



CARDIAC ARREST AND POSTPONED TRANSPLANTATION IN AN ADOLESCENT WITH UHL'S ANOMALY

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SUMMARY – We present a case of a 16-year-old male adolescent with Uhl's anomaly who initially suffered from cardiac arrest at the age of ten. Although his parents refused treatment at first, he managed to survive for five years without any surgical intervention before the heart transplantation was performed. It is the first case of an adolescent who underwent heart transplantation several years after resuscitation had been done due to Uhl's anomaly.

Keywords: *Uhl's anomaly; Heart transplantation; Cardiac imaging; Cardiac surgery*

Introduction

Uhl's anomaly is an extremely rare congenital heart defect of unknown cause characterized by the absence of the myocardium in the right ventricular free wall, which is replaced by nonfunctional fibroblastic tissue¹. In most cases, it is diagnosed prenatally or in newborns². The diagnosis can be suspected after clinical signs of right heart failure and can be made using echocardiography (ECHO), computed tomography (CT) and/or magnetic resonance imaging (MRI)^{2,3}. Definitive diagnosis is confirmed by histopathologic findings⁴. The main differential diagnosis is arrhythmogenic tachycardia of the right ventricle⁵. A case of a 16-year-old adolescent with six years between the time of cardiac arrest due to Uhl's anomaly and heart transplantation is presented. Both parents and the adolescent signed an informed consent form for this report.

Case Report

At the age of ten, the child experienced dyspnea and vertigo in the school yard. Emergency medical services were called. During transport to the hospital, the patient had cardiorespiratory arrest. He was intubated immediately upon admission, and an initial rhythm of ventricular fibrillation was noted. After successful resuscitation, the sinus rhythm reappeared. There were no neurological complications. Dilated cardiomyopathy of the right ventricle was observed during the first transthoracic ECHO (Fig. 1). Electrocardiography (ECG), as well as 24-hour ECG

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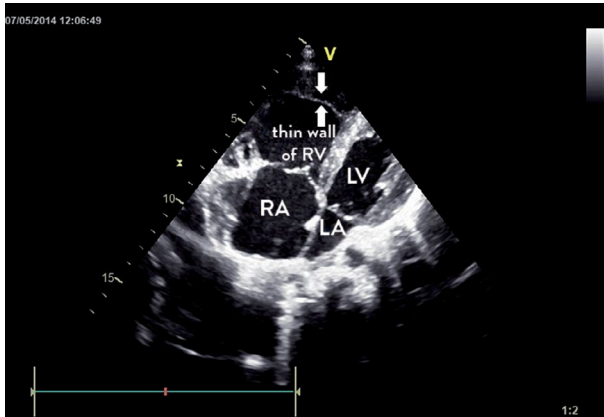


Fig. 1. Echocardiography of Uhl's anomaly at admission; RA = right atrium; RV = right ventricle; LA = left atrium; LV = left ventricle.

recording showed sinus rhythm, right atrial abnormalities, with several isolated ventricular extrasystoles; 90% of them originated from the right ventricle and 10% from the left ventricle. The antiarrhythmic treatment with digoxin and amiodarone was started. The first MRI of the heart showed significant dilatation and an impaired function of the right atrium and right ventricle. The right ventricle volume in diastole (RVEDV) was 245 mL (188 mL/m²) with ejection fraction (EF) of the right ventricle (RVEF) of 19%. The right ventricular wall was thin with an almost complete absence of the right ventricle free wall myocardium and without fibrofatty infiltration. Follow up heart MRI performed five months after the cardiac arrest showed progression in dilatation of right ventricle dimensions (Fig. 2). RVEDV was 412 mL (305 mL/m²) with RVEF of 13%. Functions of the left heart structures were within the normal range. Uhl's anomaly was the most probable diagnosis with arrhythmogenic dysplasia of the right ventricle (ARVD) as the differential diagnosis. The recommended biopsy, as well as insertion of an implantable cardioverter defibrillator (ICD) were rejected by the patient's parents. Metabolic diseases were excluded. Three and a half years after the cardiac arrest, he was dyspneic in everyday activities, exhibited peripheral edema, felt tired, and occasionally complained of tingling in the legs. The next follow up MRI of the heart was performed, showing the RVEDV of 525 mL (348 mL/m²) with RVEF of 25%. Fibrotic changes

