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ANALYSIS OF THE NUMBER OF REGISTERED PATIENTS WITH RARE DISEASES IN SOME REGIONS OF UKRAINE

Key words: rare (orphan) diseases, registered cases, orphan patient, children, adults

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АНАЛІЗ ПОКАЗНИКІВ КІЛЬКОСТІ ЗАРЕЄСТРОВАНИХ ХВОРИХ НА РІДКІСНІ ЗАХВОРЮВАННЯ В ДЕЯКИХ РЕГІОНАХ УКРАЇНИ

Ключові слова: рідкісні (орфанні) захворювання, зареєстровані випадки, орфанні хворі, діти, дорослі

Rare diseases (RDs) is an emerging global public health priority. RDsare diseases which affect a small number of people compared to the general population and specific issues are raised in relation to their rarity. In Europe, and in Ukraine also, a disease is considered to be rare when it affects 1 person per 2 000.To date, by the orphanet data, 6 172 unique rare diseases (71.9% of which are genetic and 69.9% which are exclusively pediatric) have been discovered and new diseases are regularly described in medical literature [1]. The number of rare diseases also depends on the degree of specificity used when classifying the different entities or disorders.

Point prevalence is the most appropriate indicator for RDs as it provides a measurement of the population burden of disease, and can thus inform focused service delivery targeted at the specific needs of RD patients, pharmacoeconomic evaluation of orphan drugs, appropriate health and social service commissioning [2].

According to the Directive of EU, patients suffering from rare conditions should be entitled to the same quality of treatment as other patients; it is therefore necessary to stimulate the research, development and bringing to the market of appropriate medications by the pharmaceutical industry; incentives for the development of orphan medicinal products have been available in the United States of America since 1983 and in Japan since 1993, in Ukraine since 2014 [3, 4].

Economic and social worth of the RDs consists in their severity as chronic diseases, and often life-threatening.The other rare diseases may for instance be rareinfectious diseases, rare cancers or rare auto-immune diseases. The 30 million Europeans and 25 million Americans suffering from a rare disease coincide with six to eight per cent of thetotal population [1, 4]. The reliability of epidemiological data has improved, but remain inadequate for most of therare diseases to give firm details on the number of patients with a specific rare disease.

In 2013 the impact and burden of disease is beginning to receive more attention frominvestigators in public health and are better funded at national and European level. As noted early, activization of attention to RDs in Ukraine has had place from 2014 till now.

Basic knowledge about diseases, list of available drugs, lists of specialists or consultants specialised in a given disease, are still not widely available in Ukraine, however, it is fair to say that this problem exists in the world as a whole. WHO highlights the dissemination of information as a key issue in the field of RDs. Without information,diagnosis and treatment cannot be improved, the patients are notempowered and there is ineffective use of clinical and financial resources. Nowdays In Ukraine, about 5% of the population is diagnosed with orphan, or rare, diseases. 80% of these diseases are genetically determined [5].

Given the above, an important issue today is the understanding and analysis of the registered cases of orphan diseases in Ukraine, which will effectively plan the need for therapy and forecast the cost of it.

Aim of the investigation was to analyze the available data on the registered cases of orphan diseases among children and adults in all Ukraine's regionsand determining the share of each disease in the overall structure in terms of age group of patients.

Materials and methods

The object of the investigationaredata on the cases of rare diseases, thathave been registered in Ukraine as of January 2020 and have been obtained from the Center of Medical Statistics of the Ministry of Health on the basis of a cooperation agreementat the request of the public association «Rare Diseases of Ukraine», because the data are not publicly available. Open access data of Medical Genetics Centerhave been used in the study alsoby four orphan diseases: phenylketonuria, hypothyroidism, cystic fibrosis, adrenogenital syndrome.

The research was carried out usinginformative methods of analysis (collection and data processing), systematization and generalization, mathematical and statistical calculations.

