

Case Report

Successful Heart Transplantation in Patients With Muscular Dystrophies: A Case Series

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ABSTRACT

Muscular dystrophies constitute a group of disorders characterized by muscular weakness and the involvement of the other systems such as the cardiovascular system. In some patients, cardiac involvement is severe and necessitates heart transplantation. Although there are some concerns regarding heart transplantation in these patients due to post-transplantation complications and the deleterious side effects of immunosuppressant drugs, there are several cases of heart transplantation in these patients worldwide. There is, however, no reported case in our country, Iran. Herein, we present 3 successful heart transplantations in patients with muscular dystrophies in Iran. (*Iranian Heart Journal 2020; 21(3): 141-144*)

KEYWORDS: Muscular dystrophy, Heart failure, Heart transplantation

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Inherited muscular dystrophies are characterized by progressive muscle weakness with diverse severity and distribution. The most salient muscular dystrophies are Emery–Dreifuss, Becker, Duchenne, facioscapulohumeral, oculopharyngeal, distal, and limb-girdle. The majority of muscular dystrophies have multisystem involvements. The diagnosis of muscular dystrophies is based on creatine phosphokinase levels, electrocardiography, electromyography, muscle biopsy, and DNA analysis. A specific treatment has yet to emerge for these diseases.¹

Heart transplantation in muscular dystrophies has been done in several patients with end-

stage heart failure; nonetheless, there is no reported case in this regard in our country, Iran. We herein describe 3 successful heart transplantations in patients with muscular dystrophies for the first time in Iran.

CASE PRESENTATION

Case 1

The patient is a 38-year-old man. He had muscular symptoms, mainly difficulties in climbing stairs and rising from a seated position, from the age of 20. Seven years prior to the writing of this article, he was diagnosed with dilated cardiomyopathy (DCM). Neurological examinations and muscle biopsy led to the establishment of the diagnosis of

Becker muscular dystrophy. The patient also suffered pulmonary thromboembolism 6 years before heart transplantation. He was given guideline-directed medical therapy upon the diagnosis of heart failure; nonetheless, he had several admissions during the year preceding the transplantation procedure due to decompensated heart failure attacks, leading to the administration of inotropes and intravenous diuretics in each episode. He was listed for heart transplantation, which was performed a few months later (1 year prior to the writing of this article). The post-transplantation period was uneventful, and cardiac function and right-heart catheterization findings were acceptable. The endomyocardial biopsy results showed no rejection. The patient was discharged in good condition. About 2 months later, he had an admission due to respiratory infection, which was resolved with medical therapy. In the last 12 months since the heart transplantation, he has suffered no deterioration in his muscular function.

Case 2

The patient is a 21-year-old man. He underwent orthopedic surgery 9 years prior to the writing of this article due to leg tendon problems and had difficulties walking since. A year later, he was diagnosed with biventricular dysfunction without significant valvular heart diseases. Muscular biopsy and genetic study helped establish the diagnosis of Emery–Dreifuss muscular dystrophy. Heart failure medical therapy was initiated for him. Endomyocardial biopsy showed focal myocytolysis with interstitial fibrosis, as well as patchy and irregular hypertrophy without evidence of myocarditis. He had several decompensated heart failure admissions with dyspnea and gastrointestinal symptoms despite optimal medical therapy. He was considered for heart transplantation, which was performed 4 years before the

writing of this article. A day after surgery, reoperation via sternotomy was done due to surgical site bleeding. The remainder of the postoperative course was uneventful.

