EUROPLAN National Conferences

CONFERENCE FINAL REPORT

Writing a clear Final Report is as important as the flawless execution of the National Conference on Rare Diseases. Report writing is an essential element to collect the main findings and the outcomes of the debates, so that they can be usefully fed into the work carried out at the national level to develop a RD strategy and into the EUROPLAN Recommendations to draft a National Plan or Strategy on Rare Diseases.

I. General information

Country	Romania
Date & place of the National Conference	**18-19 June, Bucharest, Palatul Parlamentului (The Houses of Parliament)**
Website	**www.bolirareromania.ro** **http://anbraro.wordpress.com** **www.apwromania.ro**
Organisers	RONARD (Romanian National Alliance for Rare Disease), Romanian Prader Willi Association, Romanian Society of Medical Genetics, Orphanet Romania, University of Medicine and Pharmacy Timisoara
Members of the Steering Committee	Dan Dorica – president ANBRaRo Prof dr. Maria Puiu – vicepresident ANBRaRo Prof. Dr. Emilia Severin – University of Medicine and Pharmacy Carol Davila Bucharest Dr. Răzvan Chivu – Adviser Ministry of Health Prof. Dr. Mircea Covic – University of Medicine and Pharmacy Iași, Prof. Dr. Marius Bembea – University of Medicine and Pharmacy Oradea Adrian Severin – Member of European Parliament Rodica Nassar – President of Health

	Commission in Chamber of Deputies Dr. Cristina Skrypnyk – University of Medicine and Pharmacy Oradea Conf. Dr. Cristina Rusu – UMF Iasi, Orphanet Conf. Dr Vlad Gorduza – University of Medicine and Pharmacy Iași Irinel Rădulescu, PR manager APWR Dr. Mihai Gafencu – University of Medicine and Pharmacy Timișoara, Save the Children Dr. Crisitna Vladu Dr. Cristina Isar – GP
Names and list of Workshops	*Workshops* W1 Taining in RD W2 Research, registries for patients in RD W3 Treatment and rehabilitation, orphan drugs, food products W4 European cooperation - EU networks for RD *Plenary Sessions* Official opening. EU recommendations on RD Europlan Project Current situation in Romania P1 Centers of reference and screening P2 Information, access to information P3 Patient Empowerment P4 Monitoring and Sustainability of National Plan of Rare Diseases
Chairs and Rapporteurs of Workshops	Chairs: Dan Dorica – president ANBRaRo Prof dr. Maria Puiu – vicepresident ANBRaRo Prof. Dr. Mircea Covic – UMF Iași, Conf. Dr. Cristina Rusu – president Orphanet Dr. Mihai Gafencu – UMF Timișoara, Salvați Copiii Dr. Cristina Skrypnyk – UMF Oradea Dr. Ioana Rotaru – medical director Fundația ACASA

	Dr. Ancuceanu Robert – vicepresident ANM Prof. Dr. Emilia Severin – UMF Carol Davila București Dr. Cristina Vladu Rapporteurs: Ionela Moacă, Amalia Sabău, Darius Porumb, Iulia Joldeș, Iulia Jurcă-Simina, Florin Jurcă-Simina, Cosmina Băican, Zsuzsa Lazar, Mihaiela Fazacas,
Attachments (programme, list of participants, etc.)	154 participants

II. Main Report

The **Conference Final Report is based on the structure of the National Conference on Rare Diseases (RD), which is common to all EUROPLAN Conferences**. This has been illustrated in the document "CONTENT OUTLINE - Minimum requirements and recommended content for the WORKSHOPS of EUROPLAN National Conferences" (27/11/2009).

Each National Conference is configured in Plenary Session and Workshops. The Workshops will be set up according to specific Themes and will deliver concrete proposals for the Plenary. Each Workshop will be devoted to a specific Theme and will report to the plenary on that Theme.

Conference organisers will have to account for how each Theme and Sub-Theme has been dealt with in the National Conference and what practical solutions or proposals emerged in that area. Therefore the **Conference Final Report will be based on the Workshops' reports and cover the proposal emerged for each of the Workshops' Themes**:

Main Themes

Official opening

Official opening. Dr. Mihai Gafencu introduces participants of the presidium: Cristian Irimie Anton - Secretary of State Ministry of Health, Razvan Chivu – Adviser MS, Petru Armean-professor doctor , Lisen Mohr-from FRAMBU, Dorica Dan – president APWR, Mircea Covic - genetician, Maria Puiu-vicepresident ANBRARO, Luminita Valcea – Romanian Medical College, Mirel Talos – Member of Parliament Salaj County

Dorica Dan emphasizes the importance of the moment, stressing the existence of "a million reasons to act." She gives thanks to those who provided support in making this conference and calls for joint actions of all stakeholders.

