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BARRIERS AND OPPORTUNITIES IN PROVIDING HEALTHCARE TO PATIENTS WITH RARE DISEASES

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The aim of the study was to evaluate the aspects related to the informational capabilities of the health system for rare diseases, the self-assessment of family doctors' knowledge, the doctors and patients' perception regarding medical assistance within the national health system.

OBJECTIVES: Evaluation of barriers and opportunities regarding the management of rare diseases from the perspective of professionals in the health system; Analysis of the experience of patients and children's parents with cystic fibrosis and phenylketonuria in relation to the assistance received within the healthcare system in the Republic of Moldova.

METHODOLOGY: Type of study – cross-sectional, descriptive, analytical. The quantitative part of the research was carried out between February 1 and March 25, 2022 on a representative sample of family doctors (418 people), using questionnaires developed for this purpose. For the qualitative part of the research, was used semi-structured interview guide applied to in-depth interviews with health experts. In order to analyze the patients' experience with rare diseases, a survey was developed and the standardized validated questionnaire EQ-5D-5L was used. The data were processed statistically using the SPSS program.

RESULTS: The research identified the existence of the following barriers in providing medical assistance by family doctors to patients with rare diseases: insufficient knowledge level about rare diseases, the difficulty in identifying and diagnosing rare diseases, difficulties in identifying the patients' course with a rare disease and ensuring the continuity of it, difficulties in interaction and collaboration with specialist doctors. Family doctors' level of knowledge regarding rare diseases determined by self-assessment can be considered low. Some of the main causes are the workload that exceeds the norms recommended by the legislation and deficiencies in the professional training of doctors.

More than half of the surveyed patients declared a high satisfaction level regarding the assistance received within the health system. The low level of satisfaction correlated with the existence of some problems that patients encountered during the evolution of the disease: lack of specialists in the field, lack of information about the rare disease, lack of communication. Anxiety and depression were more frequently reported by patients compared to other symptoms related to quality of life and health status (according to the EQ-5D-5L questionnaire).

CONCLUSIONS: In the Republic of Moldova there is a fragmentation of information and a discontinuity of actions related to rare diseases. The creation of the national register of rare diseases on an electronic platform would ensure case reporting and monitoring mechanisms, would facilitate the processes of developing strategies for the management of these diseases and a good coordination of actions. Strengthening the genetics service, increasing the potential of family doctors by improving their knowledge about rare diseases is necessary.

Keywords: rare disease, public health, family medicine, continuing professional education, cystic fibrosis, phenylketonuria.

INTRODUCTION

"Rare disease" or "orphan disease" is a pathology defined by low prevalence and affects less than 5 in 10,000 people, with variations worldwide [1]. For the European region, the prevalence of rare diseases is 1 per 2000 inhabitants. According to current data, the number of rare diseases identified constitutes more than 7000 distinct nozologies, affecting 6-8% of the world's population. According to the European Commission data, 27-36 million Europeans live with rare diseases, of which 50% do not have an established diagnosis [2]. Of all rare diseases, 70% begin exclusively in childhood [3]. Delaying the diagnosis of these pathologies is associated with incorrect, ineffective therapies, negative perceptions and dissatisfaction from the services provided and from the interaction with the medical system [4].

The results of the Rare Barometer Voices research on the impact of rare diseases on daily life show that at European level, for 52% of patients and families evaluated, the rare disease has a severe (30%) or very severe (22%) impact. The results of this research, on the impact on daily life for patients in Romania, demonstrate: for 85% of patients and families evaluated, the disease has a severe (44%) or very severe (41%) impact [5]. Most people affected by rare diseases and their carers believe that social services specialists are poorly prepared to provide support [5]. One of the major problems faced by patients with rare diseases is communication deficiency. They want a better coordination of actions between health professionals involved in the management of rare diseases, about 45% of patients [5]. According to the results of numerous studies, doctors declare a low level of knowledge they possess about rare diseases and uncertainty about the reference of the patient with rare disease. The research carried out by Greulich aimed to specify the level of own knowledge reported by specialists pneumologists, internists, generalists about pulmonary manifestations of one of the rare diseases - alpha-1 antitrypsin deficiency. About 38% of German specialists and 46% of Italian specialists declared low knowledge. This gap in knowledge can be extrapolated to multiple pathologies [10]. The research conducted on a representative group of pediatricians in Australia shows that nearly a third of them (28%) said they were unprepared to assist patients with rare diseases. Less than half of pediatricians said the topic of rare diseases was sufficiently reflected during university studies (40%) or during continuing education [11].

