Gefördert aus den Mitteln
GEMEINSAME GESUNDHEITSZIELE
aus dem Rahmen-Pharmavertrag
eine Kooperation von österreichischer Pharmawirtschaft und Sozialversicherung

pro rare austria
allianz für seltene erkrankungen

EUROPLAN
European Project for Rare Diseases National Plans Development

Austria

8th Congress on Rare Diseases and

2nd Austrian EUROPLAN-Conference EUROPLAN

EUROPLAN NATIONAL CONFERENCE

in the framework of the EU Joint Action RD-ACTION

Vienna, 19th – 21st October 2017

Final Report

RD ACTION
DATA AND POLICIES FOR RARE DISEASES

Co-funded by
the Health Programme
of the European Union

EURORDIS
RARE DISEASES EUROPE

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, 41 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	Austria
National Organizers	Rare Disease Forum Pro Rare Austria
National Conference Date and Location	19th – 21st October 2017
Websites	http://www.forum-sk.at/ http://www.prorare-austria.org/
Members of the Steering Committee	Assoc.-Prof. Priv.-Doz. Dr. Till Voigtländer OA Dr. Vassiliki Konstantopoulou Ulrike Holzer Dipl.-Ing. Victoria Mauric
Topic	Social Care for Patients Suffering from Rare Diseases in Austria
Appendix	I. Program II. List of participants by stakeholders' categories

FINAL REPORT

I Introduction

"It would sadden me to sit here again next year and work thorough the same items. It would be important to achieve something together based on the results!", explained Dipl.-Ing. Dr. Thomas Kroneis (KEKS). With these words, he found the appropriate closing statement to the **8th Congress on Rare Diseases**, which took place in the **Vienna Museumsquartier** from the **19th through the 21st of October 2017**.

This year's event was dedicated to the three **dimensions of rare diseases** and was organized by the **Forum Rare Diseases**, particularly by Dr. Till Voigtländer and Dr. Vassiliki Konstantopoulou, and by **Pro Rare Austria – the Alliance for Rare Diseases**.

While the first day of the congress offered an insight into the medical dimension of rare diseases and convinced with sound scientific presentations, day two focused on the dimension of health policy.

The focus was initially on the **European Reference Networks (ERN)**, as well as the Austrian **designation** process for **'centers of excellence for rare diseases'**. This designation is a prerequisite for full membership in an ERN. These core elements of the National Action Plan for Rare Diseases (NAP.se) were supplemented by the presentation of additional measures, which Pro Rare Austria has been supporting since September 2016 through the **"ProNAP - Supporting the implementation of NAP.se" grant** project. The pharmaceutical industry is also affected by these developments, as rare diseases remain a challenging topic. The subsequent **panel discussion** brought together an interesting and heterogeneous group of stakeholders including civil servants, medical experts, patient representatives and members of the pharmaceutical industry.

The 2nd Austrian EUROPLAN Conference and, with that, the social dimension of rare diseases was initiated with a presentation of the Vienna Social Fund, its social services portfolio and the newly established health hotline '1450'. After that, a **pilot project** using **case managers in Romania** was presented. This pilot attempts to address existing gaps in the coordination between medical, social and support services in the EU Member States which are part of the **EU project 'INNOVCare'** (Innovative Patient-Centered Approach to Social Care Provision to Complex Conditions).

The daily challenges people with rare diseases face were presented concretely and made tangible, by **reports from relatives** of patients, such as Yvonne Otzelberger (Austrian Angelman Association) and Ernst Leitgeb (HHÖ). In addition, over the course of the second panel discussion important statements were made regarding social care and welfare of people with rare diseases and possible new approaches of care in Austria were discussed.

After a brief presentation of current activities and recent developments for the members, the general assembly of Pro Rare Austria was held on the morning of the third congress day. At the core of the event were three simultaneous multi-stakeholder workshops dealing with questions of psychosocial health, challenges of everyday life, as well as financial security and administrative errands. After the itemization and categorization of current issues, each one of the three focus groups discussed possible solutions.

II The social care of patients suffering from rare diseases in Austria - Themes

Based on the presentations and the subsequent podium discussion of day two of the congress, important core resolutions regarding the social care of patients and their relatives were made.

