EUROPLAN
European Project for Rare Diseases National Plans Development
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Slovenská Aliancia Zriedkavých Chorôb

EURORDIS
Rare Diseases Europe

SLOVAKIA

EUROPLAN NATIONAL CONFERENCE

FINAL REPORT

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EUCERD Joint Action

FOREWORD

The EUROPLAN National conferences are aimed at fostering the development of a comprehensive National Plan or Strategy for Rare Diseases addressing the unmet needs of patients living with a rare disease in Europe.

These national plans and strategies are intended to implement concrete national measures in key areas from research to codification of rare diseases, diagnosis, care and treatments as well as adapted social services for rare disease patients while integrating EU policies.

The EUROPLAN National conferences are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS – the European Organisation for Rare Diseases. For this purpose, EURORDIS nominated 10 EURORDIS-EUROPLAN Advisors - all being from a National Alliance - specifically in charge of advising two to three National Alliances.

EUROPLAN National conferences share the same philosophy, objectives, format and content guidelines. They involve all stakeholders relevant for developing a plan/strategy for rare diseases. According to the national situation of each country and its most pressing needs, the content can be adjusted.

During the period 2008-2011, a first set of 15 EUROPLAN National Conferences were organised within the European project EUROPLAN. Following the success of these conferences, a second round of up to 24 EUROPLAN National Conferences is taking place in the broader context of the Joint Action of the European Committee of Experts on Rare Diseases (EUCERD) over the period March 2012 until August 2015.

The EUROPLAN National Conferences present the European rare disease policies as well as the EUCERD Recommendations adopted between 2010 and 2013. They are organised around common themes based on the Recommendation of the Council of the European Union on an action in the field of rare diseases:

1. Methodology and Governance of a National Plan;

2. Definition, codification and inventorying of RD; Information and Training;

3. Research on RD;

4. Care - Centres of Expertise / European Reference Networks/Cross Border Health Care;

5. Orphan Drugs;

6. Social Services for RD.

The themes "Patient Empowerment", "Gathering expertise at the European level" and "Sustainability" are transversal along the conference.

I. General information

Country	**Slovak Republic**
Date & place of the National Conference	**February 28, 2013** **Austria Trend Hotel Bratislava**
Website	**www.zriedkave-choroby.sk, www.sazch.sk**
Organisers	**Slovak RD Alliance** **Slovak genetic organization of Slovak medical Society** **Slovak pediatric organization of Slovak medical Society**
Co – partners	**EUCERD** **ORPHANET** **DebRA SR** **Comenius University in Bratislava, Faculty of Pharmacy,** **WHO in Slovakia** **Comenius University in Bratislava, University Children's Hospital, Medical School 2nd Department of Pediatrics** **University Hospital in Žilina** **Oncology Institute St. Elizabeth Bratislava** **National Health Information Centre in SR**
Members of the Steering Committee	**Frantisek Cisarik, MD., CSc., EUCERD, Ministry of Health** **PharmDr. Tatiana Foltánová, PhD., Comenius University in Bratislava, Faculty of Pharmacy** **Ing. Beáta Ramljaková, Slovak RD Alliance, DebRA SR** **Janette Fartelová, MD., Genzyme and Sanofi Company** **Mrs. Dorica Dan, EURORDIS – EUROPLAN advisor**

Names and list of Workshops ***Plenary Sessions:*** ***Panel Sessions:*** ***Panel I.*** ***Panel II.***	**Opening Remarks, Current situation in Slovakia, The legislative tools for creating National plan for RD, current legislation and best practice, EUROPLAN Project** **1. Methodology, Governance and Monitoring of the National Plan** (Creation of the NP for RD) **2. Definition, codification and inventorying of RD** (definition, information and education in the field of RD) **3. Research on RD** (National registry of RD, research and science in RD) **4. Centres of Expertise and European Reference Networks for Rare Diseases** (Screening and diagnostics of RD) **5. Orphan Drugs** **6. Social services for RD** **The speakers prepared the presentations of the main themes in close** **co-operation with the Steering Committee of theConference. In additional, critical aspects of every theme were disscused in Panel Sessions as well.**
Chairs and Rapporteurs of Workshops	**Chairs:** • **František Cisarik, MD., CSc. EUCERD, Ministry of Health SR** • **Ing. Beáta Ramljaková - Slovak RD Alliance, DebRA SR** **Doc. RNDr. Ľudevít Kádáši, DrSc. – Slovak genetic organization of Slovak medical Society** • **Prof. László Kovács, MD., DrSc., MPH –**

	Orphanet coordinator in Slovak Republic, Slovak pediatric organization of Slovak medical Society • **PharmDr. Tatiana Foltánová, PhD. – Faculty of Pharmacy, Comenius University Bratislava** • **Prof. Svetozár Dluholucký MD., PhD. - Children's University Hospital Banská Bystrica, Newborn Screening Centre** **Speakers:** • **František Cisarik, MD., CSc. EUCERD, Ministry of Health SR** • **Mrs. Dorica Dan, EURORDIS – EUROPLAN advisor** • **Mrs. Amalia Egle Gentile, ISS (Italian Institute for Health, EUROPLAN coordinator)** • **Prof. László Kovács, MD., DrSc., MPH – Orphanet coordinator in Slovak Republic, Slovak pediatric organization of Slovak medical Society** • **Anna Hlavatá, MD., PhD. MPH, Children's University Hospital Bratislava** • **Assoc. Prof. Eva Goncalvesová, MD., CSc. - The National Institute of cardiovascular diseases, Bratislava** • **PharmDr. Tatiana Foltánová, PhD., Comenius University in Bratislava, Faculty of Pharmacy PharmDr. Ján Mazag, State Institute for Drug Control** • **Beata Havelková, MD., MPH, VŠZP – Public Health Insurance Company** • **Prof. Svetozár Dluholucký MD., PhD., Children's University Hospital Banská Bystrica, Newborn Screening**

	Centre • **Anna Baráková, MD., NCZI – National Health Information Centre** • **Ing. Beáta Ramljaková, Slovak RD Alliance, DebRA SR** • **Mária Duračinská M.Sc., Patient organization of muscular dystrophy in the SR** • **Assoc. Prof. Dr. Jitka Prevendárová, PhD., Comenius University in Bratislava, Faculty of Education**
Annexes:	**I. Office Meeting of the Interdeparmental Working Group for Rare Diseases of the Ministry of Health** **II. Programme of the conference** **III. List of participants,**

II. Main Report

Opening Remarks

Ing. Beata Ramljakova opened the conference, introduced and welcomed the chairs as well as the guest from Europlan and all audience. František Cisarik, MD., CSc started the conference with the first speech. Afterwards the conference followed the programme.

Overview current situation in Slovakia

There is no specific budget dedicated to RD in Slovakia

- **December 12, 2011** – Establishment of the Slovak RD Alliance – Slovenská aliancia zriedkavých chorôb (Aliancia ZCH).
- **December 2011** – small workshop regarding RD was held on Ministry of Health
- A working group was established in **January 2011** at the Ministry of Health to work on a strategy of health care improvement for patients with rare diseases.
- **February 20, 2012** – Press conference/Round Table with specialist of RD in Slovakia. Slovakia actively participated in campaign for the first time.
- **February 29, 2012** – The first Slovak RD Conference organized by Orphanet was held in Bratislava on the occasion of Rare Disease Day. The aim of the conference was to raise awareness about rare diseases, as well as to improve access to the treatment. More than 150 invited participants attended the conference. Various specialities were represented as health care providers, patient organizations, universities and research institutions, public insurance companies and social affairs institutions.
- **October 2012** - National Strategy for RD was approved by the Slovak Government. It means - next step is to create and implement a National Plan for RD. The deadline for the approvement of the National Plan is December 2013.
- **November 2012** - Working group of 13 people was created (after the NS was approved) - by Ministry of Health (MoH). There are representatives of physicians, Orphanet, EUCERD, Slovak RD Alliance, insurance companies, national screening centre specialist, 3 people from the Ministry of Health, pharma industry, regulatory institutions, representative of COMP.
- **February 2013** – creation of Interdepartmental Working Group designated by MoH, Ministry of Education, research and Ministry of Labour, social affairs and family with next specialists from other spheres.
- **February 27, 2013** – The first session of Interdepartmental WG on MoH. Cooperation between members and stakeholders started.
- **February 27, 2013** – Press conference/ “Patient without diagnose” was held with experts in the field of RD. Rare Disease Day campaign continues (2012-2013).

