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REVIEW ARTICLE
Rare diseases

Establishing a curriculum on rare diseases for medical students

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Abstract

Healthcare workers often have insufficient knowledge on rare diseases that may lead to delay in making a diagnosis and providing appropriate care. The fifth area of the Europlan entitled: "Gathering The Expertise On Rare Diseases At European Level" includes education and development and exchange of knowledge and experience in the field of rare diseases. This area was ranked high priority in rare diseases, because it determines appropriate and fast diagnosis and high level of healthcare. In the present review we summarize European Union recommendations on the implementation of education systems in member states and a suggested educational plan for the course on rare diseases for medical students. We also show the implementation of the programme in the Jagiellonian University Medical College. JRCD 2015; 2 (3): 74–76

Key words: programme, medical university, knowledge, education, rare diseases

Introduction

Rare diseases are life-threatening or significantly reducing quality of life diseases that are observed in the population less often than 5 cases per 10,000. In total, 5,000–8,000 rare diseases have been described so far. It is estimated that approx. 6–8% of people suffer from a rare disease, so in total 27–36 millions of European Union citizens have a rare disease.[1,2]

Approximately 80% of rare diseases is caused by genetic factor. Mean life expectancy of almost 60% of patients with rare diseases is significantly shorter than mean for general population. Many rare diseases are severe, complicated, and lead to chronic cachexia, whereas others, if appropriately and early diagnosed and treated, allow normal life.

Rare diseases influence all *aspects of patients' lives* – their physical and psychical abilities, behavior and cognitive abilities, and often lead to disability. Disability, in some cases severe, may lead to discrimination and limitation of educational, occupational and social chances.

Knowledge on rare diseases among health care professional

Healthcare workers often have insufficient knowledge on rare diseases that may lead to delay in making a diagnosis and providing appropriate care. Early diagnosis and monitoring require appropriate medical knowledge. However, the number of experts in rare diseases is low. Limitations of the majority of healthcare systems, diagnostic delay and inappropriate treatment lead to isolation and social exclusion of patients with rare diseases.[3,4]

According to British data, in 46% patients with rare disease the time from the first symptoms to the diagnosis is more than a year, in 20% – more than 5 years, and in 12% – more than 10 years. Approximately half of patients with rare diseases had had wrong diagnosis before they got an appropriate diagnosis, whereas almost one third of the patients had had 3 or more wrong diagnoses. Diagnostic delay leads to the treatment delay and inappropriate costs generated by the treatment of wrongly diagnosed diseases. If the diagnosis is made late, the patient is not able to make a conscious reproductive decisions.[5]

One of the major ways to reduce diagnostic delay in rare diseases is to educate healthcare workers. Although nobody expects that every doctor will know every single rare disease (from more

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than 6,000 entities described so far), it is necessary to know when rare disease should be suspected and where the patient should be referred for further diagnostics. It is estimated that 1 out of 17 Europeans will develop a rare disease. Therefore it is possible that the majority of doctors will regularly see such patients in their clinical practice.

Education on rare diseases in European Union

In 2010, recommendations for development of national plans for rare diseases were published as a part of European Project for Rare Diseases National Plans Development (EUROPLAN). These recommendations reflect European Council guidelines.[6]

EUROPLAN recommendations are focused on 7 areas. The fifth area entitled: "Gathering The Expertise On Rare Diseases At European Level" includes education and development and exchange of knowledge and experience in the field of rare diseases. This area was ranked high priority in rare diseases, because it determines appropriate and fast diagnosis and high level of healthcare.

The following actions related to education on rare diseases are included in the 5th area of EUROPLAN recommendations:

- Recommendation 5.4. The curriculum of the medical degree course includes an education package on rare diseases and on the relevant, specific provisions in the healthcare services.
- Recommendation 5.5. Training of medical doctors (general practitioners and specialists), scientists and new healthcare professionals in the field of rare diseases is supported.
- Recommendation 5.6. Continuing education programs on rare diseases are made available for health professionals.

The recommendations are based on European Council Recommendation 2009/C 151/02 in the following wording:

Recommendation 17. Gather national expertise on rare diseases and support the pooling of that expertise with European counterparts in order to support:

a) sharing of best practices on diagnostic tools and medical care as well as education and social care in the field of rare diseases;
b) adequate education and training for all health professionals to make them aware of the existence of these diseases and of resources available for their care;
c) development of medical training in fields relevant to the diagnosis and management of rare diseases, such as genetics, immunology, neurology, oncology or pediatrics;
d) development of European guidelines on diagnostic tests or population screening, while respecting national decisions and competences;
e) sharing member states' assessment reports on the therapeutic or clinical added value of orphan drugs at community level where the relevant knowledge and expertise is gathered, in order to minimize delays in access to orphan drugs for rare disease patients.[7]

According to EUROPLAN recommendations education in rare diseases should include:

- clinical aspects including diagnostic algorithms in rare diseases;
- organizational aspects including the knowledge on patients referring to the expert centers;
- communication skills including communicating the diagnosis, information on what to do in case of disease progression, emergency or terminal stage of the disease, discussion of the possibility to obtain additional care, including social care.

