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Long term Marfan survivor after orthotopic heart transplantation – Case report

Baskar Sekar MRCP, Simon G Williams MD, Steven M Shaw PhD

Abstract:

Orthotropic heart transplantation in patients with Marfan syndrome remains controversial due to the increased risk of vascular complications. There is subsequently limited clinical experience of outcomes after HTX in this group. We report a 65 year-old-lady with MFS who remains alive 21 years after heart transplantation.

Introduction:

Orthotopic heart transplantation (HTX) in patients with Marfan syndrome (MFS) remains controversial due to the increased risk of vascular complications. Earlier reports suggested a higher incidence of aortic dissection and mortality in this cohort and as a result there was a significant concern regarding listing these patients for HTX. This article describes a case of MFS who underwent successful HTX 21 years ago and reviews the literature.

Case Report:

We report a 65 year-old-lady with MFS who underwent successful HTX 21 years ago for congestive cardiomyopathy. She had been diagnosed with MFS at the age of 21 at the referring unit based on Ghent Nosology(1). The features included ectopia lentis, skeletal abnormalities, positive family history but no aortic root involvement. She had recurrent admissions with cardiac decompensation, so was referred to our centre for urgent consideration for HTX. Her other significant co morbidities included traumatic amputation of left knee making her wheel chair bound and restrictive lung defect secondary to her skeletal deformity. At the time of assessment, she was initially deemed unsuitable for HTX due to concerns over future vascular complications and post operative rehabilitation issues. However, as she did not have aortic involvement, she was added to the active transplant waiting list after extensive discussion and underwent HTX at the age of 44.

Post operatively, she developed high blood pressure (BP) due to improvement in cardiac function, steroids and calcineurin inhibitors. She required three antihypertensive which included an ACE- inhibitor, Beta-blocker (BB) and alpha-blocker. Her ACE-inhibitors was subsequently switched to a Calcium Channel Blocker (CCB) as she developed significant renal impairment.

Twelve years post HTX she required haemo dialysis due to end stage renal failure (thought to be secondary to calcineurinin inhibitors and diabetic nephropathy). However, during her last clinic visit in March 2012, she was well with no signs of overt heart failure and Trans thoracic Echocardiography showed moderate concentric left ventricular hypertrophy, good biventricular function and no aortic pathology.

Discussion:

MFS is a multisystem, autosomal dominant connective-tissue disorder which results from mutation in the fibrillin-1 (FBN1) gene on chromosome 15. It is characterized by clinical manifestations involving ocular, musculoskeletal and cardiovascular systems. Aortic dissection in early adult life is the leading cause of death in MFS. Drugs that are used to lower blood pressure and/or the inotropic state of the heart are frequently used to prevent aortic complications in MFS. Of these, Angiotensin

II type 1 receptor blocker was found to more helpful in a recent study(2), as it antagonizes transforming growth beta signaling pathway which is implicated in pathogenesis and disease progression in MFS. However, Beta-blocker may also play an important role as it slows heart rate and delays aortic wave travel, in addition to lowering central and peripheral pulse pressure(3).

There is a significant concern regarding listing of patients with MFS for HTX, with previous reports suggesting a higher incidence(40%) of aortic dissection and 45% mortality in less than 3 year of follow-up (4). Increased stress placed on the aorta by the transplanted heart, collagen-weakening and hypertensive effect of immunosuppression are felt to be possible contributors. However contrary to this, Knosalla et al in 2007(5) demonstrated post HTX survival in 10 patients with MFS to be similar to patients without MFS. Our patient to date has survived 21 years after heart transplantation. Possible reasons include no aortic root involvement at the time of surgery and aggressive BP control with BB, ACE inhibitor and CCB. We propose that each case of MFS referred for HTX should be considered on an individual basis and we hypothesize that those patients without obvious aortic involvement may carry a more favorable prognosis.

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Correspondence to:

Dr Baskar Sekar
Clinical Research Fellow,
The Transplant Centre,
Wythenshawe Hospital,
University Hospital of South Manchester NHS Foundation Trust
Wythenshawe,
United Kingdom.
Tel.: +44 161 998 7070
E-mail: cardiologist1978@gmail.com

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