

Fetal Cardiac Rhabdomyoma as a Representing Sign of Tuberous Sclerosis: Case Report

Tuberoz Sklerozun Eşlik Ettiği Fetal Kardiyak Rabdomiyom

Gülengül NADİRGİL KÖKEN,^a
Mine KANAT PEKTAŞ,^a
Ayhan PEKTAŞ,^b
Bekir Serdar ÜNLÜ,^a
Reşit KÖKEN^c

Departments of
^aObstetrics and Gynecology,
^bPediatric Cardiology,
^cPediatric Neurology,
Afyon Kocatepe University
Faculty of Medicine,
Afyonkarahisar

Geliş Tarihi/Received: 16.06.2013
Kabul Tarihi/Accepted: 28.12.2013

*This case report has been presented at
11th National Congress of
Gynecology and Obstetrics which was held at
Antalya between 15-19 May 2013.*

Yazışma Adresi/Correspondence:
Mine KANAT PEKTAŞ
Afyon Kocatepe University
Faculty of Medicine,
Department of Obstetrics and Gynecology,
Afyonkarahisar,
TÜRKİYE/TURKEY
kanat.mine@gmail.com

ABSTRACT Tuberous sclerosis is one of the neurocutaneous syndromes which are defined with tumoral and non-tumoral proliferations and anomalies. Cardiac rhabdomyomas are diagnosed in 43% to 60% of the individuals who are affected by tuberous sclerosis. Due to this close relationship, the individuals who are diagnosed with cardiac rhabdomyomas should be assessed in aspect of tuberous sclerosis and the individuals who are identified with a high index of suspicion for tuberous sclerosis should be examined to specify any existing cardiac tumor. This case report describes the diagnosis and spontaneous regression of an intrauterine cardiac rhabdomyoma which eventually turned out to be a representing sign of tuberous sclerosis during neonatal period.

Key Words: Pregnancy; rhabdomyoma; tuberous sclerosis

ÖZET Tuberoz skleroz, tümoral ve nontümoral proliferasyonlar ve anomaliler ile tanımlanan nörokutanöz sendromlardan biridir. Bu sendromun etkilediği olguların %43-60 kadarında kardiyak rabdomiyom mevcuttur. Rabdomiyomlar, çocukluk çağında en sık görülen kalp tümörleridir. Tuberoz skleroz ile kardiyak rabdomiyomlar arasındaki yakın ilişki nedeniyle, kardiyak rabdomiyom saptanan olgular tuberoz skleroz bakımından araştırılmalı ve tuberoz skleroz varlığından şüphelenilen olgular kardiyak rabdomiyom yönünden taranmalıdır. Bu yazıda, prenatal dönemde kardiyak rabdomiyom tanısı konulduktan sonra postnatal dönemde yapılan incelemelerle tuberosklerozun belirlendiği ve klinik izleminde kardiyak rabdomiyomun gerilediği bir olgu sunulmuştur.

Anahtar Kelimeler: Gebelik; rabdomiyoma; tüberosklerozis

Türkiye Klinikleri J Case Rep 2014;22(4):266-9

Tuberous sclerosis is one of the neurocutaneous syndromes which are defined with tumoral and non-tumoral proliferations and anomalies. This hamartomatous polyposis syndrome causes the involvement of central nervous system, skin, retina, heart, kidneys, lungs, bones, gastrointestinal system and endocrine glands. Thus, tuberous sclerosis results in cerebral cortical anomalies, subependymal tumors, seizures, mental retardation, renal angiomyolipomas and cardiac rhabdomyomas.^{1,2} It is well known that cardiac rhabdomyomas exist in 43% to 60% of the individuals with tuberous sclerosis.² Due to this close relationship, the individuals who are diagnosed with cardiac rhabdomyomas should be assessed for tuberous sclerosis and the individuals with a high index of suspicion for tuberous sclerosis should be examined for any existing intra-cardiac tumor.¹

This case report describes the diagnosis and spontaneous regression of an intrauterine cardiac rhabdomyoma which eventually turned out to be a representing sign of tuberous sclerosis during neonatal period.

CASE REPORT

A 21-year-old, gravida 2 para 1 woman was admitted to the Department of Obstetrics and Gynecology at the study center. The patient revealed that she was married to her first degree cousin and she gave birth to a healthy baby boy by cesarean section three years ago. According to her last menstrual period, she had a singleton 32 week old pregnancy. The ultrasonographic examination showed a living fetus with biometrical measurements that were compatible with those of 32 week gestation. Moreover, a hyperechogenic mass of 13 mmx12 mmx10 mm was detected within the fetal heart (Figure 1). Due to previous history of cesarean delivery, a baby boy with a birth weight of 3200 grams (25-50 p), length of 49 cm (10-25 p) and head circumference of 34 cm (10 p) was delivered by cesarean section at 39th week of pregnancy.

