EUCERD Joint Action: Working for Rare Diseases
Progress Update

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Frambu, Norway 9th October 2014

Introduction

- Joint Action = Co-funded activity between EC and Member States
- March 2012-August 2015
- Action supporting the work of EUCERD, now CEGRD
- We will:
  - Improve the visibility and recognition
  - Increase access to higher quality RD healthcare
  - Facilitate the exchange of policies and practices.
- Ultimate outcome should be realisation of the Council Recommendation

Associated Partners

- Newcastle University (Kate Bushby)
- INSERM Paris (Segolene Ayme)
- ISS Rome (Domenica Taruscio)
- EURORDIS (Yann Le Cam/Dorica Dan)
- CIBER (Francesc Palau)
- Finnish Ministry of Social Affairs and Health (Helena Kaariainen)
- Goethe University Hospital Frankfurt (Thomas Wagner)
- Portuguese Ministry of Health (Gloria Isidro)

- All MS are collaborators via EUCERD/CEGRD

Scope

- Support for NP/NS for Rare Diseases
- Specialised Social Services
- Coding of RD and harmonisation of ontologies
- Cross-border genetic testing for RD/ application of NGS
- RD Registration and data sharing
- ERNs and 'networking the networks'
- Communications and Dissemination
- Real-life operations of CEs

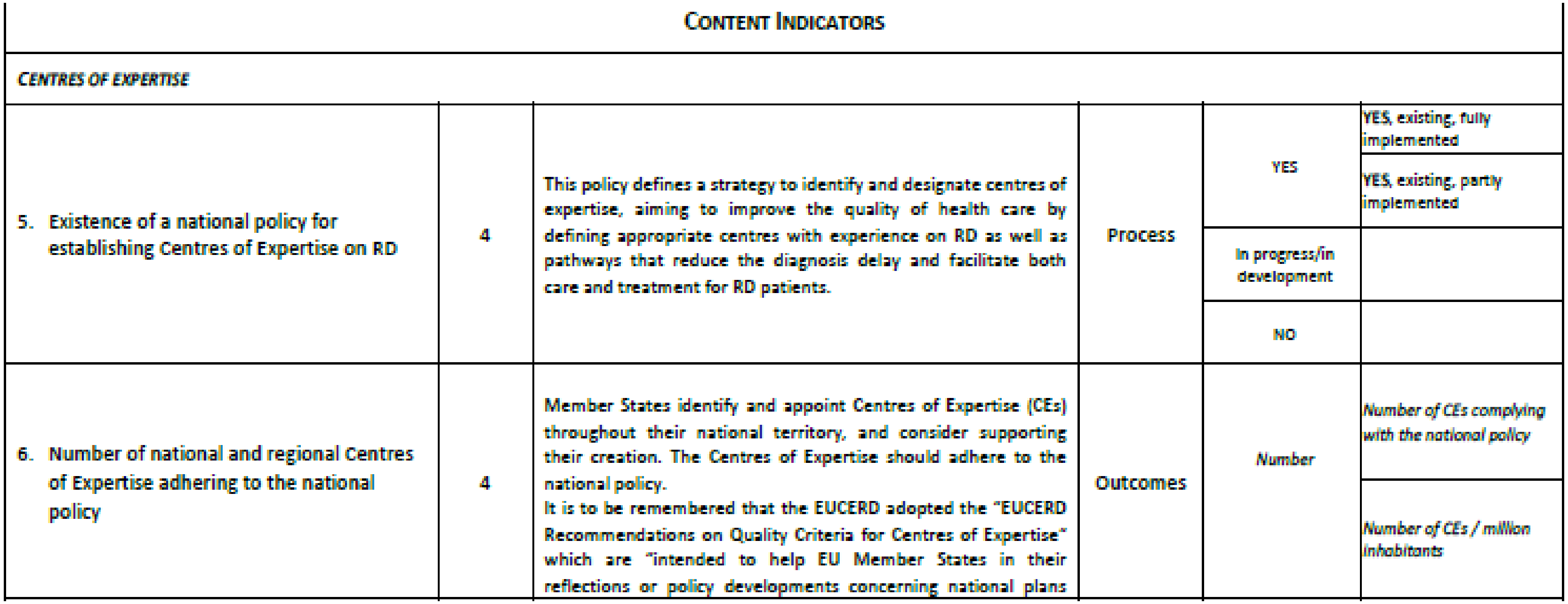
WP2

WP2 – State of the Art Report. Published in 2012, 2013, 2014

National Plans (WP4)

