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Rare Diseases in Public Health

Rare diseases (RD)s are a serious public health problem and represent unique challenges in many Countries, including EU and USA.

RDs are life-threatening or chronically debilitating diseases characterized by low prevalence; this entrains limited resources as well as insufficient knowledge available for most of these individual diseases. However, RDs are numerous (possibly more than 6,000) and overall a considerable number of citizens can be affected. Special combined efforts are needed to reduce the global population effects of RDs, such as early mortality and a considerable reduction in an individual's quality of life.

There is no internationally accepted definition for RDs. In Europe, low prevalence is taken as a prevalence of less than 5 per 10 000 persons in the European Union. While this number seems small, it translates into approximately 246 000 persons in the EU within the 27 Member States. Most of the people represented by these statistics suffer from less frequently-occurring diseases affecting one in 100 000 people or less. It is estimated that between 5 000 and 8 000 distinct RDs exist today, affecting between 6% and 8% of the population in total - in other words, between 27 and 36 million people in the European Union. In USA a RD is "any disease or condition that affects less than 200,000 persons in the United States". In Japan, the legal definition of a RD is one that affects fewer than 50,000 patients in Japan, or about 1 in 2,500 people.

RDs are not limited by geographical or historical boundaries and global partnerships are rapidly expanding across the community of RDs. In this Issue four contributions are presented, starting from collaborative research efforts and related activities of RD research at the USA National Institutes of Health (NIH), through to the European Union action in the field of RDs, to a wide picture of RDs and orphan drugs in Eastern European Countries, and finally a specific approach to tackle RDs in a EU Country such as Italy.

Several collaborative efforts of the Office of RDs Research (ORDR) at the USA NIH are presented, including the RDs Clinical Research Network, the Bench-to-Bedside research program at NIH, the Genetic and RDs Information Center, the genetic test development program, and the information on clinical research studies made available through Clinical Trials.Gov. The value of an appropriate family medical history is discussed as are the provisions of the Genetic Information Non-Discrimination Act of 2008 (GINA).

In Europe the focus on RDs is a relatively new issue, made possible by undertaking a global approach to RD as a whole rather than a piecemeal approach to individual conditions. The European Commission, recognizing that a number of healthcare and knowledge issues are common to RDs in spite of the wide variety of clinical manifestation, has initiated the promotion of national plans and comprehensive strategies for improving the provision of care to RD patients. The specificities of RDs - limited number of patients and scarcity of relevant knowledge and expertise - single them out as a unique domain of very high European added-value, where the limited resources available can be efficiently pooled together. This can be achieved through, e.g., networks linking centres of expertise in different countries, as well as by making use of new information and communication technologies ("E-Health"). The Commission will build on successful existing actions, such as the previous health programme on RDs, the Research and Technological Development Framework Programmes, and the specific regulatory framework already in place to provide additional incentives for the development of 'orphan' drugs.

Eastern European countries confront the same challenges in health financing as established market economies: how to mobilise and allocate resources equitably and efficiently to satisfy a growing need and demand for health services. Reforms have reduced direct state involvement and promoted the involvement of alternative sources of funding with wider participation of the non-governmental sector. Eastern Europe is in a transition state. Many countries are already members of the EU and others are candidate members, but at this stage it is difficult for the national governments to allocate resources on the RD issue. The problems and rights of people with RDs are also insufficiently discussed. It is often not recognised that people with RDs are experts due to their personal experience. People can improve their own situation by exchanging solutions for certain problems; organisation of 'advocacy groups" via patient organisations or a patient platform can improve such patient empowerment, counteracting the social isolation.

Finally, the experience of Italy to tackle RDs is illustrated, where a number of initiatives have been undertaken to provide quality care to RD patients, in some instances anticipating the EU recommendations.

A key legal instrument has been the establishment of the national network on RDs dealing with surveillance, diagnosis, treatment and cost exemption. Other regulations facilitate access to drugs and to assistance for RD-associated disabilities. More recent initiatives have been adopted to foster research on RDs and patients' advocacy.

A further pivotal step in the development of an effective strategy has been the establishment of the National Centre for RDs; the Centre is currently developing a wide number of initiatives, both together with the national network as well as autonomous actions including research, public health consultancy and information.

While tackling issues from different standpoints, the four papers show how the unmet needs of the RDs community require additional innovative research and educational programs to reach the extensive global populations affected by the thousands of different conditions.

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