

CASE REPORT

An Interesting Case of Fetal Cardiac Rhabdomyometa in Pregnancy

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ABSTRACT

Mrs S was detected to have fetal cardiac rhabdomyometa at 30 weeks of gestation, as the fetal echo showed multiple non-obstructive cardiac rhabdomyometa with normally connected heart and premature atrial contractions. Magnetic resonance imaging (MRI) showed no obvious cranial tubers. As the condition was isolated, patient was conservatively followed-up. Patient had an uneventful vaginal delivery. Post-delivery, fetal echocardiography showed multiple masses on the left and right side of the cardia on the tricuspid valve and was opined conservative management in view of high risk, had limited benefit for surgical intervention and as the condition would likely to improve over time. Baby was stable at discharge and was put on tablet propranolol 10 mg ¼ thrice daily for self-limiting arrhythmias.

Keywords: Arrhythmias, Cardiac rhabdomyoma, Propranolol, TSC.

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INTRODUCTION

Fetal cardiac rhabdomyoma is a rare and most common primary pediatric tumor and constitutes more than 60% of antenatally detected cardiac tumors.¹ For a fetus (unborn baby), the diagnosis accounts for 1% of all cardiac disorders detected by prenatal ultrasound. Incidence is 0.002 to 0.25% at autopsy, 0.02 to 0.08% in live-born infants and 0.12% in prenatal reviews, either sporadic or with certain genetic disorder. The natural history of cardiac rhabdomyometa is one of spontaneous regression. The fastest growth time for these tumors is between 2nd and

the 3rd trimester. More than 85% of them are detected after 24 weeks of gestation. Growth slows as the pregnancy nears its end and stops when the baby is born. The biggest concern when a rhabdomyoma is seen on prenatal ultrasonography (USG) is its association with tuberous sclerosis which is seen in 40 to 90% of the babies.

Here, we report a case of fetal cardiac rhabdomyometa without tuberous sclerosis diagnosed antenatally and also discuss regarding the importance of diagnosing the condition in the prenatal period.

CASE REPORT

Mrs S, G2A1 23-year old, a well booked case, with second degree consanguinity was having regular uneventful antenatal care with us. Nuchal translucency-nasal bone and anomaly scans done were normal. She came with reduced fetal movements at 30 weeks of gestation. Non-stress test and biophysical profile done for fetal well being detected missed beats in the fetal cardia, and hence was advised fetal echocardiography. The fetal echocardiography done showed multiple nonobstructive cardiac rhabdomyometa with normally connected heart 1.5 × 1.3 cm hyperechoic solid space occupying lesion arising from the free wall of the right ventricle noted just under the tricuspid valve leaflet. Similar, small 7 mm lesion seen in the free wall of the left ventricle. Every third atrial contraction was premature followed by a compensatory pause. 1:1 atrioventricular conduction sequence noted. Color Doppler study showed normal blood flow across the atrial and semilunar valves (Fig. 1). Due to a strong association between rhabdomyometa and tuberous sclerosis, magnetic resonance imaging (MRI) was done to rule out tubers in the brain, which showed no obvious cranial tubers or any significant abnormality, which was reassuring (Fig. 2).

There was no family history of tuberous sclerosis. Pediatric cardiologist opined that as the condition was isolated, the baby had a good prognosis and the lesions were likely to disappear over time and needed follow-up once in 2 weeks during pregnancy for heart rate and rhythm and also for tuberous sclerosis postnatally. Patient was monitored regularly with Doppler and nonstress test. Heart rate was maintained between 118 to 131 beats per minute. There was no supraventricular tachycardia.

