

Case Report

Delayed Contrast Enhancement on Cardiac MRI Unmasks Subclinical Cardiomyopathy in a Case of Myotonic Dystrophy Type 2

KONSTANTINOS SPENGOS¹, ELIAS GIALAFOS², SOFIA VASSILOPOULOU¹, PANAGIOTIS TOULAS³, PANAGIOTA MANTA¹

¹University of Athens Medical School, Department of Neurology, Eginition Hospital, ²Service of Cardiology, Eginition Hospital, ³Euromedica Encephalos Diagnostic Institute, Athens, Greece

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Current evidence suggests cardiac involvement and electrocardiographic changes of increasing frequency with age in patients with myotonic dystrophy type 2 (DM2). Myocyte hypertrophy with concurrent fibrosis seems to be the anatomical correlate. Moreover, morphological and functional changes indicative of subclinical cardiomyopathy have been demonstrated by means of cardiac magnetic resonance imaging (CMRI) and spectroscopy in patients with no overt cardiac disease. We present a 68-year-old woman with genetically established DM2 and no clinical, electrocardiographic or echocardiographic signs indicative of cardiac involvement. CMRI revealed delayed contrast enhancement of the anterior portion of the interventricular septum, indicating myocardial involvement. Contrast-enhanced CMRI might be a useful diagnostic tool in assessing cardiac involvement in cases of DM2. The role of delayed contrast enhancement should be further investigated in order to elucidate the cardiac features of this fascinating multisystem disease.

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Address:
Konstantinos Spengos

Department of
Neurology, Eginition
Hospital
Vas. Sofias 72
115 28 Athens, Greece
e-mail: kspengos@otenet.gr

Type 1 myotonic dystrophy (DM1), or Steinert's disease, and type 2 myotonic dystrophy (DM2), also known as proximal myotonic myopathy, are well recognised autosomal dominant, progressive, multisystem disorders that are based on the expansion of an unstable repeat sequence in a non-coding part of two different genes. They share a series of common features, such as myotonia and cataracts. Whereas cardiac conduction abnormalities are frequently seen in DM1, the incidence of arrhythmias in DM2 seems to be less common. However, there is evidence that cardiac involvement and electrocardiographic changes become more frequent with age. Unexpected fatal arrhythmias may occur. There is a single report of myocardial biopsy in a case of clinically typical, though genetically not established DM2, which demonstrated myocyte hypertrophy with concurrent fibrosis as

the anatomical correlate to the presence of typical electrocardiographic findings.¹ A recent study used functional cardiac magnetic resonance imaging (CMRI) and magnetic resonance spectroscopy (CMRS) to reveal morphological and functional changes indicative of subclinical cardiomyopathy in a series of DM2 patients with no overt cardiac disease. More precisely, CMRI demonstrated an increased left ventricular volume and an increase of the left ventricular mass in older DM2 patients, whereas CMRS showed a not age-dependent reduction of phosphocreatine and adenosine triphosphate in the cardiac muscle, while skeletal muscles remained unaffected.²

