

Clinical Genetics

AB036. Cardiac features in Taiwanese patients with mucopolysaccharidosis IVA

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Background: Mucopolysaccharidosis IVA (MPS IVA; Morquio A syndrome) is a rare lysosomal storage disorder caused by the deficiency of N-acetylgalactosamine-6-sulfatase, an enzyme involved in the catabolism of glycosaminoglycans, keratan sulfate and chondroitin-6-sulfate. Secondary cardiac abnormalities are common in patients with MPS of any types, with the most documented abnormalities being cardiac valve thickening, valvular regurgitation and stenosis, and cardiac hypertrophy.

Methods: We reviewed medical records, echocardiograms, and electrocardiograms (ECG) of 32 Taiwanese patients with MPS IVA (16 males and 16 females; median age, 10.8 years; age range, 1.1 to 29.1 years).

Results: The most common ECG findings (n=27) were sinus tachycardia (37%), sinus arrhythmia (33%), and right or left axis deviation (19%). Echocardiographic examinations

(n=32) revealed the mean z scores of left ventricular mass index (LVMI), interventricular septum diameter in diastole (IVSd), left ventricular posterior wall diameter in diastole (LVPWd), and aortic diameter at 0.94, 2.70, 0.39, and 3.26, respectively. Z scores >2 were identified in 25%, 50%, 29%, and 69% for LVMI, IVSd, LVPWd, and aortic diameter, respectively. Diastolic dysfunction defined as reversed ratio between early and late (atrial) ventricular filling velocity or (E/A ratio <1) was identified in 4 (13%) patients; however, ejection fraction was normal (50–75%) for all patients. Twenty-five (78%) patients had valvular heart disease. The z scores of LVMI, IVSd, LVPWd, and aortic diameter, the severity scores of aortic stenosis and regurgitation, and the existence of the thickened IVS were all positively correlated with the increasing age (P<0.05). Of these 14 patients with valve thickening, the z scores of LVMI, IVSd and aortic diameter were all larger than those of 18 patients without valve thickening (P<0.05).

Conclusions: Most patients with MPS IVA had valvular heart disease and cardiac hypertrophy. The severity of cardiac abnormalities increased with advancing age as the progressive nature of this disease.

Keywords: Cardiac; echocardiography; electrocardiography; mucopolysaccharidosis IVA

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