Cardiac ultrasound findings in children with mucopolysaccharidosis

Lazea C. (1,2), Grigorescu-Sido P. (1,2), Al-Khzouz C. (1,2), Bucerzan S. (1,2), Nascu I. (2)
University of Medicine and Pharmacy „Iuliu Hatieganu”, 1st Department of Pediatrics, Cluj-Napoca, Romania (1)
Emergency Children Hospital, Clinic Pediatrics I, Cluj-Napoca, Romania(2)

Introduction. Cardiac involvement in mucopolysaccharidosis (MPS) is variable, consisting in severe cardiac valve disease and ventricular hypertrophy and has a major contribution into the morbidity and mortality of these patients. The aim of the study was to characterise the echocardiographic abnormalities in children with different types of mucopolysaccharidosis and their evolution after 12 months of enzyme replacement therapy.

Methods. We evaluated the function of the mitral and aortic valves, left ventricular chamber dimensions, septal and posterior wall thicknesses and ventricular function in 20 patients (5 patients with MPS type I, 14 patients with MPS type II and one patient with MPS type IV), aged 1-16 years.

Results. At diagnosis, all patients presented echocardiographic alterations. Mitral valve thickening with variable grades of regurgitation was diagnosed in all patients; mitral stenosis was present in 10% of patients. Aortic regurgitation was present in 68% of patients and aortic stenosis in 5%. Left ventricular hypertrophy was diagnosed in 40% of patients and there was no systolic dysfunction. Mild pulmonary hypertension was present in 26% of the patients. Thirteen patients (3 patients with MPS type I and 10 patients with MPS type II) underwent enzyme replacement therapy. After 12 months of treatment we obtained stabilization of cardiac valvular disease in 69% of patients, mild improvement in 8% and worsening of disease in 23% of patients.

Conclusions. Left valve lesions, ventricular hypertrophy, and pulmonary hypertension are the most common findings in children with mucopolysaccharidosis. Enzyme replacement therapy had little effect on cardiac valve disease.