Through the integration of the experiences of the attending audience, it was underscored that rare diseases carry elevated complexities and demand special support systems. This particularity makes appropriate attention in the Austrian framework of rigid structures and elementary universalism very difficult. Regional differences, health-care and social system obstacles and federalism inhibit the implementation of national strategies (e.g.: physician centralization, limited exchange of information across regions, etc.). In spite of solid basic health care for the general public and several donation-funded 'light house' projects, like the EB-Haus in Salzburg, patients suffering from rare diseases are dependent on the goodwill of the dedicated few. There is, both, a lack of political interest and of awareness of health-care provision gaps.

Due to the special circumstances for patients suffering from rare disease, services of support agencies like the Vienna Social Fund only apply to them in a limited form. For instance, assisted living and adult daycare settings do not offer the adapted services necessary for rare disease sufferers (e.g.: Prader-Willi-Syndrome).

An improved quality of life through holistic and individualized care (for instance through supportive physical and psychotherapy, nutrition and mobilization programs and social integration initiatives) can often be achieved, especially when such therapies are not available. Ideally, specialized day care centers offering social interaction, supporting programs addressed above and trained personnel are established.

Experts, patients and care giving relatives discussed the above-mentioned items and related matters and worked out possible solutions and ideas for future endeavors in addressing the current needs in **three workshops** focusing on the sub topics of **psycho-social health**, **challenges of everyday life** and **financial security and administrative errands**.

THEME 1: Psycho-social health

Focus group 1 was moderated by Mag. Jay Ladurner (National Coordination Center for Rare Diseases) and dealt with the topics of **psycho-social health of rare disease sufferers and their relatives** and the care giving environment of these patients.

In total, there were 13 participants in attendance (9 women and 4 men, between the ages of 20 and 60 years). The group consisted mostly of patients and their relatives, who are active in their support groups, and two physicians, a psychology student, a social sciences researcher from the center of social innovation and an employee from Pro Rare Austria.

Two points of view were developed regarding psycho-social stresses: on the one hand, the **intrinsic view**, which affects patients and their relatives themselves, and on the other hand, the **external view** focusing on the outside world. Regarding the external view, several possible solutions and measures were developed.

1.1. Personal points of view

A touching quote from this working group shows how such everyday stresses manifest: “Visible deformities make my child an outsider and allow for it to be picked on. To witness this situation, is barely bearable.”

It became clear that **fear is a dominating sentiment.** This feeling manifests itself in various ways: fear of the future, fear of not being taken seriously or fear of stigmatization, along with blurred perspectives and dependencies. This fear turns into an intrinsic prison and renders one speechless. This is often about recurring issues such as "how do I integrate or blend with surroundings without frightening them?" or "how do I reconcile the different needs (people affected, relatives)?" and, last but not least, "how can I handle my illness positively?".

There was consensus in the group that the **power to overcome these fears** must come from within the individual. It was reported that it is absolutely possible to successfully face these challenges with a certain calmness through humor, openness and love. By actively approaching the respective life situation, by finding new perspectives and ways to develop one's own "normalness", it is possible to gain a bit more self-determination. The exchange and support of each other in the support groups is deemed essential.

1.2. External points of view

Lack of psycho-social resources in patient care (without expected improvement over the medium term), diffuse explanations of non-experts on the psychological causes and **physician disregard of the social dimension during treatment** shape the picture of rare disease patient care. Lack of knowledge on the part of the social environment (Kindergarten teachers, teachers, employers, etc.) and the constant effort for monetary support for psychotherapeutic measures (parallel accompaniment is not yet an accepted practice in the domestic health-care system) complicate the situation.

The effects around the treating physician are central. Regarding the care of people with rare diseases (but also in general), it should be ensured that **doctors receive training on the social aspects of dealing with patients.** This may be realized by shorter waiting periods, reasonable time spent in conversation with the patient, a confidence-generating and respectful conversation climate and the redesign of health care services (including accompanying relatives).

