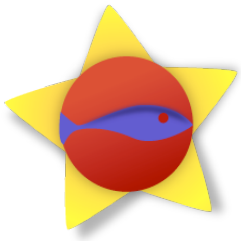

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Together for a better health care

Génodermatoses et Méditerranée
Thanks to the involvment of dermatologists from Mediterranean and Middle-Eastern countries, *Génodermatoses et Méditerranée* has been initiated in 2003 by the Fondation René Touraine and one of its founding members, Laboratoires Pierre Fabre.

Through the exchange of experience and expertise at an international level, *Génodermatoses et Méditerranée* aims at improving in each partner countries the health and social care of patients affected by severe genetic skin diseases.

Handicap, limited life expectancy, exclusion, are factors that make the patients and their families extremely vulnerable. To day, only symptomatic treatments are available. They are of the utmost importance. The health care is complex and involves numerous specialities. Social life is extremely difficult for the patients and their families: access to school, to work and leisures is almost impossible.

The partner countries have targeted 6 severe genodermatoses:

- Inherited Epidermolysis Bullosa

This form of severe genodermatose 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 is similar to that of severe burn victims, as the barrier function is highly impaired. Without treatment patients are likely to die prematurely from malnutrition and infections. The objective of treatment is to give the best possible well-being to the5: skin care, prevention of complications, management of pain, psychological support for the patient and his/her family, genetic counselling.

- Severe Ichthyosis

Severe ichthyosis is due to a genetic defect of the desquamation. The objective of treatment is to reduce skin thickening and to prevent complications thanks to skin care. Without treatment the body is covered with large scales making social life very difficult.

- Palmoplantar Keratoderma and Meleda syndrome

Hereditary palmopantar keratoderma and Meleda syndrome are characterized by the appearance of patches of hard skin on the feet and on the hands. Treatment aims at cleaning out patches of hard skin. Without treatment, the skin on the hands and the feet acquire a horny texture making movement extremely difficult.

- Neurofibromatosis

Neurofibromatosis type 1 (NF1) is an inherited, multi-system, neurocutaneous disorder that predisposes to the development of benign and malignant tumours. Children should be reviewed annually to check skin, spine, vision, blood pressure and progress at school. The treatment, which is symptomatic, can include surgery for symptomatic neurofibroma, progressive scoliosis and pseudoarthrosis. Children with learning disabilities need an early support.

- Xeroderma pigmentosum

Xeroderma pigmentosum is caused by a genetic defect in DNA excision repair following exposure to the sun. Treatment is mainly prevention from the sun and skin tumors' exeris. Without protection from the sun's ultraviolet radiation, affected children quickly succumb to a variety of fatal skin cancers.

- Other severe genodermatoses such as anhidrotic ectodermal dysplasia and incontinenia pigmenti.

Together Against Genodermatoses
Since 2008, *Génodermatoses et Méditerranée* is developing at a European level through the project *Together Against Genodermatoses (TAG)*. This project is co-financed by the European Union in the framework of the public health programme. TAG is a pilot project for the development of European Networks of Reference for Rare Diseases.

Get together the North and South countries around a public health project focused on the patients

The partner countries of the project are Algeria, Belgium, Bulgary, Croatia, Cyprus, Czech Republic, Egypt, France, Germany, Greece, Iran, Iraq, Italy, Kuwait, Lebanon, Libya, Malta, Mauritania, Morocco, Oman, Palestinian Territories, Portugal, Romania, Saudi Arabia, Slovenia, Spain, Tunisia, Turkey and United-Kingdom.

Answer the needs of patients

Six working groups are focused on groups of diseases. These international groups are composed of doctors and representatives of patients' associations. The mission of these groups is to define the best strategies for health care for each disease while focusing on the daily life of the patients and the economic and social realities of each country: health, nurse and social care. A transversal working group has also been introduced. One of its missions is to set up a network of laboratories focused on the genetic diagnosis and counseling.

Together for a better care

Since 2003, each year, the partners of *Génodermatoses et Méditerranée* and since 2009 those of *TAG* gather. Each year, the meeting is organised in a different country by the national association of dermatology or of pediatric dermatology under the auspice of the Ministry of Health of the host country and partner countries.

• 2003, France

The inaugural meeting took place in Paris in the framework of the first Sunstainable Development Forum.

• 2004, Algeria

On the invitaton of the Algerian Minister of Health, the first Euro-Mediterranean on Health enabled the discussion on the actions to be done in each country and on the international level to improve the health care of severes genodermatosespatients.

• 2005, Lebanon

Each country presented the patients' situation and suggestions to improve medical and social care.

• 2006, Tunisia

The meeting on the theme « genodermatoses, a severe skin handicap », focused on the strategies to make the genodermatoses part of the major public health issues such as chronic diseases, rare diseases, handicap, cancer.

• 2007, Egypt

Participants divided themselves into five working groups focused on the following pathologies : epidermolysis bullosa, ichthyoses, palmoplantar keratoderma, xeroderma pigmentosum and other severe genodermatoses.

• 2008, Morocco

Five years after the launch of *Génodermatoses et Méditerranée,* partners of the project made an assessment of the evolution of the patients' situation in each country and on the widening of the project to new partners.

• 2009, Greece

At the first TAG meeting, each partner presented the available epidemiological data, the assessment of patients' need and cost of the disease, the development of centres of expertise, the actions of patients' organisations, health care strategies, development of national networks and European and international collaborations. The 6 working groups gathered and defined a 3-year working programme. A transversal working group has been created to improve the access to diagnosis for the patients and their families.

• 2010, Italy

This second TAG meeting will focus on the working groups' results, the presentations of the last advances in the field of care and research, and an inter-religious debate on bioethics.

Develop synergies

Génodermatoses et Méditerranée and *TAG* are developing thanks to the involvment of experts in each country and the support of associations of dermatology and pediatric dermatology, Ministries of Health, and patients' organisations. European Union, Orphanet, Eurordis and Laboratoires Pierre Fabre bring specific support to *Génodermatoses et* Méditerranée and TAG.

TAG - Together Against Genodermatoses

A window on Europe

TAG has been initiated in 2008 for 3 years with the aim of improving health and social care of severe genodermatoses patients and families. *TAG* is co-financed by the European Commission and *TAG* is in the continuity of *Génodermatoses et Méditerranée.* This new project, chosen as a pilot project for the development of European networks of reference for rare diseases, is a window on Europe. It widens the exchange of experience by crossing widely the barrier of the Mediterranean and Middle-Eastern regions and helps developing new complementarities between the different partners.

A project focused on the patients and their families

In synergies with the existing initiatives, the *TAG* project's objectives are to:

- raise general public' awareness on severe genodermatoses,
- know better the needs of the patients,
- develop recommendations for prevention (genetic counseling, antenatal diagnosis) adapted to each country's realities,
- develop recommendations for health care adapted to each country's realities,
- develop solutions to improve the access to drugs, cosmetics and medical devices,
- promote training for health personnel,
- gather the actors of the health care of severe genodermatoses patients,
- develop links between specialists and paramedics at a European and international level,
- develop collaborations with patients' organisations at a European and international level,
- foster the development of national networks linked to the health authorities.

An international approach to improve patients health care in each country

The European and international nature of *TAG* helps raising health authorities awareness to the issues that represent the health care of severe genodermatoses patients and the necessity of developing a coordinated approach. One of the objectives of each partner, by leaning on the international nature of the network, is to get help from their health authorities for the patients by:

- collecting epidemiological data,
- identifying the patients' needs and the costs of the health care,
- identifying, in particular, the unsatisfied therapeutical needs to try to mobilize the pharmaceutical industry and obtain preferential deals,
- developing community networks enabling diagnosis and the earliest management possible, even in places really far away from university hospitals,
- favouring the development of patients' organisations,
- developing the training of health personnel,
- thinking about the local possibilities of developing genetic counseling and antenatal diagnosis by leaning on the international network that is under construction.

