



# ROMANIA

## EUROPLAN NATIONAL CONFERENCE

### FINAL REPORT

24 - 25 May 2013, Bucharest

# FOREWORD

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

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

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

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

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

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

1. Methodology and Governance of a National Plan;
2. Definition, codification and inventorying of RD; Information and Training;
3. Research on RD;
4. Care - Centres of Expertise / European Reference Networks/Cross Border Health Care;
5. Orphan Drugs;
6. Social Services for RD.

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

## General information

The Romanian EUROPLAN Conference was organized under the auspices of the Ministry of Health Romania, in close collaboration of the Romanian National Alliance for Rare Diseases, RSHG and Ministry of Health. The Conference had a clear mandate to involve all relevant national stakeholders in the field of RDs. These included patients and their families, representatives of patient organizations and the National alliance, public authorities, academics, healthcare professionals, medical societies, industry, personal assistants, social workers, educators and therapists.

In order to face the challenge of the transferability of the EU policy at national level, we had decided to invite some EU experts as speakers, while we have tried to ensure that we have an entire national approach.

Our national conference was organized with an Opening Session – a short update of the national situation and an overview of the expectation of the audience from the implementation of a national strategy for RD in Romania; a Plenary Sessions dedicated to the EU documents (Commission Communication and Council Recommendation on RD, EUCERD Recommendations), reports from the workshops, as well as the EUROPLAN, EUCERD Joint Action for Rare Diseases and the Research in RD at EU and international level. In the workshop the main common themes selected by EUROPLAN team were debated.

<b>Country</b>	<b>Romania</b>
<b>Date and place of the National Conference</b>	<b>May 24 to 25, 2013 Hotel Ibis Parliament Bucharest</b>
<b>Websites</b>	<a href="http://www.bolirareromania.ro">www.bolirareromania.ro</a> <a href="http://anbraro.wordpress.com">http://anbraro.wordpress.com</a> <a href="http://www.apwromania.ro">www.apwromania.ro</a> <a href="http://www.edubolirare.ro">www.edubolirare.ro</a>
<b>Organizers</b>	<b>ANBRaRo - Romanian National Alliance for Rare Disease (RONARD)</b> <b>SRGM - Romanian Society of Medical Genetics</b> <b>Ministry of Health</b> <b>EURORDIS – The European Organisation for Rare Diseases</b>
<b>Members of the Steering Committee</b>	1. Dr. Crisitna Vladu – Adviser Ministry of Health 2. Dan Dorica – president ANBRaRo 3. Prof dr. Maria Puiu – vicepresident ANBRaRo

	<ol style="list-style-type: none"> <li>4. Prof. Dr. Emilia Severin – University of Medicine and Pharmacy Carol Davila, Bucharest</li> <li>5. Conf. Dr. Cristina Rusu – UMF (University of Medicine and Pharmacy) Iasi, Orphanet</li> <li>6. Conf. Dr. Vlad Gorduza – University of Medicine and Pharmacy Iași</li> <li>7. Dr. Crisitna Vladu – Adviser Ministry of Health</li> <li>8. Conf. Dr. Dana Craiu – Pediatrics Clinic “ Al. Obregia” Bucharest</li> <li>9. Conf. Dr. Corin Badiu – Parhon Institute Bucharest</li> <li>10. Adriana Harja, patient ANBRaRo</li> <li>11. Radu Ganescu, patient ANBRaRo</li> </ol>
<p><b>Name and list of Workshops</b></p>	<ol style="list-style-type: none"> <li>1. Research on Rare Diseases</li> <li>2. Caring for patients with rare diseases - "patient circuit"</li> <li>3. Methodology and Governance PNBR</li> <li>4. Centers of Expertise and European Reference Networks for Rare Diseases</li> <li>5. Treatments and therapies for rare diseases, orphan drugs;</li> <li>6. Social Services in Rare Diseases</li> </ol> <p><b>Plenary Sessions</b></p> <p><b><i>Official opening. EU recommendations on RD EUROPLAN Project</i></b></p> <p><b><i>Current situation in Romania</i></b></p> <ul style="list-style-type: none"> <li>• <b><i>Information, access to information, e-Health, e-Learning</i></b></li> <li>• <b><i>Definition, codification and inventoring of rare diseases</i></b></li> <li>• <b><i>Research and health care in rare diseases</i></b></li> <li>• <b><i>Treatment and social services in RD</i></b></li> <li>• <b><i>Methodology and Governance of NPRD (National Plan for Rare Diseases)</i></b></li> <li>• <b><i>Centers of Expertise and European Reference Networks for Rare Diseases</i></b></li> <li>• <b><i>Treatments (Orphan Drugs) and Social Services in the field of RD</i></b></li> </ul>

<p><b>Leaders Workshops (rapporteurs, where applicable)</b></p> <p><b>Rapporteurs:</b>          WS1. Ghile Emanuela si Iulia Simina,          WS2. Breban Florina si Cosmina Festeu,          WS3. Brazdau Maria si Tegzes Florica,          WS4. Persa Alexandra si Culcear Ingrid,          WS5. Ghile Emanuela si Iulia Simina si          WS6. WS3. Brazdau Maria si Tegzes Florica.</p>	<p><b>Chairs: WS1. Research in RD</b>  <i>Prof. Dr. Maria Puiu – vicepresident ANBRaRo</i>  <i>Ioana Ispas – ANCS (National Authority for Scientific Resource)</i></p> <p><b>WS2. Caring for patients with rare diseases - "patient pathway"</b>  <i>Prof. Dr. Dana Craiu Carol Davila University of Medicine Pediatric Neurology Clinic, Alexandru Obregia Hospital Bucharest</i>  <i>Dorica Dan – president ANBRaRo</i></p> <p><b>WS3. Methodology and Governance NPRD</b>  <i>Cristina Vladu - Adviser Ministry of Health</i>  <i>Prof. Dr. Emilia Severin - EUCERD</i></p> <p><b>WS4. Centres of Expertise and European Reference Networks for Rare Diseases</b>  <i>Conf. Dr. Cristina Rusu – Medical Genetics Center Iasi, Orphanet</i>  <i>Dr. Vlad Gorduza - University of Medicine and Pharmacy Iași</i></p> <p><b>WS5. Treatments and therapies for rare diseases, orphan drugs</b>  <i>Dr. Ioana Bianchi – Ministry of Health</i>  <i>Marius Savu - President of National Drugs Agency</i></p> <p><b>WS6. Specialised Social Services in RD</b>  <i>Lisen Julie Mohr – Frambu Resurce Centre</i>  <i>Daniela Coța – Vice Mayor Zalau</i></p>
<p><b>Annexes</b></p>	<p><b>I. Programme</b>  <b>II. Stakeholders Groups that attended the conference</b></p>

## The main report

The 6 main topics of the common format of the EUROPLAN National Conferences have been addressed along the Romanian EUROPLAN Conference. However, we have added the theme “**Caring for patients with RD (Patients’ pathways)**”, selected by conference organizers as a theme of interest at national level.

