National Strategy for the Development of Health Care for Patients with Rare Diseases for the Years 2012-2013

CONTENTS

1. Introduction

The aim of the draft of the National Strategy for the development of health care for patients with rare diseases for the years 2012-2013 in the Slovak Republic (SR NSRD) is to prepare the development of NPRD SR and support its implementation into health and social care.

In Slovakia, there is as yet no uniform concept of diagnosis, treatment and overall care of patients with RD. NSRD SR builds on the existing development in EU and relevant recommendations. The current deficit in the development in Slovakia should be removed by prompt fulfilment of the recommendations which are defined in EUROPLANE, by joining Slovakia to the work of EUCERD and to other most significant European activities.
The existing European initiatives identified several key problems:

- Insufficient identification of the scope of issues of very heterogeneous diseases, insufficient identification of RD within the International Classification of Diseases, version 10 (hereinafter referred to only as "ICD-10");
- Serious deficiencies in diagnosing and treatment, due to the extreme rarity,
- Incorrect or delayed diagnosis often leads to irreversible damage to health or to the death of a patient,
- Uneven and insufficient quality of provided services,
- Very limited selection of drugs for the treatment of rare diseases,
- Research of RD is demanding, in the area of research of drugs for RD it is economically inefficient,
- Deficiencies in data collection at national and regional levels,
- Lack of awareness of these diseases among professional as well as lay public,
- Due to the chronic and often progressive course of these diseases, deficiencies in social care and legal assistance are common.

Development of national plans for RD should contribute to a substantial improvement in the overall care of patients with RD in individual Member States and to a more comprehensive improvement of the situation in EU.

2. Characteristics of rare diseases

Within EU, a rare disease is considered such a disease which occurs in less than 5 persons in 10,000, that is 1 patient per 2,000 or more people. **It is estimated that in total they affect6 – 8 % of the population in the developed countries, which means within the countries of the European Union an affliction of 27 – 36 million people.** The aetiology is very heterogeneous, in roughly 80% these are hereditary diseases. Other RD are conditioned by infections, immune disorders, allergies, etc. They usually have a significant impact on quality of life and social inclusion or they threaten the life of the individual.

RD diagnosing is often very complicated also because within medical expertise there are more than 6,000 different rare diseases. The list of RD and other detailed information for the diagnosis, medical care and treatment are listed on the ORPHANET portal (www.orpha.net).

Many RD become apparent early after birth and collectively they occur in up to 4% of newborn babies (congenital developmental defects, inherited metabolic disorders, genetically determined diseases, children's tumours). About one-third of patients suffering from severe rare diseases do not live past the age of five. Many of these diseases are associated with motor and mental deficits, sensory impairment, dysmorphias or threats to life. More than 50% of RD may start to manifest during adolescence. For a large proportion of patients the aetiology of their disease remains undetected. In the case of faulty or delayed diagnosis irreversible damage to health occurs, even despite the possibility of treatment for some patients. The consequence is traumatism of the family and distrust of the health care system.

3. Initiatives and intentions of the European Union (hereinafter "EU") in the issue of RD and their transfer to the environment of the Slovak Republic

3.1 Recommendations within EU

a. In the White Paper of the European Commission "Together for Health: A Strategic Approach for the EU 2008 - 2013", adopted on 23 October 2007, by which the EU strategy in the field of health is developed, RD was ranked among the priorities for acceptance of measures in the organization of health care.

b. The European Commission on 11 November 2008 adopted the "Communication of the Commission to the European Parliament, the Council of Europe, the European Economic and Social Committee and the Committee of Regions about RD: "European Challenges" (hereinafter only the "Communication of the Commission") and on 8 June 2009 the Council of Europe adopted the recommendation on activities for the improvement of the health care for patients with rare diseases (Council Recommendation of 8 June 2009 on action in the field of rare diseases)
(http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:SK:PDF)

c. The European Parliament and the Council of Europe adopted Directive of the European Parliament and of the Council 2011/24/EU of 9 March 2011 on the application of patients´ rights in cross-border healthcare.
(http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2011:088:0045:0065:SK:PDF)

3.2. The general objectives of EU in improving the care of patients with RD

The general objective of the EU in the area of RD is:
a. to improve identification of RD
b. to support the development of health policy and care of patients with RD
c. to develop European cooperation, coordination and control in this area

Recommended are:

- support to improve the access to diagnosis, care and treatment for patients with RD, for which should also be used a compilation of National Plans for rare diseases in individual EU Member States on the basis of equal treatment and solidarity in the whole EU ,
- introduction of an adequate definition, codification and list of RD, inclusion of RD in the innovated International Classification of Diseases, ICD 11,
- support for the research and treatment of RD,
- establishment of centres of expertise and coordination of the development of European reference networks for RD,
- gathering of expertise on RD at a European level,
- strengthening of cooperation with patients´ organizations for RD,
- to ensure the development of activities in the field of RD and sustainability of the health care system with regard to patients with RD.

