UHL'S ANOMALY IN A 60-YEAR-OLD ADULT FEMALE PRESENTING WITH MASSIVE DILATATION OF THE RIGHT VENTRICLE

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INTRODUCTION

Uhl's anomaly is an extremely rare cardiac malformation. Most patients are diagnosed prenatally or in infancy and rarely survive to adulthood. There is no ideal treatment and patients have poor prognosis and high mortality.

CASE PRESENTATION

A 60-year-old woman was referred for evaluation of presumed pulmonary hypertension. She reported progressive shortness of breath, fatigue and lower extremity (LE) edema over the past year. Her past medical history included bullous emphysema, hypertension, PE/DVT, hypothyroidism, osteoporosis, and 35 pack year smoking history.

Physical exam revealed prominent right ventricular (RV) heave, 4/6 systolic murmur in left parasternal area with mildly elevated closure sound of pulmonic valve and 2+ pitting LE edema.

INVESTIGATIONS

TTE revealed:

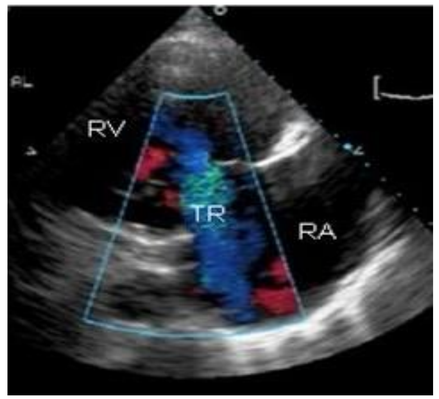
Marked right atrial (RA) and RV dilatation with thinned RV wall. Severe RV dysfunction. Preserved left ventricular ejection fraction (LVEF) between 55-60%. Severe tricuspid regurgitation (TR).

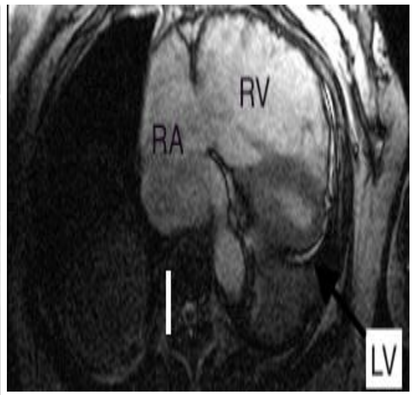
Right cardiac catheterization:

Pulmonary artery pressure: 52/25 mmHg, mean 33 mmHg. Pulmonary capillary wedge pressure: 12 mmHg. Cardiac Output: 2.9 L/min, Cardiac Index: 1.6L/m2. Right ventriculogram: marked RV dilatation with severe dysfunction. Severe TR.

Left cardiac catheterization:

Normal LV diastolic pressure. Normal coronary arteries.





ADVANCED IMAGING

Cardiac magnetic resonance imaging with and without gadolinium contrast to evaluate for RV dysplasia or infiltrative heart disease revealed; Marked RV dilatation with thinned wall. Severe RA enlargement. Severe tricuspid annulus dilatation with severe TR. Preserved LVEF of 55-60% No evidence of hyperenhancement or fibro-fatty infiltration

MANAGEMENT

Patient was diagnosed with Uhl's anomaly and started on captopril, furosemide and low dose carvedilol. Due to lack of clinical improvement, low dose dobutamine infusion and intravenous diuretics were initiated. Patient improved after 3 days on this therapy with complete resolution of symptoms. A Hickman catheter was implanted for continuous dobutamine infusion, and an implantable cardioverter defibrillator (ICD) was placed.

Cardiac transplantation work-up was initiated and patient was discharged home. 2 days after discharge, patient had cardiac arrest and died. ICD interrogation revealed an appropriate shock with a terminal rhythm of ventricular fibrillation.

DISCUSSION

First described by Dr. Henry Uhl in 1952, Uhl's anomaly is an extremely rare cardiac malformation characterized by complete or partial absence of the right ventricular (RV) myocardium, which is replaced by parchment-like nonfunctional fibroelastic tissue. The myocardium may be affected focally or diffusely through the RV free wall. It was previously considered to be a congenital development failure in early stages of embryogenesis, leading to the absence of the RV myocardium. However, more recent observations implicate apoptotic destruction of the myocardium after the heart has fully developed. Nevertheless, the etiopathogenesis still remains uncertain. It is rarely associated with other cardiac malformations. Congestive cardiac failure is the most common mode of presentation, but arrhythmias or heart block can also be seen in these patients.

DISCLOSURES

Authors have no disclosures.

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