- **20 out of 24 EJA / “EUROPLAN” NCs have been organised**
- **Multi-stakeholder** events attracting over 100 participants
  - To gather all relevant parties to reinforce the dialogue amongst them;
  - To discuss and define concrete proposals for issues raised in Council Recommendation;
  - To discuss together content of the NP/NS on RD and its implementation
- Each conference shares the same format and content guidelines whilst SUCCESSFULLY TAILORED TO NATIONAL SPECIFIC NEEDS
- *EUCERD Recommendations on Core Indicators for RD NPs*

Example Indicator



CONTENT INDICATORS					
CENTRES OF EXPERTISE					
5. Existence of a national policy for establishing Centres of Expertise on RD	4	This policy defines a strategy to identify and designate centres of expertise, aiming to improve the quality of health care by defining appropriate centres with experience on RD as well as pathways that reduce the diagnosis delay and facilitate both care and treatment for RD patients.	Process	YES	YES, existing, fully implemented
					YES, existing, partly implemented
				In progress/in development	
				NO	
6. Number of national and regional Centres of Expertise adhering to the national policy	4	Member States identify and appoint Centres of Expertise (CEs) throughout their national territory, and consider supporting their creation. The Centres of Expertise should adhere to the national policy. It is to be remembered that the EUCERD adopted the "EUCERD Recommendations on Quality Criteria for Centres of Expertise" which are "intended to help EU Member States in their reflections or policy developments concerning national plans	Outcomes	*Number*	*Number of CEs complying with the national policy*
					Number of CEs / million inhabitants

WP4 – Conclusions to date from the National Conferences

Final Conferences Reports at: www.eurordis.org/content/reports-europlan-national-conferences-2012-2015

➔ Recurring needs and problems

- not all areas are covered and/or
- most actions are without funding allocations and/or
- many policy measures difficult to implement and/or
- some disease areas left uncovered

➔ Possible areas for more coordinated action, exchanges amongst MS and policy guidance via the Expert Group

➔ 16 adopted NP/NS – shift to implementation & evaluation

➔ Social Services (preparatory document)

Coding & Classification (WP5)

- Inadequacy of current coding systems
- Contribution to the ICD11 beta draft is complete
- Cross-referencing with other ontologies e.g. SNOMED-CT, UMLS and HPO
- Promotion of the OrphaCode in healthcare systems
- Draft CEGRD Recommendations

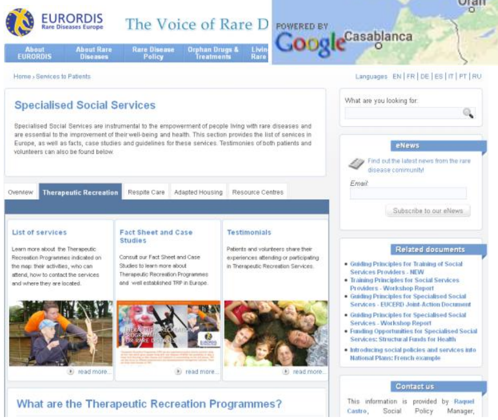
WP6 (Specialised Social Services)

Map of Specialised Social Services:

- Constantly updated, now shows 81 services from 21 countries

List of Services, Case Studies and Testimonials on:

- Therapeutic Recreation Programmes (TRP)
- Respite Care Services (RCS)
- Adapted Housing Services(AH)
- Resource Centres (RC)



Specialised Social Services

Specialised Social Services are instrumental to the empowerment of people living with rare diseases and are essential to the improvement of their well-being and health. This section provides the list of services in Europe, as well as facts, case studies and guidelines for these services. Testimonies of both patients and volunteers can also be found below.

List of services

Learn more about the Therapeutic Recreation Programmes indicated on the map: their activities, who can attend, how to contact the services and where they are located.

Fact Sheet and Case Studies

Consult our Fact Sheet and Case Studies to learn more about Therapeutic Recreation Programmes and well established TRP in Europe.

Testimonials

Patients and volunteers share their experiences attending or participating in Therapeutic Recreation Services.

What are the Therapeutic Recreation Programmes?

Find out the latest news from the rare disease community!

