LITHUANIA

EUROPLAN NATIONAL CONFERENCE

in the framework of the EU Joint Action RD-ACTION

Vilnius, 16 March 2018

FINAL REPORT

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FOREWORD

The EUROPLAN national conferences or workshops are organised in many European countries as part of a coordinated and joint European effort to foster the development of comprehensive National Plans or Strategies 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/ workshops are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS–Rare Diseases Europe. **Rare Disease National Alliances and Patient Organisations have a crucial role to shape the national policies for rare diseases.**

The strength of EUROPLAN national conference/ workshop lies in its shared philosophy and format:

- **Patient-led**: National Alliances are in the best position to address patients' needs;
- **Multi-stakeholders**: National Alliances ensure to invite all stakeholders involved for a broad debate;
- **Integrating both the national and European approach to rare disease policy;**
- **Being part of an overarching European action** (project or Joint Action) that provides the legitimacy and the framework for the organisation of EUROPLAN national conferences/workshops;
- **Helping national authorities adhere to the obligations stemming from the Council Recommendation of 8 June 2009 on an action in the field of rare diseases.**

Since 2008, National Alliances and EURORDIS have been involved in promoting the adoption and implementation of National Plans and Strategies for rare diseases. Altogether, 30 EUROPLAN national conferences took place in the framework of the first EUROPLAN project (2008-2011) and the EU Joint Action of the European Committee of Experts on Rare Diseases – EUCERD - (2012-2015).

Within RD-ACTION (2015-2018), the second EU Joint Action for rare diseases, National Alliances and EURORDIS continue to get involved in a coordinated European effort to advocate for and promote integrated national policy measures that have an impact on the lives of people living with rare diseases.

The EUROPLAN national conferences or workshops taking place within RD-ACTION focus on specific themes identified by the National Alliances as the most pressing priorities to tackle with national authorities. These thematic priorities are addressed in sessions where all the stakeholders discuss relevant measures to be taken or ways to sustain the full implementation of already approved measures.

Each National Alliance prepares a final report on the national workshop, based on a common format such as the one that follows.

GENERAL INFORMATION

Country	Lithuania
National Alliance (Organiser)	Vaikų retų ligų asociacija
Date & place of the national workshop/conference	"Crowne Plaza Vilnius", M. K. Čiurlionio 84, Vilnius, 16 March 2018
Website	https://www.rarediseases.lt/archyvas/konferencija-retu-ligu-gydymo-inovacijos/ http://www.creativa.lt/retosligos
Members of the Steering Committee	Rimantė Čerkauskienė, MD, PhD, prof. Birutė Tumienė, MD, PhD Elena Jurevičienė, MD Danas Čeilitka Rūta Udraitė-Mikalauskienė
List of Themes addressed	- State of the Art of the Lithuanian National Rare Disease Plan - Research and innovations in rare disease
Annexes :	I. Programme in English II. List of Participants (by stakeholders' categories)

FINAL REPORT

I. Introduction

The Lithuanian EUROPLAN national conference, organized within the framework of the 2nd EU Joint Action for rare diseases – RD-ACTION (2015-2018) and alongside with the annual 9th International Rare disease conference, took place in Vilnius, on 16 March 2018.

The patients were represented by two Lithuanian patient organizations - **Children Rare Disease Association** and **Spina Bifida and Hydrocephalus Association**, and **EURORDIS**. The audience also included representatives of the Ministry of Health, the Ministry of Education and Science, the National Health Insurance Fund, healthcare specialists and researchers of rare diseases from Lithuania, Czech Republic, Italy, Norway, Belgium, Germany, and medical students (including residents in training, PhD students and under-graduates).

The first session of the Conference was devoted to the National Rare Disease Plan: the state-of-the-art presentations, evaluation of the achievements of the last 5 years (2012-2017), and discussions on the actions that have to be implemented in the forthcoming 5 years. Although substantial developments and achievements marked this 5-year period, many problems still remain. At the end of the session, Memorandum of Actions was adopted.