Results and discussion

The study summarizes the regional distribution of orphan patients by all registered nosologies. There was no information from 4 regions: Chernivtsi, Odessa, Zacarpatskiy regions and the Kyivcity, which were not included to the analysis from Center of Medical Statistics of the Ministry of Health data The generalization of information on the total number of orphan patients according to the 18 nosologies that took part in the study, allowed to determine that 5 regions had been characterized by the highest total number of orphan patients: Kyiv (1 518 cases), Poltava (898), Dnipro (777), Ivano-Frankivsk (703) and Kharkiv (617) regions (Fig. 1).

Luhansk (106 cases), Lviv (118), Kherson (273), Sumy (290) and Volyn (294) regions had been characterized by the lowest number of orphan patients.

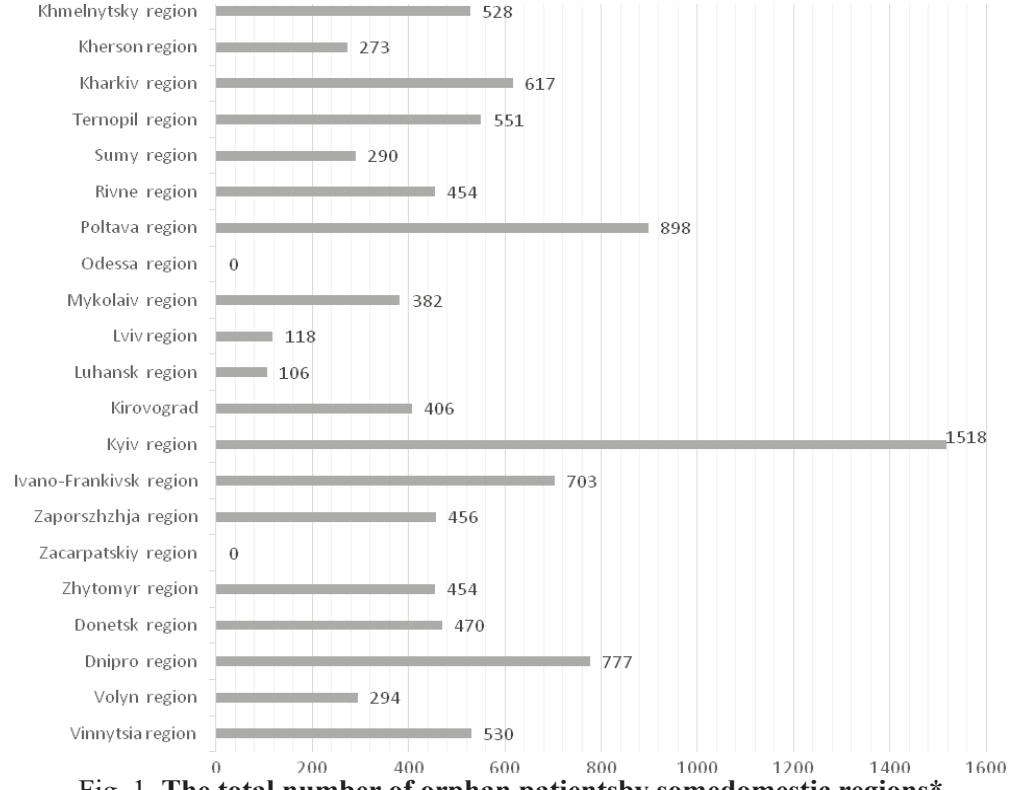

Fig. 1. **The total number of orphan patientsby somedomestic regions***

N o t e: * – by the Center of MedicalStatistics of the Ministry of Health data in 2020.

The absence of registered cases of RDs according to any of the 18 nosologies in the four regions of Ukraine, in our opinion, can not indicate the absence of orphan patients in these areas. This may indicate a delay or imperfection of the information center's data transfer algorithm, which can be a threatening phenomenon to ensure proper registration of orphan patients. It can be concluded that such significant variations in the number of patients registered in different regions are due to differences in the organization of the accounting system.

During the research it has been established that in Ukraine screening of newborns and registration of it's cases is carrying out by the Medical Genetic Center by four orphan diseases (form – N 49, table 2350 «Newborn screening programs»): phenylketonuria, hypothyroidism, cystic fibrosis, adrenogenital syndrome.

