A CASE REPORT

Fetal cardiac rhabdomyoma: a case report

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ABSTRACT

Background: Fetal cardiac rhabdomyoma is a rare condition. Case: We report a case with cardiac mass discovered in utero by prenatal ultrasonography at 33 weeks of gestational age. An echogenic round-oval shape mass at the interventricular septum protrudes to left ventricle was observed. Results: After birth, the baby was followed up for 7 months with echocardiography, physical examination, and computerized tomography scan, to rule out anything related to tuberous sclerosis. The prognosis depends on the size, site, number of tumors, and co-existing congenital abnormalities. Management highly depends on the presence of outflow tract obstruction of the heart. However, some cases may regress after birth.

Keywords: rhabdomyoma, fetal heart, tuberous sclerosis.


INTRODUCTION

The presence of a primary congenital tumor of heart is a rare occurrence. Its incidence is 1-2/10000 and 90% of these tumors are benign out and rhabdomyoma is the most common.1 Fetal heart rhabdomyoma is a benign tumor closely related to tuberous sclerosis. Even though it is rare, however, rhabdomyoma is the most common tumor found among other heart tumors (50-78%).1,2 Rhabdomyoma is often found in neonates and toddlers but rarely diagnosed before birth.3,4 Usually rhabdomyoma is diagnosed when complications such as heart valve obstruction, present at the first year of life.

Prenatal detection can be performed in the second trimester of pregnancy.5 Until now the etiology still remains unknown. The pathogenesis is unclear. It is stated that embryonal myoblast proliferation, which forms a single or multiple solid mass which can reside anywhere within the myocardium of the atrium or ventricle protrude into the heart cavity. The differential diagnoses are teratoma, myxoma, hemangioma, and mesothelioma.1,2 The prognosis depends on the size, site, number of tumors, and co-existing congenital abnormalities. Management highly depends on the presence of outflow tract obstruction of the heart. However, some cases may regress after birth. The most important point is that there should be an evaluation on tuberous sclerosis in the patients or their families.1,2 We reported one case of fetal cardiac rhabdomyoma, which found incidentally during a routine antenatal visit.

CASE REPORT

A 30-year-old woman on her third pregnancy with two offspring came to the obstetrics-gynecology outpatient clinic in Sanglah General Hospital on 20th of October 2004 to have her routine prenatal care. She has had two caesarean sections for her previous pregnancies. The last menstrual period was on 26th of February 2004. On the ultrasonography examination, a single live fetus was found, 33 weeks and 6 days’ gestational age, the estimated due date was on 2nd of December 2004. In the fetal heart, we found a round-oval shape mass protruding into the left ventricle, and the distal part of the tumor was attached to the interventricular septum. The tumor moved freely following the heartbeat. The size of the tumor was 17 mm × 9 mm, with uniform echogenic internal structure (figure 1). The mass did not fill the entire heart ventricle (figure 2) and did not obstruct the fetal heart outflow tract. (figure 3). On routine prenatal care, the fetus did not suffer growth restriction. It was then decided to perform elective cesarean section on 24th of October 2004. A vigorous baby was born weighing 3200 grams, and no major anatomical anomalies were found. During physical examination of the baby on 25th of October, hypopigmented macules were found on the medial part of left arm, right thigh, and back with varying size between 4-8 mm (figure 4).
A CASE REPORT

On echocardiography (26th of November 2004) by a Pediatric Cardiologist in Sanglah General Hospital, the heart valves were found to be normal; a mass was found in the left ventricle which moves freely (during systolic and diastolic); VSD, ASD, or PDA were not found.

There was no proof of tuberous sclerosis upon examination of the patient’s parents.

On 6th of April 2005, the baby had tonic-clonic seizures for 5 minutes, no fever was detected. Head CT-Scan found multiple calcifications in left lateral ventricles of the brain and it supports the diagnosis of tuberous sclerosis (figure 5).

