MR Imaging of Brain in Maple Syrup Urine Disease: a case report

MIN-SZU YAO1 YUH-FENG TSAI1 LIANG-KONG CHEN2 CHENG-TAU SU2 WING P. CHAN1,3

Department of Radiology1, Taipei Medical University-Municipal Wan Fang Hospital
Department of Diagnostic Radiology2, Shin Kong Wu Ho-Su Memorial Hospital
Department of Radiology1, School of Medicine, Taipei Medical University

Maple syrup urine disease (MSUD) is a rare inherited metabolic disorder due to decreased decarboxylation of branched-chain amino acids [1]. Accumulation of branched-chain amino acids and their related compounds leads to characteristic maple syrup urine odor and severe neurological deterioration. Only two cases with diffusion-weighted MR imaging (DWI) have been reported in the literatures [2,3]. We herein report the third case of MSUD with DWI study.

CASE REPORT

A newborn boy was delivered with a birth weight of 2500 gm. His mother had no complication during pregnancy. The baby was regularly fed with milk formula. At his eighth day of life, poor feeding, lethargy, respiratory distress, and seizure-like attacks became evident. He was then admitted for medical care in our hospital. Routine laboratory tests revealed compensatory metabolic acidosis and high blood ammonia level (103 \( \mu \)g/dl). Urine analysis was not abnormal. Electroencephalogram showed poorly organized activity with areas of suppression in the brainstem.

Magnetic resonance (MR) imaging (1.5-T, Symphony, Siemens, German) of the brain disclosed hyperintensities at the dorsal aspect of the brainstem, corticospinal tracts, deep cerebellar white matter, thalami, and posterior limbs of the internal capsule on both T2-weighted imaging (T2WI) (TR=2500ms, TE=90ms) and DWI (TR=5300ms, TE=135ms) (Fig. 1). Delayed myelination of white matter or metabolic disorder was highly suspected. Laboratory analysis of plasma amino acids revealed a typical chromatographic pattern of MSUD. However, the MR software of apparent diffusion coefficient (ADC) map was not available during the examination.

Thereafter, the newborn patient was fed with milk composed of MSUD formula (i.e. restriction of branched-chain amino acids). After the dietary treatment, the infant achieved clinical and neurological improvement. The baby was discharged at age 65 days.
Menkes et al. [1] first described MSUD in 1954. It is an inherited metabolic disorder due to decreased decarboxylation of branched-chain keto acids triggering an accumulation of leucine, isoleucine, and valine. The prevalence of MSUD is 1:224000 [4]. In classic form, the patients usually present acutely, with poor feeding, vomiting, lethargy, seizures, and even coma in first day of life. Neuropathologic changes may occur when there are increased brain water content, reduced number of oligodendrocytes and astrocytes, and deficient myelination with spongy degeneration of white matter and basal ganglia [5,6].

CT scan for cases of MSUD frequently revealed negative findings during the first few days, and then marked, generalized, diffuse brain edema appeared [7-10]. The edema remained as long as 6 or 7 weeks in untreated patients. Ultrasonography showed symmetric increase in echogenicity of periventricular white matter, basal ganglia (mainly pallidi), and thalami in acute stage of the disease [11].

In our case, conventional MR imaging enabled us to detect hyperintense brain edema, also referred to as MSUD edema, but failed to assess the entire extent of the disease. DWI enhanced depiction of the MSUD edema in the centrum semiovales, which was not shown on T2WI. It has been reported that DWI is more sensitive than spin-echo T2WI in assessing MSUD edema [2, 3]. The ADC values of these areas are markedly decreased. It was suggested that MSUD edema was

**DISCUSSION**

Figure 1. Axial T2W (a, b, c) and diffusion-weighted (d, e, f) images show typical location of MSUD edema. T2W images showing hyperintense signal change in a. dorsal aspect of brainstem (arrows), b. posterior limbs of internal capsule (white arrows) and posterior aspects of thalami (black arrows). c. No demonstrable abnormal signal at level of parietal lobe. DWI showing more diffuse change of hyperintensity in d. the entire brain stem, e. the thalami than that seen on T2W images. f. Hyperintensities in the centrum semiovales, which are not apparent on T2W imaging (in c).
cytotoxic edema rather than vasogenic edema [2] and the resulting dysmyelination was a reflection of disorganized tissue integrity [3].

The lack of ADC maps and follow-up DWI after treatment are the major limitations of this report. However, we report the third case of MSUD with DWI to show the sensitivity of this technique to determine extent of the disease. The infant received early dietary treatment and achieved a good clinical outcome.

REFERENCES

楓糖漿尿症在磁振造影之表現：病例報告

姚敏思¹ 蔡裕豐² 陳良光² 蘇誠道² 陳榮邦¹²

臺北醫學大學市立萬芳醫院 放射診斷科¹
新光吳火獅紀念醫院 放射診斷科²
臺北醫學大學 醫學系 放射線學科³

楓糖漿尿症是一種隱性正染色體代謝失調的病變，發生在新生兒因缺乏某種分解胺基酸之酵素所致的疾病。我們報告一例新生兒罹患此病，由於磁振造影DWI影像比傳統T2W影像對偵測腦水腫之範圍更具靈敏性，該患者得以早期診斷與治療且預後良好。

關鍵詞：腦部水腫，擴散係數影像，磁振造影，楓糖漿尿症