

An international public health project launched by

FONDATION RENE TOURAINE

A EUROPEAN FOUNDATION PROMOTING THERAPEUTIC ADVANCES IN DERMATOLOGY

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Technology and Expertise Development (Tunisia)

GENODERMATOSES AND MEDITERRANEAN

Working together toward better care

Promoting the creation of national networks
ensuring medical treatment and social protection
for patients suffering from severe genodermatoses:
the current situation, the needs of patients
and their families, problems encountered, solutions proposed

Working meeting

26-27 May 2005

Université Saint Joseph, Beyrouth

An action supported by



and

Ministry of Health, France / Ministry of Health, Morocco

French Embassy in Iran, French Embassy in the Sultanate of Oman, French Consulate in Jerusalem

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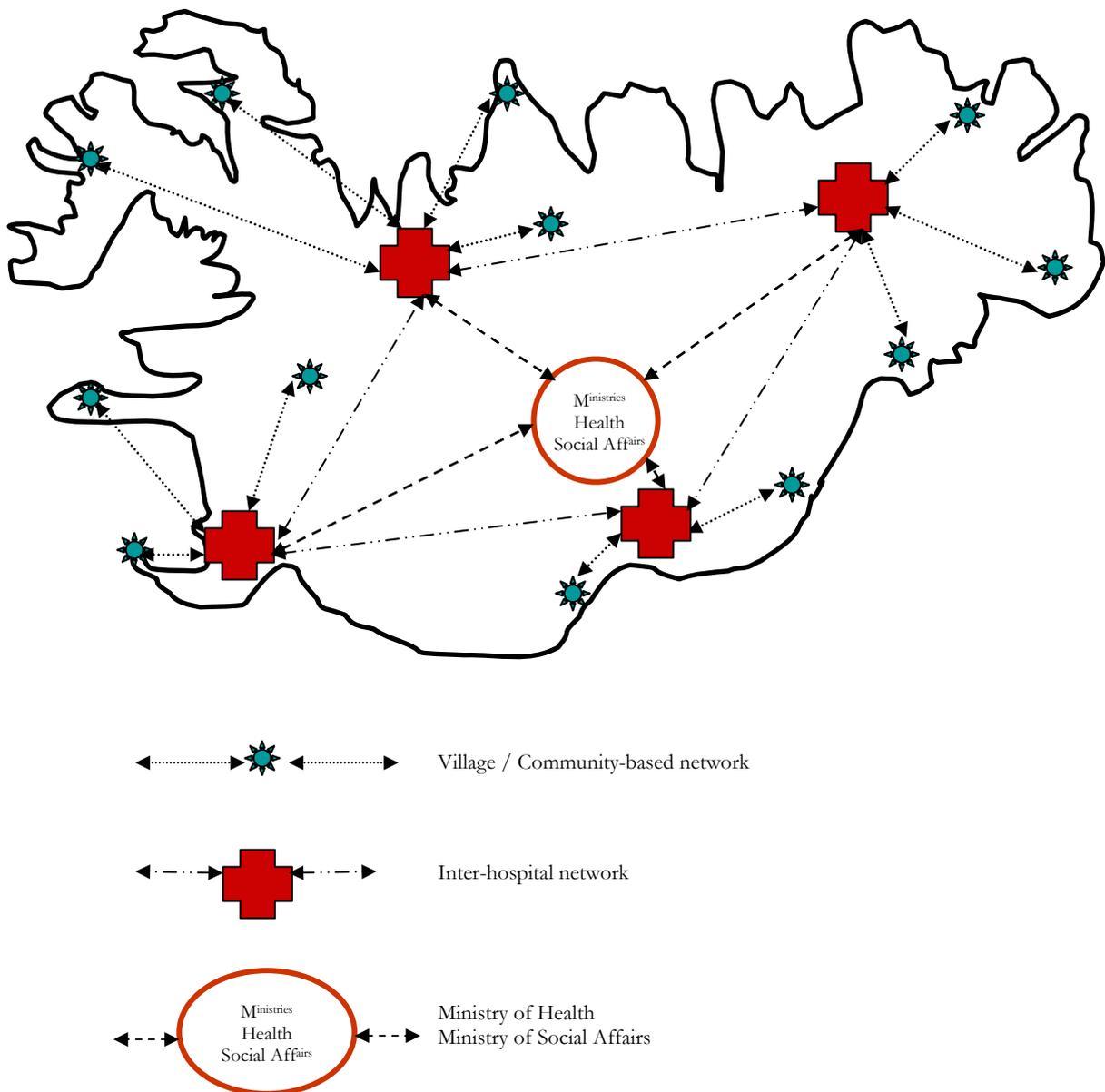
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EGYPT	EL-GAMAL	Emad
FRANCE	DURAND-DROUHIN HADJ-RABIA	Jean-Louis Smail
GREECE	KATSAROU STAVRIANEAS	A. Nikolaos
IRAN	MOSLEHI	Homayoun
IRAQ	KUSAIMI	Nuha T.
ITALY	EL-HACHEM ZAMBRUNO	Maya Giovanna
LEBANON	MEGARBANE NASSIF TABET TOMB	André Hana H. May Haddad Roland
LIBYA	BENGHAZIL	Mohammed
MOROCCO	IDRISSI AZZOUZI BENCHIKHI	Abdellatif Hakima
OMAN	ABLA REDHA MUSTAFA	Al-Ismaïli Mohamed
PALESTINE	ARDA	Hisham
TUNISIA	KAMOUN NOUIRA	Mohamed Ridha Rafaa

OUR GOAL

To improve the quality of medical care and social protection offered to patients suffering from severe genodermatoses and their families through the creation of care networks under the auspices of Ministries of Health and Social Affairs in Mediterranean and Middle Eastern countries



SEVERE GENODERMATOSES

A major handicap for patient and family

A public health problem linked to consanguinity
in the Mediterranean basin and the countries of the Middle East

4 targeted pathologies

◇ **Epidermolysis bullosa** (e.g.: 12 000 / 10 000 000 in Tunisia)

This form of severe genodermatosis is due to a permanent defect in dermal–epidermal adhesion



*Blisters appear spontaneously or following minor trauma, mucous membranes frequently erode
The condition of the skin in these patients is similar to that of severe burn victims, as the barrier function is highly impaired*



With recurrent scarring, the toes or fingers may fuse together

Without treatment, patients are likely to die prematurely from malnutrition and infections

◇ **Ichthyosis** (e.g.: 1 150 / 10 000 000 in Tunisia)



Without treatment, the body is covered with large, brown scales

◇ **Palmoplantar Keratoderma and Meleda syndrome**



Without treatment, the skin on the hands and feet acquires a horny texture, making movement extremely difficult

◇ **Xeroderma pigmentosum** (e.g.: 1 000 / 10 000 000 in Tunisia)

This form of severe genodermatosis is caused by a genetic defect in DNA excision repair following exposure to the sun



With no protection from the sun's ultraviolet radiation, affected children quickly succumb to a variety of fatal skin cancers

EXECUTIVE SUMMARY

The project

- ◇ **Launched in 2003 by Fondation René Touraine**
- ◇ **In partnership with Laboratoires Pierre Fabre**
- ◇ **Set up by dermatologists as well as dermatology and paediatric dermatology societies**

- ◇ **Focused on patients and families suffering from severe genodermatoses**
 - ✗ these diseases are experienced as nothing less than a family curse; due to the fact that they are genetic disorders, several children of the same family may be affected
 - ✗ they are particularly stigmatising, giving rise to feelings of failure and shame
 - ✗ they result in the isolation of the affected child (difficulty of access to school and leisure activities appropriate to her/his age group) and consequently to that of the family as well

- ◇ **In the countries of the Mediterranean basin and the Middle East**

26-27 May 2005

- ◇ **An international gathering of more than 40 experts**
 - ✗ physicians, researchers, public health officials
 - ✗ who self-funded -or obtained funding- for their travel and accommodation

- ◇ **Objectives**
 - ✗ identify hospital requirements to improve the quality of care for severe genodermatoses
 - ✗ with the collaboration of public health officials, strengthen care networks in order to allow for diagnoses as quickly as possible after birth, community-based care, providing of information to families, education of children, co-ordination with hospitals
 - ✗ consider ways in which national and international health authorities can be involved in this public health project

- ◇ **The adopted strategy combines**
 - ✗ a broad-based approach, incorporating
 - the perspective of public health officials on
 - public health planning constraints
 - the creation of care/health networks for the treatment of severe genodermatoses
 - the involvement of community associations in the care and management of severe genodermatoses
 - the sharing of information through the use of networks such as Orphanet or the Centre for Arab Genomic Studies
 - ✗ a country-by-country approach, focusing on
 - the current status of the care and management of severe genodermatoses and the problems encountered
 - recommended solutions to improve medical care and social protection for patients suffering from severe genodermatoses and their families, in the areas of care delivery, education, training, information and research

Numerous initiatives

◇ To improve the quality of care

multidisciplinary consultation services, specialised centres, diagnosis procedures, patient support organisations, special measures for the most disadvantaged patients, etc.

◇ To promote the communication of information and training

informational Web sites, educational brochures

◇ To support research

on the national and international levels: many co-operative projects, private and public funding

◇ To reinforce ties with

Ministries of Health, Social Affairs, Education and Research, universities, non-governmental organisations, regional offices of international organisations (UNICEF, UNDP, WHO), patients' associations

◇ Spurring the development of national plans and programmes

a national genodermatoses committee and the creation of vigilance networks in Algeria, national programmes for the prevention of genetic diseases in Iran, national health programmes in Libya, centres officially recognised as specialising in rare diseases in France and in Italy

Sources of continuing difficulties for patients and their families

◇ Overspecialisation of medical practice / challenges faced in mobilising the necessary skills and expertise

◇ Lack of awareness -among patients as well as their physicians- of the existence of centres specialising in the treatment of these diseases

◇ Obstacles to the identification of community medical and paramedical resources / distance of patients from health care centres / late diagnoses

◇ Insufficient co-ordination between independent physicians and hospitals through hierarchies or networks in need of correction to support a more comprehensive approach to the patient covering all aspects of his or her life

◇ The high cost of treatment and limited availability of medicines

◇ Insufficiently trained caregivers (physicians, nurses, social workers)

◇ Limited access to information, affecting physicians, patients, families and the general public

Original and realistic proposals

◇ Registry of activities in the area of genodermatoses

- ✗ collect and maintain epidemiological registers
- ✗ target 4 or 5 severe genodermatoses
- ✗ identify work groups

◇ Obtain recognition for severe genodermatoses as chronic diseases and/or handicaps

- ✗ by national health and social affairs authorities
- ✗ by the WHO (also propose the creation of a regional genodermatosis observatory)
- ✗ granting of disabled identity cards to improve the quality of medical care and social protection

◇ Promote the development of national associations for severe genodermatoses

- ✗ creation of multidisciplinary centres, community-based care networks adapted to each country, installation of diagnostic facilities, prevention programmes, appropriate training, better information provided to caregivers, etc.

◇ Build on existing networks

- ✗ associations, genetic disease prevention programmes, primary care networks, networks formed by NGOs (combating leprosy, HIV/AIDS), Orphanet, Geneskin (1 July 2005): a European project advancing diagnosis, management and awareness of genodermatoses

◇ Facilitate access to medicines

- ✗ establish an international distribution facility for medicines
- ✗ build ties with biotechnology firms and pharmaceutical companies interested in the development of orphan drugs

◇ Improve training and access to information

- ✗ medical treatment guidelines / protocols
- ✗ hands-on training for caregivers
- ✗ inter-hospital and inter-university agreements

◇ Encourage the growth of patients' associations

- ✗ promote their creation or reinforce ties with existing groups
- ✗ request their participation in furthering the development of networks

◇ Support research

- ✗ many topics to study: prevalence of diseases in each country, evaluation of care-giving strategies, etc.
- ✗ national and international collaboration (networks) is essential to promote rapid and efficient progress in research and to assist in the arrangement of funding for genodermatoses

◇ Working together toward better care, 26-27 May 2006 in Tunisia

- ✗ evaluation of projects implemented in participating countries (working languages: English and French, with slides in English)

CHALLENGES IN THE CARE AND MANAGEMENT OF PATIENTS SUFFERING FROM SEVERE GENODERMATOSES

M.R. Kamoun
Tunisian Society of Dermatology
PanArab League of Dermatology Representative

Broad spectrum of manifestations complicates management

◇ Genetic

- ✗ mode of transmission: recessive, dominant
 - ✗ whether or not gene responsible has been identified
 - ✗ possibility of prenatal diagnosis
- hence the need for genetic counseling**

◇ Clinical

- ✗ age at onset: at birth, early or late
 - ✗ nature of lesions: blisters, redness, scales, etc.
 - ✗ extent and location of the affliction
 - ✗ impact on other organs: nervous system, skeleton, kidneys, digestive tract, etc.
- hence the need for a multidisciplinary approach to treatment**

◇ Prognostic

- ✗ life-threatening: epidermolysis bullosa, xeroderma pigmentosum
 - ✗ functional: ichthyosis, palmoplantar keratoderma, epidermolysis bullosa, xeroderma pigmentosum
 - ✗ social: visibility, social illness
- hence the need for an approach to treatment involving local communities**

◇ Therapeutic

- ✗ existence of regrettably expensive possible care strategies: ichthyosis, palmoplantar keratoderma, epidermolysis bullosa, xeroderma pigmentosum
- hence the need for treatment provided free of charge and an international distribution facility for medicines**

◇ Possibilities for prevention

- ✗ communicating the dangers of consanguinity
 - ✗ early (prenatal) diagnosis
 - ✗ photoprotection
- hence the need for information provided to entire populations**

Prevention

◇ The consanguineous marriage issue

- in the general population: information and education

- resilience of traditional behaviour
- relatively limited impact of prevention campaigns on behaviour despite greater awareness of risks related to consanguinity
- in high-risk families: genetic counselling
- lack of required skills

◇ **Prenatal diagnosis for pregnancy termination**

- lack of technical resources and equipment
- lack of human resources and skills
- absence of a legal framework
- ethical principles (justification)

◇ **Addressing vulnerability factors**

✗ xeroderma pigmentosum: UV

- education of children and parents concerning photoprotection: behaviour, clothing, sunscreen products, medication

✗ epidermolysis bullosa: trauma, secondary infections

- education of children and parents: local and systemic treatments

✗ ichthyosis and palmoplantar keratoderma: trauma, secondary infections, bacteria, fungi

Treatments

◇ **Preventive and symptomatic treatment regimens relying on frequent hospital checkups in co-ordination with community-based caregivers**

✗ xeroderma pigmentosum: surgical destruction of precancerous lesions, early ablation of skin cancers, photoprotection

✗ epidermolysis bullosa: local and systemic antiseptic and antibiotic treatments, bandages, nutritional supervision

✗ hereditary keratinisation disorders: keratolytic and antimycotic agents, retinoids

Psychological care

◇ **Impact of genodermatosis**

✗ on the parents: guilt, powerlessness, divine punishment, stigmatisation, exclusion, fears for the future

✗ on the child: self-image, stigmatisation, exclusion

Socio-economic aspects

◇ **Entirely dependent on the political will of all partners focused on solidarity**

PUBLIC HEALTH PLANNING CONSTRAINTS

A. Idrissi Azzouzi

Consultant Dermatologist and Head of Dermatological Disease Unit

“Direction de l’Epidémiologie et de la Lutte contre les Maladies”, Ministry of Health, Morocco

⇒ In order to promote efficiency, public health planners must consider

- ✗ the significance of the health issue in question
 - frequency: prevalence and incidence
 - seriousness: morbidity, mortality, incapacity, costs
- ✗ the issue’s technical and operational vulnerability
- ✗ the acceptability of proposed solutions”

THE CREATION OF COMMUNITY-BASED CARE NETWORKS

J.-L. Durand-Drouhin

Regional Hospital Agency Director, Limoges, France

What is a network?

