MAIN PROBLEMS OF PATIENTS WITH RARE DISEASES ON THE TERRITORY OF THE REPUBLIC OF BULGARIA

Marina Yordanova

Department of Economics and Health Care Management, Faculty of Public Health, Medical University of Varna

ABSTRACT

Rare diseases are life-threatening and chronic conditions that occur in a limited number of patients. The problems with this group of patients are related to the lack of knowledge on the part of doctors and patients, which requires the cooperation of all individuals involved.

Rare diseases are a challenge both at European level and worldwide. Taking the issue seriously, all member countries have developed national plans and strategies to organize and plan activities to help and prevent illness. As a difficult and unpredictable area in healthcare, all interested parties are in the position of learners in the process of research and treatment. It is important to build strong relationships between medical professionals, patients and their loved ones, continuous communication and data exchange, trust in the work process to overcome a number of problems and obstacles in the field of rare diseases.

The article presents the author's survey on the attitudes of specialists in the field of rare diseases regarding the main problems of their patients. The aim of the study is to identify weaknesses in the system and to focus on measures to improve the problem-solving process. The analysis and evaluation of these attitudes should be the basis for the creation and implementation of effective changes by health authorities.

Keywords: *rare diseases, health problems, patients with rare diseases, orphan drugs, satisfaction*

INTRODUCTION

Rare diseases are life-threatening or chronic debilitating diseases with low prevalence and high complexity.

According to the generally accepted European definition, a rare disease is considered to have a prevalence of no more than 5 per 10,000 people in the EU. These are diseases that affect a small number of people compared to the general population. For this reason, the activities regarding their treatment, access and funding are related to specific issues and are predetermined by their rarity.

Impressive progress has already been made on some rare diseases, showing a constant and continuous effort in research and social solidarity.

All those affected by these diseases face similar difficulties in their quest for diagnosis, relevant information, and proper referral to qualified professionals. Specific issues regarding access to quality health care, general social and medical care, effective communication between hospitals and GPs, as well as professional and social integration and independence, are raised by all patients and their relatives.

Patients with rare diseases are characterized by greater psychological, social, economic and cultural vulnerability. Due to insufficient scientific and medical knowledge, many patients remain undiagnosed or without adequate treatment selection. These are also the people who suffer the most from the difficulties in gaining access to drug therapy and appropriate support.

A further challenge for patients with rare diseases is the impact of the disease on their loved ones. The family and social environment affect the mental peace and balance of the patient, which in turn has an impact on improving the health of the affected person. The problem arises from the lack of sufficient competence and training to deal with unforeseen situations at home, which are normal for this type of disease. The combination of financial impact on the family, stress and anxiety complicate the overall process of treatment and self-care of the patient and his relatives.

Patients do not always have access to the best treatment for their disease. It often happens that medicines are either not available on the market or

there is no standard principle for selecting patients for them. Orphan drugs are authorized for use by all members of the European Union under the central procedure. The assessment of whether they can be administered in a particular country and whether they should be included in the list of medicines for reimbursement is taken at national level. Unfortunately, there are countries where a treatment with proven effectiveness cannot be given to patients in need, and in others it is allowed to be used.

The entire process from diagnosis to the final stage of remission and follow-up is based on research, studies and proven theories in the field. Awareness of both patients and the medical team is essential for building trust, communication and improving the health of those affected. It is necessary to increase the number of scientific studies in the field, conducted both by pharmaceutical organizations to prove the effectiveness of treatment and among patients and their relatives to cover the missing niches of information about their needs. The creation of information campaigns, training seminars and self-help lessons build in the patient confidence and hope for improving the health condition.

Patients with rare diseases often feel the need to pay more attention to medical professionals and pharmaceutical organizations. Understanding the seriousness of their disease and the feeling of insecurity from the lack of sufficient information puts them in a number of dilemmas and lack of trust in the main participants in the process of their treatment.

MATERIALS AND METHODS

The methodological basis of the research is a combined approach between quantitative and qualitative methods: content analysis, statistical methods and sociological method—an in-depth interview with two groups of respondents.

The main goal is to establish the level of access to medicines for patients with rare diseases in Bulgaria based on a survey of medical specialists in the field of rare diseases.

An in-depth interview was conducted to gather information related to the possibilities for improving the access of patients with rare diseases to orphan drugs. The information was gathered through two groups of respondents: doctors, specialists in the treatment of rare diseases, and representatives of patient organizations for rare diseases from all over the country. All participants are part of the teams of centers for rare diseases in the country.

