Spontaneous Regression of Cardiac Rhabdomyoma with Tuberous Sclerosis

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Fetal cardiac rhabdomyoma is the most common cardiac tumor diagnosed antenatally. It has a strong association with tuberous sclerosis.

A case of cardiac rhabdomyoma was diagnosed at 22 weeks of gestation through an anomaly scan. The scan revealed multiple masses related to the ventricular myocardium with no vessel obstruction. The diagnosis of cardiac rhabdomyoma with tuberous sclerosis was confirmed at birth. Repeated echocardiography at three months of life showed slight regression of the tumor size and no deterioration of the cardiovascular status.

The most common cardiac tumor in children is rhabdomyoma, which may disappear spontaneously without treatment. They could be asymptomatic or present as heart failure and respiratory distress which requires surgical intervention. All these children, even those who are asymptomatic, need regular serial follow-ups including echocardiographs.

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Antenatal diagnosis of cardiac rhabdomyoma was first reported by De Vore et al in 1982[1]. It presented in up to 90% of cardiac tumors in pediatric populations[2]. The incidence of congenital cardiac tumors was 1 in 20,000 births, a prevalence of 0.25% in autopsies and 0.08% among live-born[3,4]. Rhabdomyomas are benign hamartomas of smooth muscle derived from embryonic myoblasts.

Echocardiography has been established as the primary diagnostic tool for the evaluation of cardiac tumors[5]. Rhabdomyomas could be localized within the ventricles but could be found in the vessels which may lead to obstruction of cardiac valves or inflow/outflow tracts. They are typically asymptomatic but may also cause atrial or ventricular arrhythmias, congestive heart failure and heart block.

Studies reported a high association between cardiac rhabdomyomas and positive tuberous sclerosis around 50 to 80%[1,6,7]. Most patients with tuberous sclerosis complex (TSC) have inactivating mutations in either TSC1 or TSC2 gene resulting in activation of mechanistic target of rapamycin (mTOR), which is responsible for cellular activation and response[8].

The prognosis is favorable if detected prenatally; obstruction to the inflow or outflow tract indicates a poor prognosis, the risk of fetal death is 4-6%[1].

Surgical resection of the tumor is not usually done unless it causes severe arrhythmias, valvular obstruction or congestive heart failure[9].

Early therapy with a mechanistic target of rapamycin (mTOR) inhibitors may cause regression of the tumor size, as well as preventing the development of tuberous sclerosis manifestations[10,11].

The aim of this presentation is to report a case of cardiac rhabdomyoma which was diagnosed at 22 weeks of gestation.

THE CASE

A nineteen-year-old Bahraini primigravida of a first-degree consanguineous marriage and spontaneous conception had a regular antenatal follow-up. She is a known case of bicornuate uterus diagnosed in her early pregnancy. Her last menstrual period was on 11 June 2017. An early scan showed the expected date of delivery on 18 March 2018.

An anomaly scan revealed a single fetus with microcephaly and intraventricular rhabdomyoma.

Antenatal fetal echo study confirmed multiple masses associated with the ventricular myocardium, the largest of which measured 18x12mm in the left ventricle, the other two masses measured 14x8mm and 15x7mm associated with the posterior interventricular septum, see figure 1. There was no obstruction to blood inflow or outflow from the ventricles, see figure 2. The presence of multiple cardiac tumors is consistent with rhabdomyoma most likely secondary to tuberous sclerosis, see figure 3.

