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SOCIAL PROBLEMS OF STUDENTS WITH RARE DISEASES IN ELEMENTARY AND SECONDARY EDUCATION

Abstract: Rare diseases are complex, chronic, often degenerative and life-threatening diseases. They are characterized by a variety of diseases, symptoms that vary not only from disease to disease, but also in the disease itself.

Common characteristics of people with rare diseases are: the start of the disease in most people occurs in early childhood, they have no information about the disease itself, have a reduced quality of life, have a large psychological burden in terms of daily care for their health, difficult relationships with families and the community, stigmatization, discrimination, and difficulties in integrating into the community.

Students with rare diseases, due to the complexity of the diseases, symptomatology and treatments for their treatment and care, face health, social and educational problems in primary and secondary education.

The main goal of the research is to gain knowledge about social problems parents and their children face while the children are attending primary and secondary education.

Specific objectives of the research:

- Gaining knowledge about students with rare diseases for social problems they face during their schooling.
- Gaining information about the parents of students with rare diseases for social problems they face during their schooling.

The empirical research was conducted on the basis of application of quantitative methodology, for that purpose the survey technique was used for which two instruments were prepared: a structured questionnaire for students with rare diseases and a structured questionnaire for the student's parent.

A sample of research are students with rare diseases who attend primary and secondary education and their parents in the city of Skopje.

Keywords: Social problems, Education, Rear diseases, Students, Parents

Rare Diseases

A disease or disorder is defined as rare in Europe when it affects less than 1 in 2000. One rare disease may affect only a handful of patients in the EU, and another touch as many as 245,000.

There are more than 6000 rare diseases. On the whole, rare diseases may affect 30 million European Union citizens. 80% of rare diseases are of genetic origin, and are often chronic and life threatening¹.

¹ EURORDIS RARE DISESES EUROPE,

The existing definition of rare diseases in the European Union was adopted with the Parliamentary Action Program for Rare Diseases 1999-2003, that is, rare diseases represent a prevalence of no more than 5 per 10,000 people in the European Union. The same definition is established in the regulation of the European Commission 141/2000, and based on that it is used by the European Commission to determine the so-called orphan drugs.

Defining rare diseases is quite complex, as different countries have adopted different definitions of a rare disease, and researchers are continuously identifying new diseases or disease variants. Therefore, the epidemiology of rare diseases—including the determination of prevalence (the number of people affected at any one time), incidence (the number of new cases in a given year), and disease patterns (for example, age distribution) in a population is inexact.²

Their number of people with different types of rare diseases is constantly increasing in global and national level. However, 80% of all rare disease patients are affected by approximately 350 rare diseases. Paradoxically, although rare diseases are of low prevalence and individually rare, collectively they affect a significant proportion of the population in any country, which according to generally accepted international research is between 6% and 8%.

Rare diseases include genetic diseases, rare types of cancer, contagious tropical diseases and degenerative diseases.

Due to the low prevalence of each disease, medical expertise is rare, knowledge is scarce, care offerings inadequate and research limited.

In the area of diagnosis, treatment and rehabilitation of people with rare diseases, provision of effective drugs for people with rare diseases, quality health care differs significantly depending on the availability of expert services and health care opportunities for these people at the level of individual countries. In any case, the development of the health systems of individual countries plays a big role, in relation to their national health policies towards people with rare diseases. More non-governmental organizations for people with rare diseases are becoming active partners in the creation of health policies in terms of protection, promotion and representation of the rights of these people in several areas (health care, social care and education, etc.).

Based on EU criteria, according to the estimate that 6-8% of the population suffers from a rare disease, there should be about 189,000 patients with a rare disease in the Republic of North Macedonia.