Theme 1 - Methodology, Governance and Monitoring of the National Plan

Sub-Themes:

1.1 Mapping policies and resources
1.2 Development of a National Plan /Strategy
1.3 Structure of a National Plan /Strategy
1.4 Governance of a National Plan
1.5 Dissemination and communication on the National Plan
1.6 Monitoring and evaluation of the National Plan
1.7 Sustainability of the National Plan

Current legislation and recommended procedures

President and guarantee of Forum of Experts, František Cisarik, MD., CSc. - Faculty Hospital Žilina, Department of medical genetics, Slovakia, set out proposals appropriate and most important for Slovakia.

The creation process of the National Plan for Rare Disease in the Slovak Republic (NP RD SR) follows the governmentally approved document of National Strategy for Rare Disease Patient of Health Care Development for years 2012 – 2013 (RD strategy). According to the RD strategy, 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 targets, which are a part of European initiatives and will form the content of the National Plan for Rare Disease in the Slovak Republic, follow this triad:

Centres of expertise – European reference networks – Registries of rare diseases

Centres of expertise (CoE) are the cornerstone for the diagnosis, treatment, and management of rare disease patients. Both big and small countries depend 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 CoE. It is not a coincidence that EUCERD drew the first Recommendations on quality criteria for CoE; 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 and preparation for active implementation into the **European reference networks (ERN)**.

Another advantage, which might the CoE at least partially solve is the existence of **registries** of rare diseases and thus epidemiological as well as clinical registries. CoE might present a

source for data gathering about the real demand and experience with the treatment of rare disease patients. Prepared implemantation of e-health might also improve the detection of rare patients and their implementation into registries.

The Rare disease strategy was drawn by a small working group at the Ministry of Health of the SR in 2012. The creation of National Plan for Rare Diseases in the Slovak Republic is, however, a much more extensive project, and an inter-branch workgroup for rare diseases was charged with it. Yet, National Plan for Rare Diseases 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 Diseases in the Slovak Republic implementation regularly. This role can be played only by a permanent working group at the Ministry of Health or by an internal body of the Ministry of Health focused on the already mentioned issues.

1.1 Mapping policies and resources

In the SR no epidemiologic figures on rare diseases were published, neither the impact of national health or social care is known. However since 2010 several activities were performed. Rare diseases were presented at national conferences of the subgroups of Slovak medical society (as Slovak genetic organization of Slovak medical Society, Slovak pediatric organization of Slovak medical Society) as well as in 2012 for the first time Slovakia actively participated at the International rare diseases day. All these activities raise the awareness between the professionals, at the patients' side Slovak RD Alliance was established, which increases the public awareness.

- No report has been made to report on of the existing resources and actions on RD (or of which RD patients can benefit) in the national health care and social system.
- The unmet needs of rare disease patients have not been evaluated for now.
- All relevant European documents are taken into account as 141/2000, 847/2000, 2009/C 151/02, 1350/2007/ES, Crossborder directive 2011/24/EU
- The Europlan national conference 2013 speed up the monitoring of the current situation, now precise review and analysis are needed.
- According to the results of the Europlan national conference three key points were detected and thus formation of CoE, their participation in European reference networks and need for registries of rare disease patients.

1.2 Development of a National Strategy

In the formation of national strategy patients organisations were involved. The national strategy is mainly from the legislative point of view a key document to support the creation of National Plan.

1.3 Structure of a National Plan /Strategy

For now only a National Strategy exists. It has a common structure, starting with general characteristic of the importants terms as well as organisations in this field, following with diagnosis, treatment and finishing with social care.

Clear deliverables and measurable results have to be implemented into the National Plan, which has to be prepared till the end of 2013. Now no timeline exists only dates of 4 meetings of the interdepartmental group were set. The aim of the national plan is to be directly implemented into the national health care and social system, mainly on the principle of exceptions, as is the case of biologic and innovative treatment for now. This has to guarantee immediate integration, comprehensiveness and sustainability in the health care and social needs. In the social care rare disease patients must be accepted as one of the goups of disabled, defined already in the legislation.

1.4 Governance of a National Plan

Stakeholders cover all areas of expertise relevant to the NP, such as pharmacology, regulatory, clinical, health and social services, epidemiology, administrative policies. However more involvement of administrative policies and healthcare and social authorities might be an advantage. These stakeholders participate in the interdepartmental group: healthcare, research and social affairs authorities, patients, healthcare professionals, academics, health insurances, representatives of the industry. Patients are a proper part of the interdepartmental group, thus they will be present in all parts of the NP formation. The limitation of the interdepartmental group is nonexistence of its status, and it also doesn't have "Rules and procedures" for now. The dates of the meetings are set in advance. The Steering Committee doesn't have any working groups. Untill now there was only one meeting and thus the plan of work is not clear.

1.5 Dissemination and communication on the National Plan

The dissemination and communication on the national plan didn't start until now. It is planned to be published on the website, to make it available for the public, easy to follow and to make it transparent.

1.6 Monitoring and evaluation of the National Plan

The EUROPLAN Indicators are planned to be used as a basis for monitoring and evaluating the activities stemming from the NP. The process of further evaluation is not clear for now.

1.7 Sustainability of the National Plan

There is no specific budget attached to the NP. Big effort is planned to implement it into the health care system. This has to be one of the factors which may guarantee the sustainability. The cost estimates for the health care of rare diseases are not known for now and are a part of the analysis of current situation, as mentioned at the conference.

To plan the funding by Structural Funds 2014 – 2020 will be taken into account.

ACTIONS	INDICATORS	TYPE	ANSWERS
Development of Regulations/ Laws	**Existence of Regulations / laws that support the creation and development of a RD plan**	**Process**	-Not existing, not clearly stated -Existing, clearly stated, partly -implemented and enforced **x Existing, clearly stated and substantially implemented and enforced**
	National / regional (percentage of regions)	**Process**	Index based on the number of regions with a Plan divided by total number of regions. A National plan will account for this index equal 100% **NA**
Establishment of Coordination mechanisms	**Existence of a coordination mechanism**	**Process**	-Not existing, not clearly stated **X Existing, clearly stated, partly implemented and enforced** -Existing, clearly stated and substantially -implemented and enforced
	Existence of an expert Advisory Committee	**Process**	-Existing and meets regularly **X Exists but partly functioning** -Does not exist

ACTIONS	INDICATORS	TYPE	ANSWERS
Establishment of an external evaluation of the plan/strategy Procedure	**Existence of an external evaluation body / Procedure**	**Process**	Number of meetings held by year: **4**
Degree of comprehensi-veness	**Number of priority areas included**	**Process**	Number ranging from 0 to 10 : **3**
Establishing of a budget for developing the plan/strategy	**Budget of plan/strategy**	**Process**	Overall budget allocated: **No budget**

Theme 2 - Definition, codification and inventorying of RD

Sub-Themes:

2.1 Definition of RD

2.2 Codification of RD ad traceability in national health system

2.3 Registries and databases

2.4 Information on available care for RDs in general, for different audiences

2.5 Help Lines

2.6 Training healthcare professionals to recognise and code RD

2.7 Training healthcare professionals

ORPHANET – INFORMATION, EDUCATION AND EXPERT CENTRES FOR RARE DISEASES

Prof. László Kovács, MD., DrSc., MPH et al, briefly described the role of ORPHANET as an informational and educational source for rare diseases. Most attention was given to the CoE and European Reference Networks. CoE are the priority for action in the EC Public Health Program (2008 – 2013). The authors suggested an easy procedure how to get the basic data about the readiness of the clinics to be recognised for Centres of Expertise at the national level. EUCERD Recommendations on quality criteria for centres of expertise were introduced. The coordinators of the potential CoE should be contacted and asked to fill in the questionnaire designed to check whether the centre matches the EUCERD Recommendations or not. In order for the process to be transparent, the selection criteria of expert resources are listed on national website (www.orphanet.sk).

The analysis of the questionnaires has to be carried out at the national level, to map the basic data about the current status. One questionnaire per department or clinic shall be filled in order to allow the evaluation. Clinics will be divided in two groups: the ones which achieved the threshold and could be recognised as CoE at the National level and the ones which need to be further monitored to reach the threshold. On the other hand Dr. Cisarik mentioned the importance of consistent assessment of the national criteria for the designation of CoE as well as EUCERD criteria. This will be the subject for further discussions at the interdepartmental meeting.