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Génodermatoses et Méditerranée - Together Against Genodermatoses : together for a better health care

An annual international meeting to share experience and develop new projects
The 2009 working session took place in Greece, May 22-25. It was organized by the Hellenic Society of Dermatology and Venereology under the auspices of the Ministry of Health of Greece and Social Solidarity and National Kapodistrian University of Athens, in the framework of the *Together Against Genodermatoses* project, co-financed by the European Union.

Algeria, Croatia, Cyprus, Egypt, France, Greece, Italy, Libya, Morocco, Palestinian Territories, Portugal Romania, Slovenia, Tunisia, and Yemen participated in this meeting. Each participant presented the advances accomplished in the health care of genodermatoses patients: collect of epidemiological data, evaluation of the needs of the patients and their families, assessment of the cost of the disease, development of centres of expertise for patients and families, development of patients' organisations, strategies for genodermatoses health care, development of a national network, development of collaborations at the European and international levels.

Seven working groups to improve diagnosis and health, nurse and social care
There are six working groups (some groups include several diseases): epidermolysis bullosa, ichthyosis, palmoplantar keratoderma, neurofibromatosis, xeroderma pigmentosum, other genodermatoses (anhidrotic ectodermal dysplasia, incontinentia pigmenti). These working groups aim at defining the best management strategies for each disease in an extremely concrete way: medical, nurse and social care. There is a transversal working group: "Trans-sectional approach of genodermatoses for a preventive synergistic action". One of the main tasks of this group will be to set up a network of laboratories for antenatal diagnosis and genetic counseling.

These groups are co-ordinated by a European coordinator and a non-European coordinator. They gathered during the 2009 working session.

Extending the project to new partners
- Kuwait: Dr. Arti Nanda, Centre of Dermatology As'ad Al-Hamad, Al-Sabah hospital
- Bulgaria: Dr. Lj. Pavlova, Dermatology Department of University Hospital, Faculty of Medecine, University St. Cyrille et Méthode
- Spain : E. Makow, Debra Association
- Spain : A. Hernandez, University Pediatric Hospital Niño Jesus, Madrid
- Eurordis : non gouvernmental « alliance » of patients' organisations, managed by the patients. It gathers more than 434 organisations of rare diseases in more than 43 countries.

A network recognized at the European and international level

The European *Together Against Genodermatoses* project is co-financed by the European Union
The European *Together Against Genodermatoses* project (TAG N° 2007 335) received a financing from the European Union, in the framework of the Public Health Programme for 3 years, from December 1st 2008 to December 31st 2011. *Together Against Genodermatoses* is a pilot project for the European networks of centres of reference for rare diseases.

Sharing information, a necessity for the progress of the health care of genodermatoses patients

A new website for the European TAG project
Thanks to Orphanet, the *Together Against Genodermatoses* has been set up. To learn more about the TAG project, its objectives, its partners, its working sessions, its working groups, go to our new website, www.tag-eu.org.

A new website for the working groups

This new tool is developed thanks to Orphanet. This website, in French and English, with a restraint access, enables the exchange between the working groups' participants. Thaks to this website, they discuss recommendations to improve the care of severe genodermatoses patients and families. The aim of these groups is also to think about the constitution of a network of laboratories focused on the help to patients and families enabling the development of antenatal diagnosis.

A newsletter

A newsletter, in French and English, is published every 2 months to evaluate the situation on the *Génodermatoses et Méditerranée* and *Together Against Genodermatoses* projects with 3 rubrics:

- Country news
- Organisations of patients news
- Working groups news

These letters are available under PDF on www.tag-eu.org and www.genodermatoses-et-mediterranee.org.

The 2009 working session

A working session organized by the Hellenic Society of Dermatology and Venereology under the auspices of the Ministry of Health and Social Solidarity of Greece and the National Kapodistrian University of Athens and with the EU support

The sixth *Génodermatoses et Méditerranée* working session and the first*TAG* meeting were held from October 22-24 in Greece. It was organized by the Hellenic Society of Dermatology and Venereology under the auspices of the Ministry of Health and Social Solidarity of Greece and the National Kapodistrian University of Athens.

Fifteen countries have presented a state of the art of the health care of genodermatoses patients.

Algeria, Cyprus, Croatia, Egypt, France, Greece, Italy, Libya, Morocco, Portugal, Romania, Slovenia, Palestinian Territories, Tunisia and Yemen presented a state of the art of the health care of genodermatoses patients (epidemiological data, progress in the assessment of the patients' needs and the cost of the disease, progress in the development of centres of expertise for patients and families, progress in the development of patients' organisations, strategies for the health care of genodermatoses patients, development of a national network, development of collaborations at a European level).

A working programme focused on the exchange of experience within the working groups, on the setting up of consultations for genodermatoses and on the development of a national network.

This state of the art permitted to see the different problems and difficulties encountered in each country and to define a working programme to:

- Develop new initiatives,
- Involve people all over the territory and promote the setting up of a national network,
- Spread information in each country,
- Organise multidisciplinary teams (and if needed, with a coordinator for each genodermatoses group)
- Develop contacts and relationships with patients' organisations,
- Promote severe genodermatoses educational and training programmes as well as specific working sessions for medical personnel and paramedics, psychologists and social workers
- Develop communication tools (websites, newsletters) to spread the information at the national and international level,
- Promote national and international collaborations to guarantee a diagnosis and a management best adapted to the patients.

Six working groups by group of diseases gathered, one tranvsersal working group has been created

The 6 working groups gathered : epidermolysis bullosa, ichthyosis, palmoplantar keratoderma, neurofibromatosis, xeroderma pigmentosum, and other genodermatoses (anhidrotic ectodermal dysplasia). A transversal working group was created: «Transsectional approach of genodermatoses for a preventive synergistic action».

A preview of the situation in 15 countries

A great diversity of situations, numerous initiatives and the will to answer the needs of the patients and their families

• A great diversity, reflect of the general situation of the country

This diversity depends on the general situation of each country and more specifically of the development of health services, development of national plans for rare diseases, the support of health authorities, the will of medical teams and the involvement of patients.

• Numerous initiatives

These initiatives are developed at every level: collect of epidemiological data, evaluation of the patients' needs, assessment of the cost of the disease, development of centres of expertise for patients and families, development of patients' organisations, definition of strategies for the management of genodermatoses, development of a national network, development of collaborations at a European level.

• Common needs

Access to diagnosis, multidisciplinary health care, access to drugs and medical devices, better knowledge of the disease, psychologic follow up, social support, meetings with other patients are needs (fulfilled or not) expressed by the medical personnel and patients.

• The necessity to develop solutions adapted to the realities of each country

With this diversity, it is of the utmost importance to adapt the recommendations issued by the working groups according to the political, economic, cultural and social context.

The available epidemiological data

The presented data concern the following pathologies : epidermolysis bullosa, ichthyosis, palmoplantar keratoderma, neurofibromatosis, xeroderma pigmentosum, other genodermatoses (including anhidrotic ectodermal dysplasia, incontinentia pigment). Data start to be collected in most countries with the development of some national registers. These data don't concern all the targeted pathologies. They are fragmented and don't cover the entire territories. Some hospital services/ specialised centres don't collect epidemiological data yet.

The presented data are from:

- national or regional registers focused on rare diseases (France, Italy, Libya)
- hospital dermatology departments or specialised centres (Algeria, Croatia, Cyprus, Egypt, Greece, Morocco, Portugal, Romania, Slovenia, Tunisia, Yemen)

In **Algeria**, data have been collected in 2 University Hospitals of Algiers, Bab El Oued and Mustapha. Data on epidermolysis bullosa, recessive ichthyosis, palmoplantar keratoderma have been communicated.

In **Croatia**, epidemiological data have not been communicated. The activity is focused on the health care of epidermolysis bullosa patients.

In **Cyprus**, epidemiological data are collected at the Cyprus Institute of Neurology and Genetics (CING) of Nicosia. Data on epidermolysis bullosa, severe ichthyosis, palmoplantar keratoderma and neurofibromatosis have been presented.

