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CASE REPORT

# Isolated Asymptomatic Fetal Intracardiac Mass: A Case of Rhabdomyoma

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**Introduction:** Fetal cardiac rhabdomyoma is one of the rare benign cardiac masses which is commonly associated with the tuberous sclerosis complex (TSC). Though mostly fetal cardiac rhabdomyoma is asymptomatic it may lead to life-threatening conditions like outflow obstruction, arrhythmias, hydrops fetalis, or sudden fetal death.

**Case Report:** We are reporting an isolated, asymptomatic fetal intra-cardiac mass (rhabdomyoma) that was discovered at 32 weeks of gestation and was followed as an outpatient until 39 weeks plus one day, at which point a cesarean section was performed. After delivery, the child underwent evaluations at the 1st day, 7th day, 30th day, 7th month, and 12th month of age. Following a checkup, the child's anthropometric and neurobehavioral growth were both healthy. Except for the tumor, which was neither growing nor shrinking in size, none of the clinical diagnostic criteria for tuberous sclerosis complex were met for this child up to the age of one year.

**Conclusion:** The most common primary benign fetal cardiac tumor is cardiac rhabdomyoma, which is usually associated with tuberous sclerosis. In developing nations where it is challenging to obtain MRIs and genetic studies, and in a similar patient like ours with no other features of tuberous sclerosis, the child needs to be followed in the future, bearing in mind that tuberous sclerosis manifestations will continue to develop over a patient's lifetime.

**Keywords:** cardiac, rhabdomyoma, tumor, fetal, tuberous sclerosis, case report, Harar, Ethiopia

## Introduction

Rhabdomyomas are the most frequently detected fetal cardiac tumor and are found within the ventricular cavities, ventricular wall and septum. Sonographic features of fetal cardiac rhabdomyomas include: multiple (in 90% of the cases, while single in 10% of them), well-circumscribed, rounded, homogenous, and non-encapsulated white or gray-white intramural or intracavitary nodules. Though in most of the cases fetal cardiac rhabdomyomas are asymptomatic, the symptoms depend on the number, size, and site of the tumor. When it is symptomatic it may cause inflow/out flow obstruction of the ventricles, arrhythmia, hydrops fetalis, cardiac failure and sudden fetal death. In more than 80% of the cases fetal cardiac rhabdomyoma is associated with the multisystem, autosomal dominant neurocutaneous disorder called Tuberous sclerosis complex. In a fetus who has cardiac rhabdomyoma; searching for other feature of tuberous sclerosis is mandatory with detailed anatomical sonography, fetal MRI, family history of tuberous sclerosis and genetic test. Though the tumor regresses usually if it persists; treatment of the cardiac rhabdomyoma depends on the symptom. Options of treatments can be medical with the sirolimus or everolimus or it can be surgical excision of the tumor.[1–7]

## Case Description

A 29-year-old gravida V par IV (all alive, 3 spontaneous vaginal deliveries, and the last child was delivered by cesarean section for the indication of a failed induction 4 years prior to the current pregnancy) came for ANC follow-up at a gestational age of 32 weeks from her LNMP.

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After taking a medical history, it was discovered that all four of her children are healthy, doing well in school, and have no known history of genetic or seizure disorders. She was investigated with the Venereal Disease Research Laboratory (VDRL), Hepatitis B surface antigen (HBSag), and urine analysis, all of which were negative. All cell lines in the CBC were normal, her blood group is A, and Rh is positive, according to the Complete Blood Count (CBC), blood group, and RH. Obstetric ultrasound was also performed showing normal anatomical scan of the all body parts of the fetus except the heart. Detailed fetal echocardiography evaluation was done with findings of: both atria have comparable size and normal situs. Both atrioventricular and semilunar valves are normally positioned with normal opening and closure. Both ventricles are comparable in size and contractility; in both 2D and color flow, the left ventricle forms the apex of the heart without any ventricular septal defect. But on the papillary muscles of the left ventricle there were two circumscribed, round, echogenic mass measuring 18.2 mm by 8.3mm and 13.5mm by 8.3 mm (Figure 1). Upon evaluation of the outflow tract, both the LVOT (left ventricular outflow tract) and RVOT (right ventricular outflow tract) have normal anatomy and function using 2D and CF ultrasound evaluation (Figure 2). According to the fetal echo finding, a diagnosis of cardiac rhabdomyoma was made. Since there is a high chance of tuberous sclerosis in cardiac rhabdomyoma, detailed neurosonography and other system exams were done to look for other signs of tuberous sclerosis. Despite searching for the other features of tuberous sclerosis, no other sign of it was found other than the tumor. She had regular ANC follow-up from 32 weeks of gestation up to 39 weeks without any complications.

