Rare Disease European Reference Networks

Concept Paper, EURORDIS Membership Meeting 2015 Madrid

How did we start the process together? What important steps have been achieved?

In 2004, EURORDIS and its members started considering solutions for pooling together resources and expertise across Europe to ensure proper diagnosis and quality care for rare disease patients. This reflection process culminated in a major milestone at the 2008 EURORDIS Membership Meeting: the adoption of the Declaration of Common Principles on Centres of Expertise and European Reference Networks. This document recognised that it was not possible to have a Centre of Expertise (CE) for every single rare disease in every Member State and that it was necessary to create European Reference Networks (ERNs) of CEs and healthcare providers. From the patients’ perspective, the focus was placed on the need for a multi-disciplinary approach and the combination of both medical and social care.

EURORDIS advocacy work led to the adoption of the Cross-Border Healthcare Directive in 2011, which provides the legal basis to sustain the establishment of ERNs in Europe. To support the implementation of the Directive, a Committee of Member States was set up and both Delegated/ Implementing Decisions were adopted by the Commission in 2014. The call for applications for networks of CEs wanting to become ERNs will be published in December 2015.

Patient-centred European Reference Networks

ERNs create a clear governance structure across the EU for knowledge sharing and care coordination for complex conditions requiring highly specialised healthcare, such as rare diseases. They are networks of CEs, healthcare providers and laboratories, organised across borders. Through these networks, doctors will have the most recent and expert knowledge possible and will be able to make better-informed decisions on how to adapt treatment and care pathways, thereby minimising the need for patients to travel across borders to access expertise.

EURORDIS advocates for the establishment of ERNs that aim at improving patient health outcomes in a measurable way through accurate diagnosis, therapies and medical/social care. This will help to structure healthcare pathways at a national level and improve the quality criteria.

How can we better organise the grouping of rare diseases for ERNs?

In order for ERNs to effectively serve the over 6000 existing rare diseases, they need to be organised according to disease areas or ‘groupings’, rather than by each individual disease. This grouping of diseases neither prevents a patient from being able to go to a disease-specific CE, nor from benefiting from the expertise of several ERNs when needed.

EURORDIS aims to ensure that every rare disease patient, including the undiagnosed, will be covered by at least one ERN so that every patient will find a “home” within the European healthcare system.
**Some important facts on how ERNs function**

In order to ensure that every rare disease patient has an ERN to refer to in the long run, it is necessary to gather rare diseases into overarching ERNs based on the following principles:

- An overarching ERN for rare diseases should be understood as an **operational ERN**, covering a broad scope of activities and serving as research and knowledge hub. These ERNs will cover a wide range of rare diseases, regrouped around the concept of medical specialties and body systems, diagnostic and therapeutic areas. This will ensure that a relatively limited number of rare disease ERNs ultimately cover all RDs.
- Each of these overarching/operational ERN will be divided into sub-networks, organised around the concept of **clinical network** in order to build on existing informal networks, and expand these further in the future.

At EURORDIS we strongly feel that this grouping structure is only a starting point. The development of ERNs should remain flexible as they mature in response to: the accession of new Member States, the addition of more rare diseases organised in networks in a step-wise approach according to the maturation level of their networking activities, evolving designation of CEs at the national level and CEs flexibility to join/leave an ERN.

**The ultimate goal of ERNs is to meet the needs of both clinicians and patients.** The grouping structure should ensure that clinicians can work effectively, that all patient groups and diseases are fairly represented and that **all** rare disease patients can access expertise.

**EURORDIS aims to ensure that rare disease patient organisations are fully engaged in the governance of the overarching networks, as much as in their specific activities and evaluation.**

The EU Commission has heard our plea to ensure that all patients have access to the best possible diagnosis and care through ERNs. A grouping model was brought forward in the 2013 **EU Committee of Experts on Rare Diseases (EUCERD) Recommendations on Rare Disease ERNs**. EURORDIS represented the patient voice when contributing to the drafting of these Recommendations. It is now up to all of us to make sure that this structure best corresponds to the needs of rare disease patients. An addendum to these Recommendations, covering both the ERN grouping and also full engagement of patient representatives in ERN activities and governance, should be adopted at the Commission Expert Group on Rare Diseases meeting in June.