In **Egypt**, epidemiological data are collected within the departments of dermatology of Damiette, Alexandria, of Kasr Al Aini hospital – Cairo University and Al Haud al Marsoud at Cairo, of the National Research Centre of Cairo and of the Research Medical Institut of Alexandria. They are available for every group of genodermatoses targeted by the partners of the project except for the ectodermal dysplasia.

In **France**, data are from the data base CEMARA. They are available for every groups of targeted pathologies. CEMARA registers a limited number of informations on all patients managed for one or several rare diseases. Informations are about patients and care units. They are the common core necessary to fulfill the needs expressed by all the stakeholders. Patients

are followed thanks to the registration of events such as change in the health care, pathology, change of health care unit or death.

In **Greece**, data have been collected in the department of dermatology of Athens and Larisa. They are available for all the targeted genodermatoses except for the incontinentia pigmenti. An online database is under development. Of simple use, it will enable a real-time gathering all over the territory.

In **Italy**, all the Italian regions have initiated registers for rare diseases. The first national epidemiological data will beavailable at the end of 2009 at the National Centre for Rare Diseases (NCRD). The data currently available are from the national registers of epidermolysis bullosa, anhidrotic ectodermal dysplasia and xeroderma pigmentosum.

In **Libya**, data concern all the targeted pathologies. Persons have been put in charge of the data collect in the 3 main regions in Libya. These data are from a national register gathering the data from Tripoli Central Hospital, the Tripoli Medical Centre and the Gamhoria Hospital.

In **Morocco**, retrospective studies are available at the University Hospital Ibn Sina, Rabat.

In the **Palestinian Territories**, data are not available. In these territories we face all the targeted pathologies, and also very rare disease such as the Atrichia with papular lesions and new syndroms such as the H syndrom or other syndroms not identified yet.

In **Portugal**, data have been collected in Lisbon and Porto. Because of the very recent development of genodermatoses consultations and due to the really low number of patients, the data are not representative.

In **Romania**, national data on epidermolysis bullosa are available. Epidemiological data for other genodermatoses are available for the department of dermatology of Cluj-Napoca.

In **Slovenia**, data are collected by the 4 departments of dermatology. They are mostly available for the Darier disease and the epidermolysis bullosa.

In **Tunisia**, national data are available for the xeroderma pigmentosum (mapping for the xeroderma pigmentosum). For the epidermolysis bullosa, ichthyosis and palmoplantar keratoderma, data are available in the 4 departments of the University Hospitals – Charles Nicolle Hospital (Tunis), Military Hospital (Tunis), Sousse Hospital and Sfax Hospital.

In **Yemen**, data have been collected at the Taiz Hospital and concern 3 pathologies.

	National Register	Regional Register	Dermatology Department Register	Mapping
Algeria			x	
Cyprus			x	
Croatia			x	
Egypt			x	
France	x			
Greece	Under process		x	
Italy	x	x		
Libya	x			
Morocco			x	
Portugal			x	
Romania			x	
Slovenia			x	

	National Register	Regional Register	Dermatology Department Register	Mapping
Palestinian Territories			x	
Tunisia			x	Xeroderma Pigmentosum
Yemen			x	

Available epidemiological data for the 6 groups of diseases targeted by the partners

The assessment of the needs of patients and the costs of the disease for the patients

The evaluation of the needs of patients has been done really differently from one country to another. This evaluation has been made, in most of the cases, by doctors and in some cases in collaboration with patients' organisations.

These evaluations show the diversity of health and reimbursement systems, the disparity of access to adapted care and drugs, cosmetics and medical devices. Patients' management is closely intertwined to the integration of genodermatoses in the framework of public health priorities such as rare diseases, chronic diseases, handicap and cancer.

In most of the cases, the assessment of the costs is the one for the patients –and not for the care system as a whole-. The expenses taken into account are essentially those for drugs, dressings, medical devices and in some cases transportation to the health centers. The Epidermolysis Bullosa is the pathology for which we have the most data, because of the implication of Debra's organisations in this assessment. The cost of the care for the patients is very changeable for the same pathology from one country to another. The cost of drugs, medical devices, cosmetics for an epidermolysis bullosa patients is, for examinationple, assessed to 12.000 euros in France per year, 2.856 euros in Romania, 1.100 euros in Tunisia and 225 euros in Algeria.

In **Algeria**, the evaluation of the patients' needs and the costs of the disease for the patients has been made in the framework of a budget request for drugs to the Algerian Ministry of Public Health, in 2008.

In **Croatia**, the evaluation of the needs of epidermolysis bullosa patients has been made by Debra, patients' organisation.

In **Cyprus**, a study is in process to better evaluate the patients' needs. The needs and costs of the disease for the patients are covered and mostly reimbursed by the services of public health.

In **Egypt**, the evaluation of the patients' needs is made through the identification of centres for the health care of patients and the definition of strategies for management. The assessment of the costs of the disease for the patients has not been made.

In **France**, an evaluation of the needs of the patients and the costs of the disease for the patients has been made. Thecost of epidermolysis bullosa has been assessed to 1.000 euros per month. Ichthyosis patients have their ointment reimbursed by the health system, the palmoplantar keratoderma patients have their care (pedicure) reimbursed by the health system, the xeroderma pihmentosum patients have their sunscreen reimbursed by the health system, the anhidrotic ectodermal dysplasia and incontinentia pigmenti patients have their dental care reimbursed, and the support activities to overcome the learning difficulties are reimbursed by the health system to neurofibromatosis patients.

In **Greece**, the evaluation of the patients' needs points out the need of the reimbursment of:

- the disease's expenses by the health insurance
- an easy access to drugs, cosmetics and medical devices
- a development of specific medical centres or expertise centres for a better treatment
- better care
- genetic counseling
- a better knowledge of the disease

- the future of the patient and new treatments.

In **Italy**, the health insurance covers, for the patients affected by the rare diseases listed by the Ministry of Health,all the costs of the genetic diagnosis examination including the molecular and antenatal diagnosis, the cost of the laboratory examinations, the cost of consultations with specialists and of hospitalizations, the cost of drugs, of medical devices and in some regions of antiseptic creams, food supplement, sunscreen, dressings, etc. But the transportation is not covered.

In **Libya**, the evaluation showed the limited number of dermatologists in comparison with the expanse of the territory, the diffcult access to a specific diagnosis (including antenatal one), the classification of cases, the genetic counseling and specialised care.

In **Morocco**, the evaluation of needs of severe genodermatoses patients is intertwined with the definition of public health dermatology priorities. For the destitute, the system of medical assistance for the destitute (RAMED) has been developed.

In **Portugal**, the needs' evaluation shows the insufficient social support of the government: genodermatoses are considered as rare chronical diseases, patients have specific but insufficient rights. Care are free of charge in the public health structures with an additionnal financial support (+50% per child until they are 18 years old). A more complete evaluation of needs is planned. A study « best answer the needs of NF1 patients » has been initiated by the Department of Genetics of the Santa Maria Hospital of Lisbon.

In **Romania**, the needs have been evaluated for the epidermolysis bullosa patients. The cost of the disease for the patients affected by epidermolysis bullosa has been assessed to 238 euros per patient per month, and 2.856 euros per year.

In **Slovenia**, the management of epidermolysis bullosa patients is made by the health and social system.

In the **Palestinian Territories**, the political situation limits the travels of doctors and patients. The access to diagnosis, health and social care is extremely difficult.

In **Tunisia**, an evaluation of the patients' needs and the cost of drugs, cosmetics and medical devices has been made for a year for the epidermolysis bullosa (1.100 euros), ichthyosis (2.200 euros) and palmoplantar keratoderma (1.150euros) patients.

In **Yemen**, access to primary health care is very diffucult, and the health care of genodermatoses patients is even more difficult. Genodermatoses patients have free health care.

