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Case Report

Fetal Rhabdomioma: Report of Three Cases

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Abstract

Rhabdomyomas are the most common fetal cardiac tumor and are generally associated with tuberous sclerosis, a disease in which tumor suppressor genes mutate, leading to abnormal cell proliferation. They can cause a variety of manifestations, with larger tumors potentially causing hemodynamic instability due to outflow tract obstruction, requiring surgical intervention. Therefore, sirolimus has emerged as a conservative management option. This drug is an mTOR inhibitor of rapamycin, which possesses antitumor activity. We present three cases of fetal cardiac rhabdomyomas with different characteristics. Two were treated with sirolimus, resulting in complete regression without adverse effects or the need for surgical intervention. The third patient, with small rhabdomyomas and no hemodynamic compromise, was followed up in an outpatient clinic with excellent progress.

Keywords: Fetal Rhabdomyoma; Cardiac Rhabdomyoma; Prenatal Diagnosis and Tratamiento

Introduction

Congenital cardiac tumors are rare, with an incidence of 0.009% to 0.2%, but thanks to advances in imaging, they are increasingly being detected prenatally [1]. Among these, rhabdomyomas are the most common in fetal life (60-86%) and present as benign, multifocal growths in the ventricular walls or atrioventricular valves [2,3,5]. While most are asymptomatic and regress spontaneously, they can cause severe complications such as flow obstruction, arrhythmias, or heart failure [3]. Their appearance is often pathognomonically associated with Tuberous Sclerosis (TSC), an autosomal dominant genetic disorder (mutations in TSC1 or TSC2) that overactivates the mTOR cell proliferation pathway, leading to the formation of multiple benign tumors in different organs such as the brain, kidney, liver, and heart [4]. When rhabdomyomas cause hemodynamic instability and surgical resection means high morbidity and mortality [4], Sirolimus (Rapamycin) emerges as an effective medical alternative, inhibiting the mTOR pathway and stopping the tumor cell cycle [6], although its use requires strict monitoring due to possible adverse effects such as bone marrow suppression, mucosal alteration or fetal growth restriction [2].

Clinical Cases

- A 26-year-old pregnant woman underwent an ultrasound at 28 weeks gestation, which revealed echogenic nodular foci

in the left ventricle. A fetal echocardiogram performed in our unit described a homogeneous, hyperechoic, space-occupying lesion in the left ventricle. This lesion originated from the interventricular septum and protruded into the left ventricle and outflow tract. It appeared more separated from the aortic valve than in previous studies and showed a slight decrease in size, measuring 12.4 x 13.5 mm (short axis) and 15.1 x 9.9 mm (long axis) (Figure 1). Caudal to this lesion, a pedicle extended, connecting to another lesion with similar characteristics but smaller, measuring 10.8 x 4.2 mm (long axis), which appeared to be attached to the anteroseptal chordae tendineae of the mitral valve. Left ventricular outflow tract flow was measured, yielding a peak velocity of 1.07 m/s and a peak gradient of 4.54 mmHg. The aortic valve had three leaflets, with normal opening and closing, adequate antegrade flow, and no regurgitation. The pregnancy was monitored as high-risk obstetrics, and at 38 weeks, a weak newborn was delivered, requiring tactile stimulation and suctioning of secretions, with a good response. CPAP support was provided at 5 cm H2O for 10 minutes, with a maximum FiO2 of 40%. Apgar scores were 4-8-9, weight 3470 grams, NT 49 cm, and HC 35.5 cm. The patient was admitted to the Neonatal Intensive Care Unit for management and initiation of sirolimus. The patient experienced a slight decrease in tumor size, but it appeared more separated from the aortic annulus and with a decreased gradient. The patient remained asymptomatic and without complications during hospitalization. The patient was discharged home, maintaining treatment with sirolimus until 8 months of age, achieving total regression of the rhabdomyomas.