Theme 1 - Methodology and Governance of a National Plan / Strategy (NP)

Sub-Themes

1. Mapping exercise before developing a National Plan
2. Development and structure of a National Plan / Strategy
3. Governance of a National Plan
4. Monitoring the National Plan
5. Sustainability of the National Plan

Steps taken by Romania in the field of RD

- 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 9**th**,** 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 with the Ministry of Health Romania – "Rare Diseases, a priority for health care in Romania" in order to finalize and implement the NPRD in Romania;
- (Rare Disease's Day campaigns continued in 2009 and 2010);
- 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;

Partners: Innovation Norway, EURORDIS, Romanian Society of Medical Genetics, Ministry of Health

Current situation in Romania – NPRD (National Plan for Rare Diseases), RONARD (Romanian National Alliance for Rare Diseases) represented by its Chairman, Dorica DAN

The goal to develop the National Plan for Rare Diseases in Romania consists in: 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.

Specific objectives:

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

Work methodology: The Development of the National Plan for Rare Diseases, steps:

1. Identifying needs, areas of intervention and components problems;
2. The actual stage of plan development;
3. Stage of public debate and adjustment - in accordance with the views expressed in meetings organized by the working groups with Ministry of Health, Ministry of Labour, 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 Naţională pentru Protecţia Copilului şi Adopţie), 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"**;
4. Signing the partnership agreement with Ministry of Health on February 29th, 2008: "Rare **Diseases: A Public Health Priority** in Romania";
5. Detailing objectives and activities of the National Plan for Rare Diseases during the meeting of the RDTFR - Rare Diseases Task Force Romania (final version NPRD - sent for approval to more formal forums);
6. Implementation of the National Plan for Rare Diseases in the National Public Health Strategy.

Answers Europlan Indicators - Methodology and governance of the NPRD

RDTFR - Rare Diseases Task Force Romania was created through RPWA – Romanian Prader Willi Association projects (funded by CEE Trust and Innovation Norway). This Committee is composed of representatives of various social actors involved in rare diseases (doctors, patients, representatives of the Ministry of Health, educators, psychologists, social workers, etc...). Up until now several meetings were held together with the representatives of the committee.
Since 2008 there is a budget for Rare Diseases National Program funded by Ministry of Health (treatment).

Several campaigns organized by RPWA – Romanian Prader Willi Association and RONARD – Romanian National Alliance for Rare Diseases showed that rare diseases is a matter of national level issue, starting from the reality that only 6,000 patients with rare diseases are included in national statistics, knowing that are 1.3 million potential patients.
RPWA has created a map of social services in Romania, a small fraction of it is for patients with RD. RPWA intention is to organize staff training in the management of these services for RD patients, for not being necessary to move from another county.

Currently we have started a research project on quality of life of the patients with rare diseases in Romania. First results (unfinished) were presented at the Europlan Conference.
A copy of the NPRD was sent to the Ministry of Health together with the Europlan Conference resolution in order to be introduced in the Public Health Strategy.
In the NPRD there is no timetable activity development set. The schedule and its budget will be completed by the Ministry of Health.
As planned in the project, the members of the committee meet quarterly.
For now we cannot say that in Romania there isn't a funding mechanism for rare diseases.
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 below.

Prof Dr. Mircea COVIC
This conference takes place 1 year after the European Council adopted the European Union Recommendations measures in the field of rare diseases.
It has been advised by prof. Dr Mircea Covic to never get tired of saying that rare diseases are one of the five largest public health priorities according to EU action since 2006.
In Romania there is no complete information on rare diseases. It is not known exactly what we do, how we do, how much we do, as reports from doctors is poor, questionable facts appear.
Regarding the diagnosis and prevention of recurrence, the situation in Romania is more critical than the situation of Bulgaria, Hungary, and Czech Republic. Early identification of patients with rare disease by the family doctor is defective. There is lack of information, doctors are not aware of what is different from what exists as a result delays appear in diagnosis.
80% of rare diseases are genetic diseases and for their correct diagnosis and treatment it is needed to involve all the specialties (possibly reference centers). Regarding diagnostic tests, there isn't a complete and general battery. Practical part conducted by geneticists, creak.
Although in 1996, genetics has been recognized as a clinical specialty, a genuine network of medical geneticists has not been created. Residents have nowhere to be employed and they end up in laboratories doing analysis or research and less work on the clinic. From another

perspective, diagnostic explorations is needed, there are labs, but haven't been authorized as a specialty in the medical laboratory; specialists are needed.

Recommendations:

- In the case of rare diseases with genetic risk of recurrence, the family should be involved!
- Family doctors must be trained in the field of rare diseases!
- Problems related to care - the plan exists, but implementation and acceptance not always depend on specialists;
- The causes must be understood and etiological treatment made!

Problems identified:

- Insufficient involvement of decision factors;
- Professional pride;
- No communication, reduced communication;
- Professionalism is not always updated;
- It required willingness to work as a team!

Adrian SEVERIN - PSD Member of European Parliament

Mr. Adrian Severin presents the recommendations of the European Commission on Rare Diseases. He talks about EU tenders being used in political action plan to consolidate the idea of European institutions, EU involvement in solving important national problems in the field of rare diseases. It calls for joint actions of all citizens for an efficient action of rare diseases.