The analysis of the results of screening for 2020, which is presented in open access, is presented in Table 1 [6].

Table 1

Domestic regions with the confirmed RDs cases by «Newborn screening programs» in 2020*

N	Region	Confirmed diagnosis, cases			
		Phenyl-keto-nuria	Hypo-thyroidism	Cystic fibrosis	Adrenogenital syndrome
Ukraine		*42*	*31*	*26*	*13*
1	Volyn region	11	8	2	1
2	Dnipro region	1	3	3	2
3	Donetsk region	2	1	2	0
4	Zaporszhzhja region	0	0	2	0
5	Ivano-Frankivsk region	1	3	1	2
6	Kyiv region	2	3	1	1
7	Lviv region	6	3	3	1
8	Odessa regoin	5	3	3	2
9	Kharkiv region	4	3	3	3
10	Cherkasy region	1	1	2	0
11	Kyiv city	9	3	4	1

N o t e: * – by the Center of Medical Statistics of the Ministry of Health data in 2020.

The presented data indicate the presence of patients with phenylketonuria, hypothyroidism, cystic fibrosis, adrenogenital syndrome in 2020 in Odessa region and in Kyiv (table 1).Evaluation of newborn screening results confirms the authors' assumptions about the lack of information on registered orphan patients for 18nosologies in Zakarpattia, Odessa, Chernivtsi regions and the city of Kyiv at the time of the study, and not the absence of orphan patients.It is important to emphasize that the absence of nosology «adrenogenital syndrome» in the list of orphan diseases accounted by the Center for Medical Statistics of the Ministry of Health, needs further discussion about the feasibility of expanding the list of accounted RDs.

Accordin gto the Medical Statistics' Center of the Ministry of Health, atthetime of the study, data on register edpatientsin 18 nosologie shave bee no btained. By the 18nosologies only cases of RD in analyzed regions have been accounted. The totalnumber of patients was 10786 patients, of whom children accounted for 54% (table 2).

Among given diseases the largest proportion of patients had been registered with a diagnosis of «juvenile rheumatoid arthritis» – 2291 patients, most of whom – almost 80% – children. A rare hematological disease, hemophilia, has taken the second place in terms of the number of registered patients – 1784 patients, the fourth partwas children.

Table 2

Quantity of accounted RDs incidents in Ukraine*

N	Nosology	Total number of patients, persons		Total, persons	Share in the general structureRDs	
		children	adults		children	adults
1	Juvenile rheumatoid arthritis	1 798	493	2 291	0,78	0,22
2	Hemophilia	464	1 320	1 784	0,26	0,74
3	Nanism of various origins	1 042	313	1 355	0,77	0,23
4	Crown's disease	51	1 297	1 348	0,04	0,96
5	Phenylketonuria	728	320	1 048	0,69	0,31
6	Cystic fibrosis	517	101	618	0,84	0,16
7	Spinal muscle atrophy	269	325	594	0,45	0,55
8	Pulmonary arteryhypertension	124	334	458	0,27	0,73
9	Primary (congenital immunodeficiency)	382	58	440	0,87	0,13
10	Bullosis epidermolysis	99	119	218	0,45	0,55
11	Osteogenesisimperfecta	128	75	203	0,63	0,37
12	Tuber sclerosis	122	39	161	0,76	0,24
13	Wilson-konovalov's disease	13	86	99	0,13	0,87
14	Mucopolysaccharidosis	46	17	63	0,73	0,27
15	Idiopathic family dystonia	37	20	57	0,65	0,35
16	Gaucher disease	17	20	37	0,46	0,54
17	Pump's disease	8	0	8	1,00	0,00
18	Fabri's disease	4	0	4	1,00	0,00
	Total	5 849	4 937	10 786	0,54	0,46

N o t e: * – by the Center of Medical Statistics of the Ministry of Health data

It is important to indicate pathologies that are diagnosed mainly in children, but are also characteristic of adults.