2.1 Definition of RD

In the SR official definition of rare diseases is accepted. However rare diseases are not identified in the health and social legislation.

2.2 Codification of RD ad traceability in national health system

For the official codification system is used ICD 10. ORPHAnet coding is currently not used in the Slovak hospitals, neither at the specialised clinics. ICD10 is mainly used in the reimbursement process, in the communication with the insurance companies as well as with the National centre for Health Information.

To identify the patient with rare diseases in the health care system proper coding would be an advantage. The CoE could support the use of proper code. In CoE well trained staff has to work, and the cooperation with all other stakeholders has to be supported, thus the importance of proper coding must be clear.

2.3 Registries and databases

No official list of RD is compiled in the SR. Official governmental RD registry doesn't exist. Specific registries are provided by the specialist in the hospitals – "potential CoE". The issues of the information flow have to be further discussed. The conference presented the idea that information flow was to share the epidemiologic information at the national level and these have to be implemented also into the international registries. The clinical registries have to be provided by the CoE or specialized medical societies. Details have to be specified.

2.4 Information on available care for RDs in general, for different audiences

Official information sources about RD in the SR are the initiatives to raise awareness on RD, e.g. Rare Disease Day.

Orphanet Slovak translation is used to provide information about RD in Slovak.

2.5 Help Lines

No help lines exist in the SR

2.6 Training healthcare professionals to recognise and code RD

That level of awareness and knowledge about the proper coding between healthcare professionals is relatively low and could be increased in CoE. In advance set training dates of every healthcare professional in the CoE in the field of the importance of proper coding and use of ICD 11 could help to solve the issue of ICD 11 implementation.

2.7 Training healthcare professionals

Further training of healthcare professionals: pregraduate as well as postgraduate is a matter of further discussion.

ACTIONS	INDICATORS	TYPE	ANSWERS
To officially adopt the EC RD definition (no more than 5 cases/10,000 inhabitants)	**Adoption of the EC RD definition**	**Process**	**X Yes** No EU definition modified with an additional information
To include the best Rare Diseases classification currently existing into the public health care related services	**Type of classification used by the health care system**	**Process**	· ICD-9 **X ICD-10** · OMIM · SNOMED · ORPHAN · MESH · Others
	Developing policies for recognising RD by the care information systems	**Process**	**x Not existing, not clearly stated** Existing, clearly stated, partly implemented and enforced · Existing, clearly stated and substantially implemented and enforced
Defining a surveillance system based on a patient outcomes registry	**Registering activity**	**Process**	Centralised RD registry Multiple RD registries but well coordinated and standardised **x Multiple RD registries not standardised** No registry at all
	Number of diseases included	**Outcomes**	Number ranging from 1 to 20 **Not standardised 7** **EB, MD, DMD, ONKO, HEM, INBORN ANOM,**

ACTIONS	INDICATORS	TYPE	ANSWERS
Existence of information sites for professionals provided by the plan/strategy	**Existence of a comprehensive national and/or regional RD information system supported by the government**	**Process**	· Yes, covers most RD · Yes, covers only some RD **X No formal decisions have been taken**
	Help lines for professionals	**Process**	· Yes, covers most RD · Yes, covers only some RD **X No formal decisions have been taken**
	Clinical guidelines	**Outcomes**	Number ranging between 0 to 30 **NA**
Promoting training activities & awareness educational campaigns among professionals	**Number of such as activities (*training & awareness educational*) promoted by the plan/strategy**	**Outcomes**	Number ranging between 0 to 30 **2**
Building – Supporting the existence of comprehensive help line and information sites for patients provided by the plan/ strategy	**Availability of Help line for RD**	**Process**	· Own help line · Referred RD help lines **x No formal decisions have been taken**

Theme 3 - Research on RD

Sub-Themes:

3.1 Mapping of existing research resources, infrastructures and programmes for RDs

3.2 Dedicated RD research programmes and governance of RD research funds

3.3 Sustainability of research programmes on RD

3.4 Needs and priorities for research in the field of RDs

3.5 Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects

3.6 RD research infrastructures and registries

3.7 EU and international collaboration on research on RD

National Health Information Centre task regarding the data availability on rare diseases in the SR

Anna Baráková, MD. from National Health Information Centre, Bratislava, Slovakia, informed the auditorium about options and current state of the data availability on rare diseases in Slovakia. She summarized needs and priorities in this field.

3.1 Mapping of existing research resources, infrastructures and programmes for RDs

The key moment enabling us to know not only the local, but also the wide-area incidence of the relevant group of diseases should be the interest of expert communities (often based on the activity of relevant authorities within them) and their central reporting. The SR is finalizing the preparation of individual steps in solving this issue. The basis for a successful progress is to analyse the current state.

3.2 Dedicated RD research programmes and governance of RD research funds

The current support of the health care system and science is not including rare diseases separately.

3.3 Sustainability of research programmes on RD

There is no specific RD research programmes in Slovakia.

3.6 RD research infrastructures and registries

The National Health Information Centre (NHIC) is a source of rare diseases in the following databases:

- The National Register of Congenital Anomalies-NHIC has at its disposal the data on congenital anomalies dating from 1965, which were processed within the federation

separately for the Czech and Slovak Republic until 1992. After the end of the federation, the SR has been collecting this data separately.

Reports on congenital anomalies were for a long time focused on somatic mutations, less on metabolic disorders, which was related to the diagnostic availability and reporting duty of physicians. The rate of reporting, including rare diseases, was low since the general practitioners for children and adolescents who received the positively confirmed results sent by Screening Centre in Banská Bystrica, did not additionally report these results to NHIC, although the reporting duty concerned children up to 1 year of age until 2011.

The change of the system of reports to a register in terms of collecting data on congenital anomalies was a very positive change. Individual identification of these cases enabled solely by the register was a great contribution.

- National cancer registry- provides information on rare cancers and hemato-oncological diseases.
- Registry of patients with diabetes mellitus 1. Type - registers also newborns with diabetes.
- National register of congenital anomaly of heart- registers also rare congenital diseases of heart.

The most important source of data for the future should be the Registry of patients with congenital anomalies. However, it is necessary for individual clinical and laboratory workplaces to accept also changes in the system of congenital anomalies registration.

3.7 EU and international collaboration on research on RD

There is no conception of collection and processing of data regarding rare diseases, neither is there any unified conception of diagnosis, treatment, and overall health care of patients with rare diseases. The central availability of data concerning rare diseases is very low for different medical specialisations which deal with this issue. There exist only partial pieces of information, which are spread across different workplaces. The SR should participate in European information network on rare diseases. The SR is a member of ICBDSR but not EUROCAT, which is necessary to be changed.

Since 2012, the reporting duty of congenital anomalies (according to the model of the Czech Republic) has been extended with reports of cases up to the age of 15. The reporting duty was thus assigned also to general practitioners for children and adolescents who should take the burden of reporting the congenital anomalies to specialists. The report form concerns mainly epidemiological data (including prenatal diagnostics). It is possible that in the future, congenital anomalies reporting without reference to the age will be adopted.

In order to control the data on rare diseases (in terms of available databases), an effective collaboration of NHIC with the Newborn Screening Centre in Banská Bystrica is necessary (in addition to the regular communication with clinical and outpatient physicians). In addition, it is beneficial to cooperate also with health insurance companies, which register also cases which require specific treatment for individual diagnoses. The clinical registries, which are important for physicians from the point of view of clinical state monitoring, treatment, and quality of patients' lives, should be under the full competence of the relevant expert centres.

The key to the knowledge of the real state of disease incidence rate in the population is an effective collaboration between relevant partners (including implementation of systematic control mechanisms), which is a basis for success. We hope that the inter-branch working group for congenital anomalies in the SR reaches its goal.

