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Metachromatic Leukodystrophy

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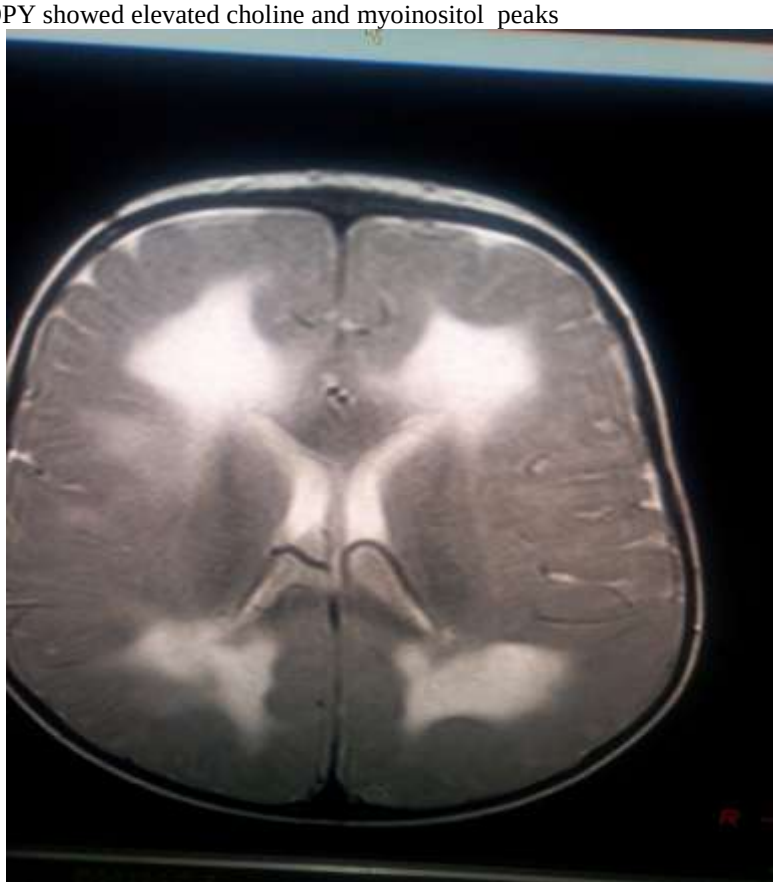
I. Materials And Methods

A one and half year female child by name aishwarya came to the bapuji hospital with chief complaints of spasticity of the limbs,loss of attained milestones,difficulty in standing ,swallowing difficulties and excessive crying

