A rare cause of neonatal metabolic encephalopathy – Maple syrup urine disease
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Section: Paediatric radiology
Area of Interest: Neuroradiology brain
Procedure: Diagnostic procedure
Imaging Technique: MR
Imaging Technique: MR-Diffusion/Perfusion
Special Focus: Metabolic disorders Case Type: Clinical
Cases
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Patient: 6 days, male

Clinical History:
A 6-day-old full term baby born to second degree consanguineous marriage was brought with history of lethargy and poor feeding. There was no history of birth asphyxia. On examination there was hypotonia and absent reflexes. The workup for sepsis was negative. An MRI brain was performed.

Imaging Findings:
Extensive areas of restricted diffusion with associated T2W hyperintense signal with symmetrical distribution was noted involving the medulla, the dorsal pons, bilateral middle cerebellar peduncles, cerebellar white matter, midbrain, cerebral peduncles - especially the corticospinal tracts, both thalami, optic radiation, posterior limb of internal capsule extending superiorly into the corona radiata and up to the high parietal cortex. Mild diffuse white matter oedema was seen in both cerebral hemispheres. The abnormal signal intensity in the medulla was also noted to extend into the cervical spinal cord. The above features were consistent with Maple Syrup Urine Disease (MSUD).

Discussion:
MSUD, first described by Menkes et al, is a rare autosomal recessive disorder of branched chain amino acid (BCCA) metabolism caused by deficiency of the α-ketoacid dehydrogenase complex, leading to accumulation of the branched chain amino acids (leucine, isoleucine, and valine) and their products (ketoacids) in the blood and urine, giving the characteristic maple syrup odour [1]. Four forms have been described: Classic, intermediate, intermittent, and thiamine-responsive.
As in our case, the affected neonates are usually normal at birth and develop lethargy, poor feeding and vomiting by day 6 to 7. No maple syrup odour was detected in the urine in our case as it has been described that it might take nearly a week to identify in the urine [2]. Elevated levels of leucine have been reported to cause neurological manifestations.
The best diagnostic clue on MRI is the presence of marked diffusion restriction in the deep cerebellar white matter, cerebral peduncles, dorsal brain stem, globus pallidus, thalamus and corticospinal tracts reflecting cytotoxic oedema in the myelinated white matter [3]. The increased signal on diffusion-weighted imaging has been found to be due to vacuolating myelinopathy where there is trapping of water molecules in vacuoles between the myelin sheet layers, leading to isotropically restricted water diffusion. Recent literature suggests the phenomenal role of diffusion...
weighted MRI in earliest detection of this classical MSUD oedema [4, 5].
Following MRI, a Tandem mass spectrosopy was done which showed elevated valine, leucine and isoleucine. Plasma amino acid levels were also found to be significantly elevated. The baby was started on special formula feeds (BCCA free), following which there was improvement in the tone with sluggish deep tendon reflexes.

Our case highlights the usefulness of MRI, especially DWI, in a neonate presenting with features of encephalopathy. Earlier diagnosis is essential as MSUD has a favourable outcome with proper dietary control and treatment measures. Prompt treatment will prevent complications like seizures, coma and death [6, 7].

**Differential Diagnosis List:** Maple syrup urine disease, Hypoxic ischaemic encephalopathy, Mitochondrial mutation disorders

**Final Diagnosis:** Maple syrup urine disease

**References:**


Description: Axial diffusion-weighted MR image showing hyperintense signal in deep cerebellar nuclei and pons. Origin: Department of Radiology and Imaging sciences, Billroth hospitals, Chennai, India.
Figure 2

Description: Coronal reformatted diffusion-weighted MR image showing hyperintense signal in bilateral corticospinal tracts, cerebral peduncles and brain stem. Origin: Department of Radiology and Imaging sciences, Billroth hospitals, Chennai, India.
Description: Axial diffusion-weighted MR image showing hyperintense signal in bilateral thalami.
Origin: Department of Radiology and Imaging sciences, Billroth hospitals, Chennai, India.
Description: Apparent diffusion coefficient (ADC) map shows restricted diffusion in the pons and deep cerebellar white matter. Origin: Department of Radiology and Imaging sciences, Billroth hospitals, Chennai, India.
Description: Apparent diffusion coefficient (ADC) showing restricted diffusion in bilateral posterior limb of internal capsule and globi pallidi. Origin: Department of Radiology and Imaging sciences, Billroth hospitals, Chennai, India.
Description: Axial T2W image showing hyperintense signal in pons, deep cerebellar nuclei and middle cerebellar peduncle. Origin: Department of Radiology and Imaging sciences, Billroth hospitals, Chennai, India.