From care network to health network: towards a single definition

⇒ Hospital-physician health network

✗ definition of a care network

“In order to better meet the requirements of the population (...) health care institutions may form care networks focusing on specific care delivery aims and resources (...) or certain diseases (...) to ensure a more efficient handling of patient referrals, facilitating co-ordination and the continuity of care (...). These networks may include independent medical practitioners and other health professionals (...).”

✗ definition of a health network

“The objective of a health network is to facilitate access to care, the co-ordination, continuity or multidisciplinary of treatments, health education, prevention, diagnoses, care delivery, evaluation. Such a network is comprised of independent health professionals, occupational physicians, health care institutions, health centres and patient representatives.”

⇒ 3 types of networks: from the hospital network and the physician network to the hospital-physician network

✗ hospital network: between private and public health care institutions

✗ physicians’ network: between independent medical practitioners

✗ hospital-physician network: between health care institutions and independent medical practitioners

⇒ 1 challenge

✗ placing the patient at the heart of the health care system: access to care, continuity, cohesiveness, comprehensiveness

⇒ 2 objectives

✗ break down barriers in the health care system: co-ordinated intervention of health professionals from a variety of specializations

✗ enhance the skills and expertise of participants: exchanges of techniques, guidelines, training

⇒ **4 essential components**

- ✗ project that associates professionals: voluntary participation, co-operation, sharing of common goals
- ✗ medical or medico-social co-ordination: integration and interaction
- ✗ training of and providing of information to participants: introduction to network procedures, respect for established principles, feedback
- ✗ evaluation of practices, quality and costs

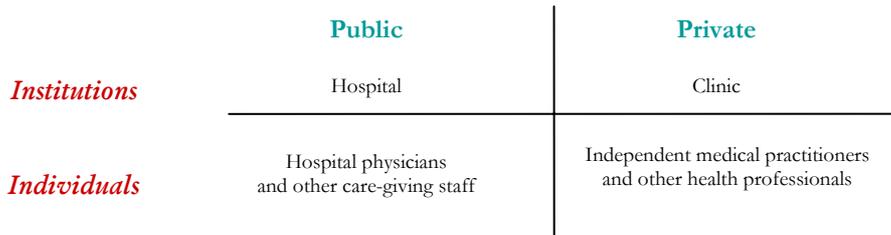
⇒ **4 imperatives**

- ✗ free choice by the patient: information provided on how network operates
- ✗ sharing of information and confidentiality
- ✗ single medical or medico-social file for each patient: standardised, capable of being shared, possibly computerised
- ✗ information system: knowledge of health status at any moment, conformity of medical practices

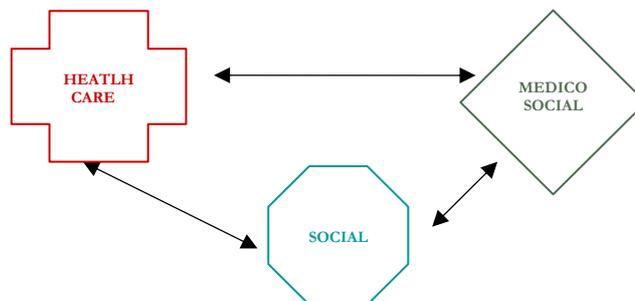
What is the environment of a network?

⇒ **The health care system**

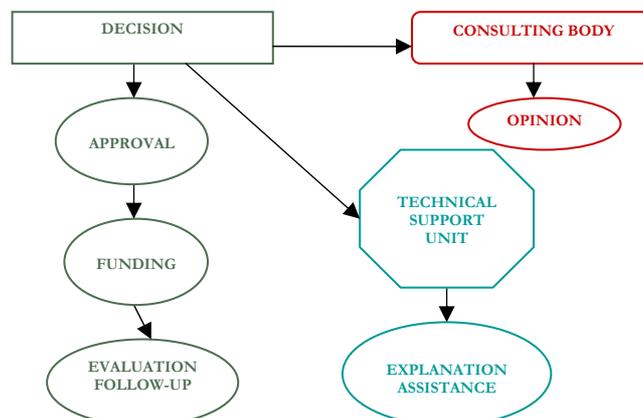
- ✗ the health care sector



- ✗ the social and medico-social sector



⇒ **The decision-making process**



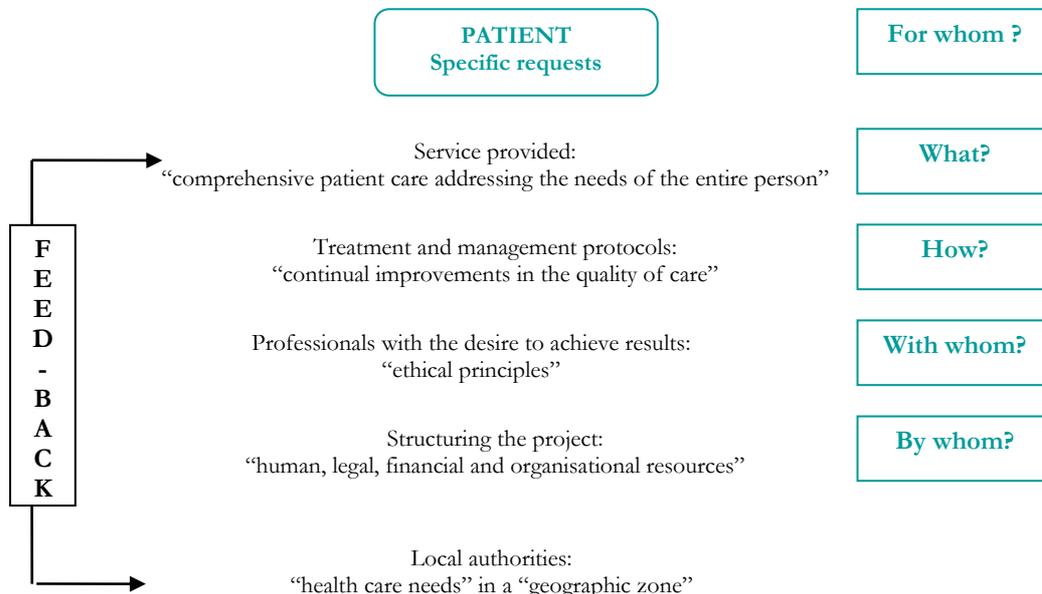
⇒ Funding that is identified, long-lasting, specific and geared to the network's particular needs

How to build a network?

⇒ Providing a structure for the network requires

- ✗ the identification of a need
- ✗ the determination of quantitative and qualitative goals
- ✗ the demarcation of a territory
- ✗ the designation of participants and their roles
- ✗ the commitment to a multi-year action plan
- ✗ the determination of funding resources
- ✗ the selection of assessment indicators and the implementation of monitoring procedures

⇒ Questions to be asked (G. Poutout)



An example: the diabetes network

⇒ Assessment

- ✗ widespread, the world's fastest growing disease
- ✗ 20,000 patients with diabetes in the Limousin region
- ✗ elderly population (12% of inhabitants are 75 or older out of a total population of 710,000)
- ✗ chronic and serious health condition
- ✗ high risk of degenerative complications
- ✗ certain treatments are not widely available (chiropractors, nutritionists, etc.)
- ✗ prevention, health education and monitoring are insufficient

⇒ Objectives

- ✗ pooling of knowledge and experience, skills and practices, harmonisation of monitoring and treatment procedures (protocols, guidelines, personalised patient diaries), organisation of multidisciplinary staff meetings
- ✗ educating patients and families about the disease, prevention of complications (self-monitoring

and treatment techniques, life hygiene, chiropodical care, diet monitoring, etc.)

◇ **Participants**

- ✗ the patient: a participant in her/his own care
- ✗ the general practitioner: plays a pivotal role, inclusion, orientation and individualised protocols, guidance for patient diary maintenance
- ✗ specialists: prevention and monitoring of complications (endocrinologists, cardiologists, ophthalmologists, nephrologists, etc.)
- ✗ nurses, chiropodists: 6 consultations per year and per foot (grade 2 and 3 ulcers), nutritionist: 2 consultations per year, etc.

Tools employed in the French experiment

◇ **A convention of establishment**

document creating the network: organisation, operations, assessment

◇ **A network charter**

document laying down the ethical principles for the network: rights and duties of network members

◇ **An informational document provided to patients**

describes how the network operates, the services it offers, the resources brought to bear to communicate information

◇ **A patient membership form**

provides a formal record documenting the patient's desire to be served by the mechanisms implemented by the network, thereby ensuring enlightened consent

◇ **Multi-year budget planning**

◇ **A legal support structure for the receiving of funds**

A successful health network requires

G. Poutout

- | | |
|---------------------------|--|
| ◇ A heart | the person, the patient |
| ◇ A soul | professionals with a desire to achieve results |
| ◇ Legs | treatment protocols |
| ◇ A nervous system | information system |
| ◇ A head | means of co-ordination |
| ◇ A skeleton | legal status |
| ◇ A stomach | financial resources |

THE CONTRIBUTION OF THE ASSOCIATIVE APPROACH IN THE PREVENTION AND TREATMENT OF SEVERE GENODERMATOSES

M.R. Kamoun
Tunisian Society of Dermatology
PanArab League of Dermatology Representative

General contribution of the associative approach

- ◇ **Community-based**
 - ✗ better appreciation of the issues at hand, better capacity for appropriate intervention
- ◇ **Flexibility**
 - ✗ considerable capacity for adaptation
- ◇ **Motivation**
 - ✗ members of associations have freely chosen to participate
- ◇ **Force**
 - ✗ pooling of capacities

Specific contribution for genodermatoses

- ◇ **Mobilising the necessary skills and expertise**
 - ✗ health professionals, educators, communicators, researchers, social workers, politicians
- ◇ **Building solidarity networks**
 - ✗ between governmental structures and community organisations
 - ✗ between the various disciplines
 - ✗ between administering physicians and researchers
 - ✗ between the northern and southern shores of the Mediterranean: complementarity

CREATING A PATIENTS' ASSOCIATION IN THE FIELD OF DERMATOLOGY

H. Benchikhi
Dermatology Dept., CHU Ibn Rochd, Casablanca, Morocco
Secretary-General, Gildi, an association offering support to persons suffering from dermatological diseases

- ◇ **Find a leader**
- ◇ **Build a plan**
- ◇ **Locate sources of private (and not public) funding (e.g., banks)**

ORPHANET

A. Mégarbané

Director, 'Unité de Génétique Médicale', St Joseph University, Lebanon
Head, 'Pôle d'excellence régional en génétique médicale'

on behalf of

S. Aymé

Director, Inserm SC11, France

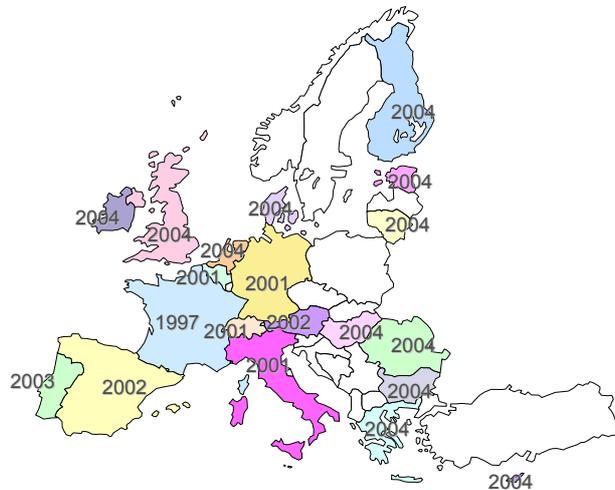


↳ Created in 1997

↳ To address identified problems...

- ✗ lack of information → encyclopedia of rare diseases
- ✗ scarce expertise → directory of experts
- ✗ too few collaborations → directory of research projects
- ✗ difficult recruitment → on-line service to register as a volunteer
- directory of clinical trials
- ✗ lack of partnerships → OrphanXchange

↳ In 20 countries



◇ **12 000 daily users**

- ✗ patients: 33,3 %
- ✗ health professionals: 51,6 %
- ✗ other: 15,1 %

◇ **Current content of the database**

- ✗ 3 713 diseases and synonyms
- ✗ 2 463 abstracts
- ✗ 624 review articles
- ✗ 751 diagnostic labs about 943 diseases
- ✗ 1 952 research projects about 1 154 diseases
- ✗ 858 patients organizations linked to 1 431 diseases
- ✗ 1 312 specialized clinics
- ✗ 4 832 professionals
- ✗ 4 379 other web pages

◇ **Conclusion**

- ✗ + 8% per month of visitors since the beginning
- ✗ serving all types of public
- ✗ real customers + high customer satisfaction
- ✗ funding: Inserm + French Ministry of Health + European Commission
- ✗ the most comprehensive set of review articles in the world
- ✗ the directory of services has to be expanded to reflect the real situation in all European countries

Extension of the network

◇ **Principles**

- ✗ any European country if the resources are identified and if a local professional is willing to run the project

◇ **New partner countries**

- ✗ Lebanon + Morocco + Tunisia
- ✗ grant from AUF

◇ **New candidates**

- ✗ Czech Republic, Slovakia, Sweden
- ✗ contacts are starting with Poland and Turkey

◇ **Orphanet in Mediterranean countries**

- ✗ Orphanet accepts new partners immediately if
 - they have the resources to collect data
 - they accept to work with our procedures
- ✗ new applications for funding next year

Orphanet Newsletter

◇ **Monthly electronic Newsletter in English**

- ◇ **Content: Scientific and Political news + Events + Books**

- ◇ Funding from European Commission
- ◇ Rare Disease Task Force
- ◇ To receive it, please send your information to sgoodman@orpha.net

Services to boost R&D

- ◇ Directory of research projects in Europe
- ◇ A tool to better inform patients about clinical trials: Eclor
- ◇ A tool to boost the development of diagnostic tools and therapeutic products: OrphanXchange

CENTER FOR ARAB GENOMIC STUDIES

A. Mégarbané

Director, 'Unité de Génétique Médicale', St Joseph University, Lebanon

Head, 'Pôle d'excellence régional en génétique médicale'

