A Case of Idiopathic Hypertrophic Obstructive Cardiomyopathy Causing Severe Right Ventricular Outflow Tract Obstruction in Infancy

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SUMMARY
An infant with idiopathic hypertrophic obstructive cardiomyopathy, presenting as severe right ventricular outflow obstruction, is described. The nonspecific clinical features and an unexpected angiocardiographic appearance made determining the correct diagnosis difficult. This infant was mistakenly diagnosed as having severe pulmonary stenosis. Attempted surgical correction was unsuccessful and at necropsy this diagnosis was confirmed. It is suggested that cross-sectional echocardiography may be diagnostic for infants with idiopathic hypertrophic obstructive cardiomyopathy.

Additional Indexing Words:
Asymmetric septal hypertrophy  Cross-sectional echocardiography

IDIOPATHIC hypertrophic obstructive cardiomyopathy presenting as right ventricular outflow obstruction in infancy is very rare. Only 4 cases have been reported,1–3) to our knowledge. This report describes an infant with severe right ventricular outflow obstruction due to idiopathic hypertrophic obstructive cardiomyopathy. The difficulty encountered in diagnosis and the important cross-sectional echocardiographic features are discussed.

CASE REPORT
This male infant was the product of a normal full-term pregnancy and an uneventful delivery (birth weight 3.3 Kg). Though a cardiac murmur had been detected at birth, the neonatal period was uncomplicated. At 6

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weeks of age, he was referred for assessment of a cardiac murmur and failure to thrive. The family history was not contributory.

Physical examination on admission showed a heart rate of 156/min and a respiratory rate of 58/min. Blood pressure was 86/40 mmHg. Body weight was 4.1 Kg. Cyanosis was present with crying or exertion. The second heart sound split normally and the pulmonic component was diminished in intensity. No ejection click was heard. A grade 3/6 systolic ejection murmur was loudest at the third left intercostal space. There was no diastolic murmur. A third heart sound gallop was present at the apex. No rales were present in either lung field. The liver was palpable 4 cm below the right costal margin. Peripheral pulses were normal and equal in amplitude.

The chest X-ray showed pronounced cardiomegaly (cardiothoracic ratio = 0.73) and slightly diminished pulmonary vascular markings (Fig. 1). The electrocardiogram showed marked right axis deviation, right atrial enlargement, and abnormal right ventricular preponderance (Fig. 2). The M-mode echocardiogram revealed no pulmonary valves and the ventricular septal
thickness was remarkably increased. (Septal to postero-basal left ventricular wall thickness ratio was 3.2.) The cross-sectional echocardiogram revealed severe ventricular septal hypertrophy which was obstructing the right ventricular outflow (Fig. 3).

The results of the cardiac catheterization are shown in Table I. The right ventricular systolic pressure was abnormally elevated to a level above systemic arterial pressure. The right atrial pressure curve showed a prominent 'a' wave. Systemic arterial oxygen saturation was only 92%, but there was no evidence of a left to right shunt. The selective right ventricular angiocardiogram was interpreted as showing severe right ventricular outflow obstruction secondary to either a pulmonary stenosis or a cardiac tumour, and the pulmonary valve cusps were thought to be thickened and domed (Fig. 4). Because of poor clinical progress with the development of increasingly severe congestive heart failure despite digoxin and diuretic therapy, an operation was performed at the age of 2 months.

Utilizing cardiopulmonary bypass, profound hypothermia and complete circulatory arrest, surgical relief of the obstruction was attempted. The
pulmonary valve, though deformed, was not stenosed. Instead, the right ventricular outflow obstruction was the result of pronounced hypertrophy of the interventricular septum. Despite partial resection of the obstructing muscle mass and right ventricular outflow plasty using a Gore-Tex® patch, the patient did not survive the operation because of low cardiac output failure.

