Analysis of the prevalence of some rare diseases in Ukraine

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Since 2001, mucopolysaccharidosis type I has been diagnosed in 15 patients under 18 years of age and one over 18 years of age. A total of 80 patients were registered in the database of patients with mucopolysaccharidosis in Ukraine. In France, about 8-10 new cases of mucopolysaccharidosis (MPS) of type I are recorded each year. Mucopolysaccharidosis of type II was diagnosed in 23 patients under 18 years of age in Ukraine; the incidence of MPSs type II ranks first among other types. According to statistics, two patients under the age of 18 were diagnosed with MPS type VI. The prevalence of this disease in Europe is 0.23 per 100 000 newborns, the frequency range of MPS type VI in the world is from 1 in 238 095 to 1 in 1 300 000.

Cystic fibrosis is the most common inherited disease that causes death. Prevalence averages 1:2500-1:3500 newborns, ranging from 1:1700 in Northern Ireland and Denmark to 1:40,000 in Finland. According to the neonatal screening results for cystic fibrosis, the average incidence of cystic fibrosis was 1:8400 in Ukraine.

Phenylketonuria belongs to orphan metabolic diseases, so consider it as one of the examples of metabolic disorders. It occurs in approximately one person per 15 000 newborns. The prevalence of phenylketonuria in different regions of Ukraine ranges from 1:6000 to 1:10000; the average incidence of this disease among infants is 1:7697-1:7027.

There were 55 patients with Gaucher disease in Ukraine, including 23 children and 32 adult patients. Among them with type I – 53 people (22 children, 31 adults), type II – 2 people (1 child, 1 adult).

According to research, the prevalence of hemophilia in most European countries is 13-18 cases per 100 000 male population, or according to the WHO and the World Federation of Hemophilia 1:10,000 newborn boys. There are about 350 000 hemophiliacs worldwide. Among hereditary coagulopathies, a significant place is occupied by Willebrand's disease caused by a qualitative and quantitative deficiency of the Willebrand factor. Willebrand's disease occurs with a frequency of 1:100 inhabitants. However, in most of these individuals, no symptoms of hemostasis disorder are observed. The frequency of clinically significant cases is much lower and is 1:10000 inhabitants.

Bullous epidermolysis is a rare genetic disease with a prevalence of 1:30000 in European countries. According to the official data, there were 154 people in Ukraine suffering from bullous epidermolysis.

According to official information, the prevalence of juvenile arthritis among children under 17 in Ukraine in 2008 was 0.34 cases per 1 000 (2 700 children), in 2009 0.37 (3 084 children), and in 2017 0.36 cases (2 987 children). Almost half of the patients become disabled after 3-5 years of illness. Priority diseases include pulmonary arterial hypertension. It is impossible to analyze prevalence in Ukraine in connection with the lack of the register of patients.

Given that data were collected from different sources that have taken much time. Lack or sometimes absence of epidemiological data of rare diseases (RD) is a huge problem. It is impossible to provide an appropriate assessment of therapy of adequate volume of patients' supplying with medicines because of the inability for effective budgeting of RD by nosologies. A significant feature of RD treatment – is a very high cost.

As a solution to the problem is the initiation of Patients' Register by RD. The aspect should be described in the national legislation – prioritization (especially criteria of RD's prioritization from registered 275 nosologies given in the Order of Ministry of Health).

It is possible through co-working of government, scientific organization (universities), doctors' communities, and patient organizations. The availability of up-to-date data allows providing practical health technology assessment in RD pharmaceutical providing of patients with RD who are a socially vulnerable category.

“ Podgaina M.V., Sliptsova N.A. (2020). Analysis of the prevalence of some rare diseases in Ukraine. *Фармакоекономіка в Україні: стан та перспективи розвитку : матеріали XII наук.-практ. INTERNET-конф., м.Харків, 22 травня 2020 р*. 249-250. ”