

CASE REPORT

Cardiac resynchronization improves heart failure in one patient with Myotonic Dystrophy type 1. A case report

VINCENZO RUSSO, ANNA RAGO, ANDREA ANTONIO PAPA AND GERARDO NIGRO

Chair of Cardiology, Second University of Naples, Monaldi Hospital, Naples, Italy

We report an improvement in symptoms of heart failure, a reduced left ventricular dysfunction and induced reverse remodeling in one patient with Myotonic Dystrophy type 1, showing an early onset ventricular dysfunction secondary to a complete left bundle branch block (LBBB) who underwent cardioverter defibrillator CRT (ICD- CRT) implantation.

Key words: myotonic dystrophy, cardiac resynchronization therapy, sudden death

Introduction

Myotonic dystrophy type 1 (MD1), is an autosomal dominant disorder associated with the presence of an abnormal expression CTG trinucleotide repeat on chromosome 19q13.3. The phenotype is characterized by myotonia and muscle weakness, but multisystemic involvement is frequently associated. Cardiac involvement affects the conduction system in about 80% of cases and causes palpitations, lypotimia or syncope (1-4). Myocardial contractile function is less commonly impaired and heart failure (HF) often occurs late in the course of the disease as the final stage of cardiomyopathy (1, 2). Cardiac resynchronization therapy (CRT) is an innovative therapy that can relieve HF symptoms by improving the coordination of the heart's contractions (5). We report an improvement of symptoms of heart failure, ejection fraction and cardiac output values, and a decrease of left ventricular end diastolic diameter (LVEDD) in one patient with Myotonic Dystrophy type 1 showing an early onset of ventricular dysfunction secondary to complete left bundle branch block (LBBB), who underwent implantable cardioverter defibrillator CRT (ICD- CRT) implantation.

Case report

A 35-year-old woman with Myotonic Dystrophy type 1 was referred to our division for dyspnoea. The diagnosis of DM1 was previously made on the basis of the clinical features (frontal balding, bilateral temporal and masticatory muscle atrophy, distal weakness of all four limbs, and confirmed by molecular testing. On physical examination crackles at the basal fields of lungs were detected. Electrocardiogram (ECG) revealed sinus rhythm, left axis deviation, normal PR interval and complete left bundle-branch block with a QRS duration of 180 ms. Trans-thoracic echocardiography showed dilated cardiomyopathy, an ejection fraction (EF), calculated by the Simpson's biplane method, of 25% (n.v. > 55%) while Tissue Doppler echocardiography showed a significant intraventricular mechanical dyssynchrony. The electrophysiological study (EPS), performed using a non aggressive stimulation protocol, revealed a non sustained ventricular monomorphic tachycardia. In presence of overt ventricular dyssynchrony, complete LBBB, and inducible ventricular tachycardia, a biventricular ICD was implanted according to our clinical experience (6).

Methods and follow-up

According to our clinical practice based on previous studies (7-11), the right ventricular lead was placed – via fluoroscopy – in an apical position and the atrial lead positioned in the Bachmann Bundle's region, and secured through active fixture. The left ventricular lead was placed in the postero-lateral branch of coronary sinus.

At 1-month follow up, the echocardiographic optimization of the atrio-ventricular and inter-ventricular intervals during cardiac resynchronization was performed. At six-months follow-up, the patient experienced a symptom relief. ECG revealed paced ventricular rhythm with

narrow QRS complexes; echocardiogram showed and increased EF and LV stroke volume, while LV mechanical dyssynchrony was significantly reduced. The ICD analysis showed no significant modification of the electrical parameters, no ventricular or atrial arrhythmias, and the presence of a 99% biventricular pacing rhythm. Twelve months later, ambulatory interrogation of the device revealed two episodes of ventricular tachycardia stopped by anti-tachycardia pacing (ATP) and one proper and effective ICD shock, occurring during an episode of ventricular fibrillation.

Discussion

Heart failure is rare in Myotonic Dystrophy type 1 and often occurs late in the course of the disease. The clinical recognition of heart failure in muscular diseases is more difficult than in patients with a normal muscular function, as fatigue is inherent to the muscular weakness and exercise tolerance is already impaired by the muscular disease itself.

In DM1, the conduction system is always more extensively affected than the contractile myocardium and high degree AV blocks requiring pacemaker therapy are a well known complication of the disease. The typical ECG of DM1 patients depicts complete LBBB (5 to 25%) with first-degree AV block (20 to 40%). According to ESC 2007 Guidelines for Cardiac Pacing, permanent pacemaker implantation is indicated in DM1 patients with acquired third-degree or second-degree atrioventricular (AV) block (class I B). There is also a class II B indication for first-degree AV block in neuromuscular diseases, when a family history of sudden death is reported.

However, neither a clear consensus about biventricular pacing nor the usage of implantable cardiac defibrillator for patients with Myotonic Heart Disease exists.

Basing on the progressive deterioration of the left ventricular function, progression of AV conduction disturbances and occurrence of ventricular tachyarrhythmia, Said et al. (12) hypothesized a role for biventricular ICD in DM1 patients who need a permanent pacemaker implantation. Kilic et al. (13) described the first case of beneficial cardiac resynchronization in one DM1 patient with heart failure, complete LBBB and ventricular asynchrony, who was not implanted of an intracardiac defibrillator, because no serious life threatening ventricular arrhythmias were induced in the EPS.

In our patient, the early onset of heart failure could be related to the electromechanical delay caused by both intra- and inter-ventricular asynchrony, that leads to regional molecular changes in a non coordinate contracting myocardium and accelerates the progression of the heart failure. The spontaneous ventricular tachycardia, occurred in our patient at twelve months follow up, suggests that the improvement in ejection fraction may not reduce the arrhythmic risk in these patients.

Conclusion

ICD-CRT can be a useful therapy in DM1 patients presenting with heart failure, cardiac dilatation with low EF, complete left bundle block and inducible ventricular tachy-arrhythmias because it improves left ventricular function, induces reverse remodelling and relieves symptoms of heart failure. It can be considered as a life-saving treatment, especially in patients at high-risk of inducible malignant ventricular arrhythmias, although the improvement in ejection fraction seems to not reduce the arrhythmic risk. Whether a biventricular ICD should be the first choice in the management of DM1 patients with early onset heart failure and complete left bundle-branch block needs further investigation.

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