Portugal

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Genodermatoses: from chronical diseases to rare diseases status

Genodermatoses are considered as chronical diseases by the government, which provides the patients with some rights: free care in public health services, 50% additional financial support from State until the age of 18. These rights are said to be insufficient by the medical staff.

On November 12, 2008, a National Plan for Rare Diseases was adopted by the Portuguese government with the following priorities: information, specialised services for rare diseases patients and national centres of expertise on rare diseases.

Epidemiological data collected in Lisbon and Porto

- Data collected in Lisbon

Disease	Number of cases
Epidermolysis Bullosa	3
Severe Ichtyosis	9
Palmoplantar Keratoderma	6
Neurofibromatosis	225
Other Genodermatoses *	66
Total	309

* Other Genodermatoses	Number of cases
Anhidrotic Ectodermal Dysplasia	8
Icontinentia Pigmenti	10
Tuberous Sclerosis	31
Eye and Skin Albinism	6
Cutis Laxa	4
Bloom Syndrome	1
Piébaldism	4
Werner Syndrome	2
Total	66

- Data collected in Porto

Disease	Number of cases
Epidermolysis Bullosa	10
Severe Ichtyosis	4
Palmoplantar Keratoderma	1
Neurofibromatosis	1
Other Genodermatoses *	8
Total	24

* Other Genodermatoses	Nombre de cas
Anhidrotic Ectodermal Dysplasia	5
Progeria	1
Syndrome LEOPARD	1
Piedbaldisme	1
Total	8

The development of a Portuguese centre of reference and a first project focused on neurofibromatosis patients' needs

- The development of a Portuguese centre of reference

A weekly multidisciplinary consultation for genodermatoses patients has been introduced at the Santa Maria Hospital of Lisbon. The multidisciplinary team is composed of M. Marques Gomes (Head of the Department of Dermatology and President of the Potuguese Society of Dermatology), Carolina Gouveia (Potuguese Pediatric Society of Dermatology), Isabel Cordeiro (Head of the Department of Genetics), Ana Berta Sousa (Geneticist), Juliette Dupont (Genetics Resident), L. Soares de Almeida (Dermopathologist), Marisa André (Dermatology Resident). To date, the number of addressed cases was limited.

- The objectives of the Portuguese centre of reference

- Collect epidemiological data,
- Involve in patients management specially trained nurses, social workers and psychologists,

▸ 2008 & 2009

- Improve information and training of parents and families for health care and prevention,
- Evaluate the needs of patients and families,
- Develop patients' organisations.

• The NF project of the Genetics Department

This project aims at best answering the needs of neurofibromatosis 1 patients through an evaluation of their needs: clinical evaluation, psychologic evaluation, learning abilities evaluation, patients and families social environment evaluation.

The development of a national network close to the patient

Due to the difficulty of gathering for all the patients of the national territory, a national network is developing. The aim of this network is to reach out to each patient. In this way, every dermatologist, general practitioner, geneticist and pediatrician will adress his genodermatoses patients to the genetic consultation they geographically depends from.

The national network for health care of genodermatoses should be organised around three centres: Porto in the North, Coimbra in the Centre, Lisbon in the South, Madere and Acores.

Awareness raising of medical staff and general public

Awareness raising on severe genodermatoses is made on several levels: medecine students, health professionals, general public through media, presentations in school, active participation of general practitioners, of pediatricians, of dermatologists and geneticists, spreading of centre of reference's activities during meetings and in specialised journals dedicated to general practitioners, pediatricians, dermatologists and geneticists.

Identification of patients' organisations

All rare diseases patients are gathered in RARÍSSIMAS, the Portuguese Organisation for rare diseases. There are patients' organisations for the following pathologies:

• Neurofibromatosis

The Portuguese Organisation for neurofibromatosis patients is the APNF.

• Epidermolysis bullosa

The Portuguese Organisation for epidermolysis bullosa patients develops few activities. A collaboration is in process with Spanish dermatologists, nurses, patients and families in the training field to create a DEBRA organisation in Portugal.

Collaborations with European and international centres of expertise would be useful to improve the health care of patients in Portugal

These collaborations and interactions with centres of expertise for genetic diseases would allow improving diagnosis (molecular diagnosis, electron microscopy, specific biochemical tests, immunology studies) and treatments. These collaborations would also offer a multidisciplinary approach to the patients.