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CASE REPORT

CASE REPORT OF NEURORADIOLOGICAL IMAGING - MAPLE SYRUP URINE DISEASE

***Masudi Sheetal, Vishal, S., Nimbal, Satish Patil, Praveen M. M. and Saurabh Rathi**

Department of Radiology, Shri B M. Patil Medical College, Bijapur Karnataka, India

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ABSTRACT

Maple Syrup Urine Disease (MSUD) is a rare inherited autosomal recessive disorder caused by a deficiency of the branched-chain alpha-keto acid dehydrogenase complex (BCKDC), leading to a buildup of the branched-chain amino acids (leucine, isoleucine, and valine) and their toxic by-products (ketoacids) in the blood and urine. It can present with life-threatening cerebral edema and dysmyelination in affected individuals. Imaging is characterestized by MSUD oedema affecting the myelinated white matter. We report diffusion-weighted imaging (DWI) findings in a newborn child with MSUD who presented with acute metabolic encephalopathic crisis.

INTRODUCTION

Maple syrup urine disease (MSUD) is a metabolic error involving catabolic pathway of the branched-chain amino acids(BCAA) leading to build up of ketoacids, which give rise to the classic 'maple syrup' or burnt sugar smell. MSUD affects an estimated 1 in 185,000 infants worldwide. It is divided into four major categories: (Chuang and Shih, 1995) Classic, (Zinnanti *et al.,* 2009) intermediate, (Naughten *et al.,* 1982) intermittent, and (Mascalchi *et al.,* 2005) thiamine responsive, which carry differing symptoms and prognostic factors (Chuang and Shih, 1995). The exact cause for brain injury is not clearly understood. According to a study by Zinnanti *et al*. they suggest two converging mechanisms of brain injury in MSUD including:

(i) Neurotransmitter deficiencies and growth restriction associated with BCAA accumulation and (ii) energy deprivation through Krebs cycle disruption associated with branched-chain ketoacid accumulation. This disease leads to accumulation of BCAA and metabolites (neurotoxic). The most common and severe form of the disease is the classic type, which becomes apparent soon after birth. Beginning in early infancy, this condition is characterized by poor feeding, vomiting, lethargy and developmental delay. If untreated, MSUD can lead to seizures, coma, and death (Naughten *et al.,* 1982).

****Corresponding author: Masudi Sheetal,***
Department of radiology, Shri B M patil medical college, Bijapur Karnataka, India.

Case Report

A 17-day-old male baby, born of primigravida mother of Indian origin through second degree consanguineous marriage was well for 2 days after birth and then developed marked lethargy, progressing to seizures and coma. Antenatal period was uneventful with a normal vaginal delivery.

At referral to our hospital, baby presented with acute metabolic encephalopathic crisis, unresponsive to pain except for tonic movements of upper limbs. Markedly boggy anterior fontanelle found and rest of clinical examination was normal (no significant skin/urine odour included). Cerebrospinal fluid (CSF) analysis was normal. MRI showed restricted diffusion involving bilateral cerebral hemispheres, cerebellar white matter, entire brain stem, bilateral thalami, internal capsules and corticospinal tracts (Figures 1).

Subtle T2 FLAIR hyperintensity is seen in the above regions (Figure 2). Small cephalhematoma was seen in right fronto-parietal regions. Cavum septum pellucidum was seen. Aminoaciduria was suggested as the possible diagnosis with a suggestion to rule out MSUD. Tandem mass spectroscopy showed increased levels of leucine and isoleucine (=1787 nmol/ml; ref < 350) but normal valine. Urine gas chromatography-mass spectrometry (GC-MS) confirmed MSUD. The patient was treated with dietary modification and measures to reduce cerebral edema.