According to the results of the analysis it has been found that the largest proportion of children patients in the age structure (children/adults) at the time of the study were characterized by the following pathologies: primary/congenital immunodeficiency (87% – the share of children in the overall structure of registered patients), cystic fibrosis (84%), juvenile rheumatoid arthritis (78%), nanism of various origins (77%), tuber sclerosis (76%), mucopolysaccharidosis (73%), phenylketonuria (69%), idiopathic family dystonia (65%) and osteogenesis imperfecta (63%).

It should be noted that among the current list of 18 rare diseases, for which there are data on the registered casesin Ukraine, 2 nosologies, or 11%, were registered only among pediatric patients – Pump's disease, Fabri's disease.

The described statistic data correlate with the available data on the quantity of cases of RDsin the world. In particular, the significant dominance of cases among children in such pathologies as cystic fibrosis, Fabri's disease, juvenile rheumatoid arthritis, Pump's disease tuber sclerosisetc. [1–4, 6–7].

Today in Ukraine there is an order of the need's for the purchase of medicinesdetermination by health care institutions and institutions that are fully or partially financed from the state and local budgets, approved by the Order of the Ministry of Health of Ukraine 11. 07. 2017 № 782, however, this Procedure does not apply to determining the need for purchase of medicines to provide patients with Gaucher disease, to plan and calculate the need for special foods for the treatment of patients with phenylketonuria, to plan and

calculate the need for test kits (reagents) for screening newborns for phenylketonuria, cystic fibrosis, for specific treatment of children, patients with cystic fibrosis, for the specific treatment of children with primary immunodeficiency, for children with dwarfism of various origins, for children with mucopolysaccharidosis, for the treatment of children with juvenile rheumatoid arthritis, to provide children with hemophilia type A or B or Willebrand's disease, blood coagulation factors and medical devices [9–11].

In fact, the need for all orphan patients registered in Ukraine is determined by the number of patients who need pharmacotherapy. It should be noted that the number of registered patients and the number of patients in need of treatment may differ, which should also be taken into account in the implementation of social and economic applied and scientific research.

At the same time, actual and up-to-date data on the prevalence of orphan diseases are essential for effective disease management from the perspective of the health care system. Therefore, the next stage of the study was to estimate the total number of orphan patients by nosology, that can be used to plan priority activities and priorities in the organization of medical and pharmaceutical support of orphan patients. The obtained results are shown in Fig. 2.

