pISSN 1738-6586 / eISSN 2005-5013 / J Clin Neurol 2022;18(2):227-229 / https://doi.org/10.3988/jcn.2022.18.2.227

Basilar Artery Dissection in Myotonic Dystrophy Type 1

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Received October 14, 2021
Revised December 6, 2021
Accepted December 6, 2021

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Dear Editor,

Myotonic dystrophy type 1 (DM1) is caused by a genetic malfunction involving the overexpression of the CTG sequence in *DMPK*. This affects the skeletal, cardiac, and smooth muscles, presenting with abnormalities in several body areas. In particular, cardiac fibrosis caused by invasion into cardiac muscle is associated with a high incidence of atrial fibrillation in patients with DM1.[1] Atrial fibrillation has been reported as a major causal factor of stroke in DM1.[2] Although previous studies have shown that DM1 is associated with an abnormality of the vascular smooth muscle,[3] there has been no report of DM1 being related to a compromise of the macrovascular system, which includes arterial dissection. Dissection of the basilar artery (BA) is a very rare disease with an annual incidence of 1/400,000. We report the case of a patient with DM1 who was diagnosed with an acute cerebral infarction due to BA dissection without trauma. Artery dissection could be another etiology of ischemic stroke in patients with myotonic dystrophy.

A 58-year-old female visited our clinic due to a gait disturbance, which had worsened 7 days before her visit. She was diagnosed with myotonic dystrophy in her 20s. Her older sister had also been diagnosed with DM1 in her 20s. A neurological examination revealed sensory deficits following light touches in the left facial region and the upper and lower extremities. Moreover, a cerebellar function test revealed left-limb dysmetria and a left-sided falling tendency during gait (National Institutes of Health Stroke Scale score of 5 and modified Rankin Scale score of 3). Percussion myotonia was observed in the abductor pollicis brevis. The findings of routine laboratory tests including of thyroid function were normal. PCR-Southern analysis with a biotin-(CTG) 10 probe revealed that *DMPK* (CTG) was amplified with over 550 repeats. The characteristic myotonic discharge was observed in the overall muscles. Electrocardiography revealed atrial fibrillation, but no conduction defect or tachycardia was present. Acute infarction of the right pons was confirmed in brain diffusion-weighted magnetic resonance angiography (MRA) (Fig. 1A). Dissection was suspected in the middle portion of the BA (Fig. 1B, C). Transfemoral cerebral angiography was performed to confirm dissection, which revealed flame-like tapering with long-segment stenosis at the same site (Fig. 1D). She was administered 5 mg of apixaban twice daily and 40 mg of atorvastatin once daily, and was discharged after her limb ataxia and gait disturbance improved.

DM1 continuously weakens and depletes the muscles in the face, neck, and extremities, and it is characterized by myotonia when the invaded muscle is percussed.[4] The severity of DM1 may vary in an individual patient. The adult-onset type is the most common, and its symptoms become distinct from the age of 40 years. DM1 is caused by mutations in *DMPK* located on chromosome 19q13.3.

DM1 presents with abnormalities in several areas of the body, including the musculoskeletal, nervous, circulatory, and endocrine systems, and the gastrointestinal (GI) tract. DM1 presents with mainly musculoskeletal clinical symptoms. However, it also often presents with symptoms associated with the smooth muscle, including vomiting, constipation, diarrhea, and dysphagia resulting from the invasion of the smooth muscle of the GI tract, which can in-