- ◇ Sheikh Hamdan Bin Rashid Al Maktoum Award for Medical Sciences
- ◇ www.cags.org.ae
- ◇ Inaugurated in June 2003
- ◇ Educate the public and professionals on
 - ✗ the important impact of genetic diseases in the Arab World
 - ✗ the methods and benefits of early genetic diagnosis
- ◇ Goals
 - ✗ to provide comprehensive genetic services by translating research achievements into well-integrated patient treatment programs
 - ✗ to address the ethical, legal and social issues that may arise with the implementation of such programs

ALGERIA

Bakar Bouadjar
CHU Bab El Oued, Algiers
Ismail Benkaïdali
CHU Mustapha, Algiers
Mourad Redjimi

CURRENT SITUATION

General information



◇ Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

total population	31 266 000
GDP per capita (Intl \$, 2001)	4 104
total health expenditure per capita (Intl \$, 2001)	169
total health expenditure as % of GDP (2001)	4,1

✗ consanguinity: 25-30%

◇ Health care institutions by type

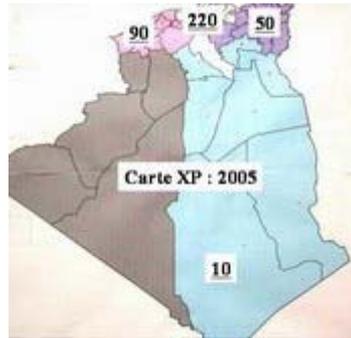
- ✗ 13 university hospitals
- ✗ 32 specialised hospitals (Etablissement Hospitaliers Spécialisés, EHS)
- ✗ 220 hospitals
- ✗ 185 “secteurs sanitaires” (regional health agencies)
- ✗ 487 polyclinics
- ✗ 1,243 health centres

◇ Dermatology

- ✗ 2 departments in Algiers
- ✗ 1 department in Tlemcen
- ✗ 1 department in Oran
- ✗ 1 department in Constantine
- ✗ 1 department in Anaba

◇ More than 30 genodermatoses are prevalent, of which 3 are particularly severe

✗ xeroderma pigmentosum, with 370 cases in 90 families in 2005



✗ recessive ichthyoses, with 150 cases in 80 families, Meleda syndrome

✗ hereditary epidermolysis bullosa, among 60 families

✗ other genodermatoses encountered (not an exhaustive list):

epidermodysplasia verruciformis, Kindler syndrome, familial psoriasis in 70 families, Papillon-Lefèvre syndrome, vitiligo in 20 families, tuberous sclerosis (Bourneville disease) in 10 families (28 cases), Chanarin-Dorfman syndrome

Care

◇ Diagnosis

✗ 2 anatomic pathology laboratories (pathologists specialising in dermatology need to be trained and electron microscopy needs to be developed further)

✗ 2 biological laboratories (equipment for the biochemical diagnosis of metabolic diseases needs to be installed)

✗ 4 molecular biology units including 3 in Algiers (DNA extraction / explorations for certain pathologies); human resources for cytogenetics and lymphocyte culturing need to be supplemented

✗ 4 DNA banks

◇ Consultation services

✗ 1 genodermatosis consulting service in each of Algiers' 2 dermatology departments, at Mustapha University Hospital and Bab El Oued University Hospital

◇ Hospital care

✗ hospitalisation depending on the number of beds available (125 in Algiers)

✗ patients are kept under observation and checkups are performed

✗ blood samples are collected from families in order to conduct genetic analyses

✗ treatments: retinoids, emollients, sunscreens

✗ treatments involving plastic surgery (XP), ophthalmology, neurology and paediatrics are handled on a case-by-case basis

✗ psychological treatment is ensured by the department's psychologists

◇ Extra-hospital care

✗ patients are advised to consult private or public health service dermatologists in their area (care often haphazard)

✗ patients are advised to consult physicians having taken the special genetics training course (CES), a recently introduced resource in need of development

✗ it is difficult to check on these patients' progress regularly due to their distance from referring centres and the financial situations of the families

◇ **Medicines**

- ✗ insufficient, particular the retinoids due to their cost, €3/day for a box of 30 capsules = ½ SMIG (legally guaranteed minimum wage)
- ✗ not reimbursed by social security, since genodermatoses are not classified as chronic diseases

◇ **Transportation**

- ✗ not reimbursed

Diseases presenting specific difficulties for care

◇ **Xeroderma pigmentosum**

- ✗ patients are from low-income families and live in rural areas
- ✗ they arrive for consultation when in third stage, with the appearance of numerous malignancies
- ✗ medicines (retinoids, sunscreens) are not widely available

◇ **Recessive ichthyoses**

- ✗ few units specialise in neonatology
- ✗ medicines are very expensive

Assistance for families

◇ **Patients' associations**

- ✗ 2 attempts
 - 1 support group for xeroderma pigmentosum lasted one year
 - 1 support group for psoriasis
- ✗ patients' associations may be long-lasting if endorsed by a health professional

Education and training

◇ **Instruction in genetics**

- ✗ during the 3rd year of medical school

◇ **A two-month course focusing on genodermatoses**

- ✗ during the 2nd year of the dermatology specialisation

◇ **A year-long course in genetics (CES)**

- ✗ objective: setting up a vigilance network for the treatment of genetic diseases
- ✗ since 2004
- ✗ 40 physicians either trained or in training (20 in 2004, 20 in 2005)
- ✗ dermatology is one of the specialisations covered

Information

◇ **General press**

- ✗ articles on genetic diseases published on the occasion of scientific and medical conferences treating genetic diseases

◇ **Medical press**

✗ 3 or 4 national journals

◇ **Television**

✗ information campaigns on vaccination, prevention of road accidents, etc

Research

◇ **On the national level**

✗ 1995: research project with the Ministry of Health, *XP in Algeria: a clinical and genetic study* (mapping, clinical classification, establishment of a multidisciplinary team in Algiers, genetic study)

✗ 1998: research project by the Agence Nationale pour le Développement de la Recherche en Santé (ANDRS), *Epidermodysplasia verruciformis in Algeria: a clinical, genetic and therapeutic study*

✗ 2000: thesis on *Tuberous sclerosis (Bourneville disease)*

✗ 2004: Inserm project, *Rare diseases*

✗ 2005: research project with the Ministry of Health (ongoing), *Hereditary epidermolysis bullosa*

◇ **On the international level**

Collaborations with

✗ Centre National de Genotypage (CNG) and Généthon

✗ Human papillomavirus unit, Institut Pasteur, Paris

✗ IRSC-CNRS, Villejuif

Recent accomplishments

◇ **Establishment of 2 consulting services specialising in genodermatoses**

✗ consultations offered once a week

✗ 1 in Mustapha University Hospital

✗ 1 in Bab El Oued University Hospital

◇ **Observation of recessive ichthyoses to arrive at a genotype-phenotype correlation**

◇ **Participation in the implementation of the vigilance network**

◇ **Contact achieved with the Ministry of Health**

PROPOSALS

Creating a national committee for genodermatoses
in collaboration with the Comité Médical National de Génétique (CMNG)

⇒ **Promote multidisciplinary consultation services in the university hospitals**

⇒ **Implementation of a vigilance network**

⇒ on the model of that developed by the CMNG, the Algerian human genetics commission

⇒ in the 13 university hospitals, 32 EHSs, 220 hospitals, 185 'secteur sanitaires' (health districts), 487 polyclinics and 1,243 health centres

⇒ establish 4 or 5 pilot centres

⇒ **Facilitate access to medicines**

⇒ by building awareness among government authorities (Ministries of Health, Social Affairs and

Solidarity)

- ⇒ thanks to the introduction of contractualisation as one of several hospital reforms
 - from 01/01/2006
 - collaboration with Ministries of Health and Social Affairs for the care and management of disadvantaged patients
- ⇒ by requesting the recognition of genodermatoses as chronic diseases (psoriasis and eczema are already on the list of chronic diseases)
- ⇒ by encouraging donations by pharmaceutical firms
- ⇒ by establishing an international distribution facility for medicines

↵ **Promote and endorse patients' associations**

↵ **Initiate training programmes**

- ⇒ in the different university hospitals
- ⇒ in close collaboration with the CMNG
- ⇒ decentralise this training (physicians, nurses, communication professionals, patients) in order to raise awareness, particularly among non-specialists (general practitioners)

↵ **Communicate to government officials and politicians**

- ⇒ via the legislative assemblies

↵ **Communicate to professionals**

- ⇒ via Orphanet and the specialised press

↵ **Communicate to the general population**

- ⇒ via religious conferences: collaboration between physicians and the Ministry of Religious Affairs

↵ **Establish a national registry for genodermatoses**

- ⇒ co-ordinated by the national dermatology and paediatric dermatology associations

↵ **Evaluate care**

↵ **Confirm diagnoses**

↵ **Establish additional DNA banks (there are 4 banks at present)**

↵ **Establish guidelines for prenatal diagnosis focusing on severe or debilitating genodermatoses**

↵ **Implement and evaluate new therapeutic tools and methods**

↵ **Encourage sociological research**

↵ **Foster international partnerships**

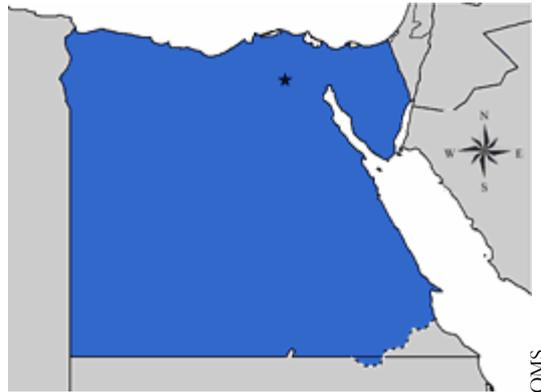
- ⇒ for diagnosis (tools, training of qualified staff)
- ⇒ for care (medicines)
- ⇒ for research (in genetics and pharmacology, equipment for genetics laboratories)
- ⇒ exchange of treatment experiences
- ⇒ for the training of nurses
- ⇒ to obtain emollients and sunscreens

EGYPT

Emad El Gamal
Vice-president, Alexandria Society of Dermatology

CURRENT SITUATION

General information



◇ Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

total population	70 507 000
GDP per capita (Intl \$, 2001)	3 901
total health expenditure per capita (Intl \$, 2001)	153
total health expenditure as % of GDP (2001)	3,9

◇ Intermarrying is very common

◇ Statistics available in every university

◇ Many initiatives to improve the management of genodermatoses

Care

◇ Diagnosis

✗ genetic counselling and antenatal diagnosis available in a few places all around the country

◇ Consultations

✗ a few health care centers for genodermatoses

Assistance for families

◇ Genetic Counselling Society in Alexandria

Recent accomplishments

The top priority: the medical and social management

◇ Target: the most common genodermatoses

◇ Information and partnerships

✕ health authorities

- head of the dermatology department in the Ministry of Health
- under-secretary of Health in Alexandria
- authorities for health in schools

✕ social affairs authority

- under-secretary for Social Affairs

✕ universities

✕ scientific societies

- Egyptian Society of Dermatology and Venereology (ESDV)
- Alexandria Society of Dermatology Venereology and Andrology (ASDVA)

✕ NGOs

- Egyptian NGOS Network against AIDS (ENNA)
- Regional NGOS Network against AIDS (RNNA)
- Family Health International (FHI)
- Caritas

✕ International Organizations

- UNICEF
- UNDP

◇ Set up a support association

✕ contact in Cairo

PROPOSALS

The top priority: the medical and social support

↵ A pilot project in Alexandria

⇒ multidisciplinary hospitals: dermatologists and specialists can be easily reached out

↵ A project in Cairo

↵ Set up a national programme of surveillance for 5 genodermatoses

⇒ together with NGOs, the Ministry of Health, Universities

↵ Raise funds with the support of

⇒ national and international organizations

⇒ NGOs

⇒ pharmaceutical companies

↵ Improve training and education for a better diagnosis

⇒ to train MDs, paramedics and social workers

↵ Support patients to set up association

⇒ in Cairo

- ↪ **Improve genetic counseling**
- ↪ **Develop research on severe genodermatoses**
- ↪ **Include talks about genodermatoses at dermatology and pediatrics conferences (one-day symposium)**
- ↪ **Set up an information network for the patients and their family**
- ↪ **Use 2 national networks**
 - ⇒ the network against leprosy (Ministry of Health and Caritas) reaches out every patient in all villages and gives lectures for paramedics and social worker
 - ⇒ Egyptian NGOS Network against AIDS (ENNA)
- ↪ **Include lectures about genodermatoses in the ones given on Leprosy in Upper Egypt**
 - ⇒ together with UNICEF, Caritas and the Ministry of Health
- ↪ **Coordinate the many initiatives through a national network**

FRANCE

Dr. Smail Hadj-Rabia

Prof. Christine Bodemer

Dermatology Dept. (Prof. Y. de Prost), Hôpital Necker-Enfants Malades, Paris

CURRENT SITUATION

General information



◇ Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

total population	59 850 000
GDP per capita (Intl \$, 2001)	26 809
total health expenditure per capita (Intl \$, 2001)	2 567
total health expenditure as % of GDP (2001)	9,6

◇ Rare diseases project

- ✗ national project in support of rare diseases involving accreditation of centres of excellence in the treatment of rare diseases including genodermatoses
- ✗ official recognition of these diseases, their prevalence and their requirements for treatment and care
- ✗ potential for greater assistance for research

◇ Centres of excellence: definitions and missions

- ✗ structure care delivery around accredited centres of excellence
 - medical and scientific expertise
 - organisation of care delivery in collaboration with regional or inter-regional structures (centres of expertise)
- ✗ definitions
 - centre of excellence: centre specialising in the treatment of a disease or group of diseases
 - secondary centre: centre of excellence located beyond the local hospital's population area: inter-regional, national, international
- ✗ missions
 - establish or confirm diagnoses
 - definition and diffusion of therapeutic and/or care delivery organisation protocols
 - provide information and training for health professionals, patients and their families, through inter-hospital and hospital-physician networks
 - research and epidemiological vigilance
 - creation and co-ordination of networks (health care-oriented and medico-social), supporting the development of regional centres of expertise enabling community-based

monitoring of patients, in close co-operation with the centre(s) of excellence

⇨ **Centres of excellence, leading to**

- ✗ the implementation of care delivery schemes that are comprehensible for the patients and their families, guiding patients throughout the cycle of care (respecting the patient's freedom of choice)
- ✗ the definition and the implementation of guidelines and recommendations for clinical practice in association with the national health authority, the diffusion of these documents, contributing to improvements in professional practice
- ✗ optimisation of initial prescriptions for orphan drugs
- ✗ a national network of expertise to liaise with the health insurance system (registry of patients suffering from long-term diseases, etc.) and other mechanisms of reimbursement

Optimal care

⇨ **Diagnosis**

- ✗ links between clinics and fundamental research (quality of clinical expertise and diagnosis, research laboratories within hospitals with national and international collaboration)

⇨ **Hospital care**

- ✗ centres specialising in clinical care for children from birth (specificity of care depending on age, multidisciplinary paediatric environment)
- ✗ continuity of care for adults with day hospitals and consultation services

⇨ **Extra-hospital care**

- ✗ adequately trained caregivers at the local level (community physicians and care-giving teams)

⇨ **Prevention**

- ✗ comprehensive care for families including offers of genetic counselling, prenatal diagnosis, pre-implant diagnosis in accredited centres

