SLOVENIA
EUROPLAN NATIONAL CONFERENCE
in the framework of the EU Joint Action RD-ACTION

28 February 2018, Brdo pri Kranju
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 and workshops are jointly organised in each country by a National Alliances of rare disease patients' organisations and EURORDIS–Rare Diseases Europe. **National Alliances for Rare Diseases and Patient Organisations have a crucial role to shape the national policies for rare diseases.**
The strength of EUROPLAN the 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 and 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, 41 EUROPLAN national conferences took place in the framework of the first EUROPLAN project (2008-2011) and the EU Joint Action of the European Union 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 and 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	Slovenia
National Alliance (Organiser)	Rare Disease Association of Slovenia
Date & place of the national workshop/conference	28 February 2018, Brdo pri Kranju
Website	www.zrbs.si
Members of the Steering Committee	Robert Medved (Ministry of Health) Jože Faganel (President of the Rare Disease Association of Slovenia; President of the Slovenian Haemophilia Society) Majda Slapar (Vice president of the Rare Disease Association of Slovenia) Tea Černigoj Pušnjak (Secretary of the Rare Disease Association of Slovenia; Representative of the Muscular Dystrophy Association of Slovenia)
List of Themes addressed	European reference networks National contact point for rare diseases The aspects of financing treatment of rare diseases in Slovenia Rare diseases Registry Screening tests Ethical aspects of rare diseases The legal basis for dealing with rare diseases Rare diseases in psychiatry Round table – Living with rare disease
Annexes :	I. Programme in English II. List of Participants (by stakeholders' categories)

FINAL REPORT

I. Introduction/ Plenary session

Official opening

The 4th National Conference on Rare Diseases was opened on 28 February in Brdo pri Kranju, Slovenia, with a speech from the representative of the Ministry of Health, Milojka Kolar Celarc.

The Conference was organised by the Rare Disease Association of Slovenia, in cooperation with the Ministry of Health, European Organisation for Rare Diseases – EURORDIS, Clinical Institute of Medical Genetics, UMC Ljubljana, and the Paediatric Clinic in Ljubljana. It was organised under the auspices of the President of the Republic of Slovenia, Borut Pahor and the Minister of Health, Milojka Kolar Celarc.

The Conference lasted one day on 28 February with the participation of about 150 participants (members of the Alliance, representatives of Slovenian patient organisations, medical experts, representatives of institutions and representatives of the pharmaceutical industry).

Theme 1 - European reference networks

1. *Prof. Borut Peterlin, MD PhD (Clinical Institute of Medical Genetics, UMC Ljubljana)*

On the basis of Article 12 of the EU Directive on the application of patients' rights in cross-border healthcare, the European Commission adopted a decision defining the establishment of European Reference Networks (ERNs), which will provide better quality and cost-effective healthcare for patients with conditions requiring a special concentration of resources and expertise.

Prof. Borut Peterlin, MD PhD said that as a small country, Slovenia has a great interest in joining the European Reference Networks, which provide access to state-of-the-art health services to every individual, regardless of where they live in the European Union. Namely, in a small country, it is very difficult to provide experts for every single rare disease.

At the European level, 24 reference networks have been established. Nine health care providers in Slovenia have applied for nine different networks. All nine providers were successful and have managed to get included. For such a small nation as Slovenia, the successful candidacy of nine healthcare providers for the ERNs is a great achievement.

Theme 2 - National contact point for rare diseases

1. *Assist. prof. Mojca Žerjav Tanšek, MD PhD (Paediatric Clinic, UMC Ljubljana)*

One of the most important achievements in the last period was the presentation of the successfully completed project of the National Contact Point for Rare Diseases, presented by assist. prof. Mojca Žerjav Tanšek, MD PhD. The ultimate goal of the project was to establish the website www.RedkeBolezni.si and an information point that will allow for future activities and contacts among patients with rare diseases and among healthcare professionals.
The website includes a list and information on institutions for rare diseases in Slovenia, contacts of existing patient societies, and a collection of major international online links. The website also includes the possibility of adding new online contacts for activities of individual rare diseases or rare disease groups, where relatives or patients can offer their own contact as a possibility for the formation of new societies and to begin building interactive cooperation with other patients and families.
In 2017, the financing of the Slovenian national contact point for rare diseases was passed with the assistance of the Ministry of Health of the Republic of Slovenia in a joint agreement with the Ljubljana University Medical Centre (UMC), which enabled the employment of a graduate nurse for office hours. The presence of a contact person enables continuous telephone and electronic contact during office hours from Monday to Friday between 11 am and 3 pm.

