



Figure. (A) Axial T2-weighted image at age 18 months shows slightly swollen, diffusely hyperintense medulla and cerebellar dentate nuclei. (B) Axial T2-weighted image at age 3 years shows normal findings. (C) Axial T2-weighted image at age 6 years shows mild hyperintensity of the outer aspect of the medulla and dentate nuclei. (D) Axial diffusion-weighted image at age 6 years shows high signal in the same areas.

## MRI in acute intermittent maple syrup urine disease

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An 18-month-old girl presented with a 3-day lasting episode of ataxia and lethargy. Following spontaneous recovery, neurologic examination, MRI, amino acids, and organic acids were normal. Five years later, a second episode occurred. During the acute decompensation, branched chain ketoaciduria and increased plasma branched chain amino acids were depicted, allowing the diagnosis of maple syrup urine disease (MSUD). After recovery

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clinical, neuroradiological and biochemical examinations became normal again.

Acute intermittent MSUD may present in childhood, adolescence, or adulthood, and between attacks the patients are entirely normal. MRI is specific (figure).<sup>1</sup> In particular, high signal abnormalities shown by diffusion-weighted images (figure, D) are related to cytotoxic or intramyelinic edema caused by a deficit of  $\text{Na}^+/\text{K}^+$  ATPase activity as a result of impairment in energy production secondary to branched chain amino acids accumulation. These changes must lead to prompt biochemical investigations that are diagnostic, only analyzing biologic material collected during acute metabolic decompensation.

1. Jan W, Zimmermann RA, Wang ZJ, Berry GT, Kaplan PB, Kaye EM. MR diffusion imaging and MR spectroscopy of maple syrup urine disease during acute metabolic decompensation. *Neuroradiology* 2003;45:393-399.

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