INTRODUCTION

Accounting 60% of all primary cardiac tumors, rhabdomyomas are the most common primary cardiac tumors during fetal life and childhood.\textsuperscript{1-3} Rhabdomyoma is a benign mesenchymal tumor of skeletal muscle, separated into two major categories based on site as cardiac and extracardic; and further separated by histology into fetal (myxoid and cellular), juvenile and adult types.\textsuperscript{1} Fetal-type rhabdomyoma is a benign tumor described almost always in extracardiac localisations.\textsuperscript{4,5}

Herein, we describe a hemodynamically unstable female neonate with a single large intracardiac mass unresponsive to medical treatment, who underwent surgery and diagnosed as fetal-type cardiac rhabdomyoma on postmortem examination of the mass.

CASE REPORT

A female neonate was born to a 19-year mother with a birth weight of 2800 grams by cesarean section at term due to fetal distress. She was referred to the Pediatric Cardiology Department at the first day of her life because of prenatal diagnosis of a cardiac mass. On physical examination, her heart rate was 170 bpm, blood pressure was 60/40 mmHg, respiratory rate was 50 per minute, and oxygen saturation was 85% to 90%. An II/VI grade systolic ejection murmur best heard on the left-sided second intercostal space and hepato-splenomegaly were also detected in physical examination. Family history for tuberous sclerosis complex (TSC) was negative.

Chest X-ray (Figure 1) demonstrated cardiomegaly with a cardiothoracic index of 0.7. There was nothing abnormal displayed in electrocardiography. Preliminary echocardiogram revealed a hyper-echoic left-ventricular mass with a 3.5 - 4 cm diameter in the ventricular and apical septum, resulting in the regression of the left-ventricular function and mild left ventricular inflow and outflow obstruction (Figure 2). In addition, a patent foramen ovale with left-to-right shunting, moderate tricuspid and mitral valve regurgitations were also noted. Thoracic computerised tomography (CT) demonstrated a single, heterogeneous, solid, hypo-dense, mass measuring 3.5 x 3.5 x 4.5 cm and occupying the whole left ventricle.

Neurologic examination was normal and Wood's light examination of the skin, cranial and abdominal ultrasonography did not reveal any lesions in terms of TSC. The tumor was thought to be a rhabdomyoma without TSC stigmas. Furosemide therapy was started for the treatment of heart failure. Cardiac surgeons evaluated the patient, but they deemed the mass as ineligible for surgical resection because of extensive intramural myocardial involvement. Instead, because of the presumptive diagnosis of cardiac rhabdomyoma, everolimus treatment (Certican 0.25 mg tablet) with the dosage of 0.25 mg every 6 hours, 2 days per week, was started. Complete blood cell count, hepatic and renal function tests, lipid profile, and lymphocyte subsets were monitored during treatment. We also monitored the serum level of the drug between 5 - 15 ng/ml.

After 20 days trial of everolimus treatment, because of the lack of regression of mass and heart failure, surgical resection was offered. After median sternotomy and cardiopulmonary bypass, the large tumor to have extensive myocardial involvement and invasion to the coronary arteries. Unfortunately, the patient expired because of ventricular fibrillation developed during the operation at 28th day of her life.
Histopathological examination of well-circumscribed lesion revealed fibro-myxoid stroma and isolated bland immature spindle cells with eosinophilic cytoplasm compatible with fetal rhabdomyoblasts; and immuno histochemical staining was positive for desmin and SMA. There was no evidence of spider cells typical of rhabdomyoma (Figure 3). So, the final pathologic diagnosis was fetal-type rhabdomyoma without any evidence of TSC.

**DISCUSSION**

Herein, a fetal-type cardiac rhabdomyoma is reported which was unresponsive to the medical treatment, but the infant expired due to ventricular fibrillation during surgery. To the best of authors’ knowledge, this type of cardiac rhabdomyomas are extremely rare, with only 3 cases reported before; and their behavior is also unknown.6,7

Rhabdomyomas are the most common cardiac tumors in the pediatric age group that are often associated with TSC, and their presentation generally depends on the size and location of the mass.8 On the other hand, fetal-type rhabdomyomas are benign tumors described almost always in extracardiac localisations, especially on head and neck region of infants. Histologically, the typical cells of cardiac rhabdomyomas are spider cells that closely resemble embryonic cardiac muscle cells. However, the histologic marker is the elongated fetal myoblasts (spindle cell) presenting with various degrees of differentiation in fetal-type rhabdomyomas.4,5 Gao et al. reported a huge right ventricular fetal-type rhabdomyoma with 5 cm diameter in a female infant of 3 months and 10 days old.6 Later, Viscardi et al.7 reported two siblings with tricuspid valve fetal-type rhabdomyoma that both were symptomatic in neonatal period. In first sibling, presented with severe tricuspid stenosis and pulmonary hypertension, elective surgical excision of the tumor was performed at 11 months of age without recurrence of the mass at first follow-up after operation. The second sibling had severe tricuspid stenosis and pulmonary hypertension and required emergent surgical intervention at the 21st day of his life and died because of sepsis and recurrent respiratory failure. Recurrent pulmonary embolism was also detected in this patient. In all those 3 cases, the lesion was located on right heart; but in this case, it was a left ventricular mass.

In general, the natural history of the cardiac rhabdomyomas is to regress and most patients can be managed conservatively including frequent monitoring with echocardiography and electrocardiography, and surgical interventions are generally preserved for severely symptomatic patients.3 Besides, everolimus is an inhibitor of mTOR that was reported to be effective against some cardiac rhabdomyomas associated with TSC since increased activation of mammalian target of rapamycin (mTOR) was reported in TSC.9 Although there was no any sign of TSC in our patient, since she was severely symptomatic and initially thought to be ineligible for surgery, so we started everolimus treatment together with the symptomatic therapy for heart failure. However, the patient was not responsive to the medical treatment and she had to undergo a surgical intervention.

Behaviour of the fetal-type rhabdomyomas, when localised in the heart, is unknown since they are extremely rare. So, there is no general consensus about the treatment strategy of this condition due to unpredictable pathologic and clinical evolution of the tumor. In case of inconsistent features for cardiac rhabdomyomas, early surgical excision should be recommended for the exact diagnosis and prevention of life-threatening complications such as arrhythmias, ventricular outflow tract obstruction, and thromboembolic events in these patients.

**REFERENCES**

3. Jozwiak S, Kotulska K, Kasprzyk-Obara J. Clinical and