Another goal would be the **creation of interdisciplinary boards** (doctors, psychologists, caretakers, social workers, self-help, etc.) under the motto "**professionals help professionals**". This may come in the form of a system services and partners exchange and through the development of sustainable psycho-social expertise. Also, models for an improved interaction with the social environment may be developed (keyword: case management). **Strengthening self-help** also seemed **desirable** to the working group. This may be realized through supervisions or trainings for patient organizations (legal, medical, psychological, socially competent, etc.).

THEME 2: Challenges of everyday life

Together with **Mag. Dominique Sturz** (Usher Syndrome Forum), eleven participants (**focus group 2** consisting of nine women and two men between the ages of 20 and 60) addressed the **challenges of everyday life** from the point of view of those affected and their relatives. To define the term "everyday life", the following categories were used as anchor points and appropriate troubleshooting efforts and ideas for possible solutions worked out: **education and work; everyday life** (household, purchases, personal care, and the like); **mobility and transport; treatment, care and rehabilitation; psycho-social aspects** (social environment, mental state).

2.1. Education and work

One of the biggest challenges for patients (and their relatives) is **becoming and remaining gainfully employed.** The sentiment of being “unemployable” was repeatedly verbalized. Also, a perceived reduction in availability and capability in their social and professional environment carries a negative connotation for affected persons. For care-giving relatives who are employed, generating enough time away from work during emergencies is often difficult. This led to the question whether relatives who provide care really can remain employed in good faith (for example: mothers).

The solution may come in the form of **additional information and creating improved awareness** amongst kindergarten teachers, teachers, employers and colleagues. Further, the topic of disability in the workplace may be tackled with a different approach, as the corresponding legal framework in Austria is available. Several examples such as "adapted probationary periods", "higher penalties for non-employment" or "more money for less work (30 hours = full time)" were mentioned. With regard to required educational opportunities, it is anticipated that the Public Employment Service takes into account the special circumstances when planning training programs or when supporting educational institutions in establishing of exemptions (for example, taking tests on a laptop, longer testing time permitted, higher tolerance for missed lessons).

2.2. Everyday life (household, shopping, personal hygiene, etc.)

Depending on the particular condition and the course of the disease, **there are problems in coping with everyday challenges** such as climbing stairs, getting dressed, cooking, cleaning, shopping, personal hygiene, dealing with sleep disorders, weakness or a general lack of energy. Not much is possible without the support of a third party. Accordingly, the group developed suggestions such as the creation of shorter waiting periods through, for example, separate check-out counters for the disabled at the supermarket or reduced waiting periods at the doctor’s office. Financial support, for example, for the renovation of sanitary facilities (handicapped accessible) or the exemption from lift costs in apartment buildings, as well as the availability of short-term support staff and increased exchange of experiences regarding self-help were mentioned. **It takes "experienced people who deal with illnesses and have alternative solutions for challenges of everyday life**," said a patient.

2.3. Mobility and transport

The main topics included the **dependency on one’s intimate social environment** and the resulting lack of flexibility and time for the immediate care-giving family member (for instance, mothers who have to give up their jobs or drastically reduce working hours), and the **dependency on support and transportation services including associated costs**, and the **often-inadequate technical standards in the public sphere**.

Legally binding entitlements with regard to transportation services through taxis, the Austrian federal railways, and other such services were discussed. There is a need for further and more detailed information regarding this topic.

2.4. Treatment, care and rehabilitation

Available therapies and rehabilitation efforts were basically positively evaluated, however, **frequent rejections** by and **tedious application processes** to the insurance fund were lamented. Once approved, therapies can be tedious, time-consuming and exhausting, which in turn affects the already challenging daily life (for example, high absentee hours during training and work). A request for simplified approvals (for instance, reduced head physician approval for the chronically ill), better coverage, and the appropriate availability of treatment slots was mentioned. When dealing with issues in a private setting, one's intimate social environment needs to be utilized for solutions (for example, networks).

2.5. Psycho-social aspects (social environment, psychological constitution)

The main issues focused on questions like "How am I being perceived?" and "Am I strong enough to keep up?" and revolved around a lack of respect from third parties and a lack of understanding for the limitations that are associated with a particular disease. A potential solution would be the psychological support during the day care setting, which may be offered as a treatment option immediately upon

diagnosis.

