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【 PICTURES IN CLINICAL MEDICINE 】

Cerebrotendinous Xanthomatosis with Nodular-hypertrophy of the Lumbosacral Roots

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Key words: cerebrotendinuous xanthomatosis, sterol 27-hydroxylase (*CYP27*) gene, root hypertrophy, magnetic resonance neurography

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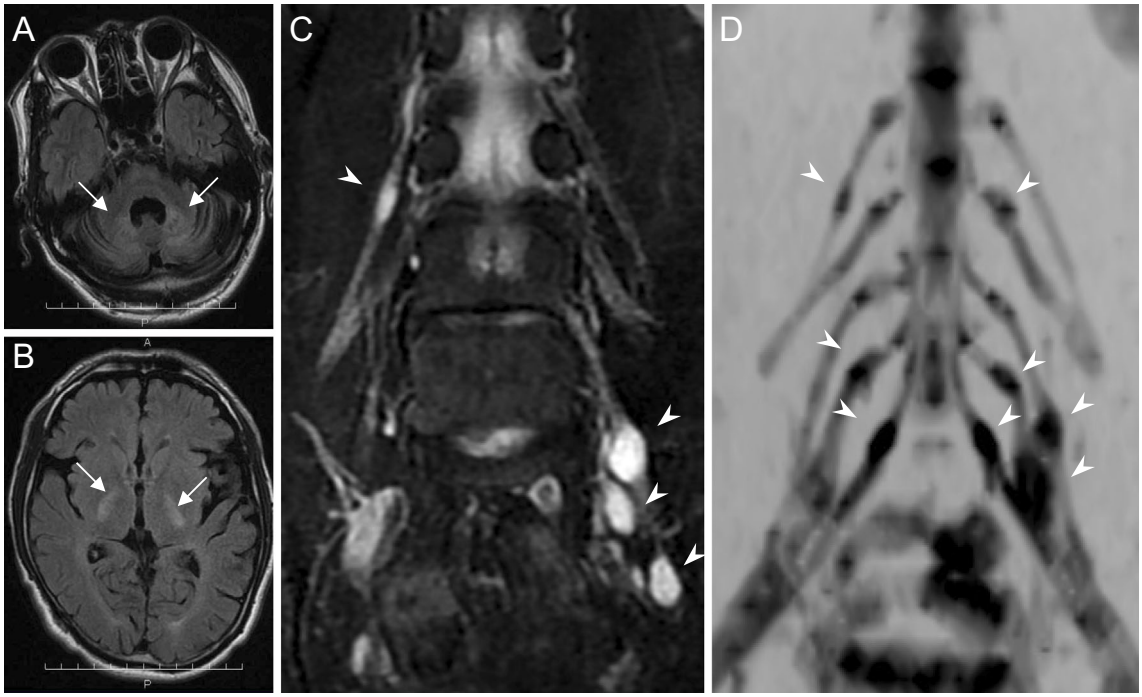

Picture.

A 39-year-old man with cerebrotendinous xanthomatosis (CTX) had a history of mental retardation (from age 4), cerebellar ataxia and spastic paraparesis (age 33), and cataracts (age 39). The cholestanol level was elevated in the serum (49.6 μg/mL, normal 2.35±0.73 μg/mL). A genetic analysis revealed c.1421G>A homozygous type in the sterol 27-hydroxylase (*CYP27*) gene (1). Brain magnetic resonance imaging (MRI) showed cerebral and cerebellar atrophy with signal abnormality around the dentate nucleus and pyramidal tract (arrows Picture A and B; T1-weighted fluid-attenuated recovery sequences). The lesions demonstrated a high signal on T1- and T2-weighted images. Multiple nodular thickening of the nerve roots and trunks of the lumbosacral plexus was detected without gadolinium enhancement on MRI and MR neurography (MRN) (arrowheads Picture C; T2-weighted MRI, Picture D; diffusion-weighted MRN obtained with post-processing with maximum intensity projection). Given that the peripheral nerves are involved in CTX (2), the CTX diagnosis may be enhanced by the detection of spinal root hypertrophy on MRI.

The authors state that they have no Conflict of Interest (COI).

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