

## Case Report

# Huge cardiac rhabdomyoma in neonate: an unusual presentation

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### ABSTRACT

Cardiac rhabdomyoma is the most common primary cardiac tumor in neonatal age group. A full term male neonate, whose antenatal ultrasound revealed mass in foetal heart, became symptomatic on day two of life. Echocardiography revealed pericardial mass with global left ventricular hypokinesia. Diagnosis was confirmed by histopathological examination of autopsy specimen. Due to social stigma, encouragement of the parents for clinical autopsy was of prime importance for definitive diagnosis and preventive measures in future pregnancies.

**Keywords:** Cardiac tumors, Echocardiography, Rhabdomyoma, Tuberous sclerosis

## INTRODUCTION

Primary cardiac tumors are unusual in neonate, most of them are rhabdomyomas. Intracardiac tumors are very rare in neonates and infants and are mostly teratomas. The incidence of cardiac rhabdomyoma varies from 0.002% to 0.25% at autopsy, 0.02 to 0.08% in live born infant and 0.12% in prenatal period.<sup>1</sup> Now a days, rhabdomyoma is increasingly diagnosed because of improvement in prenatal diagnostic facilities. It is usually associated with tuberous sclerosis, an autosomal dominant disorder that results in abnormal cellular proliferation and differentiation which are responsible for hamartomas that involve brain, kidneys, lungs and skin.<sup>2,3</sup> Though cardiac rhabdomyomas are known to regress spontaneously, hemodynamic impact depends on location and size of mass and the presence of arrhythmias, systolic and diastolic dysfunction and ventricular mechanical obstruction which can even lead to death.<sup>4</sup> Here we describe a case of cardiac rhabdomyoma with severe left ventricular dysfunction leading to death in early neonatal period.

## CASE REPORT

A full term male neonate, born of non-consanguineous marriage, to primigravida mother by caesarean delivery, whose antenatal ultrasonography at 30 weeks of gestation revealed mass in foetal heart. The baby cried immediately after birth, physical examination was normal and baby was asymptomatic at birth. However, in view of antenatally diagnosed cardiac mass, baby was admitted in the NICU for observation.

At 26<sup>th</sup> hour of life baby developed respiratory distress (RR 72/min, nasal flaring, and chest retraction), tachycardia (heart rate 176/min) and prolonged capillary refill time. Peripheral pulses were palpable but feeble. On auscultation, heart sounds were normal. Liver was palpable 4 cm below costal margin. Baby was treated as per our NICU protocol with decongestive and inotropic agents. Chest X-ray revealed huge cardiomegaly occupying whole thoracic cage (Figure 1). A two dimensional echocardiography showed pericardial mass with global left ventricular hypokinesia (Figure 2). Cardiac MRI could not be performed because of

hemodynamic instability. In spite of supportive therapy baby's condition worsened and baby expired on fourth day of life.



**Figure 1: Chest radiograph showed cardiomegaly.**



**Figure 2: Echocardiography showed huge mass in left ventricle.**

Partial autopsy was performed and on gross examination, heart was enlarged and distorted in shape. Mass was well circumscribed, solid, fleshy, and homogenous and tan colored. On cut surface, a 5×4×2.5 cm mass was seen completely occupying the left ventricle. The mass was obliterating the left ventricle and causing obstruction to outflow and inflow tract. It was also compressing and narrowing the right ventricular chamber (Figure 3). Histopathological examination revealed normal myofibrils in periphery, centrally placed oval nuclei and eosinophilic granular cytoplasm having thin radiating process, comprising typical features of spider cells (Figure 4). Because of known association of rhabdomyoma with tuberous sclerosis, family members were screened for tuberous sclerosis but no stigmata was found.

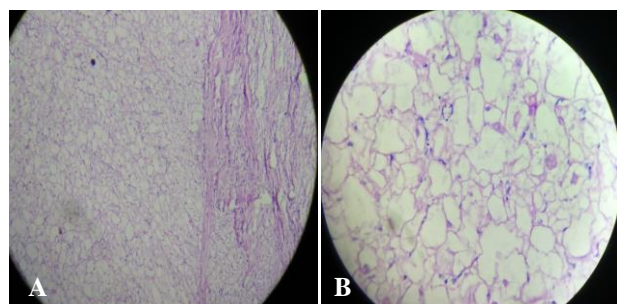
## DISCUSSION

Neonatal cardiac tumors are rare entities. Kwiatkowska et al reported 73% cases of rhabdomyoma in a series of 30 children of cardiac tumors over 20 years.<sup>5</sup> The etiology of

rhabdomyoma is unknown. The maternal estrogen is thought to be responsible for growth of tumor in utero. It resembles a hamartoma derived from embryonal myoblasts. It is thought that the tumor is caused by mutation in the TSC1 and TSC2 genes. So, they are usually associated with tuberous sclerosis with hamartoma formation in brain, kidneys, lungs and skin. Parents and family members of our case were not found to have stigmata of tuberous sclerosis. Rhabdomyomas in neonate should be differentiated from other cardiac tumors such as cardiac myxoma, teratoma, hemangioma and fibroma.<sup>6</sup> They are usually diagnosed in prenatal life or during early infancy by prenatal ultrasound or fetal echocardiography. Our case was diagnosed at 30 weeks of gestation by prenatal sonography.



**Figure 3: Gross and cut surface of cardiac mass.**



**Figure 4: A) and B) histological features of cardiac rhabdomyoma.**

The symptomatology can vary greatly from the absence of any symptoms to even life threatening complications like arrhythmias, congestive cardiac failure and even death, depends on location and size of mass. Very few case reports of hemodynamically significant left ventricular inflow and outflow obstruction by cardiac rhabdomyoma has been published and most were detected in early infancy and neonatal period. Similar to other authors, in our case a giant rhabdomyoma (5×4×2.5 cm) almost completely occupied left ventricle and caused obstruction to inflow and outflow and also compressing and narrowing the right ventricular chamber.<sup>3,7-9</sup>

Cardiac rhabdomyomas are circumscribed, un-encapsulated, yellow tan solid masses. The characteristic of this lesion is the presence of spider cells. The spider

cell is a large clear cell with cytoplasmic strands composed of glycogen extending to the plasma membrane. Ultimately, we confirmed the definitive diagnosis of rhabdomyoma by histopathology on autopsy specimen.

Rhabdomyomas tend to regress spontaneously and are not usually operated upon, unless they become obstructive or hemodynamically compromised, causing life threatening complications. Surgical resection of benign cardiac tumor is safe, usually curative and provides excellent long term prognosis. In one series 10% cases of cardiac rhabdomyoma required surgical subtotal tumor excision.<sup>5</sup> On the contrary, malignant cardiac tumors still remain highly lethal. In in-operable cases because of tumor size or position medical treatment with everolimus with therapeutic dose of 5-15 ng/ml regress the size of tumor and complete resolution has been reported by various authors.<sup>10,11</sup> Recurrence of tumor can be minimized by longer therapy and by slow decrements in doses. Derangement in lipid profile and significant side effect in lymphocyte subgroups are reported by Demir et al.<sup>12</sup>

## CONCLUSION

Cardiac rhabdomyoma is most frequent primary cardiac tumor in neonates. Early identification is possible with prenatal ultrasound and fetal echocardiography. Cardiac MRI and CT provide further help in characterization of cardiac tumors for diagnostic purpose and to assess cardiac and extracardiac involvement for treatment purpose. It is also important to encourage the parents for clinical autopsy for definitive diagnosis in cases which succumb early before correct diagnosis.

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