

Short Communication

Social and welfare aspects of rare diseases

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Abstract

Social and welfare aspects of rare diseases are discussed paying attention to the activities of patients' organizations, which often become the most significant and the most important for patients ally in the struggle to improve their situation and quality of life. Giving more subjectivity to patients, their families and organizations, in particular providing opportunities to participate in decision-making, known as empowerment, is shown in a broader context.

Comprehensive and systematic solving of the problems which are associated with rare diseases is one of the elements of the prevention of social exclusion of patients and their families. Rare diseases are a particular group of diseases distinguished, not because of the clinical symptoms, but because of the low frequency of occurrence of different disorders in populations, which leads to the common difficulty in gaining access to the right, broadly understood, medical care and appropriate forms of assistance and has a high risk of isolation, discrimination and stigmatization and social rejection and exclusion of patients and their families [1]. In addition, both in patients and their family members may appear psychological problems, such as the feeling of guilt and blaming themselves for the emergence of the disease and its consequences. We have to remember that most of these diseases are inherited autosomal recessive diseases, which should be carefully explained to both patients, their parents and families by a doctor, often a clinical genetician who would provide expert, medical information. Still most rare diseases cannot be cured by the cause and no effective pharmacological methods are available for their treatment. The quality of life of patients with rare diseases, like in the case of other heavy chronic diseases, is primarily determined by the scope of the broader support available to the patients and their families.

The chronic nature of rare diseases (10 - 25% of chronic diseases in adults), their severe course, with the presence of symptoms that can be life-threatening, the occurrence in childhood (half of them is revealed shortly after birth - for example, a large part of inborn errors of metabolism- and is associated with abnormal physical, motor and mental development) leads to a number of problems, for example the need to ensure the patient round the-clock care, prevents usually the mother from keeping work. The deterioration of the material situation of the family, often poverty is associated with low levels of benefits, the need to conduct a costly long-term treatment, care and rehabilitation. Sometimes, the illness of a child leads to the breakdown of the family and a single parent raising a child faces the whole difficulty and responsibility for the fate of the child. In Poland, the provision for sick children and their caregivers (80% of these are single mothers), amounting to a few hundred Polish zloty a month, is insufficient.

Proper patient education and preparation for life in institutions for school children with special needs brings more financial and organizational burden. One example of such activities is to organize and fund the integration of disabled children with healthy peers in

the branches and classes in mainstream schools and kindergartens. In child patients the occurrence of disorders in the psycho- motor and cognitive development creates a need to find other, innovative solutions in the education system tailored to the needs of this group of pediatric patients.

We need other tools of support, especially for adult patients, such as personal assistant or the possibility of obtaining long-term care to gain access to nursing and medical care services at a reduced or lost independence. It is desirable that, depending on the patient's condition, the care was conducted in the patient's home, nursing and care institutions and care and treatment institutions, wards for the chronically ill or nursing homes.

Solving these problems should be to a vast extent based on efficient, coherent, consistently carried out program and on the system of institutional activities complemented by social initiatives and efforts, conducting charitable activities, social involvement of non-governmental organizations, foundations, etc.

The popularization of general knowledge on rare diseases and problems of patients and their families in the community is also of considerable importance, as in the slogan 'Knowledge is the first step to change'. Popularizing is helped by World Rare Disease Day, celebrated on February 29th in a leap year and on February 28th in other years. The date is not accidental, it emphasizes the similarity of the uniqueness of this day/these days in the calendar and the occurrence of rare diseases. In 2013, a leading nationwide campaign slogan of support for patients and families with rare diseases: I can see, I can hear, I act, and the campaign of "Hope. We have it in our genes" best capture the essence of the actions needed. Extensive use of media and modern means available to communicate information, greatly facilitate this task.

The impact referred to, is also artistic creation: books and films on

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difficult topics of dramas of patients and their families. The best example is nominated for an Oscar and other awards, the movie "Lorenzo's Oil" made in the USA in 1992, directed by George Miller. The film is based on a true life story of Augusto and Michaela Odone, parents of a several year-old Lorenzo. So far, a healthy baby shows the first alarming signs of a mysterious illness. The boy's parents find out that in the case of adrenoleukodystrophy (ALD) - a rare, progressive degeneration of the brain, diagnosed in a child, there is no effective treatment. The film poignantly gives a psychological portrait of the child's parents (starring Susan Sarandon and Nick Nolte), who are striving to save their son.

The facts were also the inspiration behind the book by a journalist dealing with the issues of rare diseases "Geeta Anand Drug" and the movie „Extraordinary Measures" in 2010 (on the history of John and Aileen Crowley, parents of two children with Pompe disease, who have contributed to the rapid formation of enzyme replacement therapy used in the treatment of the disease) [2].