THEME 3: Financial security and administrative errands

Assoc. Prof. Priv. Doz. Dr. Till Voigtländer (Rare Disease Forum, National Coordination Center for Rare Diseases, Medical University of Vienna) led focus group 3 on the topic of financial security and the management of administrative errands. The eight participants (five women, three men between the ages of 20 and 60 years) were either patients or relatives of patients working in the self-help arena. Also, three physicians (including an employee of the National Coordination Center for Rare Diseases) took part in the session.

3.1. Financial security

The workshop underscored that due to the **high need for care and support when dealing with rare diseases**, patients are directly dependent on their nursing care allowance, their actual level of allowance disbursed, increased child support payments, a minimum income and other public funding schemes. In particular, there seem to be inconsistencies when setting the nursing care allowance level for one and the same disease. In addition, the allocation of increased child support often seems tedious and momentous, as many other social benefits depend on it. Even though there are rare diseases that may manifest at any age, most of them are congenital. This **intensified need for care and support** represents a **significant challenge to families** and, at times, render them dysfunctional and broken. In some cases, single mothers who depend on the nursing care allowance as their sole source of income are financially ruined after their child dies. Rather than on administrative errands, the main issue revolves around laws that off-set nursing care allowances with minimal income payments.

Another factor are the **exceedingly high cost and deductibles for medication and specialized services** (such as transport), which are only partially covered by additional, private health insurance. Things are complicated further because **decision making** happens **at the provincial level**. This holds true not only for patients suffering from rare diseases, but also for people with chronic diseases or other permanent impairments. It would be advisable to utilize existing patient support structures and "operate in a group setting". It may be advisable to provide ample **documentation of diagnostic findings and even clinical studies** to the assessor to allow the patient to present his or her situation appropriately.

3.2. Administrative errands

Managing administrative errands (health insurance fund, federal social services departments and other public service offices) is generally cumbersome. Head physicians at the insurance funds often lack appropriate information or reject applications for treatments or supportive measures when the disease manifestation is not immediately visible. In addition, the public service offices set a high bar for required documentation to approve support efforts and entitlements. Expenditures of time can be significant. Again, there are considerable differences in how fast and standardized provinces handle such requests. **Harmonized standards** regarding approvals and processes across provinces and the various insurance funds, and the increased coordination of public stakeholders like insurance funds, administrative district offices, provincial governments and the responsible federal department will improve these efforts. Above that, the establishment of a **guideline for patients and their families** for a diverse set of applications and the inclusion of patient representatives in working groups may contribute to diffusing the situation.

III Conclusions

“Living with a rare disease is a path. There is lightness and heaviness, there is difficulty and beauty. I recognize that and focus on the positive.” – this was one of the contributions from the focus groups.

This quote appears to point the way in how to deal with a rare disease. There are many challenges and unsolved problems – in the political, medical and social sphere – which can intensify dramatically on a personal level. The medium-term solution lies in the creation of a constructive, solution-oriented and knowledge-based approach. The **2nd Austrian EUROPLAN-Conference** underscored that **medical and social care** for patients suffering from rare diseases are **intrinsically connected**. The extramural setting is an example of this interconnectedness.

Rare diseases do not fall in one particular political area, but encompass many aspects, which require full integration (health, social aspects, science and research, business and industry).

This respective knowledge seems apparent within the system. However, the bottom line relates to carrying the financial burden in times of dwindling financial resources. The **federal separation of competencies** as outlined in the constitution seems to be an insurmountable obstacle. But, the Austrian political landscape is also one of the social partners, which allows for the **possibility of an open dialogue**. This is only possible if an educated community couples well prepared information with realistic ideas for such discussions. **Patient organizations** are important players and act as **translators and mouthpieces**. On the one hand, patients and their relatives have to be heard, and on the other hand, a homogeneous group needs address the stakeholders of the system to create awareness for certain problems. The fate of an individual produces consternation, evidence-based data creates pressure to act.

There needs to be a methodical approach that renders the topics workable for the political strata. The **“National Action Plan for Rare Diseases (NAP.se)”** is such an instrument, but primarily addresses the medical Dimension of rare diseases. In its realization, NAP.se also faces the same obstacles inherent to the Austrian health care system.

