ANALYSIS OF PHARMACOTHERAPY SCHEMES FOR THE MOST SOCIALLY SIGNIFICANT RARE DISEASES

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Rare diseases are also known as “orphan diseases", but there is no satisfactory definition of rare diseases around the world. In the United States of America (USA), a rare disease is defined as a disease that affects fewer than 200,000 individuals, but in Japan the number is 50,000 and in Australia it is 2,000. The European Union (EU) definition is less than 5 in 10,000. The World Health Organization (WHO) defines a rare disease as all pathological conditions affecting 0.65–1 out of every 1,000 inhabitants. These numbers clearly relate to the population sizes of these countries, but even adjusting for that, the definitions vary from about 1 to 8 in 10,000. In Ukraine rare disease (RD) - a disease that threatens human life or that progresses chronically, leads to a reduction in life expectancy or disability, the prevalence of which among the population is not more than 1: 2000. The most common and socially priority nosological rare diseases have been identified in Ukraine.

From 275 positions of the List of RD as priority orphan diseases on the principle of "social solidarity" are considered 12 nosological units: juvenile rheumatoid arthritis (JRA), bullous epidermolysis, congenital hypothyroidism, hemophilia, pulmonary arterial hypertension (PAH), cystic fibrosis, mucopolysaccharidosis, orphan metabolic diseases, including phenylketonuria, primary immunodeficiency, Willebrand's disease, Gaucher's disease. RD are differ in symptoms, pathogenetic mechanism of development, methods of screening and diagnosis. The incidence, morbidity and mortality rates vary. Therefore, it is necessary to pay more attention to the available data on these indicators. From 12 priority RD becomes impossible to investigate the tendency of distribution and development of a disease for primary (congenital) immunodeficiency, seeing the absence of the unified clinical protocol. The categories of patients for priority rare nosologies are children, with the exception of pulmonary arterial hypertension (adults also suffer). Unified clinical treatment protocols have been developed for 11 nosological units, except of primary (congenital) immunodeficiency. Basic schemes of therapy are made for 10 nosologies. Exceptions were: primary (congenital) immunodeficiency, bullous epidermoli. According to the analysis of the State Formulary, it was established: medicines for 6 nosologies therapy are included to the State Formulary, namely for the treatment of juvenile rheumatoid arthritis, Willebrand's disease, cystic fibrosis, pulmonary arterial hypertension, congenital hypothyroidism and hemophilia.

Of the 12 priority RD, 6 nosologies have in their treatment regimens medicines that are included to the National list of essential medicines - cystic fibrosis, pulmonary arterial hypertension, congenital hypothyroidism, juvenile rheumatoid arthritis, bullous epidermolysis, hemophilia. It should be noted that this is symptomatic, replacement and / or pathogenetic therapy.

According to unified clinical protocols, each nosology has its own resource supply, which consists of medicines for treatment. The exception is therapy of primary (congenital) immunodeficiency, for which no treatment protocol has been developed, so it becomes impossible to identify the necessary medicines for the treatment of the disease.

The analysis of pharmacotherapy regimens showed that medicines for the treatment of Gaucher disease, Willebrand's disease, phenylketonuria and mucopolysaccharidosis were not included to the National List of essential medicines of Ukraine and to the WHO’s Basic List of medicines. Medicines for the treatment of pulmonary arterial hypertension, bullous epidermolysis, congenital hypothyroidism, hemophilia and juvenile rheumatoid arthritis are fully included to the both - National List of essential medicines and the WHO’s Basic List of medicines. Medicines for the treatment of cystic fibrosis are partially included in the Lists with the possibility of generic replacement.