

PROSPECTS FOR THE DEVELOPMENT OF A DIAGNOSTIC ALGORITHM FOR THE MAIN CLINICAL SIGNS OF MUCOPOLYSACCHARIDOSIS

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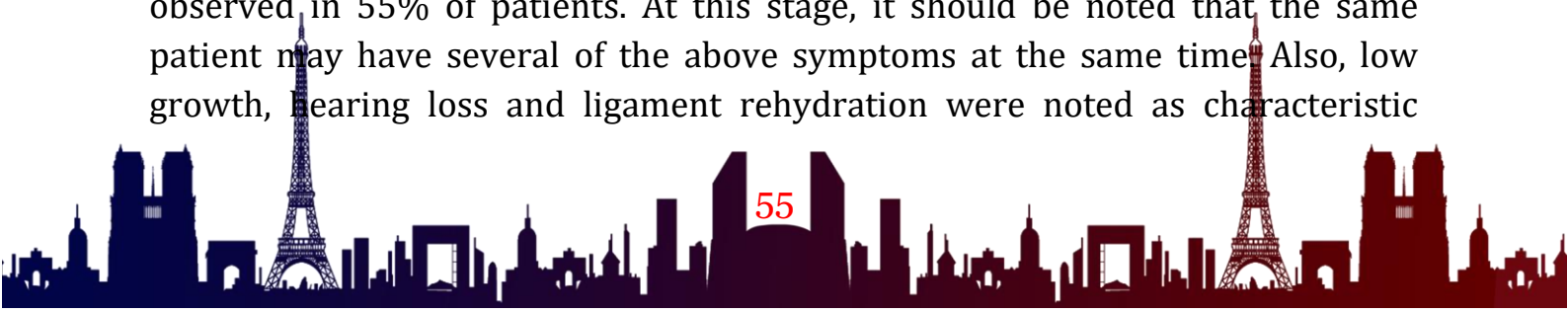
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Introduction. Mucopolysaccharidoses are a group of rare lysosomal accumulation diseases that exhibit numerous organic and severe symptoms. Although they are relatively rare, mucopolysaccharidoses are found all over the world in various forms. The manifestation of this serious disease in childhood or the appearance in children indicates a more serious problem. Early diagnosis of mucopolysaccharidosis at an asymptomatic stage can be effective for preserving organic functions and improving results. However, delayed diagnosis is common due to the latent onset of the disease and limitations of sensitive laboratory parameters [1,2,3]. As a result, it is extremely important to study new diagnostic methods based on molecular mechanisms and pathological changes caused by mucopolysaccharidoses. In this regard, the presented manuscript examines the main clinical signs specific to each type, which are most often found in the diagnosis of mucopolysaccharidosis [4,5, 6].

The purpose of the study. The main purpose of this presented manuscript is to study the features of the main clinical signs specific to each type in the diagnosis of mucopolysaccharidosis.

Materials and methods of research. As an object for determining the specificity of clinical signs characteristic of each type of mucopolysaccharidosis, 40 patients were selected whose diagnosis of mucopolysaccharidosis was confirmed by clinical laboratory instrumental methods. Anatomical pathologies, cognitive impairment, changes in visceral organs, structural and functional changes in the organs of the oral cavity, occurring in 7 types of mucopolysaccharidosis, were noted as the main clinical signs.

Results. In studies, cognitive impairment was observed in 95% of patients, visceral organ pathologies in 75% of patients, diseases related to the nervous system in 80% of patients, and other types of pathologies were observed in 55% of patients. At this stage, it should be noted that the same patient may have several of the above symptoms at the same time. Also, low growth, hearing loss and ligament rehydration were noted as characteristic



signs of all 7 types of mucopolysaccharidosis in patients. On the other hand, the first 3 types of mucopolysaccharidosis were observed significantly more often among the studied patients compared with the other types. The study analyzed both common and differentiated clinical signs in order to determine the specificity of clinical signs characteristic of all types of mucopolysaccharidosis and occurring in each patient

Conclusion. Thus, mucopolysaccharidosis is a serious and urgent disease for the health care system and is often found in a genetic form. Detection of this disease at an early stage is important for its treatment and improvement of the quality of life of patients. Not only the types, but also the clinical signs of mucopolysaccharidosis are diverse, which creates a number of difficulties and problems in its diagnosis. Even in the course of the study, it was found that it is with almost all types of clinical signs and in each patient that several signs are observed. This, in turn, requires doctors to develop more optimal methods and more careful study.

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