France

Smail Hadj-Rabia, Christine Bodemer, MAGEC, Necker-Enfants Malades, Paris, France

Salah Ferkal, Clinical Research Docteur, Neurofibromatosis Centre of Reference, Henri Mondor Hospital

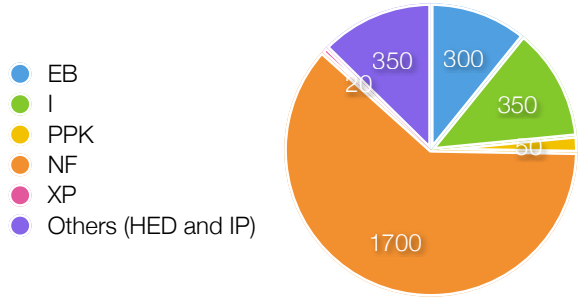
The first national plan for rare diseases (2005-2008) enabled the development of a national network organised around "centres of reference" and "centres of competence". In the framework of the first national plan for rare diseases (2005-2008), centres of reference gather university hospital teams highly specialised. A rare diseases "centre of reference" provides an expertise role for a disease or a group of rare diseases and a role enabling it to exercise an interregional, national and international attraction. Necker Hospital site gathers 15 centres of competence including MAGEC, a "centre of reference" for genetic skin diseases.

"Centres of reference" don't aim at managing all the rare diseases patients. One of their mission is to progressively organise a network of existing structures throughout the country: the "centres of competence". "Centres of competence" aim at providing health care and follow up to patients, near their home, and participate in all the missions of "centres of reference".

CEMARA : a national database

Approved by European authorities, CEMARA in a national database for rare diseases. It gathers the data from 25 centres.

Diseases	Number of cases
Epidermolysis Bullosa	300
Ichtyosis	350
Neurofibromatosis	1700
Palmoplantar Keratoderma	50
Hypohydrotic Ectodermal Dysplasia	150
Incontinentia Pigmenti	200
TSC	50
Total	2800



Number of patients with epidermolysis bullosa (EB), recessive icthyosis (I), palmoplantar keratoderma (PPK), neurofibromatosis (NF), xeroderma pigmentosum (XP) and other genodermatoses (hypohydrotic ectodermal dysplasia and incontinentia pigmenti)

5 centres of reference for rare dermatological diseases

- Paris: Neurofibromatoses Center of Expertise for NF1 patients (Adults: Hôpital Henri Mondor, Children: Hôpital Necker and Hôpital Trousseau) and NF2 patients (Adults: Hôpital Beaujon). Website: neurofibromatoses.aphp.fr
- Paris: Genetic Skin Diseases Center of Expertise. Website: www.magec.eu.
- Paris: Tuberous Sclerosis Center of Expertise
- Bordeaux/Toulouse: Rare Dermatological Diseases Center of Expertise
- Nice: Inherited Epidermolysis Bullosa Centre of Expertise

6 centres of competence for genodermatoses

26 centres of competence specifically for neurofibromatosis

Costs of the disease for the patients

The costs of the disease for patients have been assessed for:

- Epidermolysis Bullosa: 1000 euros/month
- Ichthyosis: cream reimbursed by the National Health Service

‣ 2008 & 2009

- Palmoplantar keratoderma: pedicure reimbursed by the National Health Service
- Neurofibromatosis: learning disabilities covered
- Xeroderma Pigmentosum: sunscreen reimbursed by the National Health Service
- Anhidrotic Ectodermal Dysplasia: dental care reimbursed by the National Health Service under some conditions
- Incontinentia Pigmenti: reimbursed by the National Health Service under some conditions

Projects at MAGEC to improve patients' health care

MAGEC follows the patients according to a multidisciplinary approach. The centre develops several projects to improve the health care of patients:

• Epidermolysis Bullosa

The projects focus on:

- improving quality of life - includind sexual life- to answer a frequent request of patients
- gene therapy
- healing and dressings.

• Ichthyosis

The projetcs focus on the definition of common terms to describe the disease: clinical definition, histology, etc.

• Neurofibromatosis (children)

Projects concern learning difficulties and their prevention.

• Xeroderma Pigmentosum

A National Protocol of Diagnosis and Care is available on the "Haute Autorité de Santé" website, www.has.fr (ALD n°31: Xeroderma Pigmentosum : www.has-sante.fr/portail/jcms/c_556980/ald-n31-xerodermapigmentosum)

• Anhidrotic Ectodermal Dysplasia

The writting of a National Protocol of Diagnosis and Care is in process.

• Incontinentia Pigmenti

Projects focus on the neurologic aspects of the disease.

Patients' organisations in France are well developed

Patients' organisations for severe genodermatoses are very active and well developed.

• Epidermolysis Bullosa

L'Association d'Entraide EBAE, www.ebae.org, gathers epidermolysis bullosa patients.

• Ichthyosis

L'Association Nationale des Ichtyoses et Peaux Sèches Pathologiques, ANIPS, www.anips.net gathers ichthyosis patients in France. Athina is located in Monaco, www.aaimonaco.org.

• Palmoplantar Keratoderma

Association Pachyonichie Congénitale, www.pachyonychie-congenitale-lecoeuraupied.com gathers palmoplantar keratoderma patients.

• Neurofibromatosis

Recklinghausen, www.anrfrance.org, and Ligue Française Contre les Neurofibromatoses, asso.orpha.net/LFCN/cgi-bin, gathers neurofibromatosis patients.

• Ectodermal Dysplasia

The Association Française des Dysplasies Ectodermiques, www.afde.net, gathers ectodermal dysplasia patients.

• Incontinentia Pigmenti

Incontinentia Pigmenti France, www.incontinentiapigmenti.fr gathers incontinentia pigmenti patients.

• Xeroderma Pigmentosum

"Enfants de la Lune", asso.orpha.net/AX/debut.htm, gathers xeroderma pigmentosum patients.

ZOOM>>> Complete photoprotection, 13 years of hindsight: the experience of "les Enfants de la Lune"
The "enfants de la Lune" association gathers the French xeroderma pigmentosum patients. For 13 years, the association has tried out a complete photoprotection of patients. This complete photoprotection is made possible by the laying of UV filters on the windows, the use of a dosimeter, cremes or suits. The excellent results of the UDS (Unscheduled DNA Synthesis) tests show the efficiency of this complete photoprotection.
The "Enfants de la Lune" gives advice, brings a financial help, organises activities enabling the patients to live protected from the sun and making schooling and their social integration easier. Gathering of families, activities (including scuba diving, caving, skiing), summer camps, sharing of experience, debates, awareness raising are the numerous activities organised by the association.
The "Enfants de la Lune" supported the creation of 2 associations: one at Mayotte and one in Tunisia.