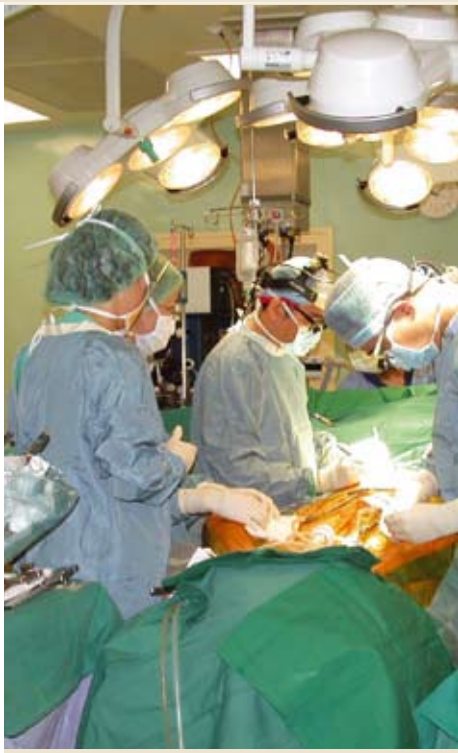
A One-year old infant with multiple cardiac masses and congenital heart disease (A case report)



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Abstract:

We present a one-year old male infant with heart murmurs discovered at birth. Transthoracic echocardiography revealed a perimembranous ventricular septal defect (VSD) as well as multiple cardiac masses. Pediatric cardiologists recommended closure of the VSD and biopsy of the uncertain cardiac masses. The VSD was repaired, and one of the masses was excised and sent for histopathological examination.

Here, we discuss a case of multiple rhabdomyomas in an infant whose associated finding was congenital heart disease, rather than tuberous sclerosis. He was discharged in good clinical condition and his parents were given instructions to have routine follow-up visits for the evaluation of the possible regression of the remaining masses.

Key words: Congenital heart disease- Cardiac masses - Rhabdomyoma

Introduction

Rhabdomyomas are hamartomatous lesions of cardiac myocytes (1), most common in infancy and childhood (2). A well-known association exists between rhabdomyomas and tuberous sclerosis; however, sporadic cases of rhabdomyomas which are solitary and endocardial-based may be seen as well (1,3). The latter is linked with congenital heart disease (2,3). Grossly, the lesions are well-demarcated and yellow-tan; they may exist singly or as multiple nodules or even numerous minute lesions, varying in size from 1 mm to 10 cm (1- 4).

Case Report

We present a one-year-old male infant who was first found to have cardiac murmurs soon after birth. In light of the abnormal heart examination, a transthoracic echocardiographic examination was performed shortly afterwards, which revealed a perimembranous ventricular septal defect (VSD) as well as multiple cardiac masses (Figure-1).

His follow-up by serial echocardiography studies showed two masses in the right atrium and three others in the right ventricle. Some degrees of mitral and tricuspid regurgitation were also noted.

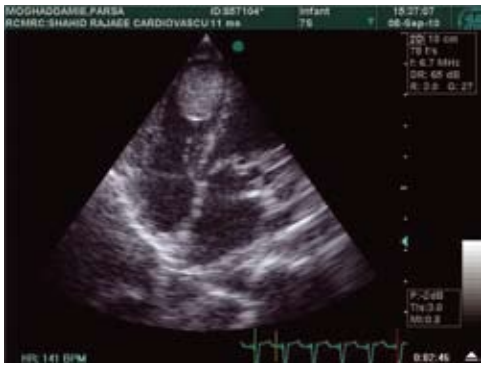

Figure-1: A well-defined mass is shown protruding into the right atrium.

Among other echocardiography findings, subsystemic pulmonary hypertension, as well as a right-sided aortic arch, was noteworthy.

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The only positive physical finding was the enlargement of the liver and spleen; other studies were, however, unremarkable.

The recommendation offered by the pediatric cardiologists was VSD closure and biopsy of the cardiac masses to determine their nature.

The patient underwent a surgical operation, during which the VSD was repaired and one of the more accessible masses in the right atrium was excised and sent to the pathology laboratory. The postoperative course was fortunately uneventful and he left the hospital, advised only to have outpatient visits for a scrutiny of possible size regression in the masses.

The surgical specimen was fleshy and brown in appearance, measuring 1.5 cm in the greatest diameter. The cut surface was solid and brown in color.

Microscopically, a well-circumscribed lesion was seen, with sheets of large cells that had clear cytoplasm (Spider cells). There was no evidence of malignancy in this specimen, and a diagnosis of cardiac rhabdomyoma was established.

The clear appearance of the cytoplasm is in consequence of the cells being embedded with glycogen, which is dissolved during the routine process of histopathology staining.

The reason why the characteristic cell is called "Spider Cell" is in the finding that only a few cytoplasmic projections are extended from the nucleus to the periphery, thus mimicking the shape of spider legs (Figure-2).

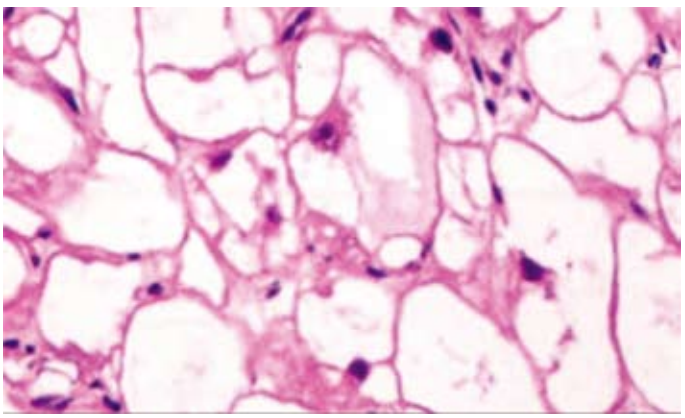

Figure-2:Plump and large clear cells are seen with central nuclei "Spider Cells".
(H& E, X 400)

Discussion:

Rhabdomyomas are considered hamartomatous lesions of cardiac myocytes (1-4) and
are the most common cardiac mass lesions seen in infancy and childhood (2,4).

Rhabdomyomas have a well-known association with tuberous sclerosis and their myriad manifestations. The lesion may, however, regress with time, if the patient survives the first month of life. Sporadic cases of rhabdomyomas, on the other hand, are solitary and endocardial-based. Nevertheless, patients in both of these groups may show certain degrees of morphological overlap (1). These masses also have an association with congenital heart disease (2,3).

Macroscopically, the lesions are well-demarcated and yellow-tan (1,3); they may exist singly or as multiple nodules or even numerous minute lesions, varying in size from 1 mm to 10 cm (2).

The hallmark of histopathological diagnosis is the presence of glycogen-rich Spider Cells. The differential diagnosis may include vacuolated myocardial cells due to glycogen storage disease, in which a diffuse distribution is found in the myocardium (1,3).

Large and single masses are amenable to surgical excision with good long-term results (2).

We herein discussed a case of multiple rhabdomyomas in an infant whose associated finding was congenital heart disease, rather than tuberous sclerosis. He was discharged in good clinical condition, while his parents were advised to have routine follow-up visits for an evaluation of possible size regression in the masses (4).

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