

# Tuberous sclerosis presenting as neonatal cyanosis because of rhabdomyoma causing tricuspid valve obstruction needing a Blalock-Taussig shunt



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We report a newborn female baby who presented at 6 hours of age with cyanosis without any signs of respiratory distress. Cardiovascular and systemic examination was unremarkable apart from cyanosis (saturation 75%). An echocardiogram showed multiple echogenic and homogeneous masses in the interventricular septum, one of which was big and protruding through the tricuspid valve causing right ventricular inflow obstruction. There was a small atrial septal defect (ASD) shunting right to left and patent ductus arteriosus (PDA) shunting left to right. The provisional diagnosis was rhabdomyoma. Blalock-Taussig shunt was done to preserve the tricuspid valve, because these masses tend to regress spontaneously, which was the case after few months. Subsequently, the patient was diagnosed with tuberous sclerosis.

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## Introduction

Studies report that 60–80% of children affected by tuberous sclerosis have cardiac rhabdomyomas, whereas these tumors are found in only around 20% of adults with tuberous sclerosis [1].

## Case report

A term female baby was born with uneventful prenatal and natal history. At 6 hours of life she started to develop cyanosis. She was having good feeds and was active. Examination showed cyanosis with pulse oximeter saturation of 75%. The patient was otherwise well with no signs of

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respiratory distress. Chest and cardiovascular examination was unremarkable. Blood tests including electrolytes, blood gas, glucose and septic workup were normal. Chest X-Ray (CXR) was unremarkable. An echocardiogram showed multiple masses in the interventricular septum, one of which was big and protruding through the tricuspid valve causing right ventricular inflow obstruction and extending to the outflow tract. The others were small and not causing any hemodynamic consequences. There was a small ASD shunting right to left and PDA shunting left to right. The patient was maintained on prostaglandin E1 and saturations were maintained at 85–90%. After discussion with the team and surgeons, the most likely diagnosis was rhabdomyoma and the decision was to do a Blalock-Taussig shunt to preserve the tricuspid valve, because rhabdomyomas regress spontaneously with time. The surgery was done uneventfully on the 5th day of life and the patient left hospital after a few more days. The patient developed ash leaf spots and seizures after a few months and this confirmed the diagnosis of tuberous sclerosis. The patient was seen at 7 months of age and the mass was smaller.

## Discussion

Cardiac tumors are extremely rare in children (0.027–0.17%). Most primary cardiac tumors in children are benign, whilst approximately 10% are malignant [2].

The most common cardiac tumors in children are rhabdomyomas (45%) [3]. These are benign tumors of the heart that are rarely found in patients not having tuberous sclerosis (TSC). They are highly specific to TSC and often the first noted manifestation of disease [4]. The presence of a cardiac rhabdomyoma prenatally is associated with a 75–80% risk of TSC, with multiple rhabdomyomas conveying an even higher risk [4]. The presence of multiple tumor masses in our case led us to think of rhabdomyomas and tuberous sclerosis as a primary diagnosis. No other features of TSC were found initially to confirm the diagnosis and genetic testing is unavailable in our center. The patient subsequently developed ash leaf spots and seizures which confirmed the diagnosis of tuberous sclerosis [5].

Rhabdomyomas are most frequently found in the ventricles, where they can affect ventricular function and sometimes impair valve function or obstruct inflow/outflow tracts [6]. Rhabdomyomas are also associated with cardiac arrhythmias including atrial and ventricular arrhythmia and

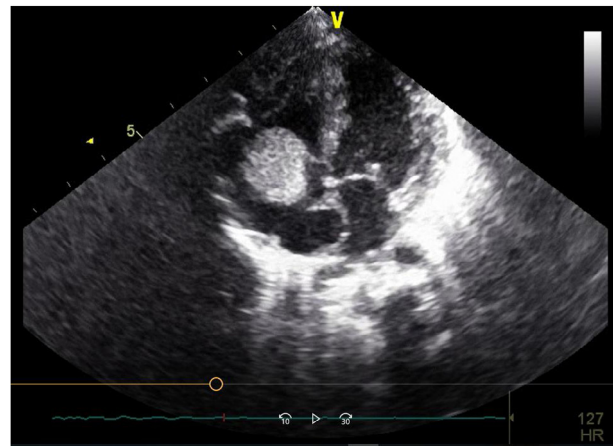


Figure 1. Four chamber view showing hyperechogenic tricuspid valve (TV) mass. Notice the bowing of the interatrial septum to the left.