The newborn had systolic arterial blood pressure of 72 mmHg, diastolic arterial blood pressure of 43 mmHg and arterial oxygen saturation of 98%. There were no cardiac murmurs or dysrhythmias on cardiac auscultation. Complete blood count of the newborn was reported as hemoglobin: 17.1 g/dL, hematocrit: 47.6%, leukocyte count: 16 600/mm³ and platelet count: 334 000/mm³. The hepatic and renal function tests of the newborn were within the normal limits. Echocardiography demonstrated four rhabdomyomas (with diameters of 17 mm, 9 mm, 3 mm and 1 mm) which were situated inside the left ventricle and over the intraventricular septum (Figure 2). Moreover, 4 mm-wide patent foramen ovale and 2 mm-wide ductus arteriosus were visualized. Telecardiographic and electrocardiographic findings were normal.

Cranial tomography of the newborn indicated the existence of subependymal tubers (Figure 3). Electroencephalography and neurological examination were completely normal. Both dermatolog-

ical examination (under Wood's lamp) and ophthalmological assessment revealed nothing pathological. Genetic analysis could not be carried out due to technical inadequacy.

Since cardiac rhabdomyomas did not cause obstruction and dysrhythmias, neither surgery nor anti-arythmic treatment was initiated. It was noted that intracardiac masses of the infant underwent spontaneous regression at the third month of life. However, phenobarbital treatment (5 mg/kg, orally) was initiated as he began to experience grand mal seizures.

DISCUSSION

Primary cardiac tumors are rarely encountered in children. As declared by the autopsy studies, the incidence of primary cardiac tumors is 0.27% during childhood. Rhabdomyomas are the most frequently observed cardiac tumors at the pediatric age group. The incidence of cardiac rhabdomyomas has been reported as 1/40 000 during the first month of life. These tumors usually appear as multiple masses that are situated inside the ventricles and near the interventricular septum.³⁻⁵

Cardiac rhabdomyomas may remain asymptomatic or cause symptoms related with intracardiac obstruction, atrioventricular valve dysfunction and



FIGURE 1: Ultrasonography demonstrated a hyperechogenic mass of 13 mm x12 mm x10 mm within the left ventricle of the fetal heart (as shown by white arrows).

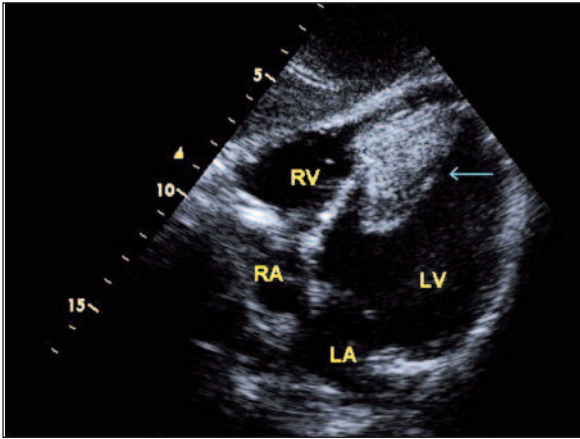


FIGURE 2: Echocardiography demonstrated a 17 mm-wide rhabdomyoma which was located over the intraventricular septum (as shown by the blue arrow).

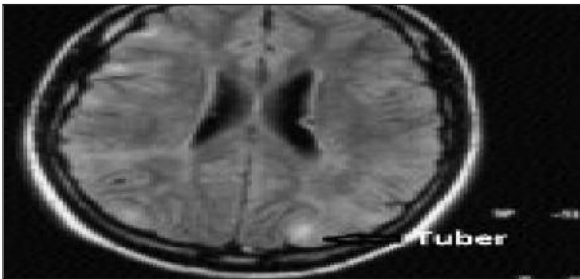


FIGURE 3: Subependymal tubers (as shown by the black arrow) were visualized by cranial tomography.

cardiac failure. Thus, these intracardiac masses may lead to arrhythmia/dysrhythmias, pericardial effusion, fetal hydrops and even sudden fetal/neonatal demise. Ventricular tachycardia, supraventricular tachycardia and Wolff-Parkinson-White syndrome are the dysrhythmias that are associated with cardiac rhabdomyomas.⁶⁻¹⁰

Echocardiography is the primary method in the diagnosis and clinical follow up of cardiac rhabdomyomas. These tumors can be visualized as hyperechogenic masses by means of echocardiography which is performed after 32th week of pregnancy and during postnatal period. Due to their close relationship, echocardiographic demonstration of a cardiac rhabdomyoma should be addressed as the first sign of tuberous sclerosis in case of positive family history or consanguineous marriage.^{1,2,6-10}

