Abstract

Myotonic dystrophy is a rare inherited condition affecting primarily skeletal muscles and commonly associated with cardiovascular pathology. We report a case of a 50-year-old female with myotonic dystrophy type 1 who presented with wide-QRS complex tachycardia and hemodynamic compromise which was treated with direct current cardioversion. Echocardiogram did not reveal any structural heart disease and coronary angiogram was normal. Electrocardiogram in sinus rhythm showed non-specific intraventricular conduction delay and borderline PR interval. She refused cardioverter-defibrillator (ICD) implantation and was discharged on oral amiodarone. The case report shows that life-threatening arrhythmia could be the first cardiac manifestation in patients with myotonic dystrophy.

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Key words: rare disease, ventricular tachycardia, myotonic dystrophy, implantable cardioverter-defibrillator, cardioversion

Background

Myotonic dystrophy (DM) belongs to the group of neuromuscular disorders. It is a rare inherited condition (incidence 1 in 8000 live births) affecting skeletal muscles and associated with multiorgan involvement including cardiac pathologies [1,2]. There are two forms of DM: DM type 1 (DM1 or Steinert disease) and the rarer DM type 2. We report a case of DM1 patient who presented with hemodynamically instable broad QRS complex tachycardia as a first cardiac manifestation.

Case presentation

A 50 years old lady was brought to hospital by ambulance with a history of first episode of sudden-onset palpitations with general malaise and lightheadedness. Electrocardiogram (ECG) on admission (Figure 1) showed broad QRS complex tachycardia 220 bpm with left axis deviation and atypical right bundle branch block (RBBB) pattern. She was deteriorating rapidly, systolic blood pressure dropped to 60 mm Hg. Blood tests including electrolytes (potassium, magnesium, sodium), full blood count, creatinine and thyroid-stimulating hormone level were all normal. She was not on regular medication prior to admission and there was no previous history of palpitations, chest pain or syncope. She was diagnosed with DM1 in the past. There was no family history of cardiac conditions or sudden deaths. She was a non-smoker.

Due to tachyarrhythmia with hemodynamic compromise the patient required emergency direct-current cardioversion (DCC) which was successful (single delivery of 150J energy). ECG post cardioversion showed sinus rhythm 83 bpm with borderline PR interval (210 ms), prolonged QRS duration (140 ms) due to non-specific intraventricular conduction abnormalities, left axis deviation due to left anterior fascicular block (LAFB) and isolated ventricular ectopics with different morphology as compared to VT (Figure 2). The patient improved clinically – blood pressure...
Majewski, et al. raised to 95/55 mm Hg. She was commenced on loading dose of intravenous amiodarone by on-call team. Transthoracic echocardiogram did not show any evidence of structural heart disease. There was normal left and right ventricular size and systolic function. Left ventricle ejection fraction was 55%. There were no valvular pathologies. She also underwent coronary angiography which showed no pathologies. The case was discussed at cardiac multidisciplinary team meeting and the consensus was to implant ICD as a first step and perform EPS with possible ablation in case of recurrence of arrhythmia in the future. However, the patient refused the proposed intervention and was therefore discharged from hospital on oral amiodarone 100 mg once daily. She will be followed-up in cardiology clinic with regular ECGs and 24 hour Holter ECG Monitoring. The indications for device implantation will be re-discussed with the patient at each follow-up visit. She was also advised to seek medical advice in case of any symptoms and also if she changes her mind regarding the implantation of ICD.

**Discussion**

Clinically, DM manifests with progressive wasting and weakness of skeletal muscles. It may also involve other organs causing cardiovascular abnormalities, thyroid dysfunction, diabetes, hypogonadism, cognitive impairment, cataracts and gastrointestinal disorders. The disease is transmitted in autosomal dominant pattern. Pathogenesis of DM is complex [2,3]. The main alteration is a repeat expansion of trinucleotide sequence (CTG) in the myotonic dystrophy protein kinase (DMPK) leading to dysregulation of various cardiac proteins including CACNA1C and SERCA controlling calcium metabolism, connexins (miR-1, connexin 43 GJA-1) and sarcomere proteins (TNNT2, MYH7,LDB3). RNA toxicity and splicing defects also play a role. Cardiac involvement occurs in 60–80% of patients and manifests as a progressive conduction disorder, atrial arrhythmias (atrial fibrillation or atrial flutter), ventricular arrhythmias and systolic dysfunction of left ventricle [1,4–7]. The most common ECG changes are first degree atrioventricular (AV) block, intraventricular conduction delay (prolonged QRS complex) and prolongation of QT interval [4,5]. Cardiac biopsy in affected individuals reveals fibrosis, fatty deposits and hypertrophy which may constitute a substrate for arrhythmias [8]. DM1 is characterized by seven times higher mortality as compared to age-matched general population. Respiratory failure and cardiovascular diseases are the most common causes of death. Sudden cardiac death accounts for one third of all deaths [5]. It was documented that the presence of conduction abnormalities is associated with poorer prognosis and prolonged HV interval measured during electrophysiological testing (EPS) is predictive for the development of AV block. The need for implantation of pacemaker or ICD in DM1 patients is quite common [9,10]. Current guidelines [5] recommend annual follow-up in patients with muscular dystrophies even in the absence of symptoms or ECG abnormalities. Permanent pacemaker implantation may be considered in patients with DM1 and first degree AV block due to the risk of rapid progression to higher grade AV block (class IIb recommendation). ICD implantation may be considered when there is an indication for pacing and evidence of ventricular arrhythmia (class IIb recommendation). Ventricular tachycardia (VT) may be the first cardiac manifestation of DM1 [11].

![Figure 1. Electrocardiogram on admission. Broad QRS complex tachycardia 220 bpm with left axis deviation and atypical right bundle branch block (RBBB) pattern](image-url)
The mechanism of VT in some patients with DM 1 was reported as bundle branch reentry which could be potentially treated by ablation [12,13]. Other possible mechanism includes ectopic tachycardia originating from right ventricular outflow tract [14]. Our patient presented with hemodynamically instable broad complex tachycardia most likely representing VT (Figure 1). The morphology of QRS complexes was not typical for RBBB with monophasic R waves in V1 and rS in V6 [15,16]. The potential mechanism could be bundle branch reentry or fascicular tachycardia [12]. However it is well known from the literature that the accuracy of ECG in differentiating VT from supraventricular tachycardia with aberrant conduction is limited [17]. Unfortunately the patient refused EPS and as a consequence the exact mechanism of tachycardia could not be established. The alternative mechanism could be atrial flutter with 1:1 aberrant conduction especially in view of LAFB in sinus rhythm. The case shows that the management of patients with DM and broad QRS tachycardia in whom EPS is not performed might be challenging.

References