⇨ **Effective patients' associations with high visibility**

Research

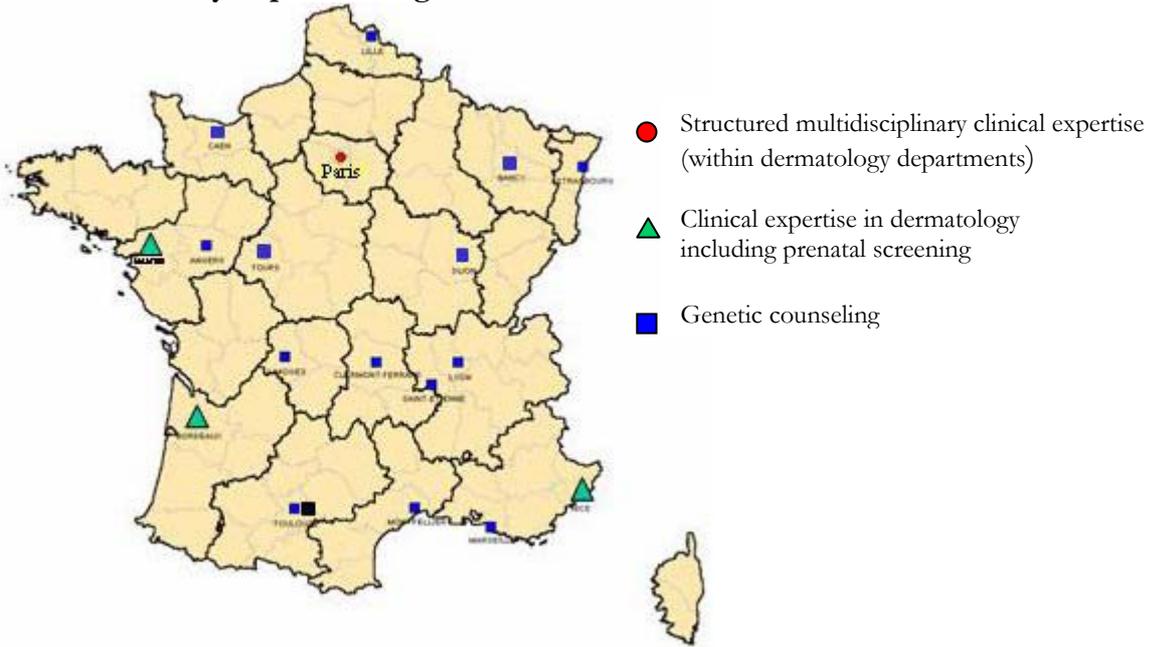
- ⇨ **Active and productive research (molecular, fundamental, therapeutic)**

Reality and difficulties

⇨ **National clinical activities**

- ✗ few centres of expertise
 - clinical expertise, multidisciplinary care
 - obstetrics departments
 - accredited laboratories for prenatal and pre-implant diagnosis
- ✗ community-based care networks
- ✗ insufficient early detection in clinics (lack of education)
- ✗ funding for clinical research (epidemiology)

⇨ Clinical activity in paediatric genodermatoses

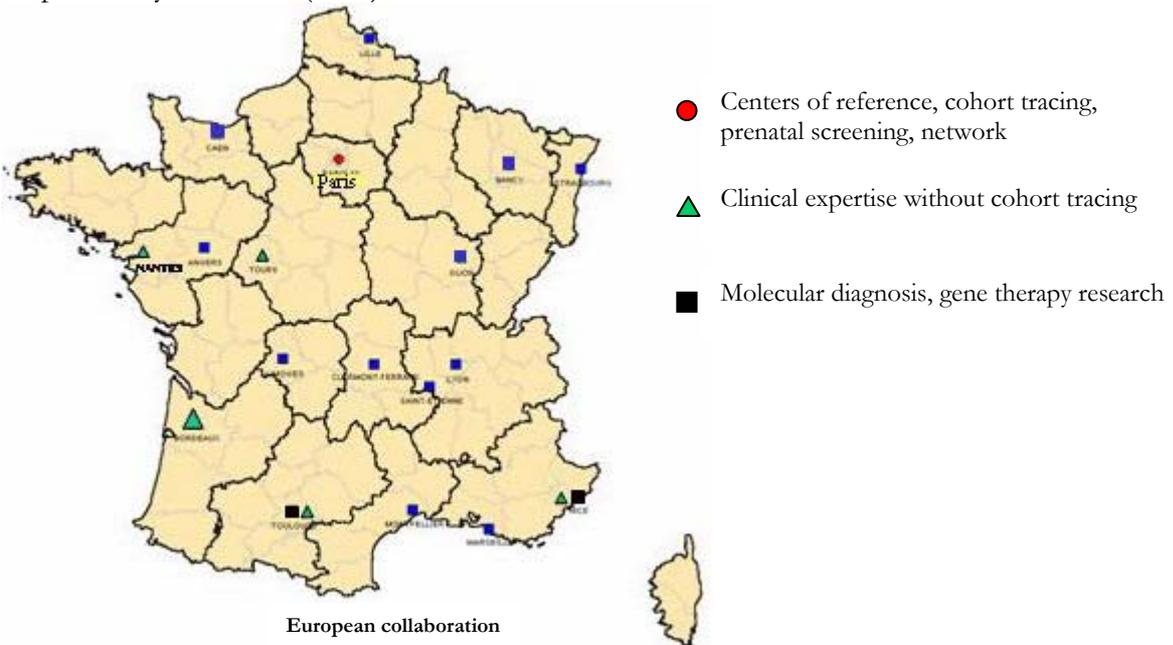


⇨ National research activities

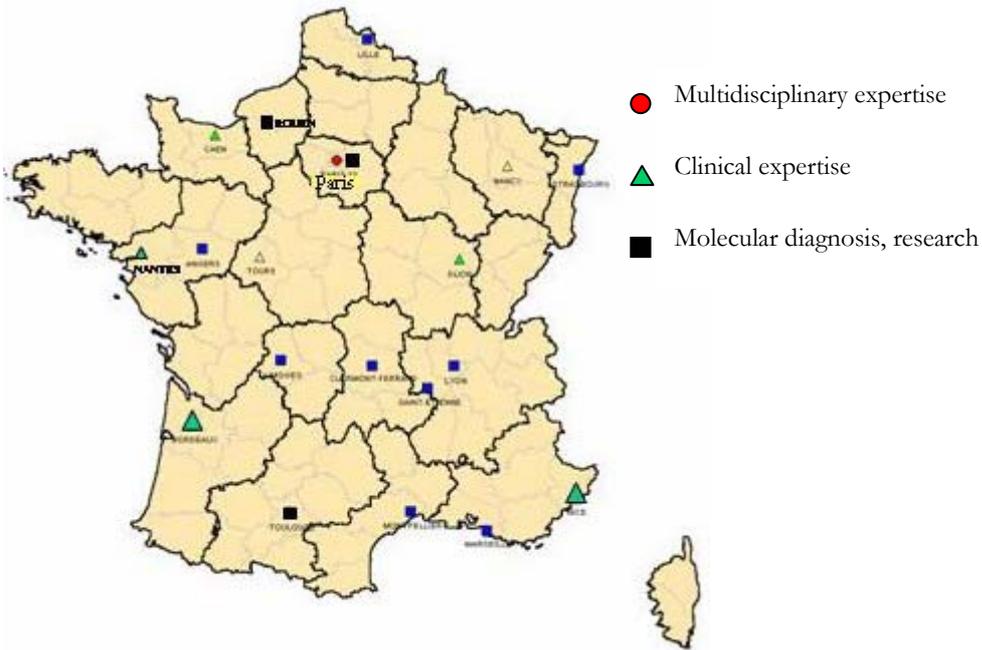
- ✗ need for greater comprehensibility
- ✗ need for a registry of research activities
 - emerging teams
 - synergy and emulation
- ✗ need for better funding
 - programmes
 - status of researchers
- ✗ insufficiency of early detection in clinics due to lack of information

⇨ National clinical and research activities

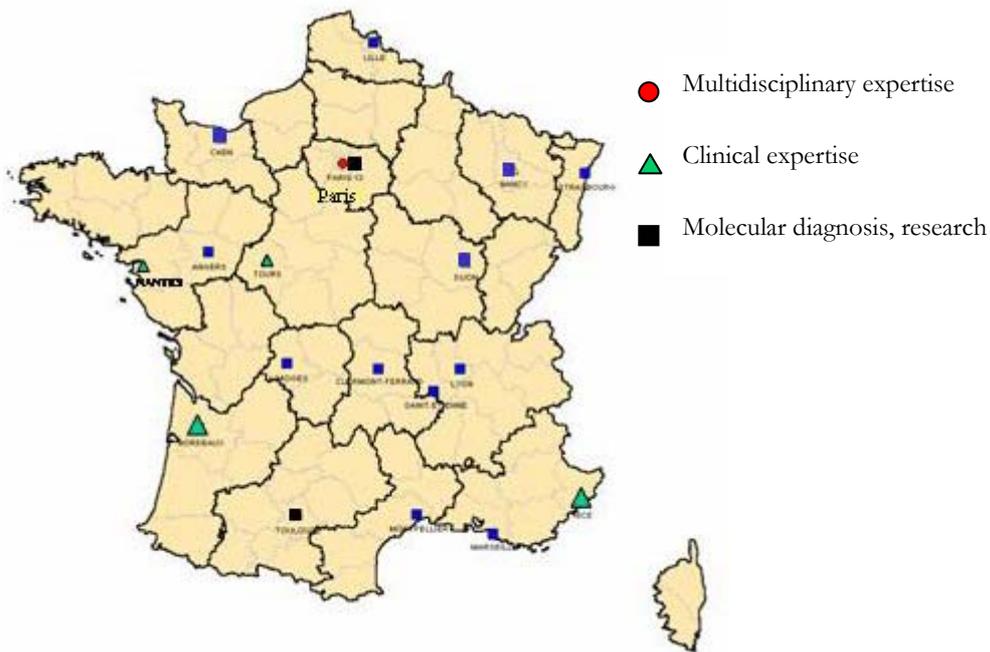
- ✗ epidermolysis bullosa (2005)



X ectodermic dysplasia (2005)



X ichthyoses, palmoplantar keratodermas



Overview of work carried out in 2004

34 official “rare disease” centres in France

◇ 1 type 1 neurofibromatosis centre

◇ 1 centre for genetic skin diseases

◇ multi-site centre:

- paediatric genodermatoses Hôpital Necker (C. Bodemer)
- adult genodermatoses I Hôpital St Louis (C. Blanchet-Bardon, N. Basset-Seguin)
- adult genodermatoses II Hôpital Avicenne (F. Caux)

Work in progress

- ⇒ **Registry of national genodermatosis activity**
- ⇒ **Work group**
 - ✗ a medical representative for each region of France (letters sent to all hospital department heads)
 - ✗ objective: develop a care network
- ⇒ **Co-ordinating secretariat at Necker Hospital in Paris**
 - ✗ centralises information (care networks, informational meetings, etc.)
 - ✗ relations with health networks, families, associations
- ⇒ **Medical treatment guidelines / protocols**
- ⇒ **Creation of databases for epidemiological studies**

PROPOSALS

- ⇒ **Working with patients' associations**
- ⇒ **Reinforcing community-based care networks**
- ⇒ **Improving social protection, specialised care, orphan drugs**
- ⇒ **Offering specific training courses designed for obstetricians, midwives, paediatricians, etc.**
- ⇒ **Providing logistical support and reinforcing clinical and fundamental research programmes**

GREECE

N. G. Stavrianeas
President of the Greek Society of Pediatric Dermatology
Athens University, Chairman 2nd Dermatological Clinic
A. Katsarou
Athens University, 1st Dermatological Clinic

CURRENT SITUATION

General information



Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

total population	10 970 000
GDP per capita (Intl \$, 2001)	16 247
total health expenditure per capita (Intl \$, 2001)	1 522
total health expenditure as % of GDP (2001)	9,4

2004: A. Sygros Hospital & General University Hospital Attikon (University of Athens)

total number of patients examined: 98 537

43 new genodermatoses cases

- 9 patients with Keratoderma Palmo-Plantaris
- 8 with Neurofibromatosis
- 7 with Ichthyosis
- 6 with Darrier's Disease
- 5 with Pseudoxanthoma Elasticum
- 2 with Ehlers-Danlos syndrome
- 1 with Hailey-Hailey, 1 with Erythrokeratoderma variabilis, 1 with Tuberous Sclerosis

Care

National Reference Center for Genodermatoses founded in 2004

at the 1st and 2nd Dermatology Departments in Athens University

Outpatients clinics for Pediatric Dermatology began in January 2005

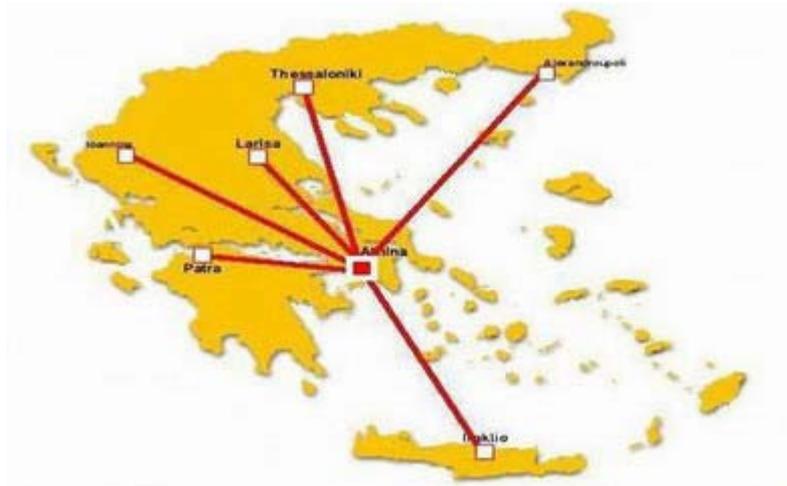
at the 1st and 2nd Dermatology Departments in Athens University

Medical staff

principal: dermatologists, histopathologists, psychologists, nurses

✕ consultants: paediatricians, geneticists, neurologists, cardiologists, others

⇨ Network of Dermatology Departments



■ Reference Center for Genodermatoses (University of Athens)

□ Existing Dermatology Departments in Medical Schools

PROPOSALS

Strengthen the national dermatological network



■ Reference Center for Genodermatoses (University of Athens)