Tuberous sclerosis is generally identified with the triology of mental retardation, epilepsy and adenoma sebaceum. Despite being inherited by autosomal dominant pattern, family history is negative in nearly 75% of the affected individuals due to the existence of new mutations. Molecular characterization of related mutations of TSC1 and TSC2 genes might be helpful in predicting short- and long-term neurodevelopmental outcomes.⁷

Cardiac rhabdomyomas are detected in about half of the patients with tuberous sclerosis. The majority of the tuberous sclerosis patients with cardiac rhabdomyomas are asymptomatic while the symptomatic patients are lost during the first two weeks of life.⁶⁻¹¹

The most noteworthy characteristic of this intracardiac tumor is that it may undergo spontaneous regression. That is, prominent rhabdomyomas which were spotted before birth may even disappear during childhood. Previously published studies claim that cardiac rhabdomyomas resolve spontaneously in approximately 70% of the patients who are able to achieve four years of age. Since the overall outcome of isolated cardiac rhabdomyoma appears to be favorable, conservative approach based on echocardiographic assessment is usually recommended for these intracardiac tumors. Surgery may be required in case of mechanical obstruction or life-threatening dysrhythmias.^{3,4}

This case report describes the diagnosis of a rhabdomyoma which appeared as multiple intracardiac masses residing within the interventricular septum and left ventricle at the 32nd week of pregnancy. Clinical follow up with echocardiography was scheduled since there were no clinical signs or symptoms and mechanical obstruction could be specified. A diagnosis of tuberous sclerosis is made when subependymal tubers were visualized by cranial tomography. Repeat echocardiography at the third month of life indicated spontaneous dissolution of the intra-cardiac tumor.

In conclusion, prenatal evaluation of the fetus by ultrasonography should include a detailed assessment of the fetal heart so that any existing intra-cardiac mass could be identified. Whenever

the presence of an intra-cardiac mass is established, rhabdomyomas are the first to be considered in differential diagnosis. Any fetus diagnosed with cardiac rhabdomyoma should be examined thoroughly by echocardiography and any subsisting mechanical stenosis, obstruction or dysrhythmia should be determined carefully. A newborn with cardiac rhabdomyoma should be treated in accordance with the clinical symptoms and signs

and the parents should be counseled about the life-threatening complications such as mechanical and rhythmical problems. Moreover, the close association between cardiac rhabdomyoma and tuberous sclerosis should be taken into account. Systematic postnatal evaluation of tuberous sclerosis should be performed even in cases of cardiac rhabdomyoma without a family history of tuberous sclerosis.

REFERENCES

1. Borkowska J, Schwartz RA, Kotulska K, Jozwiak S. Tuberous sclerosis complex: tumors and tumorigenesis. *Int J Dermatol* 2011;50(1):13-20.
2. Bezircioglu IE, Cetinkaya B, Bicer-Bulbul MA, Yavuzcan A, Baloglu A. [Fetal cardiac rhabdomyoma with tuberous sclerosis: a case report]. *Turkiye Klinikleri J Gynecol Obst* 2008; 18(4):280-3.
3. Ozbarlas HS. [Benign soft tissue tumors]. *Turkiye Klinikleri J Surg Med Sci* 2006;2(52): 45-8.
4. Suma S, Polat P. [Cardiac masses]. *Turkiye Klinikleri J Radiol-Special Topics* 2011;4(3): 76-83.
5. Pucci A, Botta G, Sina N, Tibaldi M, Valori A, Grosso E, et al. Life-threatening tumors of the heart in fetal and postnatal age. *J Pediatr* 2013; 162(5): 964-9.e1.
6. Anaya-Reyes P, Rodriguez-Rábago MJ. [Prenatal diagnosis of cardiac rhabdomyoma: case report]. *Ginecol Obstet Mex* 2013;81(8):477-81.
7. Lee KA, Won HS, Shim JY, Lee PR, Kim A. Molecular genetic, cardiac and neurodevelopmental findings in cases of prenatally diagnosed rhabdomyoma associated with tuberous sclerosis complex. *Ultrasound Obstet Gynecol* 2013;41(3):306-11.
8. Atalay S, Aypar E, Uçar T, Altuğ N, Deda G, Teber S, et al. Fetal and neonatal cardiac rhabdomyomas: clinical presentation, outcome and association with tuberous sclerosis complex. *Turk J Pediatr* 2010;52(5):481-7.
9. Madueme P, Hinton R. Tuberous sclerosis and cardiac rhabdomyomas: a case report and review of the literature. *Congenit Heart Dis* 2011;6(2):183-7.
10. Buyukkurt S, Has R, Eker Omeroglu R. [Fetal cardiac rhabdomyoma and tuberous sclerosis: case report]. *Turkiye Klinikleri J Gynecol Obst* 2003;13(3):249-52.
11. Bostan OM, Cil E. Different cardiac symptoms in two tuberous sclerosis cases. *Turkiye Klinikleri J Pediatr* 2002;11(1):33-6.