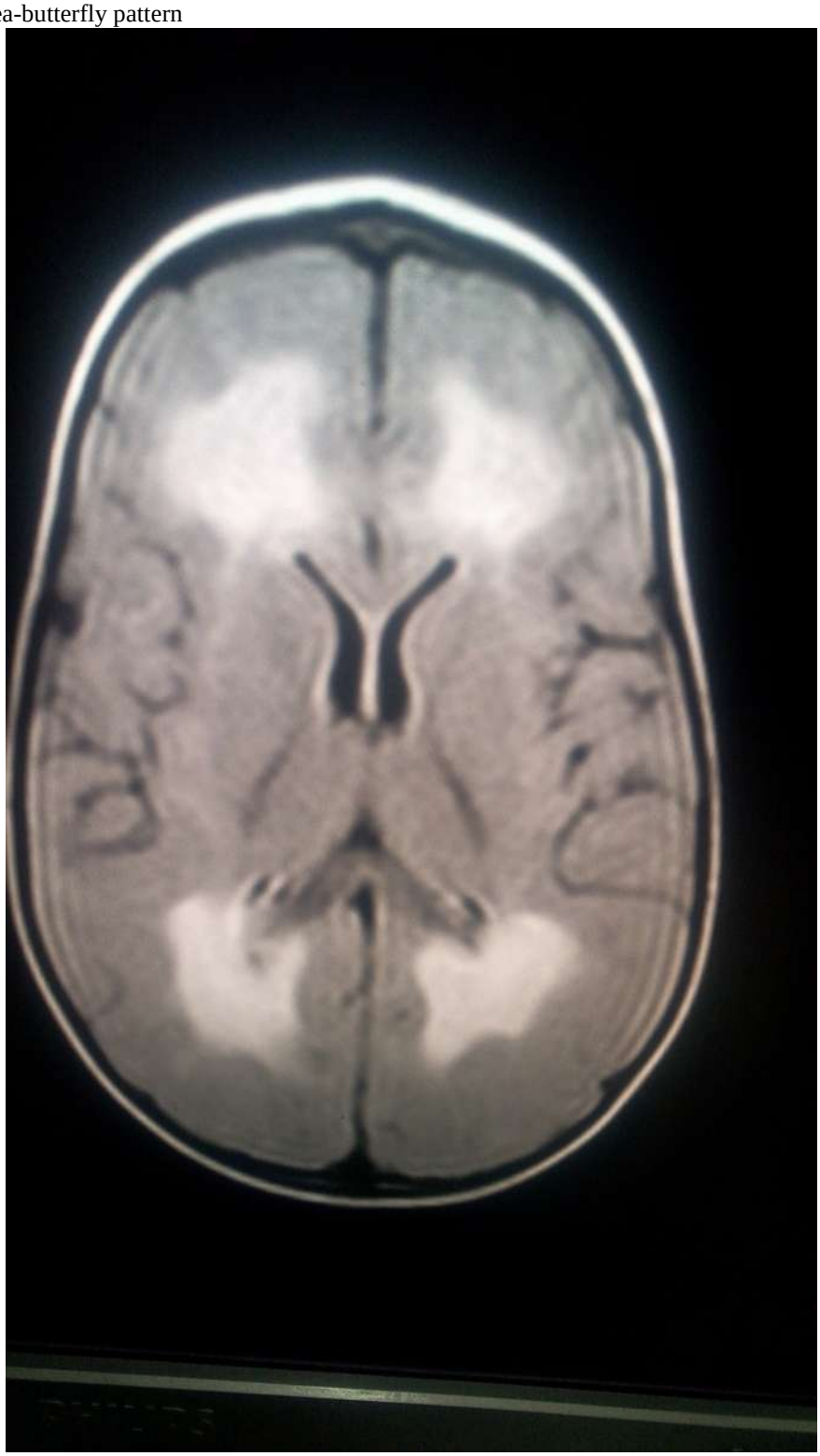
MRI of brain was done in the bapuji hospital radiodiagnosis department using 1.5 tesla Philips machine.mri was done with contrast study