Trying to solve, in a complex way, the problems of patients with rare and ultra-rare diseases and the problems of their families by ensuring adequate social support is an essential factor in improving the quality of their lives. Very active patients' organizations can play a large role in this.

Patients' organizations

In the last decades, internationally as well as in individual countries, including Poland, issues related to rare diseases are of particular importance. In addressing a number of specific problems and difficulties patients' organizations are actively involved [3]. Their origin is related to the situation a few decades ago, when patients with rare diseases and their families were often deprived of the opportunity to reach professionals who could provide appropriate diagnostic tests and treatment. There was also no information or research funding. There were big problems with health insurance and actually very little possibility of effective action. As a result, many patients for many years were not diagnosed and treated accordingly. This prompted the patients and their families to assist each other on the basis of established organizations to create links between patients and experts in this specific branch of medicine. The purpose of this organization was primarily to provide comprehensive support for patients and their families.

One of the most important activities in the field of helping people with rare diseases is to create a common platform for active researchers, clinicians, the pharmaceutical industry and patient organizations for research development and production of new drugs and medical products used in the treatment of patients with rare diseases [4-6].

An example of joint action of many organizations of patients with rare diseases in the United States is, celebrating its thirtieth anniversary this year (1983 - 2013) National Organization for Rare Disorders (NORD) and formed in 1986, the Genetic Alliance. The former comprises more than 2,000 patients' organizations, and the latter more than 600.

In Europe, founded in 1997, the European Organization for Rare Diseases (Eurordis) comprises 1673 patients' organizations from 43 countries. There is also freely available Internet website Orphanet - an extensive database in the field of rare diseases, taking into account the information about patients' organizations.

In Poland, the National Forum for Rare Disease Therapy - ORPHAN is an organization of more than twenty associations of patients with rare diseases. The primary objective of these organizations is to improve



Figure 1. The logo of NORD is a figure emerging from the darkness into the light (out of the darkness, into the light).

the quality of life of patients with rare diseases, efforts to improve their situation in terms of social, welfare and medical, educational and informative aspects.

These institutions and organizations also play an important role in education, have their own websites where you can find very accurate, useful and practical information on various aspects of rare diseases, for example, about the available diagnostic and treatment possibilities, and professionals working on these very specialized issues.

These organizations work together on a daily basis with doctors and other health system professionals, researchers and people associated with the pharmaceutical industry as well as the administrative and political structures of the country.

Patients' organizations can have a real impact on the efforts to fund research, orphan drug and product market, educational and popularizing knowledge actions. They can influence political and legislative decisions for example through lobbying. One example of such activities can be active participation of representatives of patients' organizations in the Committee for Orphan Medicinal Products (COMP) of the European Medicines Agency (EMA - European Medicines Agency).

Organizations of patients with rare diseases and their families and support groups also play an increasingly important role in the development of scientific research, their organization and financing. They are now among the most active in the successful impact on the health sector. They run a long struggle to notice the difficulties and problems of rare diseases and to improve the existing situation. They also have an impact on the decisions made at international and parliamentary forum, among others, by lobbying and other forms of influence.

Empowerment and rare diseases

Empowerment is a translation from English: authorization, plenipotency, giving rights, validation, giving power, giving strength, and also the opportunity to achieve your goal. The word's origin (according to the Online Etymology Dictionary and the Random House Dictionary, © Random House, Inc. 2013) is: em + power = giving force (verb) and a derivative form of a noun: empowerment. Although the first use of the word empower - in the years 1645-1655 - is associated with the works of Milton, it has gained popularity only since 1986.

The concept of empowerment has a number of meanings, definitions and interpretations in different sciences, disciplines and fields, from

psychology and philosophy of the social sciences and economics.