- Guiding Principles for Training of Social Services Providers - NEW
- Training Principles for Social Services Providers - Workshop Report
- Guiding Principles for Specialised Social Services - EUCERD Joint Action Document
- Guiding Principles for Specialised Social Services - Workshop Report
- Funding Opportunities for Specialised Social Services: Structural Funds for Health
- Introducing social policies and services into National Plans: French example

This information is provided by Raquel Castro, Social Policy Manager,

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WP6 Cont.

Advocacy Documents have been generated:

- **To raise awareness of the social needs of people living with RD**
- **To support the integration of RD into mainstream services**

THERAPEUTIC RECREATION PROGRAMMES FOR RARE DISEASES

ADAPTED HOUSING SERVICES FOR RARE DISEASES

RESOURCE CENTRES FOR RARE DISEASES

RESPITE CARE SERVICES FOR RARE DISEASES

EUCERD JOINT ACTION GUIDING PRINCIPLES ON TRAINING FOR SOCIAL SERVICES PROVIDERS TO THE EUROPEAN COMMISSION, MEMBER STATES AND ALL STAKEHOLDERS

April 2014

- [Rare Diseases: Addressing the Need for Specialised Social Services and Integration into Social Policies](#)'(Nov. 2012)
- ['Guiding Principles for Specialised Social Services'](#) (2013)
- ['Guiding Principles on Training for Social Services Providers'](#) (2014)

Policy Fact Sheets

Therapeutic Recreation Programmes

Respite Care Services

Adapted Housing Services

Resource Centres

WP7– Centres of Expertise and Quality of Care (CIBER)

- Designed framework integrating dimensions of quality of care (QoC), EUCERD recommendations and identification of good practices.

- Completed in-depth ethnographic research informing collection of data and approach.
- Completed research with 16 CEs
- Now compiling inventory of good practices for improving QoC for RD patients

EUCERD Joint Action

Select Conclusions of the workshop

- Good practices highlighted incl. regular clinical meetings; Laboratory testing; continuity of care; informal networking
- Many interviewees found the EUCERD Recommendations important as a guide to the creation of CEs
- **CEs directors and staff value the capacity to provide a holistic experience of patient-centred care**
- CEs increasingly play the role of 'information hub'

- Challenges:
  - Many CEs are in 'survival mode' due to lack of resources;
  - Coordination of care is a key challenge– specialist nurses & social service professionals need to be more closely involved;
  - Continuity of care/transition from paediatric to adult services

Integration (WP8)

Registries

- *EUCERD Recommendations on RD Registration and Data Collection*

- T. Wagner leads this work from Germany
- Minimum Datasets (purpose-specific)
- JRC collaborations and pilot

- **Minimal Data Set (MDS):**
  - smallest possible set of items needed to answer a single question (e.g. pseudonym plus diagnosis code)
  - *proven to be rather theoretical, not very practical;* instead:
- **Common Data Set (CDS):**
  - core set of items common to several registries
  - prerequisite for registries to be included in the European platform
- **Specific Data Sets (SDS):**

ERNs for Rare Diseases

- European Reference Networks for RD
- 1st Calls expected 2015
- Recommendations of EUCERD - 2013
- Delegated and Implementing Acts -2014
- Workshop Rome 28-29th October
  - 1st level, attempting to support National MS authorities

Genetic Testing for RD

- Helena Kääriäinen exploring the volume of cross-border genetic testing for RD
  - Reasons to sell/buy tests on cross-border basis
  - The magnitude of the practice
  - Problems faced by the laboratories
  - Problem faced by the clinicians
  - Is there equality in access to genetic testing in EU?

Genetic Testing Results

- Survey performed from Jan-March 2014, response gained from :
  a) 170 molecular genetic testing laboratories; and
  b) 105 genetic counselling clinics

- In some countries cross border testing is not at all reimbursed (Romania) or very rarely (Poland) or only for children (Croatia) or the quality may be poor (Ireland)
- Some countries do practically all their own testing (UK, France etc)
- Some countries easily allow cross-border testing (Finland)

WP8 External Collaboration

PARENT
cross-border
PAtient REgistries iNITiative

European Partnership
Action Against Cancer

eunethta
EUROPEAN NETWORK FOR HEALTH TECHNOLOGY ASSESSMENT

Rare
Best
Practices

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