of the left ventricle were also noticed. The function of the left heart structure was reduced (the left ventricle LVEF of 40%) with diminished left ventricular systolic volume of 31 mL/m², diastolic volume of 50 mL/m², and EF of 38%. Four years after the cardiac arrest, the patient was transferred to a collaborating institution for further MRI evaluation, where progression of the right ventricle dilatation was observed with global akinesia and a severely reduced systolic function of the right ventricle (RVEDV 420 mL/m², RVEF 18%)⁶. The parents finally gave consent for a heart transplant, which was performed in January 2019, almost five years after the cardiac arrest. One month later, the patient was discharged from the hospital without complications and with normal heart function (Fig. 3). The diagnosis of Uhl's anomaly was confirmed by histopathologic biopsy. Since then, five myocardial biopsies of the transplanted heart were performed, the last one in February 2020, without cellular or humoral rejection. A year and a half after the heart transplantation, the patient was stable and without major complications.

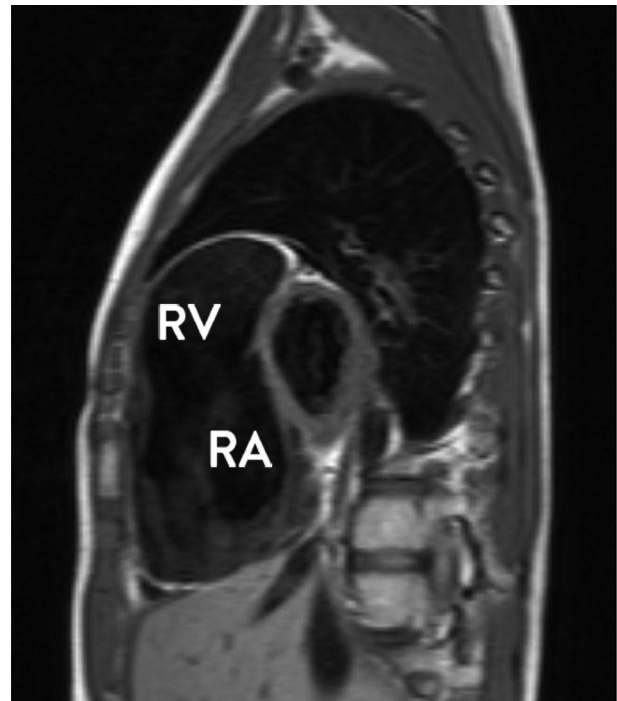


Fig. 2. Magnetic resonance imaging showing dilatation of the right side of the heart; RA = right atrium; RV = right ventricle.

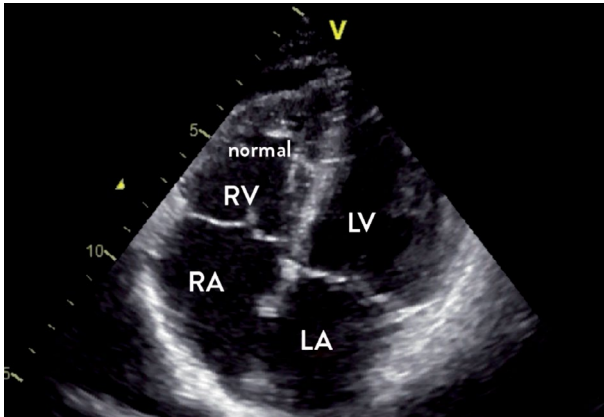


Fig. 3. Echocardiography after heart transplantation; RA = right atrium; RV = right ventricle; LA = left atrium; LV = left ventricle.