Experience in establishing the Slovenian contact point for rare diseases

- The goal of the point is to inform patients, their families and healthcare professionals about rare diseases, but includes crucial bilateral cooperation, as patients and societies need to co-create the website and promote its development. Without the active participation of the co-creating users, the point will not flourish.
- In 2017, there were almost no contacts or other activities by Slovenian health workers. Although other similar lines of aid for rare diseases identify a low share of active inquiries by health professionals (both in the case of inquiries about rare diseases and in the transmission of new information), we conclude that it is necessary to further increase visibility. As the contact point does not have the means to advertise in the media, it is important that the users themselves are promoters of its visibility.
- A share of inquiries at the contact point is related to social rights and the rights of chronically ill children. Although the basic documents are posted on the website, legal assistance and the assistance of social workers will need to be considered. The contact point also raises the question of volunteering, which in the European area is a frequent form of aid in the field of rare diseases, especially with telephone helplines. The involvement of volunteers in the counselling could contribute to the better functioning of the national point and achieve one of its main purposes: connecting and helping patients, their relatives and health care professionals on their journey with a rare illness.
- Published analyses on the type and extent of information on individual rare diseases available on informational websites show that the content must be brief and understandable, while at the same time, patients have slightly differing expectations on the key sources and information on diseases to professionals.

Theme 3 - The aspects of financing treatment of rare diseases in Slovenia

1. *Marjan Sušelj, MSc (Health Insurance Institute of Slovenia)*

According to Marjan Sušelj, MSc the director of the only public health insurance company (HIIS), they finance all the necessary specific medicines with one-seventh of their expenditures, which due to their small number are exorbitantly expensive. However they do intend to connect with similar insurance companies in Europe and apply for joint orders, which will effectively curb the prices.

Theme 4 - Rare diseases registry

1. *Eva Murko, MD PhD (National Institute of Public Health): Rare diseases registry at NIPH. How far are we?*
2. *Assist. prof. Damjan Osredkar, MD PhD (Paediatric Clinic, UMC Ljubljana): Neuromuscular disease registry (example of a comprehensive treatment registry)*
3. *Assist. prof. Ana Kotnik Pirš, MD PhD (Paediatric Clinic, UMC Ljubljana): Comprehensive treatment of rare congenital pulmonary diseases*

According to rough estimates, there are around 150,000 patients with rare diseases in Slovenia (Slovenia's population is estimated at 2.08 million). In order to properly formulate health policies,

evaluate the effects of health policies and monitor patients with rare diseases, it is essential to have epidemiological data, including data on the treatment and use of orphan drugs.
The strategic importance of rare diseases registries is highlighted by the European Commission in the 2008 document 'Rare Diseases: Europe's challenges'. The importance of such registers is also emphasised in various development and strategic documents by the former EUCERD (European Union Committee of experts on rare diseases) and the European Commission Expert Group on rare diseases. National rare diseases registries already exist in France, Italy, Spain, Slovakia and in Belgium. A Bulgarian national rare diseases registry is currently in preparation.

Eva Murko, MD PhD of the Institute of Public Health said that the national rare diseases registry in Slovenia has not yet been established, although its establishment is listed as one of the key activities of the Work plan in the field of rare diseases in Slovenia. In line with the strategic indications of the rare diseases Work plan, a targeted research project titled 'Analysis and development in the field of the rare diseases in Slovenia' was held between October 2015 and September 2017, as part of which the concept for a national system for the monitoring of rare diseases was planned, as well as the development of a pilot rare diseases registry. The pilot rare diseases registry was conceived as a web-based application by which competent sub-specialised healthcare providers would report data associated with a diagnosed rare disease. During the development of the pilot rare diseases registry, five reporting institutions participated in the project, both in the concept of the pilot rare diseases registry as well as in its testing and final optimisation. The construction of the pilot rare disease registry was based on the OpenEHR methodology, which in recent years is increasingly being used to achieve semantic interoperability in the field of healthcare, and was also used in the PARENT JA project for the creation of the Arthroplasty Registry of Slovenia. Renowned Slovenian experts in the field of rare diseases participated in the project for the preparation of the pilot rare diseases registry.

The World Health Organization is preparing the 11th edition of the ICD, which contains in its basic layer approximately 5,400 codes for rare diseases and which is expected to be issued in 2018. The 11th edition, with adaptations, is expected to be accepted by Slovenia, which will make coding of rare diseases more precise and will enable us to better understand the incidence of these diseases.

