

**INTRODUCTION/WPROWADZENIE**

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## **WHY THE RARE DISEASES BECOME TO BE A CHALLENGE FOR MEDICINE OF TODAY?**

Rare diseases defined as conditions affecting less than 5:10,000 population (according to the definition used in European Union), refer to limited number of patients in Poland, Europe and in the USA, but the collective number of disorders is very large. Currently evaluated by Orphanet (the portal for rare diseases and orphan drugs, [www.orpha.net](http://www.orpha.net)) the number of identified rare disease is over 7,000 and at least two new rare diseases are discovered and reported in the scientific literature each month. Poland has become a member of Orphanet since 2006 and nowadays Polish version of Orphanet website called Orphanet Poland is available and may be helpful not only for professionals (see article of Jezela et al).

Most of rare diseases are of genetic origin and cause high mortality, severe disability and impair somatic and mental development, reducing significantly quality of life of the patients, but also of their families. These diseases create a very heterogenous group of disorders come from e.g.: metabolic medicine, immunology, endocrinology, neurology, pediatric oncology, as well as stomatology.

A limited number of patients and a scarcity of relevant knowledge and expertise cause diagnostic difficulties and delayed diagnoses (see articles of Stępień et al, Magner et al). Though actually tremendous technological and scientific progress provides nowadays the great possibilities of proper diagnosis and medical care of rare diseases. Among them various modern methods of genetic testing are available, either in diagnostic or in therapeutic context (see article of Wertheim et al). Previously the problem of rare diseases was neglected because majority of patients with these diseases remained without proper diagnosis. Existence of such patients was documented by famous painters (see article of Limon).

Therefore the basic issues in rare diseases area are as follows:

- Early and correct diagnosis, including prenatal diagnosis (see article of Bekiesińska-Figatowska)
- Early and correct treatment
- Effective complex multidisciplinary care for patients and their families.

That is why the rare diseases are not only a medical problem, but also the social and psychological ones, as well as question how to: speed up scientific knowledge of rare diseases, develop new efficient diagnostic tools and orphan drugs, educate medical doctors (but also the society), organize the effective help in everyday life of the families. In a multidisciplinary team taking care of such families, many physicians – pediatricians, neonatologists, obstetricians – experts of different clinical specialisations, should cooperate with geneticists, radiologists, stomatologists, orthodontists, psychologists, and physiotherapists.

Meanwhile holistic approach of the diagnostic and therapeutic process should be recommended, focusing not only on curing an organ or one system but the entire person with his/her complicated psycho-somatic structure. It is very important not only for a child but also for the parents.

The major concern is financing all above necessary activities in order to improve the situation of these families (see articles of Borski).

The good example how to cope with the above problems is development and performing newborn screening for inborn errors of metabolism in Poland. Based on the best practice and results from the national population newborn screening coordinated by the Institute of Mother and Child in Warsaw since the sixties (initially for phenylketonuria), each Polish neonate is now investigated by tandem mass spectrometry method for around fifteen inborn errors of metabolism within the extended newborn screening programme.

Taking into consideration the fact that the early diagnosis and early treatment of patients with inborn errors of metabolism evidently prolong the patients' life and improve its quality, the problem of procreation of family with rare disease arises as a new challenge for medicine of today (see article of Wertheim et al and article of Taybert).

In order to improve the coordination and coherence of local, regional and national initiatives addressing rare diseases and cooperation between the centres, relevant national actions in the field of rare diseases should be integrated into plans or strategies for rare diseases. Such global approach (determined in the Recommendation of the Council of the European Union on an action in the field of rare diseases in 2009) based on special and combined efforts will prevent significant morbidity or avoidable premature mortality and improve the quality of life and socio-economic potential of affected persons (see article of Jezela-Stanek et al).

Many crucial initiatives in the rare diseases field are driven by the patients support groups, which play significant role, either in terms of a direct support to individuals living with the disease or in terms of collective work they carry out in order to improve conditions for the community of rare diseases patients, as a whole.

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