The development of specialised consultations, centres of expertise, centres of reference

The identification and organisation of these centres is different from one country to another with, in some countries, centres of reference, centres of competence, centres of expertise. These centres are labelled or not by the health authorities. The criterions of labelling are different from one country to another and in some countries from one region to another. Many centres propose a multidisciplinary approach. Due to the low number of patients, the development of centres of expertise for each of the six diseases is not always relevant, the development of these centres has to answer the needs of each country and adapt to the possibilities of neighbour countries. Collaborations for diagnosis and care are developing between different centres.

In **Algeria**, patients are followed in 2 departments of dermatology of University Hospitals of Algiers, Bab el oued and Mustapha.

In **Cyprus**, the Cyprus Institute of Neurology and Genetics is the centre of reference of the island for all the genetic diseases.

In **Croatia**, the centre of reference of the Department of Dermatology and Venerology of the University Hospital of Zagreb manages epidermolysis bullosa patients.

In **Egypt**, 11 centres have been identified all over the Egyptian territory: 9 dermatology departments, 2 genetics departments.

In **France**, 5 centres of reference for the rare dermatological diseases, 6 centres of competence for genodermatoses and 26 centres of competence specifically for neurofibromatoses guarantee a multidisciplinary health care and are labelled by the Ministry of Health.

In **Greece**, there is a centre of reference at Athens for ichthyosis and palmoplantar keratoderma and a centre at Larisa for neurofibromatosis.

In **Italy**, there are regional and interregional centres of reference. There are 35 recognised centres for epidermolysis bullosa, 43 for congenital ichthyosis, 62 for neurofibromatosis, 31 for xeroderma pigmentosum, 38 for incontinentia pigmenti. To day, there are no interregional centre for genodermatoses.

In **Libya**, specialised consultations are organised within the 3 dermatology departments of the Tripoli Central Hospital, of Tripoli Medical Center and Gamhoria hospital.

In **Morocco**, the identification of centres of reference is part of the objectives of the National Programme against Leprosy and Dermatological Diseases. The official recognition of these centres of reference concern the University Hospitals of Rabat and Casablanca and should also concern those of Fès and Marrakech.

In **Portugal**, a centre of reference for genodermatoses has been developed with the creation of a multidisciplinary consultation for genodermatoses once a week at the Santa Maria Hospital at Lisbon.

In **Romania**, two centres of reference exist for the health care of epidermolysis bullosa, one at Brasov and one at Cluj-Napoca.

In **Slovenia**, all the children affected by genodermatoses are managed in the department of Dermatology and Venerology of Ljubljana with a multidisciplinary approach, this department also manages adults affected by epidermolysis bullosa. Adults affected by other genodermatoses than epidermolysis bullosa are managed by the 3 other dermatology departments.

In the **Palestinian Territories**, patients are seen by dermatologists in the framework of general consultation of dermatology.

In **Tunisia**, the centre of expertise for the xeroderma pigmentosum is located at the Habib Thameur hospital at Tunis. Patients affected by other genodermatoses are seen in specialised consultations in pediatric dermatology at the Charles Nicolle Hospital, Children Hospital, Military Hospital (Tunis). A specialised consultation in pediatric dermatology has been created in 2009 at La Rabta Hospital. There are also 3 medical genetics departments (Tunis, Sousse and Sfax). There are 3 laboratories of molecular biology in Tunis (Institut Pasteur, Faculty of Medecine, Private), 1 in Sousse and 1 in Sfax.

In **Yemen**, patients are seen in the dermatology department of Taiz.

	Epidermolysis Bullosa (EB)	Ichthyosis (I)	Neurofibromatosis (NF)	Palmoplantar Keratoderma (KPP)	Xeroderma Pigmentosum (XP)	Other
Algeria	2					
Cyprus	1*					
Croatia	1*					
Egypt	11 under development					
France	5* + 6 ** + 26 ** for NF					
Greece	2					
Italy	35*	43*	62*		31*	Incontinentia Pigmenti : 38*
Libya	3					
Morocco	2					
Portugal	1*					
Romania	2					

	Epidermolysis Bullosa (EB)	Ichthyosis (I)	Neurofibromatosis (NF)	Palmoplantar Keratoderma (KPP)	Xeroderma Pigmentosum (XP)	Other
Slovenia	1* + 3					
Palestinian Territories						
Tunisia	4 + 1* for XP					
Yemen	1					

Number of consultations managing patients of the 6 groups of diseases targeted by the partners
** : Centre of reference ** : Centre of competence*

Patients' organisations play a key role in the improvment of the health care
Patients' organisation play a key role in the evolution of patients' health care. The development of these organisations relies on:

- the will of patients to develop these organisations,

- the support of doctors, for the identification of patients and in some countries their implication (the participation of doctors is necessary to the creation of organisations in some countries),
- the general political context favouring or hindering the development of organisations.

Due to the low number of patients and the lack of organisations in some countries, existing organisations are often in touch with patients from other countries. In some cases, they can favour and support the creation of organisations of patients abroad. In this way, Les Enfants de la Lune, the French xeroderma pigmentosum patients' organisation has supported the creation of the Tunisian organisation for xeroderma pigmentosum patients. Some organisations are structured internationally like the epidermolysis bullosa patients' organisations, through the international Debra network.

In **Algeria**, associations tend to develop and patients' organisations start to be created for metabolic and dermatologic diseases. Organisations support hospitalized children and patients affected by chronic diseases, including genodermatoses patients. A volunteer group at Oran (West Algeria) brings its help and support to xeroderma pigmentosum patients.

In **Cyprus**, due to the low number of patients because of the small population, there is no organisation but the awareness of the patients to the importance of these organisations is being raised.

In **Croatia**, Debra Croatia is the organisation for epidermolysis bullosa patients. Patients affected by rare diseases are gathered within the Croatian Association for Rare Diseases Patients. Patients' organisations are gathered in a federation, the coalition of health organisations.

In **Egypt**, the mother of an epidermolysis bullosa patient brings her support through donations to families.

In **France**, severe genodermatoses patients' organisations are very active and well developed. Epidermolysis bullosa patients are gathered within the organisation: Epidermolyse Bulleuse Association d'Entraide, www.ebae.org; the ichthyosis patients within 2 organisations: l'Association Nationale des Ichtyoses et Peaux Sèches Pathologiques, ANIPS, www.anips.net and Athina based in Monaco, www.aaimonaco.org; palmoplantar keratoderma patients within the Association Pachyonichie Congénitale, www.pachyonychie-congenitale-lecoeuraupied.com; neurofibromatosis patients within 2 organisations: l'association Recklinghausen, www.anrfrance.org and the Ligue Française Contre les Neurofibromatoses, asso.orpha.net/LFCN/cgi-bin; ectodermal dysplasia patients within the Association Française des Dysplasies Ectodermiques, www.afde.net; Incontinentia Pigmenti patients within the IPF, www.incontinentiapigmenti.fr; the xeroderma pigmentosum patients within Les Enfants de la Lune, http://asso.orpha.net/AXP/debut.htm.

In **Greece**, genodermatoses patients' organisations are not developed. Patients affected by genetic diseases are gathered in the TO MELLON Organisation, an organisation dedicated to genetic diseases patients. Rare diseases patients' organisations are gathered within PESPA, the Greek alliance of rare diseases.

In **Italy**, severe genodermatoses patients' organisations are very active and well developed. Epidermolysis bullosa patients are gathered within DEBRA Italy, www.debraitaliaonlus.org; ichthyosis patients within UNITI, www.ittiosi.it; neurofibromatosis patients within 5 organisations: ANANas, www.ananasonline.it; ANF www.neurofibromatosi.org, IOCISONO www.associazioneiocisono.it; LINFA www.associazionelinfa.it; AMINF www.associazioni.milano.it/itsos/aminf/index.html; ectodermal dysplasia patients within ASSOANDE, www.assoande.it.

In **Libya**, there is no patients' organisation for severe genodermatoses. Social support is brought through the following organisations: Safe childhood and the International African Arab Organization for Disabled Rights (IAAODR). Collaborations will be developed with Waatasimo, Shabab Libya Organization, Amal Orphan Association.

