



**6th Genodermatoses in Mediterranean working session  
1st TAG meeting**

May 22 - May 25, 2009, Greece

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

# Epidemiology: diseases

- Epidermolysis bullosa : 300 patients
- Ichthyosis : 350 patients
- Palmoplantar keratoderma : 50 patients
- Neurofibromatosis : 1700 patients
- Xeroderma Pigmentosum : 20 patients
- HED : 150 patients
- IP : 200 patients
- TSC : 50 patients

# Epidemiological data

- Based on the information of CEMARA database
- National Database, 25 centres of rare disease
- Role of the statistical department
- Contract
- Approved by the European Institutions

# Needs and costs

- Epidermolysis bullosa : 1000 euros/month
- Ichthyosis : free ointment
- Palmoplantar keratoderma : free pedicure
- Neurofibromatosis : learning disabilities management
- Xeroderma Pigmentosum : free solar protection
- Other genodermatoses
- HED : free dental care
- Incontinentia Pigmenti : free dental care

# Expertise

- 5 centers of expertise for genodermatoses
  - Paris
    - Neurofibromatosis
    - MAGEC
    - TSC center
  - Bordeaux/Toulouse
    - Rare skin diseases
  - Nice
    - Epidermolysis bullosa center

# Competence and Network

- 6 centers of competence for genodermatosis
  - Designated by a common decision of RC
  - Can not localized in a region with a known reference center
  - Contract

# Management of Diseases

- Epidermolysis bullosa :
  - Quality of life with the focus on affective and sexual life (special asking of patients)
  - Genic Therapy
  - Wound healing and Dressing
- Ichthyosis
  - Common definition of the terms used for the description of the disease : clinical, histological....

# Management of Diseases

- Neurofibromatosis
  - Learning disabilities and their prevention
- Xeroderma Pigmentosum
  - Protocole National de Dépistage et de Soins
- Hypohidrotic Ectodermal Dysplasia
  - Protocole National de Dépistage et de Soins
- Incontinentia Pigmenti
  - Neurological aspect of the disease



# Associations of patients

- Epidermolysis bullosa : EBAE
- Ichthyosis : ANIPS and Althina
- Palmoplantar keratoderma : pachyonychia
- Neurofibromatosis : Two associations
- Xeroderma Pigmentosum : Enfants de la Lune
- Hypohidrotic Ectodermal Dysplasia : AFDE
- Incontinentia Pigmenti : IPF

# National Network

- Reference centers/Competence centers
- Reference centers for other diseases
  - in Necker : 15 centers
  - outside : rare dental diseases....
- European
- Genodermatoses and Mediterranean