Less than a third of the specialists surveyed in Spain have attended postgraduate continuing education courses, less than 40% have attended continuing education courses in the last 5 years, and 36.4% have attended continuing education courses on rare diseases [12]. A large proportion of primary and hospital healthcare specialists encounter problems and uncertainty about the patient's route with the established diagnosis of rare disease (66.7% vs. 62.2%) [7]. According to the results published in 2017 by Zurynski, who assessed pediatricians' experience with rare diseases, one third of them (35%) face the problem of uncertainty of reference of the patient with rare disease [6]..

The health system of the Republic of Moldova has experienced an insignificant dynamic in the process of aligning with European standards on assistance to patients with rare diseases. With the recognition of rare diseases as a major public health problem, policies in public health systems have taken on a focus on prioritizing the management of rare diseases and ensuring fairness for this category of patients, in the Republic of Moldova, the procedure for developing the national program on rare diseases control has been initiated [8]. The results of the research provide further information on existing problems and perceptions of patients with rare diseases.

RESEARCH OBJECTIVES

The purpose of the research was to evaluate the experience of patients with rare diseases and the opinion of family doctors on existing problems related to assistance to patients with rare diseases, to analyze the self-assessment of family doctors' knowledge on the subject of rare diseases and to identify existing particularities.

MATERIALS AND METHODS

A transversal, descriptive and analytical study was conducted on a representative sample of family doctors (for the opinion of professionals). The volume of the calculated sample was 418 family doctors, with the non-response rate estimated at 10%. The surveys developed for the research were collected in paper and online format using the electronic form on the Google forms platform. The distribution of research units was made randomly, in a proportional manner between the Central, South, North and Chisinau. The research included 50 patients with rare diseases: Phenicletonuria (PKU) and cystic fibrosis (CF). Patients or parents of underage children completed an online questionnaire developed for this purpose, as well as the validated standardized questionnaire EQ-5D-5L for health status. The results were analyzed using the SPSS software program.

RESULTS AND DISCUSSIONS

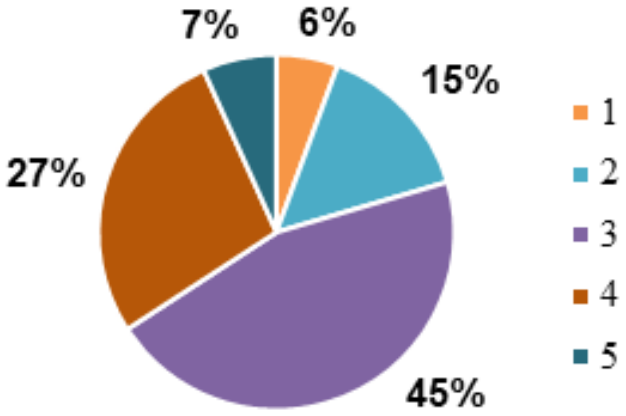
Primary medicine holds a central place in the management of rare diseases by suspecting rare pathology and monitoring patients with a large spectrum of rare pathologies, the family doctor, in this sense, has an important role to play..

The majority of respondents mentioned more than 1500 patients per sector (87%). Only 13% of family doctors have fewer than 1500 patients per sector, and 28% serve more than 2000 patients.

According to point 17, paragraph III of the *Regulation on the registration of the person with the family doctor of the medical-sanitary institution providing primary health care under mandatory health insurance* (no. 1087/721-a of 30 December 2016): "It is recommended that the practice of the family doctor should not exceed 1500 inhabitants per 1.0 family doctor function."

It was proposed to *self-assess the knowledge of family doctors* about rare diseases using a scale of values from 1 to 5 (Likert scale), 1 signifying the lowest level of knowledge and 5 signifying an excellent level of knowledge. A large proportion of respondents (44.3%) rated their knowledge of rare diseases as average, selecting 3. Over a quarter of respondents rated their knowledge as very good (26.8%) and 6.6% excellent. Only 5.5% of family doctors surveyed believe that the level of knowledge held in this field is minimal, and 14.5% declare a very low level of knowledge about rare diseases, selecting the value 1. The graphical representation of the data is shown in Figure 1.