The current Healthcare Databases Act (Official Gazette of the Republic of Slovenia, nos. 65/00 and 47/15) does not provide a legal basis for the collection of data on patients with rare diseases. Between November 2017 and December 2017, a proposal for the Act Amending the Healthcare Databases Act, including the non-malignant rare diseases registry, was held for public hearing. Currently at the time of this conference, the proposal is in further legislative procedure with the Government of the Republic of Slovenia.

Assist. prof. Damjan Osredkar, MD PhD presented the Children's neuromuscular disorders registry as an example of good practice and stressed that for the affected children and their families, as well as for society as a whole, the burden of these diseases is very high. With a precisely managed registry, they have a good overview of the situation of children with neuromuscular diseases in Slovenia, and at the same time they can quickly respond to initiatives for participation in various researches in which patients are offered new forms of treatment. At the Paediatric Clinic in Ljubljana, the first children with spinal muscular atrophy (SMA) are already being treated with nusinersen and children with Duchenne muscular dystrophy (DMD) are treated with ataluren, which alters the expression of genes, thereby inhibiting the progression of the underlying disease.

The emerging rare diseases registry will consolidate the good experiences of existing registries to 'all' rare diseases. The so-called house registers of individual departments, as the Children's neuromuscular disorders registry presented by assist. prof. Damjan Osredkar, MD PhD that is operated with foreign knowledge and support, must not only connect, but provide a legal basis.

Theme 5 - Screening tests

1. *Prof. Tadej Battelino, MD PhD with colleagues (Paediatric Clinic, UMC Ljubljana)*

How to diagnose congenital diseases as soon as possible was presented by Prof. Tadej Battelino, MD PhD. In Slovenia, for more than 30 years the programme of neonatal biochemical screening includes only two diseases (phenylketonuria and congenital hypothyroidism), thus placing Slovenia at the bottom of the scale.
In 2017, the Paediatric clinic at UMC Ljubljana began to undertake activities for the purchase of the necessary new MS/MS equipment, which will allow the programme of expanded neonatal screening by about 20 diseases to begin operating in the second half of 2018. The Ministry of Health is also preparing an appropriate policy change.
The new programme will bring a greater chance (probability) of discovering IEMs and/or faster treatment; the removal or reduction of hardly manageable symptoms and problems; prevention or reduction of secondary illness; better control of chronic disease; prevention or reduction of disability; increasing life years with less disease burdens; slowing down the course of an incurable disease and preventing premature death (prolongation of survival); increasing self-sufficiency (movement, eating, personal hygiene), normal working and family activities and enjoying free time; added physical or mental health and higher quality of life.

Theme 6 - Ethical aspects of rare diseases

1. *Prof. Božidar Voljč, MD PhD (The National Medical Ethics Committee of the Republic of Slovenia): The role of the National Medical Ethics Committee in dealing with rare diseases*
2. *Simona Borštnar, MD PhD, Miha Oražem, Professor, Božidar Voljč, MD PhD (Institute of Oncology, UMC Ljubljana): Patient involvement in research - presentation of the situation and dilemmas*
3. *Prof. Lenart Škof (Science and Research Centre of Koper): A Philosopher Before the Ethical Aspects of Rare Diseases*

Prof. Božidar Voljč, MD PhD accurately demonstrated to all participants the high standards supported by the National Ethics Committee at the Ministry of Health in the field of medicine, and especially in their relation to patients with rare diseases.

Miha Oražem, MD PhD from the Institute of Oncology picturesquely presented the realisation that patients with rare diseases should be included in research not only as 'guinea pigs', but as partners, as it may happen that a patient involved with the research will offer an original idea.

Prof. Lenart Škof kept his contemplation on ethics in the philosophical field and linked it to the problems of the status of rare diseases within society, and health and scientific policies. With this, he highlighted the role of the principles of justice and solidarity in treating rare diseases, while also touching some of the more serious ethical aspects of bioethics that affect the position of individuals - as subjects that are subject to different policies and attitudes within a democratic community.

Theme 7 – The legal basis for dealing with rare diseases

1. *Barbara Jan Bufon, MSc*

Barbara Jan Bufon, MSc discussed how legal regulation in the field of health is lagging behind the actual state of modern treatment in Slovenia. She underlined the need for the normative regulation of education of laypersons in difficult self-treatment procedures, which, for certain rare diseases, entrust the treatments and medicines intended only for inpatient treatment to the patient and those closest to them (both laypersons) for self-treatment at home (self-infusion, peritoneal dialysis, respirators).
Specific standards should be set, the compliance of these monitored, and documentation should be requisite. Lastly she mentioned that the Medical Chamber of Slovenia, with the authorisation it has in the field of health policy, will also have to regulate the range and nomenclature of specialisations in relation to the modern state of medicine, especially for the problem of rare diseases. Consequently, specialists with a license for a specific rare disease will be able to treat patients from birth to old age, because they will no longer hold only a paediatrician or an internist license, but rather a license for a specific disease, including rare diseases.

