



**Alianța Națională  
pentru Boli Rare  
România**



*Romania*

## **EUROPLAN NATIONAL CONFERENCE**

*in the framework of the EU Joint Action RD-ACTION*

*Zalău, 16-17 November 2017*

**FINAL REPORT**



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# FOREWORD

The EUROPLAN national conferences or workshops are organised in many European countries as part of a coordinated and joint European effort to foster the development of comprehensive National Plans or Strategies for Rare Diseases addressing the unmet needs of patients living with a rare disease in Europe.

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

The EUROPLAN national conferences/ workshops are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS–Rare Diseases Europe. **Rare Disease National Alliances and Patient Organisations have a crucial role to shape the national policies for rare diseases.**

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

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

Since 2008, National Alliances and EURORDIS have been involved in promoting the adoption and implementation of National Plans and Strategies for rare diseases. Altogether, 41 EUROPLAN national conferences took place in the framework of the first EUROPLAN project (2008-2011) and the EU Joint Action of the European Union Committee of Experts on Rare Diseases - EUCERD - (2012-2015).

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

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

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

## GENERAL INFORMATION

<b>Country</b>	Romania
<b>National Alliance (Organiser)</b>	Alianța Națională pentru Boli Rare România / Romanian National Alliance for Rare Diseases
<b>Date &amp; place of the national workshop/conference</b>	16 <sup>th</sup> -17 <sup>th</sup> of November 2017, Zalău, Romania
<b>Website</b>	<a href="http://www.bolirareromania.ro">www.bolirareromania.ro</a>
<b>Members of the Steering Committee</b>	Dorica Dan – chair Prof. Dr. Emilia Severin – co-chair Members: Prof. Dr. Maria Puiu Prof. Dr. Cristina Rusu Florina Breban Zsuzsa Almási
<b>List of Themes addressed</b>	<ul style="list-style-type: none"> <li>– State of the art of Rare Diseases in Romania – monitoring the NPRD;</li> <li>– Accreditation of Centers of Expertise (CE); Participation to ERNs;</li> <li>– Integration of the rare diseases in social policies and social services at national level;</li> <li>– Electronic patient registries;</li> <li>– Improving the access to treatment;</li> <li>– Training in rare diseases;</li> <li>– Promoting Romanian research projects for RDs.</li> </ul>
<b>Annexes:</b>	I. Programme in English II. List of Participants (by stakeholders' categories)

## FINAL REPORT

### I. Introduction / Plenary session

#### - The current situation of rare diseases in Romania - monitoring of NPRD - priorities

The topics addressed in monitoring the NPRD were, among others: access to information, treatment and rehabilitation therapies, Centres of Expertise (CE) and European Reference Networks (ERNs), patient registers (NoRo registry), specialized social services, research, training.

Romania has taken important steps to improve access to treatment and care for patients with rare diseases. Romania has representatives in the Committees of Experts of the European Commission, the National Committee for Rare Diseases was established, the Centers of Genetics had been officialized, the first Centers of Expertise have been accredited and have joined the European Reference Networks.

In Romania, the National Plan for Rare Diseases (NPRD) was adopted by the Ministry of Health at the end of 2013 and was subsequently included in the National Public Health Strategy for 2014-2020, through Executive Order no. 1028/2014. RARE DISEASES, A PRIORITY FOR PUBLIC HEALTH!. (<http://www.ms.ro/wp-content/uploads/2016/10/Anexa-1-Strategia-Nationala-de-Sanatate-2014-2020.pdf>)

National Public Health Programs provide patients with rare diseases access to a range of treatments and orphan drugs, their number increasing every year. The National Program for Treatment of Rare Diseases (<http://www.cnas.ro/page/programul-national-de-tratament-pentru-boli-rare.html>) and other programs for the treatment of patients with rare diseases (<http://www.cnas.ro/category/lista-programelor-nationale-de-sanatate.html>) are funded through the legislation approving the implementation methodology of the National Public Health Programs and the National Health Care Programs for the years 2015, 2016 and 2017.

Today, rare diseases in Romania are recognized as a priority of the public health system. The Romanian National Alliance for Rare Diseases (RONARD) has taken actions in this sense and in 2008 signed the first partnership with the Ministry of Health to implement the NPRD, partnership renewed in 2014.

