IMAGES DAY TO DAY

Leukodystrophy of Pelizaeus – Merzbacher's type Presenting as Multiple Neurological Deficits

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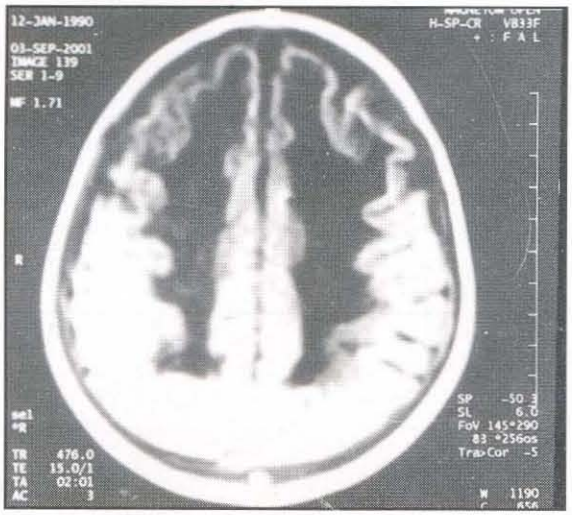

MRI scan of a 11 year old male child showing difuse increase in signal intensity of white matter in fronto-occipito partietal lobes on T2 W image

The term leukodystrophies referes to the dysmyelination in which there is a defect in the formation and maintenance of myelin. Pelizaeus-Merzbacher's disease is a genitically determined familial disease with extensive involvement of myelin, probably resulting from an aplasia of the myelin sheath with fatal course. The disease starts in the infancy and may go on for as long as 30 years. It is predominantly transmitted as sex-linked recessive but other subtypes have been described with possible autosomal dominant or recessive transmission. The clinical features consist of abnormal asymmetrical pendular nystagmus of the eyes, tremors of the head, ataxia, extrapyramidal signs and mental deterioration.

Our patient, an 11 year old boy presented with progressive gait disturbance, behaviour changes and mental retardation of two years duration. The child has markedly low IQ and is to be supported by the parents. He has nystagmus in all gazes and dysarthric speech with nasal twang. His palatal and gag reflaxes are diminished and has occasionally difficulty in swallowing. He has abnormal movements of hands and arms suggestive of choreoathetoid type. There are bilateral pyramidal tract signs in upper and lower limbs. His gait is spastic and ataxic. There is no family history of such illness. His c.s.f. examination shows proteins of 100 mgm% with no cells. There is no aminoaciduria. MRI scan is suggestive of the diagnosis of leukodystrophy of Pelizaeus Merzbacher's type.

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