CASE REPORT

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A Giant Heart Tumor in Neonate with Clinical Signs of Pierre - Robin Syndrome

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ABSTRACT

Introduction: Pierre Robin syndrome is a congenital condition of facial abnormalities in humans. The three main features are: cleft palate, retrognathia and glossoptosis. Rarely heart tumors are associated with syndromes, mostly are isolated. **Case report:** In this presentation we describe a 3-weeks-old girl with Pierre-Robin syndrome and giant left ventricle tumor, diagnosed initially by transthoracic echocardiography. The purpose of this report is to review the literature on the fetuses and neonates with cardiac tumors in an attempt to determine the various ways which cardiac tumors differ clinically and morphologically in this age group.

Keywords: cardiac tumors, Pierre-Robin syndrome, echocardiography, prenatal diagnosis

1. INTRODUCTION

Cardiac tumors in infants and children are extremely rare, mostly are benign and differ in types when compared with those in adults. Their clinical manifestations vary widely from asymptomatic presentations to life-threatening cardiac events. Improvements in diagnostic techniques, such as those offered by echocardiography, have made early detection of cardiac masses possible, with or without the presence of clinical symptoms. Most of the benign tumors are rhabdomyoma, followed by teratoma, fibroma, oncocytic cardiomyopathy, vascular tumors, and myxoma (1).

2. CASE REPORT

A 3-weeks-old girl was admitted in our intensive care unit with clinical signs of respiratory distress. The clinical signs of Pierre-Robin syndrome were also presented (micrgonathia and retrognathia with respiratory difficulties, especially when supine). She was the second child from a second normal pregnancy. During the pregnancy a few routine ultrasonography examination by obstetricians was done and normal findings were referred. It was a spontaneous delivery at term in Regional Hospital. The baby girl weighed 2820 grams, with Apgar score 7 at 1 minute and 8 at 5 minutes. On the third day after the delivery, baby was discharged at home without any objection. Two days before admission in our clinic, a viral respiratory infection was noted and, due to rapid deterioration of condition, at our institution was referred. X-ray chest showed massive cardiomegaly. ECG showed normal sinus rhythm with ST-segment depression on the lateral derivations. On the second day of hospitalisation routine transthoracic cross-sectional echocardiography was performed and giant tumor in the posterolateral wall of left ventricle was noted, measuring maximal diameter 24.9 x 27.9 mm (Figure 1). Tumor was located in the ventricular myocardium, near the insertion of posteromedial leaflet of the mitral valve and uniform echogenicity was noted. CT-scan revealed tumor mass located on the lateral wall of the left ventricle, filling the most surface of ventricle (Figure 2). Heart rhythm was normal and there was no signs of hemodynamic disturbances and heart failure.

3. DISCUSSION

Primary tumors of the heart are uncommon in the fetus and neonate with an autopsy frequency of 0.001-0.030 %. Cardiac tumors are known to originate in any part of the heart, including the endocardium, myocardium and pericardium. Most tumors are benign and can regress spontaneously but malignant and metastatic tumors are described too. Malignancy, associated malformations and aneuploidy are infrequently observed. Cardiac tumors, though rare, are now being diagnosed with increasing

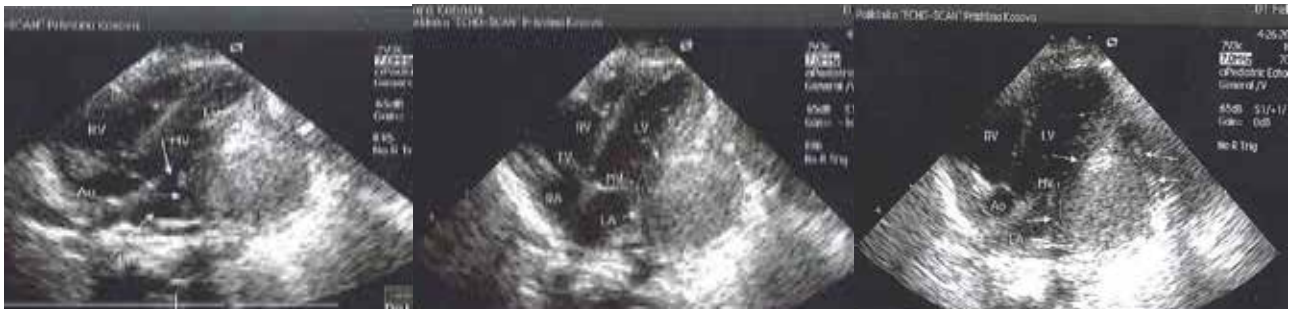

Figure 1. Tumour mass localised on the lateral wall of left ventricle, near posterolateral leaflet of mitral valve

frequency and great accuracy by prenatal ultrasound (2). Of the cardiac tumors found in the fetus and neonates, about three-fifths are rhabdomyomas, with less than one-fifths being teratomas and one-eighth are fibromas (3). The incidence of fetal and neonatal tumors has been increased due to generalization of prenatal evaluation and improvement of imaging techniques. The wide speed use of echocardiography and other non-invasive diagnostic methods in recent years have resulted in increase in detection of children and fetuses with cardiac tumors. The combination of sonographic features and their location allows reliable prediction of histological type in the majority of tumors diagnosed in fetal and neonatal period. Rhabdomyomas are usually multiple, while fibromas are always single, but both are usually located in ventricular myocardium, and have uniform echogenicity. Teratomas are usually extra cardiac, attached at the base of arterial trunks, with heterogeneous echogenicity.

