Teaching NeuroImages:

Oculodentodigital dysplasia

Hypomyelination and syndactyly

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Figure Oculodentodigital dysplasia

T2-weighted images with hyperintensities (black arrows) of the white matter in (A) precentral and (B) parietooccipital areas, as also the posterior limbs of the internal capsule. Hypointense signal changes (white arrows) affecting the precentral and parietooccipital cortex (A and B), the globus pallidus and thalamus (B). Syndactyly of the IV-V fingers (C).

A 44-year-old woman presented to our department with an 8-year history of slowly progressive spastic paraparesis, urinary and bowel incontinence, depression, and forgetfulness. The patient was born with syndactyly of the fourth and fifth fingers (figure, C). Cranial MRI revealed a distinct leukoencephalopathic pattern (figure, A and B), which together with familial syndactyly was suspicious of oculodentodigital dysplasia, an autosomal-dominant hypomyelination disorder,¹ presenting with oculofacial and dental abnormalities and syndactyly.² Neurologic symptoms and leukoencephalopathy are common.² Sequence analysis of the connexin 43/gap junction protein α -1 (*Cx43/GJA1*) gene revealed a novel missense mutation (c.283C>T; p.H95Y) in a heterozygous state.

DISCLOSURE

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