Although there is no single optimal grouping model, our reflections on how each ERN gathers a range of diseases together will be key in helping shape their scope and progressive development in the patient’s interest.

It is important that ERNs evolve as the needs of the rare disease community develop. The unity of our views will help to ensure that every patient organisation - irrespective of their scope and progressive development, size or existing influence on a European level - is fairly represented within the wider ERN system.

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In order to be fully engaged in the governance activities of the ERNs, patient groups will need to agree on representatives from specific patient group(s) to take the lead in each network on behalf of all patients and diseases represented in that network. The most effective way forward seems to be that the most mature/experienced patient groups will be taking the lead on behalf of the other patient groups within an overarching ERN.
Call for action: why should you contribute and what can you do?

The EUCAERD Recommendations stress the critical role of patient representatives and organisations as experts by experience and as producers of knowledge in all ERN activities. The evaluation of rare disease ERNs should also be multi-stakeholder and include patient representatives.

The EU Commission Delegated Decision on ERNs builds on these Recommendations and translates the principles of empowerment, participation and patient-centric care into legal criteria that all ERN applicants must fulfil.

Because of the limited knowledge, experience and expertise available in the field of rare diseases, rare disease patients play a fundamental role in contributing to ERN governance, activities and evaluation.

It may take time until the direct impact of ERNs for all patients is seen and until ERNs covering all diseases across all countries are set up, but EURORDIS Members need to take this opportunity to be part of the early stages of the implementation of ERNs.

The call for applications for networks of CE wanting to become ERNs will be published in December 2015. Therefore the moment for patient organisations to take action is NOW.

Organisations with common symptoms or medical needs will have to reach a consensus on which ERN they want to join and how to integrate into larger groupings.

Today

We would like to take the opportunity in today’s breakout sessions to discuss the proposed groupings of rare diseases.
Progress in Establishment of European Reference Networks

Since 2006

April 2006
EURODIS
EURODIS Membership Meeting – ‘Centres of Expertise for Rare Diseases: How can we make it happen?’

May 2008
EURODIS
EURODIS members adopt a Declaration of Common Principles on Centres of Expertise and European Reference Networks

2006-2009
EURODIS
EU Commission pilots 10 ERNs

2007-2010
EURODIS
RAPSODY project
Collects patient feedback on Centres of Expertise through qualitative workshops and quantitative EURODISCARE survey

2011
EURODIS
POLKA project further explores added-value of ERNs and patient representative preferences in involvement based on experience of pilots

June 2009
EURODIS
European Council adopted the Council Recommendation on an Action in the Field of Rare Diseases.

November 2008
EU Commission adopts the Communication Rare Diseases: Europe’s Challenges along with a proposal for a European Council Recommendation

2014
EU Commission publishes a delegated decision listing the criteria/conditions that healthcare providers and ERNs should fulfil and an implementing decision containing criteria for establishing/evaluating ERNs.

May 2015
MADRID
EURODIS Membership Meeting: session dedicated to deliberating best ERN grouping model to ensure a “home” for all RD patients within the European healthcare system

End of 2015
First call for applications from networks of CES wanting to become ERNs

Today

2011-2013
Directive 2011/24/EU clarifies rules on patients’ rights in cross-border healthcare. EU countries have until 25 October 2013 to pass their own laws implementing the Directive which requires the Commission to support the development of ERNs (article 12).

2015-2016
EURODIS
EURODIS leading EL Commission Tender to develop a technical proposal for:
- A manual including operational criteria for the assessment of all applications
- A toolbox providing guidelines for network applicants and independent assessment bodies to facilitate the application process

Ongoing through 2025
ERNS will be established and developed