Fatal Case of Hypertrophic Cardiomyopathy in a Donor Heart: A Case Report

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ABSTRACT

Hypertrophic cardiomyopathy is a genetically determined heart muscle disease, and patients are at an increased risk for sudden cardiac death.

We report the case of a 39-year-old White man who was found dead at home unexpectedly. He had a cardiac transplant for congenital heart disease at the age of 12 and his condition was maintained with immunosuppression ever since with good cardiac function and right bundle branch block.

At autopsy, the heart was enlarged with a weight of 591 g and had fibrous adhesions of the pericardium with endothelialized sutures in the atria and great vessels in keeping with heart transplant. There was focal septal hypertrophy noted on short axis cut. There was diffuse thickening of the coronary arteries, but no significant stenosis was noted.

On microscopic examination of the heart, sections of right and left ventricle showed myocyte hypertrophy with extensive widespread myocyte disarray and replacement fibrosis. The histologic appearance was that of hypertrophic cardiomyopathy, which was responsible for his sudden unexpected death.

This postmortem diagnosis of hypertrophic cardiomyopathy in a transplanted heart has major implications for the donor family because of the inherited nature of the condition. Follow-up with the donor family is essential in this unique case.

This case highlights the importance of autopsy in transplant death cases. Hypertrophic cardiomyopathy in the donor heart most likely did not manifest phenotypically at the time of transplant in this case because the majority present in adolescence and early adulthood, rarely in childhood. This is first report of such a case.

HYPERTROPHIC cardiomyopathy (HCM) is a genetically determined heart muscle disease with a prevalence of 1 of every 500 adults [1]. It has an autosomal dominant inheritance pattern, and patients are at an increased risk for sudden cardiac death [2,3].

PATIENT PROFILE

We report the case of a 39-year-old White man who was found dead at home unexpectedly. He had a cardiac transplant for congenital heart disease at the age of 12 and his condition was maintained with immunosuppression ever since with good cardiac function and right bundle branch block. Consent for research was obtained from the deceased's next of kin.

At autopsy, the heart was enlarged with a weight of 591 g and had fibrous adhesions of the pericardium with endothelialized sutures in the atria and great vessels (Fig 1A and 1B) in keeping with the bicaval approach of heart transplant [4]. In this technique, the circumferential stitching in both the aorta and pulmonary trunk as well as atria use a polypropylene suture (Fig 1A and 1B). There was focal septal hypertrophy noted on short axis cut (Fig 1C). There was diffuse thickening of the coronary arteries, but no significant stenosis was noted.

On microscopic examination of the heart, sections of right and left ventricle showed myocyte hypertrophy with extensive

Funding Source: Cardiac Risk in the Young sponsor the Cardiac Risk in the Young cardiovascular pathology laboratories.

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0041-1345/20
https://doi.org/10.1016/j.transproceed.2022.10.038

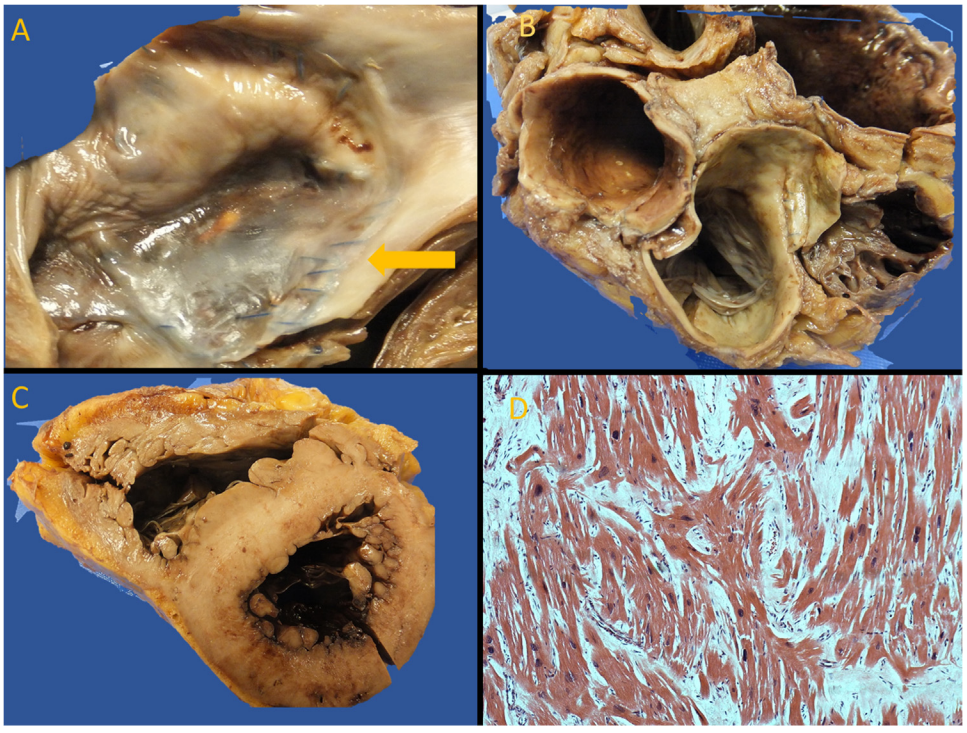

Fig 1. Donor heart showing hypertrophic cardiomyopathy. **(A)** Atrium showing re-endothelialized stitching (yellow arrow) in keeping with transplant. **(B)** Macroscopic image of the greater vessels showing stitching. **(C)** Midventricular slice showing asymmetric septal hypertrophy in keeping with hypertrophic cardiomyopathy. **(D)** Myocyte disarray confirming hypertrophic cardiomyopathy (hematoxylin and eosin).

widespread myocyte disarray and replacement fibrosis (Fig 1D). There was no infarction or significant transplant vasculopathy. The histologic appearance was that of hypertrophic cardiomyopathy, which was responsible for his sudden unexpected death.

DISCUSSION

This postmortem diagnosis of hypertrophic cardiomyopathy in a transplanted heart has major implications for the donor family because of the inherited nature of the condition. An HCM is caused by mutations in ≥11 genes encoding thick and thin contractile myofilament protein components of the sarcomere or the adjacent Z-disc [3]. Follow-up with the donor family is essential in this unique case.

This case highlights the importance of autopsy in transplant death cases. This autopsy revealed underlying genetic heart disease, which was responsible for the death of the patient and has significant implications for the donor family. An HCM in the donor heart most likely did not manifest phenotypically at the time of transplant in this case because the majority present in adolescence and early adulthood, rarely in childhood. This is first report of such a case.

Limitations

Given the nature of this case as a postmortem study, the authors do not have access to the patient's investigations in life but acknowledge that access to clinical investigations such as electrocardiogram and echocardiogram would prove very useful in retrospectively determining if there was any evidence of HCM in the transplanted heart. The authors postulate that regular cardiac surveillance with electrocardiogram and echocardiogram for recipients of heart transplants could prove very useful in detecting abnormalities in life and preventing premature death.

DISCLOSURE

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this article.

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