

Cardiac Rhabdomyomas and Congenital Hypothyroidism: A Coincidence or Hamartia

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ABSTRACT

Cardiac rhabdomyomas are the most common primary cardiac tumors in children. These tumors are generally asymptomatic, although they may be associated with neonatal tuberous sclerosis complex. Despite the fact that thyroid dysfunction rarely occurs in tuberous sclerosis, papillary adenomas (hamartomas) of the thyroid gland have been reported in a number of autopsies. Herein, we present the case of an infant with tuberous sclerosis, congenital hypothyroidism and multiple cardiac rhabdomyomas, leading to atrioventricular node dysfunction. Considering the rarity of this co-occurrence, we decided to report the present case. The co-occurrence of these two disorders in one patient may be a mere coincidence or related to hamartoma of the thyroid gland as a consequence of tuberous sclerosis gene products.

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Introduction

Cardiac rhabdomyomas are classified as hamartomas and are the most frequently found tumors in children. These tumors are associated with tuberous sclerosis in about 50-80% of patients. Moreover, 50-60% of patients with tuberous sclerosis present with rhabdomyomas (1-3). Cardiac rhabdomyomas are frequently multiple, involving ventricular free and septal walls.

Although rhabdomyomas are often clinically silent, they may lead to hemodynamic compromise and rhythm disturbances such as ectopic atrial tachycardia, atrioventricular node dysfunction and ventricular preexcitation. Herein, we report the case of an infant, diagnosed with tuberous sclerosis and cardiac rhabdomyomas. We discuss the noteworthy electrocardiographic (ECG) findings and the associated congenital hypothyroidism.

Case report

A 13-day-old female infant with a prior history of sluggish feeding and diagnosis of tuberous sclerosis was referred to our center for

cardiac evaluation. The infant had been delivered vaginally at 39 weeks of gestation, with no complications. Examination revealed a pulse rate of 70 bpm, respiratory rate of 32 bpm and blood pressure of 70/40 mmHg.

On physical examination, no abnormality was clinically detected. ECG showed right ventricular hypertrophy, right axis deviation and 2:1 second degree atrioventricular (AV) block. Chest radiography showed cardiomegaly, with normal pulmonary blood flow (PBF). Laboratory tests indicated normal results, except for hypothyroidism. No tumors were detected in the patient's brain or visceral organs.

Two-dimensional echocardiograms showed multiple homogeneous echogenic sessile mass like lesions in the interventricular septum (Figures 1 & 2), posterior wall of the left ventricle and papillary muscle of the mitral valve; moreover, a solitary lesion was detected in the right ventricle. No obstruction of intracavitary inflow or outflow tract was reported in the ventricles (Figure 3).

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Left ventricular systolic and diastolic functions were reported to be normal. There were no hypokinetic or akinetic regions, and the size and location of the proximal segments of the right and left main coronary arteries were normal on two-dimensional echocardiogram. The AV block was transient and spontaneously reverted to normal sinus rhythm within two hours. The infant did not develop hemodynamic instability during AV block. She otherwise remained healthy and her feeding improved after starting thyroxine supplementation therapy.

Discussion

Cardiac tumors are extremely rare in children (0.027-0.17%), and rhabdomyomas are regarded as the most common type of cardiac tumors (1). Tuberous sclerosis is a neurocutaneous syndrome, affecting the brain, heart, skin and other organs. In this regard, Józwiak and colleagues evaluated 154 patients with tuberous sclerosis, and cardiac rhabdomyomas were reported in 48% of cases (4). The tumors were most often detected in the right ventricle (35%), interventricular septum (33%), left ventricle (22%), left atrium (5%) and right atrium (5%);



Figure 1. Apical four-chamber view by 2D echocardiogram showing multiple homogenous echodense sessile mass lesions in the interventricular septum (red arrows)



Figure 2. Parasternal short-axis view by 2D echocardiogram of papillary muscles showing multiple homogenous echodense sessile mass lesions in the interventricular septum and papillary muscle of the mitral valve (white arrows)

