

## FETAL CARDIAC RHABDOMYOMA WITHOUT TUBEROUS SCLEROSIS: A CASE REPORT

Ahmet Karatas<sup>1</sup>, Zehra Karatas<sup>2</sup>, Tulay Ozlu<sup>1</sup>

<sup>1</sup> Department of Obstetrics and Gynaecology, Abant Izzet Baysal University Medical School, Bolu, Turkey

<sup>2</sup> Department of Pediatric Cardiology, Abant Izzet Baysal University Medical School, Bolu, Turkey

Correspondence to: Ahmet Karatas (akaratas1973@hotmail.com)

DOI: 10.5455/ijmsph.2013.290820131

Received Date: 02.08.2013

Accepted Date: 29.08.2013

### ABSTRACT

We report a 34 year old pregnant women in the fetus of whom a 6×7 mm of homogenous, hyperechoic mass lesion that was supposed to be a rhabdomyoma was observed in the left ventricle at the site of interventricular septum at the 24th gestational week. No extracardiac abnormality was detected at either fetal MRI or postnatal investigations of the urinary system and eye, therefore, tuberous sclerosis was not suspected. Cardiac functions and rhythm was normal and the lesion decreased in size within 3 months after birth.

**Key-Words:** Cardiac Mass; Fetal Echocardiography; Rhabdomyoma

### Introduction

Fetal cardiac rhabdomyoma is a rare condition.<sup>[1]</sup> It is the most common cardiac tumour in children and more than 60% of the cases are associated with tuberous sclerosis (TS).<sup>[1,2]</sup> Most of them are histologically benign tumours that regress spontaneously, and usually do not require any treatment. In some cases, depending on their location, they may cause obstruction of blood flow, arrhythmias, stillbirth or sudden death.<sup>[1]</sup> With the advancements of imaging techniques such as antenatal ultrasound, fetal echocardiography and magnetic resonance imaging (MRI), prenatal diagnosis of fetal cardiac tumors has increased in last decade.<sup>[3]</sup>

Tuberous sclerosis is an autosomal dominantly inherited syndrome characterised by tumoural and non-tumoural proliferations and abnormalities affecting many different organs or systems like central nervous system, heart, kidney, skin and retina. Cerebral cortical abnormalities, subependymal tumors, convulsions, mental retardation, renal angiomyolipomas and cardiac rhabdomyomas may coexist with this syndrome.<sup>[4]</sup>

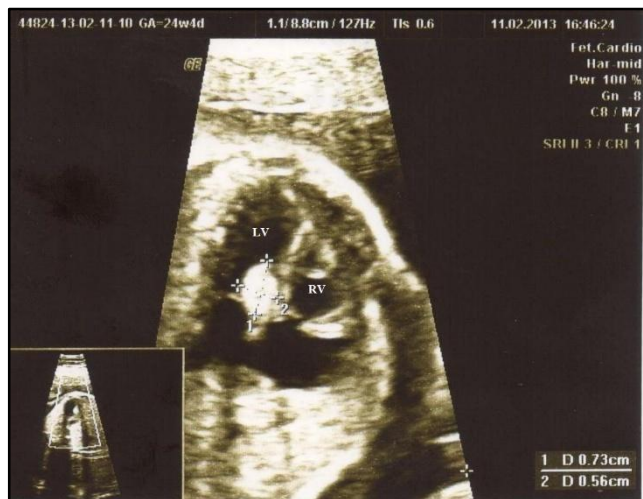
We report a case diagnosed antenatally with cardiac rhabdomyoma which was asymptomatic, isolated and mostly regressed within three

months after birth.

### Case Report

A 34-years old G2P1 pregnant woman admitted to our outpatient clinic at 24 weeks 2/7 days of gestational age for routine pregnancy control. Ultrasound examination revealed fetal biometric measurements that were concordant with the gestational age and a thick interventricular septum of the fetal heart. Examination of the fetal heart was suboptimal because of the inappropriate fetal position so another appointment was scheduled for a repeat examination. In the next examination, the fetus was at appropriate position and a 6×7 mm of homogenous, hyperechoic mass lesion that was supposed to be a rhabdomyoma was observed in the left ventricle at the site of interventricular septum (Figure 1). Fetal echocardiography showed that the mass did not obstruct the ventricular outflow and that the cardiac functions were normal. No associated congenital cardiac defect was observed. Fetal MRI was performed to find out if there is any extracardiac abnormality that could lead to a diagnosis of TS. No extracardiac abnormality was observed at fetal MRI. The size of the mass lesion at the 34th gestational week was 9×11 mm. The patient delivered a 2880 gm female infant at the 39th gestational week. There was no family history of

TS. Urinary system ultrasound and fundoscopic examination of the infant were normal, therefore TS was not suspected. Postnatal echocardiography showed a multifocal, cauliflower like mass lesion at a size of 11×13 mm at the tricuspid and mitral valve annulus (Figure 2). Cardiac functions were normal, and there was no arrhythmia. Echocardiography at the postnatal 3rd month showed that the size of the mass lesion was decreased. The infant is continuing her follow up visits at our clinic.



