International Journal of Health Sciences and Research
www.ijhsr.org ISSN: 2249-9571

*Case Report*

# A Rare Case of Fetal Cardiac Rhabdomyoma

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*Received: 24/06/2015* *Revised: 13/07/2015* *Accepted: 14/07/2015*

## ABSTRACT

Fetal cardiac tumors are a rare entity. Here we report a neonate whose fetal ultrasonogram revealed a cardiac rhabdomyoma. Postnatally the baby had as hleaf macules in the skin, subependymal nodules and cortical tubers which eventually turned out be tuberous sclerosis. Fetal cardiac rhabdomyoma serves as a prenatal marker of tuberous sclerosis.

***Keywords:*** Rhabdomyoma, ash leaf macules, subependymal nodules, tuberous sclerosis.

## INTRODUCTION

De Vore et al in 1982 first reported the prenatal diagnosis of cardiac tumor. [1] Cardiac tumors arerhabdomyoma, fibroma, myxoma, teratoma and hemangiomas. Rhabdomyomas account for 60-86% of all primary fetal cardiac tumors. [2,3] They are hamartomas of developing cardiac myocytes. They occur sporadically or in association with congenital heart diseases or in the setting of genetic disorders. Tuberous sclerosis is the most common genetic disorder associated with them. Tetralogy of Fallot, Hypoplastic left heart syndrome and Ebstein's anomaly are rare associations.

## CASE REPORT

A Term male baby born via labor naturalis was admitted in the neonatal care unit of Chengalpattu medical college hospital with the fetal ultrasonogram showing a mass attached to the interventricularseptum of the heart. Antenatal period was uneventful. There was no birth asphyxia, respiratory distress orseizures. Baby was breast feeding well and vitals were stable.

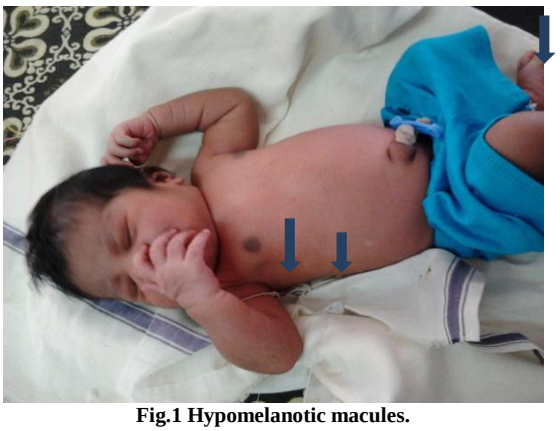

**Fig.1 Hypomelanotic macules.**

General examination revealed three hypomelanotic ma cules in the body. [fig.1] Cardiovascular system and other systems were clinically normal. Ophthalmic

examination was alsonormal. Echocardiogram revealed a mass of 20 * 17 mm attached to the interventricular septum projecting into aortic valve causing left ventricular outflow tract obstruction with a small Patent Ductus Arteriosus and Atrial Septal Defect with left to right shunt. [fig.2]

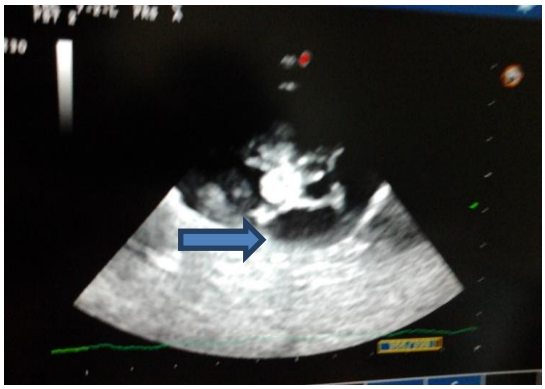

**Fig.2 Cardiac rhabdomyoma**

Cardiac rhabdomyoma and as hleaf macule were considered. With a probable diagnosis of tuberoussclerosis, CT brain and USG abdomen were taken. CT brain showedmultiple subependymalnodules and calcified cortical tubers [fig.3] while USG abdomen was normal. Hemogram, bloodglucose, renaland liver parameters were normal.

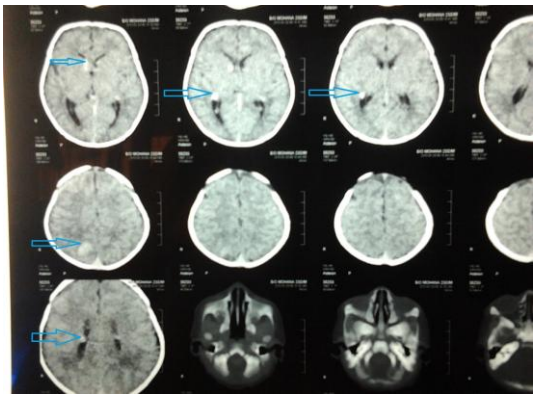

**Fig.3 Subependymal nodules and cortical tuber**

With cardiac rhabdomyoma, three ash leaf macules, cortical tuber and multiple subependymal nodules, a diagnosis of tuberous sclerosis was made. The baby was observed for arrhythmia, seizures and hemodynamic instability. Throughout the hospital course, baby was stable. Genetic counselling was given to the parents and the baby was discharged. Periodic follow up of the child had been done. There were increase in the number of ash-leaf macules, minimal decrease in the size of rhabdomyoma and persistence of neurological lesions without seizures.