□ Existing Dermatology Departments in Medical Schools

○ Future Clinics of Dermatology

⇨ Take Cypriot dermatologists on as partner

IRAN

H. Moslehi
Razi Hospital, Teheran University

CURRENT SITUATION

General information



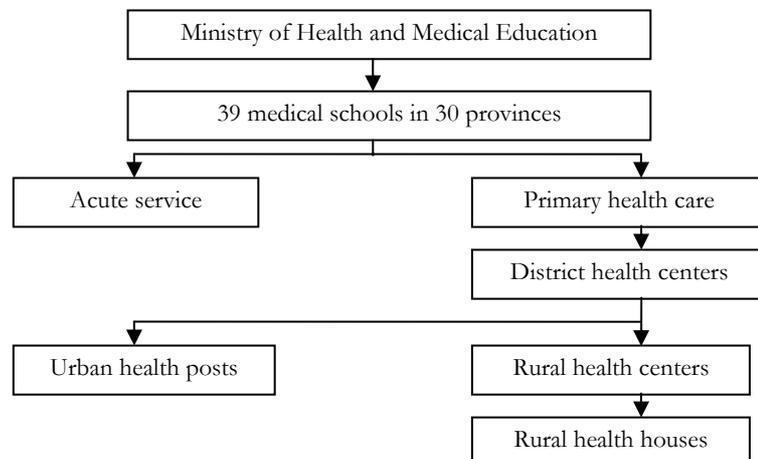
Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

✗ total population	68 070 000
✗ GDP per capita (Intl \$, 2001)	6 673
✗ total health expenditure per capita (Intl \$, 2001)	422
✗ total health expenditure as % of GDP (2001)	6,3
✗ 1 600 000 sq. km.	
✗ density: 30/sq. km (5 to 145/sq. km.)	
✗ 50% of rural population (intermarrying rate reaches 30%)	

2 health care systems coexist: public and private

- ✗ the public system is under the supervision of the Ministry of Health
 - consisting of 39 Universities of Medical Sciences in 30 provinces
 - this system encompasses primary care, medical services in university hospitals and clinics, free hospitals and clinics for the disadvantaged and research



- ✗ the private system consists of hospitals, clinics and laboratories

⇒ Dermatology

- ✗ 600 dermatologists (for the most part in private practice)
- ✗ 11 dermatology departments (training of interns)
- ✗ 29 provinces with dermatology clinics connected with universities
- ✗ 3 dermatology research centres
- ✗ Centre for Skin Diseases at Razi Hospital, University of Tehran
 - 300 to 350 patients per day on an out-patient basis
 - 1 consultation service for autoimmune bullous dermatoses since 1975 (3,000 cases of pemphigus)
 - 1 consultation service for genetic diseases from 1999 to 2001

Care

⇒ Organisation of the primary care network in Iran

- ✗ 99% of the population covered by the primary care network
 - with 15,000 “health homes” in rural areas
- ✗ primordial role
 - in the prevention of non-transmissible diseases
 - in the reduction of maternal and infant mortality and the birth rate
- ✗ staff ensure
 - the annual census of the population
 - health education
 - family health (care before and after childbirth, infant health, family planning, vaccination)
 - fight against tuberculosis, malaria, leprosy, etc.
 - basic treatments
 - public hygiene
 - collection and processing of health information
- ✗ programme for the prevention of non-transmissible diseases, an adjunct to the primary care programme in 1991

⇒ A consultation service for genetic skin diseases at Razi Hospital

- ✗ from May 1999 to April 2001 / once per week (maximum of 4 patients)
- ✗ 43 cases
 - 20 patients suffering from ichthyoses
 - 12 cases of neurofibromatosis
 - 5 cases of epidermolysis bullosa
 - 5 cases of xeroderma pigmentosum
- ✗ a positive experience
 - relatively high number of cases encountered
 - possibility of collecting information on these cases
- ✗ limits
 - dissatisfaction of patients who leave without treatment
 - lack of trained personnel
 - lack of useful collaborative arrangements
 - lack of financial assistance for tests, medicines, health insurance coverage, transportation

Education

⇒ Continuing education of primary caregivers

- ✗ when a new programme is introduced, caregivers in the 30 provinces attend a initial meeting at the ministry where the objectives of the programme, the strategy implemented and the activities to be developed are presented
- ✗ this meeting is followed by several workshops
- ✗ health personnel in the provinces then communicate this knowledge to the level immediately under their own, and the information is then relayed to lower levels
- ✗ information collected while implementing the programme is submitted following the same channels from the rural health homes up to the ministry

Research

⇒ Genetic research centres in Iran

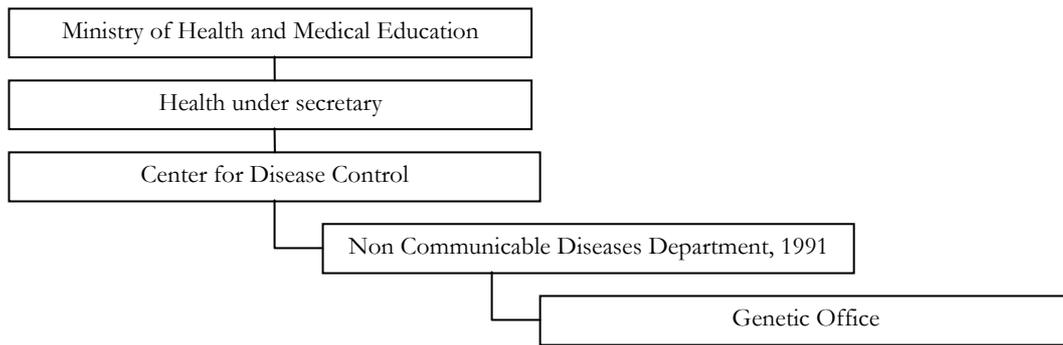
- ✗ National Research Centre for Genetic Engineering and Biotechnology: www.nrcgeb.ac.ir
- ✗ The Genetics Research Centre at the University of Social Welfare and Rehabilitation Sciences: www.uszr.ac.ir/grc
- ✗ Medical genetics units

Prevention

⇒ The national programme for the early detection of thalassemia as model for the prevention of genetic diseases

- ✗ β thalassemia
 - a public health problem in Iran (15,000 known cases, 20,000 estimated, 1,200 new cases each year)
 - model disease for the evaluation of the feasibility of the programme for the prevention of non-transmissible diseases at the primary care level
- ✗ pilot study
 - from 1991 to 1996
 - in 5 provinces
- ✗ 1997: launch of the national programme to combat thalassemia
- ✗ objectives of the programme
 - prevention of new cases
 - reduction of morbidity and mortality
 - lower treatment costs
 - prevention of side effects: impact on the family, mental health, etc.
 - training of medical personnel, students, soldiers, official registrars
 - education of the general public: posters, information provided to universities, military camps, etc.
- ✗ a success
 - model for the building of an infrastructure for the genetic disease prevention programme
- ✗ 2001
 - 2,700,000 couples underwent screening tests (10,298 couples found to be at risk)
 - all the couples at risk (10,298) received genetic counselling
 - 30% decrease in affected births
 - cost savings of USD 20 million

⇨ Organisation of the office for the prevention of genetic diseases



⇨ Actions implemented by the office for the prevention of genetic diseases in the fight against thalassemia

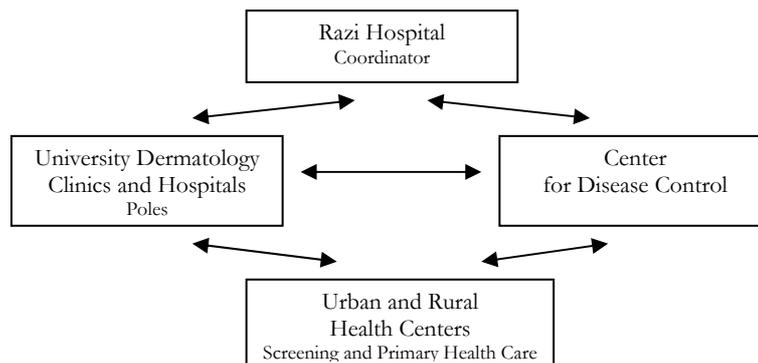
- ✗ establishment of infrastructures for early detection, genetic counselling and prenatal diagnosis with a view to future programmes (hemophilia, phenylketonuria, glucose-6-phosphate dehydrogenase deficiency, congenital hypothyroidism)
- ✗ passage into law of a bill addressing thalassemia in 1997
- ✗ therapeutic abortions made possible in 1998 in the event of severe abnormalities in the foetus

⇨ Objectives of the office for the prevention of genetic diseases

- ✗ use the programme for the prevention of thalassemia as a model for care and management
- ✗ use the prevention network established for future programmes
- ✗ include the prevention of non-transmissible diseases in the primary care network

Structure proposed for the project

- ✗ 1 co-ordinating centre: Skin Disease Centre at Razi Hospital
- ✗ treatment centres associating consultation services and university hospitals
- ✗ rural and urban health centres for screening and primary care



Recent actions

⇨ Partnerships

- ✗ with the Ministry of Health (Centre for Disease Control)
- ✗ with the Iranian dermatology association
- ✗ with university dermatology departments
- ✗ with the medical genetics department of the University of Teheran

PROPOSALS

A model: the national programme for the early detection of thalassemia

- ↪ **Take inspiration from the national programme for the early detection of thalassemia**
- ↪ **Official recognition of genodermatoses by the Ministry of Health, insurance and social welfare systems, non-governmental organisations**
- ↪ **Create centres and consultation services for genodermatoses**
 - ⇒ encourage epidemiological research on genodermatoses, compile a registry of the cases of genodermatosis in each centre (consultation services and university hospitals) and centralise this information
 - ⇒ define standard registry procedures in accordance with the International Dermatological Disease Classification
 - ⇒ specify prevalence and evaluate the disability entailed by each form of genodermatosis
- ↪ **Target main genodermatoses**
- ↪ **Include the selected genodermatoses in the project developed by the Ministry of Health for the prevention of genetic diseases**
- ↪ **Reinforce ties between primary health centres and highly specialised health services**
- ↪ **Train physicians and other caregivers**
- ↪ **Develop social protection solutions**
- ↪ **Seek new partnerships**
 - ⇒ paediatrics and genetics units in universities
 - ⇒ paediatrics and genetics associations
 - ⇒ social structures
 - ⇒ governmental and non-governmental structures for “special” diseases
 - ⇒ charitable organisations
- ↪ **Use a single language**
 - ⇒ for classification, exchanges of information, etc.
- ↪ **Exchange experiences**
 - ⇒ statistics, level of disability entailed by the disease, projects
- ↪ **Improve access to diagnostic and medical information**

IRAQ

Nuha T. Kusaimi
Department of Medicine, University of Mosul

CURRENT SITUATION

*Due to the current situation,
it's very difficult to get an overview of the health organization*

General information



◇ Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

total population	24 510 000
GDP per capita (Intl \$, 2001)	2 947
total health expenditure per capita (Intl \$, 2001)	97
total health expenditure as % of GDP (2001)	3,2

◇ Mosul

- ✗ 2nd largest city in Iraq
- ✗ main hospital for dermatology (North of Iraq)

The management of severe genodermatoses in Mosul

◇ Organization of the dermatology consultations

- ✗ morning: outpatient clinic at the Republican Hospital (teaching hospital)
- ✗ afternoon: private clinic

◇ Diagnosis

- ✗ history
- ✗ clinical examination

◇ A few drugs available in hospitals or pharmacies

◇ **Follow-up**

- ✗ depending on security and financial conditions
- ✗ depending on the hope for a cure

◇ **Information**

- ✗ MDs refer patients with severe genodermatoses to Mosul Hospital

◇ **Research**

- ✗ Ichthyosis, Acrodermatitis, Darier disease, Acanthosis Nigricans
- ✗ many data have been lost

PROPOSALS

⇒ **Improve the access to drugs and medical devices**

⇒ **Make lectures and organize training courses on genodermatoses**

⇒ **Build up partnerships with the health authorities to identify genodermatoses cases in villages**

⇒ **Promote grants for MDs**

⇒ **Develop research on genodermatoses**

⇒ **Set up support association**

⇒ build up partnership with a Jordanian support association

⇒ **Inform and heighten public awareness of the consequences of intermarrying**

ITALY

G. Zambruno
IDI-IRCCS, Rome
M. El-Hachem
OPBG, Rome

CURRENT SITUATION

General information



Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

total population	57 482 000
GDP per capita (Intl \$, 2001)	26 169
total health expenditure per capita (Intl \$, 2001)	2 204
total health expenditure as % of GDP (2001)	8,4

The Italian law on rare diseases¹ (2001)

- ✗ creation of a national network of regional centers and interregional reference centers for diagnosis and management of rare diseases
- ✗ regional centers are in charge to:
 - perform diagnosis
 - assure patient proper management and follow-up, also through a collaboration with GPs
 - keep patient's records and communicate all new disease cases to reference centers
- ✗ interregional reference centers are in charge to:
 - coordinate the activities of regional centers
 - insure application of diagnostic and therapeutic protocols/guidelines, whenever available
 - give support to GPs about rare diseases and available drugs
 - participate in training of health care personnel and in disease prevention
 - disseminate information and raise public awareness about rare diseases
 - keep contact with patients' associations
- ✗ all regions (except for 3) have identified regional centers for rare diseases
- ✗ the National Health system covers
 - all costs related to examinations required to establish the genetic disease diagnosis
 - costs of laboratory examinations
 - costs of specialists' consultations
- ✗ costs of drugs, medical devices and, in some regions, emollients, antiseptics, supplements, sunscreens, bandages, etc.

