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Case report

Prenatal diagnosis of fetal cardiac rhabdomyoma with severe cardiac dysfunction

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Abstract

Fetal cardiac tumours are rare, but rhabdomyomas are the most common. They are associated with tuberous sclerosis in 50–80% of cases. Early diagnosis of cardiac rhabdomyoma is thus important for early diagnosis of tuberous sclerosis. We describe a case of an intracardiac tumour that evolved with significant ventricular dysfunction. The report of this case teaches us an important lesson by exemplifying an unsuccessful experience of rhabdomyoma surgery.

Key words: prenatal diagnosis, fetal cardiac tumour, rhabdomyoma, cardiac dysfunction.

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Introduction

Fetal cardiac tumours are rare, with an incidence of 1–2/10,000 [1]. Among the fetal intracardiac tumours, rhabdomyomas are the most common at around of 60–86% of cases [1, 2].

The aetiology is unknown, but the fetal cardiac rhabdomyoma is associated with tuberous sclerosis in 50–80% of cases [3, 4]. Tuberous sclerosis is an autosomal dominant systemic genetic disorder with variable penetrance, which affects the central nervous system, skin, retina, kidneys, and heart. In approximately 30% of cases the cause is genetic. In 70% of cases, de-novo mutations inactivate the TSC1 (9q34.3) and TSC2 (16p13.3) genes, which encode two proteins: tuberin and hamartin [5]. Hence, early diagnosis of fetal cardiac rhabdomyoma is important for early diagnosis of tuberous sclerosis.

After birth, most of these rhabdomyomas regress without the need for surgical intervention. However, they may present complications even in the fetal period, such as arrhythmias, and progressive compression of cardiac, vascular, and pulmonary structures by a rapidly growing mass or effusion [6]. In this report, we describe a case of fetal intracardiac tumour that evolved with significant ventricular dysfunction.

Case description

We report a 31-year-old pregnant woman who was referred at 26 weeks of gestation to echocardiography examination. The patient had conceived spontaneously, without relevant medical past or familial history, and no genetic disorders were known.

Ultrasonographic examination revealed a homogeneous, hyperechogenic tumour, adhered to the interventricular septum, giving continuity to a mobile protrusion directed to the left

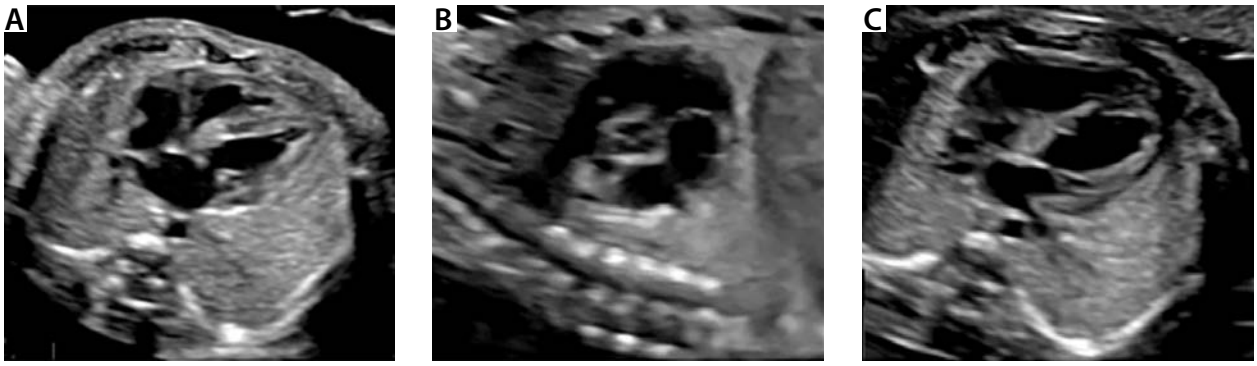

Figure 1. Fetus at 26 weeks. Homogeneous and hyperechogenic tumour, adhered to the interventricular septum (**A**), giving continuity to a mobile protrusion directed to the left ventricular outflow tract, measuring around 5.6 mm × 3.4 mm (**C**), without promising the aortic valve (**B**)