Theme 8 – Rare diseases in psychiatry

1. *Assist. prof. Marko Pišljar, MD PhD (Psychiatric Hospital Idrija)*

Assist. prof. Marko Pišljar, MD PhD from the Psychiatric Hospital Idrija presented a previously unrecognised chapter of rare diseases in psychiatry. New lessons in the field of rare disease research are becoming increasingly important for psychiatric clinical practice. With differential diagnosis, a psychiatrist must seriously consider the possibility of a rare disease, rather than establish a definitive diagnosis.
Some signs and symptoms may indicate a higher probability of a rare disease: the occurrence of a mental disorder in an atypical age, pronounced personality changes, a sudden onset of symptoms in a previously asymptomatic person, the emergence of new psychiatric symptoms with a gradual decline in cognitive abilities, episodes of stereotypical behaviour, co-occurrence of bodily or focal neurological signs, the history of rare disease in the family.
We can use genetic research on rare diseases to understand common mental disorders. Similarly, as neurological studies of focal brain injury allow for a better understanding of the functioning of the central nervous system, the disclosure of genetic changes and mutations can serve as a model of biological changes that are in the background of normal mental functioning or disease activity.
The field of rare mental disorders needs greater attention from the profession, proper development of diagnostic procedures, treatment and rehabilitation possibilities.

Theme 9 – Living with a rare disease

Round table (Moderator: Jože Faganel)

Rare diseases are extremely complex diseases, which was also presented at this year's round table entitled '*Living With a Rare Disease*'. Patients with transthyretin amyloid polyneuropathy (TTR-FAP) and hereditary angioedema were introduced at the table with their doctors - prof. Janez Zidar, MD PhD, prim. Jože Pretnar, MD PhD and prof. Mitja Košnik, MD PhD with a written contribution.
Rare diseases strongly perturb the quality of life of patients and their relatives. In addition to the psychosocial impact on patients of this type of disease, they usually require demanding medical care, which often involves complex and lengthy diagnostic procedures. Consequently, living with a rare disease becomes a daily learning experience for patients, families and carers who are faced with issues such as how to get treatment, how to define and how to manage the financial burden of everyday life with a rare disease. Each participant presented a unique and singular path from diagnosis, dealing with the first symptoms, accepting the disease, and facing the challenges and trials on their way. Their trodden paths successfully included the topical issues and invalidated the Roman phrase '*mens sana in corpore sano*' (a sound mind in a sound body). According to Jože Faganel, the President of the Rare Disease Association, with the help of current modern medicine, the creative 'spirit' can now live and thrive in a 'sick body'.

Conclusions

Indeed, why should Slovenia not compare itself in the treatment of rare diseases with France, whose system of predominantly public healthcare facilitates its foremost position in the field? Prof. Borut Peterlin, MD PhD presented Slovenia's effective inclusion in the so-called reference networks, internet channels freeing patients from having to visit foreign centres, since medical professionals can harmonise their activities locally, focusing on providing care closer to home. Consequently, Peterlin's centre at the Division of Gynaecology and Obstetrics UMC in Ljubljana now independently diagnoses practically all the known rare diseases. Assistant Professor, Mojca Žerjav Tanšek, MD PhD showcased the functioning of the national contact point for rare diseases at the Paediatric Clinic. This information system directs concerned patients or parents, by email or phone, to a competent professional or patient organisation through a dedicated lane, bypassing the paralysing system of e-referrals and inspectors tasked with scolding primary care providers for unsuccessfully treated diagnoses. Fortunately, exceptional conditions are not addressed with Lady's Mantle (attributed alchemical health benefits throughout the ages) any longer, but with modern medical measures.
According to the director of our only public health insurance company (HIIS), Marjan Sušelj, MSc, the latter finances, through one seventh of their expenses, all the necessary specific medications, at costs that are indeed exorbitant due to their small order volumes. To keep the prices in check, they are planning to collaborate with similar insurance organisations throughout Europe, placing bulk orders together. Naturally, the supply of medications and planning of treatment cannot be managed without a register of rare diseases. Eva Murko, MD PhD from the Institute of Public Health stressed that relevant databases must be designed to include a display of individual treatment. In Slovenia during the past two decades, this has only been effectively legislated for blood clotting disorders among the rare diseases, while just two such functioning registries exist for widespread diseases (cancer since 1951) or malformations. The emerging registry of rare diseases will extrapolate the good practices of the existing registers onto 'all' the rare conditions.