In Romania, the National Council for Rare Diseases (CNBR) was created in October 2013 by Ministerial Order no. 1215/2013. The National Council for Rare Diseases is an interdisciplinary scientific body of rare disease experts, without legal personality, with consultative role for the Ministry of Health. The Council's objectives are:

- technical guidance for organizing a nationwide functional network for diagnosing and treating rare diseases;

- providing expertise in the execution of therapeutic interventions in the field of rare diseases at the level of the European and international standards.

An important step in the field of rare diseases and in recognition of CEs in Romania was the official recognition of the Medical Genetics Network through Order no. 1358/2014 on the establishment of the network of medical genetics. Thus, six genetic centers were recognized at national level: Cluj, Bucharest, Timis, Bihor, Iasi, Dolj (<http://legislatie.just.ro/Public/DetaliiDocument/163135>).

Health insurance does not cover the costs of all genetic tests. Some genetic tests are free of charge (budget allocated to tests is limited) for children and adults who are enrolled in the national health program for congenital malformations. Research projects in Romania are sometimes also used to create access to free genetic testing for patients and families. Romania included provisions for cross-border genetic testing under its National Plan for Rare Disease. A number of specific genetic tests, which are not yet available in Romania, are available abroad. Patients with RD may use the S2 or E112 form (according to the situation) for medical care abroad.

Executive Order no. 1028/2014 on the approval of the National Public Health Strategy 2014-2020 and the Action Plan 2014-2020 for the implementation of the National Strategy includes interventions for rare diseases:

- Improving the quality of care for patient with rare diseases through the care chain;
  - ✚ organizing the specific services within a functional network of competence and reference centers according to the European-recommended practice and defining the mechanisms for collaboration between them
  - ✚ improving infrastructure, with priority the reference laboratories, to increase in-depth diagnostic capacity, including pre-/ post-natal diagnosis
  - ✚ expanding the use of ICT solutions in the registration of rare diseases at national level and creating rare disease registries, including the hemophilia registry (according to SO 6.1c)
  - ✚ involving first-line health services in the care of the patient with rare diseases and stimulating collaboration with community social services and patient organizations
- Ensuring access for patients with rare diseases to specific therapy and dietary treatment:
  - ✚ identifying more effective mechanisms/solutions for financing orphan medicinal products (OMPs) in a transparent allocation policy for available resources
- Reviewing treatment guidelines / protocols for patients with hemophilia in light of recent evidences and recommendations of European structures and reconsidering specific treatment access modalities, based on the principle of cost-effective, involving patient organizations
- Improving the methodological framework and the technical competences of the professionals by creating practice guides, continuous training, creating periodical recommendations of the specialized committees of the MoH for evidence based updating the educational plans of the higher education units, increasing the degree of involvement of the Romanian professionals in European and international initiatives for exchange of information
  - ✚ developing temporary therapeutic protocols, depending on the available evidences

- ✚ defining a system for "compassionate use" of orphan medicinal products for patients with rare diseases, as recommended by the EUCERD and in line with the requirements of the EMA

- Network of medical genetics - Order no. 1358/2014 on setting up the network of medical genetics
- Order no. 540/2016 on the organization, operation and methodology of certification of centers of expertise for rare diseases

## II. Specific themes:

### Theme 1: Centers of Expertise Accreditation

A Center of Expertise for Rare Diseases (CE) is a functional, non-legal structure involved in the diagnosis and provision of accessible and effective healthcare to patients whose medical condition requires a special focus of expertise in medical areas where expertise is rare.

In Romania the CE demonstrating its functional, organizational and administrative capacity in the provision of integrated services to a rare disease or a group of rare diseases, can apply for accreditation if the minimum criteria established by the Health Ministry Order no. 540 /2016 are fulfilled. (<http://legislatie.just.ro/Public/DetaliiDocument/178054>)

Applications accompanied by the self-assessment form and documents demonstrating compliance with the criteria of the EU Committee of Experts on Rare Diseases (EUCERD) must be submitted to the County Public Health Departments, which are responsible for evaluating the documents and submitting them to the Ministry of Health.

