Rare Diseases Day Policy Event at the United Nations

Resource Centres for Rare Diseases – NoRo Center
Romania

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EURORDIS.ORG
There is always a personal story ....
Our daughter’s care pathway

1. **Pre-Diagnosis:** Poor muscle tone; Distinct facial features Poor sucking reflex and difficult feeding; Almost no cry.

   1st Symptom: Hypotonic and difficulties in feeding; Poor responsiveness; Sitting up and walking — later Hypotonic and difficulties in feeding; unusually tired, respond poorly to stimulation, have a hard time waking up or have a weak cry. Sitting up and walking later.

   **Note:** My little girl seemed unusually tired, respond poorly to stimulation, have a hard time waking up or have a weak cry!

2. **18 years**

   **Note:** Care for life... **happiness as part of the treatment!**

3. **Diagnosis**

   - Unusual food-seeking behaviors, hypogonadism
   - Mild intellectual disability;
   - Delayed motor development
   - Speech problems
   - Small hands and feet
   - Thick saliva and dental problems associated
   - Overweight

   **Note:**... delayed motor skills an development ...

4. **First Treatment**

   - Delayed in motor development
   - Rehabilitation program, 3 weeks every 3 months

5. **Surgery (No)**

   - Scoliosis (gymnastic)
   - Physiotherapy

6. **Follow-up Care...**

   - Delayed in motor and intellectual development, behavior problems, Speech problems
   - Small hands and feet,
   - Overweight and sleep apnea;
   - Medication & rehabilitation
   - Support for education & school integration & inclusion into community; a proper job and qualification

**Ideally:**

- Improvement of muscle tone
- Special technics for feeding and stimulating exercises
- Medical treatment and early rehabilitation intervention

**Note:**

- Medication & rehabilitation
- Support for education & school integration & inclusion into community

**Ideally:**

- GH Treatment
- Sex hormone treatment
- Permanent rehabilitation program; Education and work Independent life skills

**Not always easy!!!**

- Improvement in general health and reduced weight

**Ideally:**

- Maintaining the weight, even reducing it
- High school, passing exams and training courses to find a proper job;
- Integration into a Center for occupational therapy and than a supported job;
- Friends, community, understanding and care at NoRo Center;
The difficulty lies in the impossibility of carrying a routine (...). The problem arises when one day you appear completely healthy, the next day you are sick, and two days later you appear completely normal again. Many people find it difficult to understand the disease and the process, and the absenteeism that entails” A.H. Romania

- Symptoms vary & can be invisible in many ways
- >90% of RDs have NO TREATMENT
- Support in coordination of care is a need!
- Heavy time burden for patients & carers

42% of patients & carers spend more than 2h/day on illness-related tasks

- Hygiene
- Administration of treatments
- Helping with house chores
- Helping patients to move
Why do we need integrated care?
Specialised Social Services for RD in EU mapped in 2014

94 Services
22 Countries

Information on activities, target population, contacts, etc.

- Therapeutic Recreation Services
- Respite Care Services
- Adapted Housing
- Resource Centres

Poster award at ECRD 2014 Berlin: “Integration of Rare Diseases into Social Services”

The map is available at www.eucerd.eu and www.eurordis.org
Romanian actions in the field of rare diseases

“RD are recognized as a priority for the health care system”

Patients and professionals as equal partners

NCRD since 2013

2008–2012 NPRDs

2008 Partnership MoH – RONARD

2014 NPRDs included in NH Strategy

2016 MoH Order for CoE

RD School for journalists 2012 - 2018
Resource Centre services include information and guidance services, training courses, provision of information concerning social benefits, and documentation and research services. They can also include daily support therapies, medical/psychological consultations and therapeutic recreation activities.

- in partnership with Centres of Expertise
- part of a Centre of Expertise themselves

Resource Centres could be defined as a ‘one-stop shop style’ service for rare disorders.
RPWA brief history:

- RPWA was established in 2003
- First international meeting in 2004 in Zalau
- Attending the 5th International PWS Conference in Christchurch, New Zealand
- Opened the Center for Information about Rare Genetic Diseases in 2005
- 1 Session about PWS at the Balkan Congress for Endocrinology Bucharest
- The 6th International PWS Conference in Cluj-Napoca, June 2007
- 1st National Conference about Rare Diseases, 1-3 November 2007
- 1st European RDD – 29 February 2008 – NPRD partnership with the MoH;
- Renewed partnership 2013; 2014 – National Strategy for Health;
- 1st National Program for Rare Diseases - 2008;
- May 2009 – 1st EE Conference on PWS, Timisoara; 2010 Zalau;
- Balkan Congress for RD, Cluj – June 2009
- Implementation of the NoRo Project: 2009 -2011
- 2010 – EUROPLAN1 and annual conferences or workshops;
- 2010 – The 2nd EE Conference on PWS
- 2011 – June 28, Official opening of the Center
- ExpertRARE Project – to organize a center of expertise
- INNOVCare (Oct. 2015- Sept 2018)

