



CURRENT LEGISLATION AND RECOMMENDED PROCEDURES

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The author elaborates on the requirements for the creation of a national plan for rare disease on the basis of three fundamental parts of the problem: Centres of expertise – European reference network – European registers of rare diseases. He presents some positive examples of the latest developments and points out the difficulties of creation of registers of rare diseases in Europe, as well as in the SR. He emphasizes the role and engagement of the Ministry of Health in the implementation of partial tasks in the process of creation of the national plan and stresses the need to concentrate on a small number of fundamental tasks.

Keywords: *rare diseases - centres of expertise – European reference networks – registers of rare diseases – national plan for rare disease*

The creation process of the National Plan for Rare Disease in the Slovak Republic (NP RD SR) follows the government approved document of National Strategy for Rare Disease Patient Health Care Development for years 2012 – 2013 (RD strategy). In the creation process of the RD strategy, we emphasized that a detailed analysis of the current state and a practical viable agreement on the implementation of National Plan for Rare Disease in the Slovak Republic into practice are necessary. A number of problems, which are a part of European initiatives and will form the content of the National Plan for Rare Disease in the Slovak Republic, follow from this triangle: **Centres of expertise – European reference network – European registers of rare diseases.**

Centres of expertise (CE) are the cornerstone for the diagnosis, treatment, and management of rare disease patients. As there are many types of rare diseases (more than 6000), Europe needs a big number of Centres of expertise to cover all special needs. As far as this is concerned, none of the countries is self-sufficient. Thus, both

big and small countries are dependent on international cooperation. At the national level we have to provide accessible diagnostics and treatment for patients with all types of rare diseases. It is possible to establish only a few quality Centres of expertise. It is not a coincidence that EUCERD drew the first Recommendation on quality criteria for centres of expertise; and application of these criteria in practice is of great importance to the SR. In Slovak health care practice, there are several centres dealing with rare diseases which might fulfil EUCERD quality criteria. Adoption of national criteria for Centres of expertise and their application in practice should be the first and easily manageable activity regarding this issue.

European reference networks (ERN) should create conditions for a meaningful flow of information and contacts necessary for solving problems of concrete patients. Today, a considerable number of specialized international groups of Centres of expertise and specialists exist. Workplaces and specialists from the SR also participate in these groups. However, the involvement in European reference networks must be systematic and it must be based on sustainable financing. Regarding European reference networks, EUCERD drew a recommendation on European Reference Networks for rare diseases. Creation of a quality European reference networks is a long-term process. A positive example of the impact of international cooperation on newborns screening in the SR will be introduced by professor Dluholucký using his experience from the Slovak Newborn Screening Centre.

The third essential section is constituted by registers of rare disease patients. The discussion about the common principles of register creation and data collection has only started in EUCERD. The importance of the issue follows from the general need to know frequencies of particular rare diseases in order to plan centres of expertise development and deal with European reference networks. The creation of a rare disease register in the SR is a subject of researching of what options the current registers of diseases and data collection in National health information centre NHIC environment allow. This process will be difficult for the SR as well as for the Europe; furthermore, it overlaps with e-Health issues.

Three other important activities determining rare disease patient health care development are reflected in these three essential pillars.

Patient organizations in the area of rare diseases associated mostly in EURORDIS have become a driving force not only for their own patients' support, but also for the formation of the health and social care itself, and for the major support of rare disease research. This is proved by the fact that this very conference was organized.

The reference portal on rare diseases – ORPHANET – is the most extensive and one of the frequently used sources of information on rare diseases also in the SR.

Financing of rare disease patient health care in the SR is provided by health insurance companies. As approximately 80% of rare disease patients have genetic aetiology, and medical genetics is developing and being introduced to practice very rapidly, while the current list of medical services is absolutely dissatisfactory, we would like to appreciate the constructive dialogue heading towards sustainable financing of genetic testing.

rare disease strategy was drawn by a small workgroup at the Ministry of Health of the SR in 2012. Creation of National Plan for Rare Disease in the Slovak Republic is, however, a much more extensive project, and an inter-branch workgroup for rare disease was charged with it. Yet, National Plan for Rare Disease in the Slovak Republic

is not only a goal that needs to be reached by the end of 2013; most of all, it is a means of long-term motivation for the development of the health care of this group of patients. Our experience in the process of rare disease strategy creation shows an essential importance of the engagement of concrete Ministry of Health employees in order for the activities to concentrate only on several decisive issues and on creating a possibility to revise the process of National Plan for Rare Disease in the Slovak Republic implementation regularly. This role can be played only by a permanent workgroup at the Ministry of Health or by an internal body of the Ministry of Health focused on the already mentioned issues.

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TVORBA NÁRODNÉHO PLÁNU STAROSTLIVOSTI O PACIENTOV SO ZRIEDKAVÝMI CHOROBAMI, SÚČASNÁ LEGISLATÍVA A ODPORÚČANÉ POSTUPY

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Autor rozvíja požiadavky na tvorbu Národného plánu starostlivosti o pacientov so zriedkavými chorobami na báze troch základných častí problému: Centrá expertízy – Európska referenčná sieť – Európske registre zriedkavých chorôb. Uvádza niektoré pozitívne príklady rozvoja z poslednej doby. Poukazuje na zložitosť vytvárania registrov zriedkavých chorôb tak v Európe, ako aj v SR.

V procese tvorby Národného plánu zdôrazňuje úlohu a angažovanosť Ministerstva zdravotníctva aj pre implementáciu čiastkových úloh a taktiež zdôrazňuje potrebu sústrediť sa na zopár základných úloh.

Acta Fac. Pharm. Univ. Comen. 2013, Suppl VIII.