**Table.1 Major Features of Tuberous Sclerosis Complex**

| |
|---|
| Cortical tuber |
| Subependymal nodule |
| Subependymal giant cell astrocytoma |
| Facial angiofibroma or forehead plaque |
| Ungual or periungual fibroma (nontraumatic) |
| Hypomelanotic macules (>3) |
| Shagreen patch |
| Multiple retinal hamartomas |
| Cardiac rhabdomyoma |
| Renal angiomyolipoma |
| Pulmonary lymphangioleiomyomatosis |

**Table 2.Minor Features of Tuberous Sclerosis Complex**

| |
|---|
| Cerebral white matter migration lines |
| Multiple dental pits |
| Gingival fibromas |
| Bone cysts |
| Retinal achromatic patch |
| Confetti skin lesions |
| Nonrenal hamartomas |
| Multiple renal cysts |
| Hamartomatous rectal polyps |

## DISCUSSION

Rhabdomyomas are usually benign and appear as round, homogenous, hyperechogenic masses in the ventricles or as multiple foci in septal wall or ventricles on ultrasound.[2] The earliest antenatal sonographic detection of a cardiac tumor was reported at 15 weeks of gestation.[4] Most cases were detected at 24 weeks of gestation in a meta-analysis by Chao et al.[5] Third trimester ultrasound at 32 weeks found out the rhabdomyoma in our case. Studies have demonstrated that the incidence of cardiac rhabdomyoma is 0.002-0.25% at autopsy, 0.02-0.08% in live-born infants, and 0.12% in prenatal reviews.[3] Because of the use of ultrasonography as part of routine prenatal screening, it is increasingly common for cardiac rhabdomyomas to be clinically recognized

in utero. [5,6] Recently antenatal MRI is instrumental in detecting rhabdomyomas and other fetal tumors. Rhabdomyomas usually increase in size until 32 weeks of gestation and regress spontaneously. Symptoms of cardiac rhabdomyomas arise because of chamber or valve obstruction, arrhythmias, or failure resulting from extensive myocardial involvement. Tumors obstructing the right-side inflow or the outflow of the ventricles can lead to decreased cardiac output, atrial and caval hypertension, hydrops fetalis and death. Arrhythmias, both ventricular and atrial, are not uncommon. The neonate in our case was asymptomatic.

The presence of cardiac rhabdomyomas (especially multiple rhabdomyomas) is strongly suggestive of tuberous sclerosis. Patients should be examined for associated clinical and pathologic features of tuberous sclerosis. [7] Cary O. Harding et al estimated that 51-86%of cardiac rhabdomyomas were associated with tuberous sclerosis. [8] They arethe first clinical manifestation of tuberous sclerosis in more than 50% of the cases. [9]

Tuberous sclerosis complex (TSC) is inherited as an autosomal dominant trait with variable expressionand a prevalence of 1/6,000 newborns. Spontaneous genetic mutations occur in 2/3 of the cases. [10] There was no positive family history in our case and we attributed spontaneous geneticmutation to be the cause. Tuberous sclerosis is an extremely heterogeneous disease with a wide clinical spectrum varying from severe mental retardation and incapacitating seizures to normal intelligence and a lack of seizures, often within the same family. The disease affects many organ systems other than the skin and brain, including the heart, kidney, eyes, lungs, and bone. [10] Diagnosis requires 2 major criteria or 1 major and 2 minor criterions. [Table 1 and 2].

**Four major criteria**

1. cardiac rhabdomyoma,
2. three hypomelanotic macules,
3. SubependymalNodule,
4. Cortical tuber was fulfilled in our case.

In addition to echocardiogram and MRI brain, ultrasonogram and CT of abdomen can be done to rule out renal angiomyolipomas. Anti epileptics are given if children develop seizures. Neurosurgical intervention can be done for obstructive hydrocephalus due to subependymal giant cell astrocytoma. Periodic follow up imaging is necessary in tuberous sclerosis to know about the extent of the lesions. Prenatal genetic studies may be offered when a known tuberous sclerosis mutation exists in a family. We advised tuberous sclerosis gene mutation studies for the mother in her next pregnancy. Neurodevelopmenal screening was necessary in tuberous sclerosis to identify abnormal behaviours and to provide early intervention. On follow up of our child at three months of age, there was a minimal decrease in size of the rhabdomyoma with persisting neurological lesions. Parents are advised to bring the child for follow up. The natural history of cardiac rhabdomyomas is that of complete or partial regression with consequent resolution of symptoms. The reported survival rates range from 81% to 92%. [3] The prognosis of patients with rhabdomyomas is chiefly determined by the size and location of the lesion. Tumors larger than 20 mm in diameter are more likely to cause hemodynamic disturbances or arrhythmias, which are associated with an increased risk of death. [11,12]Although the size of rhabdomyoma was 20*17 mm in our case, it was asymptomatic.

Most patients can be managed conservatively. Conservative management includes frequent monitoring with echocardiography and electrocardiography (ECG). Patients with arrhythmias are treated

with antiarrhythmic medications. If medical treatment fails to control the arrhythmias or blood flow is severely obstructed, surgical management is recommended.

## CONCLUSION

The incidence of fetal cardiac rhabdomyoma is rare. Fetal echocardiography and antenatal MRI helps in prenatal diagnosis of tuberous sclerosis by detecting rhabdomyomas. One should look for the other manifestations of tuberous sclerosis if a rhabdomyoma is found and genetic counselling should be given to the families at risk. This case is presented for its rarity.

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How to cite this article: Vinoth S, Lakshmi S, Anitha GFS. A rare case of fetal cardiac rhabdomyoma. Int J Health Sci Res. 2015; 5(8):631-634.


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