In the social sciences highlights are on the motivation to improve the spiritual, political, social, educational and economic capacity of individuals and communities to increase their power, especially in terms of decision-making and competence. Empowerment refers to groups and communities, which as a result of various limiting or excluding factors, are in some way disadvantaged, marginalized or stigmatized (e.g. due to disability, race, national or social origin, religion, sex, etc.) and deprived of the opportunity to take important decisions having often a big impact on their lives. In the theory of the modern marketing of service relationships, empowerment means to obtain decision-making powers by the contact personnel, and thus broadening the scope of responsibility. In more limited terms, it may mean the transfer of some power and decision-making ability directly to executive level, which in turn leads to the situation when the service recipient, the customer, in relation to the service provider, the contractor has a direct impact on everything that is related to the service and support, while a direct service contractor, with greater powers, can perform the service better, in the way more adapted to the situation, needs and customer expectations. Therefore, a deeper kind of mutual relationship follows, based on shared responsibility - more activity and intervention of staff is in fact increasing its participation in management. This results in a number of benefits, such as improving satisfaction of professional staff, their greater motivation to work efficiently and improve service quality, making their own initiative, devotion to their place of work and identification with the company, the ability to adapt to the current situation with rapidly changing environment interactions and modified expectations of the buyer. It also allows you to speed up and streamline complaint operations, which has a positive effect on customer relations. Relationships with them are stronger and make the staff maintain, enhance and expand them. This is also reflected in the improved economic and financial results of the company, where greater stability and reduced staff turnover is observed [7]. It can be concluded that empowerment is based on the belief that giving employees more decision-making powers and the possibility of action by ensuring appropriate means and resources, methods, technology, knowledge and skills, information, increase their authority and the proper motivation, will help to increase the competence and employee satisfaction, which, as a final result, leads to higher economic and financial results of the organization.

Management theory stresses the inclusion and active co-decision-making of employees about all the activities and processes of the organization, so they also take responsibility for the decisions taken. This results in an increase in awareness of incurred costs, increase in productivity, a sense of belonging and good relations among employees. This gives employees a sense of the real decision-making power and control, and strengthens their authority.

A wide, sociological and psychological meaning of the term empowerment is presented by a German sociologist Norbert Herriger [8], according to whom empowerment is:

- the ability to choose from a number of possible options, the optimal solutions for the individual and the creation of conditions to undertake alone, their own life decisions
- the ability to see their interests, needs, desires and wishes, and the ability to counter act the limiting expectations of others
- subjective experience of their life circumstances
- the ability and willingness to cope with difficulties in life

- the ability of critical thinking and rejection of aggravating habits and everyday life routine

- the ability to actively use the information, services and resources in getting help, support and networking

- measures to reduce and overcome loneliness and isolation aimed at creating cohesive communities

- to demand their rights to participation and joint action and opposition to illegal and negative behavior."

Such a broad approach to the meaning and content of the term means that it can be applied to the issue of rare diseases and orphan drugs and products, both in terms of patients' organizations and the wider "provider": a group of doctors and other professionals directly managing patients with rare diseases, scientists and researchers, both in the fields of basic and clinical medical specialists in many different sciences such as genetics and biotechnology, pharmacy, nutrition, rehabilitation, psychology, and ethics and also sociology and economics, and social welfare, or the payer of medical treatment, diagnosis and rehabilitation (eg. Polish National Health Fund) [9]. This also applies to people who have an impact on organizational, legal, legislative, educational and political activities thematically focused on patients with rare diseases and their diagnosis, treatment, rehabilitation and widely understood support, especially social.

World Bank and World Health Organization (WHO) also define the term. WHO defines empowerment as "requirements, prerequisites for health "and" partnership, active own approach of patients - a strategy to improve treatment results and quality of life of chronically ill patients".

In this concept activities are focused on the removal of formal and informal barriers and transformation of the relationship between communities and institutions. For patients with rare diseases which are chronic diseases, empowerment is a need for the patient's intensive activity in the situation when treatment is difficult, it requires coordinated action to achieve a positive effect, while the problem is not sufficiently perceived by the medical, social and political environments [10]. The first steps in this regard, were especially related to organizations of patients with chronic diseases and emerged in the 50's of the 20 century.

Knowledge and experience of patients is a valuable complement to scientific, theoretical and practical knowledge of professionals involved in this field. It should be noted that the active participation of patients in the therapeutic process should also apply to decisions taken. This brings good effects of treatment, reduces the incidence of complications, leads to healthy behaviors, and indirectly, through the actions of groups of patients it may improve access to and use of medical services. This is reflected in the views of some public health researchers who talk about a "patient - expert" and a conscious consumer" actively involved in the decisions made about their treatment.

Overall progress and technological revolution give patients access to information and ease of communication, which allows for joint action of patients' organizations around the world. It is postulated that these new and constantly expanding, opportunities served the coordinated actions of patients and professionals in the health care system. Empowerment strategy for patients with rare diseases is also on the creation of support groups and building a common relationship of patients and the medical community. This can reduce or overcome social exclusion of patients, it reduces the qualitative and quantitative differences in the obtained medical services and builds mutual bond of patients and medical staff treating them.

The groups of patients and their organizations, in relation to the concept of empowerment, can have a real impact on improving their economic, medical and social situation by taking in this regard, various initiatives and activities such as mentioned above lobbying, raising funds from different sources, etc.

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