¹ Rare disease definition: prevalence below (according to EU criteria)

✗ some genodermatoses are not included in the Italian rare disease official list

⇒ **Istituto Superiore di Sanita and rare disease**

✗ coordination of epidemiological, clinical and research activities on rare diseases

✗ creation and updating of a national registry of rare diseases

- in most cases information is not yet transmitted from regional centers

✗ establishment of guidelines for rare diseases diagnosis and management

- not yet active

✗ coordination and granting of research projects in the fields of epidemiology, disease prevention, molecular diagnosis,

- some projects on epidemiology and genetic testing quality control activated

⇒ **Agenzia Italiana del Farmaco**

✗ promotion of investments in research and development

✗ to guarantee, through the simplification of drug registration procedures, a rapid access to innovative medicinal products and orphan drugs for rare diseases

⇒ **A national register for inherited Epidermolysis Bullosa**

✗ set up by G. Tadini, Centro Malattie Ereditarie presso l'Istituto di Scienze Dermatologiche, Univ. Milano

✗ 705 inherited Epidermolysis Bullosa

Care

⇒ **Different diagnostic approaches according to the disease:**

- **Epidermolysis bullosa:** clinical findings, personal and family history → immunofluorescence antigen mapping and electron microscopy → molecular and prenatal diagnosis possible in all forms
- **Ichthyoses:** clinical findings, personal and family history → histopathology (immunohistochemistry and/or electron microscopy in selected forms) → molecular and prenatal diagnosis possible in most forms
- **Xeroderma Pigmentosum:** clinical findings, personal and family history → complementation group → molecular and prenatal diagnosis possible in all forms
- **Palmoplantar keratodermas:** clinical findings, personal and family history → histopathology → molecular diagnosis possible in the majority of forms
- **Neurofibromatosis:** clinical findings, personal and family history → instrumental examinations → molecular diagnosis possible

⇒ **Italian centers for molecular diagnosis**

✗ Epidermolysis Bullosa : IDI-IRCCS (G. Zambruno), Dept. Biomedical Sciences & Biotechnology, Brescia (M. Colombi) for dystrophic epidermolysis bullosa

✗ Ichthyoses: IDI-IRCCS (G. Melino) & Univ. Napoli (F. Salvatore), IDI-IRCCS (G. Zambruno), S. Raffaele-IRCCS, Milan (P. Arrera, M. Ferrari), CSS-IRCCS, S. Giovanni Rotondo (L. Zelante), Univ. Siena (A. Renieri), S.G. Battista, Torino (S. Gallone, B. Pasini)

✗ Neurofibromatosis type 1: Univ. Cagliari (M.A. Melis), CSS-IRCCS, S. Giovanni Rotondo (L. Zelante), Univ. Genova (F. Ajmar), Univ. Milana (L. Larizza), Univ. Padua (R. Tenconi), Ist. Mendel (B. Dallapiccola)

✗ Palmoplantar keratodermas: IDI-IRCCS (G. Melino) & Univ. La Sapienza, Rome, (P. Grammatico)

✗ Xeroderma Pigmentosum: M. Stefanini (CNR-IGM, Pavia)

⇒ Italian guidelines for diagnosis of Epidermolysis Bullosa

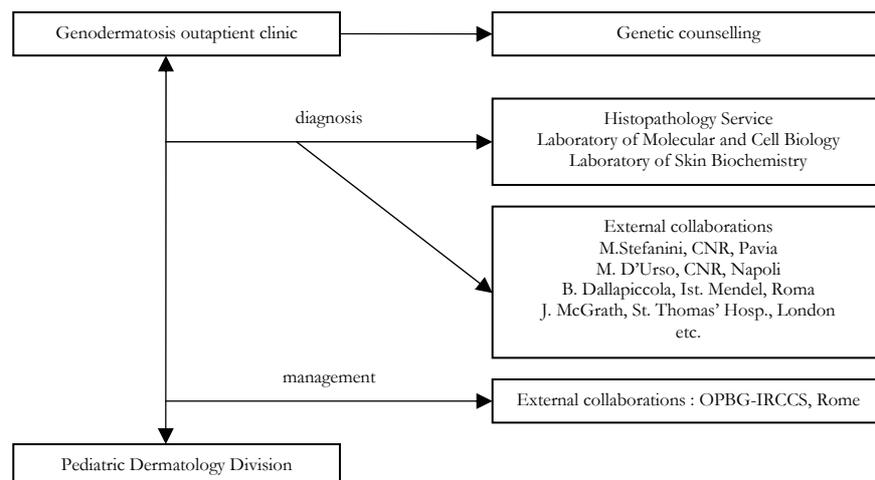
- ✗ to standardize the diagnostic approach to the EB patient with particular regard to molecular diagnosis
- ✗ proposed by a multidisciplinary study group and peer reviewed
- ✗ approved by SIDeMaST and SIGU -Società Italiana di Genetica Umana- (2004)
- ✗ diffusion to dermatologists, neonatologists, pediatricians, medical geneticists
- ✗ published online by SIGU <http://sigu.univr.it>



⇒ Pediatric Center for Rare Disease, Ospedale Bambino Gesù, Roma

- ✗ www.rarimanonsoli.it
- ✗ outpatients' department
 - a multidisciplinary team: dermatologist, endocrinologist, geneticist, immunologist, metabolism diseases' specialist, nephrologist, neurologist, orthopedist, pediatrician, psychologist
 - organization: welcome, history and examination of the patients, analysis and discussion between clinicians, discussion with the patients and/or the family
 - case manager (1 in Ospedale Bambino Gesù): takes care of the patient and guides her/him, is the co-ordinator for all the specialists involved in the management of her/his disease, organize her/his follow-up and her/his appointments, keeps in touch with GP and her/his support association, makes her/his trip to the hospital easier

⇒ IDI Genodermatosis Center



Assistance for the families

⇒ Support association

- ✗ list
 - Inherited Epidermolysis Bullosa: DEBRA Italia, Associazione Pugliese per le epidermolisi
 - Ichthyosis: UNITI
 - Ectodermal dysplasia: ANDE
 - Neurofibromatosis: ANF (national), SINCRO (Ancona), LINFA (Padova), ANANAS (Roma), UNIAMO (Venezia)
- ✗ actions
 - participate in congress and national sessions
 - spread the information among patients
 - set out patients' and their families' difficulties
 - listen to MD's proposals to support better patients

Research

Funding

⇒ At the national level

- ✗ 2004 : Italy-USA project on rare diseases activated by the Ministry of Health (funds assigned through the Istituto Superiore di Sanita)
- ✗ research on specific topics in the field of rare diseases funded by the Ministry of Health (project of IRCCS) and the Ministry of Higher Education and Research
- ✗ significant funding from private charities

⇒ At the European level

- ✗ Framework Programs of the European Commission
- ✗ significant fundings also comes from private charities (for Dermatology: DEBRA-Europe, Généthon-Téléthon)

Difficulties

⇒ Linked to the medical profession

- ✗ improve knowledge of these diseases and their complications
- ✗ develop training
- ✗ improve nurses' knowledge for specific care
- ✗ strengthen the coordination between the various specialized centers; between MDs, associations and authorities
- ✗ collect additional funding for healthcare and research
- ✗ model a national comprehensive register upon the Epidermolysis Bullosas' one set up by G. Tadini

⇒ Linked to the patients

- ✗ develop treatment for curing
- ✗ train more specialists for the management of genodermatoses
- ✗ set up new multidisciplinary centres
- ✗ promote case manager for consultations
- ✗ strengthen community-based healthcare networks
- ✗ give a better material support for the patients and their family

Accomplishments

⇒ The Italian Societies of Dermatology and Paediatric Dermatology are involved in the "Genodermatoses and Mediterranean" project

- ✗ SIDeMaST: www.sidemast.org
- ✗ SIDerP: www.dermatologiapediatrica.com

⇒ A survey of activities and needs is on progress

- ✗ a co-ordination group with clinicians and scientists has been appointed for each of the disease groups identified (Inherited Epidermolysis Bullosa, Ichtyosis, Xeroderma Pigmentosum, Palmoplantar Keratoderma, Neurofibromatosis)

⇒ Medical and scientific collaboration between clinical and diagnostic centers

- ✗ a joint centre for inherited Epidermolysis Bullosa in Roma

- ◇ **Beginning of a collaboration between clinical centers according to their competences**
- ◇ **A model for a multidisciplinary management**
- ◇ **A specific attention to relationships with patients' associations**
- ◇ **Information websites on rare disease**
- ◇ **Strengthening of the relationships with authorities**

Geneskin

An example of collaborative project on genodermatoses (EU 6° FP)

- ◇ **From 07/01/2005 for 3 years**
- ◇ **32 clinical and research centres in 12 European countries**
- ◇ **EC contribution (only for coordination): 1 238 200 euros**
- ◇ **Project objectives**
 - ✗ create a European network for 5 major groups of genetic skin diseases
 - ✗ through a dedicated website
 - ✗ improve diagnosis ; integrate, test and validate diagnostic tools and facilitate research for genetic disease
 - ✗ improve access to diagnosis, management and treatment
 - ✗ address needs of the patients and their family
 - ✗ organize training on clinical, diagnostic and management aspects of specific disease groups
 - ✗ promote pan-European communication pathways among patient's organizations, ethic committees, physicians and scientists
 - ✗ create an European task force aggregating clinical and research centers
- ◇ **Limits**
 - ✗ funding do not allow to develop comprehensive measures for all diseases
 - ✗ further support needed for the establishment of database and biological material collections
 - ✗ further support needed for specific research actions varying according to present level of knowledge of the different diseases

PROPOSALS

- ↻ **Identify the clinical reference centers**
- ↻ **Create multidisciplinary centres for a comprehensive follow-up and promote their decentralization**
- ↻ **Identify a specific "case manager" for each disease and for each centre**
- ↻ **Arrange regular meetings in between specialists and the "case manager"**
- ↻ **Promote home health care**

↵ **Organize training sessions**

⇒ 1° Giornate Le Malattie Rare in Dermatologia Pediatrica, 25 et 26 novembre 2005

↵ **Promote practical training sessions in specialized centres**

↵ **Set up medical protocols and specific directives for management and treatment**

↵ **Spur on collaboration between clinicians and scientists**

↵ **Improve coordination between the medical profession and support associations**

↵ **Involve authorities to get funding, to create an epidemiological network**

↵ **Spread information regarding actions undertaken by websites, press or conferences to inform on a regular basis patients, their family and to involve volunteers**

LEBANON

H. Nassif

“Institut de la Gestion de la Santé et de la Protection Sociale, St Joseph University”, Beirut

A. Mégarbané

Director, “Unité de Génétique Médicale, St Joseph University”, Beirut

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President, Lebanese Society of Dermatology

R. Tomb

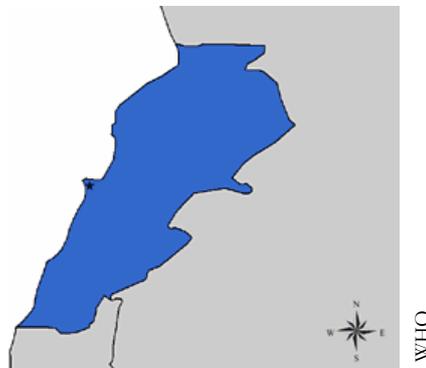
Head of the Dermatology Dept., Hôtel Dieu de France, Beirut

T. Zakhia

Director, “Institut de la Gestion de la Santé et de la Protection Sociale, St Joseph University”, Beirut

CURRENT SITUATION

General information



◇ Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

total population	3 596 000
GDP per capita (Intl \$, 2001)	5 528
total health expenditure per capita (Intl \$, 2001)	673
total health expenditure as % of GDP (2001)	12,2

◇ 10 000 sq. km

- a networked structure is not required for this country -a single centre can treat all patients

◇ A rich tapestry of communities

✗ Christian communities

- Catholic churches: Maronite, Melkite, Roman, Armenian, Syrian, Chaldean
- other churches: Orthodox, Armenian Apostolic, Syrian Orthodox, Assyrian Apostolic of the East, Orthodox Coptic, Evangelical

✗ Muslim communities

- Sunni
- Shiite (Twelvers)
- Alawite
- Ishmaelite
- Druze

◇ High rates of consanguinity

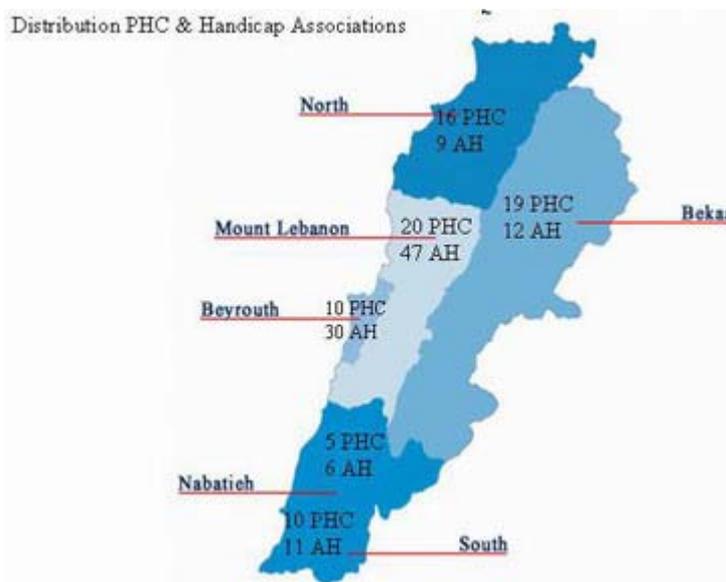
- ✗ 15% in Beirut
- ✗ 37% in rural areas
- ✗ prevalence of certain genodermatoses characterised by autosomal recessive transmission
- ✗ amplification of founder effects in isolated populations
- ✗ clinical variants of known genodermatoses or new syndromes

⇒ **The Shiite community, a special case**

- frequent endogamy (30–90%)
- 1st and 2nd degree consanguinity
- early marriages
- large families
- numerous genodermatoses are unidentified or in the process of being identified

⇒ **More or less equitable distribution of health care institutions**

- ✗ primary care centres
- ✗ dispensaries
- ✗ public and private hospitals



⇒ **Public health priorities defined by the Ministry of Health**

- ✗ cancer
- ✗ diabetes
- ✗ renal failure / dialysis
- ✗ several other chronic diseases

⇒ **Centres for the treatment of chronic diseases**

⇒ **Law No. 220**

- ✗ governs the rights of disabled persons
 - right to health care and rehabilitation services
 - right to an accessible environment
 - transportation and parking
 - education, leisure activities and sports
 - work or employment
 - social benefits and tax reductions

⇨ **Reimbursement of health care costs/medicines/medical equipment**

- ✗ chemotherapy and cardiac interventions provided free of charge
- ✗ for other pathologies, patients must be enrolled in a private social security scheme
- ✗ comprehensive coverage for patients in governmental hospitals
- ✗ no dermatological drugs are reimbursed

⇨ **Training**

- ✗ 6 universities

⇨ **Institut de Gestion de la Santé Publique et de la Protection Sociale (IGSPS)**

- ✗ dynamism and experience of civil society in the development of community health programmes (family, friends, professionals)
- ✗ availability of specialised human resources, trained in universities, and open to continuing education
- ✗ experience in the creation, support and evaluation of associations working with vulnerable populations
- ✗ ties developed with social service and health care structures in Lebanon in both the private and public sectors

⇨ **Pathologies encountered**

- ✗ list: patients suffering from ichthyosis, incontinentia pigmenti, epidermolysis bullosa (about 500 cases of Herlitz junctional EB in the southern part of the country²), xeroderma pigmentosum (very rare), keratodermas, neurofibromatosis, ectodermal dysplasia, IFAP (Ichthyosis-Follicularis-Atrichia-Photophobia) syndrome, Netherton syndrome, Werner syndrome

Care

⇨ **Individualised and fragmented care delivery**

- ✗ in private offices of physicians and rehabilitation specialists
- ✗ in rehabilitation centres run by associations depending on the disabilities involved (prostheses and orthoses, parent education, physiotherapy, psychotherapy)

⇨ **Hospital care co-ordinated by a nurse or social worker**

Assistance for families

⇨ **Care is partially funded**

- ✗ depending on the system of coverage to which the family is entitled

⇨ **82 associations caring for disabled persons**

- ✗ distributed throughout the country
- ✗ Comité pour l'Élimination de la Lèpre au Liban (CELL) is also interested in rare diseases such as dystrophic epidermolysis bullosa
 - participates in the detection of unknown cases
 - carries out an information programme
 - ensures the monitoring of treatment
 - ensures patient re-education and rehabilitation
 - provides for the insertion of children in suitable technical schools

² Figures provided by CELL (Comité pour l'Élimination de la Lèpre et des Maladies Orphelines au Liban)

- accompanies reinsertion and offers moral support
- encourages genetic research

Research

- ⇒ **Scientific publications**
- ✗ close collaboration with European teams
- ✗ genetic explorations to be developed

