WHERE ARE WE NOW IN THE INVESTIGATION OF RARE DISEASES IN THE REPUBLIC OF MACEDONIA?

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Abstract

In Europe Rare Disease (RD) is the one which affects less than 1/2000, in the USA 1/1250, while in Japan RD is the one that affects fewer than 1/50,000 patients. EU estimates that 5–8000 distinct rare diseases affect 6–8% of the Population. The impact of rare diseases in the health systems is impressive: at least 3 million patients in the UK, 4 million in Germany, and between 27 and 36 million EU citizens. There is not a precise register for rare diseases in the Republic of Macedonia. Rare diseases are becoming increasingly important as possible targets of new forms of treatment, as a valuable source of a novel insight in fundamental laws of biology, and in the specific mechanisms of many diseases. Molecular methods have created a better diagnosis and oftentimes treatment. Rare diseases pose significant problem for the patients, since their problems are often not recognized by the medical community and shunned by the health insurance. The cumulative costs of diagnosis and treatment of rare diseases is significant for any society and oftentimes not acceptable for developing countries.

Key words: rare disease, molecular diagnosis, therapy.

Molecular medicine reshapes the map of human diseases in an unprecedented dramatic and revolutionary manner. What was thought impossible becomes trivial. What was thought unreachable becomes banal. The panacea seems within our reach. Is it really so? Not quite so. The very nature of science is that by solving one, it creates many more new problems. The human quest for knowledge is an endless task.

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Rare diseases have significant consequences for the individuals, their families and the societies [1]:

1. Rare diseases impact on the families of the affected children making many parents full-time careers.
2. Patients with rare diseases often need a team approach and treatment.
3. Rare diseases are a major public health problem because of their cumulative frequency.
Rare diseases create a particular set of challenges [1]:

1. An epidemiological challenge: there is lack of registries on the epidemiology.
2. A pharmacological challenge: there is a lack of multicentre controlled therapeutic studies.
3. An organizational challenge: there is a lack of standardized referral of patients with rare diseases in Europe.
4. A legal challenge: there is a lack of legal basis for cross border genetic diagnostics.
5. An ethical challenge: there are different priorities in different European countries.

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Rare diseases pose significant problem for the patients, since their problems are often not recognized by the medical community and shunned by the health insurance [1]. The cumulative costs of diagnosis and treatment of rare diseases is significant for any society, oftentimes bearably acceptable for developing countries.

Today is the International Day for Tolerance – 16 November 2012. We should be tolerant towards the patients with rare diseases and to treat them as the other patients.

This meeting offers new ideas about diagnosis, treatment and prevention of rare diseases.

Our Center for Genetic Engineering and Biotechnology "Georgi D. Efremov" at the Macedonian Academy of Sciences and Arts offers collaboration with all of you, joint projects and comprehensive joint efforts to improve the life of patients with rare diseases.

Here, we publish the selected proceedings of the first Meeting on Rare Disease in South Eastern Europe.

REFERENCES