So far, the designation process has been finalized for nine healthcare providers in Romania:

1. The Center of expertise for Rare Diseases in the Field of Malformations, Developmental Anomalies and Rare Disability Disorders within the Regional Center of Medical Genetics, structure of the Municipal Clinical Hospital "Dr. Gavril Curteanu" Oradea;
2. The Center of expertise for rare diseases in the field of congenital malformations associated with mental impairments of the Regional Center of Medical Genetics Timis, structure of the Emergency Clinical Hospital for Children "Dr. Louis Turcanu" Timisoara;
3. The Center of expertise for rare diseases in the field of congenital malformations within the Center of Expertise in Prevention, Genetic Diagnosis and Management of Rare Diseases, Structure of the Craiova County Emergency Clinical Hospital;
4. The Center of expertise for rare diseases in the field of rare diseases and autism spectrum disorders, of the Reference Center for Rare Diseases NoRo Zalău;
5. The Center of expertise for rare diseases in the field of malformations / development anomalies and rare intellectual disabilities, within the Regional Center of Medical Genetics, structure of the Children's Emergency Clinical Hospital Sf. Maria Iasi;
6. The Center of expertise for rare diseases in the Field of Pediatric Neurology, Clinical Department of Pediatric Neurology, structure of Clinical Psychiatric Hospital Prof. Dr. Alex. Obregia București;
7. The Center of expertise for rare diseases in rare pediatric liver diseases within the Pediatric Clinic Section II, structure of the Children's Emergency Clinical Hospital Cluj-Napoca;

8. The Center of expertise for rare diseases in the field of rare cardiovascular diseases within the Cardiology Section III, structure of the Cardiovascular Disease Emergency Institute Prof. C.C. Iliescu Bucharest;
9. The Center of expertise for rare diseases in the field of thyroid cancer, parathyroid tumors and neuroendocrine tumors of the Nuclear Medicine Laboratory and Endocrinology Department, structure of the Oncological Institute "Prof. Dr. Ion Chiricuță" Cluj-Napoca.

Patient registries for rare diseases are still a challenge for this field, it is obvious the need for a National Registry of Rare Diseases with a minimum data set to support policy decisions. For the time being, there are specific registries on diseases or disease groups administered by clinicians or patient associations, some of them having international collaborations.

### **Theme 2: The participation of the Centers of Expertise in the European Reference Networks**

The objective of the European Reference Networks is to ensure improved access for patients to healthcare in medical fields where expertise is rare, safe, high-quality diagnostics and highly specialized medical care, through highly specialized cooperation and by combining knowledge. Thus, Member States with an insufficient number of patients will be supported to provide highly specialized healthcare and rapidly benefit from innovations in science and health technology.

The need to create national networks by association of accredited centers of expertise on clusters of diseases, is evident from the need to provide integrated and efficient services. Through these networks, diagnostic capabilities will be improved (most RDs are still untreated, difficult to diagnose, some undiagnosed). It will also ensure continuity of care and treatment (medicines, recovery treatments, essential social services to improve the quality of life of patients and their families), development and use of standardized tools (clinical guides, registries, patient pathways) to ensure specialized high-quality care at national level. Once accredited, the national centers of expertise can join the European Reference Networks.

European networks where Romanian service providers are members: ERN EpiCARE, Endo-ERN, ERN Skin, ERN ReCONNET, ERN GUARD-HEART, ERN ITHACA.

### **Theme 3: Specialized social services and rare disabilities**

There are 6000-8000 rare diseases, and for 5000 rare diseases there is no treatment anywhere in the world. Lack of information on illness and impact on life, lack of experts and resources, lack of good practice guides, difficulty in transferring information and knowledge to ensure continuity of care remain challenges that make life difficult for patients and families.

Research shows that 1/3 of the patients have reduced, discontinued, or never had a professional activity due to the disease. One can observe unequal access to care and treatment services depending on the patient's area of residence and level of access to information. Patients with rare diseases also argue for the need to update the criteria for classifying the disability level, considering that these are not taking into account the complexity level of these diseases.

RONARD signed the Partnership Agreement no. 4357/27.04.2016 with the National Authority for Persons with Disabilities, which contains specific actions to ensure the continuity of care through integrated services that respond to the needs of people affected by rare diseases, including collaboration with the CEs and collaboration for updating the criteria for classifying the disability level.

The National Strategy on Social Inclusion and Poverty Reduction 2015-2020, key initiative proposed for 2015-2017, has as Ob. 3. the development of integrated social services at community level - CASE MANAGEMENT.

