

## CASE REPORT

# Excision of Congenital Rhabdomyoma of the Left Outflow Tract through the Left Atrium and Mitral Valve

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## ABSTRACT

Primary rhabdomyomas obstructing the right or left outflow tract are uncommon findings in the perinatal period. The presenting symptom may be arrhythmia, cardiac murmur, complete or variable atrioventricular block, pericardial effusion, cardiomegaly, cardiac failure, or sudden death. The variety of symptoms can be explained on the basis of obstruction of blood flow, myocardial involvement, and disturbance of the cardiac rhythm. Commonly, rhabdomyoma spontaneously regresses in the majority of cases, and neonatal surgery is advocated only in case of severe left ventricular outflow tract (LVOT) obstruction or the development of arrhythmias. Herein, we describe the pathologic and clinical characteristics of neonatal presentation of a cardiac rhabdomyoma with a nearly obstructive mass in the LVOT, discovered during pregnancy and operated 4 months after delivery.

**Key words:** Left ventricular outflow tract obstruction; Neonatal tumors; Rhabdomyoma

## INTRODUCTION

Most fetal and newborn cardiac tumors are histologically benign. Compared to adults, primary tumors are found much more often than metastasis to the heart. Rhabdomyomas comprise most of the perinatal tumors.

In a 1997 study by Beghetti *et al.*, the most common tumor in infants was rhabdomyoma, with 44 examples (78%), followed by four fibromas, one teratoma, and one multicystic hamartoma of the interatrial septum. The fetal cardiac tumor study of Holley *et al.*, again, rhabdomyomas were the most common fetal tumor (17/19 or 89%) followed by one each of ventricular fibroma and atrial hemangioma [1,2].

Cardiac rhabdomyoma is the most common primary cardiac tumor in infants and children, and it is commonly associated with tuberous sclerosis. Usually, the presence of multiple cardiac rhabdomyomas prenatally may be the earliest manifestation of tuberous sclerosis complex. Clinical and hemodynamic findings

are related to the number, position, and size of the tumors and may cause arrhythmias, infant respiratory distress, cyanosis, or congestive heart failure. At echocardiography, rhabdomyoma appears as a homogeneous well-circumscribed echogenic mass in the ventricular myocardium. In the literature, a partial or complete spontaneous regression is reported in up to 50% of cases after birth, allowing conservative management for this kind of neoplasia with continuous echocardiography and electrocardiography monitoring. Surgical intervention should be reserved only for sick patients with symptoms of severe obstruction and hemodynamic compromise or intractable arrhythmias [3,4]. We report a case of primary cardiac rhabdomyoma of the left ventricle, operated 4 months after birth, with an antenatal follow-up.

## CASE REPORT

A level II obstetric ultrasound performed at 30 weeks' gestation on a mother revealed a large round mass of approximately 20 mm × 10 mm, with possible left ven-

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tricular outflow tract (LVOT) obstruction. Remote anamnesis showed a history of tuberous sclerosis (mother and sister). The subsequent fetal echocardiography at 34 weeks' gestation revealed a moderately obstructive LVOT mass, but no evidence of ventricular dysfunction, fetal hydrops, or hemodynamic compromise.

At the time of birth, the neonate, male, 3.00 kg, born from natural delivery, Apgar 9, was clinically asymptomatic. Echocardiography confirmed the presence of a large, homogenous subaortic mass arising from the interventricular septum and protruding the LVOT without a gradient (systolic gradient 10 mmHg). Therefore, the decision was to let the patient grow and carefully follow him up with echocardiography, looking for an increase in subaortic gradient (Figure 1).

At the age of 4 months, the echocardiography showed a severe increase of the pressure gradient in the LVOT (systolic gradient 58 mmHg). Due to the pressure gradient and the high risk of embolization of the mass that showed at that time only a small base pedicle keeping the neoplasm attached to the interventricular septum, the baby underwent a surgical operation for neoplasia excision.

Standard bicaval cardiopulmonary bypass (CPB) was established, and the operation was performed with mild hypothermia (32–33°C) and cardiac arrest was achieved using crystalloid cardioplegia (Custodiol). Careful manipulation of the heart was performed during cannulation and cardioplegia administration to avoid embolism and coronary ischemia, due to mass dislodgment in LVOT. The surgical approach was performed from the left atrium through the mitral valve. The inspection revealed almost a complete obstruction of LVOT by a single pedicle mass surrounded by a fibrous capsule, originated from the apical portion of the interventricular septum. Excision was performed only throughout the left atrium and the mitral valve, due to the dimension of the neoplasm compared with the aortic annulus. Mitral valve competence was checked with water test after tumor excision (Figure 2). CPB after 48 min was discontinued without complication, and patient was sent to the ICU.

At gross examination, the neoplasm showed a 14 mm × 10 mm well-circumscribed and encapsulated nodule of homogeneous whitish-grey tissue. Microscopic examination revealed the typical aspect of cardiac rhabdomyoma consisting of roundish enlarged myocytes with cleared vacuolated cytoplasm and sparse myofilaments and some spider cells. Positive desmin-immunohistochemistry proved the striated muscle nature of the cells (Figure 3).

