Metachromatic Leukodystrophy

Dr Srikanth.S\textsuperscript{1}, Dr Pramod Setty.J \textsuperscript{2}, Dr Kedarnath Dixit\textsuperscript{3}, Dr Pallavi Sinha\textsuperscript{4}, Dr Vijay .V

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 semiovale and the confluent symmetrical hyperintensities 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, outline and signal intensity

Cranial nerves appear normal in signal intensity

Intracranial vessels 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. Markers: serum/urinearylsuphatase A levels: reduced

Classification

The 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 features

late infantile form: gait abnormality, muscle rigidity, loss of vision, impaired swallowing, convulsions, dementia
juvenile form: impaired school performance; similar features as in late infantile form but slower progression
adult form: psychiatric disorders and dementia; often protracted course over 10 years

Radiographic 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 characteristic however 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 case

MR spectroscopy: (of affected white matter) Mld spectroscopy shows elevated choline and myoinositol which are seen in our case