Case presentation

We present the case of a 68-year-old woman with a slowly progressive proximal

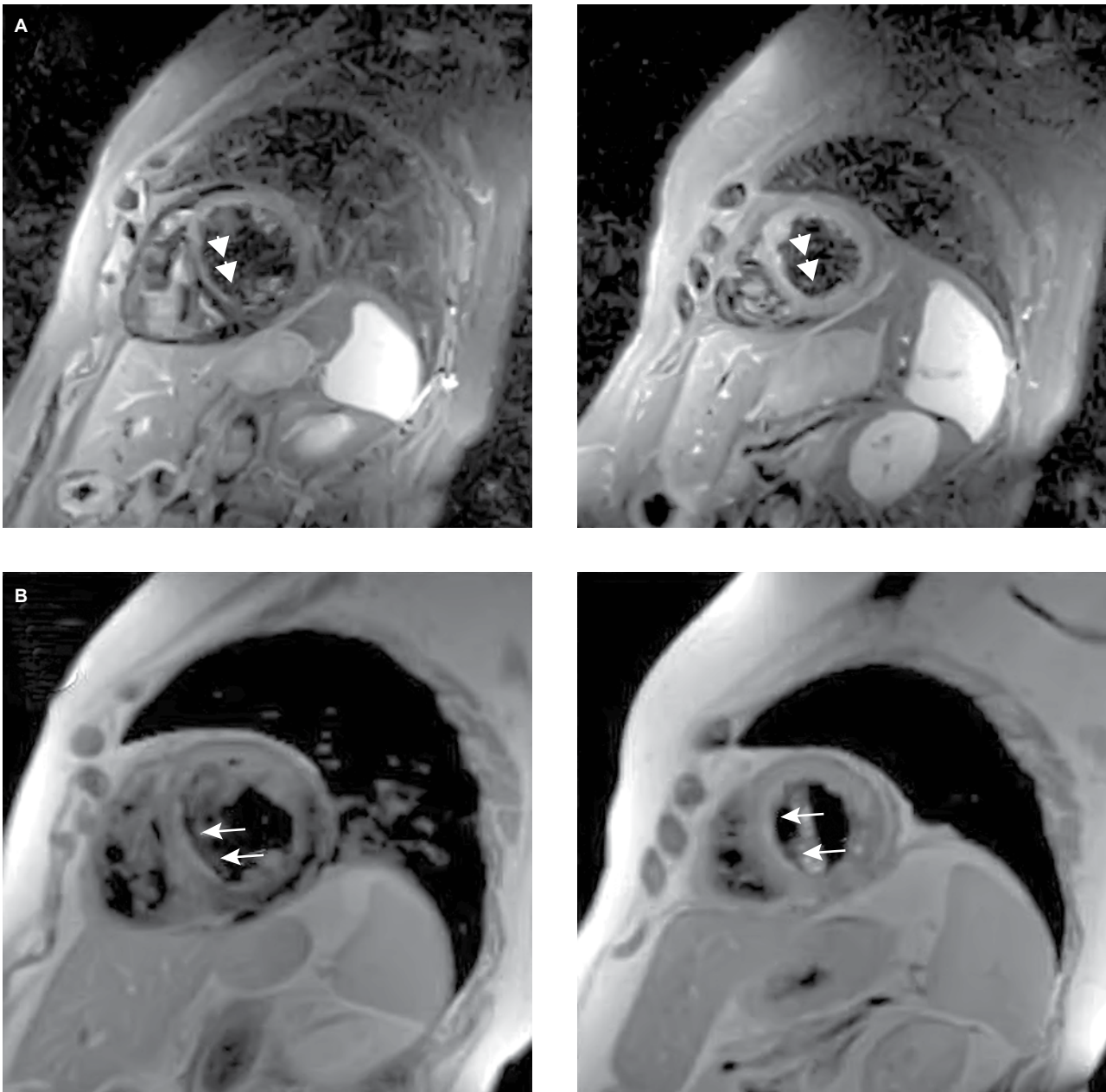


Figure 1. Cardiac magnetic resonance imaging in short-axis, two-chamber view. Signal hyperintensity (arrowheads in panel A) on T2-weighted images with fat suppression (triple IR) and delayed contrast enhancement of the anterior portion of the interventricular septum on T1-weighted images after intravenous bolus gadolinium application (arrows in panel B).

weakness of all limbs that dated back 20 years and significantly deteriorated two years previously, when diffuse myalgias and marked asthenia additionally occurred. She also had a history of surgically treated bilateral cataract. No history of hypertension or diabetes mellitus was reported, and the patient was not on any kind of regular medication. Percussion and grip myotonia were present. Electromyography revealed myopathic findings and diffuse myotonic dis-

charges. Serum CK levels were always elevated, up to four times the upper limits. Molecular evaluation using a long polymerase chain reaction (PCR)-based protocol identified an expanded ZNF9 allele and established the diagnosis of DM2. Cardiological evaluation, including electrocardiogram, 24-hour Holter electrocardiogram and transthoracic echocardiography, revealed no pathological findings indicative of clinically manifest involvement of the heart. Howev-

er, CMRI demonstrated a slight signal hyperintensity in the anterior portion of the interventricular septum on T2-weighted images with fat suppression (triple IR). After intravenous bolus gadolinium application (0.2 ml/kg) a delayed contrast enhancement was documented in the same area. No other signal abnormality or contrast enhancement was observed. There was also no increase in the volume and mass of the left ventricle (Figure 1).

Discussion

Delayed contrast enhancement has been observed in cases of myocardial injury due to myocardial infarction, hypertrophic cardiomyopathy and secondary cardiomyopathies such as cardiac amyloidosis.³ It is mostly located in the interventricular septum and it seems to be histopathologically related to myocardial disarray and fibrosis. There is also a close relation to left ventricular regional contractile function.⁴

In the present case, delayed contrast enhancement on CMRI established the presence of structural myocardial changes indicative of subclinical cardiomyopathy in a DM2 patient with no other signs of

heart involvement. Our finding is in accordance with previous studies that demonstrated myocardial involvement at different stages of DM2, suggesting fibrosis as underlying histopathology.^{1,2} Contrast-enhanced CMRI might be a useful diagnostic tool in assessing cardiac involvement in patients with DM2 and no overt cardiopathy. Its role and the significance of delayed contrast enhancement should be further investigated in order to elucidate the cardiac features of this fascinating multisystem disease.

References

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