- A 33-year-old pregnant woman with an obstetric history of two pregnancies and one delivery. No relevant medical history and a BMI of 32. At 33 weeks of gestation, she was referred with a diagnosis of Preterm Labor Syndrome and fetal pericardial effusion. Upon evaluation in our unit, fetal biometry was documented as consistent with 33 weeks of gestation at the 60th percentile. The fetal echocardiogram showed: a normal four-chamber view and normal heart size, with three hyperechoic cardiac tumors. Two were located in the free wall of the left ventricle, measuring 4 and 5 mm in diameter, and the third was in the left ventricular outflow tract, visualized as a hyperechoic image measuring 8 mm in diameter, obstructing the aortic outflow by 80-90%, with an increased velocity of 260 cm/s at the level of the aorta. The mitral valve exhibits moderate regurgitation with a velocity of 240 cm/s. Additionally, there are two small muscular ventricular septal defects. The remainder of the fetal anatomy is normal. Based on the ultrasound characteristics of the tumors, they are classified as cardiac rhabdomyomas. Transfer to a tertiary care center is indicated for evaluation and management of the tumor obstructing the left ventricular outflow tract. At the tertiary center, the diagnosis is confirmed, and an asymptomatic newborn is delivered and treated with sirolimus, resulting in tumor involution during the first year of life. Echocardiographic follow-up showed complete regression of the lesion obstructing the outflow tract, as well as of the tumors in the left ventricular free wall. Currently, the patient is asymptomatic, not taking any medication, and is under follow-up at the neurology outpatient clinic due to the association of the cardiac rhabdomyomas with tuberous sclerosis.
- A 23-year-old pregnant woman diagnosed with a large-for-gestational-age fetus underwent an obstetric ultrasound at 30 weeks and 6 days, which revealed a fetal cardiac nodule beneath the atrioventricular valve. At 35 weeks and 1 day, a fetal echocardiogram was performed, revealing a subvalvular aortic image consistent with a 1.52 square centimeter rhabdomyoma that did not cause obstruction or pathological aortic velocity increase (Figure 2). The heart appeared to be of normal size with four well-balanced chambers within the thorax. The outflow tract was normally related, and the venae cavae axis was normal. Two veins were entering the left atrium. The foramen ovale was patent with a right-to-left shunt. The aortic arch was normal. The thoracoabdominal region was normal. Three vessels were normal. The patient was hospitalized at 38 weeks and 3 days due to the onset of labor. A female newborn weighing 4000 grams was delivered, with Apgar scores of 9-9-9. Eighteen minutes after birth, the infant was given oxygen via nasal cannula due to respiratory symptoms including subcostal retractions, nasal flaring, and grunting audible without a stethoscope. Her oxygen saturation was 88%. She was admitted to the neonatal intensive care unit (NICU) for management and to complete a cardiac evaluation. An echocardiogram performed during hospitalization revealed a 0.7 square centimeter lesion in the left ventricular outflow tract and two additional lesions at the apex, one measuring 0.4 square centimeters and the other

0.3 square centimeters, without outflow tract obstruction or hemodynamic compromise. Based on these findings, sirolimus was not administered, and an outpatient follow-up appointment was scheduled for three months of age at the cardiology clinic.

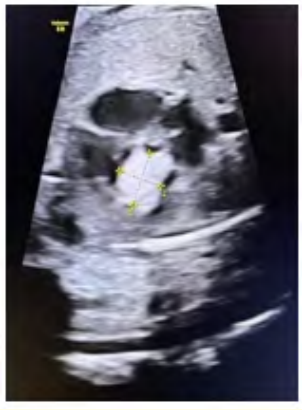

Figure 1: Shows a tumor mass occupying the left ventricular cavity.

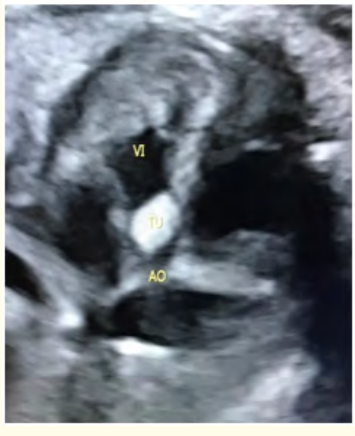

Figure 2: Rhabdomyolysis located in the left ventricular outflow tract obstructing the aorta.