In **Morocco**, the Moroccan Genodermatoses Organisation (AMDG) has been created in Rabat by doctors to improve the health and social care of patiens.

In **Portugal**, the Portuguese Neurofibromatoses Association (APNF) represents neurofibromatosis patients. RARÍSSIMAS gathers rare diseases patients' organisations. There is a Portuguese organisation for epidermolysis bullosa patients. To develop its activites, collaborations are in process with Spain to train specialists, nurses, patients and families in the aim of creating a DEBRA organisation in Portugal.

In **Romania**, epidermolysis bullosa patients are gathered in the Mini Debra Romania organisation.

In **Slovenia,** epidermolysis bullosa patients are gathered in the Debra Slovenia organisation.

In the **Palestinian Territories**, patients' organisations are not developed.

In **Tunisia**, in 2009 with the help of the Enfants de la Lune (the French organisation for the xeroderma pigmentosum patients) an organisation for xeroderma pigmentosum patients has been created. An epidermolysis bullosa organisation is under process in Sfax. A genodermatoses organisation is under process in Tunis.

In **Yemen**, patients' organisations are not developed.

	Epidermolysis Bullosa (EB)	Ichthyosis (I)	Neurofibromatosis (NF)	Palmoplantar Keratoderma (KPP)	Xeroderma Pigmentosum (XP)	Others
Algeria		under process			under process	
Cyprus						
Croatia	Debra					
Egypt	under process					
France	EBAE	ANIPS, ATHINA	Recklinghausen, Ligue Française Contre les Neurofibromatoses	Association Pachyonychie Congénitale	Les enfants de la Lune	AFDE IPF
Greece						
Italy	DEBRA	UNITI	ANANas, ANF, IOCISONO, LINFA, AMINF			ASSOANDE
Libya						
Morocco						Association Marocaine des Génoderma-toses
Portugal	under process		APNF			

	Epidermolysis Bullosa (EB)	Ichthyosis (I)	Neurofibromatosis (NF)	Palmoplantar Keratoderma (KPP)	Xeroderma Pigmentosum (XP)	Others
Romania	Mini Debra					
Slovenia	Debra					
Palestinian Territories						
Tunisia	under process				Association d'Aide Aux Enfants Atteints de Xeroderma Pigmentosum	Association Génoderma-toses
Yemen						

Patients' organisations for the 6 groups of diseases targeted by the partners

Towards a multidisciplinary care of genodermatoses and the development of recommendations adapted to each country's realities.

Each country develops its own health care strategy. This health care favours a multidisciplinary approach. It is based on the recommendations already developed in some countries and adapted to the local realities. Epidermolysis bullosa are the pathologies for which there are the most recommendations. One of the aims of *Génodermatoses et Méditerranée - Together Against Genodermatoses* is to develop health care recommendations adapted to the realities of each country, for the 6 groups of diseases targeted by the partners.

In **Algeria**, health care relies on French recommendations, some patients are treated in France in the framework of an agreement between France and Algeria.

In **Cyprus**, patients are managed according to a multidisciplinary approach (discussion on the health care strategies essentially between dermatologists and other specialists such as neurologists, neurosurgeons and oncologists).

In **Croatia**, the specialised centre for epidermolysis bullosa care of Zagreb works in close collaboration with Debra.

In **Egypt**, recommendations for health care have been developed for dystrophic epidermolysis bullosa, xeroderma pigmentosum and ichthyosis.

In **France**, a national protocol for diagnosis and care has been elaborated for the health care of xeroderma pigmentosum patients. The elaboration of two national protocols for diagnosis and care for epidermolysis bullosa and ectodermal dysplasia is under process. A consultation for diagnosis and health care is held once a week by the doctors of the MAGEC team:. this consultation provides diagnosis and management of different types of ihnerited epidermolysis bullosa, ectodermal dysplasia and incontinentia pigmenti, some ichthyosis and palmoplantar keratoderma, cutis laxa and xeroderma pigmentosum.

In **Greece**, recommendations for diagnosis of epidermolysis bullosa and neurofibromatosis are available in each department of dermatology.

In **Italy**, the national centre for rare diseases is in charge of establishing recommendations for diagnosis and care. These recommendations are prepared by a group of experts according to the Delphi method. A group of 18 experts (dermatologists, pediatricians, geneticists, molecular biologists, ethic specialists, representatives of patients' organisations) is working since 2007 on recommendations for the health care of epidermolysis bullosa. This document is being finalized.

In **Libya**, patients are managed according to a multidisciplinary approach involving pediatricians, pediatric surgeons, ophtalmologists, orthodontists, maxillofacial surgeons, pharmacists, urologists, neurologists, plastic surgeons.

In **Morocco**, a health care strategy specific to priority severe genodermatoses and adapted to the national context and the available resources is planned with the definition of recommendations for health care and a handbook of procedures.

In **Portuga**l, patients are managed according to a multidisciplinary approach at the Santa Maria Hospital of Lisbon.

In **Romania**, epidermolysis bullosa patients are managed by a multidisciplinary team (dermatologist, pediatrician, surgeon, geneticist) at Brasov and Cluj-Napoca. A centre for molecular diagnosis has been developed in the dermatology department of the University of Medecine of Cluj-Napoca.

In **Slovenia**, since 2009 a multidisciplinary team (dermatologists, pediatricians, geneticists, ophtalmologists, dentists, plastic surgeons, internists, psychologists) guarantees the health care of epidermolysis bullosa patients. Other genodermatoses patients are managed in the different dermatology departments of the country, according to a multidisciplinary approach. For the most severely affected patients, care is given at home by nurses.

In the **Palestinian Territories**, patients are seen in dermatology consultations.

In **Tunisia**, numerous initiatives to improve health care are set up in different dermatology departments in particular in the fields of education and training of patients and families, training of care personnel, psychological support, making available cosmetics and medical devices.

In **Yemen**, patients are seen in the framework of dermatology consultations.

The development of a national network

The development of a national network relies on already existing networks (such as the Leprosy network in Morocco or the sentinel doctors network in Algeria) or on networks set up in the framework of rare diseases plans with labelling of centres of reference and centres of competence. The notion of network is more or less relevant according to the size of the country. Collaborations between different centres have to be developed.

In **Algeria**, a sentinel netwrok has been introduced in 2006 and reactivated in 2009 with the involvment of pediatricians.

In **Cyprus**, the different actors of the health care (essentially geneticists, dermatologists, neurologists, neurosurgeons, oncologists) discuss the development of a national network with a more systematic multidisciplinary approach.

In **Croatia**, a national plan for the health care of rare diseases patients is under development with the first conference for Rare Diseases in Croatia in the framework of Europlan.

In **Egypt**, a website is developed to facilitate communication between different hospitals and information of doctors and patients. This website, www.genodermatosis.eg.net, has been developed by ENSTINET (Egyptian National STI Network).

In **France**, in the framework of the first rare diseases national plan, national care network have been organised through the identification and labelling of centres of reference and centres of competence. The cooperation between these centres should be reinforced. The list of the centres of reference is available on the website of the ministry of health and on the MAGEC website: www.magec.eu.

In **Greece**, a national network is under development with the introducing of two centres of reference at Athens, one for ichthyosis and palmoplantar keratoderma, one at Larisa for neurofibromatoses.

In **Italy**, the development of a national network falls into the framework of the rare diseases plan. A collaborative national network for epidermolysis bullosa has been developed these last years. Collaborative links exist in the other genodermatoses field and must be developed. The recent creation of the Italian Organisation of Hospital Dermatologists (ADOI - Associazione Dermatologi Ospedalieri Italiani) will enable the development of networks for other genodermatoses, thanks to its rare diseases working group of 27 members.

In **Libya**, coordinators have been identified for each group of pathologies targeted by Libyan doctors (epidermolysis bullosa, ichthyosis, palmoplantar keratoderma, Darier disease, neurofibromatosis, tuberous sclerosis and xeroderma pigmentosum) and for the data collection. A coordinator has been identified in the field of international collaborations and for the development of a website.