At necropsy there was generalized cardiac hypertrophy with right and left ventricular free wall thicknesses of 1.0 cm and 1.8 cm, respectively. The ventricular septum was 3.4 cm in thickness (Fig. 5). The hypertrophy of the interventricular septum was disproportionately severe and had resulted in obstruction of both ventricular outflow tracts. The pulmonary valve cusps were thickened but there was no pulmonary valve stenosis. While the right atrium showed gross dilatation, the remainder of the heart was normal. Histological examination showed the individual muscle fibers to be variably enlarged with loss of the normal parallel orientation of muscle cells which
were arranged perpendicularly and obliquely to each other (Fig. 6). The transverse diameters of muscle cells in the ventricular septum and walls ranged from 15 to 25 μ. The muscle cell nuclei were found to be large, irregular, and hyperchromatic but there was no inflammatory cell infiltration. The above gross and histological features were considered to be typical of idiopathic hypertrophic obstructive cardiomyopathy.

**DISCUSSION**

Recently, in parallel with the increasing use of echocardiography, the reported incidence of idiopathic hypertrophic obstructive cardiomyopathy in children has increased gradually. It has only rarely been recognized as originally suspected in the first 3 months of life, however, because of difficulties encountered in this age. Furthermore, it has long been recognized that although mild right ventricular obstruction may be present in this disease, it is uncommon. Nevertheless, severe right ventricular outflow obstruction in infancy is very rare and is often mistakenly diagnosed as pulmonary stenosis. This differential diagnosis is important because of the therapeutic implications.

Even in retrospect the clinical features of our case did not suggest the diagnosis of idiopathic hypertrophic obstructive cardiomyopathy. The electrocardiogram showed no left ventricular hypertrophy, ST segment depression, T wave inversion, or intraventricular conduction defects. In addition, the M-mode echocardiogram did not show systolic anterior motion of the anterior leaflet of the mitral valve and premature closure of the aortic valve. Though the septal to postero-basal left ventricular wall thickness ratio was 3.2, this could have been a result of the markedly hyper-
trophied ventricular septum. In the cross-sectional echocardiogram, the severely hypertrophied ventricular septum bulged into the right ventricular outflow tract and markedly reduced the size of its cavity. Because the left ventricular findings were not so remarkable and septal echo was not homogeneous, we had concluded the hypertrophy to be due to a cardiac tumour rather than hypertrophic obstructive cardiomyopathy. While it is at least conceivable that a similar appearance could result from septal hypertrophy secondary to severe pulmonary stenosis, we had not encountered such a case, and few had been reported previously. We therefore did not believe that this cross-sectional echocardiographic appearance strongly suggested hypertrophic obstructive cardiomyopathy. At cardiac catheterization, the pulmonary artery was not entered from the right ventricle and the left ventricle was not entered from the left atrium. In addition, no retrograde left heart catheterization was performed. As a consequence, pressure gradients could not be demonstrated across both ventricular outflow tracts and a left ventricular angiogram could not be performed. Although the appearance of the right ventricular angiogram led us to diagnose severe pulmonary stenosis, it was this appearance which, in retrospect, may have been diagnostic of hypertrophic obstructive cardiomyopathy. The angiogram showed the typical appearance of this disease due to encroachment upon the right ventricular outflow tract by the massively hypertrophied ventricular septum. However, the most striking abnormality, in our case, was the pronounced narrowing at the level of the pulmonary valve and proximal main pulmonary artery due, we believed, to their compression by the hypertrophied free wall of the right ventricle.

Surgical incision or resection of the ventricular outflow tract has been effective in some patients, but the futility of attempted surgical relief in infancy is illustrated by our case and others. Beta-adrenergic receptor blocking agents have been used successfully and are the accepted initial treatment of choice. The negative inotropic effect of these drugs has been at least a relative contraindication to their use in the presence of congestive heart failure, and this was the reason that propranolol was not used in our case. However, the remarkable response to propranolol reported elsewhere, despite congestive heart failure, suggests that beta-adrenergic receptor blockade may have been of benefit in our case.

The familial nature of this disease has always been recognized. The disease may occur in many members of the same family and demonstrates an autosomal dominant pattern with a high degree of penetration. This patient's parents and 2-year-old sister are in good health and their echocardiograms are normal. Although severe right ventricular outflow obstruction
such as was seen in our case is of unknown etiology, it can be present in utero.

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