The EU's role in RD: mobilizing resources from the European states and focus for problem solving, sharing information and expertise, sustaining at EU level the NPRD, strengthen cooperation, creation of networks, expertise centers, professional centers , flow of information on the availability of resources for diagnosis and treatment.

The European Commission recommends the development of national plans through Europlan projects which aims harmonizing the public health strategies in the field of RD. States that have adopted a national plan so far have been mentioned and calls for the adoption of the national plan in Romania.

Cristian Anton IRIMIE

The Secretary of State in the Ministry of Health presented the national program in diagnosis and treatment for rare diseases - the medical treatment is used.

Structure and program actions:

- In the treatment program;
- Involvement - for providing medicines for hemophilia, thalassemia;
- For diagnosis and management of muscular dystrophies;
- Assure dietary products for patients with phenylketonuria.

Promise which has been made: sustainability from the Ministry of Health for patients wih rare problems.

Razvan CHIVU

The adviser of Ministry of Health - presented the activities which had been made within the last six months on behalf of promises made by the Ministry of Health.

The following are mentioned:

- Maintenance of a health program for sustaining RD's;
- Collaboration on NPRD;
- Existence of financial difficulties;
- Implementation of diagnostic and treatment program since 2008;
- Optimization of diagnosis and treatment in the last few years;
- Existence of facts but not of formal documents, official;
- There is no legislative framework;
- Appreciations has been showed for patients associations which mediated the problems to the Ministry of Health;
- Need to create a formal document in which issues met by patients must be written. A resolution is needed in order to implement the NPRD in the National Health Strategy.

Lisen J. MOHR

As Information, Communication and Documentation Coordinator with FRAMBU Norway, partner in the NoRo project, working within a Rare Disease Resource Centre in Oslo, Lisen made a presentation of existing services provided for rare disease patients in Norway.

Theme 2 - Definition, codification and inventorying of RD

Sub-Themes

1. Definition of RD
2. Classification and traceability of RDs in the national health system
3. Inventories, registries and lists

Answers Europlan Indicators - Definition, coding and inventory of RD

- Romania has adopted and uses the European definition of rare diseases
- As an Information Centre for Rare Diseases, RPWA is registered on the National Supervisory Authority for Personal Data Processing
- In Romania there are no official lists of rare diseases, is accepted the Orphanet list
- Classification system used in Romania is mainly ICD 10 for reimbursement, the new system DCI 11 will be adopted when it will be available

In Romania there are at least six ongoing patients' registers: National Registry of hemophilia, National Registry of primary immunodeficiency, National Registry of Baby's Diabetes Mellitus, National Registry of Thalassemia, National Registry of Cystic Fibrosis, and National Registry of Neuromuscular Diseases.

Problem of new registers for patients is represented by the inconsistency between the idea and the implementation, the idea of necessity is the vehicle for years, without being materialized because of impediments at structure and / or management.

Often the impetus comes from setting up registers of patients associations, but the support for the project will depend on the motivations of different personal / professional as PhDs, research projects.

Registers are a necessary condition for ensuring international visibility through access to education and coherent development of research and therapeutic strategies aiming care issues, and also to establish a circuit between the patient and research information.

2.1.Information and training

4. How to improve information on available care for RDs in general, for different audiences
5. How to improve access to quality information on RDs
6. How to ensure adequate training of healthcare professionals on RDs

NPRD (National Plan for Rare Disease) contains requirements on information and training for professionals and all interested parts.

Ways to access the information:

- -Internet (www.bolirareromania.ro, http://anbraro.wordpress.com, www.orpha.net etc.),
  - Patients associations,
  - Database,
  - Information centers for rare diseases,
  - Centers of Expertise, European network of expertise,
  - 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 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.

In the project informational brochures were developed (translated into Romanian), written in an interface language. Some of the brochures are summarized. It is indicated that in the end of the brochures the addresses of all centers which may call for some tests to be included. Brochures were developed by a multidisciplinary team (doctors, psychologists).

Public funds are missing from the information initiatives, most of them are supported by the patient organizations.

Appropriate training is required for professionals. Europlan recommendation on information for specialists is focused on their awareness since college.

Interdisciplinary - complex and comprehensive information from various specialists (doctors, psychologist, social worker, etc)

Information Promotion - performed by collaboration between centers - using others experience.
The information of the general population aim is to prevent and accept / tolerate patients who are suffering from rare diseases.
Information must be permanent, appropriate, based on credible information sources and experts.