Projects implemented in partnership with Frambu:

- 2011 – Opening of NoRo Center (Norwegian-Romanian, partnership for future in rare diseases)
- AD AUGUSTA PER ANGUSTA
- NoRo- Frambu, partnership for the future
- TEAM – Together, everybody achieve more
- DocumentRARE – 2017

- RareResourceNet – to be continue!!!
NoRo – a Resource Centre for RDs

Since 2011- addressing health, psychological, social, educational, and daily life needs, abilities and wishes of people with a RD and their carers + case management in INNOVCare project;

Opened thanks to a EEA & Norway grant and our partnership with FRAMBU!
NoRo Interdisciplinary team: 24
Medical department – 2 doctors, 2 nurses;
Administrative staff: 4
Therapists - 16

Day care center: 60 beneficiaries
- Children with rare diseases and with autistic spectrum disorders
- From Zalau and county of Salaj
- Individual and group therapy
- Summer camps and therapeutic weekends for families;

RD Groups / 5 days/ month
- 12-14 Patients with the same RD
- From all over the country
- Therapies, social, medical and psychological support, support groups, registry and videoconferences;

“Trial for flight” Group for independent life skills
12-14 young people with Intellectual disabilities and RD
- From city and county
- Individual therapies, social, medical and psychological support, occupational therapy and training for life skills, summer camps and therapeutic weekends;

Therapies provided by NoRo
- Ergo therapy
- Massage
- Hydrotherapy
- Kinetotherapy (MIRA)
- Speech therapy
- Behavior therapy
- Deep oscillation
- Electric Therapy
- Psychological counseling
- Genetic counseling

Community support network
2 weeks/ year for home visits
Information campaigns
### Integrated care provision (NoRo)

<table>
<thead>
<tr>
<th>Medical services in contract with NHI – CoE Accreditation since 2017</th>
<th>Part of RO-NMCA ID &amp; ERN ITHACA</th>
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<tbody>
<tr>
<td>Different specialties, 24 permanent staff: therapists, administrative staff, medical doctors: pediatric psychiatry and geneticist;</td>
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<tr>
<th>Individual therapies:</th>
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<tr>
<td>Psychological intervention, hydrotherapy, medical gymnastic (MIRA), speech therapy, behavior, sensorial, electric therapy and social counseling;</td>
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<th>Group therapy and interventions:</th>
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<td>Psychological counseling, occupational therapy, ergo therapy; therapeutic weekends, summer camps, etc.</td>
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<th>Case management!!!</th>
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<td>Part of the Eurordis network of Help Lines</td>
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<td>Diseases management, “Trial for flight”- life skills, trainings for professionals, including RD School for journalists;</td>
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<th>Trainings and online platform: <a href="http://www.edubolirare.ro">www.edubolirare.ro</a> &amp; 2 magazines (<a href="http://www.rjrd.ro">www.rjrd.ro</a>)</th>
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<td>NoRo electronic Patient registry</td>
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<th>Registry Information</th>
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<td>Website, Facebook, blog, magazines, Radio NoRo – the voice of patients;</td>
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Quality in our services

- Demonstration of a multi-disciplinary approach
- Capacity to participate in data collection – patient registry
- Organisation of collaborations to assure the continuity of care between childhood, adolescence and adulthood – Trial for flight
- Continuity of care between all stages of the disease: diagnostic, treatment, rehabilitations, HCP, social services, education and social economy
- Links and partnerships with other CoE at national, patient organisations, Resource centres for rare diseases at EU level
- Consideration of e-Health solutions: training platform, videoconferences, mapping services, elaboration of guidelines, factsheets and documents
- Sharing resources and best practice with other HCPs
- Creating networks at national level & defining patient pathways
Networks & Partnerships:

- Ministry of Labour and Social Justice
- Ministry of Health
- National Authority for People with Disabilities
- National Authority for the Protection of the Rights of the Child and Adoption
- National Agency for Medicines and Medical Devices
- National Health Insurance House
- National Council for Rare Diseases
- Romanian National Alliance for Rare Diseases
- Romanian Prader Willi Association
- Ro-NMCA-ID RoNetwork Multiple Congenital Abnormalities with ID
- EURORDIS RARE DISEASES EUROPE
- RARE DISEASES INTERNATIONAL
- A Rare Resource NET
- A Rare Resource for Rare Cancer Association
- ÄGRENSKA
- FRAMBU
- Romanian Resource NET
- Centers of Expertise
- Public and private social services
- Case Managers
- Responsibility Groups
- Community Nurses
- ASHOKA
- ERN ITHACA
Rare Disease Day 2019 is an opportunity to be part of a **global call for action** on policy makers, healthcare professionals, and care services to better coordinate all aspects of care for people living with a rare disease!

Thank You