### I) PLENARY REPORT

#### Opening Session

#### 1. The Opening Session started with the presentation of the current status of the NPRD In Romania and the steps taken by Romania in the field of RD – Dorica Dan, President RONARD/ANBRaRo

##### a. Key moments of the development of the NPRD in Romania:

- ✓ May, 2003 - Establishment of Prader Willi Association in Romania – RPWA (Asociația Prader Willi din România - APWR);
- ✓ 16th October, 2005 - Establishment and official opening of the Information Centre for Rare Diseases (Centrul de Informare pentru Boli Genetice Rare);
- ✓ June 21 - 24, 2007 – The Sixth International Conference on Prader Willi Syndrome and Rare Diseases, Cluj-Napoca;
- ✓ August 9th, 2007 - Establishment of the Romanian National Alliance for Rare Diseases; RONARD (Alianța Națională de Boli Rare din România – ANBRaRo);
- ✓ August 10th, 2007 - first working group meeting to develop the National Plan, held at the Information Centre for Rare Diseases, Zalău;
- ✓ November 1- 3, 2007 - the first national conference on rare diseases "Rare diseases- From evaluation the needs to establish priorities";
- ✓ February 29th, 2008 - The First European Rare Disease Day Campaign finalized with a partnership established by ANBRaRo and the Ministry of Health Romania – “Rare Diseases, a priority for health care in Romania” in order to finalise and implement the NPRD in Romania;
- ✓ (Rare Disease's Day campaigns continued in 2009 and 2013);
- ✓ 2008 - Regular meetings of working groups on NPRD - National Plan for Rare Diseases;

- ✓ April 2009 - Prader Willi East European Conference, Timișoara;
- ✓ June 2009- Balkan Congress for Rare Diseases, Cluj-Napoca;
- ✓ 2009-2011- NoRo Project, the project objectives are derived from NPRD - National Plan for Rare Diseases objectives, the project developed in partnership with Ministry of Health Department and funded by Innovation Norway.
- ✓ March 2013 – Partnership "Rare Diseases, a Public Health Priority for Romania" between Ministry of Health (MOH) and the National Alliance for Rare Diseases Romania (ANBRaRo). The Romanian Ministry of Health and the National Alliance for Rare Diseases Romania (ANBRaRo) signed a new partnership to implement the National Plan for Rare Diseases in Romania. The National Plan for Rare Diseases (PNBR) developed under this partnership will be the basis for public policies and programs and dedicated resource allocation Rare Diseases for the period 2013-2020.

**Work methodology to develop the National Plan for Rare Diseases:**

- ✓ Identifying needs, areas of intervention and components problems;
- ✓ The actual plan development;
- ✓ Public debate and adjustment - in accordance with the views expressed in meetings organized by the working groups with Ministry of Health, Ministry of Labor, National Agency of Medicines, NADP- National Authority for Disabled Persons (ANPH – Autoritatea Națională pentru Persoane cu Handicap România), NACPA - National Authority for Child Protection and Adoption (ANPCA - Autoritatea Națională pentru Protecția Copilului și Adopție), representatives of the Ministry of Health from Bulgaria and EURORDIS, comments made by European expert Group, and conclusions expressed in the National Conference of Rare Diseases, November 2 - 3, 2007: " Rare Diseases: A Public Health Priority";
- ✓ Signing the partnership agreement with Ministry of Health on February 29th, 2008: "Rare Diseases: A Public Health Priority in Romania";
- ✓ Detailing objectives and activities of the National Plan for Rare Diseases during the meeting of the RD TFR - Rare Diseases Task Force Romania (final version NPRD - sent for approval to more formal forums);
- ✓ Implementation of the National Plan for Rare Diseases in the National Public Health Strategy. In 2013, during the RDD Campaign a second **Partnership "Rare Diseases, a Public Health Priority for Romania" has been signed between Ministry of Health MOH) and the National Alliance for Rare Diseases Romania (ANBRaRo)**
- ✓ Following the partnership a National Committee for Rare Diseases was set up by Ministerial Order, which creates the institutional framework for decisions in the area of RD.

**b. Objectives of the National Plan for Rare Diseases**

The **main goal** of the National Plan for Rare Diseases in Romania is:

- ✓ *Improving quality of life of people affected by rare diseases in Romania by offering the possibility to access early diagnosis, treatment and rehabilitation services for rare diseases.*

The **specific objectives** of the National Plan are:

1. Increase access to information and knowledge on rare diseases;
2. Establish a national strategy to ensure adequate measures for prevention, diagnosis, treatment and rehabilitation for patients with rare diseases;
3. Create a national data base of rare diseases;
4. Training programs for specialists in various fields on the approach of rare diseases;
5. Promote research and innovation on rare diseases, especially for treatments;
6. Develop national and international European partnerships in the field of rare diseases.

**c. The Partnership between the Ministry for Health and National Alliance for Rare Diseases Romania (ANBRaRo)**

The Romanian Ministry of Health and the National Alliance for Rare Diseases Romania (ANBRaRo) signed a partnership to implement the National Plan for Rare Diseases in Romania. The National Plan for Rare Diseases (NPRD / PNBR) developed under this partnership will be the basis for public policies and programs and dedicated resource allocation Rare Diseases for the period 2013-2020.

The **main responsibilities** of the parties in the partnership are:

**Ministry of Health Romania:**

- ✓ designate a person from MoH responsible for collaboration with ANBRaRo on the process of implementation of the National Plan for Rare Diseases;
- ✓ organize the National Committee for Rare Diseases – CNBR for methodological and scientific coordination of rare diseases services at national level. It will be an interdisciplinary scientific body without legal personality, working as an advisor of the Ministry of Health;
- ✓ provide specialized expertise both by its own team, as well as by specialized teams in different areas;
- ✓ develop criteria for designating centers of expertise in the management of rare diseases, define evaluation process and identify the Centre of Expertise in Romania;
- ✓ communicate with ANBRaRo and other institutional partners and define priority



actions to ensure continuity of care for patients with rare diseases: information, diagnosis, treatment or specific therapies, counseling and patient and family education, training and integration specialists in the community;

- ✓ National Plan for Rare Diseases adopted by December 2013; the NPRD was included in the National Strategy for Health, publicly debated in December 2013: its adoption is expected any time soon;
- ✓ Project the financial implications of actions, choose policy options and include progressive provisions in the budget priority;
- ✓ cooperate in the process of raising funds for implementation PNBR;
- ✓ PNBR includes the main components of the National Health Strategy 2014-2020.

**National Alliance for Rare Diseases Romania (ANBRaRo):**

- ✓ designate a responsible person for monitoring of the implementation of the PNBR;
- ✓ inform / consult with patient organizations in connection with the main directions of action for PNBR;
- ✓ actively assess the needs of their members and forward them to Ministry of Health;
- ✓ participate actively - through its representatives in the CNBR for the development of the quality criteria for the evaluation of Centers of Expertise in rare diseases and their evaluation;
- ✓ ensure the continuity of care for patients and their access to specialized social services, close to their home;
- ✓ help in informing, educating patients, families and the community on the issues related to rare diseases;
- ✓ engage in fundraising through specific projects for rare diseases.

