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A rare case of childhood ataxia- Ataxia telangectasia

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ATAXIA TELANGIECTASIA Also known as LOUIS BAR SYNDROME or ORPHAN DISEASE is a autosomal recessive disorder has been described as a genome instability syndrome, a DNA repair disorder and a DNA damage response (DDR) syndrome i s the most common ataxia with onset in early childhood .AT is predominantly cerebellar form of Spinocerebellar degeneration which evolves ultimately to involve anterior horn cells, posterior columns and peripheral nerves. The ATM gene 11q 22-23 encodes the protein kinase ATM – recognizes the breaks in DNA , fixes it,In absence of ATM protein, cells with genomic damage are preserved and cause cellular dysfunction, prone for oxidative damage & DNA breakage leads to chromosomal and chromatid breaks s.Cerebellar neurons are highly sensitive .Break points selectively effect chromosome 7 & 14 at sites concerned with Immunoglobulin coding and T cell receptors and with development of infection and neoplasia. characterised by-Progressive neurological impairment,Cerebellarataxia,Variable immunodeficiency with ,susceptibility to sino pulmonary infections,Impaired organ maturation, X Ray hypersensitivity,Ocular and cutaneous telangiectasia,Predisposition to malignancy.We report two cases of 7 year old, female child, born to apparently healthy consanguineous parents with no significant antenatal , perinatal or immediate postnatal complicationscomplaints of Unsteadiness during walking since 2 yr of age when the child began to walk.Fever ,cough & breathlessness since 20 days.

Progressive ataxia while walking, standing still and sitting and associated with abnormal head movements. H/o tremulousness while holding objects and slowness in performing tasks which is progressive feeding difficulties –difficulty in swallowing with drooling of salivaGait and speech disturbances worsening for last2 yrs.Gross motor, Fine motor mile stones delayed with regression of motor milestones .reccurent chest infections .

On examination: Under built and undernourished, short stature,Microcephaly,Bilateral telangiectasia of bulbar conjunctiva on temporal and nasal sides(confirmed by ophthalmologist), Café au lait spots over both arms(only 2)

Examination of CNS: Speech is slow , slurred and scanning typeMotor system:Child sitting in bed stooping forward with head tilt ,Intentional tremor present,Hypertonia -clasp knife spasticity in both lower limbs,DTR- Brisk in lower limbs, normal in upper limbs,Ankle clonus present on both sides,Gait – cerebellar gait. Sensory system- normal .Cerebellar signs: present

Investigations: X- ray B/L Wrist joints- Approx. Bone age is 3-5 yrs,HRCT – Bronchiectasis in both perihilar regions and both lower lobes, MRI Brain- Cerebellar atrophy,Immunoglobulin levels: IgA-0.17g/l (0.7-4.0), Alpha feto protein levels-217.2 ng/ml(<8.1).

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