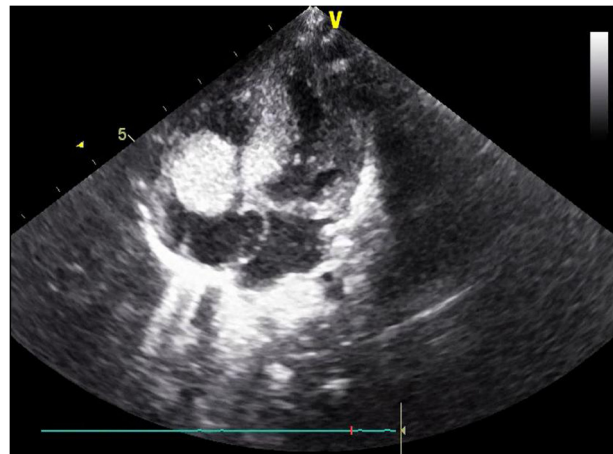


Figure 2. Two masses are seen here, one in the TV and one in the interventricular septum.



Figure 3. Short axis view showing the tumor occupying the right ventricle (RV) cavity from the inflow part and extending to the outflow.

the Wolff-Parkinson-White syndrome [7]. Our case here represents an extremely rare and severe manifestation of this histologically benign but functionally malignant tumor in causing cyanosis at such an early age.

Rhabdomyomas appear on echocardiogram as round, homogeneous, hyperechogenic, intramural or intracavitary masses and are sometimes multiple [6]. Our patient had three rounded hyperechogenic and homogeneous masses arising from the interventricular septum. The biggest one was protruding through the tricuspid valve and obstructing right ventricular inflow Figs. 1–3.

Rhabdomyoma is a condition of particular interest for the researcher due to high incidence of spontaneous regression that occurs in more than 50% of cases [8]. In one study, partial resolution of this cardiac tumor was reported in 50% of cases and complete resolution in 18%. On the other hand, they added that these tumors have been reported to grow or to appear de novo in 4% of patients with TSC [9].

The management of these tumors is usually conservative because of the benign nature of this tumor, the difficulty in removing them completely, location in the deep myocardium and the high regression rate. Excision is not usually considered unless they cause severe intractable arrhythmias, valvular obstruction, or congestive heart failure [10].

Bearing these facts in mind, we opted to preserve the tricuspid valve and to perform a

Blalock-Taussig shunt, hoping for the mass to regress. The Blalock-Taussig shunt will be closed later in the catheter lab when significant regression of the mass and convincing flow through the tricuspid valve is seen.

## References

- [1] Watson GH. Cardiac rhabdomyomas in tuberous sclerosis. *Ann N Y Acad Sci* 1991;615:50–7.
- [2] Uzun O, Wilson DG, Vujanic GM, Parsons JM, De Giovanni JV. Cardiac tumours in children. *Orphanet J Rare Dis* 2007;2:11.
- [3] Burke A, Virmani R. Pediatric heart tumours. *Cardiovasc Pathol* 2008;17:193–8.
- [4] Harding C, Pagon R. Incidence of tuberous sclerosis in patients with cardiac rhabdomyoma. *Am J Med Genet* 1990;37:443–6.
- [5] Northrup H, Krueger DA. Tuberous sclerosis complex diagnostic criteria update: Recommendations of the 2012 international tuberous sclerosis complex consensus conference. *Pediatr Neurol* 2013;49:243–54.
- [6] Di Liang C, Ko SF, Huang SC. Echocardiographic evaluation of cardiac rhabdomyoma in infants and children. *J Clin Ultrasound* 2000;28:381–6.
- [7] Myers KA, Wong KK, Tipple M, Sanatani S. Benign cardiac tumours, malignant arrhythmias. *Can J Cardiol* 2010;26(2):58–61.
- [8] Józwiak S, Kotulska K, Kasprzyk-Obara J, Domańska-Pakieła D, Tomyn-Drabik M, Roberts P, et al. Clinical and genotype studies of cardiac tumors in 154 patients with tuberous sclerosis complex. *Pediatrics* 2006;118:1146–51.
- [9] Smith HC, Watson GH, Patel RG, Super M. Cardiac rhabdomyomata in tuberous sclerosis: their course and diagnostic value. *Arch Dis Child* 1989;64:196–200.
- [10] Takach TJ, Reul GJ, Ott DA, Cooley DA. Primary cardiac tumors in infants and children: immediate and long term operative results. *Ann Thorac Surg* 1996;62(2):559–64.