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Case Report

Fetal Rhabdomyoma: Case Report.

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Abstract

Rhabdomyoma corresponds to the most common fetal cardiac tumor, and can affect any point of the cardiac muscle, generating various manifestations. Very large tumors can cause hemodynamic obstruction, requiring surgery.

Its development is related to tuberous sclerosis, which produces abnormal cell proliferation, which is why sirolimus, a rapamycin (mTOR) inhibitor, has been used for its antitumor activity. A clinical case is presented where the cardiac rhabdomyoma, despite generating great obstruction in the outlet of the left ventricle, was completely resolved after the administration of sirolimus, without generating adverse effects or the need for surgery.

Keywords: Rhabdomyoma; Cardiac Tumor; Prenatal Diagnosis

Introduction

Cardiac tumors are very rare, with an estimated incidence between 0.0017% and 0.28%. Of these, rhabdomyoma is the most common in fetal life, representing 60-86% of tumors and depending on its size and location, it can cause arrhythmias, obstruction of intracardiac blood flow, congestive heart failure and even sudden death [1].

70 to 80% of them are associated with tuberous sclerosis complex, which is an autosomal dominant disorder with TSC1Y TSC2 mutations; where both genes encode proteins that stimulate the mTOR (mammalian target of rapamycin) proliferation pathway, leading to abnormal cell proliferation and hamartoma formation in multiple organs [2-4].

Treatment will depend on the hemodynamic impact of the tumor, and may require surgical resection in massive tumors that affect cardiac function, or an mTOR inhibitor such as sirolimus (rapamycin), which has been shown to control cell growth until involution. progressive size of the lesions [2,4,5].

Prenatal diagnosis is usually made between the 20th and 30th week of gestation and growth stops after birth. Primary tumors of the heart and its pericardium are rare entities; they have an incidence of no more than 1 in 100,000 in autopsy findings. Metastases are also exceptional, but are more common than primary tumors. The tumors are generally benign. The most common is Rhabdomyoma, followed in frequency by fibroma and myxoma [6,7].

Clinical case

33-year-old pregnant woman with obstetric history of pregnancies two and one. With no relevant medical history, she had an obese nutritional status with ICM 32, Weight: 74 kilos, Height: 152 centimeters. She was 33 weeks pregnant, she was referred with a diagnosis of Premature Birth Syndrome and fetal pericardial effusion. Evaluated in our unit, fetal biometry was documented according to 33 weeks growing at the 60th percentile, with the fetal echocardiogram observing: vision of four chambers normal in orientation and size, presence of three hyperechogenic cardiac

tumors of which two were found in the free wall of the ventricle. left 4-5mm in diameter, another in the left ventricular outflow tract that is visualized as a hyperechogenic image of 8 mm in diameter that obstructs the outflow of the aortic artery between 80-90%, presenting an increase in velocity of 260 at the level of the aorta. cm/s. (Figure 1) The mitral valve presents moderate insufficiency with a speed of 240 cm/s. In addition, it has two small muscular interventricular communications.

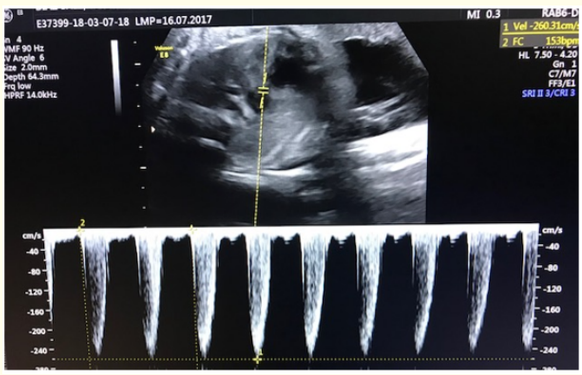

Figure 1: Shows an increase in velocity al the level of the left ventricular outflow tract with a velocity of 260 cm/s measured with pulsed Doppler.

The rest of the fetal anatomy is normal. Due to the ultrasonographic characteristics of the tumors, they are classified as cardiac rhabdomyomas (Figures 2 and 3).

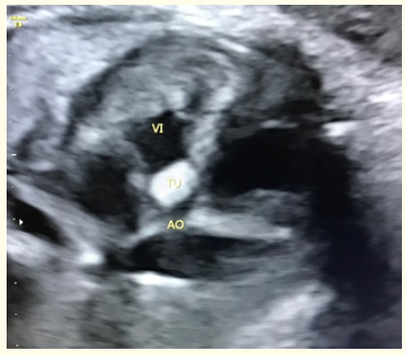

Figure 2: Shows the left ventricular outflow trac with almost complete obstruction of the aorta due to tumor. (Tu).

Transfer to a tertiary level is indicated for evaluation and management of the tumor that obstructs the left ventricular outflow tract.