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Patient delivered vaginally a healthy baby with good apgar scores at term and was shifted to neonatal intensive care unit (ICU) in view of atrial ectopics. As the saturation of the baby was falling, was put on anti-arrhythmic drugs, after which the saturation picked-up to 80%. Echocardiography done post-delivery showed multiple rhabdomyomata, moderate secundum atrial septal defect, right to left atrial ectopics seen with good biventricular function. Multiple soft tissue masses in right and left ventricle, predominately intramural, one large 22 mm close to the tricuspid valve partially obstructing the tricuspid valve inflow with no gradient, moderate tricuspid regurgitation and moderate to severe atrial regurgitation seen. Ultrasonogram abdomen and neurosonogram of the baby done was normal. Baby having

rhabdomyoma of the tricuspid valve with increased right atrial pressure, with right to left flow and was advised conservative management. The baby was put on tablet propranolol 10 mg $\frac{1}{4}$ tablet thrice daily for self-limiting arrhythmias (Fig. 3).

On day 7 of life, ECG was normal with sinus rhythm. Echocardiography showed ectopics, right sided mass on tricuspid valve, moderate tricuspid regurgitation, moderate secundum atrial septal defect with right to left flow and good biventricular function. Some amount of desaturation and tachyarrhythmia expected from cyanotic heart disease but surgical intervention had high risk and limited benefit. The location of the lesion on the cardiac valve was also crucial for surgical intervention and it was expected that the lesion would regress in early infancy.