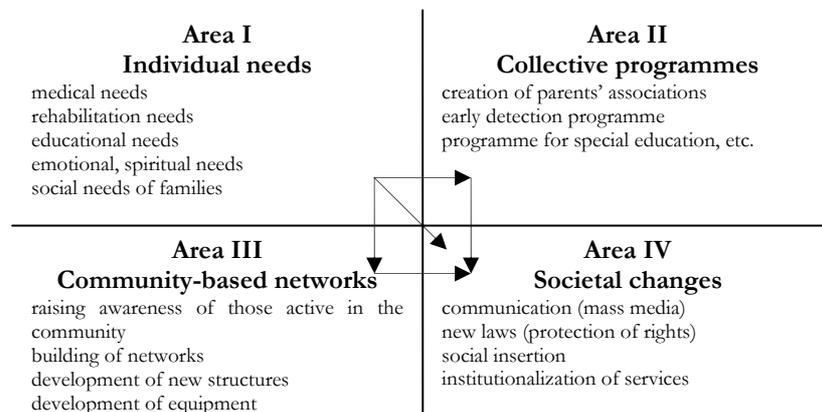
Education and training

- ⇒ **Specific training only as a part of the dermatology curriculum**
- ⇒ **Ecole de Génétique Moléculaire**
- ✗ organised by the Medical Genetics Unit of Saint-Joseph University, a member of the Agence Universitaire de la Francophonie
- ✗ for students, physicians, biologists, laboratory technicians, etc.
- ✗ theoretical instruction and hands-on training
- ✗ French-language programme

PROPOSALS

↳ **Role of the “Institut de Gestion de la Santé Publique et de la Protection Sociale” in the creation of a national network for genodermatoses**

⇒ strategy: structural approach focusing on social change and protecting the rights of vulnerable persons



⇒ the initiative is carried out simultaneously in the 4 areas

- individual and family needs
- collective needs requiring the development of new action programmes
- community resources organised into networks
- society: development of new laws, institutionalisation of services

⇒ action plan I

- determine the magnitude of the problem
 - number of persons afflicted
 - diagnoses

- social, medical and psychological needs of individuals and families
- assistance for the creation of a family support group
- educate and provide information to parents and friends concerning the structures existing in the country able to meet their needs
- inform the existing associations of this problem
- ⇒ action plan II
- assist in the creation of specialised local care facilities
 - in the regional primary care centres
 - in dispensaries (home health care)
- ensure specific training for personnel in these centres
- collaborate with non-transmissible chronic disease staff in the Ministries of Health and Social Affairs in order to
 - create a national genodermatosis programme (prevention campaigns, production of materials, development of research)
 - granting of disabled identity cards providing advantages for free or nearly free care
- ↪ **Ensure coverage for medicines, hospital and extra-hospital care**
- ↪ **Request assistance from the Ministry of Health**
- ↪ **Create centres for the treatment of genodermatoses**
- ↪ **Create a centre of excellence in Beirut**
- ↪ **Centralise research and create a regional centre of excellence**
- ↪ **Promote North-South collaborative efforts with respect to scientific research, information, training, technology transfer, medical and social assistance**

LIBYE

Mohamed BENGHAZIL
Tripoli General Hospital

CURRENT SITUATION

General information



- ◇ Large area
- ◇ Low population: 5,5 millions
- ◇ High consanguinity
- ◇ Dermatology
 - ✗ 3 Dermatology departments
 - 1 in Tripoli
 - 2 in Benghazi
 - ✗ outpatients' departments in the other regions
 - ✗ 50 dermatologists
 - ✗ 150 MDs in dermatology departements (training, specialization, etc.)

Care

- ◇ All medical services provided free of charge
 - ✗ in public hospitals and health centers
 - ✗ including available investigations and drugs

Research

- ◇ Clinical studies
 - ✗ presented in international conferences (PALD, EADV) or published in International Dermatological Journal
 - ✗ on limited samples of patients

Accomplishments

- ◇ Ministry of Health involvement for the set up of a national program genodermatoses

PROPOSALS

A national program for genodermatoses
with the Ministry of Health
to coordinate and supervise the “Genodermatoses and Mediterranean” project

- ↵ **Strengthen the relationships between Tripoli and Benghazi dermatology departments**
- ↵ **Strengthen the relationships**
 - ⇒ between public and private sectors in dermatology
 - ⇒ between dermatology and the other specialties involved in the management of genodermatoses
- ↵ **Set up pilot dermatology departments**
- ↵ **Carry national surveillance for common and severe genodermatoses**
- ↵ **Refer patients to specialized centres for further examination, investigations and management**
- ↵ **Train caregivers dealing with genodermatoses patients**
- ↵ **Start research in collaboration with the national research center**
- ↵ **Involve NGOs and charities to fund the project**
- ↵ **Heighten public awareness of genodermatoses and the problems caused by consanguinity through all media facilities (TV/Radio/Press/School/Mosque/etc.)**
- ↵ **Create patients’ associations for genodermatoses**
- ↵ **Involve the society of dermatology in the field of genodermatoses**
- ↵ **Involve the major laboratories regarding**
 - ⇒ molecular biology and genetics
 - ⇒ to set up relationships with the reference laboratories of the countries with a strong experience in the field of genodermatoses
- ↵ **To promote international collaborations with Mediterranean countries**
 - ⇒ exchange
 - ⇒ training
 - ⇒ advanced investigations and new treatments like gene therapy

MOROCCO

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General-Secretary, Gildi
A. Idrissi Azzouzi
Consultant Dermatologist and Head of Dermatological Disease Unit
Direction de l'Epidémiologie et de la Lutte contre les Maladies, Ministry of Health

CURRENT SITUATION

General information



◇ Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

total population	30 072 000
GDP per capita (Intl \$, 2001)	3 887
total health expenditure per capita (Intl \$, 2001)	199
total health expenditure as % of GDP (2001)	5,1

◇ A large country with mountainous regions

◇ The Moroccan health care sector: a landscape of reforms

- ✗ these reforms stem from
 - a political choice in favour of regionalisation
 - public administration reforms
- ✗ establishment of regional health agencies
- ✗ redefinition of the missions of central structures
- ✗ development of the regulatory function
- ✗ separation between the management function and the regulatory function
- ✗ development of results-based management
- ✗ hospital reforms are implemented through two projects:
 - Projet de Financement et de la Gestion du Secteur de la Santé (PFGSS)
 - Projet d'Appui à la Gestion du Secteur de la Santé (PAGSS)
- ✗ introduction of obligatory health insurance (Assurance Maladie Obligatoire, AMO)
- ✗ establishment of the Régime d'Assistance Médicale aux Economiquement Démunis (RAMED), a scheme serving disadvantaged persons

◇ A new social policy

- ✗ revision of legal texts affecting the social welfare domain
- ✗ redefinition of the role of social assistance
- ✗ institutionalisation of collaboration with other participants in social welfare
 - central departments of the Ministry of Health

- other departments with social portfolios
- civil society

◇ Care delivery today

- ✗ an effective community-based care network

◇ Health care costs and reimbursement

- ✗ hospital care is not provided free of charge
 - consultations: DH 60
 - hospitalisation: DH 100/day + DH 750 for hospitalisation costs + certain additional tests if performed (scanner, cardiac echography, MRI, etc.)
- ✗ ‘certificat d’indigence’ for needy patients valid for 3 months (setting up of RAMED)
- ✗ free consultations in Rabat University Hospital for patients suffering from genodermatoses thanks to the “Association Marocaine des Génodermatoses”

◇ Dermatology

- ✗ 300 dermatologists
 - half of these are in private practice, primarily based in cities
 - the other half work in the public sector
- ✗ 4 university hospitals with dermatology departments

◇ The status of dermatology in public health in Morocco

- ✗ 3 departments of the Direction de l’Epidémiologie et de la lutte contre les Maladies (DELM), the Ministry of Health’s epidemiology and disease control section, manage 3 dermatology programmes
 - a national programme to combat leprosy
 - a national programme for the fight against STDs and AIDS
 - a programme focusing on cutaneous leishmaniasis
- ✗ many dermatoses require the intervention of the health authorities due to their prevalence and the suffering involved (infectious mycotic and parasitary skin disorders, psoriasis, eczema, genodermatoses)

◇ Are genodermatoses a public health problem?

- ✗ the terms “rare” and “orphan” diseases are to be used guardedly:
 - for example, xeroderma pigmentosum is prevalent in Mediterranean countries, particularly in North Africa
- ✗ resurgence of interest due to advances in genetics and cellular biology
- ✗ no epidemiological study on the national level
- ✗ technical and operational vulnerability: genetic diseases, preventive and symptomatic treatment only
- ✗ acceptability of solutions: therapeutic abortion issue

◇ Genetic counseling

- ✗ faces the challenge of very high consanguinity
- ✗ runs up against a particular cultural context: a “sterile” woman is rejected by society



Gildi

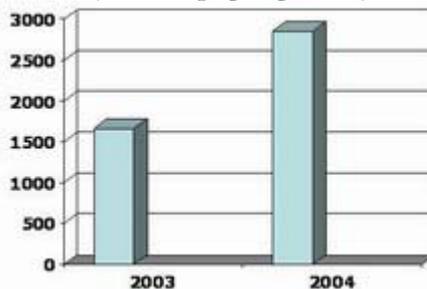
www.gildi.org

An association offering support to persons suffering from dermatological diseases

- ✗ support for needy patients suffering from dermatoses
- ✗ created in September 2001

◇ Fields of action

- ✗ assistance for disadvantaged persons
 - 947 patients receiving medicines (2–3 drugs per patient) / 2,850 units



- 82 patients have Gildi files (long-term coverage)
- 44 patients have received x-ray or biological examinations
- ✗ purchases of material (e.g., minor surgery equipment)
- ✗ maintenance works
- ✗ computerization: medical files stored and processed in an information system
- ✗ education: brochures, posters, public education sessions
- ✗ entertainment aspects: shows and gifts for birthdays and holidays
- ✗ partnerships: close collaboration with the Ministry of Health's epidemiological staff, hospital personnel, the Moroccan paediatric dermatology association, public health officials, the media, genodermatosis patient groups
- ✗ other areas
 - prevention of nosocomial infections
 - actions in support of centres dealing with precariousness and disaffection
 - creation of Gildi regional offices
 - awareness campaigns

Care

General analysis of the health care offering for genodermatoses

◇ Strengths

- ✗ interest for these diseases
- ✗ presence of associations
- ✗ 2 networks are already operational

◇ Weaknesses

- ✗ accessibility is limited geographically
- ✗ financial accessibility also limited
- ✗ lack of genetic diagnosis facilities
- ✗ insufficient support for the current treatment structures for genodermatoses

◇ Opportunities

- ✗ “Genodermatoses and Mediterranean” project
- ✗ project for a mobile structure for rural Morocco

◇ Perils

- ✗ competition instead of synergy
- ✗ other public health priorities

Care

Care delivery in Casablanca

The example of the dermatology department of Ibn Rochd University Hospital

◇ A few figures

- ✗ 651 hospitalisations in 2004
- ✗ 13,457 consultations
- ✗ 2 weekly consultation days for paediatric dermatology
- ✗ 25% of patients are children

◇ Genodermatoses: types and frequency

434 suffering from neurocristopathy, 286 xeroderma pigmentosa, 213 ichthyosis, 102 congenital epidermolysis bullosa, 98 palmoplantar keratodermas, 31 pilary dysplasias, 22 elastopathies, 4 cases of Cokayne syndrome, incontinentia pigmenti

◇ Care and management

✗ clinical care

- patients reside in areas that are difficult to access (late diagnoses and challenges to monitoring)
- diagnosis: questionnaire, clinical examination, data sheet (genealogical tree), contact information (address, telephone)
- hospitalization: treatment, education, genetic counseling, social assistance (transportation expenses, clothing, etc.)
- monitoring: quantification of treatment (dosage, duration), consideration of geographic distance and socio-economic condition

✗ therapeutic care

- surgery: services in reconstructive surgery, maxillo-facial surgery (for xeroderma pigmentosum and hereditary epidermolysis bullosa)
- medicines: donations by Gildi

✗ genetic care

- October 1995: “Généthon” project (60 families examined over a 24-hour period / genotype study for genodermatoses)
- December 1997: meeting at the molecular genetics laboratory in Villejuif, France for the launch of a genetic study of xeroderma pigmentosum
- March 2002: 1st conference of the Fédération Maghrébine de Dermatologie Pédiatrique in Algiers
- April 2003: 2nd Franco-North African medical conference in Algiers
- February 2004: “Genodermatoses and Mediterranean” round table in Algiers
- June 2004: resumption of collaboration with Généthon (Prud’hom team)
- studies in progress: ichthyoses, hereditary palmoplantar keratoderma, familial vitiligo, familial defects, familial psoriasis
- outlook: genetic research with a therapeutic focus, genetic counseling for prenatal

diagnosis

✗ social services

- social assistance: payment of transportation, tests, hospitalisation, medicines, sunscreens, emollients, genetic counseling, education

◇ **Contribution of Gildi to the treatment of genodermatoses in Casablanca**

✗ financial assistance

travel, hospitalisations, biological and x-ray examinations, treatments, transportation expenses, expenses for the collection and processing of blood samples

◇ **Medicines donated in 2004 by Gildi for genodermatoses**

emollients (992), moisturising gels (200), sunscreens (60), antiseptics (37), antibiotics (24), antihistamines (22), dermocorticoids (13)

◇ **Prevention and awareness of genodermatoses**

✗ education on a case-by-case basis

✗ raising awareness among the general public

- educational films shown in the consultation service
- television segments dealing with Gildi's work involving xeroderma pigmentosum, filmed during World Health Day on 7 April 2005 (shown on the evening news)
- complete television programmes

Research

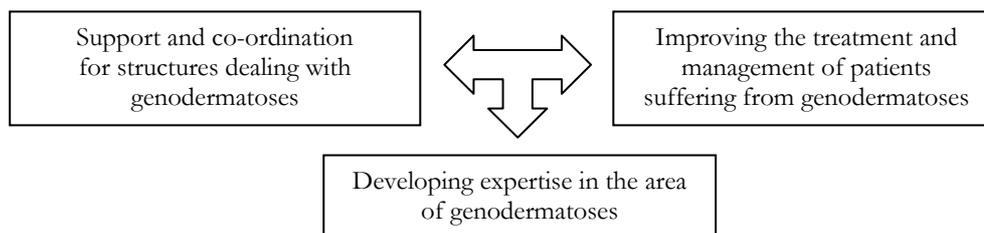
◇ **Several studies in progress**

✗ XP-associated cutaneous tumours, a study of 120 cases by Casablanca University Hospital

✗ XP-associated cutaneous tumours, a study of 100 cases (1993-2003) by Rabat University Hospital

PROPOSALS

At the national level: strategy for action



⇒ **Support and co-ordination for structures dealing with genodermatoses**

⇒ international co-ordination made possible through the “Genodermatoses and Mediterranean” project

⇒ national co-ordination between the different structures

⇒ role of the Ministry of Health

- establishment of a work group on genodermatoses
- the mission of this group will be to
 - develop national expertise in the area of genodermatoses (to create a centre of excellence)
 - carry out studies and research

- create a body of documentation
- encourage the development of a national network and regional networks

↳ **Improve social protection for patients suffering from severe genodermatoses**