The Commission for Rare Diseases, which operates under the Ministry of Health, in the "Register of Rare Diseases and Patients Suffering from Rare Diseases" includes only diseases that are listed in the ORFAN list of rare diseases – revised January 2020. This group doesn't include chronically debilitated or inadequately treated diseases, as well as patients with a rare secondary complication of a common disease.³

The program for the treatment of rare diseases in our country was adopted for the first time in 2009, for the treatment of two patients with the same rare disease, and in subsequent years the list of orphan drugs and treated patients with rare diseases has been constantly increasing. As of December 2020, a total of 575 people have been registered in the Register of Rare Diseases, of which approximately 185 people are provided with medicines through the Rare Disease Program. Through the Program for the treatment of rare diseases, a total of 35 drugs have been provided for the treatment of patients with rare diseases. All patients meet the criteria and have been given a positive opinion by the National Commission for Rare Diseases.⁴

² National Library of Medicine, National Center for Biotechnology Information, Profile of Rare Diseases – Rare Diseases and Orphan Products – NCBI Bookshelf (nih.gov)

³ Prevalence and incidence of rare diseases: Bibliographic date (https://www.orpha.net/orphacom/cahiers/docs/ GB/ List_of_rare_diseases_in_alphabetical order.pdf).

⁴ Програма за лекување на ретки болести во Република Северна Македонија за 2021 година, Министерство за здравство.

Characteristics of Rare Diseases

The majority of rare diseases appear immediately after birth and in early childhood, comprising 4-5% of newborns and infants (congenital anomalies and deformations, monogenetic diseases, hereditary metabolic disorders, rare tumors). Rare diseases are characterized by a progressive adverse course and very often by early death (30% die by the age of 5 years of life and an additional 12% by the age of 15 years)⁵.

From a medical point of view, a characteristic of rare diseases is the great variety of symptoms and disorders, which vary not only from disease to disease, but also within the disease itself. The same type of disease can have a very different clinical picture for sufferers, and very different symptoms can appear in one sufferer.

Rare diseases also vary in the severity of an individual disease, but on average, the life expectancy of people with this disease is very short. The impact of the disease on the duration of life is different from one disease to another, so some cause death at birth, many are fatal and degenerative, while others can be lived normally if they are timely diagnosed and properly treated.

The first symptoms of rare diseases affect sufferers at different ages. For many, they appear from birth and in childhood, but there are also those that are specific to adulthood. The third possibility is that the first symptoms appear in childhood, but the disease worsens only later in life. Also, symptoms that are similar to the symptoms of some common and well-known diseases can hide the fact that it is still a rare disease. The combination of different symptoms can also lead to a wrong diagnosis.⁶

Misdiagnosis and non-diagnosis are major obstacles to improving the quality of life of thousands of patients with rare diseases.

On a global scale, people with rare diseases comprise a significant percentage in relation to the total population, morbidity, mortality and quality of life and health of citizens. The untimely and inadequate recognition and diagnosis of a rare disease often leads to permanent damage and even death of the affected, and this greatly affects the health and status of the family. The most significant current challenges of every community and every country in the approach and handling of rare diseases refer to inadequate recognition, inadequate and unequal classification and codification, insufficient availability of diagnosis, limited therapeutic possibilities for certain types of diseases, insufficient awareness of people and their families for the type of disease, unequal and insufficient quality of health care and high cost of treatment and care, serious consequences and isolation and marginalization of those suffering from rare diseases.

Despite the fact that there is more and more talk about rare diseases, especially from the side of individuals, families and the civil sector, through the media, there is still insufficiently developed awareness among the general and professional public regarding the significance of rare diseases and their impact on health and the quality of life of individuals suffering from a certain rare disease.

