

Ventricular Preexcitation in Emery Dreifuss Muscular Dystrophy: A Rare Case Report

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Abstract

Case Report

Emery-Dreifuss muscular dystrophy (EDMD) is a hereditary syndrome characterized by slow but progressive locomotor involvement and cardiomyopathy. Cardiac involvement is primarily conduction defects and dilated cardiomyopathy. Here we report the case of 27 year old man who present preexcitation syndrome as a cardiac involvement of emery dreifuss muscular dystrophy. We present a rare case of a 27 years old patient who consulted for recurrent palpitations, related to ventricular preexcitation discovered at ECG holter. The neuromuscular exam and the electromyography revealed an emery dreifuss dystrophy. Cardiac imaging found a left ventricular non compaction. The patient was treated by low dose of betablocker. The outcome was uneventful. In our knowledge, the present case is the first report described emery dreifuss muscular dystrophy presenting ventricular preexcitation.

Keywords: Emery Dreifuss muscular dystrophy, preexcitation syndrome, Non compaction of the Ventricular Myocardium.

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INTRODUCTION

Emery-Dreifuss muscular dystrophy is an inherited muscular dystrophy affecting skeletal and cardiac muscles, and is characterized by slowly progressive muscular atrophy, mainly affecting humeroperoneal muscles, with muscle contractures and cardiomyopathy.

The cardiac involvement previously described is conduction defects, that may present as atrioventricular block, bradycardia, or atrial standstill that often lead to syncope and sudden cardiac death, dilated cardiomyopathy, atrial and ventricular arrhythmias[1-3]. In our knowledge no similar association has been reported in the literature.

CASE REPORT

A 27 years old man was admitted to cardiology department because of dyspnea and recurrent palpitations. The patient was born to consanguineous parents. Two of his brothers are disabled.

On examination, his blood pressure was 120/60mmHg; pulse 65beats per minute.

Cardiovascular and pulmonary examinations reveal no abnormality. Neuromuscular exam find hand arthrogyposis, weakness of belt muscles, tendon contractures of hand and elbows and equines foot deformity. The electrocardiogram revealed intermittent ventricular preexcitation. The ECG holter found intermittent ventricular preexcitation without supraventricular tachycardia (figure1). The Electromyography revealed a myogenic damage and normal nerve conduction.

Transthoracic echocardiography revealed a noncompaction diagnosis: non dilated left ventricle with global hypokinesia and numerous large trabeculations. The ejection fraction was slightly depressed (FEVG=45%). There was absence of any atrial and ventricular septal defect or coarctation of aorta. Cardiac MRI revealed a left ventricular non compaction of the apex and adjacent segments without myocardial fibrosis (figure2). The right ventricle has a mild dysfunction.

The patient was treated by low dose of beta-blockers. The outcome was uneventful and the follow up don't found conduction disorders.



Fig-1: Holter ECG of Intermittent ventricular preexcitation without supraventricular tachycardia

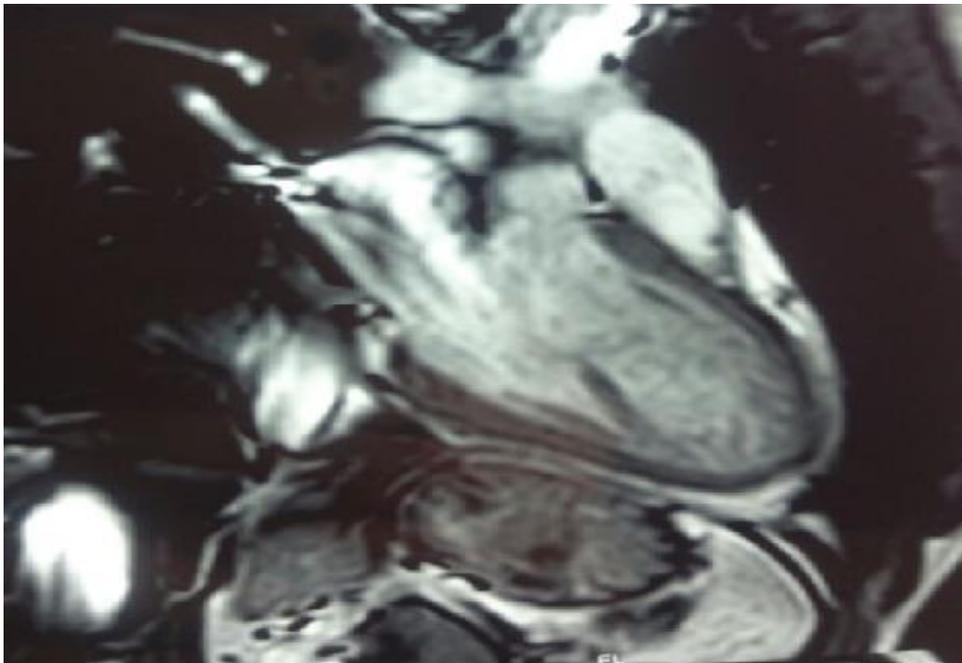


Fig-2: Cardiac-MRI: left ventricular non compaction of the apex and adjacent segments without myocardial fibrosis

DISCUSSION

Emery dreifuss muscular dystrophy is a genetic muscular dystrophy, Characterized by the clinical triad of slowly progressive muscle weakness, and wasting in a scapulo-humeroperoneal distribution; early contractures of the elbows, ankles, and posterior neck, which precede cardiac signs; conduction defects, cardiomyopathy, or both [2].