The infant's post-operative recovery was unremarkable. A post-operative echocardiogram showed an

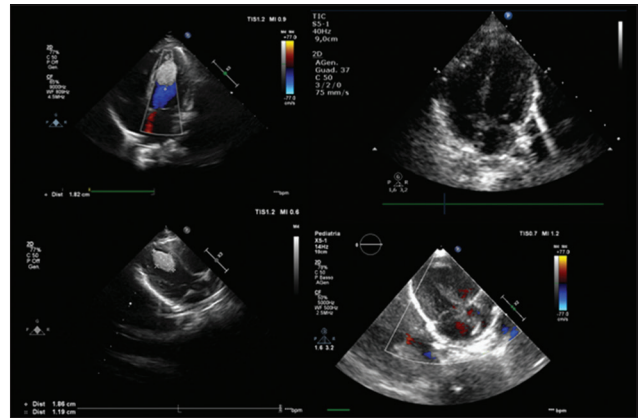


Figure 1: Pre-operative and post-operative echocardiogram compared

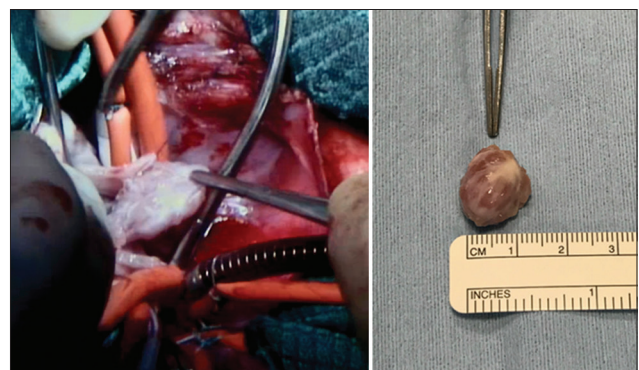


Figure 2: Intraoperative approach and view of the mass

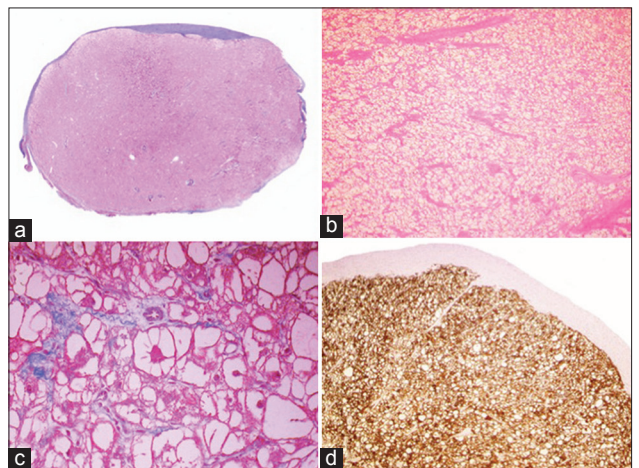


Figure 3: The nodule is almost surrounded by an incomplete fibrous capsule (a), Azan Mallory trichrome). The enlarged cleared myocytes are immersed in a slim collagen stroma (b), hematoxylin-eosin, ×100). (C), it is evident that the majority of myocytes show sparse filaments within the vacuolated cytoplasm (asterisks); in some cells, fibrillary eosinophilic strands radiate from a central nucleus to the cell membrane, resembling a spider ("spider cells"). (d) Diffuse desmin immunohistochemistry positivity

excellent EF (55%), no residual mass within the left ventricle, and mitral valve showed a trivial

insufficiency. The infant was discharged on post-operative day 11 in excellent condition. The patient started regular growth pattern and is in excellent clinical condition at 3-month follow-up (Figure 1).

## DISCUSSION

Primary cardiac tumors in children are most of the time benign while the malignants are approximately 10%. The most common benign cardiac tumor in children is rhabdomyoma, which is frequently associated with tuberous sclerosis, and often, cardiac rhabdomyoma is the earliest manifestation of this disease [5]. Even in the absence of multiple tumor masses, the presence of a strong familial history of tuberous sclerosis leads us to a suspicious of rhabdomyoma in our case.

Clinical presentation can vary from an incidental finding to congestive heart failure. Rhabdomyomas are also associated with cardiac arrhythmias including atrial and ventricular arrhythmia and the Wolff-Parkinson-White syndrome.

The natural history of cardiac rhabdomyoma leads to a spontaneous regression (in the literature, a partial or complete spontaneous regression is reported in up to 54% of cases) [6], and most incidentally found tumors can undergo surveillance with echocardiography. Indications for resection include outflow tract or cavitory obstruction and intractable arrhythmia [7].

Rhabdomyomas are most frequently found in the ventricles and occur more frequently in the left ventricle than in the right ventricle (100% vs. 81%) [8], where they can affect ventricular function and sometimes impair valve function or obstruct inflow/outflow tracts. The natural history of cardiac rhabdomyoma is the spontaneous regression that could be complete in more than 50% of patients during the 1<sup>st</sup> year. On the other hand, fetal rhabdomyoma may grow during the second and third trimester, resulting in a hemodynamically significant obstructive lesion which could lead to death in the neonatal period [9].

The management of these tumors is usually conservative because of the benign nature of the latter; the difficulties in eradicating them are the location in the deep myocardium and the high regression rate. Excision is not usually considered unless they cause severe symptoms, and nowadays, medical treatment with sirolimus or everolimus (mTOR inhibitors) has been advocated in selected cases [10]. Our patient demonstrated an unusual presentation of a neonatal rhabdomyoma that caused progressive risk of critical LVOT obstruction. Since the first diagnosis, we followed up the mother and the fetus every month. The early identification with fetal echocardiography enabled our team to plan a scheduled follow-up with an echocardiogram and leads to a postnatal treatment with lifesaving surgical resection of the obstructive mass.

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## Author's contribution

All authors contributed equally in concept, design, literature review, drafting the manuscript, and approval of the final manuscript.

## Consent statement

Authors declared that they have taken informed written consent, for publication of this report along with clinical photographs/material, from the legal guardian of the patient with an understanding that every effort will be made to conceal the identity of the patient however it cannot be guaranteed.

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