Rare diseases are characterized by:

- As much as 80% of rare diseases are of genetic origin, the rest are the result of infection, allergy, environmental factors or are degenerative and proliferative.
- In a total of 50% of people with rare diseases, the first symptoms appear immediately after birth or in early childhood
- A total of 30% of children with rare diseases live less than 5 years
- For more than 95% of rare diseases there is no registered therapy
- The most common consequence of rare diseases is permanent disability (mental or physical or both)

⁵ Предлог национален план за ретки болести на Република Македонија, Национална алијанса за ретки болест на Р. Македонија, http://challenges.mk/wp-content/uploads/2013/04/Nacionalen-plan-za-retki-bolesti-broshura.pdf

NORD, Rare diseases – Friedreich ataxia (google.com)

Most often, people suffering from a certain rare disease and their families face the same or similar difficulties that originate precisely from the rarity of the disease itself:

- Unavailable diagnosis or multi-year search for a diagnosis
- Lack of information regarding the disease itself, on where to turn for help, including a lack
 of qualified experts on the disease itself.
- Lack of scientific research, lack of drugs and appropriate medical devices
- High cost of existing drugs and treatment leads to impoverishment of the family and reduction of the availability of treatment
- Social consequences: stigmatization, isolation, discrimination, reduction of professional opportunities
- Lack of quality health care: exclusion from the health system, even when a true diagnosis is made
- Inequality: facing administrative obstacles in attempts to exercise the right to treatment or social protection rights⁷

Most common rare diseases appearing in Republic of North Macedonia are, morbus goshe, hereditary angioedema, lung arterial hypertension, multiple myeloma, phenylketonuria, myelodysplastic syndrome – MDS, GIST – recidivist solnatiniv, mesulate, acromegaly GEP – NET – tyrosinemia hereditary, galactosemia, fructose intolerance, a group of deposition diseases – mucopolysaccharidoses.

Problems of Students with Rare Diseases in Primary and Secondary Education

Children with rare diseases remain, in many cases, invisible at the educational level, due to the small number of affected children, limiting the type of resources available to children and teaching personnel. This situation requires comprehensive interdisciplinary and intersectoral measures between health services and educational services to articulate a comprehensive approach focused on the clinical needs of children. Children with rare diseases may spend a longer period in hospital or outpatient therapy. This period can be even longer if the child has a specific diagnosis where drug therapy and treatment options are still being investigated. The physical effects of certain rare diseases can make attending school in person difficult for both the child and their parents. For example, some of the effects of some neuromuscular disorders may include muscle weakness, developmental delays, and secondary conditions such as respiratory difficulties. This often means that children miss school days and, in some cases, are unable to meet significant learning and curriculum outcomes.

Evidence in literature indicate that children living with rare diseases have reduced health-related quality of life and high levels of emotional difficulties (Cohen and Bieker 2010; Cole et al. 2013). The complexity of children's care needs means that living with a rare disease often has a major impact on their schooling and mastery of curriculum content. For children with rare diseases, school experiences such as absence from work, inaccessibility to educational facilities, missing physical education (sports) and academic activities have been reported.

Health-related stigma is another potentially negative consequence of a rare disease. Health-related stigma is a social process characterized by social exclusion, rejection, discrimination,

⁷ The National Organization for Rare Diseases of Serbia – NORBS, What is rare diseases, https://norbs.rs/sta-su-retke -boesti/

Berta Paz – Luirido, Francisca Negre, Begona de la Inclesia & Sebastia Verger, Influence of schooling on the health-related quality of life of children with rare diseases | Health and Quality of Life Outcomes | Full Text (biomedcentral.com), Article number 109/2020, published 18 April 2020.

Share4 Rare, How online education can help children with rare diseases | Share4Rare

devaluation, and emotional reaction as a result of one's health problem It has a negative impact on individual and family well-being (Sentenac et al. 2011)¹⁰.

For now there isn't available a specialized center for rare diseases in Macedonia, an on health institutional level most of rare disease patients are documented and guided to the University child clinic, because rare diseases mostly occur and are discovered at early ages, just after birth or until 5 years or 15 years of age.¹¹

The challenges faced by students with rare diseases in the Republic of North Macedonia refers to the lack of a systematic approach for adequate diagnosis and treatment of children with rare diseases, lack of information about the disease itself and about getting the necessary help, lack of scientific knowledge and information, social consequences that occur in relation to school, free time with the peer group, problems that are connected to their emotional and social life. A large proportion of children with rare diseases face an uncertain course of the disease; for a large number of rare diseases, there is no adequate therapy, unavailability of specific innovative disease therapies, lack of parents' ability to cover the costs of medical treatment of children with rare diseases abroad or providing adequate available medical therapy, the need for a special regime of nutrition and care; lack of adequate follow-up monitoring for individuals with a specific disease from the specific group of rare diseases, lack of national legislation regarding rare diseases in terms of their definition, classification and provision of appropriate health, social and educational services according to the individual needs of children. Most of the families of children with rare diseases face social problems related to lack of financial resources, unemployment and poverty.

