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

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In some way each person is like all others
In some way like a few
In some way unique, like no other
(Origin unknown)

All humans are equal and yet so diverse.

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 lows 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 conesquences 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.

Rare diseases are becoming increasingly important as possible targets of new forms of treatment, as a valuable source of a novel insight in fundamental lows of biology, and in the specific mechanisms of many diseases [1]. Molecular methods have created 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 [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.

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Резиме

КАДЕ СЕ НАОЃАМЕ ВО ИСПИТУВАЊАТА НА РЕТКИТЕ БОЛЕСТИ ВО РЕПУБЛИКА МАКЕДОНИЈА?

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Во Европа за ретки болести се сметаат оние што афицираат помалку од 1/2.000, во САД 1/1.250, додека во Јапонија за ретка болест се смета таа која афицира помалку од 1/50.000 пациенти. Процена е дека во Европа 5.000–8.000 различни ретки болести афицираат 6–8% од вкупната популација. Значењето на ретките болести за здравствениот систем е импресивно: најмалку 3 милиони пациенти во Велика Британија, 4 милиони во Германија и најмалку 27–36 милиони се граѓани на Европската унија. Во Република Македонија не постои прецизен регистар на ретки болести. Ретките болести стануваат сè повеќе значајни како можен таргет за нови форми на третман, како важен извор на нови сознанија во фундаменталните закони на биологијата како и за специфични механизми за многу болести. Молекуларните методи ја подобрија дијагностиката и третманот. Ретките болести претставуваат значаен проблем за пациентите, бидејќи нивните проблеми не се препознаени од медицинската заедница и се игнорирани од здравственото осигурување. Кумулативните трошоци за дијагноза и третман на ретките болести се значајни за секое општество, често тешко подносливо за земјите во развој.

Клучни зборови: ретки болести, молекуларна дијагноза, терапија.