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Heart Transplantation in a Patient with Propionic Acidemia

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Abstract

Propionic acidemia is a rare autosomal recessive error of metabolism that may be associated with dilated cardiomyopathy and heart failure. We present the case of an 18-year-old male patient with severe heart failure secondary to dilated cardiomyopathy related to propionic acidemia, who underwent a successful heart transplant and 5 months after the procedure progressed satisfactorily, without evidence of rejection, in functional class I and with adequate medical control of propionic acidemia. Heart transplantation may be a treatment option in patients with propionic acidemia, in whom heart failure predominates.

Keywords: Propionic acidemia; Dilated cardiomyopathy; Heart transplantation; Propionyl-CoA carboxylase; Error of metabolism.

Introduction

Propionic acidemia is a rare autosomal recessive error of metabolism, consisting of the deficiency of the mitochondrial enzyme propionyl-CoA carboxylase, which is involved in the transformation of propionyl-CoA into d-methylmalonyl-CoA, which is usually diagnosed in childhood and clinically characterized by hyperammonemia, metabolic acidosis, lethargy, vomiting, hypotonia and in the medium and long term, intellectual impairment, pancreatitis, myocardial involvement with QT prolongation, muscle damage, and dilated cardiomyopathy, especially in more advanced stages of the disease [1,3]. Dilated cardiomyopathy occurs in adolescence, or in young adults who have previously been left without relevant clinical data or even with adequate medical treatment, and even after having received a liver transplant [1,4]. Its clinical manifestations require hospitalization in intensive care, circulatory support, and even heart transplantation [4,5]. We present the case of a young patient with propionic acidemia and severe heart failure secondary to dilated cardiomyopathy that required heart transplantation.

Clinical case

The patient is an 18-year-old male, weighing 56 kg and 1.7 m tall, blood group O positive, with a history of epilepsy since he was 4 years old. In September 2021, he presented with moderate COVID that required hospitalization during which pancreatitis, acidemia, hepatic encephalopathy with ammonium elevation, freckles on the face and anterior chest, gastroesophageal reflux disease were detected, with which he suspected and confirmed propionic acidemia with alteration of the PCCA gene, c.1268C>T (pathogenic) and c.2041-G>T (pathogenic). Cardiac involvement with dilated cardiomyopathy was also detected, which was not followed up and he only received treatment with oral levocarnitine, coenzyme

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Q, and oral biotin for life due to his metabolic disorder.

As of January 2023, he began with progressive functional class deterioration; with postprandial fullness and pain in the right upper quadrant of the abdomen. He was treated with prokinetics. He presented deterioration of his functional class to grade IV of the NYHA, fluid retention, dyspnea of small exertion, for which he was admitted to the hospital in April of the same year. In May, he required further hospitalization for acute decompensation of heart failure treated with an inodilator (levosimendam). The echocardiogram showed a left ventricular diastolic diameter (LVDD) of 66 mm and left ventricular systolic diameter (LVSD) of 60 mm with left ventricular ejection fraction (LVEF) of 15%, right ventricle (RV), with diameters of 48/45*68 TAPSE 14 mm and S1 0.08 cm/sec. The results of cardiac catheterization were: pulmonary artery trunk pressures (PTAP): 48/20 (30) mmHg, pulmonary capillary pressure (PcAP): 19 mm Hg, transpulmonary gradient (TPG) of 11 mmHg and pulmonary vascular resistance (PVR) 3.2 Wood units with cardiac output (CO) of 3.2 l/min. With the clinical evolution and results of the studies carried out, it is accepted to perform a heart transplant, which was performed on June 14, 2023 with a heart from a 17-year-old male donor and 60 kg and 175 cm tall, with compatibility by blood group, donor match: recipient by total heart mass of 7.6%, RV: 7.4% and LV: 7.6%, with total ischemia time: 225 minutes, aortic cross-clamping: 73 minutes, and cardiopulmonary bypass: 109 minutes. The postoperative outcome was favorable and required only milrinone and norepinephrine support at low doses, with no bleeding greater than usual.

Neurological recovery ad integrum, good ventilatory mechanics were withdrawn from mechanical ventilation and orotracheal decannulation 9 hours after admission to the Postsurgical Therapy Unit. The echocardiogram on the fourth postoperative day with a cardiac graft with 65% LVEF and VI cardiac dimensions: 38/24 mm, RV with TAPSE of 16 mm and S1 of 0.12 cm/sec, with a calculated PAH of 35 mmHg. During his hospital stay, there were no complications of hyperlactatemia, metabolic acidosis, neurological alterations, or seizures. Their diet was programmed at 0.8 g/kg/day of protein, supported by levocarnitine, cyanocobalamin intravenously and subsequently orally in conjunction with coenzyme Q. Their maximum ammonium levels were 100 mg and their minimum was 45 mg.

The immunosuppressive regimen was triple with steroids: methylprednisolone and then prednisone, tacrolimus and mycophenolate mofetil. Serum tacrolimus levels were 13.6 ng/ml. He had no renal, hemorrhagic, neurological or infectious complications. He was discharged from intensive care on the fifth day and went home with individual ambulation on the tenth day post-transplant. Endomyocardial biopsy 40 days after heart transplantation reported mild rejection of 1R (1-A) ISHLT 2012 and right catheterization with TAP: 35/12 (20 mmHg). Currently, five months after heart transplantation, the cardiac graft with LVEF 70%. RV is in functional class I NYHA: no data of acute rejection by echocardiogram.

Discussion

Dilated cardiomyopathy associated with propionic acidemia is a serious complication in which improvement has been seen when the patient receives a liver transplant, resulting in partial recovery of metabolic function [1], even in cases that have required ventricular support prior to liver transplantation [1,6]. However, given the low frequency of the pathology and despite having successful cases of liver transplantation, the complexity and even the need to combine liver-kidney transplantation, leave this option for few cases [7], since the risk of rejection reaction and immunosuppressive therapy is also added to the patient, so its indication must be carefully assessed in each case. A similar situation occurs in the presence of associated cardiomyopathy, which can be so severe that it also requires evaluating the need to use measures such as implantable cardioverter defibrillator therapy [8], and even the replacement of the heart by means of a transplant, as occurred in the case we present, where, despite carrying out an appropriate medical control, the patient evolved in the short term to severe heart failure, so it was decided to perform the transplantation, as Seguchi et al. had already done in a similar case [4], and which is also considered as an option in similar cases [9].

In the case of heart transplantation, perioperative care is essential, not only from the hemodynamic aspect. In addition, the need to avoid metabolic decompensation as much as possible, intravenous energy intake and early initiation of the oral route as recommended in the guidelines for the diagnosis and treatment of this pathology [9], as was done in this case, which is currently evolving satisfactorily. In our country, it would be the first case of propionic acidemia that he has received in a heart transplant and one of the few cases reported in the world.

Conclusion

We can conclude that propionic acidemia that progresses to severe heart failure, without any other clinical manifestation of the underlying disease, can be considered heart transplantation as a treatment option.

Declarations

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