

# Heart and cardiovascular involvement in Russian patients with mucopolysaccharidosis: effects of enzyme replacement therapy

Nato Vashakmadze<sup>1, 2</sup>, Leyla Namazova-Baranova<sup>1, 2, 3</sup>, Natalia Zhurkova<sup>1</sup>, Gregory Revunenkov<sup>1</sup>

<sup>1</sup> Research Institute of Pediatrics and Children's Health in "Central Clinical Hospital of the Russian Academy of Sciences", Moscow, Russian Federation

<sup>2</sup> Pirogov Russian National Research Medical University, Moscow, Russian Federation

<sup>3</sup> Belgorod National Research University, Belgorod, Russian Federation

**Objective.** Mucopolysaccharidosis is a rare lysosomal storage disorder. Clinical phenotype is very variable. Patients usually have short stature, multiplex dysostosis, facial dysmorphism, cardiovascular abnormalities and other changes.

**Methods.** We study cardiovascular and heart findings in patients with mucopolysaccharidosis II (MPS II) and effect after enzyme replacement therapy (ERT). 55 boys were included in our study. Cardiac problem has 49 patients (median age  $85,00 \pm 7,26$  (3–207) month). ERT was given to 46 patients since 2008 (median age  $81,54 \pm 50,24$  month).

**Results.** Before ERT, mitral regurgitation had 27 patients, thickening of the mitral valve — 17, stenosis mitral valve 1, aortic regurgitation — 20 patients, thickening of the aortic valve — 6, stenosis aortic valve — 1, tricuspid regurgitation 13 patients, pulmonary valve regurgitation — 4.

After ERT — mitral regurgitation had 35 patients, thickening of the mitral valve — 20, stenosis mitral valve — 0, aortic regurgitation — 23 patients, thickening of the aortic

valve — 35, stenosis aortic valve — 0, tricuspid regurgitation — 14 patients, pulmonary valve regurgitation — 3.

There were no statistically significant deterioration or improvement of the valve heart apparatus in the examined group of patients ( $P > 0,05$ ).

Cardiomyopathy (left ventricular hypertrophy) have 6 boys with MPS II before and after ERT, lung hypertension — 2 cases before ERT, 4 — after. The course of heart failure in MPS is progressive and we evaluated the heart condition by functional class. We have identified significantly high-performance functional class at first patient visit (I class — 44,4% patients, II class — 44,4%). After ERT (median age  $81,54 \pm 50,24$  month) 89% patients have no negative dynamics. It should be noted that most patients began receiving ERT at age 6–8 years old, they had severe somatic and neurological symptoms.

**Conclusion.** ERT is not able to reverse the cardiac damage but provides stabilization of heart failure. Early initiation of ERT is a factor preventing severe heart disease in MPSII patients.