House registers of individual departments, like the foreign-funded and conceived registry of neuromuscular disorders in children presented by assist. prof. Damjan Osredkar, MD PhD ought not only to connect with one another, but join in making sure a proper legislative foundation is set up for their operation. Namely, the informatisation of sensitive personal data, which might even include an individual's genome, is at major future risk of potential misuse. According to the theme for Rare Disease Day 2018, patients are encouraged to set up and help maintain registries themselves and directly provide data to researchers regarding their health condition.

Traditionally, the most deadly pulmonary conditions (counting aside allergies) were lung cancer and tuberculosis. Today's paediatricians, though, encounter unusual rare diseases endangering the lives of the infants. Paediatrician assist. dr. Ana Kotnik Pirš, MD PhD presented the case of cystic fibrosis, in which the insertion of pancreas enzymes prevents the onset of at least some of the formerly unmanageable complications. Due to the once-poor outcomes of diseases not promptly treated, young patients who are appropriately cared for during childhood might encounter inexperienced adult primary care physicians, who are consequently inconsiderate in this regard. This is why developed countries generally do not involve transfers from paediatricians to internists, but in the best interest of the patient, establish cooperation of the competent centres directly with the paediatricians, involving the internist in the process. This is the 'Berlin' model of rare disease management, which can be applied to most rare diseases.

At this year's conference, psychiatrist assist. prof. Marko Pišljar, MD PhD opened a new chapter in the local book of rare diseases with a lecture on the rare syndromes and conditions in psychiatry, where the stigma of patients with mental health issues is often strongly reflected in the attitude of laypersons.

The road to a rare disease's diagnosis can be as frustrating as the way of the cross. To spare patients the excruciating walk, screening tests at birth can now reveal up to 50 medical conditions (in some US states). Paediatrician prof. Tadej Battelino, MD PhD showcased, in a spirited manner, the extraordinary device that, starting this year, will help the specialised laboratory in Slovenia detect as many as 20 diseases – rather than the current two – from a single drop of a newborn's blood. The ethical precondition for the screening of a certain defect is knowledge it can be successfully treated. Fittingly, the Slovenian Minister of Health signed the selection of the procurement supplier of this fantastic instrument on Rare Disease Day 2018.

And so, we arrive to the ethics. Prof. Miha Oražem, MD PhD from the Institute of Oncology spoke on the subject of understanding that patients suffering from rare diseases should be included into research as active participants – not as mere 'guinea pigs'. Perhaps it is none other than the patients who can come up with a breakthrough idea. Prof. Lenart Škof, a philosopher from the Koper Science and Research Centre applied his ethical notions onto medicine and the subject of rare diseases. Positing health is a basic human right, he stressed medical staff should always be approaching patients with an empathetic mindset: 'What if it was me instead? His metaphor of distress (imagine for a moment, having to suddenly flee from a demolished, war-torn Slovenia to Syria across the sea) incited a moment of self-reflection, perhaps even minute revisions of everyday perspectives among the attending medical professionals. Prof. Božidar Voljč, MD PhD then reassured everyone in precise terms, especially the patients and non-physician staff present, that the National Ethics Committee of the Ministry of Health is advocating for high ethical standards across the field of medicine, especially in the treatment of patients with rare diseases. Lawyer Barbara Jan Bufon, MSc, whose master's thesis addresses the legal aspects of stem cell transplanting, shed light on the issues of our national health legislation, which appears to be lagging behind the realistic state of medical treatment in Slovenia. She stressed the necessity of establishing a normative system of educating laypersons on the demanding procedures of self-

treatment, which in the case of certain rare conditions entail tasking lay patients, or their family members, with home-administering specific procedures and medications normally reserved for inpatient treatment (self-infusion, peritoneal dialysis, respirators). Specific standards ought to be established, overseen and documented in this regard. Lastly, she also explained that the Medical Chamber will have to utilise its powers in the field of health policy to update the range and nomenclature of specialisations in line with the contemporary state of the medical profession, especially when it comes to the field of rare diseases. Thus, specialists with licenses for concrete rare conditions should be able to treat patients from birth to senior age on the basis of a specific rare disease license, rather than that of a paediatrician or internist. Eventually, the Slovenian Medical Chamber will have to tackle the unification of speciality licenses with the demands of the new international classification – the 11th consecutive (MKB 11), which this year finally includes over 2000 diagnoses of rare diseases. At last, this will allow physicians who manage to successfully treat a rare condition to enter appropriate codes absent from MKB 10.

