Metachromatic Leukodystrophy: Diffusion MR Imaging Findings

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Case Report

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Summary: Herein the case of a 10-month-old boy with metachromatic leukodystrophy is reported. Diffusion MR imaging performed with an echo-planar trace sequence revealed a cytotoxic edema-like pattern (high signal intensity on \( b = 1000 \, \text{s/mm}^2 \) images and low apparent diffusion coefficient values) in the affected white matter in the absence of an ischemic condition. This finding was unchanged at a 6-month follow-up, as revealed by diffusion MR imaging. A gradient-echo diffusion sequence, reverse fast imaging in steady-state precession, revealed hyperintense changes at the corresponding regions. It is likely that the cytotoxic edematous pattern (restricted diffusion pattern) reflected restriction of mobility of the water molecules within abnormal portions of the myelin sheath, because impaired myelin breakdown and reutilization are known features of metachromatic leukodystrophy.

Metachromatic leukodystrophy is caused by a deficiency of arylsulfatase-A resulting in impaired myelination (1–8). Diffusion MR imaging features of the condition have not been reported previously. We report diffusion MR imaging of metachromatic leukodystrophy performed by using the echo-planar and gradient-echo sequences.

Case Report

The patient was a 10-month-old boy presenting with progressive spasticity. An MR imaging examination was performed on a 1.5-T MR unit (Magnetom Vision; Siemens, Erlangen, Germany) with a gradient strength of 30 mT/m. T2-weighted images revealed hyperintense changes, mainly involving the deep white matter, and atrophy (Fig 1A). Diffusion MR images (Fig 2A) strongly suggested that the lesion was restricted diffusion (restricted diffusion pattern). The distribution of the lesions (Fig 1B) and their unchanged pattern on 6-month follow-up diffusion MR images (Fig 2A) strongly suggested that the lesions were directly related to the disease process. Restricted diffusion was manifested by hyperintense changes on heavily diffusion-weighted (\( b = 1000 \, \text{s/mm}^2 \)) images (Fig 1B) and low ADC values (eg, 0.55 \( \times 10^{-3} \, \text{mm}^2/\text{s} \)) on ADC maps (Fig 1C) of the echo-planar trace sequence. In addition, on the gradient-echo diffusion-weighted images, obtained using reverse fast imaging in steady-state precession, high signal intensity and high pixel values were evident in the deep white matter (Fig 2B) and were similar to those seen on \( b = 1000 \, \text{s/mm}^2 \) images, which again indicated definite abnormality in these regions. However, absolute ADC value calculation is not possible with the reverse fast imaging in steady-state precession sequence.

A similar diffusion MR imaging pattern on echo-planar diffusion images of patients with phenylketonuria was recently reported by Phillips et al (9). In that study, patients with phenylketonuria had highly restricted diffusion of water molecules with high signal intensity of the white matter on \( b = 1000 \, \text{s/mm}^2 \) images in association with low ADC values on ADC maps (between 0.56 and 0.63 \( \times 10^{-3} \, \text{mm}^2/\text{s} \) (9). The authors concluded that this restricted diffusion pattern in cases of phenylketonuria reflected impaired myelination, and protons within partially that de-
stroyed portions of the myelin sheath may not be as mobile as those in normal parenchyma (9). On the other hand, in a recent experiment, Branco (10) showed that transition of water from the solid to the gel state contributes to the signal intensity on diffusion MR images, and in the gel state, high signal intensity is observed on $b = 1000 \text{s/mm}^2$/images (this should result in low ADC values). Considering the observations by Phillips et al (9) and the experiment by Branco (10), in the current case of metachromatic leukodystrophy, the restricted diffusion pattern probably was consistent with some disintegration of the white matter secondary to impaired myelination, and restriction of mobility of the water molecules within abnormal portions of myelin sheath resulted in a diffusion MR imaging pattern similar to that of cytotoxic edema.

**Conclusion**

In a case of metachromatic leukodystrophy, an echo-planar trace sequence revealed a cytotoxic edematous pattern (restricted diffusion pattern) in the deep white matter in the absence of an ischemic condition. This pattern, manifested by high signal intensity on $b = 1000 \text{s/mm}^2$/images and low ADC values, probably reflected restriction of mobility of the water molecules within abnormal portions of the myelin sheath, considering that impaired myelin breakdown and reutilization are known features of metachromatic leukodystrophy. Hence, dysmyelination is a feature of metachromatic leukodystrophy. A gradient-echo diffusion sequence, reverse fast imaging in steady-state precession, revealed high signal intensity and high pixel values (165) in the deep white matter, compared with the peripheral regions of the parenchyma (99).

**References**

5. Johansson P, Ehlers L, Hansen HJ. Dementia with impaired tem-