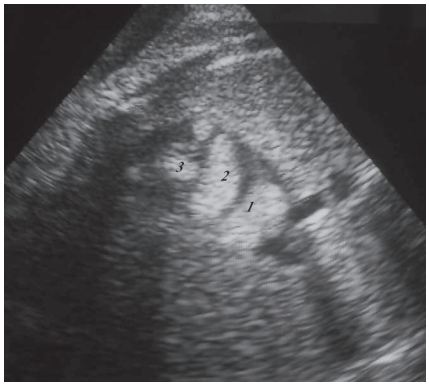

Figure 1: Large Rhabdomyoma Measuring 18x12mm Attached to the Intraventricular Septum in the Left Ventricle

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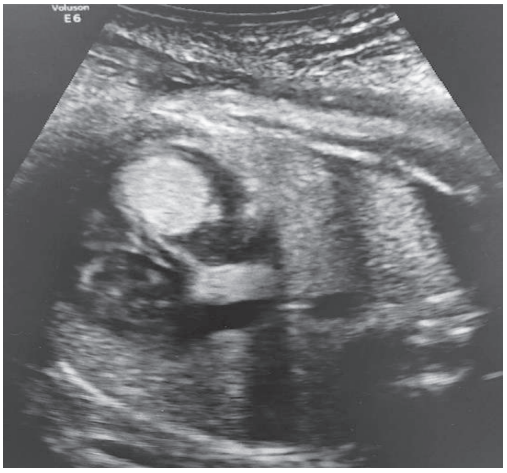

Figure 2: No Obstruction of Blood Inflow or Outflow from the Ventricles

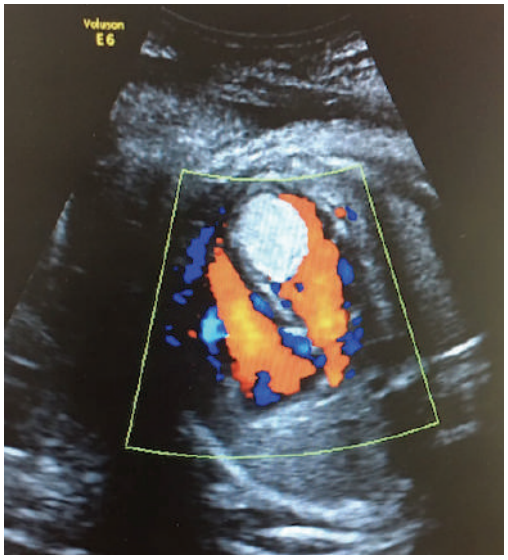

Figure 3: Basal Cut Showing Three Tumors

The patient came in early labor at 37 weeks of gestation with breech presentation and delivered by emergency lower segment cesarean section. A live male baby was born weighing 2.480 kg with an Apgar score of 9 and 10 at one and five minutes respectively. The patient's postnatal course was uneventful and the baby was discharged on the third day postpartum.

At day 1 of life, the neonate developed tachypnea and desaturation; he was kept on nasal continuous positive airway pressure (CPAP). However, oxygen requirement was still high, therefore, he was intubated and kept on patient triggered ventilation (PTV) mode. He was then transferred to high-frequency oscillatory ventilation (HFO) as he was not maintaining saturation. He needed nitric oxide and norepinephrine for persistent pulmonary hypertension (PPHTN). The patient was weaned gradually on day nine of life.

Ultrasound brain revealed normal morphology of cerebral hemispheres and normal echo pattern, no abnormal focal echogenicity, unremarkable appearances of the ventricular system with no evidence of hydrocephalus and no detectable intracerebral, intraventricular or subarachnoid hemorrhage, see figures 4 and 5. Tuberous sclerosis was confirmed.

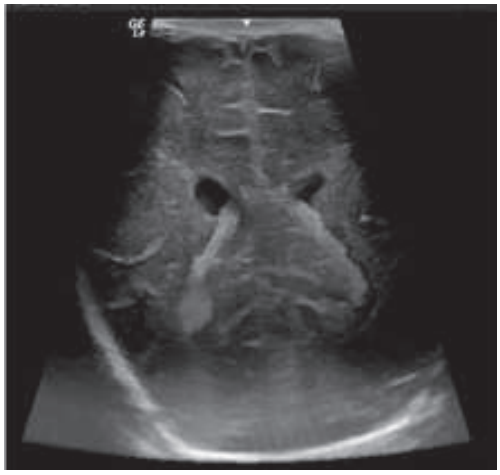

Figure 4: Ultrasound Brain Revealing Normal Morphology of Both Cerebral Hemispheres and normal Echo-Pattern

