

# Magnetic resonance imaging findings in adult-form myotonic dystrophy type 1

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**ABSTRACT** The adult form of myotonic dystrophy type 1 is a neuromuscular disorder with multisystem involvement, including the central nervous system (CNS). The presenting clinical features of this condition include distal muscle weakness, myotonia, intellectual decline, cataract, frontal baldness and testicular atrophy. Magnetic resonance (MR) imaging shows characteristic white matter changes in the CNS. The clinical presentation, characteristic white matter changes in the brain on MR imaging and electromyographic findings aid in the diagnosis of this disorder.

*Keywords:* adult form, congenital, magnetic resonance imaging, myotonic dystrophy  
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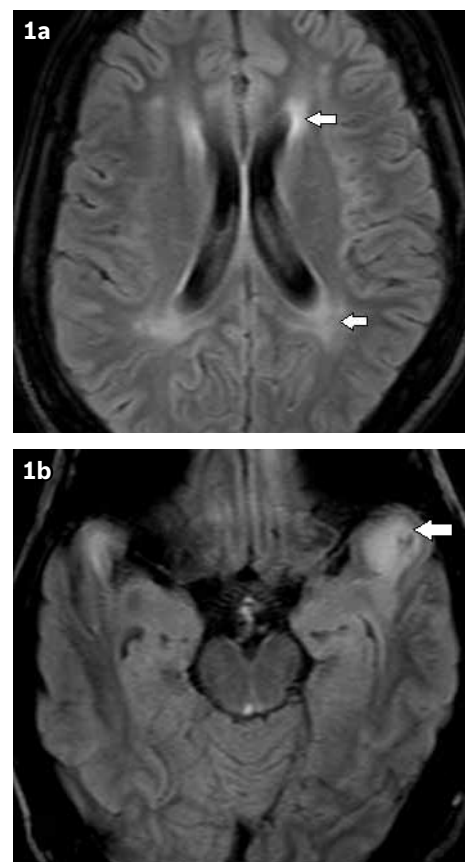
## INTRODUCTION

Myotonic dystrophy type 1 (DM1) is a multisystem disorder with autosomal dominant inheritance affecting the neuromuscular system, the eye, heart, endocrine system and central nervous system. It results from expansion of tri-nucleotide, cytosine-thymine-guanine (CTG) located on chromosome 19q13.3.<sup>(1,2)</sup> The adult form of myotonic dystrophy is a common form of DM1 and typically presents at 15–40 years of age with distal muscle weakness, myotonia, muscular atrophy, cognitive impairment, poor scholastic performance and hearing impairment.<sup>(2,3)</sup> Magnetic resonance (MR) imaging findings are of great value in the diagnosis of this disorder.<sup>(4)</sup>

## CASE REPORT

A 34-year-old Indian man presented to our hospital with a history of global developmental delay involving language and motor milestones. He had a history of poor scholastic performance, bilateral facial and distal muscle weakness in the form of poor hand grip since childhood. He also complained of visual and hearing impairment. Physical examination revealed distal limb weakness, bilateral facial weakness, ptosis and scalp baldness. Audiometric test for his hearing impairment showed mixed hearing loss, while ophthalmic examination revealed early cataract changes. Sensory examination was unremarkable. Audiometry studies showed mild mixed sensorineural hearing loss and ophthalmoscopic examination showed early cataractous changes. Metabolic work-up revealed normal serum creatine phosphokinase with mildly elevated serum lactate levels.

MR imaging of the brain showed bilateral multifocal, slightly asymmetric, patchy T2-weighted hyperintensity in the periventricular and deep white matter (Fig. 1a) and subcortical white matter of the anterior temporal lobes, which was more prominent on the left side (Fig. 1b). Similar hyperintensity was also observed in the white matter posterior and superior to the