At gestational age of 39 weeks plus 1 day, she underwent a cesarean section for the indication of full-term pregnancy plus a request for a repeat cesarean section, with the outcome of a 3200-gram female with an APGAR score of 10 and 10 at the 1st and 5th minutes. Both the mother and the neonate had a smooth post-operative period and were discharged on the third day.

After delivery, the neonate was evaluated on the 1$^{st}$, 7th, and 30$^{th}$ days for any regression or increment of the mass, emergence of skin lesions, or seizure. All physical examination results were normal, and the mass size was similar to the antepartal evaluation.

At her 7$^{th}$ month, the child was evaluated again, and upon history inquiries, the infant was doing great developmentally for her age group. The infant was examined for neurodevelopmental delay, and the child was growing appropriately

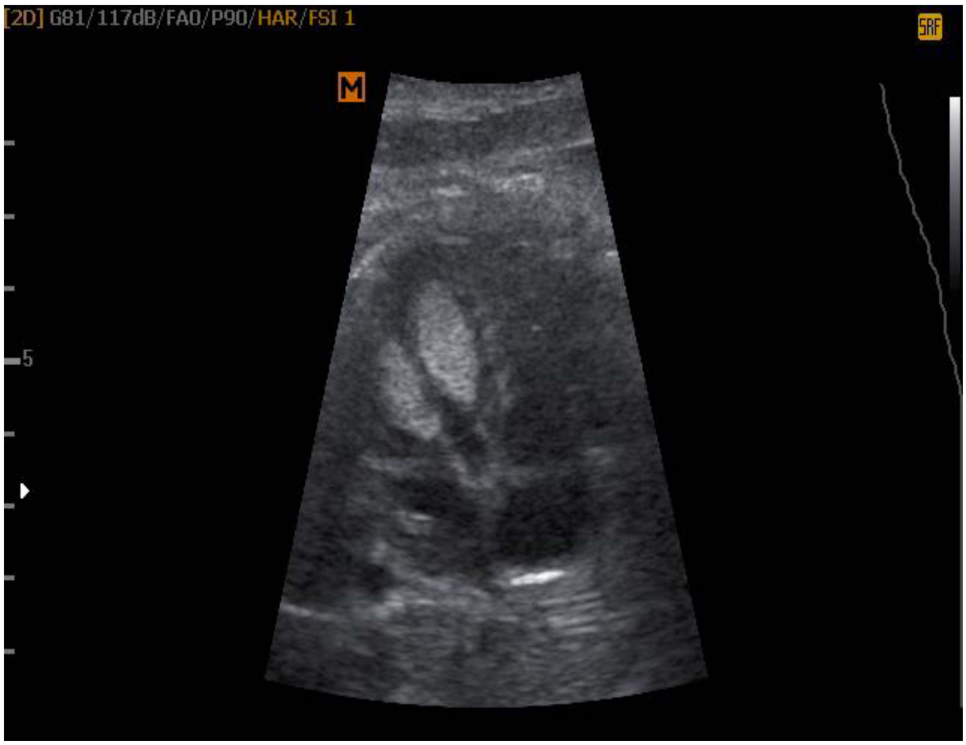


**Figure 1** Two fetal left ventricular papillary muscle echogenic mass (rhabdomyoma) at 34 weeks.

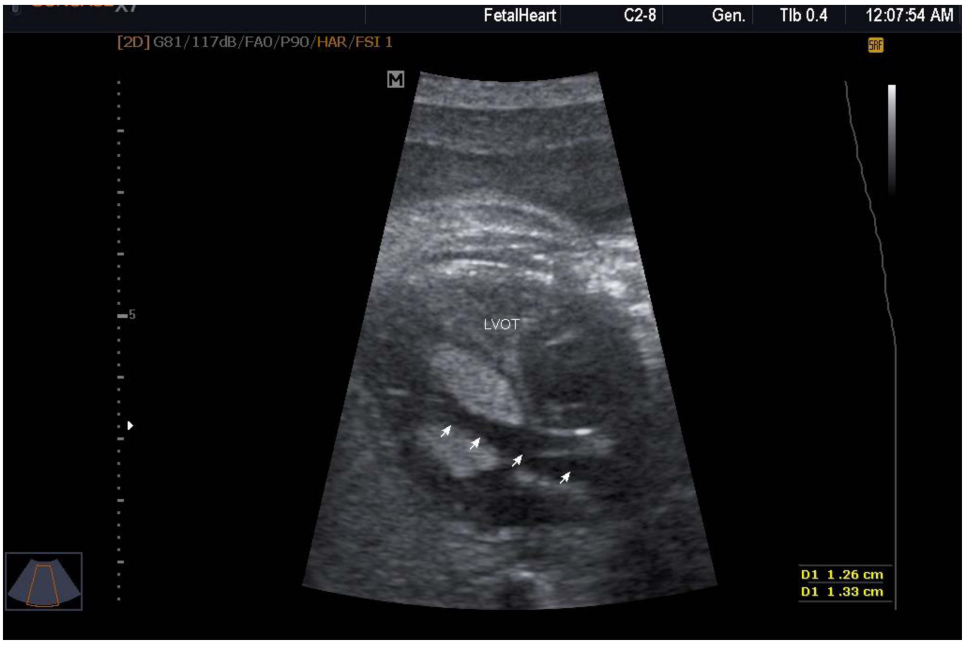


**Figure 2** Normal Left ventricular outflow tract at 34 weeks of gestation (The arrow (↑) indicates Left Ventricular Out flow Tract (LVOT)).

for her age. An echocardiography study by a pediatric cardiologist revealed well-circumscribed hyperechoic masses on both left ventricular papillary muscles, each measuring 21.8 mm by 9.2 mm and 14.7 mm by 8.5 mm (Figure 3) and creating no left ventricular inflow obstruction (Supplementary Video 1).

A history from the family was obtained, and a physical examination with anthropometric measurements was performed to assess her developmental condition during her first-year evaluation. The child was developing normally, as other children her age were. Except for the heart, all of the systems examined were unremarkable. An echocardiography study has revealed well-circumscribed hyperechoic masses on both left ventricular papillary muscles with no size increment and creating no left ventricular inflow obstruction.

