

Teaching NeuroImage: Optic Pathway Involvement in Maple Syrup Urine Disease

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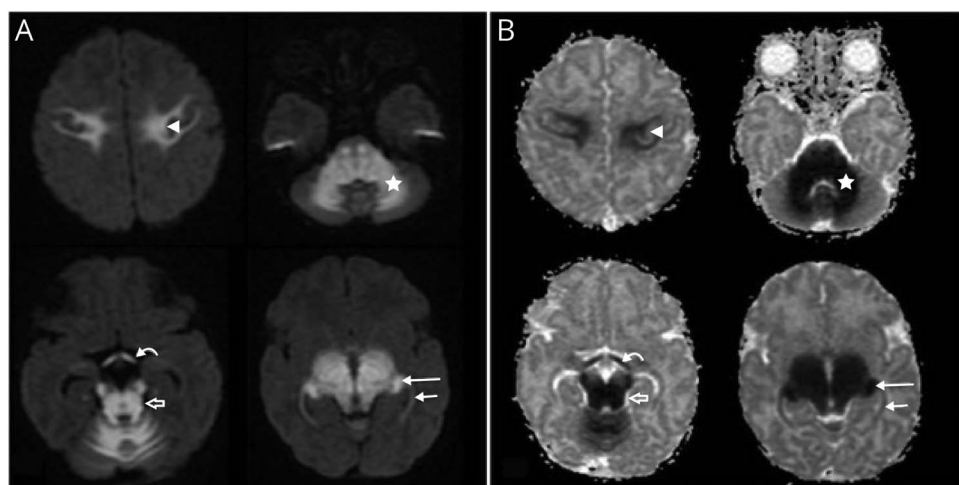
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A 10-day-old neonate presented with vomiting and irritability. Diffusion-weighted imaging showed abnormal signal intensities, including characteristic involvement of the optic pathway (Figure, A and B). Maple syrup urine disease (MSUD) was suspected. Blood tests confirmed elevated levels of branched-chain amino acids (BCAAs)—leucine, isoleucine, and valine.

Optic pathway signal abnormalities occur in 26.3% of patients with MSUD and may cause cortical visual impairment and transient blindness.¹ These abnormalities are caused by a deficiency of alpha-ketoacid dehydrogenase, elevated BCAAs, and reduced (Na⁺/K⁺ATPase) pump function resulting in water accumulation between the myelin layers (intramyelinic edema); however, they are reversible with treatment and over time.²

Figure Diffusion-Weighted MRI of a 10-Day-Old Neonate With MSUD



(A) Diffusion-weighted MRIs and (B) corresponding ADC images demonstrating diffusion restriction with corresponding reduced ADC in bilateral optic tracts (curved arrows), lateral geniculate bodies (long arrows), optic radiations (short arrows), bilateral perirolandic white matter (arrowheads), brainstem (open arrows), and cerebellar white matter (asterisks). ADC = apparent diffusion coefficient; MSUD = maple syrup urine disease.

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| Name | Location | Contribution |
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| Rama Krishna Narra, MD, DNB, DM | Department of Neuroradiology, Katuri Medical College and Hospital, Guntur, India | Drafting/revision of the manuscript for content, including medical writing for content; major role in the acquisition of data; study concept or design; analysis or interpretation of data |

Appendix (continued)

| Name | Location | Contribution |
|---------------------------------------|---|--|
| Rajendra Kumar Pamidi, MD, DNB | Department of Neurology, Katuri Medical College and Hospital, Guntur, India | Major role in the acquisition of data; study concept or design |
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