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Report of two observations of left ventricular systolic disfunction in children with mucopolysaccharidosis (MPS) type 1

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Introduction. Our purpose is focusing pediatricians and cardiologists on the problem of early diagnosis of rare genetic diseases in children with heart disease.
Methods. In the department of cardiology Scientific Centre of Children Health RAMS were admitted 2 children with a diagnosis of dilated cardiomyopathy. On physical examination both children at an early age (3 and 7 months) were observed to have coarse facial features (wide and depressed nasal bridge, large nostrils, hypertelorism, gingival hypertrophy), hirsutism, short neck, hypersalivation, noisy breathing, joint stiffness, hepatosplenomegaly. Also both children had symptoms of congestive heart failure. An electrocardiogram showed atrial hypertrophy, hypertrophy of both ventricles, marked disturbances of repolarization. According to the ultrasound examination - dilatation of the left chambers of the heart, a reduction in myocardial contractility (EF < 35%). We paid attention to uncharacteristic for dilated cardiomyopathy hypertrophy of the left ventricular wall (6-7 mm). Based on clinical findings we suspected a storage disease, in both cases, the diagnosis of MPS type 1 was confirmed by the analysis of urine glycosaminoglycans and genetically. The specific enzyme replacement therapy with laronidase (Aldurazim©) was initiated at early age.
Result and conclusions. At MPS type 1 (Hurler syndrome) heart is always affected, manifesting with ventricular hypertrophy, thickening of the heart valves, but preserved myocardial contractility, and chronic heart failure usually occurs only at late stages of the disease. In this case, the two children have shown a dramatic dilation of the left heart and decreased myocardial contractility, which led to the development of symptoms of congestive heart failure caused, apparently, by myocarditis. The presence of a cardiac dilatation and reduced ejection fraction in conjunction with myocardial hypertrophy and specific clinical features is the ground for a deep survey on storage diseases, which helps to diagnose a rare genetic disease at an early age and start specific therapy at the proper time.