Most often, students with rare diseases from primary and secondary education, in addition to health problems, depending on the type of disease, symptomatology and the changing clinical picture, most often have problems related to regularity in attending classes, transportation to school, provision of specialized services in the community, the need for educational support for mastering of the curriculum of subject programs, organizing their free time, the possibility of visiting extracurricular activities and informal education, the need for psycho-social support for students and their parents, reduced acceptance by the group of peers, occurrence of stigmatization, isolation and discrimination in certain communities.

Rare diseases not only influence the individual diagnosed with one, but also influence their families, custodians and society as whole.

Methodology

The main goal of the research is to gain knowledge about social problems parents and their children face while the children are attending primary and secondary education.

Specific objectives of the research:

- Gaining knowledge about students with rare diseases for social problems they face during their schooling.
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The empirical research was conducted on the basis of application of quantitative methodology, for that purpose the survey technique was used for which two instruments were prepared:

E.E. Adama, D. Arabiat, M.J. Foster, E. Afrira-Yamoah, K. Runions, R. Vithiatharan &a. Lin Full article: The psychosocial impact of rare diseases among children and adolescents attending mainstream schools in Western Australia (tandfonline.com), Accepted 07 Feb 2021, Published online: 22 Feb 2021.

¹¹ Предлог национален план за ретки болести на Република Македонија, Национална алијанса за ретки болест на Р. Македонија, http://challenges.mk/wp-content/uploads/2013/04/Nacionalen-plan-za-retki-bolesti-broshura.pdf

a structured questionnaire for students with rare diseases and a structured questionnaire for the student's parent.

A sample of research are 24 students with rare diseases who attend primary and secondary education and their 35 parents of the students with rare diseases.

Results

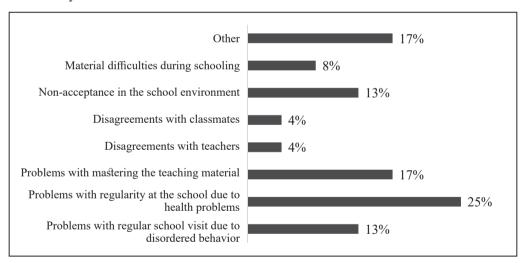
From a total of 24 students with rare diseases from primary and secondary education who were included in the survey, the following results were obtained:

- 41.6% partially feel safe in school
- 61, 9% have got attendance problems
- 50% lack of financial means for living
- 33, 3 feel discriminated, of which 44.4% feel upset about their health condition.
- A total of 45.8% of students socialize with their peers outside of school activities, and 25% of students partially socialize.

The survey covered a total of 35 parents of children with rare diseases from primary and secondary education.

Chart 1

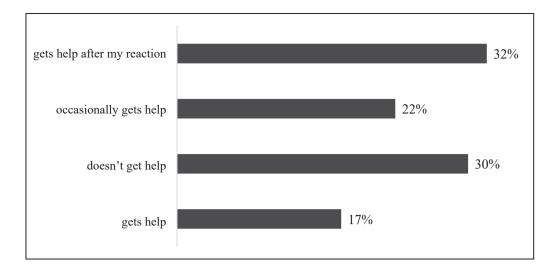
What problems are most often pointed out by parents of students with rare diseases in primary and secondary education?



To the question asked on chart number 1, a total of 25 % of the parents stated that they have problems with regular school visit due to disordered behavior, 17% have problems with mastering the teaching material and 13% with problems with regularity at the school due to health problems.

