Opportunities and challenges: ERN Eastern European HCP perspective

Laura Damian, MD PhD
Consultant rheumatologist
Emergency Clinical County Hospital Cluj
-ERN-ReCONNET

ERN- autoimmune / musculo-skeletal diseases
ReCONNET

dir. Marta Mosca

musculo-skeletal diseases cluster

- **rare**

- **systemic sclerosis** M34/ORPHA90921
- **mixed connective tissue disease** 35.1/ORPHA809
- **polymyositis** M33/ORPHA732
- **dermatomyositis** M33/ORPHA221
- **antisynthesize sdr** M33/ORPHA81
- **undifferentiated CTD** M35.9/ORPHA90002
- **antifosfolipid sdr** D68.6/ORPHA80
- **IgG4 sdr** M35.8/ORPHA284264
- **polychondritis** M94.1/ORPHA536

- **complexe**

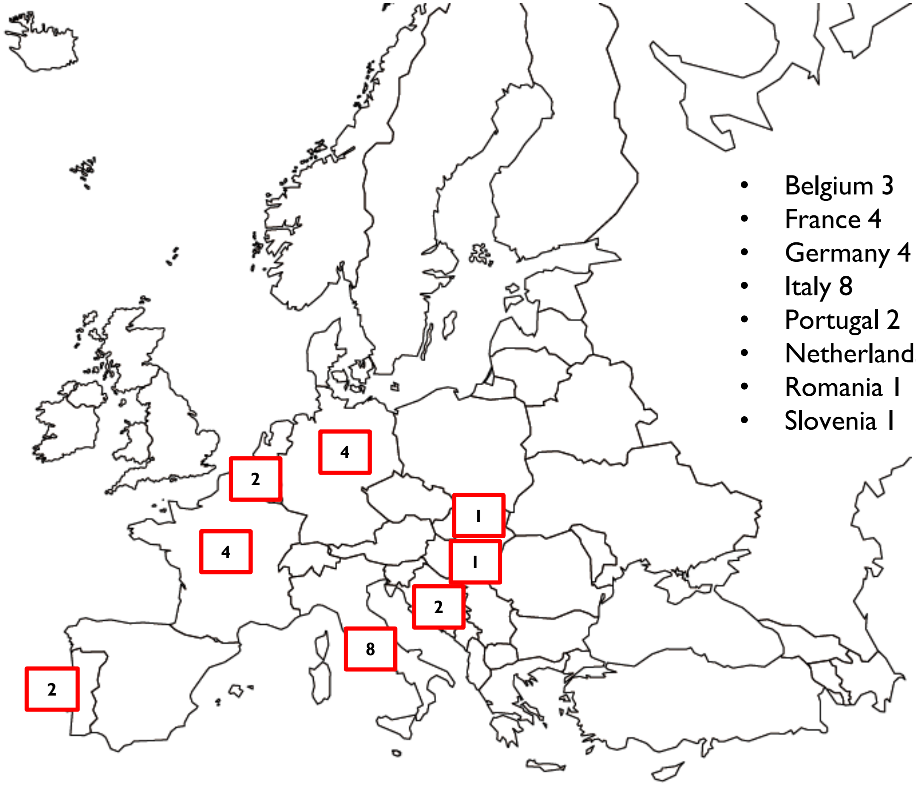
- **systemic lupus eritematos** M32/ORPHA536
- **Sjogren’s sdr** M35.0/ORPHA289390

- **hereditary**

- **Ehlers-Danlos sdr** Q79.6/ ORPHA98249

ERN ReCONNET (26 HCPs)

European
Reference
Networks



- Belgium 3
- France 4
- Germany 4
- Italy 8
- Portugal 2
- Netherlands 3
- Romania 1
- Slovenia 1

Ghent University Hospital – Vanessa Smith
Ghent University Hospital – Francisca Malfait
Clinique Universitaires Saint-Luc – Frederic Houssiau
University Hospital of Lille – Eric Hachulla
University Hospital of Strasbourg – Thierry Martin
Assistance Publique Hopitaux de Paris – Zahir Amoura
Cochin Hospital – Luc Mouthon
Charité - University Medicine Berlin – Gerd Burmester
University Medical Centre Dusseldorf – Matthias Schneider
Kerckoff Klinik gGmbH – Ulf Mueller Ladner
University of Cologne – Thomas Krieg
Azienda Ospedaliero Universitaria Careggi Firenze – Marco Matucci Cerinic
Azienda Ospedaliero Universitaria Pisana - Marta Mosca
Spedali civili di Brescia – Angela Tincani
Policlinico San Matteo di Pavia – Carlomaurizio Montecucco
Azienda Ospedaliera Universitaria San Martino Genova – Maurizio Cutolo
Azienda Ospedaliera di Padova - Andrea Doria
San Camillo Forlanini – Marco Castori
IRCCS Ca' Granda Ospedale Maggiore Policlinico Milano – Lorenzo Beretta
Erasmus MC Rotterdam – PM Van Hagen
University Medical Center Utrecht – Jaap van Laar
Leids Universitair medisch Centrum – Jeska de Vries-Bouwstra
Centro Hospitalar Lisboa Norte – Joao Fonseca
Hospital Curry Cabral, Centro Hospitalar de Lisboa Central - Francisca Fontes
Spitalul Clinic Judetean de Urgenta Cluj – Simona Rednic
University medical Centre Ljubljana – Tadej Avcin

"ERN-ReCONNET was funded by the European Union's Health Programme (2014-2020)."