ACTIONS	INDICATORS	TYPE	ANSWERS
Building a research programme for Rare Diseases	**Existing of RD National/Regional research programmes**	**Process**	Specific research programme for RD RD research programme included in the general research programme as a priority **x Not RD research programme**
	RD research programme Monitoring	**Process**	**X Not existing, not clearly stated** Existing, clearly stated, partly implemented Existing, clearly stated and substantially implemented
	Number of RD research projects approved by year	**Outcomes**	**NA**
	Clinical trials funded by public bodies	**Outcomes**	. Yes, action implemented **X No actions have been taken** . Under discussion
	E-RARE joining	**Process**	· Ongoing · In process **X Not considered**
	Including public health and social research, in the field of rare diseases	**Process**	· Yes **X No** · Under discussion
	Research platforms and other infrastructures are also funded by the research programme	**Process**	· Yes **X No** · Under discussion

ACTIONS	INDICATORS	TYPE	ANSWERS
Existence of national policy in support of the recruitment of young scientists / Researchers specifically for Rare Diseases	**Number of young scientists recruited every year to work specifically on rare diseases**	**Process**	**NA**
Allocate funds for the RD research programme	**There are specific public funds allocated for RD research**	**Process**	· Yes **X No** · Under discussion
	Funds specifically allocated for RD research actions /projects per year since the plan started	**Outcomes**	· Million Euros allocated to RD research projects · Percentage of funds allocated for RD projects by the total funds for projects **x No specific funds allocated for RD**

Theme 4 – Care for RDs - Centres of Expertise and European Reference Networks for Rare Diseases

Sub-Themes:

4.1 Designation and evaluation of CE

4.2 Scope and functioning of CEs

4.3 Multidisciplinarity, healthcare pathways & continuity of care

4.4 Access to information

4.5 Research in CEs – How to integrate research on RDs and provision of care

4.6 Good practice guidelines

4.7 Diagnostic and genetic testing

4.8 Screening policies

4.9 European and international collaboration – Cross-border healthcare and ERNs (European Reference Networks)

4.10 Sustainability of CEs

There is currently no official policy concerning centres of expertise for rare diseases and no official centres of expertise for rare diseases in Slovak Republic. Health care for several rare diseases is centralised mainly at the Departments of Clinical Genetics, the Centre for metabolic diseases, and in several specialised outpatient clinics for metabolism or few types of rare diseases. Several specialised and centralised departments would be appointed as centres of expertise in the near future .

CoE is one of the priorities to consider in creating National plan for RD in Slovakia. It is necessary to analyze and select some of EUCERD criterias the most compatible with Slovak RD situation. Current state description of Centres of Expertise is drafted in couple of next paragraphes.

The reality of care for patients with RD – inborn metabolic diseases in Slovakia.

Anna Hlavatá, MD., PhD., MPH, Centre of Inherited Metabolic Disorders, C, Bratislava
2nd Department of Pediatrics Comenius University Medical School University Children´s Hospital, described experience of health care team in the Centre of Inherited Metabolic Disorders in Children's Hospital in Bratislava in the diagnosis and management of patients with inborn metabolic diseases (IMD). She expounds the current mode of lifetime care for these patients and possibilities for improvement.

Since the most serious manifestations of IMD are usually manifested in early childhood and directly threaten health and life of patient, it is only natural that such workplace was started

to form in pediatric department. IMD Center in Children's Faculty Hospital in Bratislava uses laboratories in the Department of Laboratory Medicine and Laboratory of Molecular Genetics at the 2nd Pediatric Department. When finalizing the diagnosis, there is cooperation with the Institute of Medical Biology, Genetics and Clinical Genetics, Faculty of Medicine and UNB in Bratislava as well as international cooperation is needed.
Clinical and ambulatory care of patients with IMD is distributed to the pediatrics' and neurology departments. In recent years IMD have no longer been just pediatric problems. While in past the majority of patients did not live to adulthood, today, thanks to new treatment and therapeutic procedures, it has been changed. Improving the knowledge of the pathogenesis, diagnosis and especially IMD therapy, patients can reach the age of adulthood and they can even be diagnosed in adulthood.

Although the EU has ensured registration of majority of drugs used for IMD in EMA, there are still some long-term-used drugs (e.g. biotin) requiring exceptions for individual import by Ministry of Health of the Slovak republic (MoH SR). For treatment of metabolic patients there are also drugs used which are already registered in the Slovak republic, but not in the indication of metabolic disease, or there is no form for long-term, chronic usage registered (e.g. calcium folinate *p.o*. for treatment of cerebral folate deficiency). Also in this case we start with request for import confirmation by MoH SR, in the case of acceptance an apply to health insurance company to secure the payment for therapy of their client is following.

Having all the acknowledgements (there is always the one month period since the appeal till the adjudgement) the drug can be ordered in pharmacy.

As the diseases are genetic, it is necessary to provide the genetic advice and prenatal diagnosis, independently of the disease being treatable or not. On this purpose, to manage a complex health care, the Genetic ambulance was founded in CIMD in the 2nd Department of Pediatrics.

The postgradual training in the field of inherited metabolic diseases has not been founded in the system of postgradual training. This specialization is necessary for Slovak patients with metabolic diseases to be provided with medical care comparable to that in other countries of the EU.

Pulmonary arterial hypertension centre – a model for rare disease management

Eva Goncalvesová, MD, PhD, FESC, Ass. Prof. of Cardiology - Pulmonary Arterial Hypertension Centre, Dept Heart Failure/Transplantation, National Cardiovascular Institute, Bratislava, Slovakia

Diagnosis and management of PAH concentrate to the dedicated centres. The purpose of a reference centre is to undertake assessment and investigation of all causes of pulmonary hypertension, PAH-specific drug therapy, work with other healthcare providers to obtain best outcomes for patients, and to undertake research and education. In general, high

volume centres achieve best outcomes, because of effective concentration of the specific experience and skills needed for rare disease management.

PAH centres have to be equipped by a ward where staff have special expertise in PH, an intensive therapy unit, a specialist outpatient service, diagnostic investigations including echocardiography, CT scanning, nuclear scanning, magnetic resonance imaging, ultrasound, exercise testing, lung function testing, and catheterization laboratory (with expertise in performing vasoreactivity test) and access to the full range of specific PAH drug therapy available in the country. There should also be clearly established links to other services, which may not necessarily be on the same site. These links include services for connective tissue disease service, pulmonary endarterectomy, lung transplantation, adult congenital heart disease and genetics. Referral centres are required to undertake, register and provide patient survival analysis. It is expected that PAH centres will participate in collaborative clinical research in PAH, which includes phase II and phase III clinical trials and grant regular education about all clinical aspects of PH to appropriate healthcare professionals. Cooperation with the national and/or European pulmonary hypertension patients association is also an important part of centre activity.

Medical care of patients with PAH is well established in Slovakia. However creation of further reference centres might lead to the fact that the centres will be not able to meet quality standards of reference centres as nb. of new diagnosed pts/year, nb of treated pts/year, this fact is in rare diseases of high importance.

Newborn Screening in Slovakia – from 1985 till today

Prof. Svetozár Dluholucký MD., and PhD, Dr. Mária Knapková, - Newborn Screening Centre of SR, Children Faculty Hospital Banská Bystrica, Slovakia, presented results in newborn screening in Slovakia and international cooperation.

The International Society of Newborn Screening (ISNS) and European Union of National Experts for Newborn Screening (EUNENBS) are guarantor institutions for unification and evaluation of national NBS in Europe. Spectrum of screened disorders in NBS is chosen according to Wilson - Junger criteria, validity of which has been confirmed even after fifty years of existence.

NBS in Slovakia started in Trenčín in 1978 as phenylketonuria (PKU) screening which used Guthrie test, and simultaneously as the evaluation of the possibility to introduce the congenital hypothyroidism (CH) screening by means of radioimmunoassay (RIA) of thyroxine (T_4) from dry blood spot sample (DBS) in Banská Bystrica. The retrospective analysis of CH incidence in Slovakia confirmed incidence similar to other states of the world and the development of DBS T_4 RIA test enabled CH screening to start in the central Slovakia between 1980 and 1984. This organizational model was used to start the regular CH screening in Slovakia in 1985 in the Newborn Screening Centre of Slovak Republic (NSC SK).

After the introduction of the quantitative fluorometric test for phenylalanine (phe) in DBS, PKU screening was shifted to NSC SK and Guthrie semi-quantitative screening was abandoned. Congenital adrenal hyperplasia (CAH) screening and cystic fibrosis screening were introduced to regular screenings in 2002 and in 2009 respectively. In Slovakia NBS is provided in one national screening centre (NSC SK). Between 1985 and 2011 coverage was more than 98% of liveborn infants while escape from screening was recorded at the start of the screening (in 1985) and in the last years due to deliveries abroad.

NSC SK ranks on the sixth place in EUNENBS evaluation of thirty four EU national NBSs.

Expanded NBS in 2012 – 13

Extension of the spectrum of rare metabolic disorders (RMDs) by means of tandem mass spectrometry (ms/ms) – introduction of so called extended NBS (ENBS) − has become the main task for the future. From wide range of RMDs detected by ms/ms, nine RMDs including defects of beta-oxidation of fatty acids, organic acidurias, and carnitine pathway defects were chosen according to W-J criteria. Their common feature is a sudden disruption of energetic metabolism after various forms of stress leading to life-threatening situations, even death. In contrast, after early detection by NBS, the prevention of such life-threatening events is relatively simple and inexpensive.