The second session of the Conference was devoted to innovations and research in rare diseases and divided into three parallel sessions that covered three main themes: innovative rare disease patients' care, innovations in rare disease diagnostics and novelties in rare disease treatment.

Press conference was held during the conference with participation including patient representatives and professionals.

II. Themes

THEME 1

The National Rare Disease plan (NRDP) was adopted in 2012. It was developed by representatives of the Ministry of Health, patient organizations, university hospitals and universities. Within NRDP, a Plan of Actions for the period of 2012-2017 (5 years) was also adopted and responsibilities for the implementation of actions shared between the stakeholders.

During the period of 2012-2017, some major achievements in rare disease field took place in Lithuania:

- **Competence centres (CoC)** for rare diseases/groups of rare diseases were developed or formalized their activities in two university hospitals (**35** in the Vilnius University Hospital Santaros Klinikos (VUH Santaros Klinikos) and **21** in the Hospital of Lithuanian University of Health Sciences Kauno klinikos (LUHS));
- High quality of services provided in rare disease CoC was proven by inclusion of these centres into **European Reference Networks (ERN)**: VUH Santaros Klinikos is a True Member of **8 ERNs** (EuroBloodNet, ITHACA ERN, MetabERN, ERN-RND, ERKNet, eUROGEN, Transplant-Child, PaedCanERN), LUHS is a True Member of **4 ERNs** (ERN-EYE, EURACAN, ERN Skin, Endo ERN);
- **Coordinating Centres for Rare Diseases** were developed in two university hospitals – VUH Santaros Klinikos and LUHS - for coordination and administration of rare disease care and CoC activities;
- **Rare disease patient organizations** were created and National Alliance of rare diseases is being developed;
- **Diagnostics** of rare genetic diseases substantially improved: genomic testing methods, including molecular karyotyping and next generation sequencing, were introduced into clinical practice and regulations for reimbursement of these tests were established;
- Patient-centered, multidisciplinary, integrated and holistic **care models** for rare diseases in CoC are being developed;
- Extensive participation in rare disease **international activities** (VUH Santaros Klinikos is a national Orphanet Coordinator from 2004 and a partner of RD-Action, JARC and many other rare disease-related joint actions, platforms, projects, registries, etc.) enabled transfer of EU policies and best practices into national systems;
- Established bilateral **collaborations between CoC and patient organizations**, including common conferences, discussions, patient day-schools and campuses, awareness-raising events, etc. helps to ensure patient-empowerment and patient-centeredness;
- In Lithuania, the first **Rare Disease Day** was celebrated on the 29th of February, 2008 in VUH Santaros Klinikos, at the time when the first celebrations took place in the world. From that time, extensive dissemination activities substantially increased awareness of rare diseases amongst general public and decision-makers;

- Inclusion of rare disease **teaching** into the educational programs of all-level medical studies (including undergraduate, residents-in-training, PhD studies) and continuous medical education (CME) courses increase awareness of rare diseases among medical community;
- E-health, telemedicine and IT technological solutions are increasingly used for rare disease patient care.

However, many **problems** still have to be solved, including:

- Lack of proper **acknowledgement and prioritization** of rare disease-related problems in national legal acts;
- Lack of proper **intersectoral collaboration** (including national health, social, educational, research systems) in rare disease field;
- Improvement of rare disease patient **care coordination** along the whole clinical pathway, with special relevance given to proper engagement of primary care and regional physicians and social care;
- **Financial sustainability of CoC** and appropriate reimbursement for complex, multidisciplinary services provided in CoC;
- Coordinated approach to **undiagnosed disease** problems, including solutions for patients that have not yet been diagnosed ("stacked" in health system) and those with truly undiagnosed diseases;
- Expansion of **neonatal screening** for congenital disorders (currently, Lithuanian newborn screening includes 4 congenital disorders).