European reference networks (ERNs) in rare diseases

24 ERNs

ROMANIA

Spit. Clinic Psihiatric “Al.Obregia” **EpiCARE, Endo-ERN**

Spit. Colentina **ERN Skin**

Spit. Cl. Jud. de Urgenta Cluj **ReCONNET**

Spit. de Urg. B. Cardiovasc. “CC Iliescu” **GUARD-HEART**

IOCN Cluj- **Endo-ERN**

Reteaua de anomalii congenitale multiple- **ITHACA**

ec.europa.eu/health/ern

ERN: Mission and Vision Statement

- To deliver high quality and homogeneous care across EU borders
- To facilitate information sharing and circulation as well as patients and health care professionals movement across borders.
- To optimize available resources and deliver cost effective care.
- To empower patients in the management of their disease.
- To increase knowledge on rare and complex conditions.
- To develop recommendations and quality guidelines
- To facilitate epidemiological, clinical, translational research.

“ERN-ReCONNET was funded by the European Union’s Health Programme (2014-2020).”

Opportunities

- **Paradigm change**- putting patient in the central place in healthcare system
  - Assessment of guidelines through AGREE and ADAPTE
  - eHealth, eLearning; **CPMS**
- Steps towards **uniformisation of practice** in RD across UE
- **Actively taking part** to the activities of the scientific RD communities including the ERN-generated studies
  - Increasing participation and interest for research also to the “grass level”, of the junior doctors not belonging to university
- **Proper collection** of epidemiologic/clinic/genetic data from the country
- **Registries**
- **Legitimity** offered to the Centre/Hospital in dialogue with all stakeholders, including authorities and law makers
- **Changing/generating legisl**ation– to a better care

Opportunities

- Increased local and national inter-institutional **collaboration**
- Intra-institutional
  - officializing of the informal teams dedicated on one disease cluster
  - common examination **protocols in the hospitals**

From the Eastern countries to the Western counter-part:

- large number of patients to be included in studies for RD
- clinical expertise -often increased in low-resource settings
- ways to optimize resources use/ to decrease costs
- patients organizations' partnership with HCPs
- transition from pediatric to adulthood services in Expertise centres
- enthusiasm and genuine interest for collaboration

Challenges

- **Logistic**
  - short consultation time
  - paperwork load
  - lack of technical staff/registrators/secretary
- **Difficulties in reporting rare diseases**
  - the number of patients reported into services
  - No ORPHA codes used currently
  - only common diagnoses are being covered by the health insurance
  - reporting accuracy for RD?

Solutions

- Applied for extra-consultation time (=new services: clinical measurements and filling in the assessment forms) included in the outpatient assessment
  - *not approved yet*
- Setting a new outpatient clinic (in order to document all presentations of patients with rare diseases)
- Applied for inclusion of ORPHA codes in the informatics system
  - *Not ready yet*
- Applied for an application to retrieve all the patients on keyword search in the informatic system
  - *Not ready yet*

Challenges

- **Referral system**
  - the reform of the medical system has “democratized” the referral, the patients with RD can be seen by any specialist and do not have to be monitored in a tertiary center by law

- **Registries?**

- **Biobanking**

Solutions

- Inclusion of the specialists in the referral areas into Study groups for the Centres’ projects (on certain diseases)
- Increasing the Expertise Center’s capabilities in order to bring up added value for the patient and for the referring physician
- telemedicine

- Registries included into National Plan for RD; lack of current discussion for consensus on how to gather data and Core data set
- Turning challenge into an opportunity
  - following the data protection regulations

Challenges

- **Funding of the Expertise centre**
  - Lack of technical staff/registrators/secretary
  - No extra money for the diagnosis and therapy of difficult cases from the Health insurance

- National grants
  - scarce, mainly for Universities
  - the Hospital developed a research structure
- European grants
  - European research calls not frequent
- National Plan for Rare Diseases
  - dedicated programme for the ERN-participating centres?

Challenges

No common legislation for Expertise centers across Member Stats

- Genetic **tests** in adults are not being covered
- **drug prescription**
- (including off-label medication) for RD where there are no national programmes
- **Not uniform prescription/ covering rules in the MS**

Solutions

- Possibility of the Hospital/Expertise centre to cover the medication/genetic tests (difficult in the context of chronic shortage and legislative restrictions)
- Rare Diseases Commission of the Hospital
- Off-label Medication Law project (*did not pass yet*)

- Unlikely to change early in the absence of a common EU policy
- Possibly increasing the number of Expertise centres included in ERNs with the new wave – and mostly National networks!- could lead to law changes
- **Law changes and harmonization necessary**

Lupus Day 2019