Training in RD. Rare disease management involves interaction between different categories of professionals, patients, caregivers.
RPWA professional training provider in rare diseases field:
- Conference workshops and seminars for professionals, patients, their families and other interested persons
One approved course - personal assistant of the person with severe disabilities
Four courses in the course of approval
RPWA proposals:

- innovative training in the field of rare diseases, virtual university www.edubolirare.ro
- the importance of creating virtual channels, online training because of the large number of users of the Internet platform
- The platform aim is to offer training programs authorized by all institutions involved in the rare disease field, from Romania
- Also all courses approved or under approval will have an online component
- Accessibility for training programs - Presentation for access courses: with / without monies , with / without test
- Training methods: blended learning – modern concept implemented in the University of Tromso => combination between classical learning - face to face and learning based on modern Internet - e-learning
- Barriers: inadequate legislation in these forms of learning, national policies that limit such offers of education, lack of technological standards, reluctant attitude of potential customers

Theme 3 - Research on RD

Sub-Themes

1. Mapping of existing research resources, infrastructures and programmes for RDs
2. Needs and priorities for research in the field of RDs
3. Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects
4. Sustainability of research on RD
5. EU collaboration on research on RD
   (presentation: Cristina Skrypnyk)

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

The research appears in the proposed National Plan for Rare Diseases. Thus:

- organizing training courses on writing grant applications, advice on accessing funding from national and European funding
- considering the 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
- establishing a partnership 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.
- Establishment of the priority areas in research on rare diseases
  - Epidemiology
  - Genetics

- Pharmacology
- Treatment and care
- Social Research

- Stimulating the pharmaceutical companies for an investment in rare disease research:

❖ Establishing a partnership between the Ministry of Health and the pharmaceutical companies for 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 research priorities in 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.

Theme 4 - Standards of care for RDs - Centres of Expertise (CoE)/ European Reference Networks (ERN)

Sub-Themes

1. Identification of national or regional CoE all through the national territory by 2013
2. Sustainability of CoE
3. Participation in ERN
4. How to shorten the route to diagnosis
5. How to offer suitable care and organise adequate healthcare pathways for RD patients
6. How to ensure in CoE multidisciplinary approaches and integration between medical and social levels
7. How to evaluate CoE

Centres of reference are outlined in National Plan for Rare Disease as:

• A layered network of centres on three levels:

1. A Reference Centre - 1 / country; ultra specialised for a category of diseases selected through national competition;

- A Competence Centre - 1 / region;
- County medical offices for rare diseases - 1 / county;

2. The Centres will interact for a good functionality;

3. Centres' facilities include:

- Clinical and paraclinical 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 centres;

- 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;

County medical offices for rare diseases – attributions

- Implement screening;
- Solve simple cases and send the complex ones to the centre of competence;
- Information and education/ training (for clinicians in other specialties, patients, etc.);
- Stay in touch with patients' organizations;

Genetic testing and screening - the current situation in Romania

- The chromosome postnatal genetic testing and partial prenatal genetic testing – the lack of equipment and personnel in the main academic medical centres (Iasi, Timisoara, Cluj, Oradea, Craiova, Targu Mures, Sibiu and Bucharest) + private laboratories in Bucharest, Timisoara, Cluj → the absence of standardization and poor quality of external control + no possibility for experts' training (+ the lack of interest in this field)
- Molecular Genetic Testing – there are several laboratories in major academic medical centres (Iasi, Timisoara, Cluj, Craiova and Bucharest) + private laboratories in Bucharest → the lack of standardization and the poor quality of external control + no possibility for experts' training (+ the lack of interest in the field) + no collaboration
- The results of 25 years of activity indicates that genetic county medical services are **useful** and **necessary** in Romania, facilitating: the early identification of congenital anomalies, a more accurate diagnosis, the surveillance and controlled recovery of children with congenital anomalies and the epidemiological study.
- Starting with 1999 we have a National Screening Programme
- Activities: Training of personnel involved in screening; take samples from newborns through dry spot technique; transport samples to the reference laboratory; the neonatal dosage of TSH and phenylalanine; communication of positive results to Public Health Authorities; contacting family and guide them to a service for diagnosis confirmation; confirmation of diagnosis by dosing T 4 and TSH from serum; reference to a specialized center for treatment and monitoring.

- **Limits:** NEONATAL SCREENING IS NOT all over in Romania, delays in testing, insufficient budget, fluctuations in the supply of diagnostic kits by late bidding and contracting, fluctuations in the supply of diagnostic kits due to the discontinuity of funding, deficiencies in the collection technique, deficiencies in the registration of children at harvest, delay in harvesting the low birth weight children who require prolonged hospitalization in maternity, late transport of samples from local screening centres, **delays in identifying children at local level**, delays in the submission of children to confirm diagnosis, poor information on screening of parents in maternity, laboratory personnel migration because low salary scale, inadequate reporting and lack of a database

Proposals for improving the methods of genetic testing:

- Identify laboratories capable of testing from cytogenetics, molecular cytogenetics and molecular genetics - the collaboration of Medical Genetics Commission of the Ministry of Health and the Romanian Medical Genetics Society - and equipping them properly.
- Accreditation of these laboratories and employment of qualified personnel;
- Identify a group of 50-100 rare diseases for which molecular testing should be performed in Romania - the Medical Genetics Commission of Romanian Ministry of Health in collaboration with Romanian Medical Genetics Society;
- Ensure financial support to these laboratories for molecular testing of rare diseases with funds from the state budget or through various sponsorships;
- Identify and establish partnerships with laboratories in the European Community to carry out molecular testing in diseases for which there are no opportunities in Romania – by the Medical Genetics Commission of the Ministry of Health
- Settlement of the medical services provided abroad on the basis of endorsement by a committee of experts - by the Ministry of Health
- Providing a competent genetic counseling, before and after the genetic testing - in regional centres of medical genetics
- Expanding the screening to the national level
- Computerization of reporting (online reporting)
- Identifying new funding sources
- Establishing a network of rare diseases screening by organizing and implementing a populational screening programs
- Establishing general procedures of control which precisely define the stages of implementation and ways of assessing the correct application of the screening methods;

- A systematic assessment before and after the screening program from all stakeholders (Ministry of Health, Public Health Institute, Romanian Society of Medical Genetics, patients' organiyations, etc.)
- Developing a methodological guide for assessing the quality of screening programs - by the Ministry of Health
- Establish an independent advisory committee to examine the effectiveness of screening programs and provide ways to improve implementation
- Develop and training teams of assessors to assist the development and implementation of new methods and screening programs in rare diseases
- Achieve legal framework and detailed procedures necessary to implement screening programs in rare diseases.
- Creating national registers
- Increased international visibility
- Improve collaboration among different actors involved in screening: health policy makers, professionals, patients' organizations, nongovernmental organizations, providers of laboratory materials

4.1. Orphan Drugs (OD)

8. Future of OD
9. Access of RD patients to orphan drugs Pricing and Reimbursement
10. Compassionate use and temporary approval of orphan drugs. Off label use

Conclusions of Treatment and Rehabilitation Workshop, orphan drugs, food product
Presentations: Dr. Ioana Rotaru, Dr. Robert Ancuceanu, Carmen Cordea, Amalia Sabau, Alexandru Ciupitu

In the process of rehabilitation of the patient's, increasing quality of life and integration into society an important role has the access to treatment and appropriate therapies.

1. Rehabilitation

- The rehabilitation is important because develops compensatory mechanisms for patients
- The patient's rehabilitation depends on multidisciplinary teamwork

In Romania there is a distinction between social and medical assistance, but both are important in the rehabilitation process.
Untreated or inadequately treated symptoms can reduce the quality of life (e.g. problem dressing in Epidermolysis bullosa, special diets)

2. Orphan drugs

- 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 can not 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 - target drugs that already exist on the market but they have other indications.

3. Screening:

- Neonatal screening, prenatal in certain rare diseases is important in patient diagnosis and initiation of appropriate therapy (e.g. PKU, epidermolysis bullosa)
- Number of diseases included in neonatal screening program -2
- Number of diseases included in neonatal screening program, properly evaluated – 0

4. Various therapies have an important role in the rehabilitation process.

- should be taken into account the type of disability and patient needs:
  - Physical therapy
  - Physiotherapy
  - Occupational Therapy
  - Sensory therapy
  - Behavioral therapy

Theme 5 - Patient Empowerment and Specialised Services

Sub-Themes

1. Involvement of patients and their representatives in decision-making processes in the field of RDs
2. Support to the activities performed by patient organisations
3. Specialised social services: Respite Care Services; Therapeutic Recreational Programmes; Services aimed at the integration of patients in daily life
4. Help Lines

- The European Council recommends promoting the activities of patient organizations, networking and development of services
- It is recommended to facilitate patient's access to updated information on rare diseases
- Patients can be involved in the development of national plans, in providing information, in policy making regarding the research in the field of rare diseases, in the establishment and management of reference centers / European Reference Networks
- Support for activities carried out by patient organizations is: raising awareness, share best practices and training
- Patient organizations are involved in the decision making process in the field of rare diseases
- Levels of intensity of patient empowerment: information, consultation, involvement, influence, control

The following organizations were presented:

Romanian National Alliance For Rare Diseases – RONARD (Alianta Nationala de Boli Rare Romania - ANBRaRo)

- The goal is to gather all the rare disease organizations in a given country and developing the National Plan for Rare Diseases.

Romanian Prader Willi Association – RPWA (Asociatia Prader Willi din Romania - APWR) – **Offered services:**

- Information and counseling for persons with rare diseases and their family
- Helpline;
- Behavioral therapy for children with autistic spectrum disorders and other disabilities;
- Personalized intervention for patients with rare diseases;
- Training of personal assistants for people with severe disabilities;
- Information and training for specialists in the field of rare diseases;

- Research;
- Lobby and advocacy;
- Socialization and community integration activities for people affected by rare diseases.