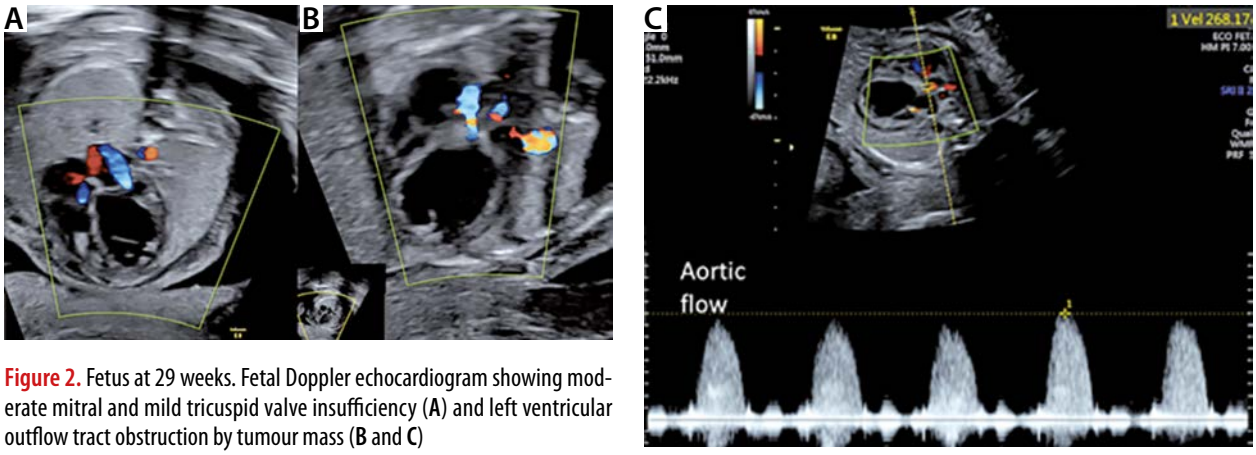

Figure 2. Fetus at 29 weeks. Fetal Doppler echocardiogram showing moderate mitral and mild tricuspid valve insufficiency (**A**) and left ventricular outflow tract obstruction by tumour mass (**B** and **C**)

ventricular outflow tract, measuring around 5.6 mm × 3.4 mm. This mass caused aortic flow obstruction with a maximum velocity of 2.7 cm/s (Figures 1 and 2). The aortic arch showed the isthmus dimension at the limit of normality, according to the *z*-score. The fetal heart was of normal size with no myocardial hypertrophy, without atrioventricular insufficiency, and preserved heart function. A fetal cardiac rhabdomyoma was suspected.

Fetal echocardiography evaluation was performed every three weeks during the pregnancy. Other tumoural lesions appeared in the right ventricular cavity (two located in the apex and one adhered to the tricuspid valve leaflet) and a punctuated lesion in the left ventricle. The tumour located in the left ventricular outflow tract, and the aortic flow decreased to a maximum velocity of 1.8 cm/s. There was a significant increase in the cardiac area, with significant left ventricular dilatation and impairment of systolic function (which probably underestimated the aortic flow velocity). Fetal heart rhythm was normal. There was no evidence of pericardial effusion or ascites. Other visceral tumours were not observed on focused scanning (Figure 3).

The patient received betamethasone for fetal lung maturation. Serial ultrasonographic examinations were performed. The size of the tumour increased in the follow-up to 14 mm × 7 mm. At 35 weeks of gestation, premature delivery by caesarean section was chosen due to deterioration of fetal cardiac function.

The male newborn was delivered, weighing 1.995 g, and the 5- and 10-minute Apgar scores were both 9. The neonate was respiratorily instable and was intubated. Neonatal echocardiography confirmed the prenatal diagnosis of cardiac rhabdomyomas and ascending aorta hypoplasia (Figure 4). There was evidence of outflow tract obstruction with hemodynamic compromise. To prevent cardiac insufficiency the neonate received anticongestive medication.

The newborn underwent pulmonary trunk banding surgery to direct blood flow to the aorta through the ductus arteriosus, to maintain appropriate cardiac output and assist in the development of the ascending aorta and transverse arch. Thus, the neonate evolved during hospitalisation, maintaining a good flow in the aorta, and with consequent increase in the dimensions of the aortic arch structures. The newborn was discharged at one month of life. At the first outpatient visit at the 36-day, he underwent a new echocardiogram that showed significant right ventricle dilation, major right ventricle dysfunction, discrete left ventricle dysfunction, and closed arterial canal. The ascending aorta had dimensions within normal range. The tumour located in the left ventricular outflow tract measuring 14 mm × 9 mm and maximum aortic flow of 3.9 cm/s. The child was hospitalised for haemodynamic stabilisation with dobutamine introduction and programming of a new surgical approach for removal of the bandage. However,