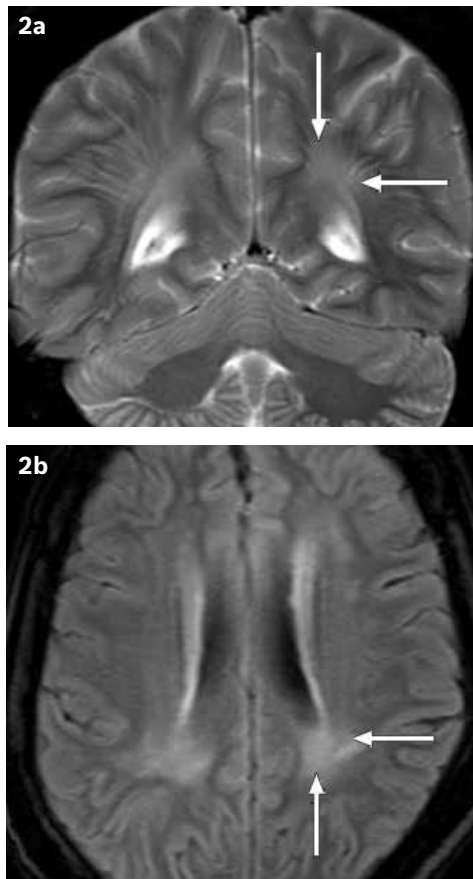


**Fig. 1** Axial fluid-attenuated inversion recovery (FLAIR) image (a) at the level of the body of lateral ventricles shows bilateral periventricular and deep white matter hyperintensity (arrows); and (b) through the anterior temporal lobes shows asymmetric anterior temporal subcortical white matter hyperintensity at the left more than right side (arrow).

trigone (WMPST) (Figs. 2a & b). Prominence of Virchow Robin (VR) spaces was also seen disproportionate to the patient's age (Figs. 3a & b). Central grey nuclei, including both basal ganglia and thalami, were normal. Brainstem and cerebellar hemispheres

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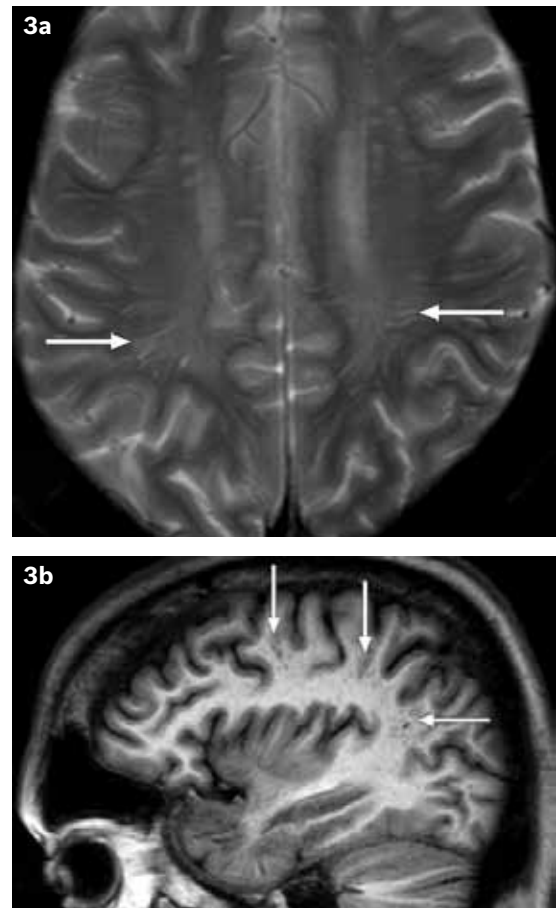
**Fig. 2** (a) Coronal T2-W image shows bilateral periventricular white matter hyperintensity posterior and superior to the trigone (arrows). (b) Axial FLAIR image shows bilateral periventricular white matter hyperintensity posterior and superior to the trigone (arrows).

were unremarkable. Based on the clinical features and MR imaging findings, we first suggested a possibility of the adult form of DM1. Electrocardiography (ECG), done to rule out associated conduction abnormality, was found to be normal. Endocrine workup showed normal blood glucose and testosterone levels.

Subsequently, the patient underwent electromyographic examination, which showed no spontaneous activity, normal motor unit potentials (MUPs) and normal interference pattern. There was evidence of myotonic discharges with small MUPs and interference pattern toward myopathy; these findings were consistent with myotonic dystrophy. Based on the clinical features, electromyography and MR imaging findings, the patient was diagnosed with the classical form of DM1. Genetic testing was carried out.

## DISCUSSION

DM1 is a progressive neuromuscular disorder with multisystem involvement. It has two major clinical phenotypes—congenital and adult form. The congenital form presents with hypotonia, severe intellectual impairment, and respiratory and feeding difficulties. Myotonia is generally absent in this form, but is characteristically seen in the adult form. Adult-form DM1 can be a mild or classical form. The mild form presents with cataract and mild muscle weakness, while the classical form is characterised



**Fig. 3** (a) Axial T2-W image shows prominent perivascular spaces (arrows). (b) Sagittal T1-W image shows prominent perivascular spaces (arrows).

by distal muscle weakness, muscle wasting, myotonia, cataract, frontal baldness, testicular atrophy and cardiac conduction abnormalities.<sup>(2)</sup>

