Louis Bar Syndrome

A young 18 years female presented with slowly progressive difficulty in standing and walking since three years of age associated with clumsiness and jerking of upper limbs for past three to four years. She had swaying movements during standing with progressive inability to stand without support leading finally to bedridden state. There was no significant past or family history. On examination bilateral telangiectasias in both temporal and nasal bulbar conjunctiva were observed (Figs. 1 and 2). No other neurocutaneous abnormalities were noticed. Neurological examination revealed normal higher mental functions with no cranial nerve or motor abnormalities. Cerebellar features in the form of limb and truncal ataxia, slow speech and nystagmus were present in addition to slow saccadic movements of eyes. There was loss of proprioception and vibration sense with preservation of other primary modalities of sensation. Investigations including special tests i.e., serum immunoglobulins (IgG and IgA), thyroid function tests and VDRL were normal. Nerve conduction study revealed evidence of demyelination in lower limbs. CT scan of brain demonstrated evidence of cerebellar atrophy.

Ataxia telangiectasia (AT) is an autosomal recessive disorder characterized by progressive cerebellar dysfunction, oculocutaneous telangiectasias, immune deficiency, premature ageing, and predisposition for lymphoreticular malignancies. Cause of AT has recently been attributed to mutation in AT gene (ATM) which has been localized to chromosome 11. The characteristic telangiectatic lesions, which are mainly transversely oriented subpapillary venous plexuses, appear at a later stage than ataxia and are most apparent over bulbar conjunctiva, ears, exposed parts of neck, bridge of nose, cheeks in a butterfly distribution, and in the flexor creases of the forearm. Thus this case is a classical presentation of ataxia - telangiectasia or Louis - Bar syndrome.

A Rohatgi*, V Pardasani**, SK Sharma***, AK Gupta****, A Gurtoo*

*Associate Professor; **Postgraduate; ***Professor; ****Senior Resident, Department of Medicine, Lady Hardinge Medical College and Associated Hospitals, New Delhi - 110 001.

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