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Maple Syrup Urine Disease: A Case Report

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Abstract: *Maple syrup urine disease is a rare autosomal recessive inborn error of metabolism of branch chain amino acids the condition is named because of distinctive sweet odour of affected infants urine. MSUD is caused by deficiency of branch chain alpha keto acid dehydrogenase enzyme complex, leading to accumulation of the branch chain amino acids (leucine, isoleucine, valine) and their toxic by products(keto acids) in the blood and urine .Imaging is characterised by MSUD affecting myelinated white matter.The typical site of involvement is cerebellar white matter, brainstem, globuspallidus, thalamus, and cerebral peduncles. The disease classically presents in early neonatal period. we report a case of MSUD presenting in a 15 day neonate with classical MRI and Biochemical findings.*

Keywords: *Maple syrup urine Disease, T2 Hyper intensities, cerebellum, brainstem, internal capsule.*

I. Introduction:

Maple syrup urine disease is a autosomal recessive disorder in new born caused by abnormal oxidative decarboxylation of branch chain amino acids, leucine, isoleucine and valine. It causes accumulation of the corresponding ketoacids which results in oxidation of metabolites with a characteristic odour[1]. MSUD is classified into five clinical phenotypes: classic, intermediate, intermittent, thiamine responsive and dihydro lipoyl dehydrogenase deficient forms of which classic type is most common and most severe type which presents in new born period as poor feeding, dystonia, vomitings and seizures. Transcranial neurosonography, CT scan and MRI show charecteristic features of the disease. If not treated earlier, they progress to raised intracranial pressure and die in a few weeks.

II. Case Report:

A 15 day old female baby presented with seizure like activity, tonic posturing, status epilepticus, not accepting feeds with associated excessive cry. There was no history of fever or cough. Routine laboratory investgations were normal. Serum Ammonia was elevated. Serum lactate levels were normal.C reactive protein was positive..Non contrast CT brain was normal . MRI showed bilateral symmetrical T2 hyperintensities in posterior limb of internal capsule, dorsal midbrain, cerebral peduncles. Bilateral symmetrical diffusion restriction noted in centrum semiovale ,the posterior limb of internal capsule, bilateral thalamus, midbrain, pons, cerebellar deep white matter and brain stem. MR Spectroscopy revealed lactate peak at 0.9 ppm [fig 1,2,3,4]. Plasma aminoacid profile revealed glutamic acid-400 umol/L, valine -802 umol/L,isoleucine-290 umol/L ,leucine-1100umol/L confirming the diagnosis as Maple syrup urine disease.
This is the third born child with normal hospital delivery ,second child of the parents expired on 26th day and the child was not investigated previously.

III. Discussion:

MSUD with classical neuro radiological findings are reported in the literature frequently[2,3,4,5]. The severity of the symptoms in the neonates presenting with the features MSUD is related to the duration of acute toxic phase. If it develops in the late part of childhood, the disease present with lethargy, irritability, vomitings. The classical neurosonographic features in neonates in this condition include bilateral symmetrical increased echogenicity of periventricular white matter, thalami, basal ganglia. The CT and MRI reveal typical signs in the form of localised edema in brainstem, cerebellar whitematter , cerebral peduncles, posterior limb of internal capsule, globus pallidus, perirolandic white matter. There may be generalised edema [7]. In MSUD encephalopathy ,there are two types of edema seen in MRI. First is intramyelinic edema and other is vasogenic edema. It has been hypothesised that myelinated areas show hyperintensity on DW images because of intra myelinic edema, whereas unmyelinated areas show hypointensity because of vasogenic interstitial edema. Hence DW imaging is more useful than other sequences as it detects both types of edema. MRS show slight reduction of NAA , slight elevation of lactate, broad peak at 0.9 ppm due to methyl protons[8] .

Janbrismar et al. described the imaging features of MSUD in 12 children and the related changes in the course of the treatment[9]. Wajanat jan et al. found that the abnormal findings on DWI are reversible and

elevated lactate, low NAA levels , elevated branched chain amino acid levels indicate mitochondrial dysfunction during metabolic decompensation[10]. The other condition with similar diffusion imaging changes include canavans disease, non ketotic hyperglycemia. The typical restriction on DWI is due to intramyelinic edema corresponding to areas that are myelinated at birth[11,12].

IV. Conclusion:

Our findings suggest that during the acute phase and early encephalopathic crisis stage of MSUD , DWI can demonstrate the involvement of myelinated white matter in new born and thus can be a valuable diagnostic tool.