For more information see (http://ec.europa.eu/health/ph_threats/non_com/rare_diseases_en.htm).

3.3 EUCERD - The European Union Committee of Experts on Rare Diseases - The Committee of Experts of the European Union for RD

The European Commission established by its Decision of 30.11.2009 a Committee of Experts of the European Union for Rare Diseases. This Committee replaces the Working Group of the European Union for Rare Diseases (Rare Disease Task Force), which was established by a Commission Decision of 25 February 2004, by which is adopted a working plan for 2004 for the implementation of the programme of activities of the Community in the area of public health (2003 to 2008), including the annual working programme for grants (2004/192/ES).

The national representative of Slovakia in EUCERD informs the Ministry of Health of SR on ongoing activities in the area of RD in the European Union and on the negotiations of the EUCERD Committee by means of a working group of the Ministry of Health of SR for RD (http://www.eucerd.eu).

3.4 Specialized workplaces for RD, centres of expertise for RD and European Reference Network for RD

In the RD group should be provided the highest quality and most effective diagnosing, treatment and management of patients through centralized health care and therefore the origin of so-called Centres of Expertise for RD is supported.

The Directive of the European Parliament and of the Council 2011/24/EU of 9 March 2011 on the application of patients' rights in cross-border healthcare assumes the creation of a reference network which will enable a full-value flow of information and contacts for troubleshooting of the individual patient with a rare disease. The European reference network for rare diseases is based on the so-called Centres of Expertise of RD / RD groups.

The creation of a European Reference Network (ERN) is complicated because of the basic characteristics of RD. There are a lot of rare diseases, there are few experts and few completely developed centres in Europe. EUCERD developed and approved as the first important document the "Recommendation on qualitative criteria for national centres of expertise (CE) for rare diseases", in October 2011.

Acceptance of the document in the EU Member States as well as in SR makes possible compliance in the characteristics, purposes and qualitative parameters of such medical workplaces. (http://nestor.orpha.net/EUCERD/upload/file/EUCERDRecommendationCE.pdf).

3.5 EUROPLAN - The European Project for Rare Diseases National Plans Development

The European Project for Rare Diseases National Plans Development (EUROPLAN) was a three-year project of the programme of the European Union in the area of Public Health (2008 - 2011) to support the development and implementation of plans and strategies for RD on a national level. The result is a guide for the development of a national strategy or national plan of development of care for patients with RD. The main objective of the project is to search for tools to implement the intentions into the legislation of EU Member States in light of the extreme variability of the health systems. Slovakia will join the EUROPLAN activities and it will use possible assistance.
(http://www.europlanproject.eu/public/contenuti/files/Guidance_Doc_EUROPLAN_2010060_1_final.pdf)

3.6. ORPHANET - The reference portal for information on rare diseases and orphan drugs – the European information portal on RD and on drugs for RD

ORPHANET is currently the largest and most widely used reference portal on RD and drugs for RD. It is developing very dynamically and its aim is to assist in the diagnosing, care and treatment of this group of diseases. It is organized by the European consortium of 40 countries and is coordinated by a French team of specialists. On the national level are created teams which are responsible for the collection of information in a national framework pursuant to a unifying scheme.

Orphanet provides data on RD, on diagnostic workplaces, research and development in the RD area, as well as on related information. It is a task for professionals and sponsors so we support the systematic usage of this database.
(http://www.orpha.net/consor/cgi-bin/index.php)

3.7 EURORDIS - European Organisation for Rare Diseases - the European NGO alliance of patient organizations and individuals

EURORDIS is a non-governmental alliance of supporting patients´ organizations and individuals active in the field of rare diseases which is aimed at the improvement

of the quality of life of patients in Europe. Currently it represents more than 500 patients´ organizations in more than 48 countries and it covers a professional space of more than 4,000 RD. It is a presentation of the voices of 30 million patients across Europe. EURORDIS has a significant impact in the support of research, drug development, and improvement of the conditions of life of patients with RD. It significantly participates in placing issues of patients with RD in priority places within European health policy and European research programmes. (http://www.eurordis.org).