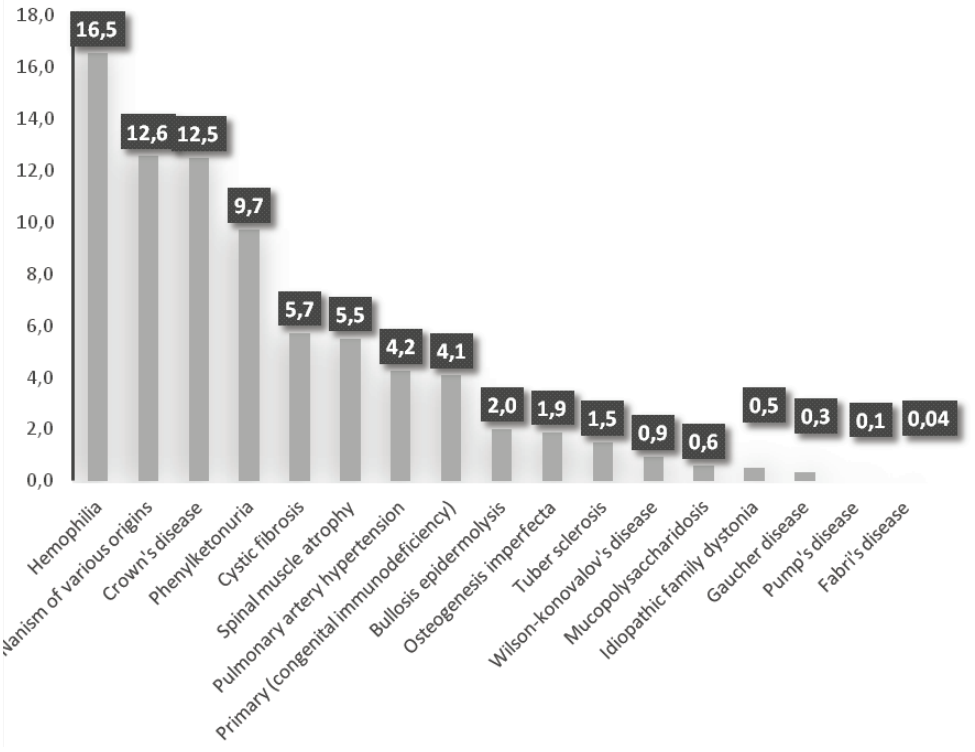

Fig. 2. **Structure of accounted rare diseases in Ukraine***

N o t e: * – by the Center of MedicalStatistics of the Ministry of Health data in 2020, share of the disease in the general structure, %.

The existing system of registration of orphan patients in Ukraine allows to analyze the whole sample of patients by individual nosologies in terms of regions of the country.

In the case of comparing the number of available cases of rare diseases by region for management purposes, it may be appropriate to compare the relative prevalence, which takes into account the number of inhabitants of the region.

The study revealed significant limitations and shortcomings in the ability to obtain information on the prevalence of orphan diseases in Ukraine in general and by region in particular. These include: lack of databases on the number of orphan patients in free access;

differences in the organization of submission and collection of information on orphan patients by regions; difficulties in obtaining summary available data and limited nosologies for which information is presented; period between the collection of information and the preparation of reporting forms.

It is fair to note that the identified limitations in the data system on orphan patients are typical for other countries. RDs present fundamentally different challenges from those of more common diseases and WHO indicates following general problems of RDs management – the small number of patients, the logistics involved in reaching widely dispersed patients, the lack of validated biomarkers and surrogate end-points, and limited clinical expertise and expert centres [4, 8].

The main positive phenomenon in the domestic system of medical and pharmaceutical care of orphan patients is the functioning of a centralized system for collecting information on the registered patients with orphan diseases. At the same time, the study showed the need and importance of improvement in the cooperation between scientists, Ministry of Health, Patients' organizations and practitioners of medicine and pharmacy, that will provide the most accurate and reliable data usage and increase the effectiveness of medical and pharmaceutical care for orphan patients.

In conclusion, it is important to emphasize the importance and necessity of presenting up-to-date data on the spread of socially significant diseases and those that are a significant burden in the health care financing system, which will contribute to effective management decisions.

Conclusions

1. Rare diseases – an emerging global public health priority. Nowdays 6 172 unique rare diseases have been discovered in the world.

2. According to the Medical Genetics Center and the Department of Statistics of the Ministry of Health the total number of patients was 10 786 patients, of whom children accounted for 54%.

3. Among given diseases the largest proportion of patients had been registered with a diagnosis of «juvenile rheumatoid arthritis» – 2 291 patients, most of whom, almost 80%, are children. Hemophilia, a rare hematological disease, hadtaken second place – 1 784 patients, where children had occupied a quarter.

4. It has been found that the largest proportion of children patients had been characterized by the following pathologies: primary/congenital immunodeficiency (87% – the share of children in the overall structure of registered patients), cystic fibrosis (84%), juvenile rheumatoid arthritis (78%), nanism of various origins (77%), tuber sclerosis (76%), mucopolysaccharidosis (73%), phenylketonuria (69%), idiopathic family dystonia (65%) and osteogenesisimperfecta (63%).

5. Among 18 rare diseases, for which there were data on the prevalence in Ukraine, 2 nosologies, or 11%, were registered only among pediatric patients Pump's diseaseand Fabri's disease.

6. It has been established thatKyiv, Poltava, Dnipro , Ivano-Frankivsk and Kharkiv regions had been characterized by the highest total number of orphan patients. Regions with the lowestregistered number of orphan patients – are Luhansk, Lviv, Kherson, Sumyand Volyn.