DISCUSSION

Primary solid tumor in the heart is a rare occurrence; however, rhabdomyoma is the most commonly found heart tumor in intrauterine life (58%), among other tumors such as teratoma (19%), myxoma, hemangioma, and mesothelioma. Other
researchers reported that the incidences of rhabdomyoma are 60%-86%;6,7 1.17%;8 36%-42%;2,9 and 60%.10

The etiology is unknown. Rhabdomyomas tumor, which is known as hamartoma, is a benign smooth muscle tumor of the heart which contains immature myocytes.1 Rhabdomyoma comes from the embryonal myoblast in which an abnormal tissue proliferation happens and produces encapsulated single or multiple solid mass, with varying sizes.2

This tumor could grow anywhere in the cardiac muscle, and the most common site is in the ventricle, rather than at heart valves, and usually protrudes inside the heart cavity. Macroscopically, it is seen as a yellow solid tumor and round-oval shape. Specific microscopic finding is spider cell, which is a large and clear cell with glycogen contain cytoplasm that spreads to the plasma membrane.2

Cardiac rhabdomyomas are also known as myocardial hamartomas. There is a strong correlation between cardiac rhabdomyoma and tuberous sclerosis, where the incidence rate of tuberous sclerosis is 50-86% in patients with cardiac rhabdomyoma.2,10,11 Tuberous sclerosis is inherited in an autosomal dominant pattern with varying expressions and high penetration. The gene related to tuberous sclerosis is found on chromosome number 9. The clinical manifestations are skin lesions (depigmentation, sebacea adenoma, shagreen patches), cerebral abnormalities (calcifications and periventricular nodules, seizures, and cerebral atrophy), and phacomata of the retina.3 Dermatologic lesions, uniform hypomelanotic macules, usually present at birth. Diagnosis is based on skin biopsy or Wood Light examination. No correlation between cardiac rhabdomyoma with other genetic abnormalities, except with tuberous sclerosis.2

The differential diagnoses are: rhabdomyoma, teratoma, fibroma, myxoma, and hemangioma. Sonographic this tumor appears as single or multiple, homogeneous echogenic mass within the myocardium. Teratoma appears as a complex mass, which covers the pericardium, and usually related with pericardial effusion. Sometimes double chambered right ventricle is seen. Cardiac fibroma has its own characteristic which is a single lesion usually found in the left ventricle wall. Myxoma is a very rare tumor, usually covers the left ventricle, looks similar to rhabdomyoma, often accompanies by orofacial cleft and cystic renal dysplasia.2,12

Prognosis depends on the number, size, site of the tumor, and whether co-existing abnormalities happen. Tumors that grow in the intracavity can cause a disruption in the intracardiac blood flow which then causes heart failure and hydrops. Cardiac dysrhythmias are often found due to pressure on the conduction system.2 The presence of cardiac dysrhythmias and nonimmune fetal hydrops indicates a poor prognosis.1 The causes of death in fetal cardiac rhabdomyoma are ventricular blood flow obstruction, cardiac dysrhythmias intrauterine myocard infarction, and loss of myocardial function due to the pressure of the large mass.13 Various studies showed that rhabdomyoma is a slow growing intrauterine tumor with little or no growth after birth. This tumor tends to regress spontaneously after birth.12,10

Management of fetal cardiac rhabdomyoma depends on the severity of the complications which arise. It is very important to do a series of ultrasound examinations to evaluate and identify signs or symptoms of congestive heart failure and dysrhythmias. Delivery should be done in tertiary health center where a pediatric cardiologist is available. Conservative management is provided for asymptomatic patients and surgery is prepared for patient with hemodynamic instability.2 Special attention should be given to the central nervous system, to find brain ventricle dilatation, and kidneys dysplasia.2,10 Ruling out tuberous sclerosis is often very challenging because brain manifestations are not sufficiently monitored with ultrasound only.14 Detailed family history especially mental retardation and epilepsy should be explored, and tests should be performed on both parents to see the existence of clinical sign of tuberous sclerosis.2,15

Postnatal management includes serial ultrasound for monitoring signs of congestive heart failure or cardiac dysrhythmias. If asymptomatic, it is advisable to treat conservatively. However, if a cardiac outflow obstruction, persistent arrhythmias, heart failure, or cardiogenic emboli appear, a resection surgery should be considered.1

REFERENCES

A CASE REPORT