In the course of the study, 22 in-depth interviews were conducted with both groups. The interview was based on a prepared questionnaire, structured specifically for the survey, consisting of open-ended questions.

RESULTS

In summary, the analysis of the interviews (classification and ranking of awareness) is the empirical basis for assessing the problems and identifying the training needs of doctors and medical teams, patients and their families and administrative teams to improve the access to expensive medicines for rare diseases.

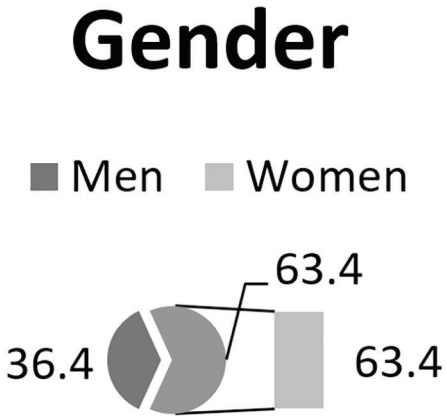

Fig. 1. Gender distribution

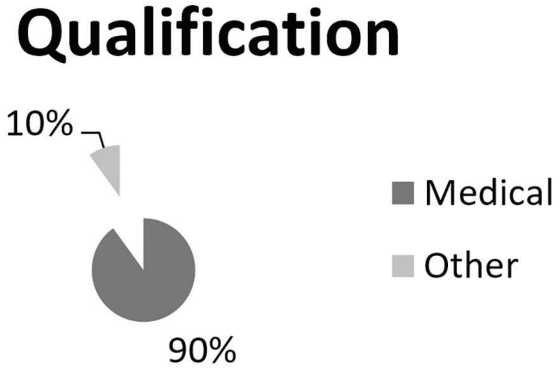

Fig. 2. Distribution by type of education

The distribution of respondents by gender is 63.6% women and 36.4% men, with an average age of 53 years. The respondents with medical education are 90.9%, and there is information about only one specialist with a different one.

Through the Likert scale and after the listed criteria, the 4 that have the highest score were derived.

It turns out that this part of the legislative framework of health care also works with poor organization of activities. Insufficient funding from the state does not allow the introduction of new therapies that will benefit the part of the population that actually needs them. Very often the treatment is quite expen-

Fig. 3. Assessment of the factors that create the greatest difficulty in the work process of patients with rare diseases

N	Criterion	Evaluation
1.	Individual / personal approaches can be used in the treatment of rare diseases	3.73
2.	**Lack of adequate legislative framework and administrative efficiency**	**4.45**
3.	Insufficient communication between institutions	3.73
4.	Lack of information about new technologies	3.27
5.	**Time and level of providing access to therapies reimbursed by the NHIF**	**3.82**
6.	**Evaluation of the effectiveness of treatment**	**3.91**
7.	Presence of frequent changes in the positive drug list of the NSCR	3.27
8.	Lack of a National Public Strategy for Improving Access to Treatment of rare diseases	2.81
9.	**Lack of public / budget funding**	**4**
10.	**Lack of sufficient public information about the rare diseases**	**4**

sive and makes it impossible for patients to cover it. At the moment on the territory of Bulgaria 350,000 people are affected by some rare disease. Only 5% of these diseases are treatable, and the remaining 95% either have no established therapy or must use maintenance therapy that requires long-term allocation of funds (2).

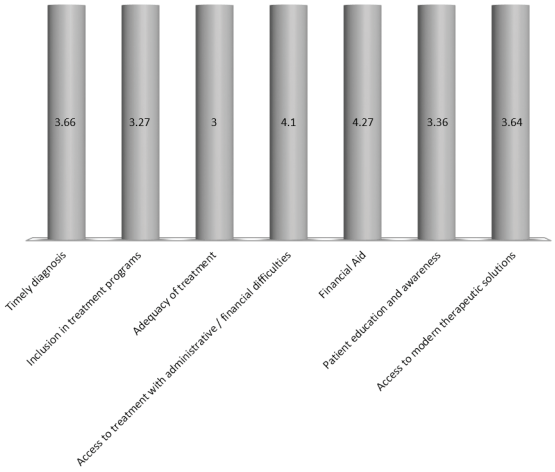

Fig. 4. Average assessment of the satisfied needs of patients with rare diseases

According to the medical specialists who participated in the study, the assessments of the level of patient satisfaction is on the positive side of the scale—over 3. The assessment is between 1 (low satisfaction) and 5 (complete satisfaction) (2).