Discussion

The first case of Uhl's anomaly was reported in 1952^{1,2,7} after autopsy of an 8-month-old infant. In 1979, Fontaine *et al.* reported a similar entity called right ventricular arrhythmogenic dysplasia (ARVD), which is characterized by local deficiency of the ventricular myocardium². Sometimes it may be hard to determine difference between these two conditions². Clinically, the first manifestation of Uhl's disease usually is heart failure, while ARVD patients mostly experience arrhythmic events and sudden death. In ARVD, the myocardium is progressively replaced by fibrofatty tissue, and the condition has been linked to mutations in the genes encoding plakoglobin and desmoplakin⁸. On the other hand, in patients with Uhl's anomaly, there is a partial or total loss of the myocardial muscle tissue in the right ventricle². This can be the result either of primary non-development of myocytes in the right ventricle, or it can be due to selective apoptosis¹. Patients with Uhl's anomaly rarely survive to adulthood^{1,2}. There are only a few case reports of successful surgical treatments after the infant period⁴. The oldest reported patients with Uhl's anomaly are two patients, a female and a male, both aged 62^{2,9}.

There are three main surgical approaches for these patients. The first one is based on the exclusion of the right ventricle by closing the tricuspid valve orifice

with atrial septectomy and a bidirectional Glenn shunt⁷. The second option is a 'one-and-a-half ventricular repair' in which the first approach is combined with partial right ventriculotomy^{4,7,10}. The third option is heart transplant, described for the first time in an 11-month-old infant^{5,10}. In our case, Uhl's anomaly was suspected, but because of the initial presentation with malignant arrhythmia and regional right ventricle dyskinesia, as proved by MRI, ARVD was also considered as a possible diagnosis². The planned diagnostic and therapeutic procedures including catheterization, ICD insertion and heart surgery, were initially rejected by the patient's parents. Although he had severe signs and symptoms of the right heart failure and was often hospitalized, he managed to survive for five years after cardiac arrest. In this period, routine heart MRI and ECHO examinations revealed progressive deterioration of the right ventricular function, and afterwards deterioration of the left ventricular function. The parents finally gave consent for the surgical procedure. Because of a major right heart and moderate left heart failure, we decided that cavopulmonary shunts could not be sufficient for the normal functioning of the heart and therefore opted for heart transplantation. A similar case has been reported by Chounoune *et al.*, where they describe a 12-year-old male patient who also presented with cardiac arrest¹¹. In their case, the parents refused heart transplantation. A repair on one and a half ventricles was performed, with a good outcome. In their case, the function of the left ventricle was normal, thus they had an option for univentricular heart circulation, or one-and-a-half repair. Conversely, in our case, when one and a half ventricle repair was an option, the parents refused the operation. In further period, although not usual in patients with Uhl's anomaly, the left ventricular function deteriorated, which was why we gave up the Fontan operation. In conclusion, Uhl's anomaly is a rare condition, and it is usually treated surgically shortly after the diagnosis. The best treatment option is still a matter of discussion. This is the first reported case of an adolescent patient receiving a heart transplant five years after the diagnosis. This case report shows that a patient can survive for a few years with significant right heart failure and without surgical treatment. Heart transplantation seems to be the only feasible option after this period.

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Sažetak

SRČANI ZASTOJ I ODGOĐENA TRANSPLANTACIJA U ADOLESCENTA S UHLOVOM ANOMALIJOM

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Prikazujemo slučaj 16-godišnjeg adolescenta s Uhllovom anomalijom koji se inicijalno, u dobi od 10 godina, prezentirao srčanim zastojem. Iako su njegovi roditelji isprva odbijali liječenje, uspio je preživjeti pet godina bez ikakve kirurške intervencije prije nego što je izvršena transplantacija srca. To je prvi slučaj adolescenta koji je podvrgnut transplantaciji srca nekoliko godina nakon što je obavljena reanimacija zbog Uhllove anomalije.

Ključne riječi: *Uhllova anomalija; Transplantacija srca; Ultrazvuk srca; Kardiokirurgija*