Left ventricular giant rhabdomyoma in an infant with no tuberous sclerosis: accidental finding and complex management

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SUMMARY: Bassareo PP, Fanos V, Tavera MC, Biddau R, Montis S, Boscarelli D, Mercuro G, Tumbarello R. Left ventricular giant rhabdomyoma in an infant with no tuberous sclerosis: accidental finding and complex management. Turk J Pediatr 2010; 52: 420-422.

The accidental finding of a giant left ventricular rhabdomyoma in a female infant with no tuberous sclerosis is described herein. This is the first report of a huge cardiac rhabdomyoma occluding the left ventricular cavity, which was not associated with tuberous sclerosis. The clinical management of the baby and the difficult therapeutical choices involved both pediatricians and pediatric cardiologists.

Key words: heart, tumor, tuberous sclerosis, imaging, drugs.

Primary cardiac tumors are rare, with a prevalence of 0.01% among congenital anomalies at autopsy[1]. The most frequent in clinical practice are rhabdomyomas, benign masses associated with tuberous sclerosis (TS) in 86% of the patients, while about 50% of those affected by TS have rhabdomyomas in their hearts[2,3]. On the contrary, malignant cardiac tumors are exceptionally rare[4]. Rhabdomyomas are uncommon in both adolescents and adults, because of their frequent trend to spontaneous regression[5,6]. They are usually small and multiple ventricular masses. Only in a very few cases are they so large as to become symptomatic (arrhythmias, ventricular inflow or outflow obstruction)[6,7].

Herein, we describe the case report of a giant single left ventricular rhabdomyoma, accidentally diagnosed in a female infant without TS. Her complex management involved both pediatricians and pediatric cardiologists.

Case Report

A four-month-old female infant was referred to our attention from the pediatric division for cardiac murmur, where she had been previously admitted for bronchiolitis. No congenital heart disease was reported in her family. She was born at term after a cesarean delivery, weighing 3.84 kg Apgar scores were 9 and 10 at 1 and 5 minutes, respectively. Echocardiographic (two- and three-dimensional) examination showed a huge mass measuring 5.2 x 3.3 cm occluding about 80% of the left ventricular cavity (Figs. 1, 2). A subsequent cardiac magnetic resonance imaging (MRI) confirmed the presence of a left ventricular solid mass coming from the inferior wall and obstructing the main part of the internal cavity. Taking into account both the early age of the patient and the MRI characteristics of the tumor (a homogeneously hyperdense mass in T2 and hypodense in T1, with no clear demarcation between its borders and the inferior ventricular wall), a diagnosis of rhabdomyoma was posed. On the basis of the ECHO draft, since this benign tumor is frequently related with TS, other examinations were performed (brain MRI, EEG, cutaneous examination by dermatologists, abdominal ECHO), but no signs of TS were reported. The only brain MRI showed an Arnold Chiari type I malformation, which is not necessarily associated with TS. Finally,

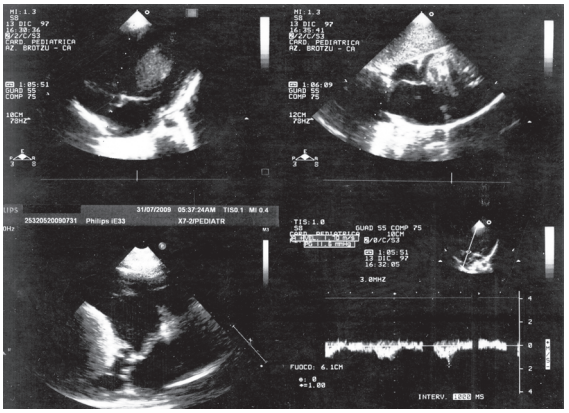

Figure 1. Two-dimensional ECHO shows a giant mass inside the left ventricle (apical four chamber view, PANEL A; subcostal view, PANEL B), which involves the papillary muscles (PANEL C), causing a mild ventricular inflow obstruction (mean gradient: 4-5 mmHg). The Doppler ventricular outflow pattern is shown as well (PANEL D), resulting from the presence of a mildly obstructive mass.

at genetic screening, no mutations involving *TSC1* and *TSC2* genes were detected, so that the concomitance of TS was excluded.

