytic leukemias; alpha-feto protein level readily distinguish ataxia telangiectasia from other ataxias and immunodeficiency syndromes. Cytogenetic studies to look for chromosomal aberrations using amniotic cell culture is important in diagnosis of ataxia telangiectasia, we can also diagnose the patient and confirm by alpha-fetoprotein levels. Genetic counseling of the parent's family is very important as this disease can be diagnosed antenatally[6].

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