During the first three months of the year 2012 the ms/ms device was tested, and ENBS software adjusted for routine use. From June 15th to the end of October 2012 (four months), the prospective pilot study of ENBS of all newborns in SK was carried out.

Regular population-wide ENBS started in January 1st 2013 under the guarantee of Ministry of Health. During January 2013, 5 343 newborns were screened for a total of 13 disorders.

During this month of ENBS, screening prevalence was extraordinary high: 1:763 liveborns which justifies the extension of NBS in Slovakia as a highly effective step of preventive care in childhood. Definitive evaluation of ENBS effectiveness and of other additional features will be possible in the next years.

ACTIONS	INDICATORS	TYPE	ANSWERS
Improve the quality of health care by defining: Appropriate centres with experience on RD And pathways that reduce the diagnosis delay and facilitate the best care and treatments	Existence of a policy for establishing centres of expertise at the national/regional level	Process	**X Not existing, not clearly stated** ·Existing, clearly stated, partly implemented .Existing, clearly stated and substantially implemented
	Number of centres of expertise adhering to the policy defined in the country	Outcomes	NA
	Groups of rare diseases followed up in centres of expertise	Outcomes	NA
	Centres of expertise adhering to the standards defined by the Council Recommendations - paragraph d) of preamble	Outcomes	NA
	Participation of national or regional centres of expertise into European reference networks	Outcomes	NA

ACTIONS	INDICATORS	TYPE	ANSWERS
Existence of information sites for professionals provided by the plan/strategy	**Existence of a comprehensive national and/or regional RD information system supported by the government**	**Process**	Yes, covers most RD Yes, covers only some RD **X No formal decisions have been taken**
	Help lines for professionals	**Process**	Yes, covers most RD Yes, covers only some RD **X No formal decisions have been taken**
	Clinical guidelines	**Outcomes**	**NA**
Promoting training activities & awareness educational campaigns among professionals	**Number of such as activities promoted by the plan/strategy**	**Process**	**NA**

ACTIONS	INDICATORS	TYPE	ANSWERS
Develop Screening Policies	Number of diseases included in the neonatal screening programme	Outcomes	13
	Number of diseases included in the neonatal screening programme properly assessed	Outcomes	13
Ensure quality of RD diagnosis laboratory	Existence of a public directory/ies of both genetic tests on Rare Diseases	Process	X Yes · No · Under discussion
	Proportion of laboratories having at least one diagnostic test validated by an external quality control	Outcomes	NA

Theme 5 – Orphan Medicinal Products

Sub-Themes:

5.1 Support to Orphan Drug (OD) development

5.2. Access to treatments

5.3. Compassionate use programmes

5.4. Off label use of medicinal products

5.5. Pharmacovigilance

In this section PharmDr. Tatiana Foltánová, PhD presented the European view and a brief review of the European incentives, to support R&D in the field of OMP. In the second presentation PharmDr. Ján Mazag, the Head of the State Institute for Drug Control, presented OMP from the regulatory point of view. The perspective of the public insurance company, which is the biggest insurance company in the SR and the only one which insures most of the patients in the SR, was presented by MUDr. Beata Havelková MPH. The presented preliminary data was from the years 2010 and 2011. According to the presentation in the year 2010, 1441 rare disease patients were treated with the OMP, in the year 2011 it was 1691 rare disease patients.

5.1 Support to Orphan Drug (OD) development

Slovakia, being a small country, does not have Slovak pharma companies, thus OMP (Orphan Medicinal Products) are not developed and no specific programmes exist to foster further development of designated OMP. In Slovakia international pharma companies are represented.

For now in Slovakia there are no CoE designated at the national level or CoE corresponding/matching the EUCERD criteria. However rare disease patients are treated in the Faculty Hospitals, thus most of the MDs are involved also in the academic field/duties. This situation is prepared to support clinical trials for small populations run by academic centres. Creation and designation of CoE will solve this problem.

5.2. Access to treatments

- The national procedure of pricing and reimbursement of OMP is known as „categorisation“, updated up to January 2012 in monthly intervals. It is defined in theSlovak legislation. The Marketing authorisation holder (MAH) has to submit all necessary documentation to the categorisation committee which is active at the Ministry of Health. The categorisation procedure lasts 60 days from submission of all needed data. For OMP no special rules exist and OMP are not defined in this law, although drugs for disease with very low prevalence are mentioned in this law (1:100 000).
- Pharmacoeconomic studies are not possible in OMP, thus categorisation committee will benefit from CAVOMP. However it is important to increase awareness about this fact. This means it is needed to adapt the legislation to profit

from the CAVOMP and improve the process of pricing and reimbursemt of OMP at the national level. On the other hand it is of high importance to educate responsible persons, who will be involved as experts in the categorisation process of OMP. At the international level, SR does not have a representative in the EUnetHTA nor in the COMP or MOCA thus is not involved in CAVOMP at the EU level. Slovak representatives are not involved in the scientific advice or protocol assistance

- Increasing awareness between profesionals as well as creation of training programmes in the field of rare diseases on diagnosis, treatment and pharmacoeconomic assessment might solve national as well as international personal problems.
- Adoption of the Cross Border Healthcare Directive increased the possibilities to support the use of the OMP delivery on patient named basis. Thie Ministry of Health is in charge of this and acts generally for all drugs, not only OMP which are not accessible on the market. The MD has to provide all information about the diagnosis of the patient as well as grounds, why the patient will benefit from this drug and that there is no alternative treatment available. According to the approval of the Ministry of Health the insurance company provides the OMP to the hospital, hospital pharmacy respectively, where the patient is treated.
- Creation of CoE will support coordinated/centralised access to OMPs, which nowadays naturally exist. The prescription of all drugs categorised in the SR, as well as OMP, is regulated by several restrictions as speciality of the MD and character of the institution. For OMP this means that they could be prescribed only by MD who has experience with treatment of the disease as well as only in hospitals, which provide care for these patients in the future CoE. Several medical specialities as well as several hospitals could prescribe the same OMP. The restrictions of the OMP prescription are set in the “categorisation list”. Following these facts it means that the “categorisation list” is prepared to serve as a tool for the regulation of OMP after the designation of CoEThe medical doctors and representatives of the CoE could be a part of the “categorisation commitee”.The “categorisation list” is prepared with active cooperation of health care insurance companies, which are responsible for the reimbursement. There is no special budget for OMPs. Because of solidaristic health care system in Slovakia, OMPs are fully or mostly (with minor patient co-payment) reimbursed, if they fullfill all the criteria set in the “categorisation”.
- In the “categorisation list” there is no special label for OMP. However at the website of State Institute for Drugs, professionals as well as public can set the filters to get the OMP categorised in the SR. However in the practice several professionals are not aware of the fact if the drug is an OMP and they mistake OMPs with other biological therapies, gene therapies, etc.

5.3. Compassionate use programmes

Compassionate use in the SR is very much influenced with the participation of the patient in the international registries. Registries of rare disease don't exist and thus they do not receive extra funds by public sources. Here the involvement of patient organisations is of big importance. According to the conference presentations, thanks to the participation in the registry supported by these organisations, two Duchenne muscular dystrophy patients were involved in the clinical trial.

5.4. Off label use of medicinal products

There is no data about the off-label use in rare disease patients. Health insurance companies are in charge of the reimbursement of drugs . These are facing several problems with correct diagnose recording, thus disabling the reimbursement according to the proper diagnosis on one hand, on the other hand disabling to follow the off label use in general. However if the MD wants to use a treatment which is not indicated for the disease, he must individually ask the insurance company to reimburse this treatment and provide them with satisfying clinical data about the benefit for the patient. Centralisation of care into CoE as well as prepared e-health program may solve this situation.

5.5. Pharmacovigilance

The State Institute for Drug Control is in charge of pharmacovigilance. No special programs for OMP exist. The data is submitted to the European database of suspected adverse drug reaction reports, at the national level it is not published now. Encouragement of the patients and health care professionals to report suspected ADR could be performed in CoE. This must be one of the duties of the CoE, which is needed to be implemented in the designation criteria of the CoE in the SR (if we decide for national designation criteria of CoE).

