

Working together at last for Rare Diseases: ERN-Skin

Professor Celia Moss OBE, 21st November 2017

People with rare conditions suffer on many levels from disconnected health care. Patients often feel isolated; support groups may feel ignored by doctors; GPs struggle to find specialist information; experts lack forums for discussion, training and research. Care is uneven – some patients are referred promptly to the right specialists while others wait months or even years. No single hospital or even country has the expertise to treat all rare conditions. But rare disease has moved up the political agenda, and at last Europe, to which happily we still belong, has provided a solution.

The “European Reference Network for Rare or Low Prevalence Diseases”, launched in March 2017, recognises the difficulties of providing high-quality, cost-effective care for these conditions which affect the lives of around 30 million EU citizens. Twenty-four specialties are covered, one of which is Dermatology. Of only 8 conditions under “ERN-Skin” one is “Ectodermal Dysplasia, Incontinenti Pigmenti and Related Disorders.” This is a triumph and puts ED firmly on the main stage.

The 56 ERN-Skin participating centres (“healthcare providers” or HCPs) in 18 European countries were selected following a tough bidding process; 7 are in the UK (see list); indications are that Brexit will not exclude us.



Further information is available via the links below. Patients and support groups are firmly embedded in the networks. Europe provides infrastructure for networking and grants but does not fund care. Continued membership after 5 years will depend on our performance.

From right to left in the picture:

Holm Schneider, Ulrika Holtzer, Smail Hadj-Rabia (Necker), Lenja Wiehe (Eurodis), Celia Moss, Johanna Hammersen (Erlangen), Olivia Khalifa

I’m writing this on Eurostar after attending the first working meeting of ERN-Skin in Paris. The ED sub-group (photo) was chaired by two experts, Holm Schneider, paediatrician from Germany and Smail Hadj-Rabia, dermatologist in Paris, and included patient representatives and medical experts. We had already met “virtually” by email and in a web-based conference. Our first task was to map available resources and define priorities for the next 5 years. We had 2 hours to put our findings together and 10 minutes to share them with the other ERN-Skin members. Like many groups, we found few up-to-date guidelines for managing these conditions; tests are not always available; training opportunities are patchy. We need more “outcome measures”, that is ways to measure the performance of HCPs and to determine whether they actually make any difference to patients. These should include basic clinical measures such as how many ED babies die

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from over-heating, patient-reported outcome measures (PROMs) such as whether families received appropriate information, and “burden of disease” questionnaires about symptoms, time lost from work or school, financial costs and quality of life. We should also measure “process issues” such as waiting times for appointments and accessibility of the multidisciplinary team.

This meeting introduced the Clinical Patient Management System (CPMS), a secure, web-based IT platform, accessible to all ERN members, which will enable experts to discuss cases. ERNs are not directly accessible to individual patients: their doctor would refer their case to the relevant ERN member in their country. The referring doctor can invite experts from any ERN, an important point for ED disorders which affect more than just skin. Patients will receive summarised conclusions from this virtual panel.

By combining resources scattered across countries, the ERNs will give healthcare providers access to a much larger pool of expertise. The networks will enable experts to share best-practice guidelines and training resources, and to focus on innovation rather than “re-inventing the wheel”. The future looks brighter for people with rare disease.

Further information

ERN website: https://ec.europa.eu/health/ern_en

ERN factsheet: http://europa.eu/rapid/press-release_MEMO-17-324_en.htm

ERN-Skin website: <http://www.genodermatoses-network.org/spip.php?article483>

ERN-Skin factsheet: https://ec.europa.eu/health/sites/health/files/ern/docs/ernskin_factsheet_en.pdf

List of UK and Ireland ERN-Skin Healthcare Providers

Barts Health NHS Trust

Birmingham Children's Hospital NHS Foundation Trust

Guy's and St Thomas' NHS Foundation Trust

NHS Tayside

Great Ormond Street Hospital for Children NHS Foundation Trust

The Leeds Teaching Hospitals NHS Trust

Cardiff and Vale University Health Board

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