Concluding with the round table discussion “Living with a rare disease”, which presented two patients with amyloidosis and a patient with hereditary angioedema who spoke on the subject of their experiences alongside their doctors (prof. Janez Zidar, MD PhD, prim. Jože Pretnar, MD PhD, prof. Mitja Košnik, MD PhD with a written contribution), the conference was characterised by an overall atmosphere of steadfast awareness that the ancient Roman 'mens sana in corpore sano' (sound mind in a sound body) is no longer as prescriptive. Bodies afflicted by grave disorders likewise carry the full radiant spirit of humanity, which can increasingly be assisted in the establishment of a vibrant life.

I. Programme

EUROPLAN NATIONAL WORKSHOP

Congress center Brdo pri Kranju, Slovenia
28th February 2018

8:30 – 9.00 **Registration**

9:00 – 9:15 **Welcome Speeches**
Milojka Kolar Celarc *(Ministry of health)*
Majda Slapar (*Rare diseases association of Slovenia)*
Vlasta Zmazek (DEBRA Croatia/*EURORDIS)*

9:15 – 9:30 **Jože Faganel** *(Rare diseases association of Slovenia)*

9:30 – 9:45 **European reference networks**
Prof. Borut Peterlin, MD PhD *(Clinical Institute of Medical Genetics, UMC Ljubljana)*

9:45 – 10:00 **National contact point for rare diseases in 2017**
Assist. prof. Mojca Žerjav Tanšek, MD PhD *(Paediatric Clinic, UMC Ljubljana)*

10:00 – 10.15 **The aspects of financing treatment of rare diseases in Slovenia**
Marjan Sušelj, MSc *(Health Insurance Institute of Slovenia)*

10:15 – 10:30 **Rare diseases registry at NIPH. How far we are?**
Eva Murko, MD PhD (National Institute of Public Health)

10:30 – 10:45 **Neuromuscular disease registry (example of a comprehensive treatment registry)**
Assist. prof. Damjan Osredkar, MD PhD *(Paediatric Clinic, UMC Ljubljana)*

10:45 – 11:15 **Coffee Break**

11:15 – 11:30 **Comprehensive treatment of rare congenital pulmonary diseases**
Assist. prof. Ana Kotnik Pirš, MD PhD *(Paediatric Clinic, UMC Ljubljana)*

11:30 – 11:45 **Rare diseases in psychiatry**
Assist. prof. Marko Pišljar, MD PhD *(Psychiatric Hospital Idrija)*

11:00 – 11:15 **Screening tests**
Prof. Tadej Battelino, MD PhD with colleagues *(Paediatric Clinic, UMC Ljubljana)*

12:00 – 12:15 **Patient involvement in research - presentation of the situation and dilemmas**
Simona Borštnar, MD PhD, Miha Oražem, Professor, Božidar Voljč, MD PhD *(Institute of Oncology, UMC Ljubljana)*

11:45 – 12:00 **A Philosopher Before the Ethical Aspects of Rare Diseases**
Prof. Lenart Škof (*Science and Research Centre of Koper)*

12:15 – 12:30 **The role of the Slovenian National Medical Ethics Committee in addressing rare diseases**

Prof. Božidar Voljč, MD PhD *(The National Medical Ethics Committee of the Republic of Slovenia)*

12:30 – 12:45 **The legal basis for dealing with rare diseases**
Barbara Jan Bufon, MSc

12:45 – 13:00 **Discussion**

13:00 – 13:15 **How rare diseases affect our everyday life! Debra Croatia – DEBRA Resource Centre**
Vlasta Zmazek (*Debra Croatia/EURORDIS)*

13:15 – 14:00 **Round table – living with rare disease (Moderator: Jože Faganel)**
Participating patients with:
Gaucher's disease
Myelodysplastic syndrome
Hereditary angioedema
Transthyretin-Related Familial Amyloid Polyneuropathy (TTR-FAP)

Participating doctors:
Prof. Janez Zidar, MD PhD (*UMC Ljubljana)*
Prim. Jože Pretnar, MD PhD (*UMC Ljubljana)*
Prof. Mitja Košnik, MD PhD (*University Clinic of Respiratory and Allergic Diseases Golnik)*