Figure 1. Self-assessment of the level of knowledge of family doctors about rare diseases



The period of the last continuing professional development course on the subject of rare diseases was one of the questions of the questionnaire. The majority of respondents (57.5%) have not attended such courses in the last 5 years, only 17.7% of family doctors have attended such courses in the last 3-5 years. Only in the last year, about 12.7% of respondents have followed continuing professional development course from which they have received information on rare diseases, and 11% followed such a course 1-3 years ago (Figure 2).

1. The time interval over 5 years since the last continuing professional development course on rare diseases was

Figure 2. The time interval from the last continuing professional development course of family doctors on the subject of rare diseases

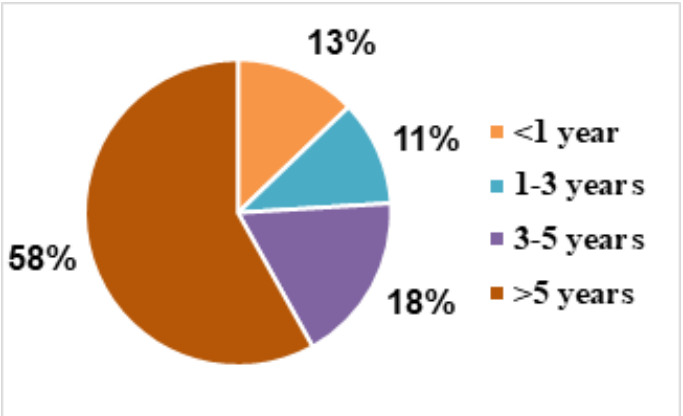
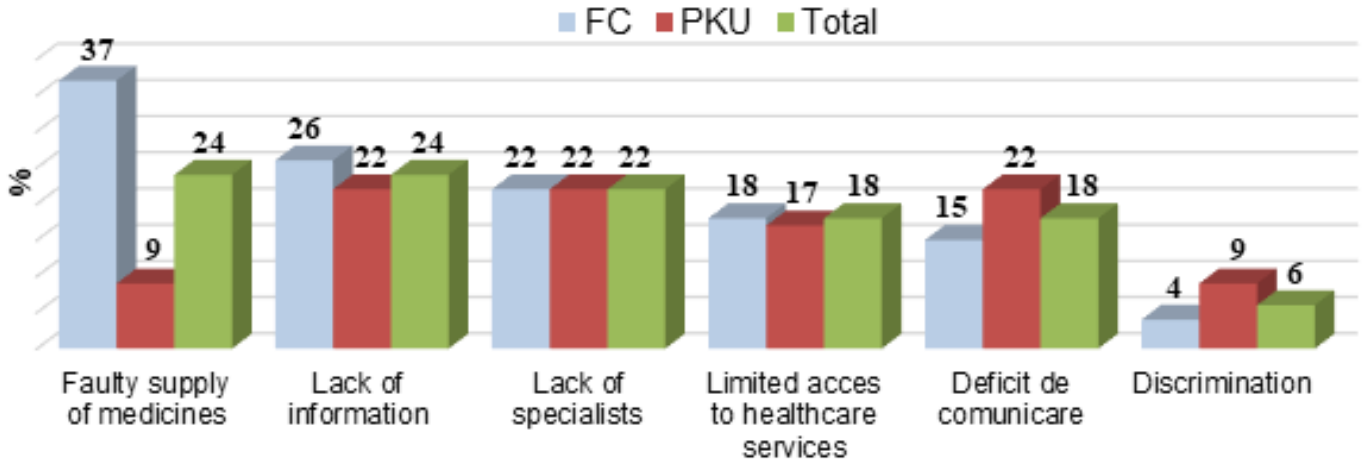


Figure 3. The problems faced by patients with CF and PKU during the course of the disease



declared by more than half of respondents, most of whom were in the more advanced age groups. Only 22% and 29% of doctors aged 41-57 and >57 years have followed continuing professional development courses in the last year or 3 years.

One third of the respondents (34%) declare they have not been able to take continuous professional training courses on the topic of rare diseases due to the lack of time or inability to leave work during the course hours. One third of respondents believe that the hours of continuing professional development on rare diseases are insufficient (33%).

Professionals' opinion

Family doctors consider the difficulty of diagnosing rare diseases (44%) and insufficient diagnostic possibilities (35%) to be the biggest challenges in providing care to patients with rare diseases. Difficulties in establishing the route and ensuring continuity of the route of patients with rare diseases are other problems of the health system mentioned, as a priority, by 31% and 30% of family doctors. Difficulties in interacting with doctors of other specialties regarding the patients' behavoiur with rare diseases is considered a priority issue by 22% of the respondents. The lack of knowledge of national clinical protocols on rare diseases is considered an acute problem by 22% of doctors. Problematic communication with patients with rare diseases is considered a priority issue by 19% of respondents.

