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, presenting with ocular, facial, and dental abnormalities and syndactyly. 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.

REFERENCE

DISCLOSURE
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