TAG 2007 335

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

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# A Portuguese referral centre for genodermatoses

- Multidisciplinary genodermatoses outpatient clinic on a weekly basis
- Team:
  - M. Marques Gomes (Director of the Dermatology Department),
  - President of the Portuguese Society of Dermatology)
  - Carolina Gouveia (Pediatric Society of Dermatology)
  - Isabel Cordeiro (Director of the Genetic Department)
  - Ana Berta Sousa (Clinical Geneticist)
  - Juliette Dupont (Genetic Resident)
  - L. Soares de Almeida (Dermopathology)
  - Marisa André (Dermatology Resident)

# 1. The Portuguese situation: Lisbon

| Disease | Number of patients |
|---|---|
| **Epidermolysis Bullosa** | **3** |
| **Severe ichtyosis** | **9** |
| **Palmoplantar keratoderma** | **6** |
| **Neurofibromatosis** | **225** |
| **Others** | **66** |
| Ectodermal dysplasia | 8 |
| Incontinentia pigmenti | 10 |
| Oculocutaneous albinism | 6 |
| Cutis laxa | 4 |
| Bloom syndrome | 1 |
| Tuberous sclerosis | 31 |
| Piedbaldism | 4 |
| Werner syndrome-Progeria *like* | 2 |

# 1. The Portuguese situation: Oporto

| Disease | Number of patients |
|---|---|
| **Epidermolysis Bullosa** | **10** |
| **Severe ichtyosis** | **4** |
| **Palmoplantar keratoderma** | **1** |
| **Neurofibromatosis** | **1** |
| **Others**<br>Ectodermal dysplasia<br>Progeria<br>LEOPARD syndrome<br>Piebaldism | **8**<br>5<br>1<br>1<br>1 |

# 2. Assessment of needs and costs of the diseases

- Insufficient governmental social support:

*Genodermatoses are considered rare chronic disorders and, as such, patients have special rights, which are clearly insufficient*

- Free health care in public health facilities
- Extra financial support (+ 50% of per child subsidy) until the age of 18
- 12th November of 2008: approvement of the *National Programme for Rare Diseases*

# 2. Assessment of needs and costs of the diseases

- Genetics department project: *“meet the best needs of the NF1 population”*
  - clinical evaluation
  - psychological evaluation
  - educational evaluation
  - social environment of the patients and families
  - evaluation of the needs of NF1 patients

# 2. Assessment of needs and costs of the diseases

Difficulty at collecting data from the cases of the entire territory:

- Until now, it has not been possible to accurately assess these items
- It’s imperative to accomplish the task of collecting all the existing portuguese cases

# 3. Development of centers of expertise

We have created a genodermatosis outpatient clinic, but we haven’t received significant number/relevant cases

# 4. Development of patient associations

- Portuguese Neurofibromatosis Association (APNF)
- RARÍSSIMAS (Association of rare diseases)
- Portuguese Epidermolysis Bullosa Association
  - almost inactive at present
  - collaboration with spanish colleagues, nurses, patients and families in order to get specific training (portuguese DEBRA?)

# 5. Strategies of the management of genodermatosis

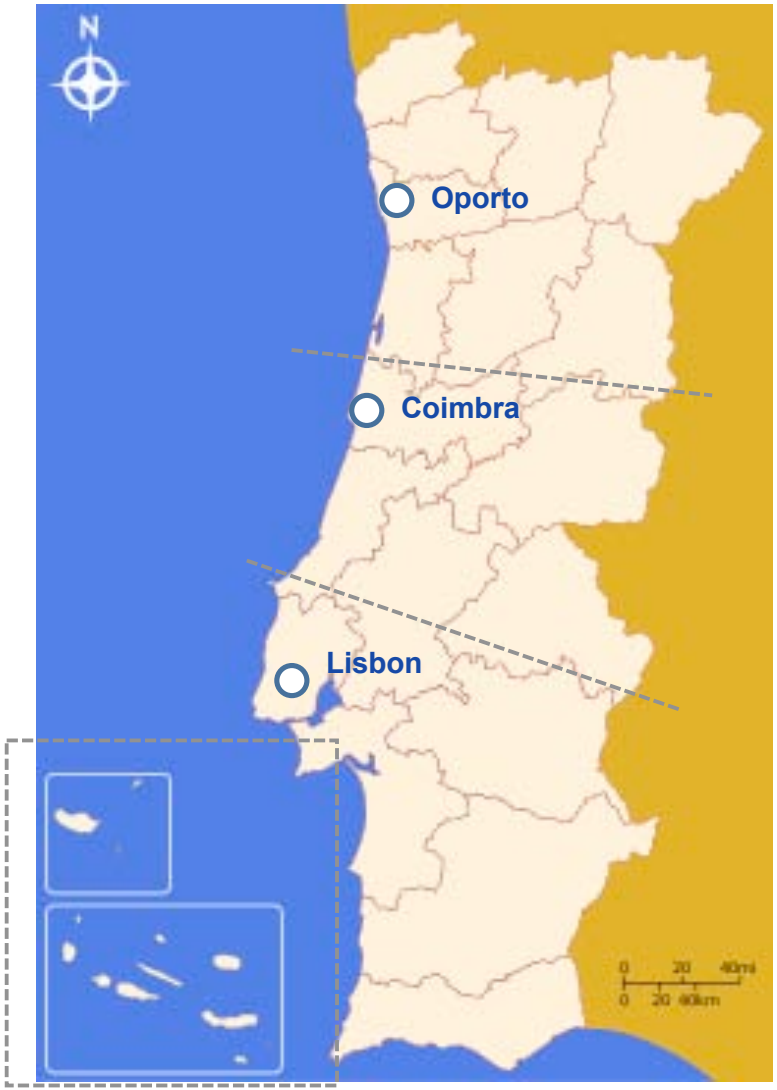
Different sensibilization level:

- medical students
- health care workers
- mass media awareness
- clarifying sessions at school´s
- active participation and spreading of our activities in General Practicioners, Pediatrics, Dermatologists and Genetics meetings and their journals

# 6. Development of a national network

- We have realized that we had to organize a national network in order to reach all the existing cases
- All the dermatologists, general practitioners, geneticists and pediatricians, with public and/or private practice will be asked to refer the patients with one of the 6 disease groups to the genetic outpatient clinic of their respective geographic area

# 6. Development of a national network




North of  Portugal: Oporto

Centre of Portugal: Coimbra

South of Portugal, Madeira and Azores : Lisbon

Centre of expertise for the 6 groups of diseases

# 7. Collaboration on an european level

- organize an European epidemiological database
- International cooperation and interaction
- improvement of diagnosis
  - cooperation with genetic diseases referral centers: molecular genetics diagnosis, electronic microscopy, specific biochemical assays, immunologic studies…
- to improve treatment, providing a multidisciplinary team approach

# 8. National program for rare diseases

MILES TO GO...:

- … to know the real national epidemiological data of these diseases
- …to involve specific trained nurse, social worker and psychologist in the management of these diseases
- …to provide information/formation for parents/family members about management and prevention
- … to assess the needs of the patients and their family
- … development of associations of patients