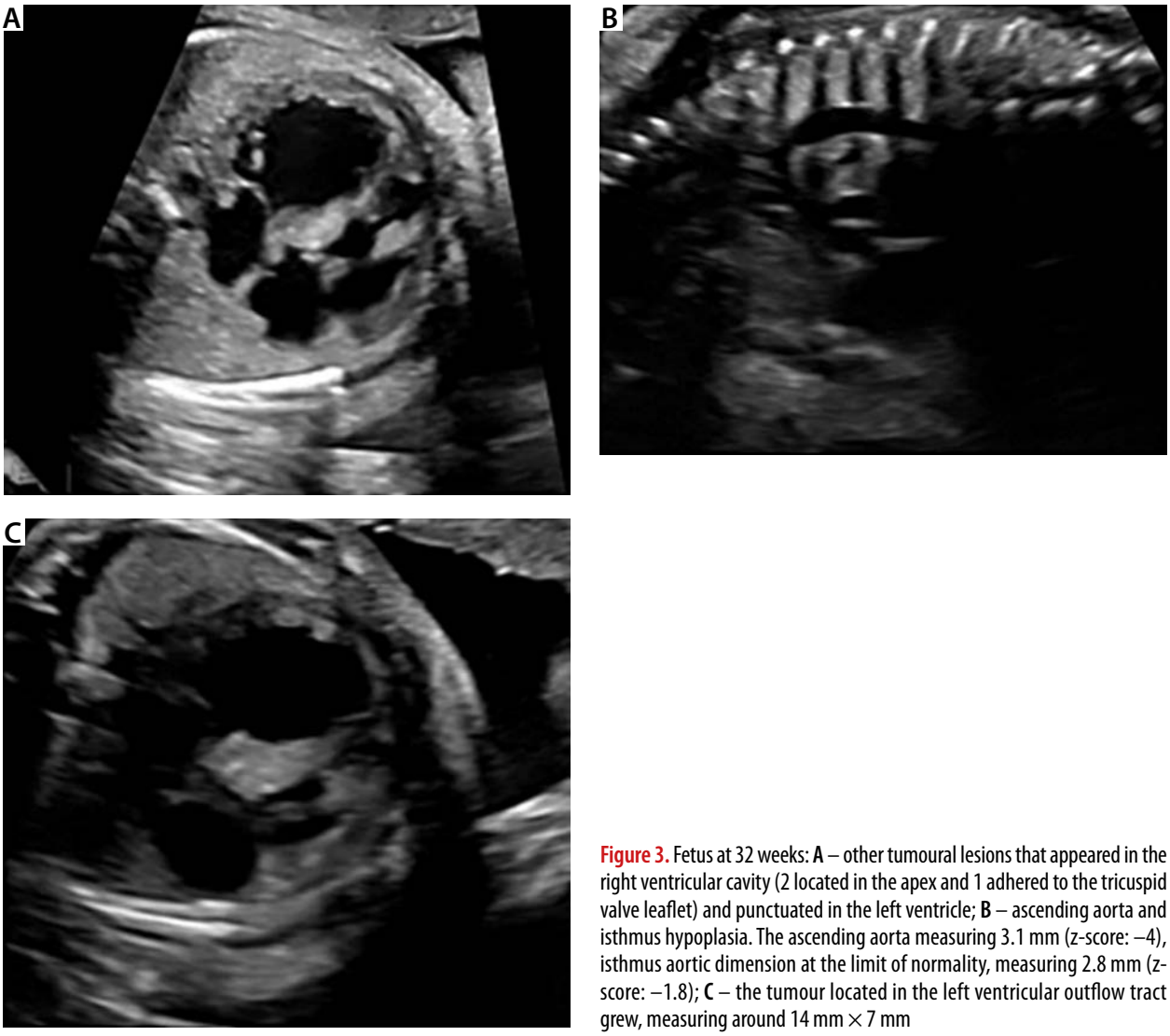

Figure 3. Fetus at 32 weeks: **A** – other tumoural lesions that appeared in the right ventricular cavity (2 located in the apex and 1 adhered to the tricuspid valve leaflet) and punctuated in the left ventricle; **B** – ascending aorta and isthmus hypoplasia. The ascending aorta measuring 3.1 mm (z-score: –4), isthmus aortic dimension at the limit of normality, measuring 2.8 mm (z-score: –1.8); **C** – the tumour located in the left ventricular outflow tract grew, measuring around 14 mm × 7 mm

the child presented infection and haemodynamic instability leading to death with 40 days of life.

Discussion

With improved ultrasonography technology and greater experience, fetal cardiac rhabdomyomas are now more frequently diagnosed prenatally. The diagnosis is often incidental [1]. In ultrasound, the rhabdomyoma are seen as non-vascular homogenous hyperechogenic masses originating from the myocardium, mostly multiple masses in the ventricles [1, 6]. They may be located in all myocardial areas but are usually detected in the septal, atrial, or ventricular myocardium [1, 6]. Differ-

