

Registers of orphan diseases in the world: international practice
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Orphan diseases are congenital or acquired diseases that occur very rarely: in one person among 2,000 people. Only 5% of people with rare diseases know about their disease. And 80% of children die before the age of five.

More than 300 million people in the world live with orphan disease. By comparison, 339 million people are diagnosed with bronchial asthma and 422 million with diabetes. This group of diseases affects more people than AIDS and cancer combined. Every week 5 new conditions and diseases are described in the world.

Newborn screening in Ukraine is performed for only four diseases, while in the United States - for more than forty. The lack of a National Registry and insufficient state funding for the treatment of orphan diseases are the main reasons that prevent orphan patients from exercising their right to life and health.

The international practice of creating registries of orphan diseases and its patients has been analyzed in a study based on the Orphanet portal. The registers will allow the development of programs for clinical and epidemiological research, and calculate the budget for medical and pharmaceutical support. The register of orphan diseases includes the distribution by EU countries and other neighboring countries that are members of the Orphanet network. It consists of a list of rare diseases, the distribution of registries by country, the distribution of registries by scale and the distribution of registries by affiliation to the organization. The portal also contains existing European and international registries of orphan diseases.

The list of rare diseases covered by this register consists of an orphan number and the direct name of the nosology. Orphan number is an identifier of the disease in the nomenclature of the portal Orphanet. The distribution of registers by affiliation is based on the assignment of the list to certain types of organizations: public, private commercial, private non-profit, or affiliation is not defined. This affiliation makes it possible to understand who developed the register, and most importantly - who is the direct source of funding. Belonging to a certain type of organization creates restrictions on access to registers. According to the Orphanet portal, out of 747 registers, 84% belong to public organizations, 6% - to private commercial organizations, 4% - to private non-profit organizations, and for 6% affiliation was not established. The list of international registers includes 86 lists. It specifies two criteria: the country that coordinated the creation and implementation, and the affiliation to the organization. The list of European countries' registers contains 54 lists, which are formed into a single European register according to the same criteria. In Ukraine register of orphan diseases has place and has consisted on 375 nosologies approved by order of the Ministry of Health № 778 October, 27, 2014 (edition from 28.01.2020).

The next stage of the study will be the analysis of countries that provide registers of orphan patients and the study of criteria for creating such registers, that will allow to substantiate expediency and to develop a methodic and algorithm of creation of the register of orphan patients creation in Ukraine.