3.8 The RD working group of the Ministry of Health SR

In 2011 the Ministry of Health of the Slovak Republic established an RD working group for with the aim of professional assistance and support in the fulfilment of the EU objectives in the area of RD in the Slovak Republic. The main task of the working group is the development of NSRD SR and support for the implementation of European activities into the health and social system in the Slovak Republic.

The working group for RD should then be transformed into an independent expert advisory body of the Ministry of Health of the Slovak Republic – the Commission for RD.
Another objective of the working group is to initiate the formation and creation of a Ministry of Health inter-departmental working group whose task will be to prepare and professionally guarantee NPRD SR.

4. The current state of health care for patients with RD in the Slovak Republic

A precondition of the quality preparation of NPRD SR is a detailed analysis of the current situation. The main objectives and measures will be prepared pursuant to this analysis.

4.1. Post-neonatal RD screening

Post-neonatal screening in SR is performed for: phenylketonuria, cystic fibrosis, congenital adrenal hyperplasia and congenital hypothyroidism. The strategy envisages the evaluation and review of screening and in the case of deciding on the suitability of the introduction of screening for other congenital metabolic diseases it assumes a procedure pursuant to the efficiency of screening and curability of the given disease.

4.2 Specialized RD workplaces, RD centres of expertise

So far in the Slovak Republic, attention has not been paid to the conceptual view on the provision of health care to patients with RD. Diagnosing, treatment and curing of patients with RD takes place at workplaces across all medical specializations. For some RD or RD groups there are now created specialized outpatient units and specialized centres. According to the newly approved EUCERD recommendations

(Recommendations on qualitative criteria for national centres of expertise (CE) for rare diseases", of October 2011), however, no workplace in the Slovak Republic would so far fulfil all the criteria required for such centres. Several workplaces in the Slovak Republic, however, could be successful in this in future.

4.3. Drugs for Rare Diseases - "Orphan drugs" and medical devices for patients with RD

The drugs for RD, mainly due to large investments into research, development and production, are very expensive. Due to the specific European legislation (Regulation (EC) no. 141/2000 and Commission Regulation (EC) no. 847/2000) which is valid in every EU Member State, stimulation for the development of drugs for RD group is taking place. The legislation is under the responsibility of the European Medicines Agency (EMEA; www.ema.europa.eu) and its specialized Committee for Orphan Medicinal Products (COMP).
(www.ema.europa.eu/htms/general/contacts/COMP/COMP.html).
Ensuring the availability of medicines for rare diseases in the individual EU Member States is the responsibility of the Member States themselves. In the Slovak Republic the drugs are available on the market only after categorization, or there are possibilities of individual import.

One part of RD patients with requires the provision of special medical devices which help compensate for the reduced quality of their life and whose availability should be ensured.

4.4. Organizations to support RD patients in the Slovak Republic

The EURORDIS members are from Slovakia: The Slovak Alliance of Rare Diseases (RD Alliance), The Slovak Association of cystic fibrosis, DebRA SR (The Dystrophic Epidermolysis Bullosa Research Association – a non-profit association uniting patients with the severe skin blistering disease Epidermolysis bullosa - disease of "butterfly´s wings") and ZOGO (The Association of Sporadic Genetic Diseases o.z.), the Pulmonary Hypertension Association. In addition to the above mentioned associations, there are also other patient organizations in Slovakia dedicated to individual RD (The Slovak Marfan Association, The Muscular Dystrophy Organization - Duchenne muscular dystrophy, The Huntington's Disease Society, The Slovak Society for Spina Bifida and Hydrocephalus, Living with Lupus o.z., The National Association for PKU, The Friends of Salty Children Civic Association).

5. Financing of care for RD patients

The diagnostic process of many RD is usually multistage and time-consuming and demanding on the expertise of highly educated specialists. The treatment of some RD is expensive. We do not have available data about the costs related with the treatment of RD. This is partly because only about 250 out of a total number of more than 6,000 RD are coded in MKCH-10, which prevents statistical monitoring of these diseases in the national statistics.