During the first session of the Conference, presentations on evaluation of rare disease activities and NRDP accomplishments were given by the representatives of the Ministry of Health (Ms A.Balčiūnienė), Children Rare Disease Association (Mr D.Čeilitka), VUH Santaros Klinikos (prof.R.Čerkauskienė, Ms E.Jurevičienė). Representative of the Ministry of Education and Science gave a presentation on integrative teaching (Ms R.Labinienė). Extensive presentation about coordination of rare disease activities and recent achievements in Czech Republic was given by prof. M.Macek, the Head of the National Coordination Centre for Rare Diseases in Prague. The EURORDIS representative, Ms Ariane Weinman, provided a presentation about the role of rare disease patient organizations in multiple dimensions of rare disease field. A review and evaluation of rare disease national plans and strategies in Europe, including 17 rare disease NP/NS, was given by dr.B.Tumienė (VUH Santaros Klinikos). Discussions and propositions at the end of the first session were finalized with the adoption of the **Memorandum of Actions** including the following issues:

- Rare disease acknowledgement and prioritization.
- Improvement of coordination of healthcare for rare disease patients.
- Further development of Competence Centres in the university hospitals.
- Assurance of timely access to high quality healthcare services for rare disease patients.
- Improvement of healthcare service accessibility for rare disease patients by assurance of proper reimbursement and regulations of healthcare services.
- Coordinated approach to problems related to undiagnosed diseases.
- Expansion of neonatal screening program for congenital diseases.
- Collaboration with Lithuanian and international educational and research institutions.

- Active participation of patient representatives and communities in decision taking and spreading of information on rare diseases.
- Dissemination of information on rare diseases and national/international activities in the field.

Welcome addresses of the conference: speaking – Viceminister of the Ministry of Health of the Republic of Lithuania Kristina Garuolienė; moderators: prof.R.Čerkauskienė, dr.B.Tumienė, E.Jurevičienė.

The main organizers of the conference: left to right – prof.A.Utkus, E.jurevičienė, dr.B.Tumienė, prof.R.Čerkauskienė, D.Čeilitka.

THEME 2

The second session of the Conference was divided into three parallel sessions.

The **first parallel session** was led by spina bifida patient organization and Children Rare Disease Association and included presentations on spina bifida patients' rehabilitation, multidisciplinary care, functional mobility possibilities, prevention and management of secondary health conditions, urotherapy, psychosocial care, gait assessment and correction measures. During and after the session, there were extensive discussions on best practices in other European countries and possibilities to transfer these practices to Lithuania.

Author: Birutė Tumienė

In the **second parallel session,** led by prof. A.Utkus (Vilnius University, Faculty of Medicine; VUH Santaros Klinikos), innovations in diagnostics of rare diseases were discussed. The use of artificial intelligence in 3D facial gestalt analysis in dysmorphology and diagnostics of genetic syndromes was presented by prof. M.Macek (Czech Republic). Strategies of stem cell use in regenerative medicine were provided by dr.E.Bernotienė (Centre for Innovative Medicine). Multiple developments, achievements and innovations in genetic disease diagnostics were presented by representatives from VUH Santaros Klinikos, including preimplantation genetic diagnostics (dr.L.Ambrozaitytė), pharmacogenetic testing (Mr K.Baronas), molecular karyotyping (Ms D.Braždžiūnaitė), genomic testing in epilepsy (dr.B.Tumienė), dysmorphology evaluation (dr.A.Matulevičienė) and applications of artificial intelligence in genetic disease diagnostics and research (Mr K.Šablauskas).

Speakers and participants of the second parallel session: the first row, left to right - dr.V.Šliužas, K.Šablauskas, D.Braždžiūnaitė, K.Baronas, dr.B.Tumienė, dr.L.Ambrozaitytė, prof.A.Utkus; the second row – dr.A.Matulevičienė, dr.A.Morkūnienė, L.Bikauskaitė, K.Grigalionienė, M.Smirnova.