2. In the opening session, following the presentation of the progress of the NPRD in Romania by Dorica Dan, representative ANBRaRo, Eurordis EUROPLAN Advisor, **Cristina Vladu - MoH counselor presented the current situation in Romania regarding the care of patients with RD.**

The main expectations from implementation of the NPRD in Romania are:

- ✓ Raising the capacity for diagnosing rare diseases;
- ✓ Raising the quality of life for patients with rare diseases;
- ✓ Raising the capacity of research in rare diseases;
- ✓ Infrastructure of care in rare diseases;
- ✓ National Centers of References assigned to the European networks in rare diseases;
- ✓ Access to clinical trials for Orphan drugs;
- ✓ Support from the MoH in the development of the strategy for RD;
- ✓ Transparency and better communication between reference centers for rare diseases

3. A **round session** of key messages for the NPRD development took place among the Romanian professionals and the foreign experts, representatives of various institutions, who identified the following problems and solutions: the assessment of the health of any person by the family doctor should be made through a grid performed by geneticist thus for achieving a “**riscograma**”. There is a grid already performed by **Dr. Adrian Pana** - Secretary of State, MoH, and in it we can propose to introduce genetic risk - riscograma (*‘riscograma’ is a tool that covers all questions for specific examinations and tests, if any, for each age group and medical conclusions, on individual risks identified, their level and recommended interventions to reduce risk. It was first developed for people with chronic diseases to enable monitoring disease-related risks*). It would be good if it appeared in the basic package of health services.
4. **Dr. Vlad Gorduza** proposed the need for a genetic cabinet in each county and attracting specialists. At the moment we have 30 geneticists in Romania. 80% of chronically ill cases are genetic cases. He also emphasized the following aspects of the rare diseases communities:
  - ✓ Small populations dispersed over wide geographic areas
  - ✓ Heterogeneous expression of disease
  - ✓ Variable diagnostic criteria and assessment measures
  - ✓ Late or incorrect diagnosis
  - ✓ Lack of access to appropriate therapies and clinical experts:
    - Limited Laboratory Scientists and Clinician Expertise and Experience
    - Limited Access to Research Resources
  - ✓ Restrictions to animal and other disease models, reagents.
  - ✓ Limited number of clinical trials (small number of patients)
  - ✓ Limited Foreseeable Profits/Financial Incentives
  - ✓ Lack of interest in developing new therapies due to limitations in the market
  - ✓ Drug/therapeutic agent development costs compared to market
  - ✓ Industry greater dependency on clinical and laboratory scientist collaboration-requires negotiations of IP issues, etc.
5. Also, as part of the Opening Session, **the EUCERD recommendations** were introduced via Skype by **Yann Le Cam – EURORDIS Chief Executive Officer in his** presentation “**PLANS AND STRATEGIES IN THE FIELD OF RARE DISEASES**”.

**a. Main points of the presentation:**

*Mission of EUCERD:* to aid the EC with the preparation and implementation of European activities in the field of rare diseases covering aspects from research to provision of

care, and sharing of expertise and tools.

**b. Three first EUCERD Recommendations adopted as of May 2013:**

- ✓ Centers of Expertise (CE) on Rare Diseases
- ✓ European Reference Networks of Rare Diseases
- ✓ Clinical Added Value of Orphan Medicinal Products information flow (CAVOMP)  
Identified as **key priorities** to be included in NPRD:
  - *Organize provision of care for RD patients;*
  - *Provide adapted services to RD patients;*
  - *Improve access to available therapies at better cost.*

**c. Upcoming EUCERD Recommendations**

*(adopted after the conference, on 5-6 June 2013 )*

- ✓ Rare Disease Patient Registration and Data Collection – These concern the core common governing principles to optimise RD data collection in all EU MS
- ✓ Core Indicators for Rare Disease National Plans - EUCERD Recommendation of a core set of 21 indicators to be used by all EU MS to monitor the planning and implementation of national plans / strategies on RDs (these indicators were selected from the list of 59 indicators adopted in EUROPLAN 2008-2011).

**d. The EUCERD**

- ✓ Advises the EC in developing and implementing its policy and actions on RDs;
- ✓ Provides guidance to EU MS to develop their plans and improve the management of RD patients;
- ✓ Member States, when developing and implementing NPs, apply the EUCERD Recommendations developed with them and for them to:
  - *Adjust their healthcare, social and research structures & budgets to the challenges of rarity;*
  - *Reinforce EU collaboration in an area where scarcity of patients, knowledge, expertise and resources call for a long term vision and an EU & MSs integrated approach.*

**Plenary Session**

The Plenary Session followed the Opening Session and focused on presentation of European documents, and other relevant initiatives such as the EUROPLAN project, the EUCERD Joint Action Working for Rare Diseases (EJA) of which EUROPLAN is an integral part, the EUCERD (EU Committee of Experts on RD) itself, as well as the EU approach on research in the field of RD.

In the **plenary session** were held **four presentations**:

**1. EUROPLAN: support for Member States initiatives in developing NPRD - Dr. Domenica Taruscio, EUROPLAN Project Coordinator**

EUROPLAN, the “European Project for Rare Diseases National Plans Development”, is a source to actively support Member States initiatives project co-funded by the EU Commission (DG-SANCO). It is coordinated by the National Centre for Rare Diseases (ISS, Italy) to promote the elaboration and the implementation of National Plans / Strategies for rare diseases by sharing relevant experiences among countries linking national efforts with a common strategy at European level.

EUROPLAN objectives:

- ✓ To implement consistent national actions within a common EU strategy
- ✓ To facilitate the coherence of national initiatives with identified priority issues and good practices
- ✓ To prepare the ground for synergies and cooperative approaches
- ✓ To establish an interactive network of actors (policy makers, ect.)
- ✓ To share experiences
- ✓ To support National Conferences coordinated by EURORDIS and organized by its National Alliances.

**2. EUCERD Joint Action for Rare Diseases (EJA) - Antonio Atalaia, University of Newcastle UK, leader of EJA.**

Antonio Atalaia introduced the “EUCERD Joint Action: Working for Rare Diseases” which started on 1 March 2012 and will support for a three-year duration the activities and mandate of the Committee. This Joint Action is led by Prof. Kate Bushby, Vice-Chair of the EUCERD. **This Joint Action will build on the achievements of previous European initiatives in the field**, such as the EC Rare Disease Task Force, Orphanet, the EUROPLAN project, and the outputs of and the several rare disease networks that have received EU funding over the past years.

This Joint Action has **five main areas of work**:

- the implementation of plans and strategies for rare diseases at national level
- the standardisation of rare disease nomenclature at international level
- mapping the provision of specialised social services and integration of rare diseases into mainstream social policies and services
- the leveraging of the value of EU networking for improving the quality of care for rare diseases
- the integration of RD initiatives across thematic areas and across Member State

**3. Research on rare diseases in Europe - Dr. Ruxandra Drăghia Akli**, Director at DG Research, European Commission -introduced to the audience the research challenges related to rare diseases. She approached the *huge unmet medical needs of RD patients* and the ways *these might be addressed*:

- Life-threatening, chronically debilitating diseases
  - ~30 million of Europeans are affected or will be affected during their lifetime, doctors often lack knowledge and tools to do correct diagnosis, lack of specific treatments: up to ~90% drug use for rare conditions is off-label.
  - Small patient populations are challenging for the development of diagnostic/therapeutic tools
  - Challenge of (standardized / harmonized) data/sample gathering challenge for clinical trials, due to difficult identification of cases and small patient samples (classic double blinded, placebo-controlled multi-centred, multi-national clinical trials cannot be envisioned...)
  - Research resources (patients, experts, budget) are scarce and scattered.