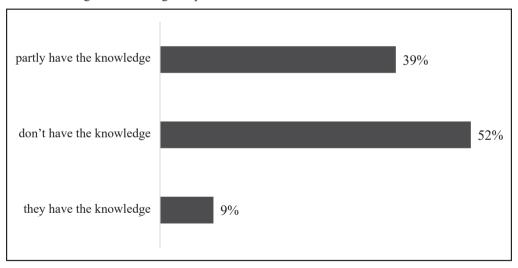
Chart 2

The support that students with rare diseases receive from the teaching staff in primary and secondary schools



A total of 32% of the surveyed parents declared that they receive help for certain problems of their children after their reaction, while a total of 30 % do not receive the necessary help.

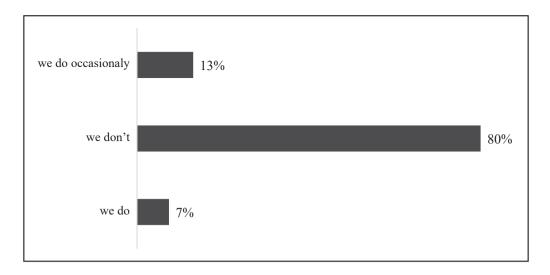
*Chart 3*The knowledge that teaching and professional staff have about students with rare diseases



This question was answered by 52% of the respondent parents who believe that they do not have basic knowledge about children with rare diseases, and 39% of them believe that they have little knowledge about children with rare diseases.

Chart 4

The psycho-social support that students receive from the professional team in primary and secondary schools



A total of 80% of respondents answered they don't receive psycho-social support from the professional teams in primary and secondary schools, while 13% receive occasional psycho-social support, and 7% of parents receive psycho-social support from the professional teams.

Conclusion

Rare diseases are diseases affecting a small number of people compared to the general population and special problems occur due to their low rate of frequency. Rare diseases are complex lifelong conditions that often require a multidisciplinary approach.

Although most inherited diseases are rare diseases, not all rare diseases have a genetic basis. There are very rare infectious diseases, as well as autoimmune diseases and rare types of cancer. A large number of children with rare diseases in our country, face health, social and educational problems. Due to the fact that for a large number of people with rare diseases, a problem arises regarding their diagnosis, thereby making it difficult to include these people in appropriate therapy.

Rare diseases are complex lifelong conditions that usually require a multidisciplinary approach. World health analyzes that also apply in our country show that 75 percent of rare diseases attack children. The first symptoms appear after birth or in early childhood. For more than 95 percent of rare diseases, there is no registered therapy or treatment protocol. The most common consequence of rare diseases is permanent disability (mental or physical, or both). Despite their mutual diversity, people with rare diseases and their families face the same or similar difficulties that originate precisely from the rarity of the disease itself. Taking into account the variable symptomatology itself, the clinical picture that constantly varies in certain rare diseases, children, i.e. students from primary and secondary schools, face a series of problems related to: diagnosis of the disease and information about the disease itself, frequent absences from the school due to the changing state of health, lack of therapy, reduced educational support for students in primary and secondary education, psycho-social support for students and their parents, poor material condition of the students' families, stigmatization, isolation and discrimination within school environments and in the community.

In terms of overcoming the social problems faced by students with rare diseases and their parents, obtaining adequate social support in the school environment and outside of it, in the community, it is necessary to include the social workers as a professional profile in primary and secondary education.

From the results obtained from the survey of the students and their parents, we received certain insights that should be directed in the area of the need for multi-sectoral action in providing health, social and educational services that will meet the individual needs of students in primary and secondary

schools, greater psycho-social support by the professional teams in schools to the students and their parents, providing greater professional support to the professional teams in solving the current problems of the students, the need to increase the awareness of the teaching and professional staff in primary and secondary schools about children with rare diseases, increasing public awareness among students and parents for acceptance and inclusion of students with rare diseases in all social streams and reducing discrimination and stigmatization towards these people.

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