

Case Report
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Delayed Contrast Enhancement On Cardiac MRI In a CASE of Steinert Disease: A Case Report

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ABSTRACT

Type 1 myotonic dystrophy (Steinert's disease) is a genetic syndrome characterized by progressive muscular weakness and multisystemic repercussions. Cardiac conduction abnormalities are frequently seen, but the incidence of dilated cardiomyopathy and heart failure seems to be less common. Current evidence suggests that subclinical cardiomyopathy can be demonstrated by means of cardiac magnetic resonance imaging (CMRI). We present a 31-year-old man with genetically established muscular dystrophy 1 and no signs of cardiac involvement. CMRI revealed intramyocardial and pericardial contrast uptake in some regions. The meaning of these findings should be investigated in order to understand and prevent future complications.

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Introduction

Muscular dystrophy connotes a heterogeneous group of inherited disorders characterized by progressive wasting and weakness of the skeletal muscles. In several forms cardiac dysfunction occurs, and cardiac disease may even be the predominant manifestation of the underlying genetic myopathy [1].

Type 1 myotonic dystrophy or Steinert's disease, is the most common muscular dystrophy in the adult life with an incidence of 1 in 8000 births and a worldwide prevalence from 2.1 to 14.3:100 000 inhabitants [2-3]. It is a well-recognized autosomal dominant, progressive, multisystem disorder associated with abnormal expansion of a CTG-trinucleotide repeat sequence in the DMPK gene [4]. Diagnosis is confirmed by genetic testing, with affected individuals having more than 35 trinucleotide repeats. Disease severity and age of onset are correlated with CTG expansion length [5].

Patients with Steinert disease have classic symptoms presenting with muscle weakness and atrophy, myotonia, cataracts, alopecia, endocrine problems and cardiac conduction defects [5]. Predominant symptoms are distal progressive muscle weakness, difficulty making fine movements with hands and feet [5, 6]. Cardiac manifestations are present in about 80% of those patients, and represents the second most common cause of death, after respiratory causes [7]. Dilated cardiomyopathy has been reported, but progressive atrioventricular or intraventricular conduction

defects and tachyarrhythmias (ventricular and supraventricular) are the most life-threatening forms of cardiac complications [8]. Early detection of muscular dystrophy associated cardiomyopathy is important and cardiac magnetic resonance imaging (CMRI) has shown promise in revealing early cardiac involvement when standard cardiac evaluation is unremarkable [9,10].

Case report

We present the case of a 31 years-old man with a slowly progressive weakness and wasting of intrinsic hand and ankle dorsiflexors. He had no history of cataracts, baldness, infertility, mental and endocrine abnormalities. He had no complains of syncope, palpitations, breathlessness or precordial pain, and he was not on any kind of regular medication. Electromyography revealed myopathic findings and diffuse myotonic discharges. 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 expansion of a CTG-trinucleotide repeat sequence in the DMPK gene and established the diagnosis of Steinert disease. Cardiological evaluation, including electrocardiogram (ECG), 24-hour Holter electrocardiogram, exercise electrocardiogram and transthoracic echocardiography, revealed no pathological findings indicative of arrhythmias or another clinically manifest involvement of the heart. However, CMRI (Figure 1) demonstrated an intramyocardial and pericardial contrast uptake in the apical and mid-apical sidewall regions. These alterations did not change with fat suppression or worsen after contrast injection. No other signal abnormality or contrast enhancement was observed. There was also no increase in the volume and mass of the left ventricle.

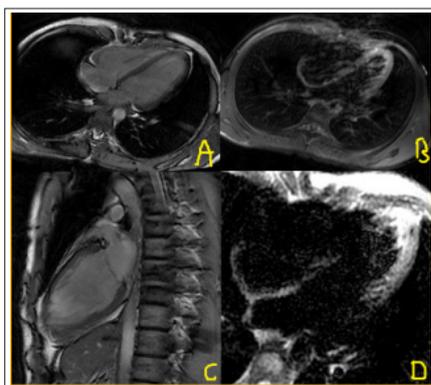


Figure 1: Post-contrast T1-weighted CMRI images demonstrate pericardial thickening and epicardial enhancement (arrows) in the apical and mid-apical sidewall (panels A and C). These alterations did not change with fat suppression (panels B and D).

Discussion

Based on the age of onset and on clinical features, type 1 myotonic dystrophy can be divided into three forms: congenital, classical, and minimal. In the classical form, the most common, symptoms become evident between the second and the fourth decade of life, with predominant distal muscle weakness and a slow progression over time [11]. Despite cardiac abnormalities are present in 80% of muscular dystrophy patients, initial assessment of cardiac involvement should focus primarily on conduction abnormalities, atrial and ventricular arrhythmias and sudden death [12]. Rarely dilated cardiomyopathy and heart failure can occur [13].

Although ECG and echocardiography are typically advocated for screening, CMRI has shown promise in revealing early cardiac involvement when standard cardiac evaluation is unremarkable [9]. CMRI can be requested to assess fatty infiltration and fibrosis in the myocardium [14]. This exam may help define the left ventricle abnormalities of the disease: dilatation, systolic dysfunction, hypertrophy, and occasionally, noncompaction. Typical late gadolinium enhancement (LGE) patterns have not been reported in muscular dystrophy [1]. Overheard et al (2011) referred that mild midwall fibrosis involving the septum is occasionally present [1].

In our patient, despite asymptomatic from the cardiovascular standpoint, his CMRI showed an atypical contrast uptake in the apical and mid-apical sidewall regions, that did not change with fat suppression or after contrast injection. The role of these alterations and their meaning remains unknown. Increased recognition of subclinical myocardial changes with advanced imaging raises challenging management questions. Early detection of cardiac involvement can permit the institution of cardioprotective medical therapies that may slow adverse cardiac remodeling and attenuate heart failure symptoms in the patients [15, 16].

Conclusion

In the present patient, with no signs or symptoms of heart involvement, delayed contrast enhancement on CMRI established the presence of structural myocardial changes indicative of subclinical cardiomyopathy. Contrast-enhanced CMRI has been proposed as a sensitive and useful screening in assessing cardiac involvement and myocardial fibrosis in patients with type 1 myotonic dystrophy and no overt cardiopathy [14].

The assumption that cardiac dysfunction can be prevented (or at least attenuated) in patients with muscular dystrophy has led to the belief that therapy should be initiated at an early stage of the

disease, rather than delayed until ventricular dilatation or systolic dysfunction becomes apparent [15]. The role and the significance of delayed contrast enhancement should be further investigated in order to elucidate the cardiac features of this fascinating multisystem disease [17].

Competing interests

The authors declare they have no competing interests.

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