

# THE GENODERMATOSES NETWORK

## The European and International Network on Rare Skin Diseases for professionals and patients

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### THE GENODERMATOSES NETWORK IN BRIEF

- The Genodermatoses Network is the European Reference Network for Rare Diseases pilot project sharing experience and knowledge with Mediterranean and Middle-Eastern countries
- Genodermatoses represent more than 400 rare skin diseases
- Since 2003, the Genodermatoses Network aims to improve health care and social support for patients with rare and severe skin diseases by promoting a patient based approach and organizing healthcare networks
- More than 75 medical and research teams as well patient groups are partners of the Genodermatoses Network
- Gathering over 30 countries, the Genodermatoses network has become the only rare diseases pilot network encompassing both European and North-South cooperation



### WHO WE ARE

- The Genodermatoses Network is the Fondation René Touraine's network on rare genetic skin diseases initiated in 2003
- The Genodermatoses Network Scientific Committee supervises and develops the activities of the Genodermatoses Network (Figure 1)
- A project manager is responsible for delivering the actions of the Genodermatoses Network
- The 2014-2017 Genodermatoses Scientific Committee will be enlarged to allow greater country and expert representativeness (including patient groups).**



Figure 1: The Genodermatoses Network Scientific Committee (2011-2014) - J. Bauer, President (Austria), C. Bodemer, President-elect (France), L. Bruckner-Tuderman (Germany), A. Hovnanian (France), M. Jonckman (The Netherlands), S. Karpati (Hungary), I. Leigh (United Kingdom), J. Mellerio (United Kingdom), H. Traupe (Germany), G. Zambruno (Italy)

### GENODERMATOSES REPRESENT MORE THAN 400 RARE SKIN DISEASES

- More than 400 monogenetic genodermatoses have been defined based on molecular research.
- Very rare in Europe (mean prevalence of 1:250.000), they are more frequent in the Mediterranean and Middle-Eastern countries. In a 2011 survey conducted by the Genodermatoses Network in Algeria, Egypt, Iran, Libya, Morocco, Saudi Arabia, Syria, Turkey and Yemen the mean prevalence for 6 groups of severe genodermatoses was estimated around 1:20.000.
- Handicap, short life expectancy and social exclusion make patients and families vulnerable.
- At present, there is no cure for rare skin diseases. Treatment is symptomatic.
- Genodermatoses Network partners have targeted 6 groups of diseases: ectodermal dysplasia and incontinentia pigmenti, epidermolysis bullosa, ichthyosis, neurofibromatosis, palmoplantar keratoderma and xeroderma pigmentosum.



Figure 2: Junctional Epidermolysis Bullosa



Figure 3: Palmoplantar Keratoderma



Figure 4: Netherton Syndrome (Ichthyosis)

### TOWARDS A EUROPEAN REFERENCE NETWORK FOR RARE SKIN DISEASES SHARING EXPERIENCE AND KNOWLEDGE WITH MEDITERRANEAN AND MIDDLE-EASTERN COUNTRIES

#### Assess patient and health care professional needs: international surveys

- 2011 survey to assess the needs of patients, the health care strategies developed by medical and paramedical teams, the use of drugs and medical devices.
- 2014 survey on Medical and Paramedical consultations for patients with rare skin diseases

#### Inform patients and health care providers (Figure 5)

- An interactive directory for patients and professionals making possible to identify in each country: Genodermatoses consultations, Research teams, Patient organisations