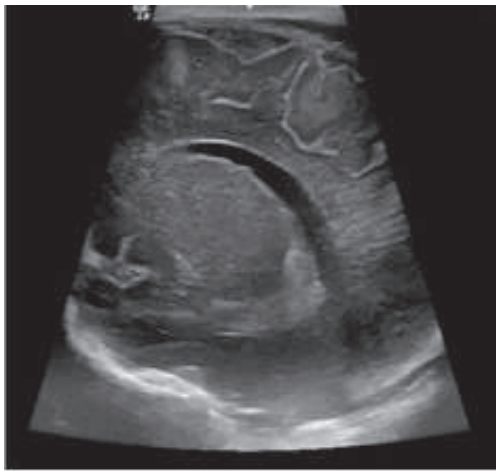

Figure 5: Ultrasound Brain Revealing Normal Morphology of Both Cerebral Hemispheres and normal Echo-Pattåern

After birth, neonatal echo revealed the same number and sizes of tumors as antenatal, with patent foramen ovale (PFO) and patent ductus arteriosis (PDA) shunt around 2-3mm with a right to left flow. There was trivial mitral regurgitation with mild tricuspid regurgitation. No coarctation of the aorta, heart dysfunction or effusion was detected. The infant was in stable condition on day 16 of life and was managed as an outpatient.

Repeat echo at age 3 and 6 months showed a reduction in the size of the largest tumor (14x11mm) as well as the other two tumors (approximately 5x4mm).

DISCUSSION

Cardiac rhabdomyoma accounts for the majority of cardiac tumors in pediatric populations, the majority are asymptomatic and associated with tuberous sclerosis complex and regress spontaneously during the first few months of life.

Kwiatkowska et al reported a newborn with multiple giant cardiac tumors; the largest measured 33x30mm with no obstruction to both inflow and outflow heart tracks. During 3 months' follow-up, the infant remained asymptomatic[12]. In this study, it was stated that asymptomatic infants need close follow-up with serial imaging and electrocardiography as they are at risk of unsuspected death.

Kwiatkowska et al reported 30 children with cardiac tumors[13]. Most of them did not need cardiac surgery except 3 due to severe hemodynamic disturbance. All of them confirmed association with tuberous sclerosis. There was only one death during the follow-up period.

A study was performed by Chen et al on 53 pregnant women with fetal cardiac tumors; thirty-seven had multiple cardiac tumors and the rest had a single cardiac tumor[7]. This study concluded that there is no significant difference between single and multiple tumors regarding the degree of fetal heart damage.

Song et al reported a large rhabdomyoma occupying most of the left ventricle causing flow obstruction[14]. The tumor spontaneously decreased to more than 70% of its size within 3 years and no obstruction.

Helen et al reported a neonate case with a single rhabdomyoma obstructing the left ventricular outflow tract[10]. Everolimus was used for 6 months (0.5mg twice weekly) which showed a significant reduction in the tumor size. The drug was tapered gradually to prevent a rebound increase in the size and late side-effect.

Hoshal et al reported a two-day-old neonate with tumor obstructing the outflow tract of the right ventricle and infiltrated into the left ventricle[11]. Everolimus was started after successful cardiopulmonary resuscitation. It resulted in a successful reduction of tumor size within two months.

Another case was reported by Demir et al of a neonatal cardiac rhabdomyoma using Everolimus[15]. The tumor regressed in size after 75 days of therapy. Three cases reported by Chang et al showed regression of tumor size in all cases after using low dose Everolimus[16].

CONCLUSION

Our case was the first case reported in Bahrain, which was diagnosed antenatally. The tumor regressed in size without medical or surgical treatment.

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