In **Morocco**, the project of a national network for severe genodermatoses falls into the scope of the action plan of the Ministry of Health for 2008-2012. This plan aims at the preservation of the achievements of the anti-Leprosy plan in a decentralised framework ; the answer to the other public health dermatological needs and the development of dermatology services. The partners of the Ministry of Health[1] programme are the Moroccan Organisation of Dermatology, the Moroccan Organisation of Pediatric Dermatology (SMDP), Gildi, the Moroccan Organisation of Genodermatoses (AMDG) and the Moroccan Organisation of Psoriasis (AMP). The hospitals concerned are those of Rabat and Casablanca but also those of Fès and Marrakech.

In **Portugal**, a national network will be organised around the centre of reference of the Santa Maria Hospital of Lisbon. It will enable an adapted health care for all genodermatoses patients. Dermatologists, general practitioners, geneticists, and pediatricians working in private practice or in hospital will be contacted in order to make them refer their severe genodermatoses patients to the genetic consultation of their region.

In **Romania**, the centre of reference for epidermolysis bullosa of Cluj-Napoca works in close collaboration with the centre of Brasov and Debra's organisation.

In **Slovenia**, the network is organised between the 2 university hospital departments of dermatology and 2 other hospitals. The cooperation with social workers is well organised.

In the **Palestinian Territories**, in 2009, a presentation about genodermatoses patients has been made at the Palestinian Medical Congress, seeking the involvment of all the medical personnel to improve the management of severe genodermatoses patients. The representative of the Ministry of Health commited himself on creating a committee of dermatology and pediatrics experts to handle this question. Meanwhile, a committee for the management of genodermatoses patients has been created during the annual dermatology meeting.

In **Tunisia**, the collaboration between the departments of dermatology has been reinforced, with, among others, the development of a closer collaboration between the departments of dermatology, genetics and molecular biology (Institut Pasteur, department of genetics of Charles Nicolle Hospital and the different departments of dermatology of Tunis).

In **Yemen**, the development of a network for the management of patients is linked to the improvment of access to primary health care.

The development of international collaborations

• Collaborations in the fields of diagnosis, health care, training and research

These collaborations exist in the fields of diagnosis, health care, training and research. Collaborations in the health care field are developed with the management of patients, under some conditions, in countries providing care which are not available in the native country. Collaborations with South countries are developed through *Génodermatoses et Méditerranée*.

• Collaborations between the different centres

At a European level, structured collaborations exist through the Geneskin network. Algeria, Morocco, Tunisia and France have also developed solid collaborations.

• Collaborations between the patients' organisations of different countries

European and international patients' organisations network are being developed. Debra international is a very structured network working really well. Ectodermal dysplasia patients' organisations also meet at an international level. Ichthyosis patients' organisations gather at a European level. Les Enfants de la Lune, the French organisation for xeroderma pigmentosum patients works closely with the Maghreb countries with the support to creation of the Tunisian organisation for xeroderma pigmentosum patients. Les Enfants de la Lune also has special links with the English organisation.

Algeria has collaborations especially with France in the fields of research and health care with the management of patients in France (special agreement between France and Algeria). International collaborations are developing through the *Génodermatoses et Méditerranée* and TAG networks.

[1] Head of Epidemiology and struggle against diseases, Department of Dermatological diseases

In **Cyprus**, collaborations are essentially developed at the European level, through the *TAG* project.

In **Croatia**, Debra Croatia has organised the 1st Regional Symposium (Mediterranean – Central Europe- East Europe) on Inherited Epidermolysis Bullosa in 2009.

In **Egypt**, collaborations are developed through the *Génodermatoses et Méditerranée* network.

In **France**, solid international collaborations exist with the Maghreb countries and are developed through the *Geneskin*, *Génodermatoses et Méditerranée* and *TAG* networks.

In **Greece**, international collaborations are developed through the *Génodermatoses et Méditerranée* and *TAG* networks.

In **Italy**, Prof. Zambruno initiated and coordinated from 2005 to 2008 a European Union financed project: *Geneskin* (Rare genetic skin diseases: advancing diagnosis, management and awareness through a European network). This network enabled the development of a European network for 5 major groups of genodermatoses.

In **Libya**, collaborations are developed through the *Génodermatoses et Méditerranée* and *TAG* networks.

In **Morocco**, collaborations are developed through the *Génodermatoses et Méditerranée* and *TAG* networks.

Portugal wants to initiate international collaborations with centres of reference to improve diagnosis and multidisciplinary care.

In **Romania**, exchange programmes for training exist at a European level. Cooperations also exist in the field of diagnosis and treatment. These cooperations are developed with other centres of expertise such as the Department of Dermatology of Freiburg of the Prof. Dr. Leena Bruckner-Tuderman. For the severe diseases that cannot be treated in Romania, patients can, under some conditions, be treated in another country.

In **Slovenia**, epidermolysis bullosa patients' organisations from Croatia and Slovenia (Debra Croatia and Debra Slovenia) organised joint meetings. Contacts with the Derpartment of Dermatology and Molecular Dermatology of Freiburg in Germany are being developed for the genetic analysis of junctional and dystrophic epidermolysis bullosa patients. In the field of training, 2 dermatologists and 2 nurses participated in symposiums on epidermolysis bullosa in United Kingdom and in Croatia in 2009.

In the **Palestinian Territories**, collaborations are developed through the *Génodermatoses et Méditerranée* and *TAG* networks.

In **Tunisia**, several teams have contacts and personal collaborations with foreign teams, essentially French ones. International collaborations are developed through the *Génodermatoses et Méditerranée* and *TAG* networks.

In **Yemen,** international collaborations are developed through the *Génodermatoses et Méditerranée* and *TAG* networks.

A state of the art in 15 countries

Each country has presented, when they were available, the following data on the 6 groups of diseases targeted by the partners:

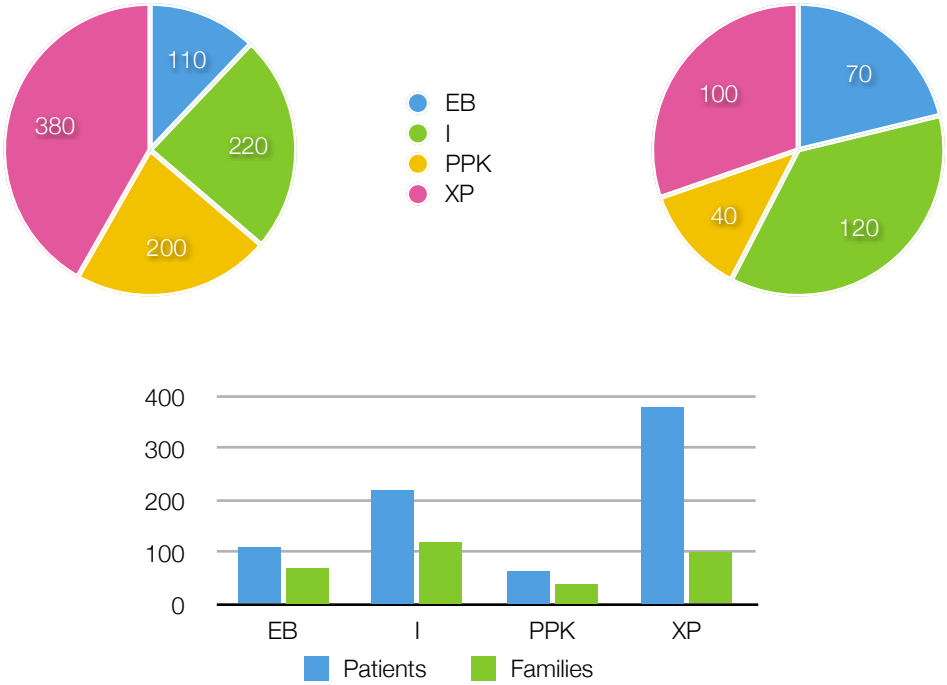
- Epidemiological data,
- Progress in the evaluation of needs of the patients and the costs of the disease for the patients,
- Progress in the development of centres of expertise,
- Progress in the development of patients' organisations,
- Strategies for the health care of genodermatoses specific to each country,
- The development of a national network,
- The development of European and international collaborations.