ACTIONS	INDICATORS	TYPE	ANSWERS
Gathering the expertise on Rare Diseases at European level **To ensure and accelerate accessibility to Orphan Designated Drugs (ODD)**	**Number of ODD market authorizations by EMEA and placed in the market in the country**	**Outcomes**	Index based on Number of ODD placed in the market by total of ODD approved by the EMEA **24/65**
	Time between the date of a ODD market authorization by EMEA and its actual date of placement in the market for the country	**Outcomes**	Average days since the date of market authorization by EMEA until the official date of placement in the market in the country **557 days, 18.6 months** **Last counted in 2010**
	Time from the placement in the market in the country to the positive decision for reimbursement by public funds	**Outcomes**	Average days since the date of placement in the market until the reimbursement decision date in the country **NA**
	Number of ODD reimbursed 100%	**Outcomes**	Number ranging 0 to 1,000 **34 out of 40** **Different package taken into account** **21 out of 24** **According the substances**
	Existence of a governmental program for compassionate use for Rare Diseases	**Outcomes**	**X No** . Yes . In process

Theme 6 –Social Services for Rare Diseases

Sub-Themes

6.1. Social resources for people with disabilities

6.2. Specialised social services for rare diseases

6.3. Policies to integrate people living with rare diseases into daily life

6.4. International–supranational dimension

Issues around EUROPLAN indicators that are considered in current situation in Slovakia were answered in the speeches of Ing. Beáta Ramljaková, V vice President of the Slovak RD Alliance and president of DeRA SR, and of Vice President of the Neuromuscular Dystrophy Association Mária Duračinská.

Beáta Ramljaková mentioned that the National Strategy of RD in Slovak Republic for 2012 – 2013 was approved on 24th October 2012 by the Government of Slovakia. It is crucial for all stakeholders to be involved in this process in order to ensure good results. Who are the all stakeholders? Physicians, academics, researchers, pharmacists, pharma industry, national authorities, regulators, medical insurance companies, social insurance, medical and social workers, politicians, patients and patients organizations as well.

People affected by rare diseases face common challenges such as:

- Diagnostic delay
- Misdiagnosis
- Lack of information
- Lack of access to experts and standards of care
- Lack of choice of treatment
- Psychological stress
- Lack of practical support for everyday life

Patient organizations need to be active and work on following in the process of creating National Plan of RD in Slovakia:

1. There is a strong need to adopt a **holistic approach** to each individual citizen. For people living with rare diseases, care should not be only restricted to medical and paramedical aspects, but should also take into account social inclusion and psychological aspect. Patient organizations should closely cooperate with physicians, psychologists and social workers.
2. Patients and their leaders should be **involved in the decision-making process** in the field of rare diseases. It would be possible only if patients are equal partner for national authorities. Educated patients.
3. **Specialised social services** are important instrument to the empowerment of people living with rare diseases and improve their health and lives. Especially Respite Care

Services (RCS), Therapeutic Recreation Programmes (TRP), Adapted Housing (AH), Resource Centres (RC). RCS is provided on a short term basis for disabled or very ill people who either live at home or attend respite centre. Their caregivers can have a break from care giving. TRP for rare disease patients are organized as recreation activity which gives children possibility to have a fun and to take a break from thinking about the disease. AH allow people living with rare diseases to develop and enjoy some autonomy in their own home under supervision of staff. It is better than being placed in the institution. RC are new type of service. It is combination of information and social and medical services.

4. People living with rare diseases (PLWRD) need to be recognized and **integrated into existing social system** and to be provided with quality services in response to their needs. It is important to consider also other factors except from functional character of disorder, such as its chronic character and degenerative processes.
5. **Help lines** – the primary role of Help lines is to **provide information** and support to the callers.

Next activities of Patient Organizations (PO) in the process of creating NP RD SR are networking and cooperation between POs, raising awareness about RD, international cooperation (EURORDIS), spreading information through websites, educational activities, collaboration with media, releasing promotional materials.

Maria Duracinska (Neuromuscular Dystrophy Association) pointed out another challenge for the associations and issues to work on.

Organization of Muscular Dystrophy in the Slovak Republic (OMD in SR) is the only specific organization in Slovakia which associates children and adults with muscular dystrophy and other types of neuromuscular diseases (hereafter only NMD) and their families. The organization was founded as an independent public association in 1993. It has been providing social counselling and other specialized social services for its members and clients (patients, but also their families, pharmacists, staff from the recreational services, etc.). In its early years, it performed the community service and non-profit activities on a voluntary basis; today, a professional team of nine takes part in its activities, seven of which are people with extensive disability, plus two assistants. This change could take place thanks to having obtained the status of a protected workplace in 2009. In accordance with EURORDIS recommendations, OMD in SR takes care of activities in 3 different areas: information services and help line, therapeutic recreational programmes, and respite care services.

As a part of the information service, it provides specialized social counselling for disabled people via phone, email, in person or in the field − at a patient's place. The organization also publishes its own quality magazine *Ozvena (ECHO),* which is distributed free-of-charge to all members four times a year. People with NMD are involved in the creation of the magazine from the position of experts and patients. However it also supports the creation or translation publications for healthcare professionals.

The organization is particularly devoted to recreational activities, the camps for children with NMD, especially boys with Duchenne muscular dystrophy. Activities organized every year include also integrative national meetings of members.

The group of relief services includes providing specialized social services in the *Personal Assistance Services Agency* founded under OMD in SR. It provides wide-range service for 180 personal assistance users in Bratislava region; however, counselling covers the whole Slovakia.

For a long time, the organization has been making effort to change regulations in the legislation which concern several aspects essential for people with NMD.

OMD in SR is actively cooperating with the health care professionals. It is organising expert presentations, supporting the implementation of the patients in the interanational registries as well as educating health care professionals how to insert new patients into the registries. In their presentation they strongly support transparent granting system implementation into the National plan.

ACTIONS	INDICATORS	TYPE	ANSWERS
Compensating disabilities caused by rare diseases	**Existence of official programs supporting patients and families with disabilities**	**Process**	**.** Not existing, not clearly stated **X Existing, clearly stated, partly implemented and enforced** . Existing, clearly stated and substantially implemented and enforced
	Existence of an official directory of social resources for patients with disabilities	**Process**	**X Yes** **.** No . In preparation

ACTIONS	INDICATORS	TYPE	ANSWERS
Supporting social services aimed at rare disease patients and their families	**Existence of national schemes promoting access of RD patients and their families to Respite Care services**	**Process**	. Yes . Yes, and it includes financial support to patients / families **X No** . In preparation
	Existence of public schemes supporting Therapeutic Recreational Programmes	**Process**	**X Yes** **.** Yes, and it includes financial support to patients / families . No · I n preparation
	Existence of programmes to support integration of RD patients into daily life	**Process**	· Yes · Yes, and it includes financial support to patients / families **X No** · In preparation
Supporting rehabilitation programmes	**Existence of programmes to support rehabilitation of RD patients**	**Process**	· Yes · Yes, and it includes financial support **X No** · In preparation

Conclusion of the Final Report

The Slovak EUROPLAN National Conference - Forum of Experts, prepared as much as possible, according to the EUROPLAN guidelines, introduced a new format of conferences for most of the participants, who attended it. On one hand it was political, policy-creating and very broad, if we take into account all the discussed topic, on the other hand also scientific, because all the papers are published in English in the scientific journal of the Comeniu University (http://www.degruyter.com/view/j/afpuc.2012.59.issue-2/issue-files/afpuc.2012.59.issue-2.xml), thus increasing its European impact and creating conditions for future cooperation and participation in European projects.

It reached its main goal to connect all stakeholders, who might be involved in the creation of the national plan. It formed a big opportunity to share the information about the EUROPLAN projects and its measures, about the European Council Recommendation on rare diseases from 2009, about the Cross Border Directive as well as other European incentives. This fact is of big importance, because Slovakia is missing in most of the European projects.

Another added value of the conference was the direct contact and common attribute. All presentations were oriented to the main topic - rare diseases. Thus it allowed to know the specialist from a new point of view. SR does not have specialists oriented only in the field of rare diseases. However, as the conference established, several specialists faced rare diseases from all points of view: diagnosing, pharmacological and social care and thus as health care providers or patients, social workers or psychologist etc. The advantage of its organisation was also that most of the participants had the possiblity to join all presentations and discussions. This approach prevented misinterpretation or lack of interest and involvement. In a country, small as Slovakia, the comprehensiveness of the view is of big importance. Another advantage is that it might guarantee more fluency in the approach to rare diseases.