7. There was no information on registered RDs in Chernivtsi, Odesa, Zakarpattia regions and Kyiv city from the Center for Medical Statistics of the Ministry of Health, but it was indicated the cases of four RDs in Odessa region and Kyiv city by newborn screening program in 2020, that has shown problems in the collection, centralization and coordination of the statistic information of RDs and needs to be resolved in the future at the state and regional levels.

8. Significant limitations and shortcomings in the ability to obtain information on the registered cases of orphan diseases in Ukraine had been revealed: lack of databases on the number of orphan patients in free access; differences in the organization of submission and collection of information on orphan patients by region (lack of data from some regions of Ukraine); difficulties in obtaining summary available data and limited nosologies for which information is presented; period between the collection of information and the preparation of reporting forms. It has been shown that identified limitations in the data system on orphan patients are typical for other countries.

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A B S T R A C T

Rare (orphan) diseases (RD) are a global priority for health systems. In Europe, including Ukraine, the disease is considered rare with a prevalence of 1 patient per 2,000 population. To date, by the orphanet data, 6,172 unique rare diseases have been discovered. An important issue today is the understanding and analysis of data on the prevalence of RD in Ukraine, which will effectively plan the need for therapy and predict its cost.

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The object of the in vestigation are data of the Department of Statistics of the Ministry of Health on the cases of rare diseases, that were registered in Ukraine as of January 2020 and Medical Genetics Center's data. The study was conducted using informative methods of analysis (collection and data processing), systematization and generalization, mathematical and statistical calculations.

According to the Center's of Medical Statistics of the Ministry of Health data the total number of registered patients was 10786 patients, of whom children accounted for 54%. Among given diseases the largest proportion of patients had been registered with a diagnosis of «juvenile rheumatoid arthritis» – 2,291 patients, most of whom, almost 80%, are children. Among 18 rare diseases, 11%, were registered only among pediatric patients: Pump's disease, Fabri's disease. It has been established that Kyiv, Poltava, Dnipro, Ivano-Frankivsk and Kharkiv regions had been characterized by the highest total number of orphan patients. Regions with the lowestregistered number of orphan patients – are Luhansk, Lviv, Kherson, Sumyand Volyn. There was no information on registered RDs in Chernivtsi, Odesa, Zakarpattia regions and Kyiv city from the Center for Medical Statistics of the Ministry of Health, but it was indicated the cases of four RDs in Odessa region and Kyiv city by newborn screening program in 2020, that has shown problems in the collection, centralization and coordination of the statistic information of RDs and needs to be resolved in the future at the state and regional levels.

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АНАЛІЗ ПОКАЗНИКІВ КІЛЬКОСТІ ЗАРЕЄСТРОВАНИХ ХВОРИХ НА РІДКІСНІ ЗАХВОРЮВАННЯ В ДЕЯКИХ РЕГІОНАХ УКРАЇНИ

Ключові слова: рідкісні (орфанні) захворювання, зареєстровані випадки, орфанні хворі, діти, дорослі

А Н О Т А Ц І Я

Рідкісні (орфанні) хвороби – глобальний пріоритет систем охорони здоров'я. У Європі, зокрема і в Україні, захворювання вважають рідкісним за розповсюдженості 1 хворого на 2 000 населення. На сьогодні, за даними бази даних Orphanet, виявлено 6 172 рідкісних хвороб. Важливим питанням сьогодення визначається розуміння та аналіз даних щодо зареєстрованих випадків орфанних хвороб в Україні, що дасть змогу ефективно планувати потребу в терапії та прогнозувати її вартість.

Метою дослідження було проведення аналізу наявних даних щодо кількості зареєстрованих випадків рідкісних хвороб серед дітей та дорослих за регіонами України та визначення частки кожного захворювання у загальній структурі за віковими групами хворих.

Об'єкт дослідження – дані Центру медичної статистики МОЗ України щодо кількості зареєстрованих випадків рідкісних хвороб в Україні станом на січень 2020 року та даних Медико-генетичного Центру. Дослідження виконували зі застосуванням інформаційних методів аналізу (збір та оброблення інформації), методів систематизації та узагальнення, математичних та статистичних розрахунків.

