Prenatal diagnosis of cardiac rhabdomyoma associated with tuberous sclerosis: report of 3 cases

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Abstract
Cardiac rhabdomyoma is the most common cardiac tumor in fetal life, accounting for 60–86% of primary fetal cardiac tumors. It is primarily benign, originating form myocardial muscles and consisting of immature myocytes. About 50–60% of these tumors are associated with tuberous sclerosis. In this report, we present the clinical course and discuss the importance of prenatal diagnosis of cardiac tumors and their follow-up after birth.

INTRODUCTION
Cardiac rhabdomyomas are the most common cardiac tumors in childhood (Allan et al. 2000). They are benign tumors from the muscle of myocardium, but they can be life-threatening, because of heart chamber obstruction by their infiltrative growth (Freedom et al. 2000; Thomas-de-Montpréville et al. 2007). Most of the cases of cardiac rhabdomyomas are asymptomatic, but they can cause arrhythmias, murmurs and sudden cardiac death (Freedom et al. 2000). They are very often associated with tuberous sclerosis (Thomas-de-Montpréville et al. 2007). The diagnosis of rhabdomyoma is made by two-dimensional echocardiography and magnetic resonance imaging. Most patients with primary cardiac tumors are diagnosed by fetal echocardiography performed during the perinatal period. In postnatal life cardiac rhabdomyoma can be detected through the symptoms including arrhythmia, murmur, respiratory distress, and cyanosis. Most of the rhabdomyomas have tendency for spontaneous regression (Black et al. 1998; Piazza et al. 2004; Thomas-de-Montpréville et al. 2007).

CLINICAL REPORTS
We performed a retrospective review of the medical records of three children diagnosed with primary cardiac tumors, who have been followed at our centre during 5 years (Table 1). The diagnosis
Localization of rhabdomyoma was in the left ventricle, especially in the interventricular septum, in all three cases. None of these patients had any complications such as arrhythmia, thromboembolism or ventricular dysfunction during the follow-up.

**DISCUSSION**

Cardiac tumors are very rare in childhood. Incidence of pediatric cardiac tumors is about 0.17% (Etuwewe et al. 2009). Most of them are benign, only 10% are malignant (Uzun et al. 2007).

Rhabdomyoma is the most common cardiac tumor in children. The tumor is benign, arising from myocardial muscles, but can be life-threatening because of its infiltrative growth throughout the heart chambers (Allan et al. 2000; Thomas-de-Montpréville et al. 2007). Most of the cases are associated with autosomal dominant neurocutaneous syndrome called tuberous sclerosis (Takehiro et al. 2000). Cardiac rhabdomyoma is often asymptomatic, but it can be accompanied by symptoms such as murmurs, arrhythmias, respiratory distress and cyanosis. The affected conductive system of the heart and intracardiac blood flow obstruction are mainly responsible for the occurrence of these symptoms (Piazza et al. 2004; Thomas-de-Montpréville et al. 2007). Moreover, epilepsy and mental retardation in early childhood as symptoms of tuberous sclerosis, can
also be a warning for the presence of cardiac rhabdomyoma (Takehiro et al. 2000).

It has been reported in the literature, that more than half of the cardiac rhabdomyomas are diagnosed during prenatal period (Thomas-de-Montpréville et al. 2007). Rhabdomyomas in all our cases were diagnosed by prenatal ultrasonography. They were localized in the left ventricle without causing ventricular dysfunction.

In two cases a mutation of TSC1 or TSC2 genes was confirmed, which is the cause of tuberous sclerosis. In the third child, this mutation was not detected, but magnetic resonance imaging showed degeneration of brain tissue typical for tuberous sclerosis. None of our patients had positive family history for this neurocutaneous syndrome. Infants with tuberous sclerosis are at risk of epilepsy or mental retardation (Takehiro et al. 2000). In our study, two children had epilepsy and one of these children had mental retardation as well. Cardiac rhabdomyoma usually has a tendency for spontaneous regression (Black et al. 1998). In the follow-up echocardiographic studies, we observed partial spontaneous regression of rhabdomyoma in all three of our patients. The most dangerous feature of cardiac rhabdomyoma is the fact, that it can cause malignant arrhythmias leading to sudden cardiac death. Miyake et al. reported, that about 16% of primary cardiac rhabdomyomas cause significant arrhythmias (Miyake et al. 2011). In our three cases, none of the children had malignant arrhythmia, but in one child, Wolff-Parkinson-White syndrome was diagnosed by electrocardiography.

The diagnosis of rhabdomyoma is made by two-dimensional echocardiography and magnetic resonance imaging. Chest radiography is helpful to show cardiomegaly and electrocardiography is important for detection of arrhythmias (Allan et al. 2000).

Echocardiography should be performed systematically in all children with rhabdomyoma and tuberous sclerosis and is also recommended for members of their families. Histological proof of the diagnosis of rhabdomyoma is not necessary in the presence of characteristic echocardiographic findings

In this study, none of our patients experienced any complications such as arrhythmia, thromboembolism or ventricular dysfunction during follow-up.

CONCLUSION

Cardiac rhabdomyoma is a rare tumor, which is usually possible to diagnose by ultrasonography during prenatal period. In our study, all three rhabdomyomas were detected in prenatal life and they were all associated with tuberous sclerosis. From the diagnostic standpoint, it seems reasonable, that after confirmed diagnosis of cardiac rhabdomyoma it is necessary to do magnetic resonance imaging of brain to exclude tuberous sclerosis and conversely, after confirmed diagnosis of tuberous sclerosis it is necessary to do echocardiography to exclude cardiac rhabdomyoma.

REFERENCES