It was first described as an X-linked disease caused by mutations in the EMD gene, located on chromosome Xq28 encoding the nuclear protein emerin, which is a transmembrane protein of the nuclear envelope inner membrane (EDMD1) [2,3]. More recently it has been found that EDMD can also be

an autosomal disorder caused by mutations in the LMNA gene, which is located in chromosome 1, encoding the nuclear proteins lamin A and lamin C (EDMD2)[1]. The finding that emerin and lamin A/C mutations cause similar disorders indicates that nuclear membrane components play a crucial role in skeletal and cardiac muscle function, and that loss of integrity of the nuclear component is an underlying cause of the muscular dystrophy[4].

Contractures are the first clinical signs of the disease and appear before muscle weakness and wasting, which is opposite of other muscular dystrophies. Contractures affect the elbows, posterior neck and achille tendons. The muscle weakness occurs

progressively humeroperoneal muscles[2]. In EDMD1, the first symptoms are generally contractures, weakness and difficulty in running. Muscle weakness and disease course tend to be more severe in EDMD2 than EDMD1 [5].

Many studies led to the relevant finding that LMNA gene mutation can cause autosomal dominant dilated cardiomyopathy with conduction system disease without EDMD phenotype[1].

Cardiac involvement in EDMD may occur at any age or may even be present at every onset[2,5]. The normal myocardium is progressively replaced by fibroadipose tissues, which result in atrial and ventricular dilatation, and conduction defects. The cardiac conduction defect is the most serious and life-threatening clinical manifestation of the disease, and is considered to be a hallmark of EDMD1, including atrioventricular blocks of different degrees (from prolongation of PR interval to complete block) and sinus node dysfunction up to atrial paralysis[5,3,3], while conduction defect, and dilated cardiomyopathy may be present in EDMD2. Patients with emery dreifuss syndrome risk not only bradyarrhythmia but also supraventricular arrhythmias (atrial fibrillation, flutter) which can cause embolic stroke at a young age[3]. Some of patients may develop atrial and ventricular dilatation with reduction in ejection fraction[3].

Finsterer *et al.* have published a review comparing arrhythmias in different muscular dystrophies. He reported that conduction defects, supraventricular tachycardia, ventricular tachycardia, ventricular fibrillation and sudden death can be found in EDMD patients. In our knowledge, this is the first report of ventricular preexcitation in EDMD. It was related in Duchene muscular dystrophies, Becker muscular dystrophy and mitochondrial disorder [6,7,3]. The WPW syndrome is thought to arise from abnormal embryologic persistence of atrioventricular muscular continuity, which can accompany the failing regression of noncompacted myocardium [8]. The left ventricular non compaction, which is congenital cardiomyopathy, presumably resulting from an arrest in normal endomyocardial embryogenesis, may be associated to emery dreifuss syndrome because the mutation of LMNA gene can cause both disorders[9].

Although EDMD is not a physically disabling disorder as other muscular dystrophies are, but cardiac involvement especially risk of sudden death is thus extremely high and unpredictable; hence the early identification of heart involvement is important. The placement of a permanent ventricular pacemaker may be lifesaving, [10,5]. Some patients can receive Pacemaker as primary prophylaxis of sudden death[6]. However, many of these patients develop ventricular tachycardia; they must be monitored, and should receive implantable cardiac defibrillator as soon as

ventricular tachycardia is recorded[6]. In our case which is the first, the patient was treated by beta blockers, and has a close follow up that not reveal conduction disorders.

However the treatment of the conduction defect does not prevent intra-atrial thrombus formation and cerebral embolization. Successful prophylaxis with anticoagulants should be considered. If there is significant ventricular involvement, diuretic therapy and ACE inhibitors may be indicated[4]. In case of end-stage cardiomyopathy, heart transplantation should be considered.

CONCLUSION

Emery-Dreifuss syndrome is an inherited muscular dystrophy affecting skeletal and cardiac muscles. The cardiac involvement is mostly characterized by conduction defects. Hence, the necessity of a close monitoring and placement of pacemaker is required if conduction anomaly was detected.

To our knowledge, the present case is the first report described the association of emery dreifuss muscular dystrophy and preexcitation syndrome. In the lack of guidelines, the management must be cautious.

List of abbreviations

EDMD: emery dreifuss muscular dystrophy
ECG: electrocardiography
MRI: magnetic resonance imaging
ACE: Angiotensin converting enzyme

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