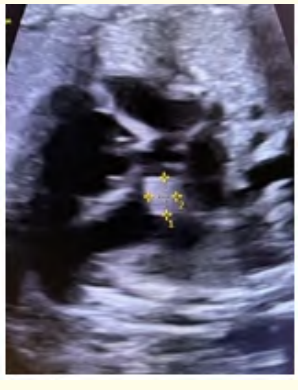

Figure 3: Aortic subvalvular rhabdomyoma.

Discussion

Although rhabdomyomas can occur in isolation, they are usually pathognomonically associated with tuberous sclerosis. The expression of the disease varies significantly among patients. Therefore, it is difficult to predict the degree to which a child will be affected [4,5]. Rhabdomyomas usually have a benign course and are found in the ventricular walls or atrioventricular valves in a multifocal pattern [3,5], as was the case in our patients. They are generally diagnosed in the second trimester and usually grow until the 30th-32nd week of gestation [8]. Our cases presented with obstructive rhabdomyoma of the left ventricular outflow tract, two lesions in the posterior wall of the left ventricle, and another in the left ventricular outflow tract. In the third case, a subvalvular aortic tumor was present.

Most rhabdomyomas are asymptomatic and undergo spontaneous regression over time [7]. However, in some cases, especially in neonates, these tumors can progress and cause hemodynamic instability due to embolization, circulatory obstruction, heart failure, and valve interference, as well as arrhythmias and heart blocks, which can lead to death [3]. Surgical resection of the tumor, which has been the standard treatment to relieve the obstruction, is not always possible and can be associated with significant morbidity and mortality [4]. These tumors require close monitoring with fetal echocardiography, as it is useful for determining the appropriate time of delivery and avoiding major hemodynamic complications.

In tuberous sclerosis, the mutation that occurs overactivates the mTOR (mammalian target of ripamycin) proliferation pathway, leading to the formation of multiple benign tumors in different organs, such as cardiac rhabdomyomas. We believe that frequent follow-up after diagnosis during the fetal period is essential to anticipate possible complications in the fetus's life and thus provide timely treatment and protect fetal viability.

Two of the cases presented were treated with sirolimus (rapamycin). This is a macrocyclic antibiotic discovered in a soil sample from Rapa Nui, produced by the fermentation of Streptomyces hygroscopicus. In addition to its antifungal properties, it was found to have immunosuppressive and antiproliferative effects. It acts as an mTOR inhibitor (mTORi), arresting the cell cycle in the G1-S phase by blocking IL-2-mediated signal transduction. Therefore, its use has been demonstrated in treating and preventing proliferative diseases such as tuberous sclerosis, psoriasis, and malignant lesions [6].

In pregnant women with tuberous sclerosis, targeted fetal ultrasound is mandatory. Once a rhabdomyoma diagnosis is made, continuous monitoring is necessary to track the progression of known lesions and the appearance of new ones. If the tumors are multiple and increase in size, jeopardizing fetal viability, sirolimus therapy has the potential to induce regression of rhabdomyomas in the fetal heart [9,10]. In this way, no other invasive treatment is required. Two of our patients were treated with sirolimus after birth with excellent results and complete regression of the rhabdomyomas.

Conclusion

Our case series is relevant because one of the cardiac tumors almost completely obstructed the left ventricular outflow tract. Postnatal treatment with sirolimus was initiated, resulting in an excellent response and a reduction in the size of the rhabdomyomas without requiring surgical intervention. Similarly, the second case presented with three rhabdomyomas that were treated with sirolimus, leading to complete tumor involution.

We believe that prenatal detection of rhabdomyomas using echocardiography and treatment with sirolimus in fetuses and newborns with large tumors causing hemodynamic compromise is a conservative option with very favorable results, thus avoiding the need for complex surgical interventions with high morbidity and mortality in both the fetal and neonatal periods.

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