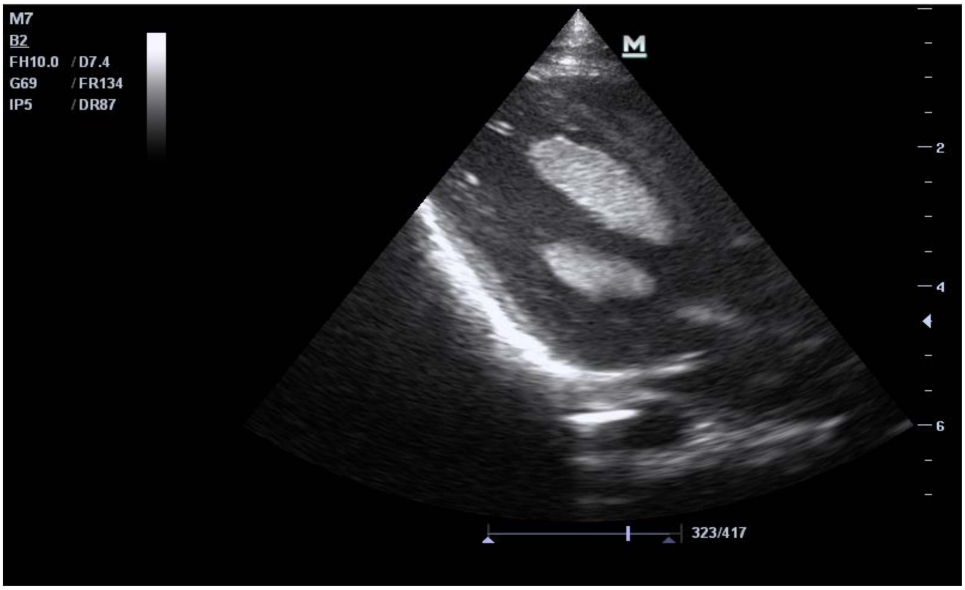


**Figure 3** Left ventricular papillary muscle echogenic mass (rhabdomyoma) at one year of age.

## Discussion

Primary fetal cardiac tumors are rare, with a reported prevalence of 0.08% to 0.2%. Though most of these tumors are benign there are few malignant cardiac tumors. Rhabdomyomas are the most common primary fetal cardiac tumors accounting for 60% and was first described by von Recklinghausen in 1862 as intracardiac rhabdomyoma, followed by teratomas, fibromas, hemangiomas, and myxomas.[3,8,9] It is associated with the tuberous sclerosis complex in 80% of cases, especially when multiple tumors and a positive family history of tuberous sclerosis are noted.[1,10–13] Tuberous sclerosis complex is a multisystem, autosomal dominant neurocutaneous disorder that results from mutations in one of two genes, TSC1 (encoding hamartin) or TSC2 (encoding tuberin). TS is characterized by the development of benign tumors in many tissues and organs like the brain, skin, kidneys, eyes, and heart and its neurological manifestations include epilepsy, autism, cognitive and behavioral dysfunction, and giant cell tumors. The diagnosis of TSC is primarily made clinically based on recently updated diagnostic criteria (two major or one major with at least two minor), which include eleven major and six minor clinical findings.[14–16] Our case, antepartally inside the uterus and after delivery the baby has only one major criteria that is cardiac rhabdomyoma with no other additional major and minor clinical features. In one series of cases, rhabdomyoma was identified in 19 fetuses; the diagnostic criteria for TSC were met after birth in 15 (79%).[17] Though we could not perform MRI and genetic analysis because of the availability and affordability problem in our set-up we did not find other features of TSC, for this reason, the child should be followed for the features of tuberous sclerosis that can develop at any time in lifetime or do genetic test before concluding that there is no tuberous sclerosis complex.

Even though most rhabdomyomas are asymptomatic, their clinical features depend on the location, size, number, and histologic composition of the tumor. The number of fetal cardiac rhabdomyomas can range from one to 20 tumors, which may be intramural, intracavitary, or in the interventricular septum. Though the above-mentioned sites are common, it can be located in any cardiac chamber. Rhabdomyomas have a biphasic growth pattern in the fetus: they increase between 20 and 32 weeks' gestation, then either begin to regress or remain the same size. The fetal manifestations range from minor signs to life-threatening conditions like arrhythmias, hydrops fetalis, ventricular outflow or inflow obstruction, cardiac failure, and even sudden death.[1–3,8,18–20] In this case, except the tumor size there was no any symptom and sign of complication both while in utero and after birth at one year of age.