За даними Центру медичної статистики МОЗ, загальна кількість хворих за рідкісні хвороби, що обліковуються, становила 10 786 осіб, із них діти становили 54%. Серед зазначених захворювань найбільша частка хворих зареєстрована з діагнозом «ювенільний ревматоїдний артрит» – 2 291 осіб, більшість з яких, майже 80%, діти. Серед 18 рідкісних захворювань 11% зареєстровано лише серед пацієнтів дитячого віку – хвороба Помпа, хвороба Фабрі. Встановлено, що найбільша кількість орфанних хворих характерна для Київської, Полтавської, Дніпровської Івано-Франківської та Харківської областей. Регіони з найменшою зареєстрованою кількістю орфанних пацієнтів – Луганська, Львівська, Херсонська, Сумська та Волинська. Інформації про зареєстровані випадки рідкісних хвороб у Чернівецькій, Одеській, Закарпатській областях та місті Києві від Центру медичної статистики МОЗ не надходило, але за програмою скринінгу новонароджених у 2020 році було зазначено випадки чотирьох рідкісних хвороб в Одеській області та місті Києві, що виявило проблеми зі збору, централізації та координації статистичної інформації щодо рідкісних хвороб і потребує вирішення у майбутньому на державному та регіональному рівнях.

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АНАЛИЗ ПОКАЗАТЕЛЕЙ КОЛИЧЕСТВА ЗАРЕГИСТРИРОВАННЫХ БОЛЬНЫХ РЕДКИМИ ЗАБОЛЕВАНИЯМИ В НЕКОТОРЫХ РЕГИОНАХ УКРАИНЫ

Ключевые слова: редкие (орфанные) заболевания, зарегистрированные случаи, орфанные больные, дети, взрослые

А Н Н О Т А Ц И Я

Редкие (орфанные) болезни – глобальный приоритет систем здравоохранения. В Европе, в том числе и в Украине, заболевание считается редким при распространенности 1 больной на 2 000 населения. На сегодняшний день, по данным базы данных Orphanet, выявлено 6 172 редких болезней. Важным вопросом сегодняшнего дня определяется понимание и анализ данных о распространенности орфанных болезней в Украине, что дасть возможность эффективно планировать потребность в терапии и прогнозировать ее стоимость.

Целью исследования было проведение анализа имеющихся данных о распространенности редких болезней среди детей и взрослых по регионам Украины и определение части каждого заболевания в общей структуре по возрастным группам больных.

Объект исследования – данные Центра медицинской статистики МОЗ Украины относительно зарегистрированных случаев редких болезней, зарегистрированных в Украине по состоянию на январь 2020 года. Исследование осуществляли с использованием информацинных методов анализа (сбор и обработка данных), систематизации и обобщения, математических и статистических расчетов.

По данным Центра медицинской статистики Минздрава, общее количество пациентов с редкими болезнями, по которым осуществляется учет, составило 10 786 человек, из них дети – 54%. Среди данных заболеваний наибольший удельный вес зарегистрирован у пациентов с диагнозом «ювенильный ревматоидный артрит» – 2 291 больной, большинство из которых, почти 80%, – дети. Из 18 редких болезней 11% зарегистрированы только среди педиатрических больных – болезнь Помпе, болезнь Фабри. Установлено, что наибольшее общее количество пациентов находилось на учете в Киевской, Полтавской, Днепровской, Ивано-Франковской и Харьковской областях. Регионы с наименьшим количеством зарегистрированных случаев редких болезней – Луганская, Львовская, Херсонская, Сумская и Волынская. Информации о зарегистрированных редких болезнях в Черновицкой, Одесской, Закарпатской областях и городе Киеве от Центра медицинской статистики Минздрава не было, но были указаны случаи заболевания четырьмя редкими болезнями в Одесской области и городе Киеве по программе скрининга новорожденных в 2020 году, что показало проблемы в сборе, централизации и согласовании статистической информации о редких болезнях и требует решения в будущем на государственном и региональном уровнях.

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