Dr. Draghia-Akli then introduced the International Rare Disease Consortium (IRDiRC) that the European Commission launched together with the US National institute of Health.

*Why working together?*

- ✓ To mobilize the necessary critical mass of expertise and resources;
- ✓ To avoid overlaps in research allowing for more diseases to be tackled;
- ✓ To deliver new cures and diagnoses to treat patients world-wide.

*Benefits with international cooperation:*

- ✓ Economy of scale as treatments and cures are universal;
- ✓ Allowing to set, and faster reach, ambitious goals;
- ✓ Easier to mobilize the necessary critical mass of expertise and resources;
- ✓ Avoid overlaps in research allowing for more diseases to be tackled.

*Co-operation at international level to:*

- ✓ stimulate
- ✓ better coordinate
- ✓ maximize output

*How does it work?*

- ✓ Identify and define shared strategic goals
- ✓ Agree to approach jointly and pool resources
- ✓ Let each organization use its own funding mechanisms/timing
- ✓ Agree to share data / standards

**4. Orphanet Romania - Definition, codification and inventorying of rare diseases - Dr. Cristina Rusu**

- ✓ A medicinal product shall be designated as an 'orphan medicinal product' when intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition affecting not more than 5 in 10 000 persons in the Community when the application is made. According to the Orphanet database, of the thousands of known rare diseases for which a clinical identification is possible, only 250 of them have a code. Types of classification used by the health care systems are: ICD – 9, ICD-10, OMIM, SNOMED, ORPHAN, MESH, others.

There were no questions after the plenary presentations and the session ended with the ceremony of **awarding winning journalist** launched after ANBRaRo Contest with the theme Solidarity in rare diseases. Marinela Frâncu – National Romanian Radio was awarded by Cristina Vladu - MoH counsellor and Dorica Dan - President ANBRaRo.

## II) REPORTS FROM THE WORKSHOPS

### Workshop 1 – Research on Rare Diseases

#### **Main topics addressed during the Workshop:**

- National Research Programs in Rare Diseases
- National Strategy for Research in Rare Diseases
- Clinical trials supported by public (bodies)
- Number of research projects on RD approved per year (if possible annually since the year before the beginning of the plan)
- The inclusion of social and public health research on rare diseases;
- European research programs;

#### **Presentations given during the Workshop:**

- 1. Rare disease research projects in Romania** - Dr. Ioana Ispas - National Authority for Scientific Research. Topics elaborated :
  - Developing a strategy/national program for research in rare diseases;
  - The existence of national policies for recruiting young researchers especially for the field of rare diseases;
  - Allocate funds for the research program in rare diseases.
- 2. Medical research on rare genetic diseases in Timisoara University Center** - Dr. Maria Puiu - UMF Timisoara
- 3. Microdeletion syndromes** - Dr. Magda Budişteanu - Pediatric Neurology Clinic Al. Obregia
- 4. Research situation in Romania, where are we?** - Dr.Vlad Gorduza, UMF Iasi.

The presentations were followed by debates on how are accomplish in Romania the EUROPLAN indicators and EU Council Recommendation.

#### **The situation in Romania:**

- small number of specialists in medical genetics and the absence of specialized diagnostic centers;
- lack of a national network of specialized medical centers where to make detection, diagnosis and monitoring of patients with rare diseases;
- lack of protocols and guidelines of good practice;
- the collaboration with the European network of centers specialized in rare diseases is sporadic and unstructured (bulk);
- low level of research on rare diseases;

- ✓ Research on RD is included in the general research program. A national strategy for research is needed and RD should be a priority; no monitoring of research programs exists.
- ✓ In Romania there is not a special program for RD, 14% of the General Plan Projects are on Health of which approx. 1% are rare diseases.
- ✓ Clinical trials are funded by public institutions (ANCS and MS). Romania participated with 500,000 Euros to E-RARE, the transnational cooperation project among rare disease research funders.
- ✓ Inclusion of social and public health research on rare diseases is under discussion, but not within the health, but the socio-economic category. Research platforms and other infrastructure are also financed by the research program, e.g.: Capacities Programs and Structural Funds.
- ✓ There is no national policy in the direction of young professionals annually recruited to work especially in the field of rare diseases.
- ✓ There are no specific public funds allocated for RD research. It's hard to say if funds are allocated annually for the action / research projects in the early RD plan, because there is no specific program in the field of RD.

→ **There is a need for a national strategy for research.**

**Research appears in the proposed National Plan for Rare Diseases.** The following activities are listed although no specific budget provisions are made for them:

- organizing training courses on writing grant applications, advice on accessing funding from national and European funding;
- considering rare diseases as a priority in the Romanian medical research: the logistical and financial support to research projects aimed to find new methods of diagnosis and new therapeutic products for rare diseases, through programs of national competitions for research in this area.

In addition, at the EUROPLAN Conference it was concluded that a partnership should be established with the structures competent for research of the Ministry of Education, Research and Innovation (National Council for Research from Higher Education, National Authority for Scientific Research, etc..), by setting a multiannual plan for research on rare diseases, which will have financial support and will enable the launch of an annual competition for research projects. These priorities should be also included in the National Strategy for Research, but at present there is not such a Strategy in Romania.



**Closing remarks of the Workshop:**

**Establishment of the priority areas in research on rare diseases:**

- Epidemiology
- Genetics
- Pharmacology
- Treatment and care
- Social Research
- Drug development. Namely, stimulating the pharmaceutical companies to make investments in rare disease research by establishing a partnership between the Ministry of Health and the pharmaceutical companies to realize clinical studies, to validate the action of these drugs, with respect laws in this area and the rights of patients with rare diseases.

**Setting the priority actions in the field of research on Rare Diseases:**

- Collaborating between research centers and patient organizations to identify the participants to research.
- Creating a database of reference works on rare diseases.
- Publication of research results in leading scientific journals in the field.
- Disseminating research results to practitioners and patients' associations.
- Developing the necessary infrastructure for research.
- Developing animal models for research into rare diseases.
- Develop the protocols for research, for cooperation.
- Initiation of joint research projects.
- Promoting a voluntary and committed research policy, particularly in clinical trials.
- Development of diagnostic tests.
- The collaboration of Ministry of Health in projects such as E-Rare / ERA-NET.

**W2 - Patient care - "patient pathway"**

"**Patient pathway**" is the path traveled by the patient from the first consultation to the physician in the diagnosis, orientation between medical, social and educational to complete treatment and community integration. This ensures continuity of care. Patient pathway gives us information on what might happen in this journey of patient access to medical and social services and this information can be used for the patient but also for planning and organizing services.

Holding this pathway is necessary to manage the quality of care in terms of standardizing care processes. Clinical pathways promote organized and efficient patient care based on evidence-based practice.