Algeria

Bakar Bouadjar, Department of Dermatology, University Hospital of Bab El Oued, Algiers

Collected data in the 2 university hospital departments of dermatology of Algiers

Available data come from the University Hospitals' dermatology departments of Algiers, Bab El Oued and Mustapha. Generally, the recruitment of patients increased with the reactivation of the sentinel network introduced in Algeria. Other genodermatoses for which data have not been communicated are present in Algeria: Kindler syndrom, Papillon-Lefèvre syndrom, Chanarin Dorfman Syndrom.



Number of patients and families with Epidermolysis Bullosa (EB), Recessive Ichthyosis(I), Mal de Meleda (PPK), Neurofibromatosis (NF), Xeroderma Pigmentosum (XP)

Disease	Number of patients	Number of families
Inherited Epidermolysis Bullosa	110	70
Épidermodysplasies verruciformes	26	22
Recessive Ichtyosis	220	120
Mal de Meleda	65	40
Scléroses Tubéreuses de Bourneville	28	10
Xeroderma Pigmentosum	380	100
Total	829	362

A sentinel network to point out the severe genodermatoses cases in link with dermatologists and pediatricians.

A medical sentinel network has been introduced in Algeria to point out the severe genodermatoses cases and address them to dermatology departments of the University Hospitals of Algiers. In the meantime, pediatrician are now involved in the management of youg severe genodermatoses patients.

A network for the management of xeroderma pigmentosum in Algiers

Sentinel doctors within the country are in touch with the departments of dermatology of the University Hospitals of Bab El oued and Mustapha for the health care of patients. This management is made in collaboration with the plastic and reconstructive surgery departments of Prof. Joucdar and Prof. Mitiche.

A budget of more than 200 000 euros for the purchase of drugs for genodermatoses promised in 2008

A budget of 19 565 000 DA = 207 867 euros has been granted by the Head of Finances of the Ministry of Health to the 2 Unversity Hospitals' dermatology departments of Algiers, Bab El Oued and Mustapha to buy drugs. This budget has been assessed as follow:

- Inherited Epidermolysis bullosa: 125 000 dinars (1 329 euros) for 50 patients = 225 euros per patient and per year,
- Ichthyosis: 12 320 000 dinars (130 893 euros) for 220 patients = around 600 euros per patient and per year,
- Palmoplantar Keratoderma: 1 400 000 dinars (14 474 euros) for 150 patients = around 100 euros per patient and per year,
- Xeroderma Pigmentosum : 5 720 000 dinars (60 772 euros) for 200 patients = around 300 euros per patient and per year.

This budget is said to be renewable each year with the possibility of increasing its amount if necessary. But, to date, the budget has not been allocated yet.

A budget of 125 000 euros for antenatal diagnosis of xeroderma pigmentosum

A budget has been granted for equipping and the reagents of laboratories whose activity is focused on diagnosis and genetic studies of rare diseases. This budget has essentially be used for antenatal diagnosis of xeroderma pigmentosum. The assessment of the budget has been made in collaboration with Dr. Zghal from the University Hospital of Habib Thameur in Tunisia.

Organisations to help patients and families

• Organisations to help patients

Organisations support hospitalized children and chronic diseases patients:

- Le Souk, an organisation of medical students

Le Souk accompanies chronical diseases patients. This organisation has already organised activities for xeroderma pigmentosum and ichthyosis patients. Their website: www.lesouk.org

- Amine, an organisation for hospitalized children

Amine is located in the University Hospital of Bab El Oued. It aims at helping hospitalized children.

• Organisations and volunteer groups bring support to dermatological and metabolic diseases patients

Organisations of patients are developing in Algeria with the involvment of doctors. But there is no severe genodermatoses patients' organisation yet.

- Organisation of metabolic diseases patients' parents
- Psoriasis organisation
- Xeroderma Pigmentosum Algeria: a volunteer group at Oran (West of Algeria) brings its help and support to xeroderma pigmentosum patients. www.xeroderma pigmentosumalgerie.org

Croatia

Mihael Skerlev, Department of Dermatology and Venerology, University Hospital and Faculty of Medecine, Zagreb University and Mrs. Vlasta Zmazek, DEBRA, Croatia, Zagreb

Services for epidermolysis bullosa patients are well developed thanks to a great mobilisation of Debra Croatia helped by the medical personnel

• Debra Croatia fights for a better health care of epidermolysis bullosa patients

In 2008-2009, Debra Croatia fought for the health care of patients above the age of 18. Debra Croatia develops numerous activities for patients and families: improvement of the daily life of patients through actions of education, social insertion, general public awareness raising through medias, organisation of exhibition and gala, a book for children.Their website: www.debra-croatia.com.

• Debra Croatia works in synergy with rare diseases patients' organisation

Debra Croatia is a member of Eurordis, of the Croatian Association of rare diseases patients, of the Croatian federation of health organisations, of the *Génodermatoses et Méditerranée* and *TAG* networks. Debra Croatia is member of Debra International and has very close links with Debra Bosnia and Herzegovina as well as Debra Serbia.

• The 1st Regional Symposium (Mediterranean- Central Europe- East Europe) on Inherited Epidermolysis Bullosa took place in Zagreb

The 1st Regional Symposium on Inherited Epidermolysis Bullosa (Mediterranean- Central Europe- East Europe) took place in Zagreb on April 3-4, 2009. It was organised by the Association of Dermatology and Venerology of the Croatian Medical Organisation, by Debra Croatia and the centre of reference on Epidermolysis Bullosa of the Ministry of Health and Social Protection.

The centre of reference for the epidermolysis bullosa within the Department of Dermatology and Venerology, ofthe University Hospital of Zagreb

This centre has been created in May 2008.

The multidisciplinary committee for the health care of epidermolysis bullosa of the Ministry of Health and Social Protection

This committee is co-ordinated by Dr. Slobodna Murat-Sušić, dermatologist-venerologist and pediatrician involved in the management of epidermolysis bullosa.

Towards a national plan for rare diseases

Debra Croatia is member of the Association for Rare Diseases of the Croatian Medical Organisation. In 2010, the 1st conference on rare diseases was held in Croatia in the framework of the Europlan project which aims at promoting the development of national plans for rare diseases.

Cyprus

Violetta Christophidou Anastasiadou, Pediatrician-Geneticist, Clinical Genetics Consultation of the Cyprus Institute of Neurology and Genetics, Makarios Hospital, Nicosia

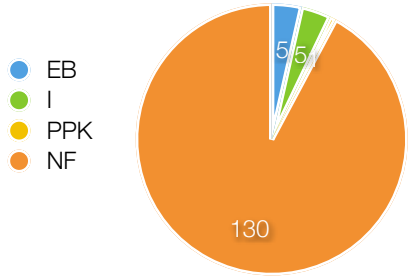
In Cyprus, the number of patients is very low number. This is mainly due to the low population on the island (around 1 million inhabitants).

The data of the register of the Clinical Genetics Consultation of the Cyprus Institute of Neurology and Genetics

Presented epidemiological data are from the register of the Clinical Genetics Consultation of the Cyprus Institute of Neurology and Genetics, Makarios Hospital. A significative number of genodermatoses patients are referred to this consultation. Patients are also seen by their dermatologist and other specialists. The following genodermatoses have been spotted:

- Xeroderma Pigmentosum and genetic diseases of the DNA repair: a few families
- Anhidrotic Ectodermal Dysplasia: a few families
- Incontinentia Pigmenti : a few families
- Other genodermatoses : a few patients

Disease	Number of patients
Epidermolysis Bullosa	5
Severe Ichtyosis	5
Palmoplantar Keratoderma	1
Neurofibromatosis	130



Number of cases of Epidermolysis Bullosa (EB), Severe Icthyosis(I), Palmoplantar Keratoderma(PPK), Neurofibromatosis(NF)

The genetic consultation is a centre of reference

The Genetic Consultation is a centre of reference. The personnel is composed of a geneticist nurse, a geneticist, and genetic counselors. The Genetics Department provides genetic diagnosis, treatment, genetic counseling. The Genetics Department also gives lectures in Cyprus and abroad. This department has research activities. It also makes awareness raising activities.

