Rare diseases: prevalence and social significance for society and the state

Podgaina M.V., Sliptsova N.A.

Organization and economy of pharmacy department

National University of Pharmacy, Kharkiv, Ukraine

economica@nuph.edu.ua

More than 300 million people worldwide suffer from one or more diseases. 72% of orphan diseases are genetically determined, and the remaining 28% are diseases that have a bacterial, viral allergic nature or are caused by adverse environmental factors. According to the European Alliance of Patients with Orphan (Rare Diseases, RD), 2 out of 3 orphan nosologies are detected in early childhood. Among them 65% of cases have a severe disabling course, 50% - a worse prognosis for life, 35 % - the cause of death before the age of 1 year, 10% - at the age of 1-5 years, 12% - at the age of 5-15 years. There are more than 6,000 RD in the world, which are characterized by a wide variety of disorders and symptoms. It is impossible to cure genetic disorders completely, but it is possible to improve the quality of life and prolong the quality of life! If the child is diagnosed in time and he receives systematic therapy, the child can grow into a full member of society.

As of 01.01.2020, according to the Kharkiv Interregional Specialized Medical and Genetic Center - Center for Rare (Orphan) Diseases, patients with various RD are registered. The most common regional RD are: hyperhomocysteinemia (749 patients, including 640 children), mitochondrial dysfunction (411 patients, including 382 children), amino acid metabolism disorders (137 patients, including 108 children), phenylketonuria (107 patients, including 67 children), vitamin B deficiency (59 patients, including 38 children), cystic fibrosis (71 patients, including 45 children), Shereshevsky-Turner syndrome (40 patients, including 32 children), homocystinuria (11 patients, including 6 children), Marfan syndrome (18 patients, with 8 children).

For many RDs have no treatment, but if it exists and if started on time as being available to patients, there is a good prognosis for them to be able for normal life. The problems of patients affected by RD are related to the lack of diagnosis and timely undergoing as well as their treatment or prevention. Orphan drugs are products intended for treatment, diagnosis or prevention of RD, but for their development and marketing the industry has not been interested in yet because of their marketing reasons. Patients suffering from a RD although belonging to the vulnerable group for their specific health needs,is becoming invisible in the health care system due to their additional needs unpropertly recognized. Ethical problems faced by patients, unequal approach to health care, inappropriately specialized social services as well as therapy and rare orphan drugs unavailability.

Solving of different aspects' problems of rare patient is possible through scientific approaches in health practice, especially by Health Technology Assessment process.