Variants Cause Spastic Paraplegia Associated with Cerebral Hypomyelination

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Résumé en anglais	Oculodentodigital dysplasia is an autosomal dominant disorder due to variants characterized by dysmorphic features. Neurologic symptoms have been described in some patients but without a clear neuroimaging pattern. To understand the pathophysiology underlying neurologic deficits in oculodentodigital dysplasia, we studied 8 consecutive patients presenting with hereditary spastic paraplegia due to variants. Clinical disease severity was highly variable. Cerebral MR imaging revealed variable white matter abnormalities, consistent with a hypomyelination pattern, and bilateral hypointense signal of the basal ganglia on T2-weighted images and/or magnetic susceptibility sequences, as seen in neurodegeneration with brain iron accumulation diseases. Patients with the more prominent basal ganglia abnormalities were the most disabled ones. This study suggests that -related hereditary spastic paraplegia is a complex neurodegenerative disease affecting both the myelin and the basal ganglia. variants should be considered in patients with hereditary spastic paraplegia presenting with brain hypomyelination, especially if associated with neurodegeneration and a brain iron accumulation pattern.
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