

Primary Hypertrophic Cardiomyopathy in Noonan Syndrome

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Abstract

We describe a case of Noonan syndrome referred to the department of pediatric cardiology for routine evaluation of cardiovascular abnormalities. Physical examination, electrocardiogram, chest X-ray and echocardiographic finding confirmed severe hypertrophic cardiomyopathy in the absence of any other cardiac abnormalities or systemic condition (*Iranian Heart Journal 2007; 8 (1): 52-54*).

Key words: Noonan syndrome ■ hypertrophic cardiomyopathy

Hypertrophic cardiomyopathy includes a thickened but nondilated left ventricle in the absence of other cardiac or systemic disease.¹⁻³ Mutations of genes for contractile protein on chromosome 14 and other chromosome loci are responsible for this condition.^{2,5,6,7}

Histological and morphologic abnormalities produce a disorder of relaxation, and sometimes left outflow tract obstruction; but overt clinical manifestations may not be present for decades.^{6,7}

More than 70 terms have been used to describe hypertrophic cardiomyopathy (HCM); emphasizing different aspects of the disorder; such as the site and asymmetry of left ventricle hypertrophy.^{3,8,9}

By the 1980s, the term hypertrophic cardiomyopathy was favored.^{3,7} Hypertrophic cardiomyopathy can occur as a congenital heart malformation. In most cases in both children and adults the condition behaves as an autosomal dominant disorder. HCM was noted in glycogen storage disease, infants of diabetic mother, and in Noonan syndrome.

Case report

A 7 year-old Iranian girl was referred to the department of pediatric cardiology. Physical examination at time of admission disclosed a body weight of 17 kg (Z-score, 1.76); a height of 104cm (Z-score, -3.03) with upper to lower segment ratio of 1.2/1, and blood pressure of 95/77mmHg.

She had a short and webbed neck; low posterior hairline, and shield chest (Fig. 1).



Fig. 1. Webbed neck and shield chest in 7-year old girl with Noonan syndrome

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HCM may be identified or cause disability and death at any age, including early childhood. During the past 40 years, our understanding of the complexity of HCM has increased dramatically. On the other hand, perhaps no other cardiovascular disease has presented the challenges and controversies with respect to diagnosis, clinical course, and management as has HCM. Development and progression of left ventricular hypertrophy can lead to ventricular outflow obstruction in 25% of patients.^{8,9}

Prognosis is significantly affected by age and mode of presentation. Mortality for HCM is twice as high in children as in adults. Presentation is by heart murmur, and congestive heart failure and arrhythmia more likely portend a poor prognosis.

Both medical and surgical treatment may improve quality of life.

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