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Prenatal diagnosis of fetal cardiac rhabdomyoma associated with tuberous sclerosis: A case report

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ABSTRACT

Cardiac tumors are rarely diagnosed in utero. Rhabdomyomas are the most common fetal cardiac tumors. They are usually diagnosed during the first year of life after obstruction of a valve orifice or a cardiac chamber; but they can be detected by echocardiography as early as the second trimester. Rhabdomyomas are usually small. Fetal hydrops and pericardial effusion are rare. The most important indication of tuberous sclerosis in the prenatal period is cardiac rhabdomyoma. Early diagnosis of cardiac rhabdomyoma is thus important for early diagnosis of tuberous sclerosis. This case report concerns the prenatal diagnosis of both multiple fetal cardiac rhabdomyomas and tuberous sclerosis.

1. Introduction

Although fetal cardiac rhabdomyomas are rare, they are the most common fetal cardiac tumors. The incidence of fetal cardiac rhabdomyomas in the postnatal phase is about 1/40000. The etiology is unknown, but there are data indicating that maternal hormones are involved [1]. With improved ultrasonography technology and greater experience, fetal cardiac rhabdomyomas are now more frequently diagnosed prenatally. They are seen as non-vascular homogenous hyperechogenic masses originating from the myocardium. They may be located in all myocardial areas but are usually detected in the septum or ventricles. The diagnosis is often incidental. It is reported that cardiac tumors are detected at the earliest in the 15th gestational week [2].

Tuberous sclerosis is diagnosed in most cases of multiple fetal cardiac rhabdomyomas and in 50% of single cases. Tuberous sclerosis is an autosomal dominant systemic genetic disorder with variable penetrance that affects the central nervous system, skin, retina, kidneys and heart. In approximately 30% of cases the cause is genetic. In the other 70% of cases, de-novo mutations inactivate the TSC1 (9q34.3) and TSC2 (16p13.3) genes, which encode two proteins, tuberin and hamartin. Pericardial teratoma, fibroma, hemangioma, myxoma and echogenic cardiac focus should be considered in the differential diagnosis [3].

Multiple and large lesions are more likely than single or small lesions to grow in utero. Enlargement reduces after the 32nd week, although they can enlarge in the third trimester. Cardiac rhabdomyomas may continue to grow postpartum, due to maternal estrogen. The later decrease in the size of tumor may be due to reduced estrogen levels [3]. These tumors are at their largest in fetal life, then shrink with age and may even disappear completely. The prevalence is therefore higher in children than in adults [4].

Cardiac rhabdomyomas commonly regress without treatment, but surgical resection should be performed if cardiac function is adversely affected. The prognosis is good if there is no complication in utero and or in the first six months postnatally, but is poor where there is cardiac dysfunction. The neonatal mortality rate is 4–6% [3]. Here we discuss the importance of prenatal diagnosis of cardiac rhabdomyomas.

2. Case Report

A 33-year-old patient, gravida five, parity four, at her 36th gestational week was referred for sonographic morphological examination of a cardiac malformation diagnosed by routine sonographic study. The pregnancy was spontaneous and the father not consanguineous. The patient had had only one prenatal visit, in the first trimester. Her medical and obstetric histories were unremarkable.

Echocardiographic findings demonstrated an intracardiac bulky mass, 14 × 13 mm, on the apical left ventricular wall and a similar mass, 19 × 13 mm, on the interventricular septum, with the tricuspid valve septal leaflet bulging. Both masses presented a hyperechogenic, homogeneous aspect and were diagnosed as cardiac rhabdomyomas (Figs. 1 and 2). Fetal cardiac function was normal. Cardiac size was normal, as was tricuspid valve function. No associated cardiac anomaly was detected. Fetal cranial sonographic examination was normal. Prenatal genetic diagnosis was not applied due to advanced gestational age. A male neonate was delivered after induction of labor at the 37th gestational week, one week after the diagnosis, weighing 3300 g, the 5- and