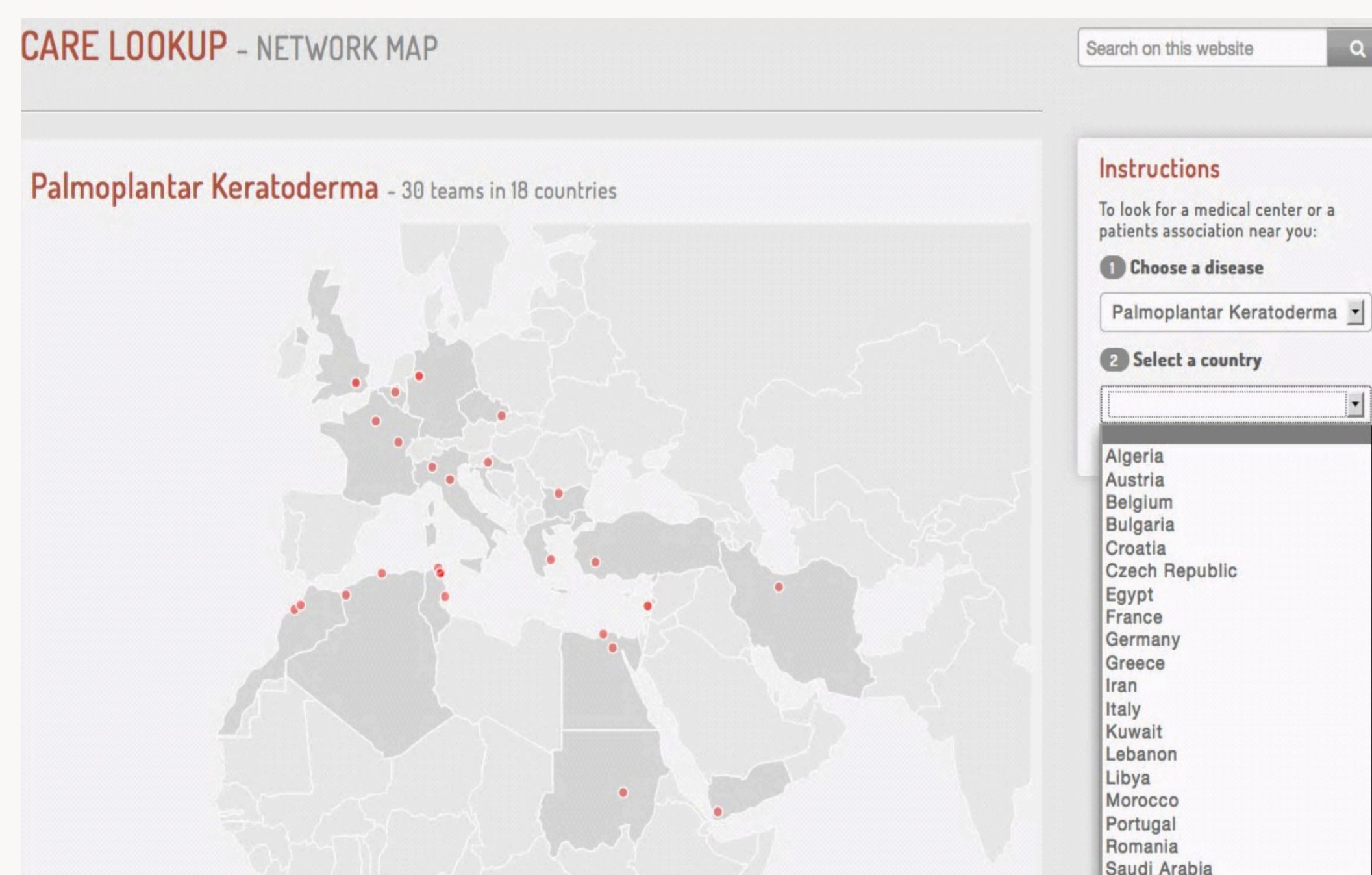


Figure 5: The Genodermatoses Network Directory: Choose a Disease - Select a country

#### Organise a European Reference Network on Rare Skin Diseases

- The First European workshop on ERN recommendations and criteria in Rare Skin Diseases** was held on October 5, 2013. The Genodermatoses Network partners have set the following objectives:

- ✓ have knowledge and expertise to diagnose, follow up and manage patients with evidence of good outcomes
- ✓ follow a multi-disciplinary approach
- ✓ offer a high level of expertise and have the capacity to produce good practice guidelines and to implement outcome measures and quality control
- ✓ make a contribution to research
- ✓ organise teaching and training activities
- ✓ collaborate closely with other centres of expertise and networks at national and international level



Figure 7: The 2011 Genodermatoses Network training session in Paris, France

#### Get together: international expert meetings (Figure 6)

- 8 international meetings** to discuss recommendations for best practice, train young teams, foster the development of new teams and national networks, develop new international cooperation projects including e-health, meet companies interested by rare skin diseases



Figure 6: The Genodermatoses Network International Expert Meetings

#### Train specialists, nurses, paramedics and general practitioners (Figure 7)

- 30 training sessions** including 15 practical training sessions for specialists and nurses in the dermatology departments of centres of expertise

### NEXT GENODERMATOSES NETWORK EVENTS!

- May-June 2014:** International Survey on Medical and Paramedical consultations for patients with rare skin diseases
- 10 October 2014, Amsterdam, Netherlands:** Second European Workshop on ERN recommendations and criteria in rare skin disease
- 30-31 October 2014, Paris, France:** Training session on rare skin diseases cross-cutting issues: Highlight on cutis laxa, ectodermal dysplasia, incontinentia pigmenti and specific common problems (pain, pruritus, nutrition)
- 6-7 November 2014, Beirut, Lebanon:** Training session towards a network of excellence for rare skin diseases in the North Africa and the wider Middle-East area

### MEET THE GENODERMATOSES NETWORK EXPERTS AT THE ECRD!



- Friday 9 May, 14.00-15.30:** Theme 1 - Centres of Expertise (Part I) - Best Practices (example 1) - Specialised centre for epidermolysis bullosa, Leena Bruckner-Tuderman
- Saturday 10 May, 9.00-10.30:** Theme 1- European Reference Networks - The example of the Austrian Epidermolysis Bullosa Centre and how it links into a broader network, Gabriela Pohla-Gubo
- Saturday 10 May, 15.45-17.15:** Theme 5 - Emerging Concept & Future Policies for Rare Disease Therapies : Emerging alternatives to medical interventions, Smail Hadj-Rabia

### ACKNOWLEDGEMENTS

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- Fondation René Touraine: since 2003
- Laboratoires Pierre Fabre: 2003-2011
- EuroScience Rammal Award: 2009

### JOIN US! SHARE YOUR EXPERTISE!

- If you wish to learn more about the Genodermatoses Network, join our group of experts or participate in our activities.
- Please contact Marie Guillou, Project Manager, [office@genodermatoses-network.org](mailto:office@genodermatoses-network.org)