Currently we do not have data on the resources spent by health insurance companies under other diagnoses for RD care. According to the recommendations of the Council of Europe as well as from the patient organizations - EURORDIS (www.eurordis.org), a reduction of the costs of treatment of complications of late-diagnosed RD can be achieved particularly by the centralization of care. Those workplaces (centres of RD expertise) which have interdisciplinary capacity, sufficient experiences with early diagnosing and prolonged treatment of RD are the guarantee of providing quality and at the same effective care.

For these reasons it is not currently possible to estimate a medium-term prognosis of the costs for the improvement of health care for RD patients. These calculations will be available only based on the MKCH-10 statistics, which will include all RD.

6. The objectives of the national strategy and proposed measures

The national strategy for the development of health care for patients with rare diseases in the Slovak Republic for the years 2012-2013 (NSRD SR) aims to prepare the development of NPRD SR and the support of its implementation into health and social care.

I.

6.1. Improvement of general public awareness of RD issues

- To present RD issues on the Ministry of Health SR portal, including links to the most important European initiatives and professional websites.
- To encourage patients´ organizations in the issues of increasing the attention of the Slovak public to patients suffering from RD and to their families.
- To encourage activities in individual professional societies of the Slovak Medical Association in favour of greater space for RD issues.
- To present Orphanet activities and support the active participation of the Slovak Republic in its activities. To present activities of European institutions in the RD issues and in the case of commitments of the Slovak Republic to request timely implementation and control of the fulfilment of the commitments
- To support working meetings, conferences and other similar professional activities thematically focused on RD issues in the period of work on NPRD SR in 2012 ¬ 2013.
- To prepare and implement a National Conference on RD issues in 2013.
- To prepare and implement a media activity within the "Rare Disease Day – The International Day of Rare Diseases" in 2012 - 2013.
- To encourage media coverage of the issues of RD also through the Ministry of Health of the Slovak Republic.

6.2. Education of professional and general public concerning RD

- To extend the scope of RD issues in curricula of medical specialisations involved in postgraduate education.

- To encourage the usage of e-learning environment also for presentation in the RD area .
- To initiate the support of education of the professional public also in the undergraduate education of health professionals.
- To support the development of a Slovak branch of Orphanet.
- To encourage the education of the lay public through medical educational training.

6.3 Improvement of RD diagnosis, RD screening and the quality of health care

- To analyze the current status of neonatal screening and review the requirements on the expansion of other diseases from the point of view of the effectiveness of screening and curability of diseases in accordance with European recommendations.
- To promote the range and quality of RD selective population screening in risk populations, according to the assessment of efficiency, need and importance.
- To support and enhance existing national centres of specialized health care for patients with RD with the aim to enable and support their recognition as a Centre of RD Expertise in the case they meet the relevant quality criteria.
- To support and enhance existing national centres of specialized laboratory diagnostics in the area of RD with the aim to enable and support their recognition as the Centre of RD Expertise , if they meet the relevant quality criteria.
- To apply professional regulations (guidelines) in the care of RD patients, where national recommendations are not developed, adopt relevant international recommendations (for example, from Orphanet, international professional societies, etc.).
- To negotiate the assurance of the quality aspects of health care for RD patients with health insurance companies.
- To define a network of outpatient and laboratory health care for RD issues, based on detailed analysis.

6.4. Support of the establishment of RD Centres of Expertise and their involvement in the European RD reference network

- To implement the *Directive of the European Parliament and of the Council 2011/24/EU of 9 March 2011 on application of patients' rights in cross-border healthcare* in healthcare legislation in the Slovak Republic, especially in those parts which are directly related to RD.
- To support the foundation of the Centres of RD Expertise in line with the implementation of the *Directive of the European Parliament and of the Council 2011/24/EU of 9 March 2011 on the application of patients' rights in cross-border healthcare* in healthcare legislation in the Slovak Republic and in line with its objectives.
- On the basis of current EUCERD recommendations on the quality requirements for the Centres of RD Expertise, to develop national criteria for the recognition and operation of Centres of RD Expertise.

- To support the establishment of the Centres of RD Expertise and support their activities only if they meet the required criteria and if they ensure the continuing operation and activities of high quality.
- To support the participation of the established Centres of RD Expertise into the European RD Reference Network.

6.5 Support of science and research in the area of RD

- To analyze the presence of RD issues in departmental research.
- To support grants for RD at the Ministry of Health SR and from other sources, including the sources from EU, with the emphasis on the international cooperation in research.
- To encourage cooperation in RD research also with other departments and the Slovak Academy of Sciences.
- To require a general simplification of access to resources for research and the reduction of administration.