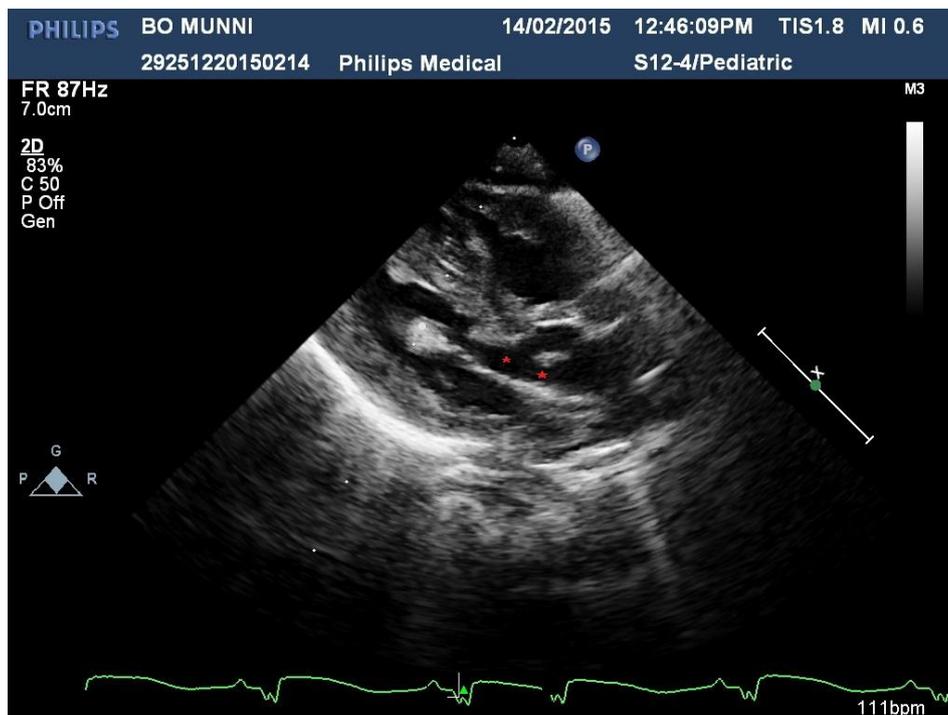


Figure 3. Parasternal long-axis view on 2D echocardiogram showing no obstruction in the inflow or outflow tract of the left ventricle (red stars)

overall, 61% of these tumors were clinically silent.

Rhabdomyomas can be intramural or pedunculated, encroaching on the intracavitary space. In fact, these tumors are hamartomas with no malignant potential. The clinical presentation of cardiac rhabdomyomas depends on the number, size and position of these tumors. They may be detected prenatally on a routine fetal ultrasound or may present as hydrops fetalis.

Postnatally, rhabdomyomas may be totally asymptomatic or present with low cardiac output due to intracardiac flow obstruction, rhythm disturbance or sudden infant death. In the present case, multiple rhabdomyomas were reported in the left ventricle and interventricular septum; we also detected obstructive manifestations.

In a study by Shiono and colleagues, 12 out of 21 patients presented with one or more ECG abnormalities (5). Ventricular tachycardia, atrioventricular block, and preexcitation syndrome are the most often reported rhythm abnormalities (6-8). Intramural rhabdomyomas are thought to interrupt the conduction pathways leading to ectopic electrical foci or giving rise to an accessory electrical circuit producing preexcitation (4).

The present case had transient AV node block, probably due to the presence of intramural tumors blocking the conduction pathway; the AV block spontaneously reverted to normal sinus rhythm. Rhabdomyomas have a propensity for spontaneous regression. According to previous findings, younger age at diagnosis results in the higher risk of spontaneous regression. In fact, complete regression is more common in the first four years of life (9); this has important implications for patient treatment. Generally, surgical intervention is indicated only in the event of hemodynamic compromise or intractable arrhythmias.

Congenital hypothyroidism is usually sporadic, although up to 2% of cases with thyroid dysgenesis are familial (10). Despite the relatively ubiquitous expression patterns of tuberous sclerosis complex gene products, the most common hamartomas associated with tuberous sclerosis complex are limited to relatively few organs, i.e., brain, heart, kidneys and skin (11).

However, endocrine abnormalities, associated with tuberous sclerosis complex, have been previously reported. Moreover, angiomyolipoma of the adrenal gland, thyroid adenoma, pancreatic islet cell adenoma, pituitary adenoma, testicular angiomyolipoma, hyperparathyroidism, as well as hypothalamic-pituitary-gonadal abnormalities, resulting in precocious puberty, have been

reported in patients with tuberous sclerosis complex (12).

Therefore, hypothyroidism in our case could be a coincidental finding or a consequence of tuberous sclerosis gene products, causing thyroid dysgenesis. Although the co-occurrence of thyroid dysgenesis and tuberous sclerosis is rare, there have been reports in the past. The present case again highlights the importance of further research on the genetic aspect of this co-occurrence (13).

Conflict of Interest

The authors declare no conflict of interest.

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