In the **third parallel session**, innovations in rare disease treatment were discussed. Novel insights in CKD and PD induced alterations of the peritoneal membrane were provided by prof.B.Shaefer (Germany). A new form of hemolytic uremic syndrome, DGKE nephropathy, was presented by Mr K.Ažukaitis (VUH Santaros Klinikos). Gene engineering and synthetic biology methods for the treatment of rare diseases prior to their manifestation were presented by Mr G.Jakutis (Vilnius University, Faculty of Medicine). A study of cognitive functions in children with spina bifida was presented by Dr. M.Hoff (Norway). Afterwards, several case presentations were given by speakers from VUH Santaros Klinikos (dr.J.Rascon, Ms A.Judicikienė) and LUHS (assoc.prof.E.Danytė).

III. Conclusions

Many important developments in the field of rare diseases took place in Lithuania in 2012-2017, during the first implementation period of the National Rare Disease Plan. Notwithstanding, in pursuance of these achievements to make a real difference for every Lithuanian rare disease patient and turn into better outcomes and improvements of their health and quality of life, many important issues remain to be solved. During this Conference, commitment of various rare disease stakeholders to strive for these common goals were expressed by adoption of Memorandum of Actions.

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Minutes from press conference: left to right – L.Rudokienė, D.Čeilitka (patient representatives), K.Šablauskas, dr.B.Tumienė, prof.R.Čerkauskienė, prof.A.Utkus.

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APPENDIX I: PROGRAMME

IXth INTERNATIONAL SCIENTIFIC CONFERENCE
"INNOVATIONS IN THE MANAGEMENT OF RARE DISEASES"

CONFERENCE VENUE: CROWNE PLAZA VILNIUS (M.K. ČIURLIONIO STR. 84, VILNIUS)
16 MARCH 2018

09:00–09:30	Registration
09:30–10:00	Welcome and opening Ministry of Health of the Republic of Lithuania; Ministry of Education and Science of the Republic of Lithuania; VUH Santaros Klinikos; EURORDIS representative.
10:00–10:30	Lithuanian national rare diseases plan: accomplished tasks and future challenges Anželika Balčiūnienė, Ministry of Health of the Republic of Lithuania
10:30–10:45	Patient organization activities: 2 years experience and further aspirations Danas Čeilitka, president of the National Rare Diseases Alliance
10:45–11:00	Integrative teaching Regina Labinienė, Ministry of Education and Science of the Republic of Lithuania
11:00–11:30	Coffee break
11:30–12:00	Coordination of rare disease activities in Czech Republic: National Coordination centre for rare diseases Prof.Milan Macek, Prague, Czech Republic
12:00–12:20	EURORDIS: role of rare disease patient organizations Ariane Weinman, EURORDIS
12:20–12:45	Coordination of rare disease activities in VUH Santaros Klinikos Prof. Rimantė Čerkauskienė, MD Elena Jurevičienė, VUH Santaros Klinikos
12:45–13:00	Review of rare diseases national plans and strategies in Europe Dr. Birutė Tumienė, VUH Santaros Klinikos
13:00–13:30	Discussions, conclusions, suggestions for memorandum, formation of memorandum work group
13:30–14:30	Lunch
14:30–18:00	Parallel sessions

TOGETHER WE CAN ACHIEVE MORE (SPINA BIFIDA)