6.6 Collection of data on patients with RD and the support of the establishment of the RD National Register

- To analyze the current data collection on patients with RD in the statistics of the National Health Information Centre.
- To define the interests of individual medical expertise in the collection of data for own needs and also to define the needs of the registration of RD diseases fulfilling the RD criteria in such a way that through this connection we would have the option of creating a RD register.
- In the necessary extent to expand the collection of data on patients with RD also taking account of trends in the development of RD registers in EU.
- To review the legal framework for the collection of data and biological samples from RD patients.
- To implement the international recommendations for the area of RD registration.

6.7. Support and assistance of RD patient organizations in SR

- To support RD patients´ organizations operating in SR and promote their association so that they can effectively and jointly implement the requirements of patients and their families on the domestic level.
- To promote the activities of patients´ organizations in the area of increasing the awareness of the general and professional public on the issues of the life of patients suffering from RD and their families.
- To invite representatives of the patients´ organizations to meetings of the umbrella patients´ organization in the Slovak Republic and to the meetings of governmental institutions dealing with health and social care for RD patients.
- To support RD patients´ organizations in SR in their involvement in international cooperation, and particularly in the work of EURORDIS.

II.

Preparation of a "National Development Plan of care for patients with rare diseases in Slovakia - Slovakia NPZCH"

A detailed analysis of the current state of the provision of health care for RD patients and defining strategic objectives are the basis for the development of NPRD SR and its implementation.

6.8. Interdepartmental RD Working Group

The RD issue goes beyond the health care sector and is also an important issue for other ministries (e.g., the Ministry of Labour, Social Affairs and the Family SR). Therefore it is also necessary to involve other bodies of state administration as well as other materially competent institutions and representatives of the professional and general public and to establish an inter-ministerial (super-ministerial) RD working group for this purpose.

The proposed composition of the inter-ministerial working group for the preparation of NPRD SR: Representatives:

- Ministry of Health of the Slovak Republic,
- Ministry of Labour, Social Affairs and the Family of the Slovak Republic,
- Ministry of Education, Science, Research and Sport of the Slovak Republic,
- Government Office,
- Social Insurance,
- Health insurance companies,
- Association of Slovak Health Insurance Companies,
- Healthcare Supervisory Authority
- Slovak Medical Association and other professional organizations,
- Slovak Medical Chamber,
- National Health Information Centre,
- State Institute for Drug Control,
- Association of Drug and Medical Devices Suppliers,
- Slovak Association of Foreign Manufacturers of Generic Drugs - GENAS,
- Slovak Association of Suppliers of Medical Devices EN + MED,
- Slovak Association of Pharmaceutical Companies Oriented to Research and Development - SAFS,
- representatives of patients´ organizations,
- Slovak Academy of Sciences,
- faculties of medicine,
- Faculty of Pharmacy,
- representatives of specialization fields:
    - endocrinology,
    - gastroenterology,
    - haematology and transfusiology,
    - clinical biochemistry,
    - clinical pharmacology,
    - clinical immunology and allergology,

clinical oncology,
medical genetics,
neonatology,
neurology,
ophthalmology,
paediatrics,
assessment medicine,
internal medicine
and others, pursuant to the agreement of specialists.

7. The period to which the national strategy is oriented.

The proposal of NSRD SR is aimed at the development of NPRD SR based on a factual analysis of the situation in the Slovak Republic and in accordance with the recommendations of EU. The implementation of the strategy is determined for the years 2012 – 2013. In the following years, the NPRD SR will be implemented.

8. Conclusion

The proposal of NSRD SR was prepared by the Working Group of the Ministry of Health of the Slovak Republic for RD as a background document for a detailed analysis of the current state of the provision of health care for RD patients and for the preparation of NPRD SR. NPZCH SR assumes not only the preparation of the document, but also agreement on the implementation of the plan into medical practice, based on a factual analysis of the situation in the Slovak Republic and in accordance with the recommendations of EU.

The range of activities described in the objectives of the "Strategy" predetermines the need for an intensive multidisciplinary cooperation and continuous establishment of arrangements in such a way that the added value of the activities would result in an improvement of actual health care.

The NPRD SR should be adopted by the Government SR by the end of 2013.