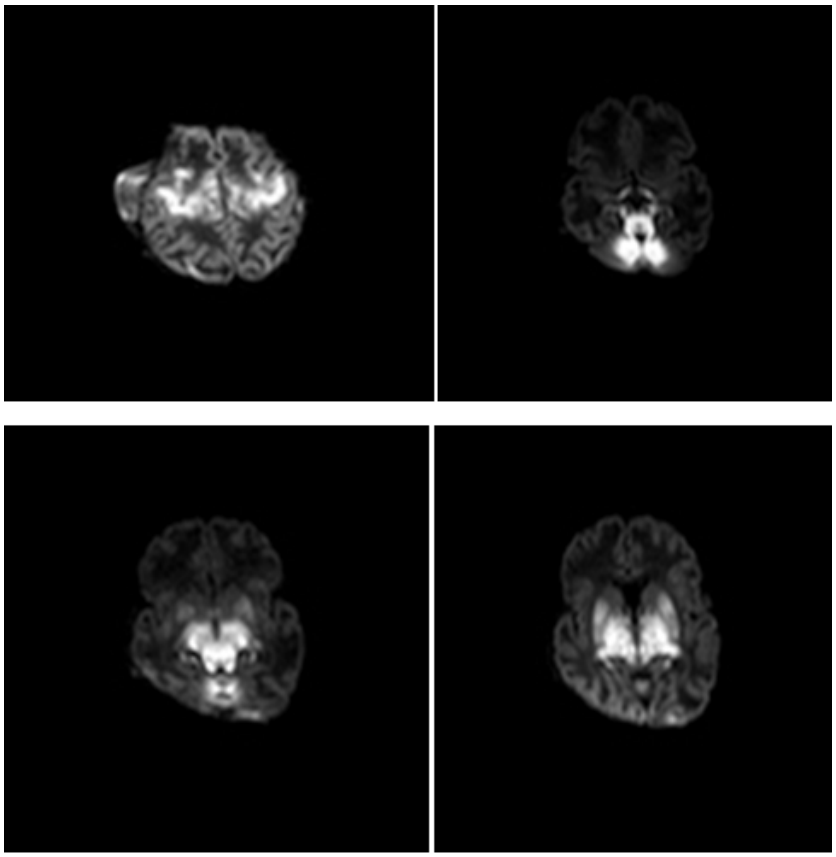

Fig. 1. DWI images show areas of restriction diffusion in bilateral cerebral hemispheres, cerebellum white matter, brain stem, thalami and basal ganglia

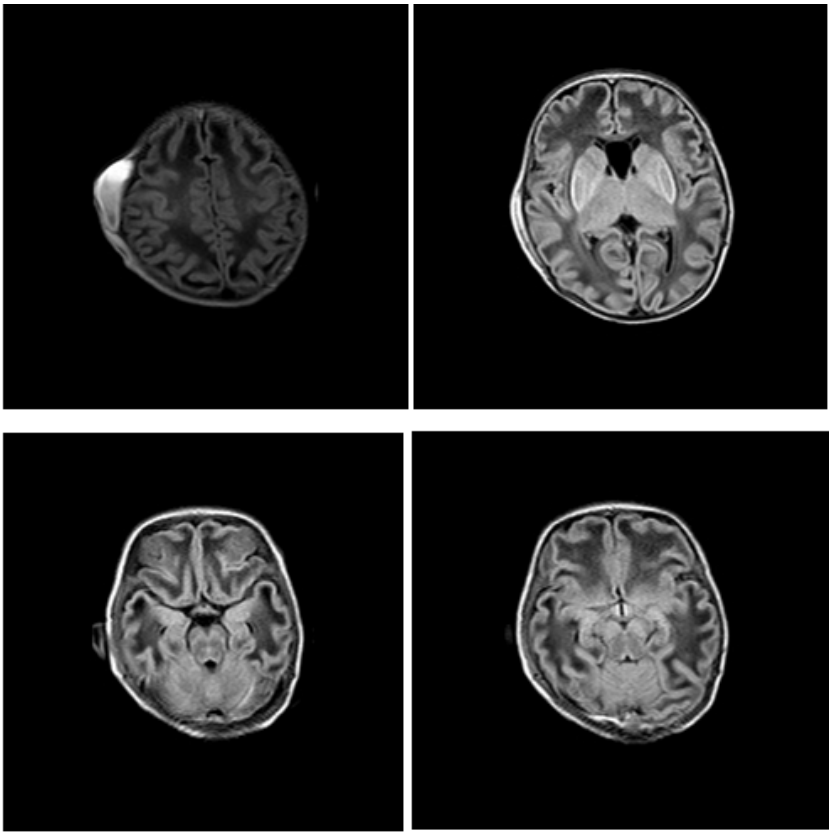

Fig. 2. Subtle T2 FLAIR hyperintensity is noted in bilateral cerebral hemispheres, cerebellum white matter, brain stem, thalami and basal ganglia. Hyperintense lesion seen in right fronto-parietal region of scalp with blooming on GRE - cephalhematoma

DISCUSSION

Diffusion weighted magnetic resonance (MR) has an important role in the assessment of brain maturation and of white matter diseases in the fetus, neonate and the child. Diffusion MR imaging enables a better characterization of the lesions demonstrated by conventional MR imaging, in hypoxic‐ischemic encephalopathy, infections, inherited metabolic diseases, etc. and is particularly important for the longitudinal evaluation of these conditions (Mascalchi *et al.,* 2005). The areas of restricted diffusion represent cytotoxic edema and damaged oligodendro‐axonal units within the affected white matter (Parmar *et al.,* 2004). Generalized diffuse edema, though not characteristic is known to occur in MSUD with localized, more severe edema involving the deep cerebellar white matter, the dorsal part of the brainstem, the cerebral peduncles, and the dorsal limb of the internal capsule (Brismar *et al.,* 1990; Jan *et al.,* 2003). Our patient showed diffuse restricted diffusion involving bilateral cerebellar white matter, entire brain stem, bilateral thalami, internal capsules and corticospinal tracts. Presence of such finding points towards aminoacidurias, including MSUD, in spite of the absence of maple syrup odor. Differential diagnoses for similar pattern of restricted diffusion include nonketotic hyperglycinemia and Canavan disease (Bindu *et al.,* 2010). Lab investigations (tandem mass spectroscopy for blood levels of amino acids and Urine GC‐MS for urine levels of amino acids) are confirmatory for the underlying disease. Though MSUD is life threatening as in our case, earlier and prompt treatment of MSUD would help reverse the decompensated state and prevent associated mortality and brain damage in surviving patients. Follow‐up MRI can reflect the reversal of findings in appropriately treated cases (Sener, 2007). Prompt recognition of the pattern of diffuse restricted diffusion involving bilateral cerebellar white matter, entire brain stem, bilateral thalami, internal capsules and corticospinal tracts and raising the suspicion of aminoacidurias, is essential in curtailing the morbidity and mortality of the affected patients.

Conclusion

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

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