By Executive Emergency Decision no.18/2017 was created the legal framework for Community medical assistance (<http://legislatie.just.ro/Public/DetaliuDocument/186978>). Community health care includes all programs, health services and public health actions provided at community level to increase the access of people and, in particular, vulnerable groups to health services, especially focused on prevention. The purpose of community health care is to improve the health of the population by ensuring equitable access to health services for all people in each community, regardless of their socio-economic status, their level of education, their location in rural or urban areas or distance from the medical service providers.

The local public administration authorities are responsible for providing community-based healthcare services to the population, especially for the vulnerable groups (from a medical, economic or social point of view), under the law and within the limits of existing human and financial resources.

Decision no. 797/2017 for the approval of the framework regulations for organizing and functioning of the public social assistance services and the guiding personnel-structure (<http://legislatie.just.ro/Public/DetaliuDocument/194962>) was also adopted, which introduces case managers to local authorities for several categories of vulnerable people, including those affected by rare diseases.

To improve access to information, there are several patient-led services in Romania, of which the only helplines included in the European Network of Rare Disease Helplines are the NoRo Helpline and the Myasthenia Gravis Helpline. The Orphanet team in Romania is coordinated from Iasi.

### **III. Gaps, challenges, needs:**

- difficult access to diagnostic and treatment services for rare diseases;
- finding solutions to prevent parallel drug exports and avoiding the disappearance from market of essential drugs (e.g. immunoglobulins);
- updating European clinical guidelines and initiating new guidelines (where not available) for the care of patients with rare diseases;
- permanently updating the list of drugs, support equipment, medicine food and other products reimbursed in the National Rare Disease Program;
- solving the problems of patients with severe respiratory problems (DMD, CF, PWS) and provision of equipment (CPAP, BPAP, ventilators etc.) through National Programs, training specialists in this field and establishing a clear circuit for access to oxygen therapy;

- promoting integrated care, inter-sectoral and interdisciplinary collaboration;
- monitoring the work of accredited expertise centers.

#### **IV. Conclusions:**

##### **Theme 1:**

- The Ministry of Health - through the message sent to the Europlan Conference - stressed that rare diseases remain a priority for MoH and encourages continuing the accreditation of centers of expertise and their participation in the European Reference Networks at the future call of the European Commission in 2018.

##### **Theme 2:**

- The need to set up the Pilot National Rare Disease Registry through the creation of a consortium of accredited centers of expertise, adopting a minimum common data set to be communicated in the national registry;
- Initiating a collaboration strategy for research projects on rare diseases, correlating the research plans of accredited expertise centers.

##### **Theme 3:**

- Project identification and collaboration with the Ministry of Labor for the training on rare diseases of the personnel in social services at local level (e.g. case management, rare disease management, recovery teacher, personal assistant of the person with severe disability etc.), community support networks to ensure case management and co-ordinate patient care;
- Active participation in the Ministry of Labor working group for updating the criteria for classifying the disability level for patients with "rare disabilities";
- The Ministry of Labor representative promised to reactivate this working group that was initiated in the previous government;
- Initiating projects that encourage the employment of people affected by rare diseases, clarifying the situation of sheltered workshops and protected units, flexible working time contracts, coaching, adapting workplaces to the needs of the person with rare diseases;
- Development of respite care services for patients and families and establishment of group homes for adults with rare diseases.

*I, the undersigned KUPÁS ZITA, authorised interpreter and translator for English and Hungarian based on license no. 36360/2013 issued by the Ministry of Justice from Romania, hereby certify the accuracy of the translations from Romanian to English, that the presented text was complete, without omissions and the text's content and meaning were not altered by translation.*

*The translation of the submitted text consists of 7 pages and it was performed following the written application forms, achived by the undersigned.*

*AUTHORISED INTERPRETER AND TRANSLATOR  
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**EUROPLAN CONFERENCE AGENDA 2017**  
**16-17 November 2017, Zalău, Severus Hotel**

