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RESEARCH ARTICLE

ATAXIA TELANGIECTASIA: A CASE REPORT.

Khaire P B¹, Dahyalkar A A², Patil M A³, Kachhap R⁴ and Kasliwal C A².

1. Associate Prof., Department of Pediatrics, Govt. Medical College, Aurangabad.
2. Assistant Prof., Department of Pediatrics, Govt. Medical College, Aurangabad.
3. JR2, Department of Pediatrics, Govt. Medical College, Aurangabad.
4. JR3- Department of Pediatrics, Govt. Medical College, Aurangabad.

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Abstract

Ataxia-telangiectasia (A-T) is a rare hereditary neurodegenerative disorder. Ataxia and telangiectasias are the hallmarks of the disease. Due to its wide clinical heterogeneity, it often leads physicians to an incorrect or missed diagnosis, and insight into this rare disease is important. There is no gold standard diagnostic test and diagnosis relies on clinical evaluation, exclusion of similar conditions, and supportive laboratory tests. More than 99% of individuals with classic A-T have mutations in ATM, the only gene known to be associated with ataxia-telangiectasia. We report 10 year old boy who was presented with respiratory tract infections, unsteady gait, frequent falls and telangiectasias of the eyes. He also has had frequent episodes of respiratory tract infections.

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Introduction:-

Ataxia telangiectasia (AT) is a rare multisystem, neurodegenerative genetic disorder. (1) Due to its wide clinical heterogeneity, it often leads physicians to an incorrect or missed diagnosis, and insight into this rare disease is important. The syndrome was not mentioned until 1941 when Louis-Bar reported a case of a 9-year-old child. (2) Boder et al recognized the familial incidence proposing an autosomal recessive mode of inheritance for the disease. (1) The ataxia telangiectasia mutated (ATM) kinase initiates a well characterized response to DNA damage, resulting in arrest of cell-cycle, DNA repair, or apoptosis. (2) A-T has led to much speculation about additional pathogenic mechanisms including oxidative stress. How ATM is involved in oxidative stress management remains unclear as do other potential roles in cellular homeostasis in the absence of DNA damage. (2) The incidence of ataxia-telangiectasia is about 1 case in 100,000 births. (3) The frequency of ataxia telangiectasia mutant alleles heterozygosity was reported to be 1.4- 2% of the general population. (2-3) Ataxia-telangiectasia occurs equally among males and females.

Case Report:-

A 10 year old boy who is the first issue of non consanguineous parents completely immunized according to EPI schedule was admitted to Government Medical College Hospital Aurangabad, on 26.01.2010 with the complaints of generalized weakness and difficulty in walking from 3 years of life and

Corresponding Author:- Dahyalkar AA.

Address:- Assistant Prof., Department of Pediatrics, Govt. Medical College, Aurangabad.



Fig 1

Fig 2

Fig.1 & Fig.2:-showing bilateral bulbar conjunctival telangiectasia``

breathlessness since 5 days. The patient initially could try to walk but later on he fell on the ground as he developed imbalance in walking that gradually involved also in standing capacity within one month. He also developed congestions of the conjunctiva that has appeared at age 1 year. The congestions were first noted in the interpalpebral bulbar conjunctiva away from the limbus. Past history - suggestive of frequent respiratory infections. Birth history was normal.

On general examination, gait was ataxic. The patient was found anemic. Congestion was seen in both eyes that is radiating from the both corners to limbus. The patient was conscious and cooperative. On CNS examination, cranial nerves were normal. There were hypotonia in all four limbs which was more marked in lower limbs with diminished jerks and flexor plantar reflexes. Sensory function was intact. Contractures were present. Cerebellar function was abnormal showed slurred speech, intention tremor, past pointing and dysdiadochokinesia. On investigation, complete blood count, lipid profile, LFT, chest x-ray, serum electrolytes, ultra sonogram of whole abdomen, serum Ig G and ECG showed normal results.

Serum α -fetoprotein was increased that is 1250 ng/L. serum Ig A level decreased 0.5 gm /l. There was no specific treatment was provided for AT.

Discussion:-

In our index case, there was respiratory infections, progressive cerebellar ataxia, bulbar telangiectasias, raised serum alpha-fetoprotein and decreased Ig A level. These clinch us to reach the diagnosis of Ataxia telangiectasia. In general, Ataxia telangiectasia (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. (1, 3, 4) Telangiectasia can also appear on sun exposed areas of skin such as face and ears, occasionally arise in liver and lungs. (5) Both humoral & cellular immunity are impaired in patients with AT. (5) The most common immunologic abnormality is the absence or decreased level of serum & salivary level of Ig A. (5) This deficiency has been found in 50 to 80 percent of these patients. Patients with AT have an increased susceptibility to sinopulmonary infection, x-ray hypersensitivity, and predisposition to malignancy. Chromosomal abnormalities in specific cell population are characteristic finding in AT patients. The specific defect has been shown as an increased tendency for spontaneous breakage & rearrangement of chromosome 2,6,7,8,14,22 & X.24,25. (2) Clinical characteristics include progressive ataxia (100%), telangiectasia of skin or conjunctivae (83.8%) and of ears (70.2%), eye movement disorder (apraxia of horizontal and vertical saccadic eye movements) (80.6%), choreoathetosis (87.1%), dysarthria in almost all cases. (4,5,6) The index case had progressive ataxia, telangiectasia over bulbar conjunctiva and dysarthria.

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