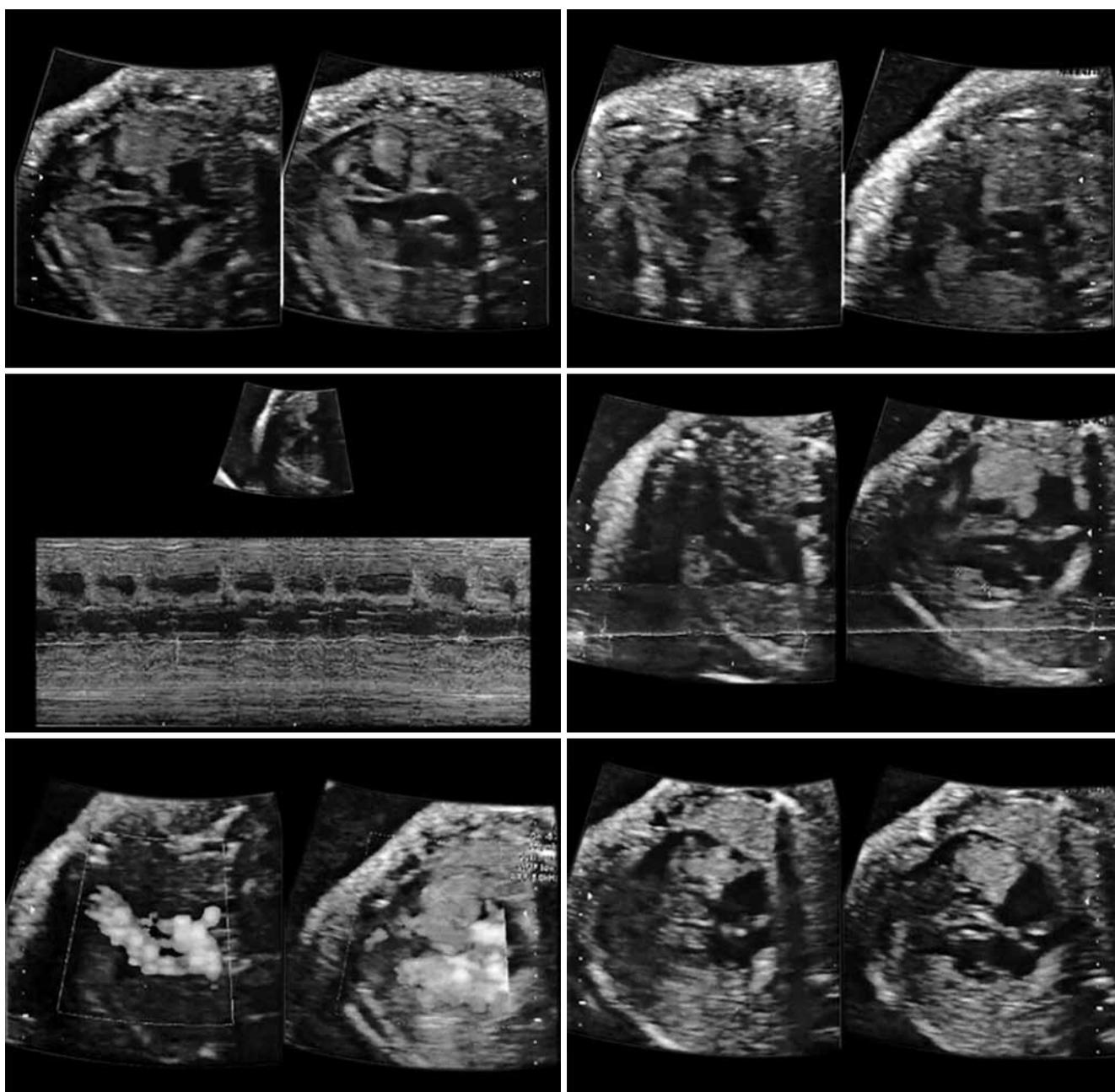


Fig. 1: Fetal echo showing multiple cardiac rhabdomyomata

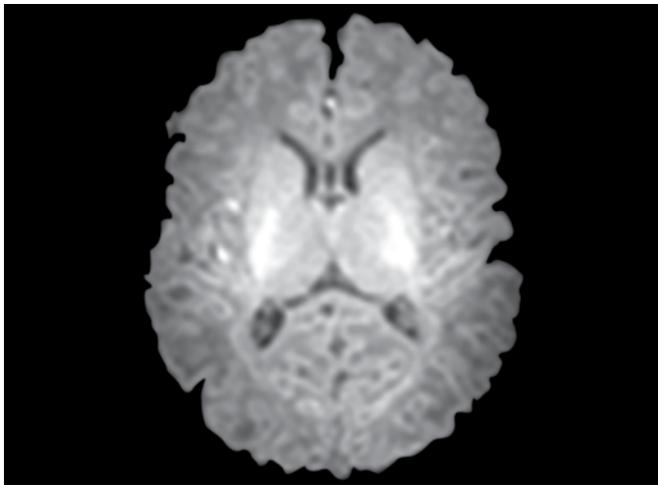


Fig. 2: Magnetic resonance imaging of the infant, with no features of tubes

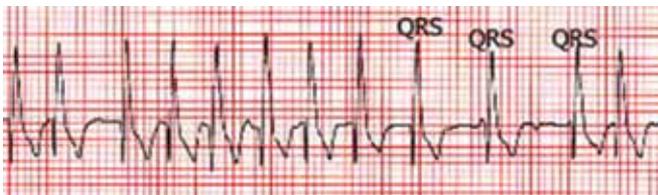


Fig. 3: Electrocardiogram showing atrial fibrillation (irregular RR interval)

The baby was discharged on tablet propranolol 10 mg $\frac{1}{4}$ thrice daily and was monitored every week for 2 months and following that every month for 6 months. The baby was continued on tablet propranolol 2.5 mg thrice daily which was changed to twice daily after 6 months of birth. It had appropriate milestones and came for regular follow-up until 6 months and was stable maintaining a saturation of around 80 to 90%. Later on baby was lost for follow-up. At 11 months of age, baby had an episode of convulsion and was admitted to neonatal ICU of our hospital and detected to have an infarct of the right anterior cerebrum. Baby died after 3 days of admission and was on ventilator when it expired.

DISCUSSION

Cardiac rhabdomyoma are the most common cardiac tumors diagnosed *in utero*. Fetal cardiac tumors constitute one to two per 10,000 births. Around 90% of these are benign, out of which 60 to 80% are rhabdomyomas while others being fibromas, teratomas, vascular tumors and very rarely myxomas.² They represent the earliest detectable hamartoma in tuberous sclerosis where they have been diagnosed in fetal life as early as 22 weeks of gestation. As they may be the only manifestation of tuberous sclerosis at this stage, probability that other features of the disease will develop with time is important for genetic counseling and patient management.³ The prenatal diagnosis of cardiac rhabdomyoma most often