Case 3

The patient is a 36-year-old woman. She had a diagnosis of limb-girdle muscular dystrophy, which was confirmed by muscle biopsy and neurologic findings. She had been symptomatic due to dyspnea since age 21. Further evaluations revealed DCM. She had frequent admissions with decompensation, leading to intravenous diuretic administration in each episode. Due to frequent syncope attacks, she received an implantable cardioverter-defibrillator 6 years prior to the writing of this article. A year later, she was admitted with hemiparesis and unilateral reduced muscular force. Brain magnetic resonance imaging showed evidence of a cerebrovascular accident. Brain angiography showed right carotid dissection and right internal carotid artery occlusion. Accordingly, conservative management was planned; she was placed on guideline-directed medical therapy including lisinopril, carvedilol, spironolactone, furosemide, folic acid, amiodarone due to atrial fibrillation rhythm, warfarin, and levothyroxine due to hypothyroidism. Two years before the writing of this article, she underwent heart transplantation. The pathological examination of the explanted heart showed DCM. Pre-transplantation echocardiography showed severe left ventricular enlargement with an ejection fraction of approximately 20% to 25%, mild right ventricular enlargement with mild-to-moderate systolic dysfunction, moderate tricuspid regurgitation, systolic pulmonary artery pressure of approximately 40 mm Hg, mild-to-moderate mitral regurgitation, and severe biatrial enlargement. The post-transplantation course was

uneventful except for a tamponade episode, which was resolved with pericardiocentesis.

DISCUSSION

The presence of cardiomyopathy, arrhythmia, myopathy, or high levels of creatine kinase in a young adult hints at the possibility of hereditary neuromuscular disease.²

Becker dystrophy is an X-linked recessive disorder that leads to disturbance in dystrophin protein. Symptoms usually begin after the second decade of life with problems climbing stairs. DCM affects up to one-third of this group of patients. Sometimes cardiac symptoms begin earlier than muscular symptoms. Several complications have been reported in the post-heart transplantation period of patients with Becker dystrophy; these complications include increased intubation time due to pneumonia and atrioventricular block.³

The range of cardiac involvement is wide, from subtle to severe heart failure necessitating heart transplantation. Heart transplantation was relatively contraindicated in inherited dystrophies previously on account of 2 major reasons: first, the effect of immunosuppressant drugs on muscle involvement progression and second, postoperative respiratory complications. Nevertheless, evidence showed similar rates of events between patients with and without dystrophies. Generally, patients with mild muscular involvement without respiratory muscle involvement stand a good chance of successful transplantation.⁴

Emery–Dreifuss muscular dystrophies have 3 characteristics: early-onset elbow, ankle, and cervical spine contractures; weakness in the humeroperoneal muscle; and cardiac involvement. Inheritance could be in the forms of X-linked or autosomal dominant, in which mutations in the *STA* or *LMNA* genes occur, respectively. Cardiac involvement includes heart failure and cardiac arrhythmias such as atrial

fibrillation. Female X-linked carriers could also be affected, so the X-linked inheritance does not rule out the disease in females.⁵

In several cases of Emery–Dreifuss muscular dystrophies, the first event is the early contracture of the Achilles tendon; consequently, in patients with this history, muscular dystrophy should be kept in mind.⁶

Limb-girdle muscular dystrophies are genetically heterogeneous diseases that present with progressive proximal shoulder and pelvic muscle weakness besides such other manifestations as cardiac involvement.⁷

Similar to other muscular dystrophies, patients with mild skeletal involvement can undergo heart transplantation, and there are several reported cases of this type around the world.⁸

Although heart transplantation is the treatment of choice in patients with end-stage heart failure and muscular dystrophy, it is advisable that such patients receive careful neuromyological assessment to prevent immunosuppressant-associated myopathy, cyclosporine-induced rhabdomyolysis, and the deterioration of muscular weakness due to post-surgical problems and also underlying dystrophia. It has been posited that the presence of the dystrophin-deleted region in these patients may lower the risk of transplantation rejection by comparison with patients without this problem.⁹

Previous research shows that heart transplantation in this group of patients may augment skeletal muscle performance and functional capacity and resolve heart failure itself.¹⁰

Despite concerns regarding post-transplantation rehabilitation in these patients, there is evidence indicating that rehabilitation in some types of muscular dystrophies may be effective.¹¹

CONCLUSIONS

Heart transplantation in patients with muscular dystrophies can be done in

selected cases with appropriate concerns regarding underlying disorders.

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