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Cardiac results of enzyme replacement therapy in Romanian children with mucopolysaccharidosis type I and II

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Introduction. The prevalence and severity of cardiovascular disease in patients with mucopolysaccharidosis (MPS) is high and progressive, consisting in severe cardiac valve disease and ventricular hypertrophy. Enzyme replacement therapy in MPS may improve the organ impairment. The aim of the study was to characterize the cardiac disease and the results of enzyme replacement therapy in children with MPS type I and MPS type II.

Methods. We evaluated 23 patients with mucopolysaccharidosis (MPS): 5 patients with MPS type I and 18 patients with MPS type II. We assessed the function of valves, left ventricular chamber dimensions, septal and posterior ventricular wall thicknesses, systolic and diastolic function of the ventricles, pulmonary hypertension at every 6 months. The treatment of these patients consisted in weekly administration of recombinant form of human alpha-L-iduronidase in dose of 0.58mg/kg iv weekly for 5 patients with MPS type I and iduronate 2-sulfatase in dose of 0.5mg/kg, iv weekly for 16 patients with MPS type II.

Results. At diagnosis, all patients presented echocardiographic alterations. Mitral valve thickening with variable grades of regurgitation was diagnosed in all patients, aortic regurgitation was present in 56% of patients and mitral stenosis in 9% of patients. Left ventricular hypertrophy was diagnosed in 35% of patients. Mild pulmonary hypertension was present in 17% of patients. The mean age of the patients at starting therapy was 6.1 year. The treatment results after 2 years were: valvular heart disease were stable in 67% of patients, mild improvement was present in 19% of patients and aggravation in 14% of patients. Ventricular hypertrophy remained unmodified in 67% of patients and worsened in 11% of patients. After 4 years of treatment the results were: valvular heart disease were stable in 67% of patients, mild improvement was present in 14% of patients and aggravation in 19% of patients. Ventricular hypertrophy remained unmodified in 55% of patients and improvement was recorded in 45% of patients.

Conclusions. The most prevalent cardiac changes in children with MPS are valvular lesions. Enzyme replacement therapy had little effect on cardiac disease in children with MPS.