**Main topics addressed during the workshop:**

- Information and guidance/orientation (HelpLine)
- Diagnosis, counseling, interdisciplinary evaluation in the CoE
- Specialized medical social services (education, personalized therapies, PIP(personalized intervention plan), psychological counseling, social, educational, orientation to other services in the proximity of patients
- Social, educational services, community integration
- Re-evaluation at CoE

**Presentations given during the Workshop:**

**1. Patient care – Dorica Dan, President of ANBRaRo**

- ✓ existence of Help Line networks (NoRo, Orphanet);
- ✓ referring the patient for a specialized service;
- ✓ good collaboration between specialists, improving the services for patients;
- ✓ The NoRo Center offers: psychological counseling for patients, interdisciplinary approach first-aid courses, training support groups;
- ✓ [www.edubolirare.ro](http://www.edubolirare.ro) – training courses for specialists;
- ✓ The patient with rare disease, with diagnosis or not, usually is looking for information about the disease; in reality, the clinician does not have the needed time to explain more about the patient's illness and what he has further to do; even if s/he receives information, the patient is tempted to seek other patients which have the same diagnosis, to share experience. As a result we have established Helpline NoRo.
- ✓ In perspective of accreditation of Centers of Expertise, it is necessary that the interdisciplinary team of specialists have good communication, longer time for a patient with rare diseases, therefore, a starting point in funding could be the allocation of ***a larger amount of money for the evaluation of patients with rare diseases within the interdisciplinary team.***

**2. The Pediatric Neurology Clinic analysis. Obregia Bucharest in light of current European recommendations – Conf. Dr. Dana Craiu**

**3. Objectives PKU: Screening and Treatment - Carmen Cordea – PKU Life România**

**4. Pediatric Clinic Fundeni experience in care of patients with thalassemia major - Dr. Adriana Diaconu – lecturer- Pediatrics Fundeni.**

The presentations were followed by debates on how are accomplished in Romania the EUROPLAN Indicators and EU Council Recommendation in relation to the care of patients with RD.

### **Closing remarks of the Workshop**

*It was taken stock of the existence of the following:*

- Services and official sources of information and guidance of people with RD in Romania such as: National Authority for Persons with Disabilities, Orphanet HELPLINE NoRo, Myasthenia Gravis Association, Romania, the websites of ANBRaRo member organizations;
- National socio-medical services for patients and their families to promote access to therapy and recovery, such as NoRo Center, Medical Center for Evaluation and Rehabilitation for children and youth "Cristian Serban", Pediatric Neurology Clinic Al. Obregia;
- Training programs for patients and professionals, such as [www.edubolirare.ro](http://www.edubolirare.ro), that should be improved, formalized and would require the allocation of financial resources.

*Needs have been identified, such as:*

- CE designation and their good cooperation with social services;
- The patient circuit, starting from its call to the family doctor via social-medical diagnosis and care.

*Ways to access the information:*

- Internet ([www.bolirareromania.ro](http://www.bolirareromania.ro), <http://anbraro.wordpress.com>, [www.orpha.net](http://www.orpha.net), etc.)
- Patients associations
- Databases
- Information centers for rare diseases
- Centers of Expertise, European Reference Networks of Centres of Expertise (when they will be set up)
- Specialist MF- specialists for children and adults
- Brochures
- Scientific meetings
- Publications in journals
- Informative books: Dr. Covic's Treaty of Medical Genetics and the book Essence in 101 rare disease
- HELPLINE services (the initiator of this service is RPWA /APWR, the Romanian Prader Willi Association, with the support of EURORDIS).

*Sources of information at European level*

- OMIM=Online Mendelian Inheritance in Men
- Orphanet = Database Line information on rare diseases Geneclinics

- EURORDIS=European Organisation for Rare Disease
- EUROCAT=European Surveillance of Congenital Anomalies
- ECARUCA=European Cytogeneticists Association Registry of Unbalanced Chromosome Aberration
- SSIEM=Society for the Study of Inborn Errors of Metabolism
- ESID= European Society for Immunodeficiency
- ESDN=European Skeletal Dysplasia Network
- EuroGentest. It is a network of excellence in genetics. Its aim is to increase access to information / genetic knowledge.

### **W3 - Methodology and Governance of NPRD (National Plan for Rare Diseases)**

#### **Main topics addressed during the workshop:**

- The existence of regulations / laws that support the development of a plan for rare diseases
- The existence of a coordination mechanism
- The existence of an expert committee of support
- The existence of an external body of evaluation / procedure
- Number of priority areas in the plan
- Budget of the Plan / Strategy

#### **Presentations given during the Workshop:**

- 1. Methodology and Governance of PNBR – Dr. Cristina Vladu – MoH counselor**
- 2. Priority objectives PNBR Romania – Dr. Emilia Severin, EUCERD member**
- 3. Ensuring continuity of care for patients with RD through the NPRD – Dan Dorica – President of ANBRaRo**

The presentations were followed by debates on how in Romania the EUROPLAN indicators are accomplished and recommendations of the National Committee of Reference for Rare Diseases.

#### **Main findings/ closing remarks:**

- ✓ In Romania there is a political will to create and implement a NPRD. An advisory committee of experts representing the specialized committees of the MoH and other stakeholders is under approval. Since 2008, a National Committee for Rare Diseases have been established and worked as part of the projects implemented by ANBRaRo/RONARD.
- ✓ There is no external assessment body of the plan at the moment. MoH specialized

committees could accomplish the evaluation procedure.

*The NPRD includes six priority areas (goals):*

1. Development of institutional framework;
2. Development of services for the diagnosis, treatment, rehabilitation and prophylaxis in the field of rare diseases;
3. Improving access to information on rare diseases;
4. Human resource development;
5. Stimulating research on rare diseases;
6. Increasing the role of organizations.

The budget for the National Plan is under discussion at MoH level and might be included in the funding sources of the national strategy of the MoH.

Since 2008 there is a budget for financing treatments for rare diseases in the National Programme funded by Ministry of Health.

Several campaigns organized by APWR, the Romanian Prader Willi Association, and ANBRaRo, the Romanian National Alliance for Rare Diseases, showed that rare diseases is a matter of national level issue. Only 6,000 patients with rare diseases are included in national statistics. However, it is known that there are 1.3 million potential patients. APWR has created a map of social services in Romania, of which only a small fraction is for patients with RD.

A copy of the NPRD was sent to the Ministry of Health together with the EUROPLAN Conference resolution in order to be included in the Public Health Strategy. For now we cannot say that in Romania there is not a funding mechanism for rare diseases, but it is not specifically addressed to the NPRD. All the speakers who attended the introductory session showed their support and stressed the importance of adopting the NPRD. Some of the details are mentioned in the section above (plenary report).

The National Plan addresses both medical and social needs, but in Romania the coordination of these actions is performed by two separate decision bodies (MoH, Ministry of Work, Family, Social Protection and Elderly).

#### **W 4 - Centres of Expertise and European Reference Networks for Rare Diseases**

##### **Main topics addressed during the Workshop:**

- The existence of a policy for the establishment and accreditation of centers of expertise at national / regional level
- Number of existing centers, number of illnesses traced/pursued

- Expertise Centers adhering to standards defined by the Recommendations of the Council-
- Participation of the national or regional expertise centers in European Reference Networks
- Existence of a national and / or regional covering system for information on RD, supported by the government, Help Line?
- The proportion of laboratories that have at least one diagnostic test validated by an external quality control

*Centres of Reference are outlined in National Plan for Rare Disease as following:*

A layered network of centers on three levels:

1. A Reference Centre – one per country; ultra-specialized for a category of diseases selected through national competition;  
A Competence Centre - one per region;  
County medical offices for rare diseases - one per county;
2. The Centres will interact for a good functionality;
3. Centers' facilities include:  
Clinical and para-clinical diagnosis;  
Specific care and social assistance;  
Screening;  
Education.

