

Effects of enzyme replacement therapy on cardiac disease in children with mucopolysaccharidosis type II

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

Methods. The treatment of these patients consisted in weekly administration of recombinant form of human iduronate 2-sulfatase in dose of 0.5mg/kg, iv. We assessed the function of the mitral and aortic valves, left ventricular chamber dimensions, septal and posterior wall thicknesses and ventricular function in 15 patients with MPS type II every 6 months after starting of treatment.

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 9 patients and aortic stenosis in 2 patients. Left ventricular hypertrophy was diagnosed in 7 patients.

Mild pulmonary hypertension was present in 4 patients. The mean age of the patients at starting therapy was 6.1 years. Duration of treatment was 12 months in 3 patients, 2 years in 7 patients, 3 years in 2 patients, 4 years in 2 patients and 5 years in one patient. The treatment results on valvular heart disease were: stable disease in 8 patients, mild improvement in 2 patients and aggravation in 5 patients. Ventricular hypertrophy remained unmodified in 6 patients and worsened in one patient.

Conclusions. Enzyme replacement therapy had little effect on cardiac disease in children with MPS type II.