14:00 – 15:00 **Lunch**

II. List of Participants

Name and surname	Organization, company ...	Stakeholder group
Milena Burnik	Retirement home of Idrija	Institution
Marija Špelič	UMC Ljubljana	
Alenka Marič Cevzar	Health Insurance Institute of Slovenia	Institution
Rozeta Hafner	Health Insurance Institute of Slovenia	Institution
Nataša Jakša	Health Insurance Institute of Slovenia	Institution
Aleš Šabeder	UMC Ljubljana	Healthcare professional
Luca Lovrečić	Clinical Institute of Medical Genetics, UMC Ljubljana	Healthcare professional
Karin Writzl	Clinical Institute of Medical Genetics, UMC Ljubljana	Healthcare professional
Helena Jaklič	Clinical Institute of Medical Genetics, UMC Ljubljana	Healthcare professional
Gorazd Rudolf	Clinical Institute of Medical Genetics, UMC Ljubljana	Healthcare professional
Nataša Teran	Clinical Institute of Medical Genetics, UMC Ljubljana	Healthcare professional
Taja Džambasović	Institute of Oncology Ljubljana	Healthcare professional
Irena Bačlija Brajnik	University of Ljubljana	Institution
Tjaša Pečnik	Institute of Oncology Ljubljana	Healthcare professional
Majda Oštir	Paediatric Clinic, UMC Ljubljana	Healthcare professional
Peter Šuštar	Health center of Medvode	Institution
Barbka Repič Lampret	Paediatric Clinic, UMC Ljubljana	Healthcare professional
Tanja Golli	Paediatric Clinic, UMC Ljubljana	Healthcare professional
Jerina Nika	Paediatric Clinic, UMC Ljubljana	Healthcare professional
Montanič Sendi	Institute Service of Slovenia for Transfusion Medicine	Institution
Barbara Faganel Kotnik	Paediatric Clinic, UMC Ljubljana	Healthcare professional
Zidar Janez	UMC Ljubljana	Healthcare professional
Peter Černelč	UMC Ljubljana	Healthcare professional
Borut Peterlin	Clinical Institute of Medical Genetics, UMC Ljubljana	Healthcare professional

Mojca Žerjav Tanjšek	Paediatric Clinic, UMC Ljubljana	Healthcare professional
Eva Murko	National Institute of Public Health	Institution
Damjan Osredkar	Paediatric Clinic, UMC Ljubljana	Healthcare professional
Ana Kotnik Pirš	Paediatric Clinic, UMC Ljubljana	Healthcare professional
Marko Pišljar	Psychiatric Hospital Idrija	Institution
Tadej Battelino	Paediatric Clinic, UMC Ljubljana	Healthcare professional
Simona Borštnar	Institute of Oncology Ljubljana	Healthcare professional
Miha Oražem	Institute of Oncology Ljubljana	Healthcare professional
Božidar Voljč	The National Medical Ethics Committee of the Republic of Slovenia	Institution
Matjaž Sever	UMC Ljubljana	Healthcare professional
Jože Pretnar	UMC Ljubljana	Healthcare professional
Samo Zver	UMC Ljubljana	Healthcare professional
Mihaela Zidarn	University Clinic of Respiratory and Allergic Diseases Golnik	Healthcare professional
Robert Medved	Ministry of Health	Institution
Milojka Kolar Celarc	Ministry of health	Institution
Vlasta Zmazek	Debra Croatia/EURORDIS	Institution
Jože Faganel	Slovenian Haemophilia Society /Rare diseases association of Slovenia	Patient representative
Marjan Sušelj	Health Insurance Institute of Slovenia	Institution
Barbara Jan Bufon		Institution
Urška Oblak	Ewopharma	Industry
Nena Pižmoht	Roche	Industry
Nataša Jenčič	Biogen Pharma	Industry
Mateja Kastelic	Medis	Industry
Darko Mofardin	Medis	Industry
Mediha Grom	Novo Nordisk	Industry
Naskov Julian	Celgene	Industry
Lampe Ivanovska Dalila	Celgene	Industry