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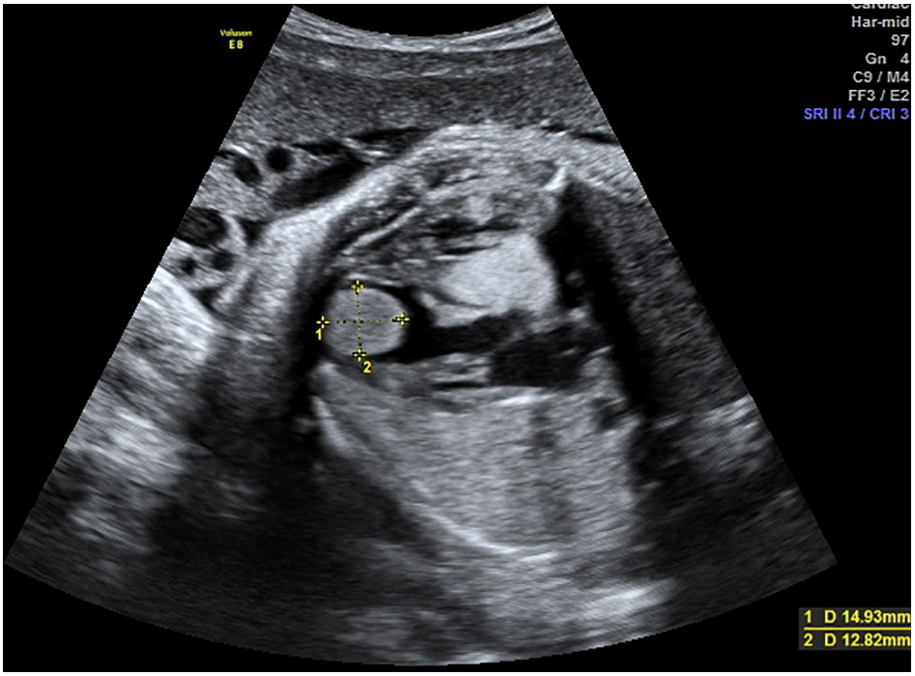

Fig. 1. Left ventricular apical rhabdomyoma.

10-min Apgar scores were 8 and 9. The neonate had unstable respiration and so was intubated and taken to an intensive care unit. Umbilical cord blood was taken for genetic testing for tuberous sclerosis.

Neonatal echocardiography confirmed the prenatal findings and additionally 5x4mm and 7x4mm rhabdomyomas were detected on the interventricular septum. Cardiac surgery was planned for the newborn because the rhabdomyoma affecting the tricuspid valve was causing obstruction. The result of the genetic analysis confirmed the diagnosis of tuberous sclerosis. A neurological examination was without pathological findings.

3. Discussion

Fetal cardiac rhabdomyomas are the most common cardiac tumors and are associated with tuberous sclerosis: they occur in 43–60% of cases [5], and tuberous sclerosis is diagnosed in most cases of multiple fetal cardiac rhabdomyomas [4]. The early manifestations of tuberous sclerosis can be detected on prenatal screening and most commonly involve the heart and brain. Rhabdomyomas are an early manifestation and may be the only finding before other clinical manifestations appear. For this reason, the diagnosis of tuberous sclerosis should be considered