*The Reference Centre – attributions*

- Managing the national registry for a category of diseases;
- Diagnosis, specialized investigations, initiating therapy, developing a patient management plan;
- Monitoring of patients;
- Correlation of research;
- Developing the best practice's guides;
- Managing the health programs;
- Regular information to interested parties;
- Cooperation with the European network of centers;
- Training of professionals and patients.

*The Competence Centre – attributions*

- Apply best practice's guides provided by the centres of reference;
- Monitor and disseminate information to reference centres and County medical offices for rare diseases;

- Organize screening activities;
- Initiates and updates the database of professionals with expertise in rare diseases;
- Current activities of prevention, diagnosis, therapy, recovery.

**Presentations given during the Workshop:**

- 1. Romanian Network of hereditary angioedema, from the pilot model reference center**  
- Dr. Brândușa Căpâlna
- 2. Multidisciplinary advice center for rare diseases** - Dr. Vasilica Plaiasu
- 3. Experience in management of rare diseases** - Dr. Maria Puiu
- 4. Genetic Center Iasi** - Dr. Cristina Rusu
- 5. Model of standardized multidisciplinary approach in rare diseases: diagnostic and therapeutic protocols in acromegaly** - Dr. Monica Gheorghiu

The presentations were followed by discussions on the current state of CE in Romania.

**Main findings/ closing remarks:**

- ✓ In Romania, currently there is no accredited examination center, but patients known centers experienced in this field through the websites of: SRGM, ANBRaRo, NoRo help-line, Orphanet.

**Premises – Centres should have these features to be qualified as Centres of Expertise:**

- The existence of protocols for diagnosis and treatment
  - Continuous Monitoring
  - A disease registry
  - Involvement of professionals and interdisciplinary teams
  - Labor monitoring
  - Specialized Social Services
- ✓ **Disadvantages** existing in Romania include: infrastructure, access to information, treatment and funding;
  - ✓ Patient organizations are represented to the NCRD – National Committee for Rare Diseases but, still the NCRD is not officially a MoH committee.

**W5 - Treatments and therapies for rare diseases, orphan drugs**

**Main topics addressed during the Workshop:**

- OD number refunded 100%; Time between the OD authorization date of EMEA and the effective date of appearance on the market in the country;
- Time from the market appearance in the country until a positive decision for

reimbursement by public funds;

- Dissemination of information on the treatment of rare diseases;
- National Programs for patients with rare diseases;
- Patient access to authorized treatment of rare diseases, including reimbursement status is recorded at national and / or EU level;
- EUCERD recommendations regarding the clinical added value of orphan medicinal products - the flow of information;
- The existence of a government program in order to use the compassionate therapy in rare diseases;
- In many cases patients have difficulties in obtaining drugs, because of insufficient funding, even if they are included in national programs of rare diseases or there are discontinuities in treatment or inadequate treatment;
- There are several ways of access to orphan drugs: Order nr. 962/2006 for approval of the application of art. 699, paragraph (1) of Law no. 95/2006;
- Compassionate use of drugs for a certain patient (a drug approved for marketing, but cannot be obtained through normal distribution channels in a reasonable time);
- Compassionate use of drugs for a group of patients with an invalidating disease, either chronic or serious, or a disease considered to be life-threatening (the provision of a centrally authorized product);
- Use of off-label drugs, i.e. medicinal products that already exist on the market but they have other indications.

**Presentations given during the Workshop:**

- 1. Evaluation of medical technologies for orphan drugs in the EU and solutions for Romania**  
- Dr. Ioana Bianchi – MoH
- 2. Apamorfina between necessity and need** - Dan Răican - Association Antiparkinson
- 3. Lack of access to specific therapy condemns Romanian patients to suffering** - Ramona Petrean - Romanian Association of Hereditary Angioedemacare

The presentations were followed by debates on how the EUROPLAN Indicators are accomplished in Romania as well as the EU Council Recommendation in relation to the care of patients with RD.

**Main findings/closing remarks**

- In Romania compassionate use programmes are not regulated by legislation.
- In Romania, in addition to the National Plan for Rare Diseases, there are no specific programs that stimulate the development of orphan drugs.
- In the establishment and management of compassionate use programs, patients can



collaborate with representatives of pharmaceutical companies (marketing authorization holders) and can come up with proposals for new indications.

- As support provided to drug manufacturers, they must be guaranteed a quick method of repayment for their products after marketing authorisation.
- In accordance with EU Regulation on Pharmacovigilance, the National Medicines Agency's website has a form where physicians can report adverse reactions.
- Not yet encourages off-label drugs reimbursement. We still need to find a way to conduct clinical trials to prove this benefit.

## **W6 - Social services for patients with rare diseases**

### **Main topics addressed in the workshop:**

- The existence of formal programs to support patients and their families;
- The existence of an official directory of social resources for patients with disabilities;
- Existing national schemes to promote access for patients and their families to Respite Care Services;
- Existing national schemes to encourage the access of patients and their families to Resource Centers for rare diseases which support the integration of patients in daily life;
- Existing public schemes supporting Respite Care Services and Therapeutic Recreation Programs;
- Existence of programs to support the recovery of patients with rare diseases.

### **Presentations given during the Workshop:**

**Workshop 6** 'frame' presentation was immediately followed by debate and in the end additional presentations were given:

- 1. Social Services in rare diseases, presentation framework - Dana Cota** - Vice Mayor City Hall Zalau
- 2. Frambu Center - Lisen Julie Mohr**, Frambu Center, Norway.
- 3. Information Centre for one group of RD - MG-RO** - Dr. Nadia Radulescu, National Association of Myasthenia Gravis

### **Main findings/closing remarks**

*The following have been identified:*

- ✓ The existence of an Official Monitor of social resources for people with disabilities that also include patients with RD – The monitor of social services conducted by Ministry of Work, Family, Social Protection and Elderly People.
- ✓ The existence of a national scheme of resources for people with disabilities, including

people with RD category - National Authority People with Disabilities - National Strategy for protection, integration and social inclusion of people with disabilities in the period 2006-2013, Decision no. 1175 of 29/09/2005, - followed by a national plan "Equal opportunities for people with disabilities - towards a society without discrimination";

- ✓ Existing social security mechanisms for people with disabilities, including people affected by RD provided by legislation;
- ✓ There are sources of information, such as patient organizations websites, information campaigns and informational materials - local and national media campaigns. Media coverage of each of the new services has been proposed;
- ✓ Social services respond to minimum quality standards, with the accreditation system of social services by MMFPSPV.

*The following initiatives need to be supported:*

- ✓ Support to the several initiatives of patient organizations to create **a network of information and coordination of people suspected with RD;**
- ✓ **Reassessment of criteria for defining the degree of disability,** which should be based on the assessment of the disability and not on the disease. The way disability is currently assessed in Romania is not favourable to RD patients, hence assessment criteria should be adapted;
- ✓ Developing a **standard cost for specialized social services** for people affected by RD through a project in this regard;
- ✓ Improving dialogue of the Ministry for Health with the Ministry for Work (MMFPSPV) and improving the collaboration of the latter with the MoH;
- ✓ **Taking best practices from some counties,** more tenaciously to support in front of authorities the cause of RD.