Romanian Association of Hemophilia (Asociatia Romana de Hemofilie)

- Collaboration with the Cristian Serban Center from Buzias for treating, educating, recovering and monitoring patients with hemophilia and offering support for the parents
- Editing the newspaper "FACTOR"
- Celebrating the World Hemophilia day – April 17th
- Participation in camp in Austria
- Camp for children in Eforie Nord
- Educational materials (Hemophilia explained to everybody, Treatment book, Hemophilia Card)

National Myasthenia Gravis Association Romania (Asociatia Nationala Miastenia Gravis Romania) **– Services for patients and families**

- Information about the disease and centers of reference in MG
- Psychological, legal, etc. support and counseling
- Creating rehabilitation, medical recovering and social integration programs
- Organizing socio-educational programs, attending training courses, cultural activities, procurement of medical equipment and devices for independent living
- Humanitarian aid for adequate medical treatment
- Informing the public and the media about issues wich are imoptant to people with MG
- Professional education of the involved health care professionals through information materials, symposiums and medical conferences
- Stimulating medical research in this field in order to find the cause and the treatment for the disease myasthenia gravis

PKU Life Romania Association (Asociatia PKU Life Romania)

- Generalization of the neo-natal screening and providing medical foods

Multiple Sclerosis National Society Romania (Societatea de Scleroza Multipla din Romania)
Information and counseling: newspaper "Columna", webpage – www.smromania.ro, Help Line – 0800 800 044, national Seminar on MS themes (addressed to people affected by MS)

Mini Debra - *Epidermolysis bullosa in Romania* (Mini Debra - *Epidermoliza Buloasa din Romania)*

- Proper information and counselling
- Organizing meetings among members
- Helping disadvantaged members

Werdnig Hoffman Association (Asociatia Werdnig Hoffman)

- information and counselling (flyers, newspaper articles, radio and tv appearances, internet)
- recreational activities

Horizontal Themes

"Horizontal Themes" 6 and 7 may be addressed within the Themes 1 to 5. It is of course also possible to organise specific Workshops for Themes 6 and 7. Should the organisers opt for the second option, the specific Sub-Themes for Themes 6 and 7 are identified in the grey boxes of the Content Outline paper. The reporting will of course follow the choice made by the Conference organisers in dealing with the Workshops' Themes.

Theme 6 – Sustainability

In order to assure the sustainability of the National Plan for Rare Diseases, the participants of the Europlan Conference prepared a Resolution with the following text. The resolution was sent to the Ministry of Health and other state authorities, in order to be accepted, so the National Plan for Rare Diseases to be included in the National Strategy for Health, which has to be updated this year.

Resolution of EUROPLAN Conference
Bucharest, Palace of Parliament,
18-19 June 2010

EUROPLAN Conference held in Bucharest in June 18 to 19, 2010, in partnership with the Ministry of Health, under the auspices of the Health Committee of the Chamber of Deputies by ANBRaRo - National Alliance for Rare Diseases Romania, SRGM - Romanian Society of Medical Genetics, and Orphanet Romania - University of Medicine and Pharmacy Timisoara – UMFT, had adopted the following resolution:

1,3 million of Romanians are affected by rare disease in their lifetime

Rare diseases are a threat to the health of EU citizens to the extent that they are life threatening or chronically debilitate of diseases with a low prevalence and high complexity. Despite their rarity, there are so many different diseases, that millions of people are affected.

The White Paper entitled 'Together for Health: A Strategic Approach for the EU 2008-2013 ", from October 23, 2007, the European Commission sets up the EU strategy on health Rare Diseases are included among the priority areas.

***According to European Council recommendations (8 June 2009), on action of rare diseases**, Member States should involve patients and patient representatives in developing policies and to promote the activities of the patients.*

In addition, Member States - hence, Romania should develop and adopt as soon as possible (and preferably no later than the end of 2013) a plan or a strategy aimed at guiding and structuring the relevant actions on rare diseases within the social and health systems.

*The Ministry of Health and the **National Alliance for Rare Diseases Romania** (ANBRaRo) have agreed a partnership which aim was development of **National Plan for Rare Diseases** (PNBR) Romania. The National Plan for Rare Diseases in developed under this partnership will underpin decisions on the allocation of resources devoted to programs of Rare Diseases since 2008.*

The National Plan for Rare Diseases

Objectives:

- *Ensure proper institutional framework for the development of preventive, diagnostic, treatment and rehabilitation for patients with rare diseases and the development of regional reference centers;*
- *Creation of the Registries for Rare Diseases;*
- *Development of screening programs - to avoid default and late diagnosis, complications and high costs of subsequent treatment.*

Increasing the quality of services for patients with rare diseases :

- *Improving the acces to information about rare disease (including the initiative of a website - **www.bolirare.ms.ro**)*
- *Development of continuous service / cross the dianostic / treatment / rehabilitation for patients with rare diseases;*
- *Ensuring access to medicines, foods medicine, assistive devices and compensation costs;*
- *Stimulate the research on rare diseases aria,*
- *Stimulating the development of human resource in diagnosis and management of rare diseases.*
- *Development of Human Resources involved in the diagnosis and management of rare diseases.*

Europlan partners ask The Ministry of Health:

- *To include the National Plan for Rare Diseases (PNBR) in the National Public Health Strategy;*
- *To include requirements of PNBR to be developed by the Ministry;*
- *To create a joint working group which include the experts from the Ministry of Health, specialists and patients. The aim is to implement the principles of PNBR as they result from the European Council Recommendation.*

By including the PNBR in the National Public Health will result the increasing of the quality of life for patients with rare diseases as well as economic efficiency of the health system.