It clearly established that there is no extra space or support for rare diseases. The implementation of health care of RD patients into the current system has to be transparent and sustainable. To reach this goal it is important to know the current situation and thus from several points of view: political, regulatory, healthcare providers, healtcare payers, patients, as it was possible at the meeting. This is also the main topic of the interdepartmental working group.

On the 27th February, there was the first official meeting of the Interdepartmental Working Group. On one hand it was more convival, with less disscusion. On the other hand the speech of Prof M. Macek from Czech Republich was very challenging and inspirative. If we take into account the similarities and thus not only systemic but also personal, this could prevent several mistakes. For further development of the working group its status as well as abilities have to be stated by the MoH.

To conclude Slovak EUROPLAN National Conference - Forum of Experts was promissing and inspirative celebration of the Rare Disease Day 2013 in Slovakia. It increased the awareness between professionals as well as public. Paradoxically identified more problems than solved and helped to concentrate at the cardinal problems (CoE, Registries, European refference network). However this means success.

It stressed the importance of active participation in European projects and formed a competetive enviroment which is the best engine of the success.

Annexe I: Office meeting of the Interdepartmental working group for Rare Diseases of the Ministry of Health of the Slovak Republic

Wednesday, February 27th, 2013

Predsedníctvo/ Chair

Dorica Dan – EURORDIS (European Organisation for Rare Diseases - Európska mimovládna aliancia pacientskych organizácií a jednotlivcov)/ EUCERD

Prof. MUDr. Milan Macek, DrSc. – Univerzita Karlova, 2. Lekárska fakulta, Ústav biológie a lekárskej genetiky, prednosta/ EUCERD

MUDr. Anna Hlavatá, PhD. – DFNsP Bratislava, II. Detská klinika, zástupca prednostu/ Childern's Hospital, Bratislava

MUDr. František Cisarik, CSc. – FNsP Žilina, hlavný odborník MZ SR pre lekársku genetiku/ EUCERD

MUDr. Mario Mikloši, PhD. – MZ SR, Sekcia zdravia, generálny riaditeľ/ MoH SR, general manager, Department of Health

1:00pm - 1:30pm
Registration

1:30pm - 1:40pm
Welcome

1:40pm - 2:00pm
Interdepartmental Working Group in preparation of National Plan RD in SR
MUDr. Mario Mikloši, PhD.

2:00pm - 2:40pm
Theme of RD in Europe
Prof. MUDr. Milan Macek, DrSc.

2:40pm - 3:10pm
Coffee break

3:10pm - 3:40pm
Patient organizations in European context
Dorica Dan

3:40pm - 4:10pm
From National Strategy of improving medical care for people living with RD (PLWRD) to National Plan of development of care for patients with rare diseases in Slovak Republic
MUDr. František Cisárik, CSc.

4:10pm - 5:00pm
Proposal of Interdepartmental Working Group programe, Discussion, End

Outcomes from the meeting will be presented and processed next day during conference

Annexe II: Programme of the National Conference

Thursday, February 28th, 2013

EUROPLAN NATIONAL CONFERENCE
FORUM of EXPERTS

8:00 a.m. – 9:00 a.m.
REGISTRATION

9:00 a.m. – 9:15 a.m.
Opening remarks
Ing. Beáta Ramljaková, Slovak RD Alliance, DebRA SR
František Cisarik, MD., CSc. EUCERD, Ministry of Health SR
Dorica Dan, EURORDIS, EUCERD member
Doc. Dr. Ľudevít Kádáši, DrSc.
Prof. László Kovács, MD., DrSc., MPH

9:15 a.m. – 9:45 a.m.
The legislative tools for creating National plan for RD, current legislation and best practice
František Cisarik, MD., CSc. EUCERD, Ministry of Health SR

9:45 a.m. – 9:55 a.m.
EUROPLAN project
Dorica Dan, Amalia Egle Gentile, EURORDIS, EUCERD

9:55 a.m. – 10:20 a.m.
Role and importance of ORPHANET network, awareness, education, definition and codification of RD, Centers of Expertise
Prof. László Kovács, MD., DrSc., MPH, Gabriela Nagyová, MD.,Eszter Hegyi, MD., ORPHANET SK

10:20 a.m. – 10:40 a.m.
Health care reality for patients with RD – specifically with genetic metabolic

diseases in Slovakia
Anna Hlavatá, MD., PhD. MPH, Children's University Hospital Bratislava

10:40 a.m. – 11:00 a.m.
Pulmonary arterial hypertension Centre in Slovakia – "Ideal"of care. Diagnostics and management conditions of treatment in Slovakia, Quality criteria for Centers of expertise for RD
Assoc. Prof. Eva Goncalvesová, MD., CSc. - The National Institute of cardiovascular diseases, Bratislava

11:00 a.m. – 11:15 a.m.
COFFEE BREAK

11:15 a.m. – 11:35 a.m.
Orphan drugs in EU
PharmDr. Tatiana Foltánová, PhD, PharmDr. Ján Mazag, Comenius University in Bratislava, Faculty of Pharmacy, State Institute for Drug Control

11:35 a.m. – 11:55 a.m.
Orphan drugs in Slovakia
PharmDr. Ján Mazag, PharmDr. Tatiana Foltánová, PhD., State Institute for Drug Control, Comenius University in Bratislava, Faculty of Pharmacy

11:55a.m. – 12:15 a.m.
VŠZP access to patients with RD
Beata Havelková, MD., MPH, VŠZP – Public Health Insurance Company

12:15 p.m.– 12:30 p.m.
PANEL SESSION I.
Creation of the NP for RD, definition, information and education in the field of RD, orphan drugs, aim of EUROPLAN project in the SR, members of multidisciplinary working group and their tasks
F. Cisarik, D. Dan, L. Kovács, A. Hlavatá, E. Goncalvesová, D. Tomek, T. Foltánová, J. Mazag, B. Havelková,

12:30 p.m. – 1:50 p.m.
LUNCH

1:50 p.m. – 2:10 p.m.
Screening of RD
Prof. Svetozár Dluholucký MD., PhD, Dr. Mária Knapková, Children's University Hospital Banská Bystrica, Newborn Screening Centre

2:10 p.m. – 2:30 p.m.
The role of the National Health Information centre in availability of the information about rare diseases
Anna Baráková, MD., Peter Hlava MD., Zuzana Vallová M.Sc., NCZI – National Health Information Centre

2:30 p.m. – 2:50 p.m.
Role of patient organizations in the field of RD, Slovak RD Alliance mission in the context of EURORDIS
Ing. Beáta Ramljaková, Slovak RD Alliance, DebRA SR

2:50 p.m. – 3:10 p.m.
Special services for people with RD – practical experience
Andrea Madunová M. Sc., Mária Duračinská M.Sc., Patient organization of muscular dystrophy in the SR

3:10 p.m. – 3:30 p.m.
Psychological aspects of rare diseases
Assoc. Prof. Dr. Jitka Prevendárová, PhD., Comenius University in Bratislava, Faculty of Education

3:30 p.m. – 3:50 p.m.
PANEL SESSION II.
Screening and diagnostics of RD, National registry of RD, research and science in RD, Patient empowerment and specialized services for people living with RD
S. Dluholucký, M. Knapková, A. Baráková, M. Ondrejčák, B. Ramljaková, J. Prevendárová, M. Duračinská

3:50 p.m.– 4:00 p.m.
Closing remarks, end of conference
František Cisarik, MD., CSc. EUCERD, Ministry of Healtth SR

Annexe III: List of Participants

- Academic/Researcher 8/164 - 5%
- Clinician/GP.68/164 – 41%
- Healthcare Professional (other than clinician or GP) 13/164 - 8%
- Industry.. 15/164 9%
- Insurer 3/164 – 2%
- Medical /Learned society 22/164 - 13%
- *other health care worker (psychologist, speech therapist...) 9/164 – 5%*
- Patient representative 17/164 10%
- Politician
- Public administration (local, regional or national) 6/164 – 4%
- Social worker
- Other 11/164 – 7%.