The diagnosis of fetal cardiac rhabdomyoma depends on fetal echocardiography, MRI, histology, and genetic analysis. On echocardiography, cardiac rhabdomyomas are visualized as homogenous multiples (90% of the cases), well-circumscribed, rounded, or oval-shaped hyperechogenic masses with a clear boundary. Detailed echocardiography is important not only to diagnose the presence of the tumor but also for its complications like outflow or inflow obstruction, arrhythmia, and affected cardiac function.

Fetal cardiac rhabdomyomas should trigger awareness of a potential coexisting tuberous sclerosis complex that can lead to a poor neurological outcome. This condition is not only uncommon but can be easily unrecognized prenatally in the absence of a meticulous neurosonogram and MRI with careful evaluation of other fetal structures, including the brain and renal parenchyma.[1,12,21,22]

Most of the time Cardiac rhabdomyoma regress spontaneously after birth. However, in some cases, these tumors may cause severe obstructions on the fetal heart and need specific treatment. Treatment of the cardiac rhabdomyomas either medical or surgical is reserved only for symptomatic fetus. Medical treatments that was studied and found to be effective in shrinking of the cardiac rhabdomyomas are everolimus and sirolimus.[14,15,18,19]

Although most cardiac rhabdomyomas have a relatively benign perinatal course, the long-term prognosis is determined by the neurological manifestations associated with tuberous sclerosis.[23]

## Conclusion

To our knowledge, there was no fetal cardiac rhabdomyoma diagnosed intrauterine and followed up to one year of age without other features of tuberous sclerosis is reported from our country Ethiopia. Even though there is no feature of TSC, one should follow the child for the development of tuberous sclerosis complex or do genetic test before concluding that the child has no feature of TSC.

## Patient Consent

Written consent has been obtained from the patient after she has seen the detailed description of the case and video to be published. No institutional approval was required for the publishing of this case report.

## Author Contributions

All authors contributed to data analysis, drafting or revising the article, gave final approval of the version to be published, agreed to the journal submitted to, and agree to be accountable for all aspects of the work.

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## Disclosure

The authors have no financial or other conflicts of interest in this study.

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