Right ventricular myxoma and pulmonary embolism in an adolescent with Carney complex

Lauren E. Parker, BS,a Cathlyn K. Medina, BA,a Berk Aykut, MD,a Jennifer Sherwin, MD,b Kimberly Jackson, MD,b Christopher Atkins, MD,b Katherine Cashen, MD,b Douglas M. Overbey, MD,a Joseph W. Turek, MD, PhD, MBA,a and Ziv Beckerman, MD,a Durham, NC

CASE DESCRIPTION

This case report is approved under Pro00101549 (approved January 3, 2019) with waiver of individual patient consent. A 14-year-old female patient (60.0 kg with body surface area = 1.73) with a history of macroprolactinoma status after transsphenoidal resection, presented to the emergency department with 1 day of chest pain and shortness of breath. On arrival, computed tomography imaging demonstrated bilateral pulmonary embolism. A large right ventricular (RV) mass was also visible on computed tomography (Figure 1). The patient was anticoagulated with heparin and transferred to our institution for surgical evaluation.

Upon arrival, she was hemodynamically stable and with normal oxygen saturations. Transthoracic echocardiography revealed a large RV mass involving the ventricular free wall, obstructing the RV outflow tract and abutting the pulmonary valve. Echocardiography further revealed mild tricuspid regurgitation with a peak gradient of 40 to 50 mm Hg and trace pulmonary valve regurgitation. RV and left ventricular function were grossly normal. Cardiac magnetic resonance imaging again demonstrated the large intracavitary RV mass measuring 6.2 × 2.9 × 3.2 cm with a stalk attaching to the tricuspid valve apparatus. The mass was heterogeneous in appearance on steady-state free precession, T1-, and T2-weighted sequences. There was no late gadolinium enhancement or fat saturation. Together, preoperative imaging suggested RV myxoma.

OPERATIVE DETAILS

The patient was brought to the operating room for mass excision and pulmonary embolectomy. A median sternotomy was performed, systemic heparin was administered, and the ascending aorta, superior vena cava, and inferior vena cava were cannulated for bypass. Cardiopulmonary bypass was initiated, and the patient was cooled to 32 °C. An aortic crossclamp was placed and antegrade cardioplegia was administered into the aortic root. An oblique right atriotomy was performed and a small atrial septal defect was created to serve as a left heart vent. The RV was visualized through the tricuspid valve, revealing a large, yellow-red mass with smooth margins, attached to the ventricular free wall. Working through the tricuspid valve, the mass was mobilized and the stalk attaching to the RV free wall was excised (Figure 2). The mass was then delivered through the tricuspid valve en bloc. The base of the stalk was further resected until normal-appearing myocardium was reached, and then sent for frozen tissue sections.

CENTRAL MESSAGE

Cardiac myxoma in a young patient with history of endocrine tumors should raise suspicion for Carney complex.

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CASE REPORT

Video clip is available online.

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Tricuspid valve competency was confirmed. Surgical pathology demonstrated clear margins at the stalk base. The right pulmonary artery was opened and a 1.8 × 1.0 × 0.8 cm mass with the same consistency as the RV mass was extracted (Video 1). After confirming absence of additional emboli, the right pulmonary artery was closed. The atrial septal defect was then closed. The left heart was de-aired and the crossclamp removed. The right atriotomy was closed and the patient was rewarmed. Postoperative transesophageal echocardiography showed no residual mass, good biventricular function, and no tricuspid regurgitation or pulmonary insufficiency.

The patient was extubated and weaned from supplemental oxygen on the day of surgery. The mediastinal drain was removed, and the patient was transitioned to the step-down unit on postoperative day 2 and discharged from the hospital on postoperative day 4.

GENETICS EVALUATION

Given the patient’s history of macroprolactinoma and presence of skin and mucosal lentigines, the pediatric endocrinology team was consulted preoperatively for clinical suspicion of Carney complex. Carney complex is a rare, autosomal dominant syndrome with roughly 750 cases reported since described in 1985.1 Carney complex is caused by inactivating mutations or deletions of PRKAR1A-encoded protein kinase cyclic adenosine monophosphate-dependent type 1 regulatory subunit alpha.1 It is characterized by the development of benign tumors, including myxomas, skin lesions, and multiple endocrine tumors, including pituitary and adrenocortical tumors.

A diagnosis of Carney complex is made by presence of 2 major diagnostic criteria or 1 major and 1 minor criteria. In this patient, her history of hyperprolactinemia and intense skin and mucosal lentigines supported a diagnosis of Carney complex.
freckling constitute 2 minor criteria, whereas cardiac myxoma constitutes a major criterion. Genetic testing identified a heterozygous intronic c.502+3A>C variant of uncertain significance in the PRKAR1A gene. Based on these findings, a diagnosis of Carney complex was made. This has received outpatient follow-up with a multidisciplinary care team, including pediatric cardiology, dermatology, ophthalmology, and endocrinology.

**COMMENT**

In this case report, we describe successful surgical management of a 14-year-old patient with an unusual presentation of bilateral pulmonary embolism secondary to RV myxoma and Carney complex. Pediatric cardiac myxomas are rare, with RV myxomas being the rarest subtype. Moreover, this patient’s history of macroprolactinoma, diagnosis of RV myxoma, and pigmented skin lesions ultimately led to clinical suspicion of Carney complex, which was validated by genetic testing. Young patients presenting with cardiac myxoma may benefit from undergoing endocrine evaluation to assess for Carney complex.

**Conflict of Interest Statement**

The authors reported no conflicts of interest.

The Journal policy requires editors and reviewers to disclose conflicts of interest and to decline handling manuscripts for which they may have a conflict of interest. The editors and reviewers of this article have no conflicts of interest.

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