Depending on the histological type, location and number of nodules, cardiac tumors have a range of presentations, going from silence to development of severe clinical manifestations, such as arrhythmias and blood flow restrictions due to obstruction of the ventricular cavities or the atrioventricular and semilunar valves, thereby causing valve regurgitation due to changes to the mobility of these valves, depending on the degree of insufficiency, presence of these tumors may lead to a hemodynamic disorder. Alteration of the ventricular ejection fraction, hydrops or congestive heart failure may occur and, if such situations develop, the prognosis will consequently be worse. Murmurs, arrhythmias, cyanosis, respiratory distress, and cardiac failure are the main presenting signs of cardiac tumors in the perinatal period. Disturbances in hemodynamic function are correlated with the size and location of the tumor. Cardiac vascular tumors have the best outcome, whereas malignant tumors have the worst (1, 2, 4).

In our presentation, tumor is single, deeply inserted in the wall of left ventricle, with homogenous echogenicity and, despite of the large dimension, tumor is conveying heart contraction. No hemodynamic, contractile and rhythm disturbances were registered.

The early detection of neonatal tumors and understanding of its imaging features are very important for maternal and neonatal care. Ultrasonography is usually used for the detection and differential diagnosis of fetal tumors, and CT-scan and magnetic resonance imaging is increasingly being used as a complementary study. Diagnosing cardiac tumors by means of two-dimensional echocardiograms is a fundamental step towards follow-up and prognostic evaluation. Many fetal tumors have different clinical and imaging features compared with pediatric tumors (5). Differential diagnosis is important, as this affects prognosis and subsequent management (2).

Despite that in children and fetuses cardiac tumors are usually benign they may induce life threatening symptoms with nonspecific symptoms and often mimic other heart disease. Cardiac symptoms predominate, further-

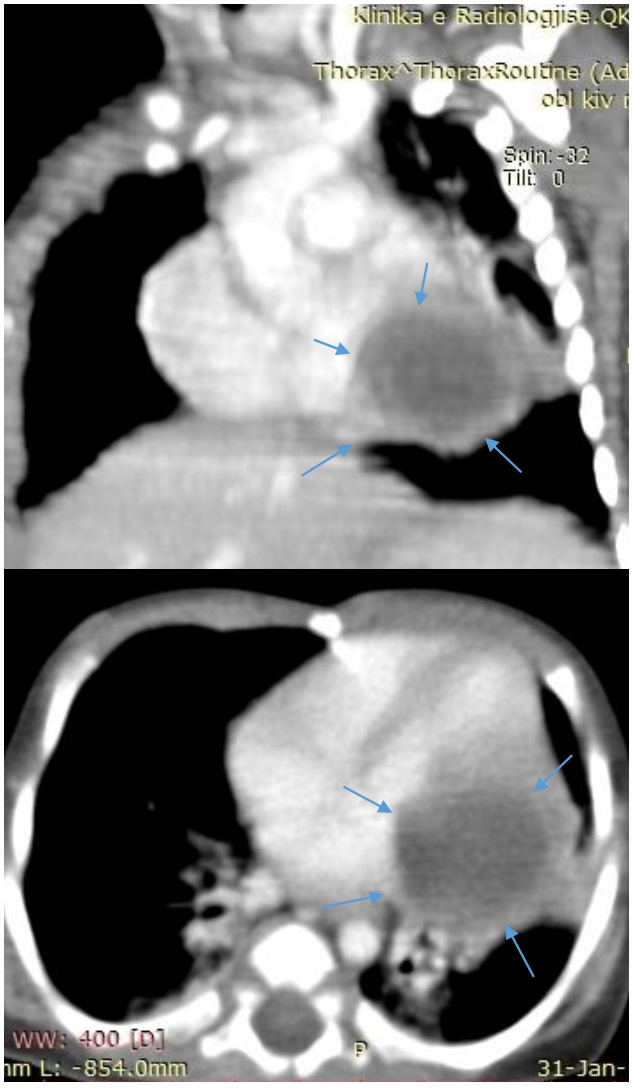

Figure. 2. CT-scan revealed tumour mass located on the lateral wall of the left ventricle

more they can also manifest systemic and embolic symptoms too. Although several fetal tumors may mimic other common anomalies, some specific imaging features may carry early accurate diagnosis of fetal tumors, which may alter the prenatal management of a pregnancy and the mode of delivery, and facilitate immediate postnatal treatment (5).

Pierre Robin sequence may be caused by genetic anomalies at chromosomes. The syndrome is generally diagnosed clinically shortly after birth. A genetic cause to PRS was recently identified. However, association with gene loci 2q24.1-33.3, 4q32-qter, 11q21-23.1, and 17q21-24.3 has been found (6). Recent studies have indicated that genetic dysregulation of SOX9 gene prevents the SOX9 protein from properly controlling the development of facial structures, which leads to isolated PRS. Similarly, KCNJ2 gene also has a role to play. Overlap with certain other genetic syndromes like Patau syndrome has also been found (7).

The infant usually has respiratory difficulties, (airway obstruction caused by backwards displacement of the tongue base) especially when supine. The cleft palate is often U-shaped and wider than in cleft palate which is not associated with this syndrome (8). In our case we registered expressed micrognatia, retrognathia and glossoptosis with clinical signs of airway obstruction in supine.

Pierre Robin syndrome may occur in isolation, but it is often part of an underlying disorder or syndrome. The most common is Stickler Syndrome. Other disorders causing PRS, according to Dr. Robert J. Sphrintzen Ph.D. of the Centre for Craniofacial Disorders Montefiore Medical Centre, are Velocardiofacial syndrome, Fetal Alcohol Syndrome and Treacher Collins Syndrome (9). In our case we noted clinical and radiograph signs of Stickler syndrome but, in the condition of limited genetic resources, more genetic investigation were not made.

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