occurs after referral for the finding of a cardiac tumor or fetal dysrhythmia on routine obstetrical ultrasound assessment without other obvious features of tuberous sclerosis at the time of diagnosis, as also seen in our case. Knowledge of the outcome of affected fetuses and the true incidence of tuberous sclerosis in fetal cardiac rhabdomyoma is critical for accurate prenatal counseling and planning of prenatal treatment.⁴ When fetal cardiac rhabdomyoma is diagnosed, careful evaluation of other fetal structures including the brain and renal parenchyma should be performed to search for signs of tuberous sclerosis.⁵ The commonest complication associated with rhabdomyoma of the heart is conduction defect and obstruction.⁶

Tuberous sclerosis is an autosomal dominant disorder affecting approximately one in 6,000 to 8,000 individuals worldwide. Sporadic mutation constitute 50%. Genetic disorder associated is tuberous sclerosis. Tuberous sclerosis is a hamartoma syndrome due to mutation in TSC1 or TSC2 in which cardiac rhabdomyoma are seen in approximately 60% of patients. The disease is characterized by the development of benign hamartomas in a number of organ systems and often has a prominent neurologic component due to the occurrence of CNS hamartomas that lead to epilepsy and developmental disorders.² Frequently cardiac, renal, and lung lesions are asymptomatic and discovered at necropsy. There are also skin manifestations of the disease in the form of shagreen patches or adenoma sebaceum. In rare cases, it is associated with Tetralogy of Fallot (TOF), Ebstein abnormality, hypoplastic left heart syndrome, Down's syndrome and basal cell nevus syndrome.⁷ Detection of cardiac rhabdomyoma in the fetal period is important as they can lead to sudden death in some while they can regress spontaneously in the others. Cardiac rhabdomyoma are typically asymptomatic and are therefore, usually not operated upon unless they are obstructive, cause heart failure or are complicated with severe intractable arrhythmias. Also, they may be difficult to remove completely as they are often located deep in the myometrium,⁸ as was in our case. Serial observations have demonstrated that the majority of these lesions become less prominent over time, with some disappearing altogether as assessed by ultrasound. So, surgical resection is performed only when they cause life threatening complications.⁷

When symptoms are present, they are generally related to the size of the tumors and their location. So, asymptomatic tumors are simply monitored by echocardiography and holter recording, in addition to usual clinical examination.⁸ Two-dimensional echocardiography offers a simple noninvasive method, which shows cardiac tumors very well and is the most useful diagnostic test

in pregnancy and early infancy.⁹ While most rhabdomyomas are multiple even a single lesion in infancy is very likely to be rhabdomyoma and if other features of tuberous sclerosis are found, the diagnosis is beyond reasonable doubt.⁶

A meta-analysis conducted by Chao et al showed that multifocality of fetal cardiac rhabdomyoma and a positive history of tuberous sclerosis are two strong predictors of neonatal outcome. Large obstructive lesions and those interfering with cardiac motility also have a poor prognosis.^{3,10} Fetuses with a tumor of ≥ 20 mm in diameter and persistence of arrhythmias in the postnatal period increases the risk of neonatal and infant death.⁹ No embolic events have been reported and there is no need for oral anticoagulation in the absence of a specific indication like atrial fibrillation.⁸ A novel therapy of treating a patient with clinically significant cardiac rhabdomyoma is by giving Everolimus a mammalian target of rapamycin (mTOR) inhibitor.¹¹

As we saw in our case, the low saturation was due to the mass leading to the shunting of blood from right to left and ectopics were due to conduction defects. Since our case was not associated with tuberous sclerosis, the outcome was considered to be good. In our case, the baby was not brought for regular check up after 6 months of age and expired at 11th month of age following a convulsion with a cerebral infarct. This case is reported because of its rarity and also it being an isolated lesion as it is not associated with tuberous sclerosis. Here, the infarct could be due to an emboli which is again very rare probably due to the crucial location of the rhabdomyoma on the cardiac valve.

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