Mucopolysaccharidoses: clinical and epidemiological characteristics, cardiac manifestations, current approaches to diagnosis and treatment

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ABSTRACT. This article presents a comprehensive analysis of the current scientific literature on mucopolysaccharidoses (MPS) – a heterogeneous group of rare inherited lysosomal storage disorders. It highlights current data from epidemiological studies and presents modern views on the etiopathogenesis and clinical polymorphism of various types of MPS.

Particular attention is paid to the systematization of general clinical markers, which allows the practicing physician to suspect the pathology in a timely manner and perform differential diagnosis with similar conditions. The integral links between types of MPS and specific lesions of the cardiovascular system are analyzed in detail. The authors have systematized the most common cardiac manifestations: acquired valvular defects (mitral and aortic valve insufficiency and stenosis), arrhythmias and conduction disturbances, cardiomyopathies, as well as the development of progressive heart failure.

An algorithm for modern laboratory and instrumental diagnostics has been established, based on a combination of imaging methods and mandatory verification of the diagnosis through enzymatic and molecular-genetic testing. The evolution of therapeutic strategies for MPS is reviewed, including the efficacy of enzyme replacement therapy, hematopoietic stem cell transplantation, and promising directions in gene engineering.

KEY WORDS: mucopolysaccharidosis, MPS, lysosomal storage diseases, glycosaminoglycans, heart defects, cardiomyopathy, ventricular hypertrophy, enzymatic diagnosis, enzyme therapy.