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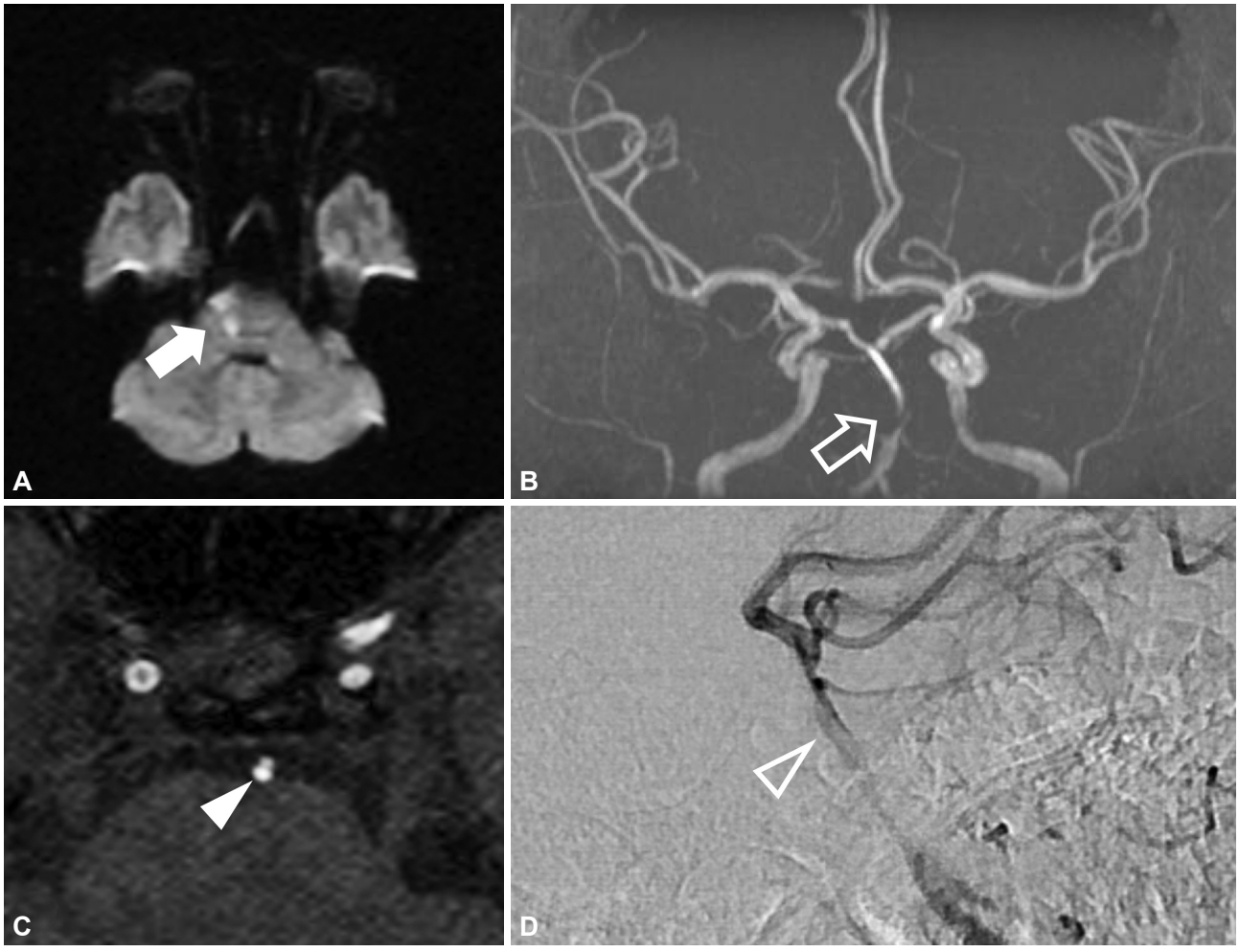

Fig. 1. A case of basilar artery (BA) dissection with acute right lateral pontine infarction. A: Brain noncontrast diffusion-weighted magnetic resonance angiography (MRA) image showing acute infarction involving the right lateral pons (arrow). B: A curved planar reformatted image generated using computed-tomography angiography shows tapered stenosis of the midbasilar artery with an intimal flap. C: Axial 2D time-of-flight MRA image in this patient demonstrates the double lumen with an intimal flap (arrowhead) around the BA.[9] D: Conventional cerebral angiography image of the BA shows flame-like tapering of the proximal vessel with long-segment high-grade stenosis (arrowhead).

duce chronic intestinal pseudo-obstruction in adults. These GI symptoms can be the initial clinical manifestations of DM1, which may be followed by musculoskeletal symptoms. Histologically, it has been reported that the muscular layer of the small intestine in DM1 patients is segmented and damaged, similar to the effects on skeletal muscle. This suggests that the pathological mechanisms of DM1 are similar regardless of the muscle type.[5]

It has frequently been reported that DM1 involves the invasion of blood vessels.[1] which may be related to dysfunction of the smooth muscle in the vascular walls. Itoh et al.[6] applied a dipyridamole thallium-201 myocardial perfusion test to DM1 patients to confirm the presence of perfusion defects in the cardiac septum. Those authors considered that microvascular dysfunction in the heart could have damaged the myocardium. Myocardium damage in patients with DM1 can cause diverse types of cardiac dysfunction, including rhythm abnormality and sudden cardiac death.[7] Moreover, the reported incidence of atrial fibrillation in patients with DM1 has been up to 30%, and this is the main cause of ischemic stroke.[8] Yoshida et al.[2] reported that 9 out of 71 patients with DM1 had ischemic stroke. Those authors also found that the atrial fibrillation rate was significantly higher in the ischemic stroke group than in the asymptomatic group. A causal analysis revealed that cardioembolism was the most common factor (six of out nine patients).

Blood vessels structurally comprise the intima, media, and adventitia, with the media being the thickest layer, and consisting mainly of smooth muscle and elastic fibers. Therefore, DM1 may involve vascular damage, such as dissection, even in the presence of minor trauma caused by structural instability of the smooth muscle of the vascular media. The present

subject was a DM1 patient without any specific history of trauma. It was suspected that intracranial artery dissection occurred incidentally because of the weakened smooth muscle in the intracranial artery as a result of DM1. There were no other intracranial atherosclerotic features other than findings suggesting BA dissection, even in brain MRA. It appears that this case is the first patient with DM1 who developed an ischemic stroke due to artery dissection rather than cardioembolism.

The present findings suggest that complications due to structural instability resulting in artery dissection in DM1 can occur. Further extensive studies are needed to clearly explain the underlying mechanisms.

Ethics Statement

Written informed consent was obtained from the patient.

Availability of Data and Material

The datasets generated or analyzed during the study are available from the corresponding author on reasonable request.

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Conflicts of Interest

The authors have no potential conflicts of interest to disclose.

Funding Statement

This research was supported by Basic Science Research Program through the National Research Foundation of Korea (NRF) funded by the Ministry of Education (No. 2021R1I1A3056006).

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