**During the report of the results of the workshops to the plenary, it was suggested to address the main problems in a “Resolution of the EUROPLAN Conference” that will be submitted to the MoH.**

#### **MAY 2013, RESOLUTION OF THE EUROPLAN CONFERENCE:**

**Conference purpose:** facilitating dialogue, participation and involvement of all stakeholders in the field of rare diseases in Romania (patients, professionals, authorities, politicians, industry, media, etc..) in order to address solutions that need to be taken to update the proposals of The National Plan for Rare Diseases.

**Conference format:** two plenary sessions, six workshops, a "debriefing session" and resolution of the conference presentation to be included in the National Plan for Rare Diseases.

**In conclusion,** we can say that **in Romania there is a political decision to implement the NPRD the National Committee for Rare Diseases that is being finalized / formalized under a Ministerial Order.**

**The following steps are proposed for the next stage:**

- ✓ updating the National Plan for Rare Diseases with the proposals made at the Conference EUROPLAN;
- ✓ finalizing the procedure for appointing the National Committee for Rare Diseases & working groups (possibly with European experts as consultants);
- ✓ achievement of the procedure for assessing the Centers of Expertise;
- ✓ breakdown of activities, objectives and identify necessary budget, responsible institutions and potential sources of funding;
- ✓ Inclusion of NPRD in the National Strategy for Health with budget allocated
- ✓ reimbursement of orphan drugs before obtaining marketing authorization, when the pathology is severe and there is no alternative;
- ✓ establishing a national strategy for research for rare diseases;
- ✓ facilitating access for rare disease patients to orphan medication based on centralized procurement;
- ✓ introduction of PTT = temporary therapeutic protocols;
- ✓ introducing genetic “riscogram” - in the basic package of health services;
- ✓ inclusion of representatives of the Ministry of Labor, Ministry of Education, National Health Care Insurance House, CNAS, National Authority for Drugs, ANM, and National Authority for Scientific Research, ANCS, in National Committee for Rare Diseases, CNBR;

**Identifying specific gaps, changes and needs in all themes.** Specific needs or gaps should be identified to stimulate ability to build the process that will continue to unfold in the EUROPLAN aftermath.

The Romanian EUROPLAN Conference offers to the Romanian national health authorities a support tool for the development and for implementing the National Plan for Rare Diseases, according to the EU Council Recommendation on rare diseases and EUROPLAN indicators. Due to the European level of the project activities, the impact is much bigger at national level.

Gaps and changes have been discussed in a one-hour evaluation session (“**Debrief Session**”) which was held immediately after the National Conference between the conference organizers and consultant, Ministry of Health officials, the Project Leader EUROPLAN (Istituto Superiore di Sanità, Rome) and the EUCERD Joint Action Coordinator (University of Newcastle, UK).

## Conclusions of the Final Report

The Romanian EUROPLAN Conference offered to the Romanian national health authorities a support tool for the development and for implementing the National Plan for Rare Diseases, according to the EU Council Recommendations on rare diseases and EUROPLAN indicators. Due to the European level of the project activities, the impact is much bigger at national level.

### Objectives achieved:

- Presentation of the EU level initiatives on rare diseases;
- Specialists and representatives of various institutions identified the following difficulties and solutions in the field of rare diseases:
  - Assessment of the health of any person by the family doctor should be made through a grid performed by geneticist thus for achieving a “riscograma”. There is a grid already performed by Dr. Adrian Pana - Secretary of State MH and in it we can propose to introduce genetic risk – “riscograma”; it would be good if it appeared in the basic package of health services;
  - The creation of a genetic cabinet in each county, able to attract professionals, was proposed. At the moment we have 30 geneticists in Romania, 80% of chronically ill cases are genetic cases;
  - The existence of national socio-medical services for patients and their families to promote access to therapy and recovery, such as NoRo Center, Medical Center for Evaluation and Rehabilitation for children and youth "Cristian Serban", Pediatric Neurology Clinic Al. Obregia;
  - Existence of training programs for patients and professionals, such as [www.edubolirare.ro](http://www.edubolirare.ro), that should be improved, formalized and would require the allocation of financial resources;
  - Services and official sources of information and guidance of people with RD in Romania: National Authority for Persons with Disabilities, Orphanet HELPLINE NoRo, Myasthenia Gravis Association, Romania, the websites of ANBRaRo member organizations;
  - The existence of an Official Monitor of social resources for people with disabilities who also include patients with RD;
  - The existence of a national scheme of resources for people with disabilities, including people in the RD category;

- In the establishment and management of compassionate use programs, patients can collaborate with representatives of companies and can come up with proposals for new indications;
  - Several initiatives of patient organizations to create a network of information and direct people suspected with RD;
  - Reassessment of criteria for defining the degree of disability, which should be based on the assessment of disability and not of the disease (not necessarily in favor of those affected by RD);
  - Developing a cost standard for specialized social services for people affected by RD through a project in this regard;
  - In Romania there is no compassionate use program;
  - In Romania, in addition to the National Plan for Rare Diseases, there are no specific programs that stimulate the development of orphan drugs;
  - Informing specialists about clinical particularities and management of the rare diseases: Prader Willi, Angelman, microdeletion syndrome, etc.;
  - Medical research in rare genetic diseases are carried out in Timisoara: *“Correlation of clinical, genetic and epigenetic, understanding the genomic etiology of diseases Prader Willi / Angelmann: multidisciplinary model approach of rare diseases in Romania”*;
  - Difficulties in diagnosing rare diseases: Prader Willi Syndrome, Angelman, microdeletion syndromes, hereditary angioedema, acromegaly by family doctors, pediatricians, neuropediatrics, which have long term implications on the evolution of the patients; due to these aspects, scientific research is carried out to facilitate early diagnostic, treatment and individualized intervention;
  - Extending the application of special education techniques to stimulate at cognitive level in rare genetic diseases.
- The final Strategy for Research and the National Health Strategy will be finalized at the end of 2013, with funds for research in rare diseases (at the very least, funds for a national registry for RDs, allocated through the ‘Norway Grants’, the norwegian funding programme, and the continuation of funds for the national programme for treatment of RD);
  - MoH has to finalise the evaluation of the innovative treatments proposed to be reimbursed and to update the list of innovative treatments. Also, a methodology for accreditation of CoE and Centers for Genetics has to be agreed by MoH.
  - Recognised need for more international cooperation in research on rare diseases:
    - to align taxonomy, diagnosis and treatment options;

- to optimise scattered and scarce resources (patients, experts, budgets) with a view to accelerate the development of new diagnostic and therapeutic options.
- Timely reflection in view of:
  - The set-up of similar research programs by research funders throughout the world;
  - Omics technologies bring new opportunities and are getting ripe to demonstrate their clinical utility;
  - Rare diseases can be seen as a model for developing personalized medicine approaches;
  - High number of orphan drug designations that need further research for reaching the patients (« crossing the valley of death »);
- There is an advisory committee of experts representing the specialized committees of MH;
- The Research Program on RD is included in the general research program as a priority. There is monitoring for the research program and it is implemented;
- Partnership of ANBRaRo with the Ministry of Health to implement the NPRD.