- ⇒ petition for the inclusion of certain genodermatoses on the list of chronic illnesses
- ⇒ guarantee treatment free of charge for the most disadvantaged
- ⇒ improve geographical access: focus on increased mobility of care delivery
- ⇒ work towards the social integration of these patients

↳ **Develop expertise in the area of genodermatoses**

At the international level

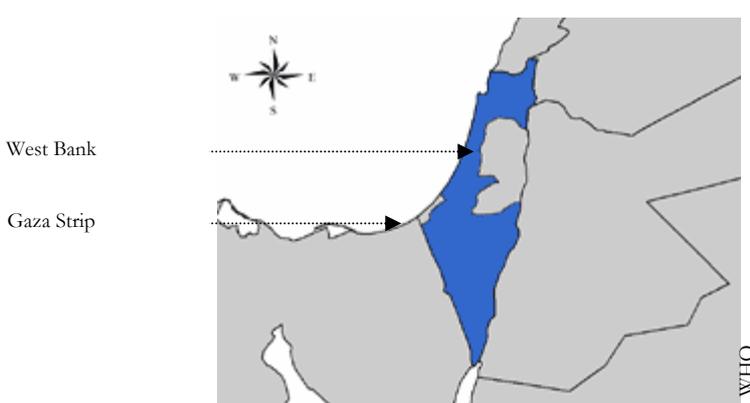
- ↳ **Create a link on the Gildi Web site to the (future) site of the “Genodermatoses in the Mediterranean” project**

PALESTINE

H. Arda
Al Quds University, Jerusalem

CURRENT SITUATION

General information



West Bank

Gaza Strip

WHO

- ⇨ Intermarrying is very common
- ⇨ Consultations available everywhere
- ⇨ Gaza 15 dermatologists in government hospital
- ⇨ Nablus 6 dermatologists
- ⇨ Incidence of congenital anomalies in skin diseases in West Bank-Palestine: 1,49 %
- ⇨ Genetic counselling: prenuptial test for Thalassemia
- ⇨  Al Quds University (2000-2004): 28 003 patients
45 patients suffering from Ichtyosis, 11 Neurofibromatosis, 10 Xeroderma Pigmentosum, 8 Epidermolysis Bullosa, 6 Congenital Ectodermal Dysplasia, 4 Acrodermatitis Enteropathica

PROPOSALS

An individual management
for health care and its reimbursement and education of the patients

- ↻ Come into contact with the Health, Social Affairs and Education Ministries
- ↻ Report genodermatoses
- ↻ Inform the media
- ↻ Involve NGOs working in the field of health
- ↻ Come into contact with the families of the patients
- ↻ Work together with the PanArab League of Dermatologists

SULTANATE OF OMAN

Abla Al-Ismaili
M.R. Mustafa
Al Nahdha Hospital

CURRENT SITUATION

General information



Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

total population	2 768 000
GDP per capita (Intl \$, 2001)	11 474
total health expenditure per capita (Intl \$, 2001)	343
total health expenditure as % of GDP (2001)	3,0

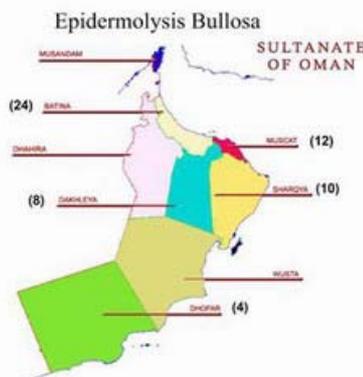
309 000 sq km

Health centers

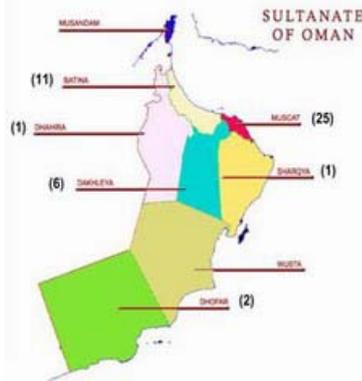
- ✗ 1970: 2 hospitals with 12 beds and 9 clinics
- ✗ 2005: 49 hospitals and 129 health centers all over Oman in each region
- ✗ the major hospitals provide 2nd and some 3rd care for the people in their catchment's area

Genodermatoses

- ✗ Total number of cases: 166
- ✗ Epidermolysis Bullosa: 126

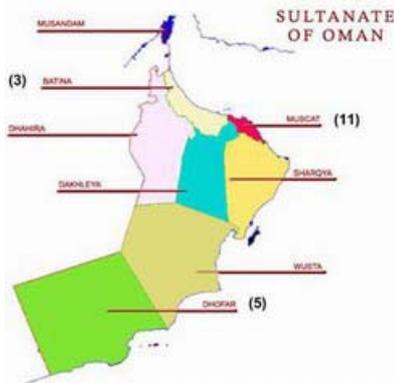


X Ichthyosis: 46



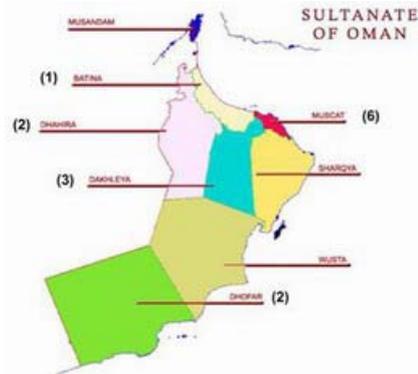
REGIONS	n°
Muscat	25
Batinah	11
Dakhleya	6
Dhofar	2
Dhahira	1
Sharqiyah	1

X Neurofibromatosis: 19



REGIONS	n°
Muscat	11
Dhofar	5
Batinah	3

X Tuberous Sclerosis: 14



REGIONS	n°
Muscat	6
Dakhleya	3
Dhahira	2
Dhofar	2
Batinah	1

X other genodermatoses

8 Darier disease, 5 Ectodermal dysplasia, 4 Palmoplantar Keratoderma, 4 Xeroderma Pigmentosa, 4 H-Haily disease, 2 Ehler-Danlos, 2 Aplasia cutis, 1 Cutis Marmorata

Care

◇ Diagnosis

X 2 pathology laboratories (Muscat region) with 1 dermatopathologist and molecular biology and biochemical diagnostic units

◇ Multidisciplinary management

X dermatologists, pediatricians, ophthalmologists, dentists, Ear, Nose and Throat specialists, dieticians, surgeons and plastic surgeons

◇ **1 Genodermatoses clinic in AlNahdha Hospital**

✗ every 2 months follow-up

◇ **Treatments**

✗ retinoids

✗ nutritional supplements

✗ laser (NF, TS)

◇ **Genetic counselling**

✗ 1 geneticist in Muscat

◇ **Social care**

✗ financial support for treatments

✗ except for transportation

✗ a few social workers

◇ **Research**

✗ 1993: *Genodermatoses in Oman*

✗ 1997-1998: *Prevalence of EB in Oman*

✗ Kindler syndrome

◇ **Information**

✗ to develop because of the fear of contagion

PROPOSALS

⇒ **Medical care**

⇒ promote home care

⇒ supply medications (sun blocks, peeling agents, etc.)

⇒ arrange new interventions: e.g., skin culture

⇒ set up antenatal screening and diagnostic lab tests

→ fetal skin biopsy

→ direct analysis of DNA from CVS or amniotic fluid cells

⇒ organize genetic counselling

⇒ **Social care**

⇒ propose financial support

⇒ propose premarital counselling (intermarrying)

⇒ improve access to care and transportation

⇒ involve social workers

⇒ **Set up a network between medical and social services in collaboration with the Ministry of Health/Social affairs and the Ministry of Religious affairs**

⇒ **Train and heighten awareness (patients and her/his family, MDs, general public)**

⇒ **Research**

⇒ set up a data bank

⇒ develop genetic studies (molecular defects)

⇒ develop gene therapy

TUNISIA

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CURRENT SITUATION

General information



Statistics

Figures are for 2002 unless indicated. Source: The World Health Report 2004

total population	9 728 000
GDP per capita (Intl \$, 2001)	7 183
total health expenditure per capita (Intl \$, 2001):	463
total health expenditure as % of GDP (2001)	6,4

160 000 sq. km

High rate of consanguinity

Organisation of the health care system

- X 4 medical schools
- X 10,000 physicians
- X 250 dermatology specialists (+ 40 in training)
- X 31,429 paramedical and auxiliary health personnel
- X 1,300 medico-social workers
- X 20 university hospitals, with a total of 7 dermatology departments
- X 148 regional hospitals
- X 2,028 primary care centres
- X 30 family planning centres
- X 100 maternal and infant protection centres

The government is very well disposed to improving the quality of medical care and social protection for disabled persons through

- X new legislation
- X non-governmental organisations (e.g., BASMA, AAGHD)

◇ **Genodermatoses are relatively prevalent in Tunisia**

◇ **A number of vulnerability factors for genodermatoses**

- ✗ high consanguinity
- ✗ socio-economically underprivileged populations
 - poverty
 - distance
 - lack of knowledge
- ✗ intense and constant insolation (xeroderma pigmentosum)
- ✗ mycoses are very prevalent (ichthyoses and palmoplantar keratodermas)

◇ **Research: 1% of the gross national product is devoted to research**

- ✗ health research: strong commitment by the Ministry for Scientific Research
- ✗ 30 research laboratories
- ✗ 60 research units
- ✗ 4 research units work in genetics
 - hereditary keratinisation disorders in Tunis
 - hereditary blistering dermatoses in Sfax
 - rare genetic diseases in Tunis
 - cytogenetics and molecular genetics in Sousse
- ✗ federal research programme
 - research subject: treatment, management and re-education of disabled persons
 - project: improve the quality of care and support for disabled persons (definition, social status, etc.)

Prevention

◇ **Communicating the dangers of consanguinity**

- ✗ ingrained tradition
- ✗ prenuptial certificate
- ✗ information, education and communication (IEC) actions

◇ **Genetic counseling for affected families**

- ✗ a few specialists
- ✗ a few structures

◇ **Prenatal diagnosis**

- ✗ operational work flow and specific equipment to be developed
- ✗ rarely performed (individual initiatives only)

Training

◇ **Study of xeroderma pigmentosum**

- ✗ during medical school and post-graduate studies (dermatology training programme)

Care and management of xeroderma pigmentosum

◇ Fight against UV

- ✗ complete photoprotection is unrealistic
- ✗ patients of simple means
- ✗ sunscreens are expensive

◇ Treatments are difficult

- ✗ develop a specific therapeutic management strategy suited to the situation, with
 - specialised consultation services
 - therapeutic consensus
 - co-ordination between the various caregivers
 - specific training for the management of the disease
- ✗ develop a specific health programme
 - prevention (consanguinity, genetic counselling and prenatal diagnosis)
 - therapeutic management
 - training

Recent actions

◇ Improving the quality of care for genodermatoses is a political decision that involves

- ✗ the Ministry of Public Health
- ✗ the Ministry of Social Affairs
- ✗ the Ministry of Higher Education
- ✗ the Ministry of Scientific Research, Technology and Expertise Development

PROPOSALS

↵ Care

- ⇒ Facilitate access to care
 - institute universal coverage for patients
 - facilitate the delivery of disabled identity cards (exemption from all health care expenses in the country's medical facilities)
- ⇒ create multidisciplinary groups in each university hospital for each genodermatosis (centres of excellence)
- ⇒ improve co-ordination between the different participants
- ⇒ expand paediatric dermatology consultation services in the various university hospitals
- ⇒ develop clinical diagnostic tools
 - by leveraging existing structures in each university hospital (molecular biology, electron microscopy)
 - by providing specific training for physicians, technicians (biologists) and scientists

↵ Medicines

- ⇒ financial assistance provided to patients for medicines

↵ Prevention

- ⇒ support policies for the prevention of genodermatoses and especially those that approach the problem from the perspective of genetics (awareness campaign targeting the general public)
- ⇒ prenuptial certificate

- mandatory and governed by legislation
- train general practitioners (training organised by the Ministry of Public Health)
- raise awareness among future spouses concerning the risks of consanguinity
- ⇒ family planning: limit the number of births in affected families
- ⇒ prenatal diagnosis: use existing structures to make this examination more accessible through the training of personnel and financial assistance

↵ **Education and training**

- ⇒ train paramedical personnel and home caregivers for disabled persons

↵ **Information**

- ⇒ improve access to information for the general public
 - make information sites accessible to physicians
 - facilitate the access to information for patients and their families
 - role of patients' associations
 - training in hospitals and in maternal and infant protection centres

↵ **Research**

- ⇒ collaborate with the existing structures so as to identify the genes responsible for diseases in the hope of devising a form of gene therapy
- ⇒ create a research unit specifically for genodermatoses involving dermatologists, paediatricians, geneticists, other scientists, auxiliary health personnel, patients' associations and social workers

↵ **North-South co-operation**

- ⇒ promoting North-South co-operation: complementarity between the two shores of the Mediterranean for technology transfer in the research areas of clinical, therapeutic and biological epidemiology
- ⇒ better cohesiveness and efficiency of scientific exchanges between the two shores of the Mediterranean and the participating countries
- ⇒ build on existing networks: for example, the Economic and Health Network in North Africa

CONCLUSIONS AND OUTLOOK

- ◇ **“Genodermatoses in the Mediterranean basin”, an opportunity and a strong commitment**
 - ✗ to work together in the countries of the Mediterranean basin and the Middle East
 - ✗ to improve medical care and social protection for patients suffering from severe genodermatoses and their families

- ◇ **Severe genodermatoses, a public health challenge**
 - ✗ a medical and social handicap for patients
 - ✗ a curse for their families
 - ✗ varying and complementary approaches in each of the countries of the Mediterranean basin and the Middle East

- ◇ **Actions relying on a wealth of national expertise in order to**
 - ✗ improve the quality of care
 - ✗ inform and educate patients and their families as well as caregivers
 - ✗ encourage the development of patients’ associations
 - ✗ support research
 - ✗ reinforce national projects through international collaboration

- ◇ **In order to ensure optimal care and management of patients and their families, it is imperative to**
 - ✗ ensure management of care, medicines and medical equipment, transportation
 - ✗ offer community-based care
 - ✗ ensure early and rapid diagnoses
 - ✗ propose a multidisciplinary approach to care
 - ✗ facilitate access to medicines
 - ✗ promote social integration
 - ✗ educate and inform
 - ✗ support research on new therapies

- ◇ **Innovative responses meeting the needs of patients and their families**
 - ✗ build work groups on the national level associating physicians, biologists and public health officials
 - ✗ establish national registries for severe genodermatoses
 - ✗ obtain recognition for severe genodermatoses as chronic diseases and/or handicaps
 - ✗ promote intervention programmes suited to each country
 - ✗ build on existing structures/care networks/associations
 - ✗ make treatment guidelines/protocols available to caregivers, organise hands-on training
 - ✗ establish an international distribution facility for medicines and build ties with the pharmaceutical industry to improve access to medicines
 - ✗ reinforce collaborative efforts for care, training and research

- ◇ **26-27 May 2006 in Tunisia**
 - ✗ assemble work groups comprised of public health officials and physicians in each country
 - ✗ evaluate the actions implemented on the national level in 2005–2006 to improve the quality of care for these patients and their families