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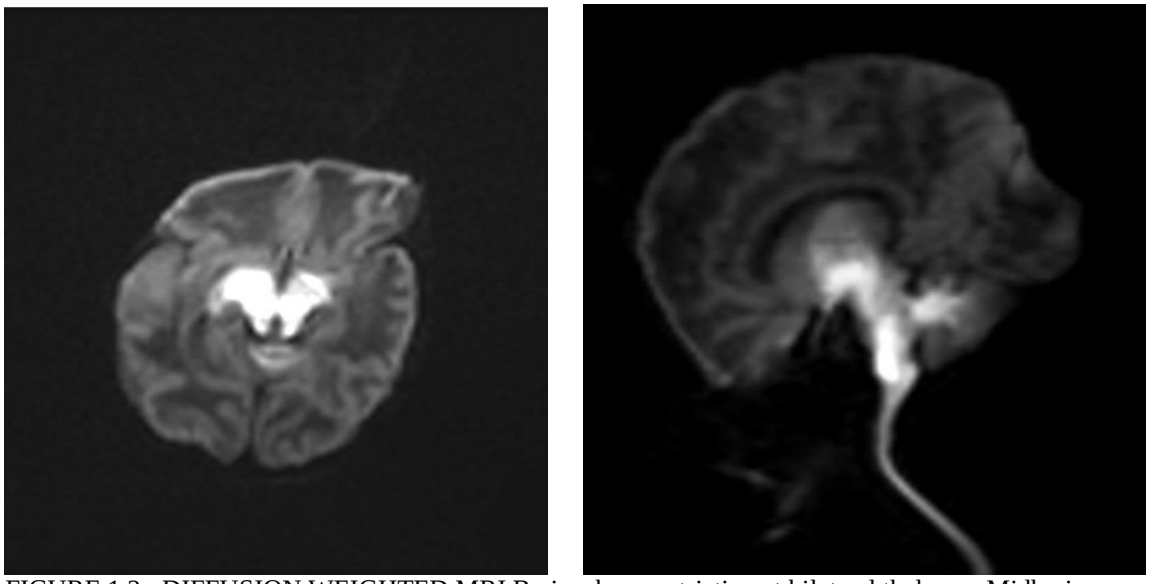

FIGURE 1,2,: DIFFUSION WEIGHTED MRI Brain show restriction at bilateral thalamus,Midbrain,pons ,cerebral peduncles,cerebellar deep white matter.

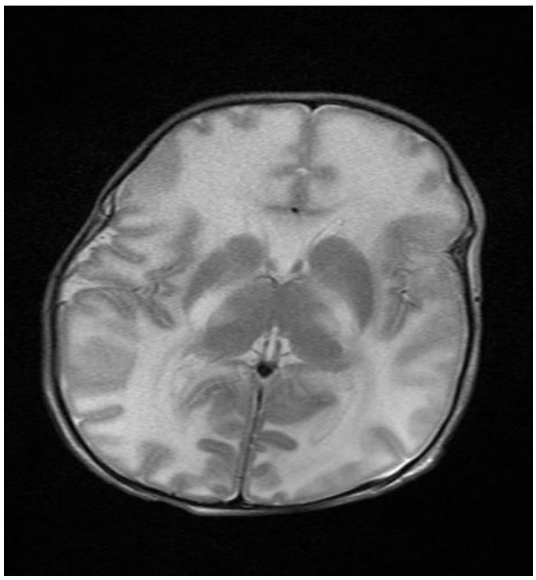

FIGURE 3: T2W axial MR image show hyperintansity at posterior limb of internal capsule..

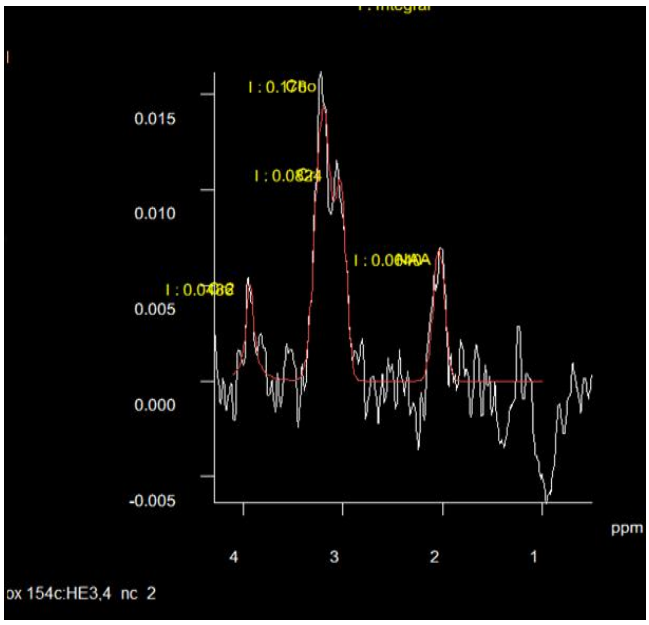

FIGURE 4: MR spectroscopy image show lactate peak at 0.9 ppm.