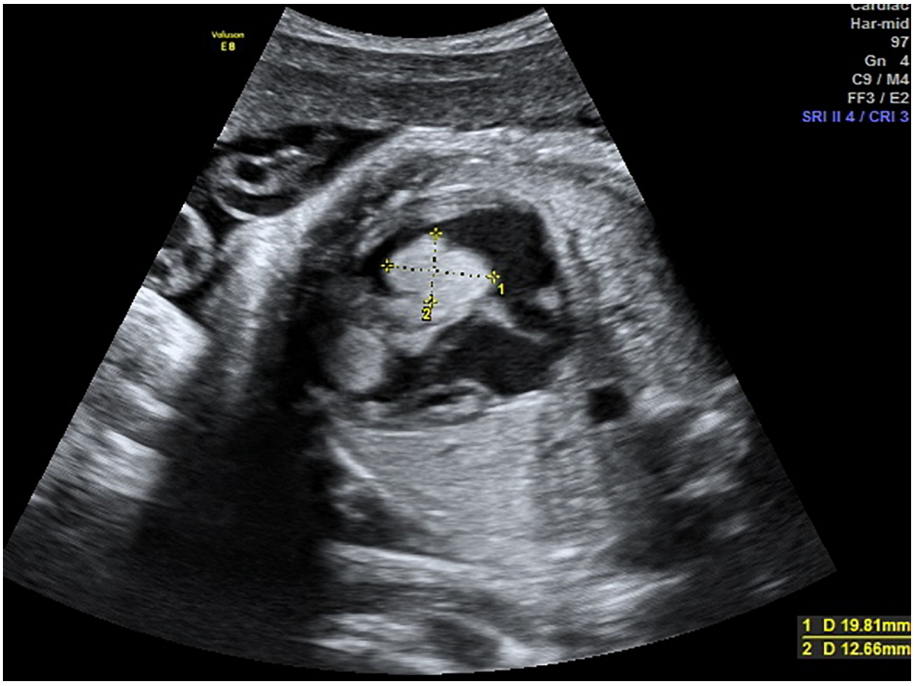

Fig. 2. Right ventricular rhabdomyoma on the interventricular septum and tricuspid valve septal leaflet.

in cases of cardiac rhabdomyomas [6]. Our patient had no cranial lesions but had multiple cardiac rhabdomyomas and tuberous sclerosis was diagnosed neonatally. This reveals the importance of genetic counseling for tuberous sclerosis in patients with fetal cardiac rhabdomyomas. In a meta-analysis, Chao et al. reported that most cardiac rhabdomyomas were detected after the 24th gestational week and only 13.7% of the cases were detected before the 24th gestational week [6]. In our case, the diagnosis was made in the 36th gestational week, as the patient did not scheduled checks in the second and third trimesters. Although rhabdomyomas may form in any part of the heart, they are usually detected in the ventricles and interventricular septal areas. They are often multiple masses [4]. In our case, rhabdomyomas were multiple and were located on the interventricular septum and ventricular walls.

The clinical manifestations of rhabdomyomas are variable. Heart failure, hydrops fetalis and stillbirth may occur in the prenatal period [3]. They may be asymptomatic during the postnatal phase or may cause mechanical obstruction, arrhythmia, heart failure and sudden death, depending on the number and location of the rhabdomyomas. Arrhythmias may be in the form of premature atrial or venous contractions, supraventricular tachycardia and sinus bradycardia [7]. Cardiac rhythm was normal in our case but mechanical obstruction due to rhabdomyoma on the tricuspid valve was detected during the neonatal period. Fetal cardiac rhabdomyomas can enlarge in the 2nd and 3rd trimester but they usually regress without treatment after birth. Nonetheless, postpartum echocardiographic monitoring is necessary. If the rhabdomyoma leads to heart failure in the newborn, mechanical obstruction or life-threatening arrhythmias, surgical resection should be performed [8]. In our case, surgical resection was planned due to rhabdomyomatic obstruction of the tricuspid valve and a fall in the newborn's blood oxygen level.

4. Conclusion

Early prenatal diagnosis of cardiac rhabdomyoma is important for perinatal follow-up and a multidisciplinary approach to treatment. Moreover, since cardiac rhabdomyomas may be the earliest manifestation of tuberous sclerosis, the infant should be evaluated accordingly.

Contributors

All authors contributed to the preparation of this case report and saw and approved the final manuscript.

Conflict of Interest

The authors declare that there are no conflicts of interest regarding the publication of this manuscript.

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Patient Consent

The patient in this study gave informed consent for her history, exam, images, and evaluation to be used in this manuscript.

Provenance and Peer Review

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