

Brain magnetic resonance image changes in a family with congenital and classic myotonic dystrophy

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摘要

Abstract

We present the clinical manifestations, brain magnetic resonance images (MRI), and genetic analysis of a family with 2 siblings with congenital myotonic dystrophy type 1 (DM1) and 4 patients with classic DM1. These 2 patients with congenital DM1 had severe mental retardation and a characteristic feature of hyperintensity of white matter at the posterior-superior trigone (HWMPST), in addition to ventricular dilatation in T2-weighted images (T2WI) of brain MRI. In 2 of the 4 classic DM1 patients, brain T2WI MRI showed hyperintensity lesions in the bilateral frontal and/or temporal regions, which were absent in congenital DM1. In conclusion, we suggest that the HWMPST in brain MRI is a characteristic finding in congenital DM1, and that the severe cognitive impairments are not only attributable to the subcortical white matter lesions. In congenital DM1, the cognitive function is a diffuse impairment, which is different from that in classic DM1.