Chair: Rūta Udraitė-Mikalauskienė, Danas Čeilitka

14:30-14:45	Rehabilitation clinic "Spinalis" (Stockholm, Sweden): multidisciplinary approach to rehabilitation of adults with spinal cord injury. Vilija Šatienė, member of SBH association administration
14:45-15:00	Functional mobility possibilities in children with spina bifida. Elena Slobodyanik, chairperson of SBH association
15:00-15:30	Overview of Spina Bifida and Hydrocephalus patients multidisciplinary care systems in Europe. Janina Arsenjeva, The International Federation for Spina Bifida and Hydrocephalus (IF), Europe Programme Manager, Belgium
15:30-16:00	Prevention and management of secondary health conditions in children with Spina Bifida: a systematic approach. Marit Rekkedal Edvardsen, physical therapist, Akershus university hospital, Norway
16:00-16:30	Coffee break
16:30-17:00	Methods and measures used in follow-up programs in Norway in relation to maintaining renal function. Using aid and medication to get the best possible control Vibeke Haahr, urotherapist, Akershus university hospital, Norway
17:00-17:15	Psychosocial approach to rehabilitation, Kaunas Clinics Pediatric rehabilitation clinic "Lopšelis" experience. Indrė Bakanienė, Pediatric rehabilitation clinic„Lopšelis", head of the department, pediatric neurologist
17:15-17:30	Neuropsychological features of spina bifida Rūta Vyšniauskė, Children's Hospital VUH Santaros Klinikos
17:30 -18:00	Gait assessment and correction in children. Dr.Aurelijus Domeika, head of KTU Institute of Mechatronics Biomechatronics laboratory; Eugenijus Mačiukas, „Aurelka" orthopedic shoes representative for Baltic states

THE FIRST STEP: DIAGNOSTICS OF RARE DISEASES

Chair: prof. Algirdas Utkus

14:30-15:00	3D facial gestalt analysis in dysmorphology. Prof. Milan Macek, Prague, CzechRepublic.
15:00-15:20	Preimplantation genetic diagnostics-Lithuanian status. Dr. Laima Ambrozaitytė, Kristina Grigalionienė, VUH SK
15:20-15:40	Strategies of stem cell use in regenerative medicine. Dr. Eiva Bernotienė, Centre for Innovative Medicine
15:40-16:00	Clinical applications in pharmacogenetic testing. Karolis Baronas, VUH SK
16:00–16:20	Coffee break
16:20–16:40	Dysmorphology today. Dr. Aušra Matulevičienė, VUH SK
16:40-17:00	The use of artificial intelligence in rare disease diagnostics. Karolis Šablauskas, VUH SK
17:00–17:20	Genomic testing in epilepsy diagnostics. Dr. Birutė Tumienė, VUH SK
17:20-17:40	Molecular karyotyping: clinical utility and practice. Deimantė Braždžiūnaitė, VUH SK
17:40-18:00	Discussions

PATH TO THE FUTURE: A NEW APPROACH TO RARE DISEASES

Chair: prof. Augustina Jankauskienė

14:30-15:00	Novel insights in CKD and PD induced alterations of the peritoneal membrane. Betti Shaefer, Germany.
15:00-15:20	DGKE nephropathy: a new form of hemolytic uremic syndrome. Karolis Ažukaitis, Vilnius University Faculty of Medicine
15:20-15:40	Gene engineering and synthetic biology methods to treat rare diseases prior to their manifestation. Gabrielius Jakutis, Vilnius University Faculty of Medicine, Vilnius-Lithuania iGEM, Society of Innovative Medicine
15:40-16:10	Cognitive function in children with spina bifida and the results of pilot study on goal management training. Dr.Marie Hoff, TRS National Resource Centre for Rare Disorders, Sunnaas Hospital, Norway
16:10-16:30	Coffee break
16:30-16:50	Atypical Presentation of Griscelli Type 2 Syndrome: Lithuanian Experience Dr.Jelena Rascon, Children‘s Hospital VUH Santaros Klinikos
16:50-17:10	Batten disease. Asta Judickienė, MD, Children‘s Hospital VUH Santaros Klinikos
17:10-17:30	Rare types of diabetes. Assoc. prof. Evalda Danytė, LUHS Kaunas Clinics
17:30-18:00	Discussions

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APPENDIX II: List of participants by stakeholders' categories

Patient representatives	16
Representatives of public institutions	4
Health care providers/ researchers	180
EURORDIS	1
Total	**201**