II. Results

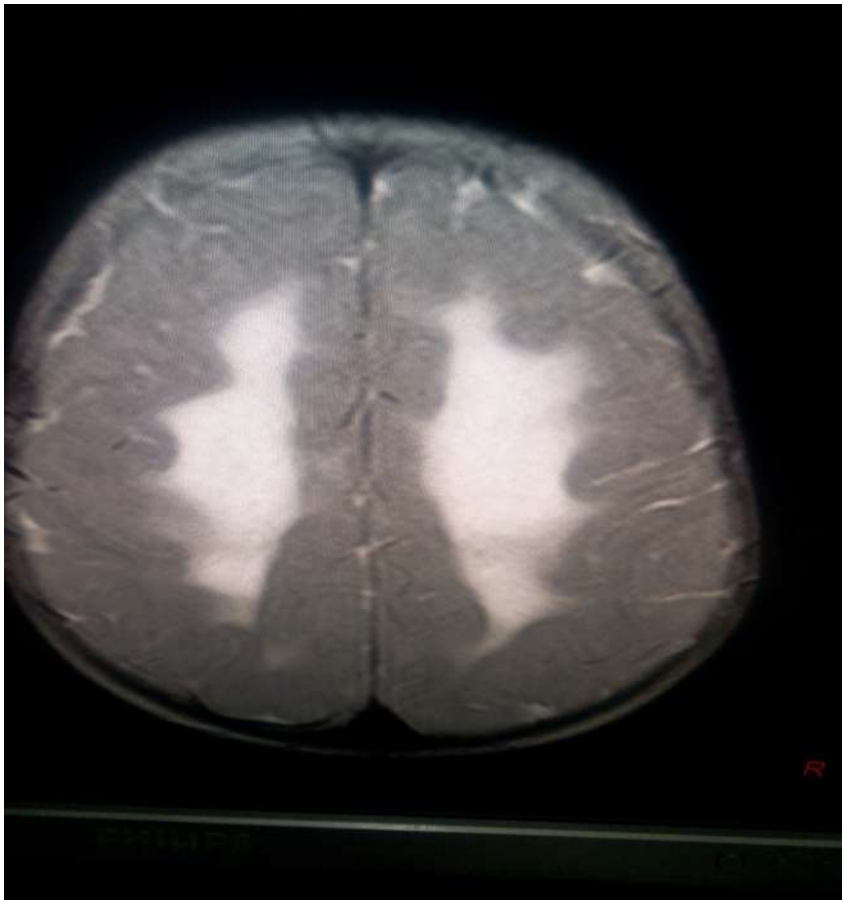
MRI T 2 and FLAIR sequences showed B/L symmetrical hyperintensities in the centrum semiovaleand the confluent symmetrical hyperinstensities around the B/L frontal horns ,peritrigonal areas -(butterfly pattern) which are hypointense on T 1
Rest of the brain parenchyma shows normal signal intensity
Myelination is appropriate for the age
Ventricles,subarachnoid spaces appear normal
Brain stem appears normal in shape,size,outine and signal intensity
Cranial nerves appear normal in signal intensity
Intracranial vesels normal course and outline
Affected areas showed diffusion restriction with corresponding low ADC values
No E/O abnormal enhancement noted on the post contrast study
NO e/o abnormal areas of blooming noted on the gradient echo sequences
MR SPECTROSCOPY showed elevated choline and myoinositol peaks



T2W axial images showing bilateral symmetrical confluent hyperintensities noted around the frontal horns and peritrigonal area-butterfly pattern



Axial T2W FLAIR sequence showing bilateral symmetrical confluent hyperintensities around frontal horns and peritrigonal areas-butterfly pattern



Axial T2W image showing bilateral symmetrical hyperintensities in the centrum-semiovale

III. Conclusion

Metachromatic leukodystrophy (MLD) is the most common hereditary (autosomal recessive) leukodystrophy and is one of the lysosomal storage disorders. It has characteristic imaging features including peri-atrial and to a lesser extent frontal horns leukodystrophy as well as periventricular perivenular sparing results in "tigroid pattern" on fluid sensitive MRI sequences.Markersserum/urinearylsuphatase A levels: reducedClassificationThe disease can sometimes be according to the time of onset:late infantile: most common ~65% (range 50-80%)juvenile (onset between 3-10 years);adult (after age 16)Clinical featureslate infantile form: gait abnormality, muscle rigidity, loss of vision, impaired swallowing, convulsions, dementiajuvenile form: imparied school performance; similar features as in late infantile form but slower progression adult form: psychiatric disorders and dementia; often protracted course over 10 yearsRadiographic features

Mri Brain

Characterised by bilateral symmetrical confluent areas of periventricular deep white matter signal change, in particular around the atria and frontal horns with sparing of subcortical U fibers (see case 1)- butterfly pattern. Progression can lead to cortical and sub cortical atrophy. Which are seen in our case

T1: affected areas are low signal

T1 C+ (Gd)

no enhancement is characteristichowever some cases may show a linear punctate enhancement pattern within lesions 2multiple cranial nerve enhancement has been reported

T2: affected areas are high signal and may show a "tigroid pattern" on axial plane or "leopard pattern" on sagittal plane: sparing along the venules which is seen in our acseMR spectroscopy: (of affected white matter)

Mld spectroscopy shows elevated choline and myoinositol which are seen in our case