After a cardiac surgical consultation, the operation was deferred considering the frequency of spontaneous reduction/regression of these masses and the high death risk during surgery. The cardiac transplant was delayed as well. In the interim, because of the high frequency of asthma crisis by bronchiolitis, a therapy with beta-2 adrenergic agonists and corticosteroids was introduced. This strategy, although very efficient in reducing the breathing difficulty, resulted in the development of many ventricular premature beats, especially triggered by coughing. This arrhythmia (Lown class 5, being even runs of 3 beats) was confirmed by 24-hour Holter ECG registration. As a result, beta-2 adrenergic agonist administration was interrupted and palivizumab (Synagis) was used to prevent respiratory syncytial virus infection. A complete remission of the respiratory illness was registered. Notwithstanding the reduction in the frequency of ventricular premature beats after beta-2 adrenergic agonist suspension, the extrasystolic arrhythmia persisted (Lown class 2). In view of the high risk of heart failure, an antiarrhythmic therapy with amiodarone was introduced. This drug was very efficient in solving ventricular arrhythmias, without producing an excessive reduction in the already compromised systolic function of the baby. A few months later, with both the iatrogenic development of subclinical hypothyroidism (low FT3, normal thyroid stimulating hormone [TSH]) and long QT (473 msec) at basal ECG, this therapy was also interrupted, with no extrasystole recurrence. Thyroid function and QT normalized as well, and the infant was discharged. She is now two years old, is in seemingly good clinical condition, and is under treatment with furosemide 1 mg/kg x 2/day. At frequent echocardiographic examinations, the cardiac mass is visible with stable dimensions.

Discussion

To the best of our knowledge, many left ventricular giant rhabdomyomas have been previously reported in literature, but never in a patient without a diagnosis of TS. The absence of this frequent concomitant condition was confirmed by clinical, instrumental and genetic examinations[8]. The origin of the tumor from the inferior ventricular wall and not from the interventricular septum is uncommon as well. In addition, this case report is very interesting considering the many problems in clinical management of the infant, from points of view of both pediatricians and pediatric cardiologists. As stated above, the echocardiographic diagnosis was accidental, since the symptoms of the baby, who was referred to our attention for cardiac murmur, were very mild. The availability of two-dimensional echocardiography for about 20 years has often allowed a trained physician a precise description of intracardiac tumors, providing a great support to cardiac surgeons

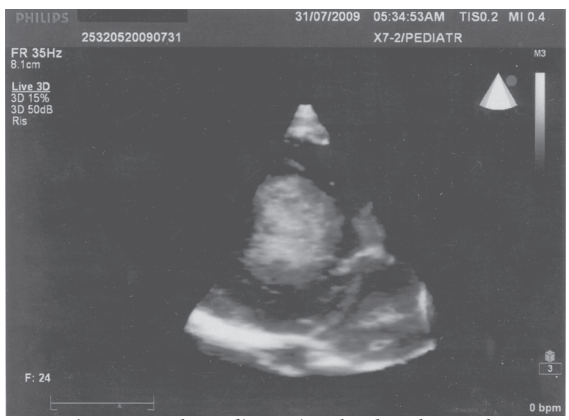

Figure 2. Three-dimensional echo shows the rhabdomyoma inside the left ventricle.

and avoiding the need for a potentially dangerous cardiac catheterization[9]. In addition, the recent option of three-dimensional echocardiographic imaging allows a perfect spatial description of heart neoplastic masses. In order to avoid X-ray exposure (computed tomography) at such a young age, the infant underwent a contrast-enhanced cardiac MRI. In this way, it is now possible to define the nature (benign or malignant) and the type of a number of intracardiac masses, such as rhabdomyoma, with no risk of endomyocardial biopsy[10]. The natural history of this benign tumor is generally favorable, with spontaneous resolution in most cases. Taking this point and the mild cardiac symptoms into account, the cardiac surgery was postponed[5,6,11]. Because of the frequent crisis of asthma with the recurrence of bronchiolitis, the infant was treated with beta-2 adrenergic agonist and corticosteroids in order to restore her health. Unfortunately beta-2 adrenergic agonists can induce cardiac arrhythmias, so ventricular premature beats and unsustained ventricular tachycardia are common, especially in subjects with a previous heart disease[12]. For this reason, this therapy was interrupted and palivizumab (Synagis) was used to prevent recurrent bronchiolitis due to respiratory syncytial virus infection[13]. Unlike what was initially reported, its safety profile is very good even in patients with congenital heart disease[14]. Because of the persistence of ventricular premature beats, even if less severe, a therapy with amiodarone was introduced. Among the antiarrhythmic drugs, it is the one that least reduces left ventricular systolic function, which had been previously compromised by the quite complete tumoral obliteration of the internal cavity. Although efficient, amiodarone in a short time caused the development of subclinical hypothyroidism and long QT, but with the interruption of this drug, they both disappeared[15]. After the suspension, no arrhythmia recurrence was registered during our frequent monitoring. In order to reduce the risk of heart failure, a diuretic therapy was introduced as well and the child is now in seemingly good clinical condition. The dimensions of the rhabdomyoma have shown no change over time. In conclusion, our unusual case report shows the difficult therapeutical choices faced by both pediatricians and pediatric cardiologists in the whole complex management of the case.

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