The later diagnosis of rare diseases caused by the lack of specialist, the lack of screening programs or the lack of information to patients and their families, lead to complications with high costs to patients - in terms of emotional development and integration into society and the system health.

Made today, 19 June 2010.

Message of Romanian Doctors Collegium

A rare disease is a disease affecting less than 1 in 2,000 people. In general, rare diseases are usually without treatment, recognition and appropriate care. Although the number of patients affected by each rare disease is small or very small; overall rare disorders would be a public health problem in any civilized country. This goal is linked to the large number of rare diseases: over 8000. According to the European Community, 6-8% of the population has one o f these diseases, which reported to the Romanian population means a minimum 1,000,000 people.

In most cases, rare diseases are little known (and implicitly recognized) by health professionals and by officials of the health system, because of the low frequency of each disease and the high number and complexity of the diseases. Lack of knowledge about these diseases often generates errors in diagnosis and delays in the implementation of the specific care, which constitutes a supplementary source of suffering for patients and families. Moreover, most of these diseases are multi-systemic, requiring multidisciplinary care and having a genetic etiology, which correlates with the risk of transmission of the mutation and thus the presence of the disease at other family members.

The severity of rare diseases is extremely variable; they often cause physical, mental and/or sensory difficulties. In addition, rare diseases are chronic conditions and in most cases they have no specific treatment, just a series of palliative measures that allow patients to improve their condition.

Medical and scientific knowledge of these diseases is incomplete, the pathogenic mechanism is known only in approximately 10% of the disease. Considering this, it is not

surprising that getting the correct diagnosis can often take months or years, and generating unnecessary expenses for the family and the society.

Since rare diseases affect over 25 million people in Europe, EU Health Commission is concerned about the stimulation of research in this area, about creating possibilities for the education of the health care professionals, patients and the general population, with the stated goal of ensuring conditions for correct and early diagnosis of these diseases; of preventing hereditary transmission and of ensuring a better life for these patients.

The Romanian context of rare diseases is very different from the European one, which requires a series of concrete measures that Romania should take for the benefit of hundreds of thousands of patients; as other countries from Europe did. These actions are very urgent, as in a rough estimation more than 95% of these patients doesn't have yet a proper and/or complete diagnosis and do not receive treatment or proper care

*Although Romanian conditions are difficult, involving a lot of generous people from various social sectors (patient associations, doctors, other professionals) it was possible to create the Romanian National Alliance for Rare Diseases – RONARD (Alianta Nationala pentru Boli Rare Romania – ANBRaRo), which has established cooperation with the Ministry of Health and the Romanian Society of Medical Genetics. In this context, it should be mentioned the tripartite partnership between Romanian Prader Willi Association, Romanian Ministry of Health and the Norwegian State; which allows the implementation of the project "**Norwegian-Romanian (NoRo) Partnership for Progress in Rare Diseases**", with financial support from the Norwegian government, through the **Norwegian Cooperation Programme for Economic Growth and Sustainable Development in Romania**.*

A National Committee for Rare Diseases was created through this project; which is composed of representatives of rare disease patient organizations, specialists (doctors, psychologists, social workers, teachers, etc...) and recently, representatives of the Ministry of Health. An important objective of this committee is finalizing the National Plan for Rare Diseases in order to be introduced in the National Strategy for Public Health (a draft of the National Plan for Rare Diseases is attached).

The EUROPLAN project supports this work by having the goal of providing tools for the development and implementation of National Plans for Rare Diseases to the National Public Health Authorities, closely following the European recommendations agreed by all Member States (2009 / C 151/02) .

The National Plan for Rare Diseases (NPDR) aims to provide a series of concrete measures to enable a coherent structure and organization of the system of diagnosis, treatment, care and prevention of rare diseases, which is able to respond to the rightful grievances of those affected by a rare disease and their families. This plan is the result of an extensive consultative process involving the participation of experts in rare diseases (geneticist and other specialties), of rare disease patient organizations and of health policy specialists from the Ministry of Health, Ministry of Finance and the Ministry of Labour, Family and Social Protection. The main priorities of the plan are:

1. *creating organizational structure by establishing a network of reference centers, with branches in the major medical academic centers and (solid and long term) financial support for their continuous functioning;*
2. *conducting epidemiological studies and creating a national registry on rare diseases;*
3. *establishing an adequate national strategy for ensuring measures of prevention and diagnoses;*
4. *establishing a national strategy for providing treatment and rehabilitation of patients with rare diseases;*
5. *recognition of the special features of rare diseases by health and social protection authorities;*
6. *improved training of medical personnel and other specialists to be able to recognize a rare disease and to provide adequate care to patients with such a disease;*
7. *improving access to information for patients, medical professionals and the general public on rare diseases;*
8. *recognizing the special needs of people affected by rare diseases and development of measures to support patient organizations;*