At the tertiary center the diagnosis is confirmed, an asymptomatic newborn is born and undergoes medical treatment with sirolimus, resulting in the involution of the tumors. During the first year of life, his echocardiographic controls showed total regression of the described lesion that obstructed the outflow tract, as well as

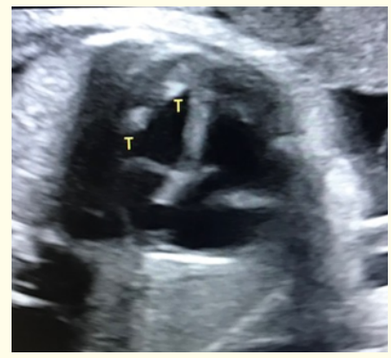

Figure 3: Where two tumors can be seen in the free Wall of the ventricle. (T)

the tumors of the free wall of the left ventricle. Currently, the patient is asymptomatic and does not take medications and is under polyclinic neurology control due to the association with tuberous sclerosis of cardiac rhabdomyomas.

Discussion

Cardiac rhabdomyomas are usually diagnosed early in life or prenatally as in our case. Most fetuses with multiple rhabdomyomas have tuberous sclerosis [8]. Our patient did not have a previous family history of tuberous sclerosis; therefore, it could correspond to a de novo mutation. Fetal cardiac rhabdomyoma is generally identified as a finding in the ultrasound study of the second trimester of pregnancy [6]. Ultrasounds of cardiac rhabdomyoma usually show the presence of echogenic masses located intramural. Our case presented two lesions in the posterior wall of the left ventricle and another in the outflow tract of the left ventricle. The natural course of most prenatally detected tumors is favorable, showing tumor regression; However, they can also progress in the uterus and cause complications in the fetal period such as arrhythmias, heart failure and, in extreme cases, fetal death. After birth, large tumors can affect cardiac output; In such cases, surgical resection or additional treatment will be necessary[9]. Close monitoring with fetal echocardiography can be useful to determine the appropriate time of delivery and avoid important hemodynamic complications in these cases.

The association of cardiac rhabdomyomas with tuberous sclerosis, a genetic disease characterized by benign and multiple tumor lesions, is clearly documented. The interesting thing about our case is that one of these tumors obstructed the outflow tract of the left ventricle and could have caused serious complications both in the fetal period and in the newborn period. We think that frequent follow-up after diagnosis in the fetal period is important to anticipate possible complications in fetal life. Frequent monitoring of patients is indicated to be able to provide timely treatment and thus protect the viability of the fetus.

Our case was treated medically with sirolimus, a first-generation inhibitor of the mTOR protein kinase that controls cell growth, proliferation and survival. Initially used as an anticancer agent, its

use as an immunosuppressant after organ transplantation has been authorized by the FEDA [5,9]. In pregnant women with tuberous sclerosis, targeted fetal ultrasound is mandatory. Once the diagnosis of rhabdomyoma has been made, continuous surveillance is necessary to control the progression of known lesions and the appearance of new ones. If multiple cardiac rhabdomyomas increase in size and put the viability of the fetus at risk, sirolimus therapy has the potential to induce regression of the rhabdomyomas in the fetal heart [5].

Our patient was treated after birth and responded well, the injury returning quickly, allowing it to progress quickly to improvement without requiring any other type of treatment.

Conclusion

Our case is relevant since a cardiac tumor almost completely obstructed the outflow tract of the left ventricle, it was diagnosed in the fetal period and closely monitored, managing to bring the pregnancy to term without the need for in utero treatment, obtaining a newborn in very good condition, post-birth treatment with sirolimus was started, having an excellent response, reducing the size of the rhabdomyomas, not requiring surgery.

Bibliography

1. Chía- Vázquez NG., *et al.* "Importancia del rabdomioma cardiaco en la población pediátrica. Experiencia de 39 años. Serie de casos". *Archivos de Cardiología de México* (2021).

2. Chen X-Q., *et al.* "Sirolimus Can Increase the Disappearance Rate of Cardiac Rhabdomyomas Associated with Tuberous Sclerosis: A Prospective Cohort and Self- Controlled Case Series Study". *The Journal of Pediatrics* (2021).

3. Martinez- Morales MC., *et al.* "Múltiples rabdomiomas intracardiacos en vida fetal simulando atresia tricuspídea al nacimiento". *Perinatología y Reproducción Humana* 36.2 (2022).

4. Dr. Marco Tulio Gómez Cerdas and Dr Johnny Alejandro Vargas Morales. "Esclerosis tuberosa". *Revista Médica Sinergia 4*.3 (2019): 21-37.

5. Park H., *et al.* "Sirolimus therapy for fetal cardiac rhabdomyoma in a pregnant woman with tuberous sclerosis". *Korean Society of Obstetrics and Gynecology* (2019).

6. Somoza Marino. "Cardiopatías congénitas cardiología perinatal segunda edición" (2015): 444.

7. Alfred Abuhamad and Rabih Chaoui. "Ecocardiografía fetal tercera edición Ediciones Journal (2019): 537-546.

8. Sasongko TH., *et al.* "Rapamycin and rapalogs for tuberous sclerosis complex". *Cochrane Database of Systematic Reviews* 7 (2016): CD011272.

9. Bader RS., *et al.* "Fetal rhabdomyoma: prenatal diagnosis, clinical, outcome, and incidence of associated tuberous sclerosis complex". *The Journal of Pediatrics* 143 (2003): 620-624.