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CT of Uhl's anomaly in an adult

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Abstract

Uhl's anomaly is an extremely rare condition of unknown cause characterized by complete or partial absence of the right ventricular myocardial layer, which is replaced by nonfunctional fibroelastic tissue. The disease causes progressive right-sided heart failure, increased right-sided cardiac pressure, massive peripheral edema, and ascites. Patients usually present in infancy and rarely survive to adulthood. The disease appears to be congenital in origin. Diagnosis was previously made at autopsy, but advances in imaging now permit diagnosis by echocardiography or cross-sectional imaging (computed tomography or magnetic resonance). We present a case of a 51-year-old patient with Uhl's anomaly who underwent placement of a ventricular assist device as a bridge to cardiac transplantation, and discuss CT findings.

Introduction

Uhl's anomaly is an extremely rare condition of unknown cause characterized by complete or partial absence of the right ventricular myocardial layer, which is replaced by nonfunctional fibroelastic tissue. The disease causes progressive right-sided heart failure, increased right-sided cardiac pressure, massive peripheral edema, and ascites. Patients usually present in infancy and rarely survive to adulthood. The disease appears to be congenital in origin. Diagnosis was previously made at autopsy, but advances in imaging now permit diagnosis by echocardiography or cross sectional imaging (computed tomography or magnetic resonance). We present a case of a 51-year-old patient with Uhl's anomaly who underwent placement of a ventricular assist device as a bridge to cardiac transplantation, and discuss CT findings.

Case report

As a 1-month-old boy, the patient developed symptoms of fever, tachypnea, and cyanosis. By age 13, he was noted to have cardiomegaly and catheterization showed identical pressure wave forms in the right atrium, right ventricle, and pulmonary artery. Cine images showed a dilated noncontracted right ventricle, and the patient was diagnosed with Uhl's anomaly [1].

At age 51, he presented with NYHA Class IV congestive heart failure and recurrent ascites, requiring paracentesis every 2 weeks of 6–10 l of fluid. One year earlier, he had experienced a near fatal malignant ventricular arrhythmia. The

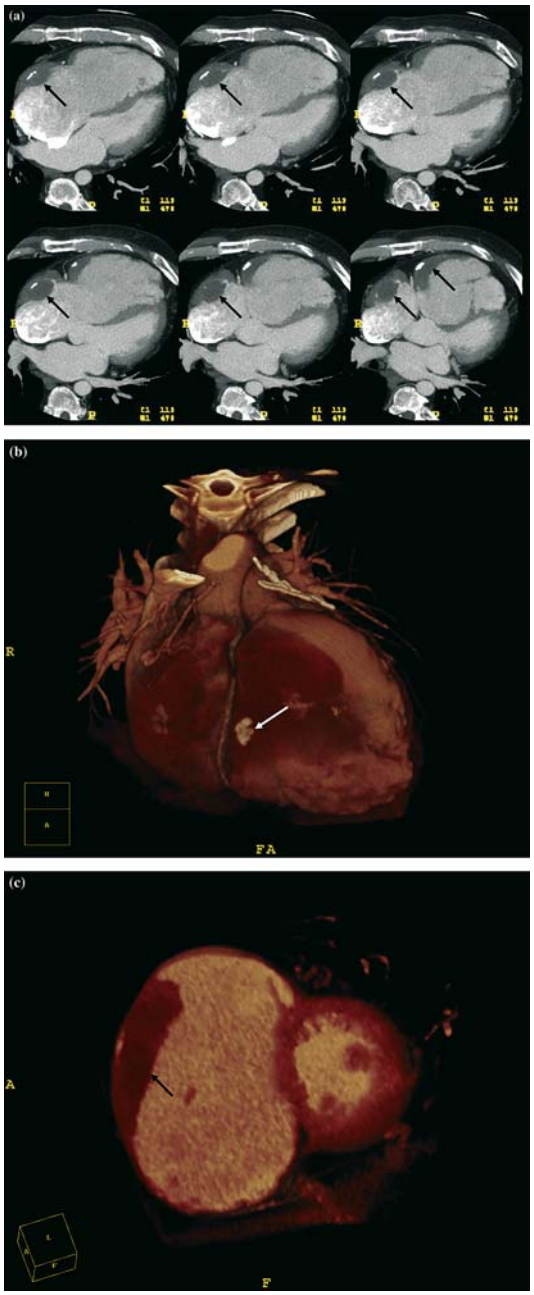

Figure 1. A 51-year-old male with Uhl's anomaly. (a) Sections from an axial contrast-enhanced CT show massively dilated right ventricle and right atrium. Abnormal low density contains foci of calcifications, representing chronic mural thrombus (arrows). (b) Volumetric CT reconstruction shows dilated right ventricle and a focus of calcium (arrow). (c) Short axis multiplanar reconstructed CT image shows dilated right ventricle and right atrium with thrombus (arrow).

patient was evaluated for cardiac transplantation, but liver congestion and refractory ascites prevented him from being a suitable candidate at that time. Thus, a right-sided ventricular assist device was placed in an effort to reduce liver congestion as a bridge to future cardiac transplantation.

ECG-gated multislice cardiac CT (MX8000IDT, Philips Medical Systems, Best, Netherlands) was performed for pre-operative evaluation after injection of 150 cc of contrast material at 4 cc/s. One-millimeter thick sections were acquired axially and multiplanar and volumetric images were reconstructed. The images revealed a massively dilated right ventricle and right atrium (Figure 1). Abnormal low density representing chronic mural thrombus and containing punctate calcifications was demonstrated within the anterior aspect of the right atrium, ventricle, and infundibulum. The mean attenuation is 26 HU. Cine images (not shown) revealed absence of motion in the right ventricular free wall. The left ventricular myocardium was unaffected and wall motion was normal.