Day 1 – 16 <sup>th</sup> of November 2017	
12.00 – 13.00	Registration of participants & Lunch
13.00 – 14.00	<b>Official – State of the Art in Rare Diseases / Implementation of the NPRD (Representatives of: Ministry of Health, RONARD, Salaj Public Health Directorate, Local Authorities), moderator Prof. Dr. Maria Puiu, president of Romanian Medical Genetics Society, Medical University Timișoara</b>
14.00 – 15.30	<b>Centers of Expertise: between accreditation and functioning - moderator Prof. Dr. Vlad Gorduza, Medical University Iași</b>
14.00 – 14.20	1. Functional relations in the Regional Medical Genetics Center – Prof. Dr. Marius Bembea, Dr. Claudia Jurcă
14.20 – 14.40	2. Research and diagnosis for rare diseases in Romania; Prof. Dr. Maria Puiu, Medical University Timișoara, president of National Council for Rare Diseases
14.40 – 15.00	3. Country report on rare diseases – Prof. Dr. Emilia Severin, Medical University București, CEGRD and National Council for Rare Diseases member
15.00 – 15.20	4. Specializes social services for rare disease patients – Dorica Dan, president of RONARD, National Council for Rare Diseases member
15.20 – 15.30	Q&A
15.30-16.00	Coffee break
16.00 – 17.30	<b>National and European networks for rare diseases, moderator Prof. Dr. Cristina Rusu, Medical University Iași, Orphanet România</b>
16.00 – 16.20	1. RO–NMCA ID Network – participation in ERN ITHACA, Dr. Adela Chiriță, Timișoara Center of Expertise;
16.20 – 16.40	2. Challenges and opportunities in participating in ERNs – Dr. Laura Damian, Cluj Emergency Hospital, ERN ReCONNET;
16.40 – 17.00	3. Center of Medical Genetics Iași – comparing plans and achievements, Prof. Dr. Cristina Rusu, ERN ITHACA
17.00 – 17.20	4. Center of Expertise in prevention, genetic diagnosis and management of congenital malformations – present and perspectives - Conf. Univ. Dr. Mihai Ioana, Craiova, ERN ITHACA
17.20 – 17.30	Q&A



<b>Day 2 – 17<sup>th</sup> of November 2017</b>	
<b>09.00 – 10.30</b>	<b>Integrated approach in rare disease care through CE and ERNs – moderator Prof. Dr. Vlad Gorduza, Medical University Iași</b>
<b>09.0 – 09.15</b>	1. NoRo Center– integrated approach, video
<b>09.15 – 09.30</b>	2. Center of expertise for rare hepatic paediatric diseases - Paediatric Clinic 2 Cluj-Napoca
<b>09.30 – 09.45</b>	3. The accreditation file of the Reference Center of Hereditary Angioedema: between agony and ecstasy, Dr. Noemi-Anna Bara, Dr. Attila Borka-Balas, Dr. Eniko Mihaly, Conf. Dr. Dumitru Moldovan.
<b>09.45 – 10.00</b>	4. Physical therapy in dystonia –Molnar Csilla, physical therapist
<b>10.00 – 10.15</b>	Q&A
<b>10.15 – 11.00</b>	<b>Coffee break</b>
<b>11.00 – 12.30</b>	<b>Specialised social services &amp; rare disabilities</b>
	<b>Debate</b> on criteria for classifying the disability level and access to specialized social services for patients with rare diseases <b>Moderator Mihai Tomescu – minister counselor of Minister of Labor</b>
<b>12.30 – 13.30</b>	<b>Coffee break</b>
<b>13.30 – 15.00</b>	<b>Monitoring the implementation of the NPRD in Romania – moderator Prof. Dr. Emilia Severin, Medical University București</b>
	<ul style="list-style-type: none"><li>- Access to information</li><li>- Treatments, therapies, rehabilitation</li><li>- Centers of Expertise</li><li>- European Reference Networks</li><li>- Patient Registries</li><li>- Specialised Social Services</li><li>- Training</li><li>- Research</li></ul>
<b>15.30 – 16.00</b>	<b>Conclusions</b>



**LIST OF PARTICIPANTS BY STAKEHOLDERS' CATEGORIES**

<b>1</b>	<b>RONARD and Patient Associations</b>	<b>40</b>
<b>2</b>	<b>Ministry of Health, Ministry of Social Security and public institutions</b>	<b>19</b>
<b>3</b>	<b>Centers of Expertise</b>	<b>14</b>
<b>4</b>	<b>Care providers</b>	<b>25</b>
<b>5</b>	<b>Laboratories and pharma</b>	<b>4</b>
<b>6</b>	<b>Others (press)</b>	<b>2</b>
	<b>Total</b>	<b>104</b>