

## Case Report

# A rare case of cardiac rhabdomyoma in the fetus

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### Introduction

The presence of a primary congenital tumor of heart is a rare occurrence. Its incidence is 1-2/10000 and 90% of these tumors are benign out of which rhabdomyomas are the most common (58%)<sup>1</sup>. Rhabdomyomas are hamartomas rather than true neoplasms and are caused by mutation in TSC1 and TSC2 genes<sup>2</sup>.

Here we present a rare case of cardiac rhabdomyoma which was diagnosed during routine antenatal ultrasound examination.

### Case report

A-30-year-old G<sub>3</sub>P<sub>2</sub>L<sub>2</sub> woman with a history of previous two cesarean sections for contracted pelvis came for her first antenatal visit at the end of third trimester. Her general and obstetric examinations were normal, corresponding with her period of gestation. She was advised ultrasonography (USG) as a routine. It showed small to moderate sized echogenic lesions in both the ventricles of the fetal heart suggestive of

rhabdomyomas. The fetal cardiac chambers were otherwise normal and there was no other congenital anomaly. At that time her gestational age was 32 weeks, biparietal diameter (BPD) 80 mm, fetal abdominal circumference (AC) 268 mm, and femoral length (FL) 63 mm. She was subjected to further evaluation by color doppler fetal echocardiography which revealed a large fetal heart with cardiothoracic ratio of 0.72 (normal upto 0.60). Multiple well defined echogenic nodular masses were seen in the ventricles. Three of them were in the left ventricle, measured 1 – 1.5 cm, and appeared to arise from interventricular septum near the base and the anterolateral and posterior walls in mid part. One large nodule of 3 cm occupied the apical wall of the right ventricle. No other anomaly was there and cardiac hemodynamic pattern was within normal limits (Figure 1 and 2).

The woman was counseled regarding the diagnosis and association of cardiac rhabdomyoma with tuberous sclerosis. A detailed genetic and family history, and a detailed history of siblings were taken for evidence of tuberous sclerosis. The woman's husband had hypopigmented spots (Ash leaf spots), tiny red nodules over the nose and cheeks (Adenoma sebaceum).

She was taken for elective cesarean section at 38 weeks of gestation and delivered a male child of 3 kg with apgar score of 7 at 1 minute and 9 at 5 minutes. The neonate was kept under strict vigilance of a

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**Figure 1.** Four chamber view showing cardiac rhabdomyoma



**Figure 2.** Echocardiography showing normal function.

neonatologist. The postnatal course of the neonate and the mother was uneventful. Both the baby and the mother were discharged on the 9<sup>th</sup> postoperative day. At the follow up after 1 month the baby had no skin lesions and the neonatal echocardiography was consistent with the previous findings.

## Discussion

Rhabdomyomas, also known as myocardial hamartomas, are benign smooth muscle tumors of the myocardium, derived from embryonal myoblast<sup>1</sup>.

Histopathology reveals typical spider cells<sup>2,3</sup> - they may arise anywhere in the myocardium, but most commonly appear in the left ventricle<sup>4</sup>. It is strongly associated with tuberous sclerosis. In cases of rhabdomyomas 51-86% also have tuberous sclerosis, while 50% of the cases of tuberous sclerosis have rhabdomyomas<sup>5</sup>. Tuberous sclerosis is an autosomal

dominant disease with variable expressibility and high penetrance. The genes of this disease are located on 9<sup>th</sup> (9q<sub>34</sub>) and 16<sup>th</sup> (16p<sub>13</sub>) chromosomes<sup>6</sup>. Clinical manifestations of rhabdomyoma range from no symptoms to dysarrhythmia due to compression of conductive system. Intracavitary growth of tumors cause disruption of intracardiac blood flow leading to congestive cardiac failure (CCF). Fetal manifestations include polyhydramnios, intrauterine death and non-immune fetal hydrops<sup>2,3</sup>. Clinical manifestation of tuberous sclerosis include classic triad of seizures, retardation, adenoma sebaceum, skin lesions which are hypopigmented or depigmented mass known as Ash Leaf spots, tiny red nodules over nose and cheeks (adenoma sebaceum) and shagreen patches, cerebral abnormalities include periventricular calcification or nodules, cerebral atrophy causing seizures and mental retardation.

Rhabdomyomas grow slowly in-utero and show no or minimal postnatal growth. Tumor growth is proportional to cardiac growth and tends to somewhat slow down towards the end of pregnancy. Some tumors are known to show regression<sup>3-5,7</sup>. Diagnosis can be done in mid-second trimester as early as 20 weeks by fetal echocardiography<sup>6</sup>. Fetal echocardiography promotes early diagnosis of tuberous sclerosis through prenatal identification of cardiac rhabdomyoma<sup>8</sup>. They appear as echogenic mass in echocardiography as they are strongly associated with tuberous sclerosis. MRI imaging combined with USG would allow improved prenatal diagnosis of tuberous sclerosis complex<sup>6</sup>.

Prognosis depends on the size, site and number of tumors as well as presence or absence of associated anomalies. Presence of dysarrhythmia or non-immune fetal hydrops indicates a poor prognosis<sup>1</sup>. The treatment depends on their location and size. When the diagnosis is made prior to fetal viability, pregnancy termination may be offered. Serial USG to identify signs of CCF or dysarrhythmia should be done. Standard obstetrical management is appropriate for uncomplicated course. Tertiary center facility with a pediatric cardiologist should be available at the time of delivery. Most of the cardiac rhabdomyomas are small and asymptomatic and should be managed conservatively with observation for growth or regression<sup>1</sup>. Large tumors that show signs of obstructing blood flow and those producing ventricular arrhythmias should be removed surgically<sup>5,8</sup>. Family screening for tuberous sclerosis is desirable.

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