Ilijev Zaharie	Celgene	Industry
Prah Klemenčič Mojca	KRKA	Industry
Urbančič Minka	ROCHE	Industry
Ivkanec Saša	SHIRE	Industry
Kuntner Irena	SHIRE	Industry
Barbara Markovič	SHIRE	Industry
Andreja Berguš	SHIRE	Industry
Silič Anja	PFIZER	Industry
Remškar Nina	ROCHE	Industry
Bajt Karmen	The Muscular Dystrophy Association of Slovenia	Patient representative
Černigoj Tea	The Muscular Dystrophy Association of Slovenia/ Rare diseases association of Slovenia	Patient representative
Ana Kovačević Matić	The Muscular Dystrophy Association of Slovenia	Patient representative
Rijavec Marko	The Muscular Dystrophy Association of Slovenia	Patient representative
Jaka Remih	The Muscular Dystrophy Association of Slovenia	Patient representative
Lah Maša	The Muscular Dystrophy Association of Slovenia	Patient representative
Mrak Iztok	The Muscular Dystrophy Association of Slovenia	Patient representative
Sabina Zorko	The Muscular Dystrophy Association of Slovenia	Patient representative
Bajec Stanislav	The Muscular Dystrophy Association of Slovenia	Patient representative
Marija Pintarič	Fabry Patients Association Slovenia	Patient representative
Žnidar Irena	Gauchers Patients Association Slovenia	Patient representative
Rajka Bavčer	Slovenian Haemophilia Society	Patient representative
Jože Berus	Slovenian Haemophilia Society	Patient representative
Aleš Blazetič	Slovenian Haemophilia Society	Patient representative
Miha Perhaj	Slovenian Haemophilia Society	Patient representative
Jože Faganel	Slovenian Haemophilia Society	Patient representative
Čuček Milan	Association of patients with cerebrovascular disease in Slovenia	Patient representative

Modic Kristina	Association of patients with lymphoma and leukemia, L & L	Patient representative
Žagar Brina	Association of patients with lymphoma and leukemia, L & L	Patient representative
Milena Remic	Association of patients with lymphoma and leukemia, L & L	Patient representative
Blaž Kondža	Lymphoma Association	Patient representative
Triller Mirko	Pulmonary and Allergic Patients Association of Slovenia	Patient representative
Andreja Gradišar	DEBRA Slovenia	Patient representative
Polona Zakošek	DEBRA Slovenia	Patient representative
Ivana Domić	DEBRA Slovenia	Patient representative
Grmek Ugovšek Slavka	Cystic Fibrosis Association of Slovenia	Patient representative
Dragica Sajko	Cystic Fibrosis Association of Slovenia	Patient representative
Dragica Iskrenovič	Cystic Fibrosis Association of Slovenia	Patient representative
Božjak Marjana	Association of patients with blood diseases	Patient representative
Erzar ŽepičA	Association of patients with blood diseases	Patient representative
Filipčič Jožica	Association of patients with blood diseases	Patient representative
Flajs Cotič Cvetka	Association of patients with blood diseases	Patient representative
Hribar Janez	Association of patients with blood diseases	Patient representative
Javornik Milojka	Association of patients with blood diseases	Patient representative
Kebe Nada	Association of patients with blood diseases	Patient representative
Kebe Zvone	Association of patients with blood diseases	Patient representative
Koletnik Marija	Association of patients with blood diseases	Patient representative
Koželj Anja	Association of patients with blood diseases	Patient representative
Kralj Štimec Anica	Association of patients with blood diseases	Patient representative
Kus Franc	Association of patients with blood diseases	Patient representative
Kus Irena	Association of patients with blood diseases	Patient representative
Levačič Avguština	Association of patients with blood diseases	Patient representative

Levačič Franc	Association of patients with blood diseases	Patient representative
Mlakar Martina	Association of patients with blood diseases	Patient representative
Mlakar Stane	Association of patients with blood diseases	Patient representative
Molka Franc	Association of patients with blood diseases	Patient representative
Molka Rozalija	Association of patients with blood diseases	Patient representative
Mrak Zdenka	Association of patients with blood diseases	Patient representative
Osredkar Lovro	Association of patients with blood diseases	Patient representative
Ovijač Darja	Association of patients with blood diseases	Patient representative
Petkovič Živorad	Association of patients with blood diseases	Patient representative
Rak Alenka	Association of patients with blood diseases	Patient representative
Rot Marta	Association of patients with blood diseases	Patient representative
Rozina Anton	Association of patients with blood diseases	Patient representative
Rozina Terezija	Association of patients with blood diseases	Patient representative
Ručigaj Boštjan	Association of patients with blood diseases	Patient representative
Sever Matjaž	Association of patients with blood diseases	Patient representative
Slapar Majda	Association of patients with blood diseases/Rare diseases association of Slovenia	Patient representative
Slapar Boris	Association of patients with blood diseases	Patient representative
Starič Antonija	Association of patients with blood diseases	Patient representative
Šimenc Judita	Association of patients with blood diseases	Patient representative
Škrlj Anica	Association of patients with blood diseases	Patient representative
Škrlj Franc	Association of patients with blood diseases	Patient representative
Uhan Mihaela	Association of patients with blood diseases	Patient representative
Uhan Jože	Association of patients with blood diseases	Patient representative
Stjepanović Tatjana	Association of patients with blood diseases	Patient representative
Škrabar Rado	Association of patients with blood diseases	Patient representative

Iskra Tea	/	Patient representative