**Figure-1:** Intracardiac mass lesion detected at antenatal ultrasound (RV: right ventricle, LV: left ventricle)



**Figure-2:** Echocardiographic image of a multifocal hyperechogenic mass lesion at the tricuspid and mitral valve annulus (RV: right ventricle, LV: left ventricle)

## Discussion

Cardiac rhabdomyomas are usually asymptomatic tumors. Prenatal diagnosis of a rhabdomyoma at 15<sup>th</sup> gestational week was firstly reported in 1982<sup>[5]</sup> and by the advancement of imaging technics in the last decades, prenatally diagnosed cases have increased. Nevertheless more than 85 % of them are detected after 24 weeks of

gestation.<sup>[3]</sup> Ultrasound evaluation of the fetal heart should be performed when the fetus is at an optimal position (fetal spine at the posterior side and fetal heart facing to the probe) as described by Allan et al.<sup>[6]</sup> Examinations performed at suboptimal fetal positions may lead to errors in diagnosis, and especially small lesions may be missed from diagnosis. The size of a rhabdomyoma may increase in utero, but rapid growth it not common.<sup>[7]</sup> Fetuses with a tumor  $\geq 20$  mm in diameter have a higher risk of perinatal death. Obstructive and electrophysiological changes that resulted in cardiovascular compromise have been reported.<sup>[8]</sup> Fetal brady and/or tachyarrhythmia were frequently reported to be associated with hydrops. Persistence of arrhythmia in postnatal life increases the risk of neonatal and infant death.

Cardiac rhabdomyoma may be an in utero sign of TS. Sonographic detection of a fetal cardiac tumor should warrant further investigation for the possible presence of associated disorders. Due to its genetic nature, first-degree relatives of a patient with TS should be investigated with cerebral computed tomography, chest X-ray, kidney, skin, ophthalmological and echocardiographic examinations.

Due to their asymptomatic nature, rhabdomyomas usually do not require surgery but they may occasionally cause obstruction, heart failure or arrhythmias. In the present case, the fetus was followed up until term since there was no hemodynamic abnormality or arrhythmia. We observed a decrease in the size of the mass lesion at the postnatal 3rd month. It is known that a large rhabdomyoma size and hydrops are significantly associated with poor neonatal outcome.<sup>[3]</sup> A meta-analysis conducted by Chao et al.<sup>[3]</sup> showed that multifocality of fetal cardiac rhabdomyomas and a positive family history of TS are two strong predictors of neonatal outcome. Gorincour et al.<sup>[9]</sup> suggested fetal MRI as an additional imaging modality for the detection of TS, despite absence of cerebral involvement by MRI could not guarantee a better postnatal outcome. They reported that MRI was able to show increased densities of parenchymal, cortical, subcortical and subependymal tubers in T1-weighted sequences.

Although the regression mechanism is not yet well understood, it has been reported that cardiac rhabdomyomas may regress in the first few years of life spontaneously. In a series of 154 patients with TS, Jozwiak et al.<sup>[10]</sup> reported a partial regression of the cardiac rhabdomyomas in 50% of cases and complete regression in 18%. They reported that early complete regression occurs before 6 years old. In addition to this, it should also be noted that they may grow or appear de novo in 4% of patients with TS.

## Conclusion

Detection of rhabdomyomas in the fetal period is important since they can lead to sudden death in some of the cases while they can regress spontaneously in the others. Fetal heart should be examined in optimal fetal position during routine antenatal ultrasonographic evaluation, and in the presence of a mass lesion in the fetal heart, rhabdomyoma should be suspected as the most possible diagnosis. Such cases should be taken into close echocardiographic follow up since these lesions can lead to obstruction or arrhythmia. Its possible coexistence with TS should be investigated.

## References

1. Isaacs H Jr. Fetal and neonatal cardiac tumors. *Pediatr Cardiol.* 2004;25(3):252-73.

2. Harding CO, Pagon RA. Incidence of tuberous sclerosis in patients with cardiac tumors in early infancy and childhood. *J Thoracic Cardiovasc Surg.* 2005;129:1358-63.
3. Chao AS, Chao A, Wang TH, Chang YC, Chang YL, Hsieh CC, et al. Outcome of antenatally diagnosed cardiac rhabdomyoma: case series and a meta-analysis. *Ultrasound Obstet Gynecol.* 2008;31(3):289-95.
4. Gomez MR. Neurocutaneous disease. In: Bradley WC, Daroff RB, Fenichel GM, Marsden CD. *Neurology in Clinical Practice.* 2nd ed. Oxford: Butterworth Heinemann, 1996:1566-71.
5. DeVore GR, Hakim S, Kleinman CS, Hobbins JC. The in uterodiagnosis of an interventricular septal cardiac rhabdomyoma by means of real-time-directed, M-mode echocardiography. *Am J Obstet Gynecol.* 1982;143:967-969.
6. Allan LD, Crawford DC, Chita SK, Tynan MJ. Prenatal screening for congenital heart disease. *Br Med J (Clin Res Ed).* 1986;292(6537):1717-9.
7. Fesslova V, Villa L, Rizzuti T, Mastrangelo M, Mosca F. Natural history and long-term outcome of cardiac rhabdomyomas detected prenatally. *Prenat Diagn.* 2004;24:241-8.
8. Emmel M, Brockmeier K, Sreeram N. Rhabdomyoma as accessory pathway: electrophysiologic and morphologic confirmation. *Heart* 2004;90:43.
9. Gorincour G, Gire C, Nassi C, D'Ercole C. Pitfalls in the prenatal diagnosis and prognosis of Bourneville sclerosis. *Ultrasound Obstet Gynecol.* 2003;21:409-411.
10. 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:e1146-51.

**Cite this article as:** Karatas A, Karatas Z, Ozlu T. Fetal cardiac rhabdomyoma without tuberous sclerosis: A case report. *Int J Med Sci Public Health* 2013; 2:1095-1097.

**Source of Support:** Nil

**Conflict of interest:** None declared