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Myotonic dystrophy type 1 with anterior temporal white matter changes mimicking cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy

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Abstract

Myotonic dystrophy type 1 is the most common type of muscular dystrophy in adults characterized by progressive myopathy, myotonia, and occasional systemic involvement. This is a case of myotonic dystrophy type 1 with cognitive decline showing brain magnetic resonance image abnormality mimicking cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL).

Keywords: Myotonic dystrophy, magnetic resonance image, cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)

INTRODUCTION

Myotonic dystrophy type 1 (DM1) is a muscular disorder caused by the expansion of cytosine-thymine-guanine (CTG) repeats in the myotonic dystrophy protein kinase (*DMPK)* gene.[1] Previous literatures delineated that DM1 can not only present with muscular involvement, but also manifest with central nervous system (CNS) dysfunction.[2,3] We report here a patient with DM1 whose brain magnetic resonance imaging (MRI) showed severe hyperintense lesions on both anterior temporal lobes mimicking cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). The institutional review board of the Hallym University Sacred Heart Hospital approved this study (IRB No 2020-03-008).

CASE REPORT

A 52-year-old man with underlying diabetes mellitus was referred to our hospital for increasing difficulty of respiratory problem. He was intubated and treated with artificial ventilator in the intensive care unit. He had progressive generalized weakness and gait disturbance for years, but diagnostic work-up was not performed before. His parents and sisters did not have any neurologic problem, but his older brother was diagnosed with myotonic dystrophy. He showed the typical phenotype of DM1, such as bald head and diffuse muscle wasting including the facial muscle. Percussion myotonia was prominently observed in the first dorsal interossei and abductor pollicis brevis muscles. Needle electromyography showed frequent myotonic discharges in the distal hand muscles. Molecular analysis of *DMPK* mutation demonstrated the 650 CTG repeats in 1 allele.

Although the detailed cognitive function could not be ascertained due to intubation and tracheostomy, progressive cognitive decline was noted by family members before admission and has been observed during admission as well. A brain MRI showed diffuse brain atrophy and hyperintensities in the external capsule and subcortical white matter, especially the anterior temporal lobes on the T2-weighted and fluid-attenuated inversion recovery (FLAIR) image which is a common finding in CADASIL syndrome (Figure 1a). In addition, his cognitive function became worse for the next 6 months and the follow up brain MRI showed worsening of the brain atrophy and increased high signal intensity lesion in bilateral anterior temporal lobes (Figure 1b). This suggested the necessity to exclude CADASIL syndrome in addition to DM1, because the imaging findings and cognitive dysfunction can mimic CADASIL syndrome. Exome sequencing for *Notch 3* gene mutation was performed and was normal.

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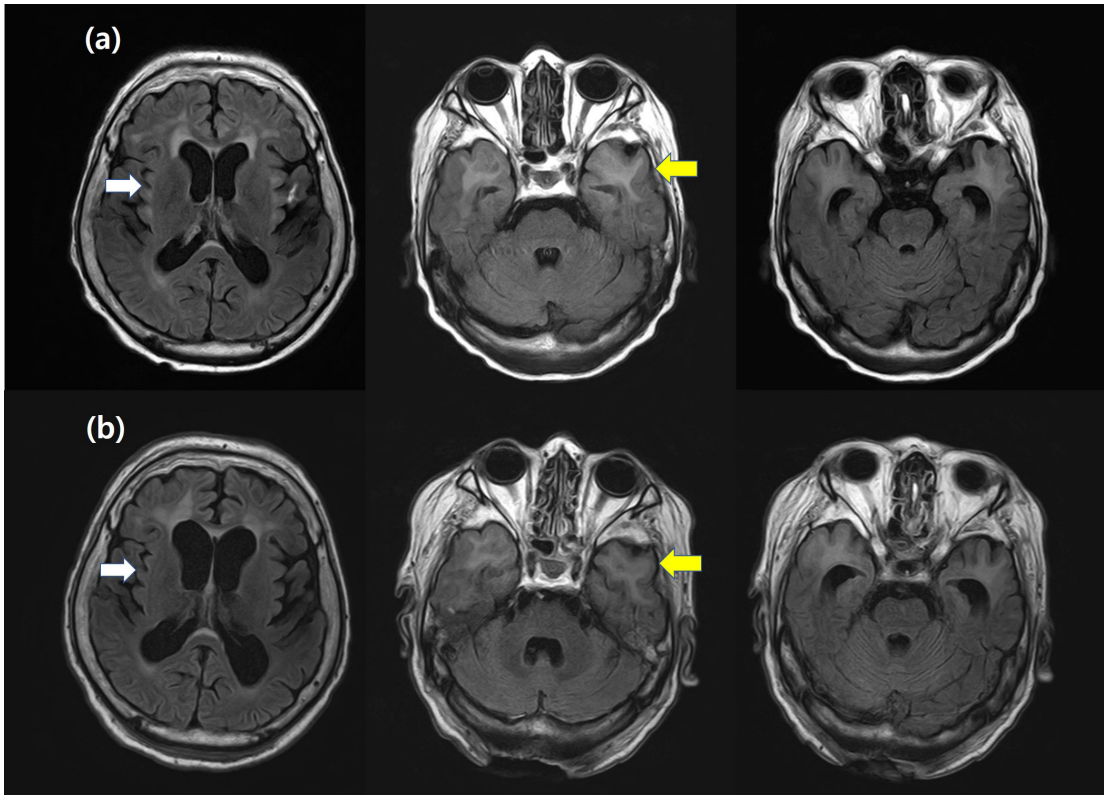

Figure 1. Brain FLAIR MRI showing diffuse brain atrophy and hyperintensity in the external capsule (white arrow) and the bilateral frontotemporal white matter (yellow arrow). (a) Initial evaluation of MRI, (b) Follow-up MRI after 6 months showing worsening of the brain atrophy

DISCUSSION

DM1 is an autosomal-dominant muscular disorder not only characterized by myotonia and muscular dystrophy, but also endocrinopathy, cardiac conduction defect, peripheral polyneuropathy, and CNS dysfunction.[4] The CNS manifestations in myotonic dystrophy are not prominent but can present as intellectual impairment, hypersomnia, personality changes, and apathy.[4]

Previous studies have reported that brain MRI in DM1 can show cortical atrophy, ventricular dilatation, and subcortical or periventricular white matter lesions (WML).[4,5] The severity of subcortical WMLs may correlate with the degree of intellectual impairments. However, poor correlation was found between cognitive impairment and CTG repeat size.[4,6,7] The involvement of external and extreme capsules in the WMLs, especially the anterior temporal lobe, well known in CADASIL, has also been reported in DM1.[8,9] We have thus excluded the mutation of *Notch 3* gene that causes CADASIL. This case shows that DM1 can manifests with cognitive dysfunction and brain MRI abnormality similar to CADASIL.

DISCLOSURE

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Conflict of interest: None

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