9. *stimulating research on the management of rare diseases, in order to improve the diagnosis and treatment processes in rare diseases;*
10. *collaboration with EU and international organizations and networks;*

Taking note about this initiative, which involves collaboration between physicians of various specialties, health policy authorities and patient organizations, the Romanian Doctors Collegium welcomes the efforts made towards modernizing the Romanian health system. In addition, as representative of all doctors in the country, the Romanian Doctors Collegium has the moral duty to engage in any activity that improves the diagnosis, treatment and prevention of any human disease, whether it is common or rare.

Prof. dr. Vasile Astărăstoae
President - Romanian Doctors Collegium

SOLIDARITY FOR HEALTH

Salaj parliamentary group wants to present the manifesto "Solidarity for Health" - in order to obtain the support of all parliamentary groups from the Chamber of Deputies and Senate to improve the quality of life of patients with rare diseases in Romania. To achieve this goal we request:

- *Equal and early access to diagnosis, treatment, rehabilitation and community integration for patients;*
- *Diversification and improvement of information and training resources for patients and specialists;*
- *Ensuring the sustainability of research projects at national level;*

- *Participation in European Networks of Expertise and nominating the national Reference Centers;*
- *Ensuring the screening at national level;*

In 1995, the WHO categorized 5000 rare diseases. Today, there is talk of over 8000 such diseases. Depending on the disease, it can register between a few to a few thousands.

In most cases, rare diseases are little known (and implicitly recognized) by specialists and by officials of the health system, because of the low frequency of one disease, the high number and the complexity of these diseases.

The lack of knowledge on the disease often causes wrong diagnosis which can lead to incorrect treatment, inefficient allocation of resource and additional suffering for the patient and family.

As from the medical point of view we need multidisciplinary teams - which involve both scientific expertise and professional competences - to approach these diseases; to address the social dimensions is needed our collaboration, regardless of political color or other personal or group interests.

We believe it is in the interest of all of us to adopt a National Strategy to address rare diseases and the National Plan for Rare Diseases is the answer to this problem.

Also, we ask the Ministry of Health to be involved in the adoption and implementation of the National Plan for Rare Diseases, as the government's response to a public health issue, a priority in the European Union.

Signed by all the deputies of Salaj County in the Romanian Parliament

Theme 7 - Gathering expertise at the EU level

Workshop: European cooperation - European network of rare diseases

Existence of a national and/or regional information system of Rare Diseases supported by government

- Orphanet network is a more important resource for specialists, but also for patients. Is a complex network of information, which develop in Romania and hopefully soon will also have sections in Romanian.
- Orphanet project became a project for the EU Joint Action, supported by European funding, the Member States.

Help line for patients

- there are some organizations which provide this kind of service
- APWR – accreditation/and government financial support for information and counseling 080 080 11 11
- EUROPEAN NETWORK OF RARE DISEASE HELP LINES (Rapsody Project)

Help line for specialists

- Nothing yet

Clinical guidelines

- We intend to translate clinical guidelines for patients (which exist at the international level)
- PWS educational package, guidelines for clinical. professionals, parents and crisis situations (IPWSO) – translated

Promoting of training and educational awareness campaigns among professionals and patients

- Rare Diseases Day
- EUROPLAN project
- Treat NMD
- NoRo
- Eurogentest

Conclusion of the Final Report

The Romanian Europlan Conference offers to the Romanian national health authorities support tool for the development and for implementing the National Plan for Rare Diseases, according to the European 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 Member States initiatives on rare diseases
- Forming a National Committee for Rare Diseases and support this committee's quarterly meetings from the Project Norwegian-Romanian (NoRo) Partnership for Progress in Rare Diseases funded by Innovation Norway;
- Partnership with the Ministry of Health;
- Support for the initiative from the Health Commission of the Chamber of Deputies, from the Parliamentary Group of Salaj County and from the Romanian Doctors Collegium;
- Prepare a list of priority areas of intervention and actions included in the National Plan or Strategy for Rare Diseases;
- Developing indicators for monitoring the implementation and for assessing the impact of the National Plan;
- Identifying past and current experiences - academic centers with expertise on different rare diseases;
- Identifying existing and accredited social services in our country, services that can be accessed by patients with rare diseases after an adequate training of the professionals working in these services;
- Development of a resolution to implement the National Plan for Rare Diseases

Debates were stimulated to achieve consensus on the importance of the National Plan for Rare Diseases. Were present representatives of patients, specialists, local and national authorities, Members of European Parliament and representatives of the Ministry of Health (154 participants, of which 51 medical specialists, 10 politicians, 4 representatives of the Ministry of Health, 35 other specialists, 1 representative of FRAMBU Center , 52 patients and 1 representative Innovation Norway);

III. Document history

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