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DIAGNOSIS AND MANAGEMENT OF THE IPEX SYNDROME

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Relevance. IPEX syndrome is an X-linked inborn error of immunity clinically characterized by the triad of: enteropathy, polyendocrinopathy and eczema. However many other clinical presentations lacking the triad above described have been reported what underpin the need of careful clinical suspicion, immunological evaluation and genetic sequencing.

Aim: to present a clinical case of the IPEX syndrome in a child aged 5 years.

Materials and methods. The object of the study was patient N., 5 years old, undergoing treatment at the 3rd City Children's Clinical Hospital and the Belarusian Research Center for Pediatric Oncology, Hematology and Immunology in Minsk. More than 100 articles from from PubMed, Google Scholar and UpToDate databases have been analyzed.

Results and their discussion. The patient was initially admitted to the hospital in 2016, at the age of 2 years, due to hypoalbuminemia, hypoproteinemia and presence of erythrocytes in the urine. The initial corticosteroid treatment initially led to positive dynamics with respect to proteinuria, which however, still remained nephrotic; no changes in the biochemical blood parameters of albumin and protein were observed leading to the requirement of human albumin infusions. Ultrasound examination and biopsy indicated the possible presence of an autoimmune enteropathy. Patient was also diagnosed with protein-energy deficiency of the 2nd degree.

The patient's subsequent admissions in 2017, 2018 and 2019 were a result of exacerbation of the nephrotic syndrome.

In 2017, the administration of corticosteroids showed an almost immediate resolution of proteinuria; less impressive changes were noted in the biochemical analysis of blood. In 2018, the patient presented with severe manifestations: edematous face, proteinuria (1.4 g/L) and wheezing in bilateral lung fields. The treatment led to a significant resolution of daily proteinuria however, several sporadic increases above 1g/L were present during the course of hospitalization.

In 2019, the patient presented with proteinuria, hypoproteinemia and hypoalbuminemia. Daily proteinuria reached 1.8 g/L. Concomitantly, a diagnosis of acute rhino-pharyngitis was made.

In 2020, the patient presented with loose stool, signs of hypoproteinemia and hypoalbuminemia. Injections of 10% human albumin was repeatedly done. A puncture kidney biopsy was performed which revealed signs of membranous nephropathy (diffuse thickening of glomerular basement membranes with the presence of intramembranous and sub-epithelial deposits). Esophagogastroduodenoscopy revealed total atrophy of the mucosa of the duodenum, superficial gastritis, insufficiency of the cardia. Colonoscopy revealed hypotrophy of the mucosa of the distal parts of the small intestine, dilatation of the lumen of the colon and signs of acute unexpressed colitis. Results of the genetic testing showed a positive result of the pathogenic variant of FOXP3 (*variant c.748_750del (p.Lys250del*)) which confirmed IPEX syndrome.

The patient's history notably included a diagnosis of the infantile form of atopic dermatitis that was made in the 1st year of life. Concomitant diseases of importance are bronchial asthma and recurrent bronchial obstruction, polyvalent allergy, immunodeficiency and minor congenital anomaly of the heart (extra chord in the left ventricle).

Conclusion: in recent years, although IPEX syndrome is characterized by inflammatory bowel disease, type I diabetes, and skin diseases, the number of IPEX cases exhibiting atypical symptoms was increasing. In our case, the patient except atopic dermatitis suffered from proteinuria, chronic gastritis and bronchial asthma. These symptoms were not the primary symptoms of IPEX syndrome. Therefore, through this case, we recognize that the consideration of IPEX syndrome is important when a child develops multiple system disorders.