To expand the focus to the social care aspect, data were collected through efforts of the **EU-project "INNOVCare"**, which supports the importance of the **social dimension** of rare diseases. The concluding recommendation of these efforts concentrates on the establishment of case managers who aim at guiding patients and their relatives through the system while helping them access the available services.

To implement this or other solutions in Austria, it is necessary to understand the topics identified in the **multi-stakeholder workshops** and apply these learnings to the **specifics of Austria and their associated leverage effects**. Qualitative socio-scientific research would allow for a concrete depiction of the reality of rare disease patients, and encourage subsequently, based on such data, the development of appropriate recommendations. The results and possible solutions generated during the 2nd Austrian EUROPLAN-Conference were further solidified.

A well-known supportive measure in this context is the development of black books and white papers. **The results** of the congress should be **summarized in a black book**. Its contents then should be openly addressed in a political discourse with the goal of **developing concrete strategies for the social welfare of people with rare diseases in Austria**. These strategies then should be **summarized in a white paper** to support next steps.

"As a first step, the National Action Plan on Rare Diseases moved the topic of medical care of people with rare diseases to the center of health policy," explains Till Voigtländer. Now the time has come for a second step: to give the social dimension of rare diseases the importance it deserves - because it shapes every day of an affected person's life.

APPENDIX I - Program

8th AUSTRIAN CONGRESS FOR RARE DISEASES

2nd AUSTRIAN EUROPLAN CONFERENCE

MUSEUMSQUARTIER Vienna
ARENA 21 and OVAL HALL
19-21 OCTOBER 2017
www.forum-sk.at

Excerpt from the congress program:

Co-Event: 2nd Austrian EUROPLAN Conference and Workshop

Social care of people with rare diseases in Austria from cradle to grave

Friday, 20.10.2017	**Part I: Existing (AT) and Innovative (EU)**	
14:00 – 14:30	Keynote: Social care offers for patients with rare diseases	S. Herbek
14:30 – 14:55	INNOVCare (Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions) – the project	R. Castro
14:55 – 15:15	INNOVCare in Austria	U. Holtgrewe
15:15 – 15:45	Challenges in Austria - patient reports: • Angelman Society Austria • Hepatitis Help Austria • Challenges from cradle to grave	 Y. Otzelberger E. Leitgeb V. Mauric
15:45 – 16:15	coffee break	
16:15 – 18:00	Podium and plenary discussion with • S. Herbek (Social Fund Vienna) • U. Holtgrewe (Center für Social Innovations) • R. Castro (EURORDIS) • M. Weigl (Pro Rare Austria) • D. Karall, D. Möslinger (medical experts)	S. Kircher
18:00 – 19:00	Humorous and tender encounter with patients - a CliniClown reports on her work	V. Vondrak-Zorell
19:00 – 20:00	Dinner	

Saturday, 21.10.2017	**Part II: New ways in Austria as well?**	
09:00 – 09:10	Welcome and summary of the previous day	V. Konstantopoulou, T. Voigtländer
09:15 – 09:45	Presentation of current activities of Pro Rare Austria	V. Mauric
09:45 – 10:45	General Assembly of Pro Rare Austria	R. Riedl
10:45 – 11:15	coffee break	
11:15 – 13:20	**Workshop: Quo vadis, Austria? New ways to provide social care for people with rare diseases in Austria**	
11:30 – 11:40	Introduction (methodology and goals)	V. Konstantopoulou, T. Voigtländer
11:40 – 13:00	Workshop-groups: • Psychological care • Challenges in everyday life (education, transport, etc.) • Financial security and administrative errands	 J. Ladurner D. Sturz T. Voigtländer
13:00 – 13:15	Reports from workshop-groups and final discussion	
13:20 – 13:30	Closing remarks	V. Konstantopoulou, T. Voigtländer
13:30	Farewell	

APPENDIX II - List of participants by stakeholders' categories

Pro Rare Austria – Austrian Alliance for Rare Diseases	10
Patients and their relatives	40
Public institutions	19
Health care providers	23
Research Centres and Pharma	22
EURORDIS	1
European Commission	1
Total	**114**

Remark: Only a part of the participants of the congress participated in the workshops of the last day of the event