Patients with more common rare diseases have the opportunity to participate in programs that give them the opportunity for financial relief and coverage of drugs available to Bulgaria. In order to take part, it is necessary to perform a number of tests, prove a certain severity of the disease or lack of effect of the standard therapy and fill in administrative documents. Of course, not all patients can participate due to lack of indications or a limited number of participants.

Awareness related to the level of awareness 3.36 is based on explanations and data provided by doctors to their patients. No information is available on the interest of patients and their level of health literacy beyond the data provided by the doctors.

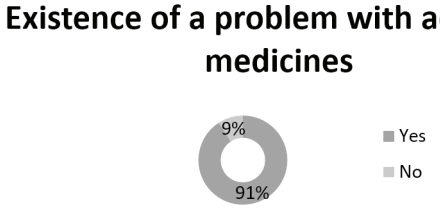

Fig. 5. Presence of a problem with access to orphan drugs

The main issue in the present study is confirmed by the results of Fig. 5. Out of all participants, 90.9% confirmed the problem of access to orphan drugs. After a detailed analysis of questions created specifically for the survey, it was determined that the problem was due mainly to the documentation related to either the registration and use of drugs in the country or the difficult process of approval of the patient applying for treatment. As the major share of the treatment for patients with rare diseases is covered by the National Health Insurance Fund, the access itself and its availability need attention (Fig. 4) (2).

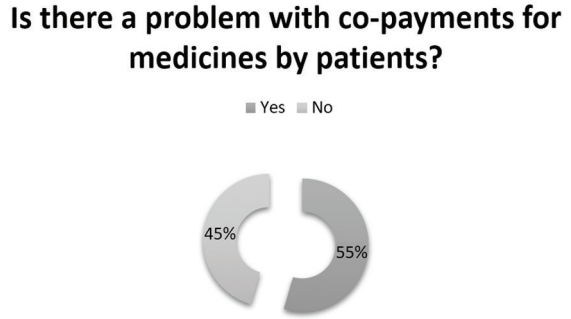

Fig. 6. Existing problems with the co-payment for medicines

According to the experts who participated in the survey, 54.5% are of the opinion that there is a problem with the additional payment for medicines, and the remaining 45.5% deny it. In recent years, some orphan drugs and drug therapies for patients with rare diseases have been paid for in full or in part by the NHIF. Another part remains out of the positive list of drugs, which creates great difficulties for patients because the price is too high or they are replaced by their generics, whose effectiveness in some cases is insufficient or not proven (Fig. 6) (2).

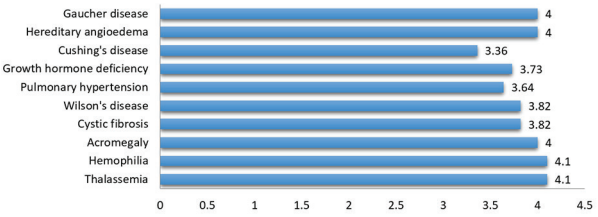

Fig. 7. Average assessment of the degree of significance for the results of the treatment

Most experts are of the opinion that the use and impact of the main drug cannot be replaced by a generic drug. On a scale from 1 to 5 (with 1 corresponding to the lack of significance, and 5 being of great importance), the respondents answered that for 9 out of 10 rare diseases the use of the original medicine is more appropriate, as well as the proven effects of it (Fig. 7) (2).

DISCUSSION

After the study, using a Likert scale from 1 to 5, with 5 being the highest point, a tendency of uniformity in the answers of the respondents was noticed. For the most part, they are unanimous on issues related to the need for change in the system—over 90% positive answer, as the main problems are related to the lack of adequate legislative framework, lack of administrative efficiency, insufficient funding and lack of information on final participants in the health system—patients (2).

The inability to participate in programs, the lack of medicines on the territory of the country, and the slow pace of document processing can lead to deterioration of their condition. The treatment process consists of two parts—the ability to use treatment methods and the availability of staff to actually apply them. The whole system suffers because the first part hinders the process.

Access to medicines for patients with rare diseases was assessed as too slow and there were cases of patients's refusal due to inability or delay in approval of treatment.

It can be characterized by two criteria—financial ability and access to available medicines. In one case we assess the ability of patients to afford additional or full cost of their therapies, and in the other case we assess the available medical resources and whether all patients suffering from a rare disease have the opportunity to find them in our country.