The education and training of specialists may be implemented in various ways, depending on their role in the treatment of patients with rare diseases. All healthcare workers should be aware of rare diseases and the related diagnostic problems as well as organization of healthcare system to provide appropriate care and manage patients' needs.

There are various activities based on our previous experience that awareness of rare diseases, including:

- easy access to the available information sources on rare diseases, e.g., a few local and regional databases such as Orphanet,
- support of medical students training on rare diseases (e.g., introduction of a specific module on rare diseases into medical syllabuses that includes specific organizational profile allowing fast diagnosis and easier access to high quality specialist knowledge),
- trainings on rare diseases for healthcare workers (e.g., on the basic features of rare diseases and understanding of medical and psychosocial need of these patients due to rarity of their ailments),
- development of medical training in the fields relevant to the diagnosis and management of rare diseases (genetics, immunology, neurology, oncology or pediatrics),
- support of development of new professions that may improve patients and family care,
- promotion of exchange and sharing of specialist knowledge between local and international centers,
- development and dissemination of evidence-based and internationally agreed guidelines and best practices on rare diseases,
- equivalent qualifications for medical specialties to unify education of healthcare workers involved in care for patients with rare diseases.

Program of Training in Rare Diseases for students of medical faculties at medical universities

The Steering Committee of the project funded by European Union titled "Development of the European Network in Orphan Cardiovascular Diseases" together with the authorities of Jagiellonian University Medical College, Faculty of Medicine decided to establish a unique educational program on rare diseases for medical students. Scientists and tutors affiliated at the Jagiellonian University as well as partner Universities and organizations took part in the project. This document includes the review of European Union recommendation on the implementation of education systems in member states and a suggested educational plan for the course on rare diseases for medical students. Educational plan is supplemented with extensive data set on respective issues that should be presented by the university teacher and description of teaching methods. The plan is supplemented with examples of case studies.

General aim of the program is to increase the awareness of rare diseases in medical students and physicians and to increase their competency and in result to improve care in patients with rare diseases.

Specific aims include education of medical students in the following topics:

- definition and epidemiology of rare diseases;
- main causes of rare diseases;
- constellation of symptoms suggesting rare diseases;
- diagnostic and therapeutic algorithms in selected rare diseases including the principles of referring a patient to reference center;
- process of asking questions and concluding in clinical settings;
- principles of orphan drugs trials and the registration of these trials;
- organization of treatment of rare diseases in Poland;
- communication of information on rare disease to the patient and his/her family;
- procedures in selected emergencies in rare diseases;
- the use of international and polish knowledge sources on rare diseases.

Target audience consists of 3-, 4-, 5-, and 6-year students of medical faculties at medical universities. It is suggested to perform the program as an optional, multidisciplinary course, i.e. the classes could be performed in different departments specialized in the treatment of rare diseases.

Implementation of the Program

In the year 2014/2015 in the framework of elective courses at the Medical Faculty, Jagiellonian University classes were held for two student groups of the 4th and 5th year with a total of 18 people. Classes lasted 30 hours and were conducted by experts from the following clinics and departments of the Medical Faculty, Jagiellonian University Medical College:

- Department of Cardiac and Vascular Diseases, Institute of Cardiology (Assoc. prof. Grzegorz Kopec, Assoc. prof. Lidia Tomkiewicz-spider, Pawel Rubiś MD, PhD, Jakub Stępniewski MD, Marcin Waligóra MD)
- Department of Medical Genetics, Polish – American Institute of Pediatrics (prof. Mirosław Bik-Multanowski)
- Department of Metabolic Diseases (Magdalena Szopa MD, PhD)
- Department of Internal Medicine and Geriatrics (prof. Thomas Grodzicki, Magdalena Strach MD, PhD, Joanna Sulicka-Grodzicka MD, PhD)
- Department of Neurology (prof. Joanna Pera)
- Department of Hematology (Assoc. Prof. Beata Piątkowska-Jakubas)

Additionally, courses for students of the scientific groups were organized.

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