Key elements in ensuring good management of rare diseases and optimal care for these patients were identified by doctors as: effective collaboration with specialist doctors (42% of respondents), creation of the national electronic register for rare diseases (37%), improvement of communication with patients with rare diseases (35%).

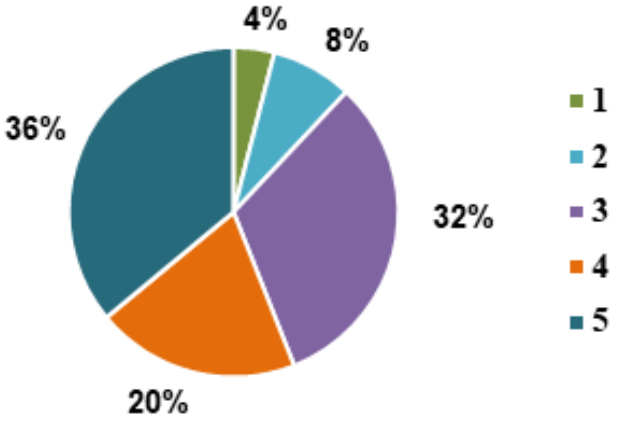
Patients' opinion

Another part of the research included patients with rare diseases - Phenylketonuria (PKU) and Cystic Fibrosis (CF), the experience of patients with rare diseases and their perceptions as a result of the assistance received within the health system has been analyzed. The average age of respondents was 13.04 (±1.21) years (14.1 years in the CF group and 11.7 years in the PKU group; p>0.05). The diagnosis of CF was established on average at the age of 7 (±2.13) months, and the time period between onset of symptoms and diagnosis was 3.9 (±1.4) months, which is not different from the results in other countries [10]

Faulty supply of medicines, lack of specialists and lack of information on pathology were the most frequently mentioned problems faced by patients and parents of sick children. Discrimination was the most rarely mentioned by respondents, with only 6% of those surveyed considering discrimination a common phenomenon (Figure 3).

Patients and parents of underage children were asked to rate with a score from 1 to 5 the degree of satisfaction with the assistance received in the health system for their rare disease, if the value of 1 corresponds to the lowest level of satisfaction, and with 5, the highest level of satisfaction was appreciated. More than half of the respondents stated a maximum and very high level of satisfaction from the assistance received under the health system (36% selected the value 5, another 20% selected the value 4). The obtained results were presented graphically in Figure 4.

Figure 4. The level of satisfaction of patients with rare diseases (CF,PKU) and parents of sick children in relation to the assistance provided within the health system



Health self-assessment

The validated standardized EQ-5D-5L questionnaire proposed to patients allowed self-assessment of health status at the time of the questionnaire, which was rated with a score from 1 to 10. The average was 7.22 (±0.288). Patients who gave a low score for satisfaction (1 or 2 points) rated their health by 6 points (± 0.966), and for patients who rated satisfaction with 4 and 5 points, the average for self-assessment of health constituted 7,6 (±0,342). About 50% of patients who rated satisfaction with 4 or 5 points have never experienced a communication deficit, and 36% - only sometimes.

The lack of information in the healthcare system affects the quality of care and the perception of satisfaction of patients, 67% of patients who rated satisfaction as low have frequently faced this problem vs. 22% of patients who gave a maximum satisfaction score.

The level of discrimination of patients has been declared low, which also reflects the satisfaction with the assistance received in the health system. About 79% of patients who reported a high level of satisfaction with the services received under the national health system have never been discriminated.

CONCLUSIONS

1. Patients and family doctors have identified common problems in the health system, with a negative impact on the quality of services provided to patients with rare diseases
2. Lack of communication, lack of information on rare diseases, limited access to healthcare are the most common problems encountered by patients with rare diseases. These issues negatively affect the satisfaction of patients in relation to the assistance received in the health system.
3. The research identified the need to strengthen family doctors' knowledge of rare diseases, with most declaring insufficient information on rare diseases and gaps in ensuring continuing professional education on the subject.
4. The workload of family doctors significantly exceeds the established norms and would be one of the causes of the existing deficiencies in providing healthcare to patients with rare diseases

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