## Annexe I: Programme

### **EUROPLAN Conference Program**

**24 -25 May 2013, Palace of Parliament Bucharest**

*An event organised under the auspices of the Romanian Health Ministry*

*EUROPLAN National Conferences are organised by EURORDIS in partnership with National Alliance in EUROPLAN project financed within EUCERD Joint Action for Rare Diseases*

<b>Day 1</b>	
<b>09.00-09.30</b>	<b>Participants Register</b>
<b>09.30 – 10.30</b>	<p><b>EUROPLAN opening speeches:</b></p> <p>Representatives of EURORDIS, ANBRaRo (Romanian National Alliance of Rare Diseases), EUCERD, Orphanet, SRGM (Romanian Society of Medical Genetics), APL (Local Public Authorities), ANM (National Agency for Drugs), CNAS(National Health Insurance Agency), MMFPSPV(Ministry of Work), Frambu, Institute Superiore di Sanita, DG Research, other guests, tbc;</p>
<b>10.30-11.00</b>	<b>Coffee break</b>
<b>11.00-12.00</b>	<b>Plenary session:</b>
<b>11.00-11.30</b>	The current situation in Romania: organization of care, information and training on rare diseases, eHealth, eLearning, - Dr. Adrian Pană - Secretary of State MoH
<b>11.30 – 12.30</b>	<p>Eurordis, ISS,ANBRaRo</p> <p>EUROPLAN Project, European Joint Action, NPRD</p>
<b>12.00 -12.25</b>	Definition, codification and inventorying of rare diseases – Dr. Cristina Vladu MoH Adviser
<b>12.25-12.30</b>	Movie – Rare diseases solidarity in Romania



<b>12.30 – 13.00</b>	<b>Session of questions</b>
<b>13.00-14.00</b>	<b>Lunch</b>
<b>14.00 – 17.00</b>	<b><i>Parallel workshops - Current situation of rare diseases in Romania</i></b> <b><i>Coffee break – 15.30 – 16.00</i></b>
<b>Workshop 1</b>	<p><b>Rare Disease Research</b></p> <ul style="list-style-type: none"> <li>- National Research Programs in Rare Diseases</li> <li>- National Strategy for Research in Rare Diseases</li> <li>- Clinical trials supported by public (bodies)</li> <li>- Number of research projects on RD approved per year (if possible annually since the year before the beginning of the plan)</li> <li>- The inclusion of social and public health research on rare diseases;</li> <li>- European research programs;</li> </ul>
<b>Workshop 2</b>	<p><b>Caring for patients with rare diseases - "patient pathway"</b></p> <ul style="list-style-type: none"> <li>- Caring for patients with rare diseases - "patient pathway"</li> <li>- Information and guidance/orientation (HelpLine)</li> <li>- Diagnosis, counseling, interdisciplinary evaluation CoE</li> <li>- Specialized medical social services (education, personalized therapies, PIP(personalized intervention plan), psychological counseling, social, educational, orientation to other services in the proximity of patients</li> <li>- Social, educational services, community integration;</li> <li>- Re-evaluation CoE</li> </ul>
<b>17.00 – 17.30</b>	<b>Reporting the results of both workshops</b>
<b>Day 2</b>	
<b>09.30 – 10.00</b>	<b>Summary of Day 1</b>
<b>10.00 – 11.30</b>	<b><i>Parallel workshops - Current situation of rare diseases in Romania</i></b>

<b>Workshop 3</b>	<p style="text-align: center;"><b>Methodology and Governance NPRD</b></p> <ul style="list-style-type: none"> <li>- The existence of regulations / laws that support the development of a plan for rare diseases</li> <li>- The existence of a coordination mechanism</li> <li>- The existence of an expert committee of support</li> <li>- The existence of an external body of evaluation / procedure</li> <li>- Number of priority areas in the plan</li> <li>- Budget of the Plan / Strategy</li> </ul>
<b>Workshop 4</b>	<p style="text-align: center;"><b>Centres of Expertise and European Reference Networks for Rare Diseases</b></p> <ul style="list-style-type: none"> <li>- The existence of a policy for the establishment and accreditation of centers of expertise at national / regional level</li> <li>- Number of existing centers, number of illnesses traced/pursued</li> <li>- Expertise Centers adhering to standards defined by the Recommendations of the Council-paragraph d) of the preamble</li> <li>- Participation of the national or regional expertise centers in European Reference Networks</li> <li>- Existence of a national and / or regional covering system for information on RD, supported by the government, Help Line?</li> <li>- The proportion of laboratories that have at least one diagnostic test validated by an external quality control</li> </ul>
<b>11.30 – 12.00</b>	<b>Coffee break</b>
<b>12.00 – 12.30</b>	<b>Reporting the results of both workshops</b>
<b>12.30 – 14.00</b>	<b><i>Parallel workshops - Current situation of rare diseases in Romania</i></b>
<b>Workshop 5</b>	<p>Treatments and therapies for rare diseases, orphan drugs;</p> <ul style="list-style-type: none"> <li>- OD number refunded 100%; Time between the OD authorization date of EMEA and the effective date of appearance on the market in the country;</li> <li>- Time from the market appearance in the country until a positive decision for reimbursement by public funds;</li> <li>- Dissemination of information on the treatment of rare diseases</li> </ul>

	<ul style="list-style-type: none"> <li>- National Programs for patients with rare diseases;</li> <li>- Patient access to authorized treatment of rare diseases, including reimbursement status is recorded at national and / or EU level;</li> <li>- EUCERD recommendations regarding the clinical added value of orphan medicinal products - the flow of information;</li> <li>- The existence of a government program in order to use the compassionate therapy in rare diseases;</li> </ul>
<b>Workshop 6</b>	<p style="text-align: center;"><b>Social Services in Rare Diseases</b></p> <ul style="list-style-type: none"> <li>- The existence of formal programs to support patients and their families;</li> <li>- The existence of an official directory of social resources for patients with disabilities;</li> <li>- Existing national schemes to promote access for patients and their families to Respite Care Services;</li> <li>- Existing national schemes to encourage the access of patients and their families to Resource Centers for rare diseases which support the integration of patients in daily life;</li> <li>- Existing public schemes supporting Respite Care Services and Therapeutic Recreation Programs;</li> <li>- Existence of programs to support the recovery of patients with rare diseases;</li> </ul>
<b>14.00-15.00</b>	<b>Lunch</b>
<b>15.00 – 15.30</b>	<b>Plenary session: Reporting the results of both workshops</b>
<b>15.30 – 16.00</b>	<b>Resolution of the conference</b>
	<p><b>Closure of the EUROPLAN conference</b></p> <p><b>Debriefing session</b></p>

## **Annexe II: Stakeholder groups that attended the Romania EUROPLAN conference**

- **Academician / Researcher: 3**
- **Clinician / GP : 10**
- **Healthcare professionals (other than clinician or GP) : 10**
- **Industry : 3**
- **Insurer: 0**
- **Medical / Learned Society : 31**
- **Representative of patients: 48**
- **Politician: 4**
- **Public Administration (local, regional or national): 3**
- **Social assistance : 2**
- **Other: 0**