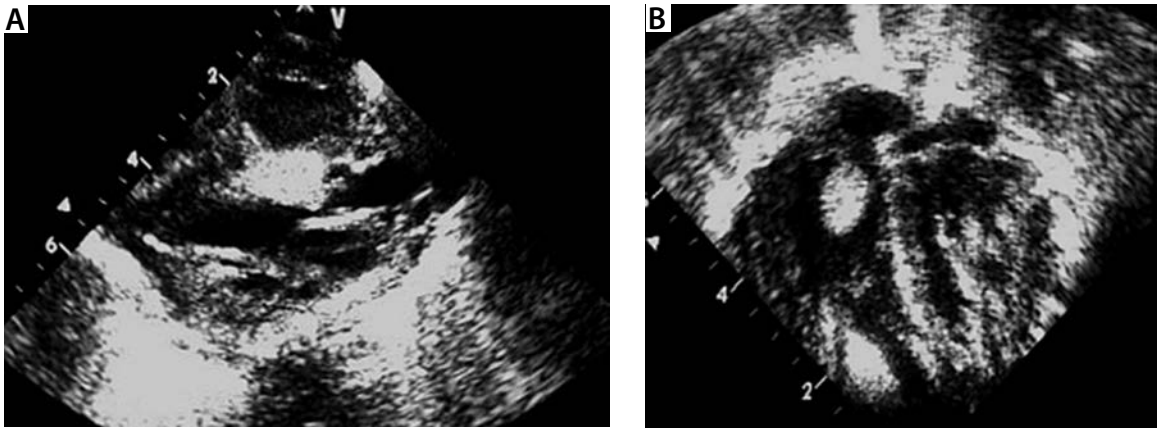

Figure 4. A – the tumour located in the left ventricular outflow tract measuring 14 mm × 9 mm and maximum aortic flow of 3.9 cm/s. B – other tumoral lesions that appeared in the right ventricular cavity (2 located in the apex and 1 adhered to the tricuspid valve leaflet) and significant right ventricle dilation

Table 1. Literature review of intrauterine complications of cardiac rhabdomyoma

Authors	*n*	Intrauterine complications				
		Hydrops	Dysrhythmia	Blood flow obstruction	Cardiac dysfunction	Death
Chao et al. 2008 [3]	11	2	1	NR	NR	1
Geipel et al. 2001 [1]	10	5	3	NR	5	1
Chen et al. 2019 [7]	53	NR	1	4	NR	0*
Bader et al. 2003 [4]	20	0	5	0	0	1
Yinon et al. 2010 [6]	40	3	2	9	1	0
Żalińska et al. 2017 [9]	13	NR	1	NR	NR	0

*NR – not reported; *in this article, termination of pregnancy was reported in 83% of cases.*

ential diagnoses are teratoma, fibroma, and haemangioma, but the presence of multiple tumours involving the ventricular myocardium is indicative of rhabdomyomas [1]. Complications can include arrhythmias, and progressive compression of cardiac, vascular, and pulmonary structures by a rapidly growing mass or effusion (Table 1) [1, 3, 4, 6, 7, 9].

In the absence of family history, cardiac rhabdomyoma may be the earliest sign of tuberous sclerosis in utero and can precede the detection of brain or kidney lesions [3]. After birth, rhabdomyomas usually regress in size, and smaller ones may even completely resolve [1, 6]. Therefore, conservative management is advocated if the cardiac function is stable [1, 3].

Surgical treatment is advisable only in selected cases, as was necessary in our case. The tumours can be difficult to remove completely, because they are often located deeply in the myocardium and have major vessel supplies from the coronary arteries [8]. This was the reason for not performing tumour resection, we believed that there would be coronary arteries nourishing this rhabdomyoma because this fetus had such a significant dilation and ventricular dysfunction.

In a retrospective cohort of 33 fetuses with rhabdomyomas, 12% died at birth, and although nine (30%) fetuses had ventricular obstruction, only one (3%) had prenatal cardiac dysfunction, but none are predictors of fetal or neonatal death [3, 6]. In our case, although the tumour was not very large, it caused ventricular obstruction leading to dysfunction, which is uncommon. Then, we postulate that such dysfunction could be associated with the flow theft from the adjacent coronary artery. So, knowing that a premature delivery is a strong predictor of neonatal death but that maintaining the pregnancy could lead to fetal death, we opted for premature delivery.

From the surgical point of view, resection of the outflow tract tumour was also avoided due to the risk of coronary supply, so it was decided not to remove the tumour as it regressed after birth. However, the pulmonary bandage to direct blood flow through the ductus arteriosus was effective, but it was insufficient because no shunt was ensured. Thus, there was closure of the ductus arteriosus and cardiovascular decompensation. The report of this case teaches us an important lesson by exemplifying an unsuccessful experience of rhabdomyoma surgery.

In summary, fetal diagnosis of rhabdomyoma is important for prenatal guidance, birth planning, and postnatal programming. Most often, these tumours are the first manifestations of tuberous sclerosis, and a thorough and continuing investigation of this child should be performed.

Conflict of interest

The authors declare no conflict of interest.

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