ANALYSIS OF THE PRIORITY DIAGNOSES AMONG THE ORPHAN DISEASES

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Pharmaceutical care of patients with rare diseases – is an actual problem of healthcare worldwide. Rare (orphan) disease - a disease that threatens human life or which is chronically progressive, leads to a reduction in the life expectancy of a citizen or his disability, which prevalence among the population is not more than 1: 2000. The prevalence of orphan diseases in the world varies, so the lists of orphan diseases vary from country to country. One rare disease may affect only a handful of patients in the EU (European Union), and another may touch as many as 245,000. In the EU, as many as 30 million people may be affected by one of over 6000 existing rare diseases. 80% of rare diseases have identified genetic origins whilst others are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative. 50% of rare diseases affect children.

In Ukraine, the current normative legal act that establishes a list of orphan diseases is the Order of the Ministry of Health of Ukraine dated 30.12.15. No. 919 "On Amendments to the List of Rare (Orphan) Diseases that caused a shortening of life expectancy of patients or their disability and for which there are recognized methods of treatment ", in accordance with the Order No. 731 dated 29.06.17." On Amendments to the List of Rare (Orphan) Diseases, which lead to a shortening of life expectancy of patients or their disability and for which there are recognized methods of treatment» Thus, in Ukraine, the current list of orphan diseases contains 275 nosologies by the code of the International Classification of Diseases and Related Health Problems - ICD-10.

According to the principle of "social solidarity", 12 nosologies were identified as priority orphan diseases in Ukraine: juvenile rheumatoid arthritis, bullous

epidermolysis, congenital hypothyroidism, hemophilia, pulmonary arterial hypertension, cystic fibrosis, mucopolysaccharidosis, orphan metabolic diseases, including phenylketonuria, primary (congenital) immunodeficiency, Villebrand's disease, Gauche disease.

Categories of patients with these nosologies are mainly children, exception - a pulmonary arterial hypertension (adults also are suffered). Unified clinical protocol of treatment for 11 priority nosologies has been developed, except primary (congenital) immunodeficiency. The basic therapies are composed of 10 nosologies. The exceptions are: primary (congenital) immunodeficiency and bullous epidermolysis.

In the study the presence of drugs for the treatment of 12 priority orphan diseases in the National Drug Formulary (10-th edition, 2018) (NDF) has been analyzed. It is established: medicines used for the treatment of 6 orphan diseases among priority states have been included in the National List of Essential Medicines, namely, for the treatment of juvenile rheumatoid arthritis, Willebrand disease, cystic fibrosis, pulmonary arterial hypertension, congenital hypothyroidism and hemophilia [7]. Medicines for treatment of two nosologies of the 12 priority orphan diseases (cystic fibrosis and pulmonary arterial hypertension) have been included in the NDF. It should be noted that this drugs are used for a symptomatic therapy mainly.

It has been established that the providing of patients with orphan diseases requires significant changes. The development of unified clinical protocols and basic therapies will increase the level of providing quality medical and pharmaceutical care. The introduction of necessary medicines in the National List of Essential Medicines and the State Formular is the important mechanism for ensuring the availability of therapy of orphan disease. Creation of criteria for assigning orphan diseases to the list of priority orphan diseases will make it possible to make adjustments in the mechanisms of securing the most socially important orphan diseases.