KIDNEY PATHOLOGY IN ORPHAN DISEASES IN CHILDREN

[1]Akhmedova Dilorom Ilkhamovna, [2]Abidova Malikakhon Davronovna
[1]DSc, Professor, Director of the Republican Specialized Scientific and Practical Medical Center of Pediatrics, Head of the Department of Hospital Pediatrics No. 2, Folk Medicine of the Tashkent Pediatric Medical Institute
[2] Clinical resident of the Republican Specialized Scientific and Practical Medical Center of Nephrology and Kidney Transplantation
https://doi.org/10.5281/zenodo.8436064

Abstract. *The article discusses the problem of kidney pathology that occurs in orphan diseases in children. Orphan diseases are rare genetic or congenital disorders that affect a small group of patients. The article is based on a review of the literature and studies conducted in the field of kidney pathology in children with orphan diseases. It discusses various types of orphan diseases that can lead to the development of kidney pathology, such as Polander syndrome, Alport syndrome, Fanconi syndrome and others. The article describes the main clinical manifestations of kidney pathology in orphan diseases in children, such as proteinuria, hematuria, hypertension and impaired renal function. In conclusion, the article emphasizes the importance of early diagnosis and timely treatment of kidney pathology in orphan diseases in children.*
Keywords: *kidney pathology, children, orphan diseases, pediatrics, nephrology*

Research results show that orphan diseases in children associated with kidney pathology are a serious problem in pediatrics. Orphan diseases, also known as rare diseases, are characterized by low prevalence and often cause significant problems for patients and their families.

According to a study conducted in 2018 and published in the journal “Pediatric Nephrology”, orphan kidney diseases account for approximately 10-15% of all cases of chronic renal failure in children [7].

Another study published in the journal “Nephrology Dialysis Transplantation” in 2019 indicates that orphan kidney diseases in almost 50% of cases can cause the development of chronic kidney failure in children in developing countries [6].

For specific orphan kidney diseases, such as Alport syndrome and Bartter syndrome, exact prevalence data may vary depending on geographical distribution and population from 1 case per 5,000 people to 1 case per 1,000,000 population [9, 11].

The purpose of the study. To analyze the scientific literature in international scientific databases and search engines about orphan diseases in children accompanied by kidney pathology.

Research materials and methods. The analysis of scientific publications – theses, articles and dissertations in international scientific databases and scientific electronic libraries eLibrary, CyberLeninka, Pubmed, Cochrane Library, Web of Science, Embase, including available works in Russian literature were performed.

Research results. Kidney pathology is one of the most common causes of orphan diseases in children. It can include various conditions, such as congenital kidney abnormalities, genetic disorders, syndromes associated with kidney failure, and other rare diseases affecting kidney function.

Kidney failure in children with orphan diseases is a serious problem that requires serious attention and specialized treatment. Orphan diseases, also known as rare diseases, are

characterized by low prevalence and often cause significant problems for patients and their families.

One of the most common orphan diseases that lead to kidney failure in children is Fanconi syndrome. This is a genetic disorder that leads to defects in reabsorption due to dysfunction of the proximal renal tubules, causing the loss of various substances in the urine, including glucose, amino acids and electrolytes. According to a study conducted by Hou et al. in 2018 (Hou et al., 2018), Fanconi syndrome occurs in approximately 1 in 100,000 newborns. Patients with Fanconi syndrome often experience polyuria (frequent urination) and polydipsia (excessive thirst) due to loss of salts and water through the kidneys, hypophosphatemia (low levels of phosphate in the blood), which can lead to weakening of bones and the development of osteoporosis, metabolic acidosis (a shift in the pH of the blood towards the acidic side), due to violations of bicarbonate reabsorption in the kidneys and the appearance of various defects in kidney development, such as cysts or deformities. Studies show that mutations in various genes, such as FANCA, FANCB, FANCC and others, may be responsible for the development of this syndrome.

One of the most studied orphan diseases associated with kidney pathology is Alport syndrome. This is a genetic disease that leads to progressive kidney failure and damage to other organs, such as the hearing aid and eyes. According to a study conducted by Shah and co-authors in 2019 (Shah et al., 2019), Alport syndrome accounts for about 2-3% of all cases of chronic renal failure in children. In this pathology, kidney damage in children is accompanied by proteinuria (increased protein content in the urine), which may be the first sign of kidney damage, hematuria (the appearance of blood in the urine), which may manifest as a red or brown tinge of urine and a gradual decrease in kidney function, which can lead to chronic renal failure. Studies show that mutations in the genes encoding collagen IV are the main cause of the development of Alport syndrome [9].

Another rare disease associated with kidney pathology is Bartter syndrome. This is a genetic disorder that leads to impaired reabsorption of certain electrolytes in the kidneys (hypokalemia, hypochloremia, metabolic alkalosis and hyperreninemic hyperaldosteronism) due to loop tubulopathy in the ascending knee of the Henle loop [4, 6]. Children with Bartter syndrome may have the following symptoms of kidney damage: polyuria (frequent urination) and polydipsia (excessive thirst) due to loss of salts and water through the kidneys, hypokalemia (low potassium levels in the blood), which can lead to muscle weakness, heart rhythm disturbances and other problems; metabolic alkalosis also develops (blood pH shift towards the alkaline side) caused by the loss of chlorides and other electrolytes through the kidneys [11]. Studies show that mutations in various genes, such as SLC12A1, SLC12A3 and KCNJ1, may be responsible for the development of this syndrome [1].

Fabry's disease (a genetic disease also known as factory disease) can cause various kidney problems in children. Fabry's disease is characterized by a lack of the enzyme alpha-galactosidase A, which leads to the accumulation of fatty substances in various organs, including the kidneys [10]. Children with Fabry's disease may experience a gradual deterioration in kidney function. This can manifest itself in the form of increased protein content in the urine (proteinuria), the appearance of blood in the urine (hematuria), as well as deterioration of kidney function, which can lead to a gradual decrease in their work. It is important to note that the symptoms and severity of kidney damage may vary in different children with Fabry's disease [10]. Some children may have a milder kidney lesion, while others may have a more serious one [2].

A multidisciplinary integrated approach is needed for the diagnosis and treatment of orphan diseases associated with kidney pathology. This includes medical and genetic counseling, laboratory research, educational support for patients and their families, as well as the choice of optimal treatment depending on the specific disease. Children with suspected orphan disease should be referred to geneticists, neurologists and other relevant specialists to conduct a detailed examination and establish an accurate diagnosis.

Treatment of kidney failure in children with orphan diseases may include conservative methods such as diet, pharmacotherapy and maintenance therapy, as well as more invasive methods such as dialysis and kidney transplantation [3]. The decision on choosing the optimal treatment should be made taking into account the individual characteristics of the patient and the characteristics of the disease.

However, the availability of effective treatment of kidney failure in children with orphan diseases remains a problem. Lack of information and experience in the field of rare diseases, as well as high treatment costs, can create barriers to obtaining the necessary assistance. Therefore, it is important to continue research and development of new methods of diagnosis and treatment, as well as to raise awareness of medical professionals and the public about rare kidney diseases in children.

Conclusion. Thus, orphan diseases of children associated with kidney pathology are a serious problem in pediatrics. Research in this area allows us to better understand the causes and mechanisms of the development of these diseases, which in turn contributes to the development of new methods of diagnosis and treatment. However, further research and improving the availability of treatment remain important tasks to improve the prognosis and quality of life of children suffering from orphan diseases associated with kidney pathology.

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