It is assumed that at the moment in Bulgaria there are two types of payments for medicines—partial, which are defined in the Positive List and published on the NHIF website (3), and full, where patients have to pay for all medical treatment. The second model influences the patient's choice, namely whether or not to purchase the prescribed medication. This often creates an obstacle to treatment and hinders the complete and successful work of medics, because in the absence of finances, patients either do not take the drug at all or determine the pattern of behavior and alternative treatment in order to save money.

Since 01.01.2010, medicines for rare diseases have been transferred from the budget of the Ministry of Health to the budget of the National Health Insurance Fund. This transfer takes place in view of the fact that medicines of this type are classified as home remedies (3).

By 2014, 125 drugs were allowed in the European Union of which only about 30% were allowed in countries such as Bulgaria, Romania, and Greece. The approval period is between 1 and 2.6 years (7). As of 2020, there are 2286 medicines available for orphan diseases, and in Bulgaria the National Health Insurance Fund reimburses 118 medicines for 73 rare diseases.

Over BGN 114.27 million were paid last year for the medicines for these diagnoses. However, they financed the therapy of only 6,113 patients, or the average cost of treatment per person per year was BGN 18,693 (1).

At the same time, the National Health Insurance Fund allocates over BGN 758 million for all medicines for home treatment. It has covered the therapy of over 1.6 million Bulgarians, and its average cost was BGN 470 or 40 times lower than that of rare diseases (1).

Currently, the Health Insurance Fund provides funding for 118 drugs for 73 rare diseases. The amounts allocated in previous periods are lower because the available drugs were smaller. The differences for 5 years are nearly BGN 50 million (5).

Over the years, health organizations and those involved in the problem have repeatedly made proposals to change the methodology of granting and re-

viewing the possibilities for financing the treatment of specific patients. About 6,000 people use over BGN 110 million, and the tendency is to increase. The strict specificity of the diseases provokes the need for various treatments and the opportunity to discover new methods for this.

The availability of medicationc for all types of rare diseases is impossible. The alternative of involving patients in various projects related to their treatment solves this problem. In 2019, as part of the proposal to a draft ordinance of the Minister of Health was the possibility for patients over 18 to be treated abroad, when it concerned methods that are not available in Bulgaria.

Another major problem for patients with rare diseases is the lack of sufficiently comprehensive and accessible language information. The main data are related to the nature of more common rare diseases, but it overlaps in most sources without being significantly enriched. The problem is bigger when it comes to information about side and complementary factors such as training for early educational needs of newly discovered patients, choice and availability of medication, concomitant diseases, possible alternatives for treatment in the country and Europe. It happens that those affected and their relatives have more information about the specific rare disease in a larger volume than the doctors who deal with their case. Patients who understand the causes, symptoms and impact of the disease on their daily lives cope with it more easily, accept it and learn to live normally with it. According to a study of patients with rare diseases in Germany, the most important information they seek is related to the types of research, possible treatments, and future life with the disease, the last of the symptoms, causes of the disease, and self-help and support groups (8). The whole process of treatment of the patients shall pass through obligatory administrative and legal regulation. Unfortunately, in Bulgaria each stage takes a lot of time and covers a number of unproven procedures. Despite the introduction of electronic filing of documents in a number of sectors, in the field of rare diseases it is still necessary to submit paper documents to health authorities. The approval of patients for the use of medicines paid for by the National Health Insurance Fund takes between one and two months, and for the preparation and research before that even more is allocated.

CONCLUSION

The principles and defining values of universality, access and quality healthcare, justice and solidarity are paramount for patients with rare diseases. The specific features of these diseases, such as the limited number of patients and the lack of knowledge and expertise, create a favorable environment for innovation. Raising awareness and improving communication between doctors and patients facilitates the overall process of diagnosis, treatment, and follow-up.

Despite the constant development of technology and the information age in which we live, there are still a significant number of questions about rare diseases that need to be answered. Their detection must be carried out through constant collaboration between all affected patients, their relatives, medical teams, and pharmaceutical organizations. Broad cooperation and participation between all the people that are involved in the process and representatives of national institutions is needed in order to ensure better, faster and more comprehensive treatment. The end result that all countries are fighting for is an improved quality of life for all patients with rare diseases.

Address for correspondence:
Marina Yordanova
Faculty of Public Health
Medical University of Varna
55 Marin Drinov St
9002 Varna
e-mail: yordanova250@gmail.com

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