

## European Reference Network (ERN): A Joint Project Focussing on Rare and Undiagnosed Skin Disorders



ANDERS VAHLQUIST, MD

Department of Dermatology, Uppsala University Hospital, SE-751 85 Uppsala, Sweden.

E-mail: anders.vahlquist@medsci.uu.se

The European Commission decided in 2014 to launch a health-oriented project aiming at improving the collaboration between EU states (plus Switzerland and Norway) for the benefit of patients suffering from rare diseases. In the case of Dermatology, rare skin diseases have been chosen for the ERN project. The background and some of the plans for this project are presented below.

Many patients suffering from rare diseases lack an exact diagnosis, are inadequately informed and receive suboptimal therapy. Most likely they would benefit from the expert services not generally available in the European countries. This body of expertise should include both high-level clinical management and research & development. The scope and context of the ERN project is shown in Fig. 1.

For patient management, it could be the mapping in each country of the services provided by expert centres such as:

- platform for a multidisciplinary management,
- medical and paramedical expertise
- guidelines
- medico-social organization
- partnership with patient groups
- e-health
- training and teaching
- therapeutic education programme
- coordination secretariat

- formal organization of a national network and charter defining the responsibilities of each network member

For research, it could focus on epidemiology and clinical research with the mapping in each country of the services/resources such as:

- registries at the local and at the national level
- mapping of laboratories for diagnosis and available tests
- quality insurance mechanisms
- guidelines for genetic counselling
- charter and guidelines for sharing information between clinical centres and laboratories

When the ERN project was discussed from a dermatological perspective it was decided to focus in a first step on rare genetic skin disorders, because there already exists embryos of network in this field: E.g. the Genodermatoses Network, Geneskin and EB Clinet.

Already in 2008, the Genodermatoses Network was co-funded by the European Commission as a reference network pilot project. The Genodermatoses Network is now co-funded by the Fondation René Touraine and the European Academy of Dermatology and Venereology.

The Genodermatoses Network is in charge of structuring the future ERN proposal for the first groups of rare skin disorders that we have identified with our partners. We focus on 5 groups of diseases (Table I). The inclusion of other groups is under discussion.

The aim is to submit a proposal early next year (2016) in which existing facilities within a network are described together with a business plan on how the collaborations should be developed in the future. Key features of the network are shown in Fig. 2.

For the Ichthyosis and PPK research group several persons have now been appointed as team members. One of our interim re-

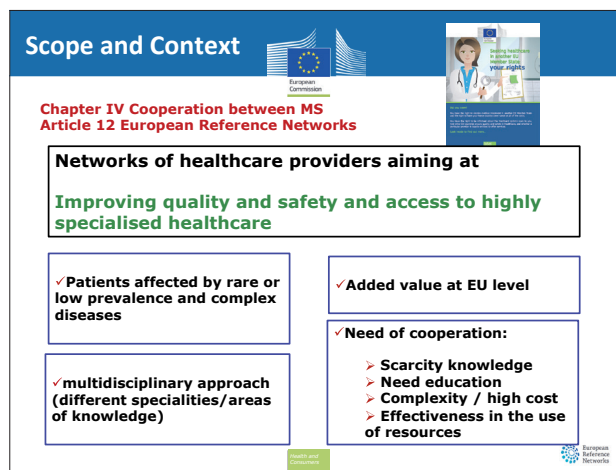


Fig. 1. The scope and context of European Reference Network (ERN).

Table I. Coordinators and co-leaders of the various task forces within the ERN (Rare and Undiagnosed Skin Disorders) project

Coordinators <i>Christine Bodemer, Maya El-Hachem, Johann Bauer</i>		
Disease	High-level patient management	Research in link with other EU initiatives
Epidermolysis Bullosa	M. Jonkman, J. Mellerio	L. Bruckner-Tuderman, I. Leigh, G. Zambruno
Ichthyosis and Palmoplantar Keratoderma (PPK)	A. Hernandez-Martin, J. Mazereeuw-Hautier	A. Vahlquist, H. Traupe
Ectodermal Dysplasia including Incontinentia Pigmenti and Skin Fragility Disorders	S. Hadj-Rabia, M.V. Ursini	S. Hadj-Rabia, M.V. Ursini
Connective Tissue Diseases	A. Chiriac, S. Karpati	B. Callewaert
Cutaneous Mosaic Disorder – Naevi & Naevoid Skin Disorders	A. Diociauti, A. Torello	L. Boon, V. Kinsler

ports is shown as e-supplement (<http://www.medicaljournals.se/forum/ERNInterimReport.pdf>)

Meanwhile, Geneskin has developed an on-line database dedicated to rare genetic skin diseases. We are also working on creating specific guidelines adopted to ERN, the working groups for ichthyosis and PPK are shown in Table II.

It is hoped that an ERN for genodermatoses will eventually be approved by the Commission and that several clinical and research centres also in the Nordic countries will qualify as hubs in this network and as referral centres for European patients who are in need of expertise help not available elsewhere in their home region.



Fig. 2. Requirements for European Research Network (ERN) competence centres.

Table II. Guidelines for Ichthyosis and palmoplantar keratoderma prepared by the subgroups (contact persons listed). Coordinator: Juliette Mazereeuw-Hautier

**Topical therapies**

Angela Hernandez-Martin, Spain, [dermicus@gmail.com](mailto:dermicus@gmail.com)

**Systemic therapy (Acitretin)**

Anders Vahlquist, Sweden, [anders.vahlquist@medsci.uu.se](mailto:anders.vahlquist@medsci.uu.se)

**Ichthyosis particularities**

Heiko Traupe, Germany, [traupeh@ukmuenster.de](mailto:traupeh@ukmuenster.de)

**Ichthyosis complications**

Vinzenz Oji, Germany, [ojiv@uni-muenster.de](mailto:ojiv@uni-muenster.de)

**Psychosocial aspects**

Anette Bygum, Denmark, [Anette.Bygum@rsyd.dk](mailto:Anette.Bygum@rsyd.dk)

**Associations of patients**

Gesje Wehr, Germany, [e-n-i@gmx.net](mailto:e-n-i@gmx.net), [geske.wehr@gmx.net](mailto:geske.wehr@gmx.net)