The patient was taken to the operating room for placement of a ventricular assist device at which time a massively dilated heart was found with findings consistent with Uhl's anomaly. The patient died postoperatively secondary to sepsis and autopsy of the heart confirmed the diagnosis of Uhl's anomaly. The right ventricle wall was replaced mostly by adipose tissue, with a few myocardial cells.

Discussion

In 1905, William Osler reported a heart with thin walls he described as 'parchment-like'. Henry Uhl reported his first case in 1952 after performing an autopsy on an 8-month-old infant [2]. That year, two articles were published describing an infant and an adult, respectively, both with virtually complete loss of myocardium in the right ventricle with replacement by fibro-elastic nonfunctional tissue [2, 3]. The etiology of Uhl's anomaly remains unknown, but is currently thought to result from selective apoptosis of right

ventricular myocytes after complete cardiac development [4].

Over the past century, fewer than 100 cases of Uhl's anomaly have been reported. Since 1952, many cases have been described combining Uhl's anomaly and other pathological conditions of the heart. Eponyms used to describe this rare cardiac anomaly include right ventricular ectasia, congenital right ventricular myocardial aplasia, fat infiltration or lipomatosis, right ventricular idiopathic myocardial dysplasia and right ventricular myocardial absence.

Although often considered together, recent studies suggest that Uhl's anomaly is a different disease than arrhythmogenic right ventricular dysplasia (ARVD), with distinct morphological and clinical features, such as age, sex predilection, familial history and clinical presentation. Patients with Uhl's anomaly typically present as an infant or during childhood, may be of either gender, and have no familial history. Congestive heart failure is common. In contrast, patients with ARVD usually present as an adolescent or adult, with a 3:1 male predominance. A family history of ARVD and exercise-induced sudden death are more frequent in ARVD [5]. Typical symptoms are arrhythmia, syncope, or sudden death. Morphologically, fibroelastic tissue separates the right ventricular endocardial and epicardial layers in Uhl's anomaly differentiating it from ARVD, in which fibrofatty tissue separates the endocardial and epicardial layers. [6]

As is typical of Uhl's anomaly, our patient had no familial history and presented with refractory congestive heart failure, necessitating an orthotopic cardiac transplantation. Survival is poor following heart transplantation for Uhl's anomaly and our patient expired 9 days after surgery due to sepsis.

Prior to the advent of cross-sectional imaging, most patients diagnosed with Uhl's anomaly died and diagnosis was confirmed at autopsy. However, in the 1970s and 1980s, the development of echocardiography, angiocardiography, and electrocardiography created the potential for premortem diagnosis [4]. With the development of CT and MRI, this rare cardiac pathology has become more readily diagnosable in the proper clinical setting.

MRI characteristics include an extremely thin-walled right ventricle with almost complete absence of right ventricular free wall myocardium, a paucity of apical trabeculations, and normal left ventricular myocardium. On MRI, the right atrium is dilated and hypertrophied as a consequence of the right ventricular restrictive cardiomyopathy and dependence on atrial contraction to augment pulmonary artery forward flow [7]. Nevertheless, imaging by MRI is somewhat limited by its contraindication for use in patients with pacemakers and the inability to detect calcified thrombus which may affect precise delineation of the extent and nature of the thrombus.

In the present case, we elected to use multislice CT rather than MRI to maximize spatial resolution prior to potential transplantation. CT images showed a massively dilated right ventricle and right atrium with thin walls. The right ventricular myocardium was not identified, compatible with the patient's history of Uhl's anomaly. The left ventricular myocardium appeared normal. Chronic mural thrombus was visualized well on enhanced CT within the anterior aspect of the right cardiac chambers. Wall motion abnormalities also were accurately depicted. To our knowledge, only a single case of Uhl's anomaly has been demonstrated using multislice CT [8]. In summary, contrast-enhanced ECG-gated multislice CT scanning provides an accurate assessment of the morphology and function of the heart in Uhl's anomaly and is valuable in surgical planning.

References

1. Zuberbuhler JR, Blank E. Hypoplasia of right ventricular myocardium. Am J Roentgenol Rad Ther Nucl Med 1970; 110(3) 491–496.
2. Uhl HS. A previously undescribed congenital malformation of the heart: almost total absence of the myocardium of the right ventricle. Bull Johns Hopkins Hosp 1952; 91: 197–209.
3. Castleman B, Towne V. Presentation of case 38201. N Engl J Med 1952; 256: 785–790.
4. Uhl HS. Uhl's anomaly revisited. Circulation 1996; 93(8) 1483–1484.

5. Loire R, Tabib A. Arrhythmogenic right ventricular dysplasia and Uhl disease. Anatomic study of 100 cases after sudden death. Ann Pathol 1998; 18: 165–171.
6. Marcus FI. Is arrythmogenic right ventricular dysplasia, Uhl's anomaly and right ventricular outflow tract tachycardia a spectrum of the same disease?. Cardiol Rev 1997; 5: 25–29.
7. Greer ML, MacDonald C, Adatia I. MRI of Uhl's Anomaly. Circulation 2000; 101(24) e230–e232.
8. Ceviz N, Kantarci M, Okur A. Electrocardiographic gated multislice computed tomography of Uhl's Anomaly. Br Heart J 2004; 8: 886.

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