1	Bert	Bartas	Clinician/GP
2	Angelika	Bátorová	Clinician/GP
3	Pavol	Baumgartner	Clinician/GP
4	Jana	Behunová	Clinician/GP
5	Zora	Brucháčová	Clinician/GP
6	Paulina	Bukovinová	Clinician/GP
7	Marta	Dobáková	Clinician/GP
8	Zuzana	Dobiašová	Clinician/GP
9	Darina	Ďurovčíková	Clinician/GP
10	Milan	Grofik	Clinician/GP
11	Katarína	Hálová	Clinician/GP
12	Zuzana	Havličeková	Clinician/GP
13	Eszter	Hegyi	Clinician/GP
14	Emilía	Kaiserová	Clinician/GP
15	Vlasta	Kákošová	Clinician/GP
16	Hana	Kayserová	Clinician/GP
17	Miriam	Kolníková	Clinician/GP
18	Peter	Kosoň	Clinician/GP
19	Adrian	Krátky	Clinician/GP

20	Zita	Kubová	Clinician/GP
21	Pavol	Kučera	Clinician/GP
22	Zuzana	Laluhová Striežencová	Clinician/GP
23	Lenka	Lisá	Clinician/GP
24	Martin	Matejka	Clinician/GP
25	Katarína	Melišová	Clinician/GP
26	Marián	Molnár	Clinician/GP
27	Gabriela	Nagyová	Clinician/GP
28	Michal	Ondrejčák	Clinician/GP
29	Mária	Ostrožlíková	Clinician/GP
30	Ivana	Plameňová	Clinician/GP
31	Beata	Slezáková	Clinician/GP
32	Juraj	Sokol	Clinician/GP
33	Peter	Špalek	Clinician/GP
34	Katarína	Štěpánková	Clinician/GP
35	Simona	Tárnoková	Clinician/GP
36	Miroslav	Urban	Clinician/GP
37	Iveta	Valachovičová	Clinician/GP
38	Adriana	Vrbjarová	Clinician/GP
39	Silvia	Vrbová	Clinician/GP
40	Jolana	Zlatošová	Clinician/GP
41	Eleonóra	Čmelová	Clinician/GP
42	Marcel	Repinský	Clinician/GP
43	Mária	Gvodjaková	Clinician/GP
44	Pavol	Sýkora	Clinician/GP
45	Katarína	Čolláková	Clinician/GP
46	Dagmar	Komadelová	Clinician/GP
47	László	Kovács	Clinician/GP
48	Svetozár	Dluholucký	Clinician/GP
49	Dagmar	Sorkovská	Clinician/GP
50	Oto	Urge	Clinician/GP
51	Monika	Václavíková	Clinician/GP
52	Miloš	Jeseňák	Clinician/GP
53	Veronika	Bencová	Clinician/GP
54	Ľuboš	Drgoňa	Clinician/GP
55	Katarína	Gaľová	Clinician/GP
56	Eva	Goncalvesová	Clinician/GP
57	Ivan	Martinka	Clinician/GP
58	Jozef	Rovenský	Clinician/GP, research
59	Jana	Šaligová	Clinician/GP
60	Marta	Kvasnicová	Clinician/GP
61	Ľubomír	Lisý	Clinician/GP, academy
62	Martina	Skokňová	Clinician/GP
63	Lukáš	Plank	Clinician/GP

64	Stanislav	Špánik	Clinician/GP
65	Alena	Počarovská	Clinician/GP
66	Eva	Korostenská	Clinician/GP
67	Anna	Hlavatá	Clinician/GP
68	Vladimír	Bzdúch	Clinician/GP, academy
69	Alica	Valachová	Healthcare Professional (other than clinician or GP)
70	Lucia	Babjaková	Healthcare Professional (other than clinician or GP)
71	Darina	Behúlová	Healthcare Professional (other than clinician or GP)
72	Martina	Hikkelová	Healthcare Professional (other than clinician or GP)
73	Darina	Holešová	Healthcare Professional (other than clinician or GP)
74	Petra	Jungová	Healthcare Professional (other than clinician or GP)
75	Mária	Knapková	Healthcare Professional (other than clinician or GP)
76	Anna	Šalingová	Healthcare Professional (other than clinician or GP)
77	Ľudevít	Kádaší	Healthcare Professional (other than clinician or GP)
78	Alena	Poľazovská	Healthcare Professional (other than clinician or GP)
79	Denisa	Ilenčíková	Healthcare Professional (other than clinician or GP)
80	Andrej	Genčík	Healthcare Professional (other than clinician or GP)
81	František	Cisarik	Healthcare Professional (other than clinician or GP)
82	Radoslav	Danko	industry
83	Janette	Fartelová	industry
84	Martin	Frčo	industry
85	Marcel	Kulich	industry
86	Radek	Korbel	industry
87	Ivan	Bakoss	industry
88	Július	Fíger	industry
89	Karin	Takačová	industry
90	Bibiana	Vallova	industry
91	Elena	Valovičová	industry
92	Jana	Meixnerová	industry
93	Michal	Kiripolský	industry
94	Jiří	Munk	industry
95	Peter	Beniač	industry

96	Jana	Meixnerova	industry
97	Anna	Ištv1vá	insurer
98	Beata	Havelková	insurer
99	Štefan	Kridla	insurer
100	Mária	Fischerová	Medical /Learned society
101	Beáta	Frčková	Medical /Learned society
102	Ján	Chandoga	Medical /Learned society
103	Mária	Klocháňová	Medical /Learned society
104	Jana	Konkoľová	Medical /Learned society
105	Ľubica	Krajčiová	Medical /Learned society
106	Lívia	Lukáčková	Medical /Learned society
107	Slavomíra	Mattošová	Medical /Learned society
108	Lucia	Minichová	Medical /Learned society
109	Iveta	Mlkvá	Medical /Learned society
110	Jana	Perečková	Medical /Learned society
111	Juraj	Šimko	Medical /Learned society
112	Hana	Zelinková	Medical /Learned society
113	Slávka	Machalová	Medical /Learned society
114	Michal	Konečný	Medical /Learned society
115	Roman	Dušinský	Patient representative
116	Gabriela	Markovova	Medical /Learned society
117	Zuzana	Michalková	Patient representative
118	Tatiana	Folt1vá	Medical /Learned society, academy
119	Veronika	Kollárovičová	Medical /Learned society, academy
120	Michaela	Korčeková	Medical /Learned society, academy
121	Magdaléna	Kuželová	Medical /Learned society, academy
122	Viliam	Foltán	Medical /Learned society, academy
123	Dominik	Tomek	Medical /Learned society, academy
124	Martina	Bekešová	other
125	Marián	Kročan	other
126	Magdaléna	Čeriová	other
127	Cecília	Marinová	other
128	Silvia	Sýkorová	other
129	Victoria	Headley	other
130	Ľudovít	Komadel	other
131	Vladimír	Pohánka	other
132	Anna	Markusová	other
133	Peter	Krajčovič	other
134	Amalia	Eagle	other
135	Dorica	Dan	other
136	Zuzana	Frčová	other health care worker (psychologist, diagnostician, speech therapist...)
137	Anna	Gabaríková	Patient representative
138	Ľubica	Kaiserová	other health care worker (psychologist, diagnostician, speech therapist...)

139	Andrea	Májková	other health care worker (psychologist, diagnostician, speech therapist...)
140	Andrea	Partlová	other health care worker (psychologist, diagnostician, speech therapist...)
141	Robert	Petrovič	other health care worker (psychologist, diagnostician, speech therapist...)
142	Gabriela	Svičeková	other health care worker (psychologist, diagnostician, speech therapist...)
143	Jitka	Prevendárová	other health care worker (psychologist, diagnostician, speech therapist...)
144	Katarína	Jariabková	other health care worker (psychologist, diagnostician, speech therapist...)
145	Mária	Duračinská	Patient representative
146	Jana	Guráňová	Patient representative
147	Radoslav	Herda	Patient representative
148	Alžbeta	Lukovičová	Patient representative
149	Andrea	Madunová	Patient representative
150	Vladimír	Makovník	Patient representative
151	Iveta	Makovníková	Patient representative
152	Ján	Mútala	Patient representative
153	Beáta	Ramljaková	Patient representative
154	Barbora	Zajackova	Patient representative
155	Libuša	Albertová	Patient representative
156	Katarína	Félixová	Patient representative
157	Kristína	Lacenová	Patient representative
158	Lýdia	Trubačová	Patient representative
159	Eva	Sladká	Public administration (local, regional or national)
160	Beata	Černáková	Public administration (local, regional or national)
161	Ján	Mazag	Public administration (local, regional or national)
162	Anna	Baráková	Public administration (local, regional or national)
163	Eva	Vivodová	Public administration (local, regional or national)
164	Zuzana	Ďurčíková	Public administration (local, regional or national)