

CLINICAL IMAGE

Maple Syrup Urine Disease (MSUD)

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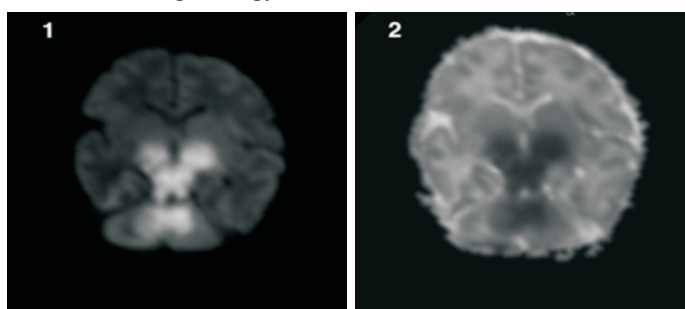
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A 2 month male infant presented with poor feeding, vomiting, poor weight gain, and increasing lethargy after birth. Laboratory testing revealed metabolic acidosis. The serum tandem mass spectrometry showed elevated plasma levels of branched chain amino acids (leucine, isoleucine, and valine). MRI revealed restricted diffusion involving the corticospinal tracts, internal capsule, thalami, globus palladi and brainstem (Figure 1 and 2). Based on these findings diagnosis of maple syrup urine disease (MSUD) was made.

Bilateral symmetrical restricted diffusion on axial diffusion weighted magnetic resonance images (Figure 1) with corresponding low apparent diffusion coefficient value (Figure 2) involving the cerebellar white matter, dorsal brainstem, cerebral peduncles and posterior limb of the internal capsule.

MSUD also known as branched-chain ketoaciduria (BCKA), is a rare autosomal-recessive disorder of branched-chain amino acid metabolism that is caused by deficiency of the branched-chain α -ketoacid dehydrogenase (BCKD) complex.¹ The initial symptoms of affected infants are complex and lack specific characteristics; therefore, the disease is often misdiagnosed. However, the disease progresses to mental retardation, spastic paralysis, cortical blindness, and eventually death.² MRI findings are diagnostic in the early weeks of life; the hyperintensity on DWI mainly involves the globus pallidus, thalamus, internal capsule, brainstem, and cerebellar white matter, which represent the typical myelinated areas in normal full-term neonates.³ Because early detection and timely treatment are very helpful for the prognosis of MSUD

patients, MRI examination should be performed in neonates who manifest poor feeding, vomiting, poor weight gain, and increasing lethargy after birth.



Conflict of Interest: None

Funding: None

Written informed consent: Taken

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How to cite this article: Shah A, Jabeen S, Shah O, Shaheen F, Gojwari T. Maple Syrup Urine Disease (MSUD). *JMS* 2022; 25 (2) 39

Received: 2022-04-27 Accepted: 2022-05-26

Access this article online



Website:
www.jmsskims.org

DOI:
<https://doi.org/10.33883/jms.v25i2.1189>



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