X-linked Charcot-Marie-Tooth Disease with Transient Splenium Lesion on MRI

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Figure 1. Axial slice of cerebral MRI at the level of the splenium. Neither T1-weighted nor gadolinium-enhanced T1-weighted images showed any abnormal intensity (A,B). Fluid-attenuated inversion recovery (FLAIR) and diffusion-weighted (DW) images showed high signal intensity in the splenium (C,D). Follow-up MRI (two months later) showed reduced signal intensity of the splenium on FLAIR and DW images without any morphological change (E,F).

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A 40-year-old man presented with slowly progressive weakness and muscle atrophy of the distal extremities with pes cavus, stork leg appearance, mild sensory disturbance of feet and areflexia. Other neurological examination findings were normal including the higher cerebral function. Blood test and cerebrospinal fluid were normal. Nerve conduction study sup-

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ported the presence of demyelinating sensori-motor polyneuropathy. He had never been exposed to toxic materials or drugs. He was diagnosed as having X-linked Charcot-Marie-Tooth disease (CMTX) according to gene analysis demonstrating non-sense mutation at codon 157 of connexin 32 gene. Brain MRI was performed, since CMTX has been reported to cause symptomatic or asymptomatic cerebral lesions (1-6). The diffusion-weighted and fluid-attenuated inversion recovery images disclosed an abnormal high signal intensity in the splenium without contrast enhancement (Fig. 1A, B, C, D). The abnormal intensity diminished two months later (Fig. 1E, F). Acute disseminated encephalomyelopathy, mitochondrial disorder and adrenoleukodystrophy must be considered in the differential diagnosis (5-7).

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