On cranial MR imaging, the adult form of DM1 shows hyperintensity involving the periventricular and deep white matter on T2-weighted sequence. Subcortical white matter involvement is a characteristic finding, with a predilection for anterior temporal white matter. The distribution of these white matter changes is asymmetric and patchy.<sup>(3-5)</sup> Dilated VR spaces is a commonly described finding.<sup>(6)</sup> Basal ganglia, thalamus and brainstem show normal signal intensity. Compared to the adult form, the congenital form of DM1 shows more severe cerebral atrophy, ventriculomegaly and hyperintensity in the WMPST.<sup>(7,8)</sup>

Abnormal signal in the temporal white matter in association with white matter cysts can be seen in a variety of conditions such as cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL), cytomegalovirus infection and megaencephalic leukoencephalopathy. Congenital cytomegalovirus infection can produce similar white matter changes as DM1, but the lesions tend to be multifocal, with the largest lesion affecting the parietal lobes with sparing of the immediate periventricular and subcortical white matter. The associated gyral abnormality, periventricular calcifications and periventricular cystic changes are unique imaging findings.<sup>(9)</sup> Megaencephalic leukoencephalopathy with sub-

cortical cysts can show similar white matter changes, with a propensity to involve external and extreme capsules. The characteristic cystic changes in the frontal and temporal subcortical white matter along with macrocephaly can distinguish this disease from DM1.<sup>(10)</sup> CADASIL is another differential for similar white matter changes, but it can be excluded based on the history of headache, dementia and stroke-like symptoms. On imaging, lacunar infarcts are characteristic of this entity.<sup>(11,12)</sup>

In conclusion, in the appropriate clinical setting, high-signal areas on T2-weighted images in the anterior temporal lobe, subcortical white matter, periventricular and deep white matter with prominent VR spaces are highly suggestive of DM1. Knowledge of MR imaging findings in the proper clinical context may help to expedite the diagnosis of this rare entity.

## REFERENCES

1. Lee JE, Cooper TA. Pathogenic mechanisms of myotonic dystrophy. *Biochem Soc Trans* 2009; 37(Pt 6):1281-6.
2. Turner C, Hilton-Jones D. The myotonic dystrophies: diagnosis and management. *J Neurol Neurosurg Psychiatry* 2010; 81:358-67.
3. Glantz RH, Wright RB, Huckman MS, Garron DC, Siegel IM. Central nervous system magnetic resonance imaging findings in myotonic dystrophy. *Arch Neurol* 1988; 45:36-7.
4. Miaux Y, Chiras J, Eymard B, et al. Cranial MRI findings in myotonic dystrophy. *Neuroradiology* 1997; 39:166-70.
5. Di Costanzo A, Di Salle F, Santoro L, et al. Pattern and significance of white matter abnormalities in myotonic dystrophy type 1: an MRI study. *J Neurol* 2002; 249:1175-82.
6. Di Costanzo A, Di Salle F, Santoro L, Bonavita V, Tedeschi G. Dilated Virchow-Robin spaces in myotonic dystrophy: frequency, extent and significance. *Eur Neurol* 2001; 46:131-9.
7. Martinello F, Piazza A, Pastorello E, Angelini C, Trevisan CP. Clinical and neuroimaging study of central nervous system in congenital myotonic dystrophy. *J Neurol* 1999; 246:186-92.
8. Di Costanzo A, Di Salle F, Santoro L, Bonavita V, Tedeschi G. Brain MRI features of congenital- and adult-form myotonic dystrophy type 1: case-control study. *Neuromuscul Disord* 2002; 12:476-83.
9. van der Knaap MS, Vermeulen G, Barkhof F, et al. Pattern of white matter abnormalities at MR imaging: use of polymerase chain reaction testing of Guthrie cards to link pattern with congenital cytomegalovirus infection. *Radiology* 2004; 230:529-36.
10. van der Knapp MS, Scheper GC. Megalencephalic leukoencephalopathy with subcortical cysts. In: Pagon RA, Bird TD, Dolan CR, et al, eds. *GeneReviews™* [online]. Available at: [www.ncbi.nlm.nih.gov/books/NBK1116/](http://www.ncbi.nlm.nih.gov/books/NBK1116/). Seattle (WA): University of Washington, Seattle, 1993.
11. O'Sullivan M, Jarosz JM, Martin RJ, et al. MRI hyperintensities of the temporal lobe and external capsule in patients with CADASIL. *Neurology* 2001; 56:628-34.
12. van den Boom R, Lesnik Oberstein SA, Ferrari MD, Haan J, van Buchem MA. Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: MR imaging findings at different ages--3rd-6th decades. *Radiology* 2003; 229:683-90.