This consultation is a centre of reference and the only one on the island, so the patients of the whole island are addressed there for every type of genetic diseases and for every question linked to diagnosis, health care, or genetic counseling. The consultation offers its services to patients of all ages, to couples and families. Due to the small population on the island, the development of centres of expertise for each disease is not the appropriate solution.

The patients' needs are essentially managed by the public health services

The patients' needs are essentially managed by the public health services. A state of the art is in process to better evaluate these needs and develop strategies to improve the health care of severe genodermatoses patients.

Develop a patient organisation: a need expressed by the patients and families

Due to the very low number of patients, there is no patients' organisation for severe genodermatoses but the patients express this need.

Strategy for genodermatoses health care and development of a national network

The development of a national network favouring a multidisciplinary health care is under discussion with dermatologists and other specialists including neurologists, neurosurgeons and oncologists.

Egypt

Presentation has been made by the Dr. El Gamal, Faculty of medecine of Damiette, University Al-Azhar, Egyp. He coordinates the Egyptian Genodermatoses Group.

The *Egyptian Genodermatoses Group* grows and develops its website

• 5 departments of dermatology join the Egyptian Genodermatoses Group

The *Egyptian Genodermatoses Group* is coordinated by the Dr. El-Gamal. 5 departments of dermatology join the Egyptian Genodermatoses Group: the dermatology departments of the Faculties of Medecine of Alexandria, Damiette, Assiout, Minieh and Sohag. Assiout, Minieh and Sohag are in the South of Egypt, a region with a high number of patients due to consaguinity, among other. These centres have been identified as the future centres of reference of the region.

Les centres identifiés en Egypte sont les suivants :

- National Research Center (NRC), Cairo: Dept. of Clinical and Molecular Genetics, Dept. of Dermatology
- Medical Research Institute, Alexandria: Dept. of Genetics
- Kasr Alainy Faculty of Medecine, Cairo University: Dept. of Dermatology
- El-Haud el Marsoud Hospital, Cairo: genodermatoses outpatients
- Shark Al Medina Hospital, Alexandria: Dept. of Dermatology
- Ain Shams Faculty of Medecine: Dept. of Dermatology
- Menia Faculty of Medecine: Dept. of Dermatology
- Sohag Faculty of Medecine: Dept. of Dermatology
- Assiut Faculty of Medecine: Dept. of Dermatology
- Damietta Faculty of Medecine: Dept. of Dermatology
- Alexandria Faculty of Medecine: Dept. of Dermatology

• The website of the Egyptian Genodermatoses Group : www.genodermatosis.eg.net

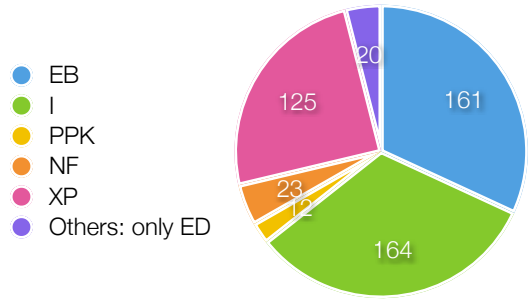
Dr. Amin Sharobim from the Dermatology Department of the National Research Centre in Cairo has developed the website of the *Egyptian Genodermatoses group* with the support of ENSTINET (Egyptian National STI Network), Prof. Ghada El Kamah and other members of the group.

Epidemiological data have been collected in 6 partner centres of the Egyptian Genodermatosis Group

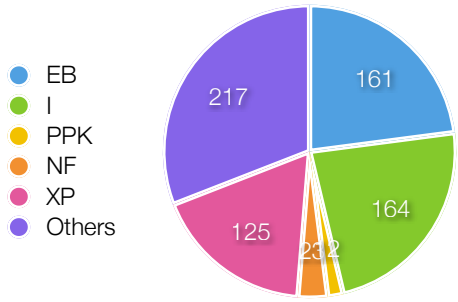
Data have been collected in the following centres:

- Kasr Alainy Faculty Hospital, Cairo University: Dept. of Dermatology
- National Research Center (NRC), Cairo
- El-Haud el Marsoud Hospital, Cairo: genodermatoses outpatients
- Damietta Faculty of Medecine: Dept. of Dermatology
- Alexandria Faculty of Medecine: Dept. of Dermatology
- Medical Research Institute, Alexandria: Dept. of Genetics

Unisvertisy Hopital / Health Center	EB	I	NF	PPK	XP	Others	Total
Kasr Alainy Faculty Hospital, Cairo University	130	55	0	0	97	0	282
National Research Center (NRC), Cairo	7	7	6	0	3	26 including 12 ectodermal dysplasia	49
El-Haud el Marsoud Hospital, Cairo	22	33	0	12	12	171	250
Damietta Faculty of Medecine	2	2	0	0	5	3	12
Alexandria Faculty of Medecine	2	59	15	0	8	7	91
Medical Research Institute, Alexandria	3	8	2	0	0	10 including 8 ectodermal dysplasia	23
Total	166	164	23	12	125	217 including 20 ectodermal dysplasia	707



Numbers of cases of Epidermolysis Bullosa (EB), Recessive Icthyoses(I), Palmoplantar Keratoderma(PPK), Neurofibromatoses (NF), Xeroderma Pigmentosum (X) and Ectodermal Dysplasia (Others: only ED)



Numbers of cases of Epidermolysis Bullosa (EB), Recessive Icthyoses(I), Palmoplantar Keratoderma(PPK), Neurofibromatoses (NF), Xeroderma Pigmentosum (X) and other genodermatoses (Others)

Health care strategy for Recessive Dystrophic Epidermolysis Bullosa, Lamellar Ichthyosis and Xeroderma Pigmentosum

Genodermatoses are managed according to the following protocols:

• Clinical trials using cell therapy for the health care of Recessive Dystrophic epidermolysis Bullosa

Prof. El-Darouti (Kasr Al Aini Hospital, Cairo) is involved in clinical trials in RDEB patients (89) using cell based therapies. He has performed intravenous administration of non-hematopoetic bone marrow stem cells (BMNHSC), from allogeneic matching donors. The medical treatment is modified (therapeutic rotation: retinoids and topical phenytoin) Prof. El Darouti was invited in US to discuss EBD treatment with 13 experts on February 20, 2009, "Genetics, Stem Cell Biology and Stem Cell Transplantation in Epidermolysis Bullosa"

• Xeroderma Pigmentosum health care

- Systemic retinoids, daily
- Systemic cyclo-oxygenase inhibitors, daily
- Topical 5 FU bid for 4 weeks repeated every 6 months
- For SCC, BCC and SK: Shave excision and postoperative INF sublesional 3x/ week for 8 weeks.

• Ichthyosis health care (lamellar ichthyosis and ichthyosiform erythroderma)

- Acitretin 0.3 mg/kg/day
- Sodium bicarbonate baths for 20 minutes twice a day followed by urea 10% cream application
- Tazarotene application(whole body in the evening to be washed next morning) 2x/week.
- Sugar paste application(as in hair removal) to most noticeable areas once every 2 weeks

Training in Cairo and Alexandria for general practitioners, pediatricians and dermatologists

In Cairo, an agreement has been signed with ENSTINET, the I'Egyptian National STI Network to organise training sessions for general practitioners, pediatricians and dermatologists. In Alexandria, the Association of Dermatology of Alexandria and the charity association Caritas-Alex got the official agreement to organise similar training sessions.

A scientific collaboration in Egypt in the genodermatoses field

This collaboration is developing between 2 institutions: the University Hospital of Kasr Al Aini (Prof. El Darouti) and the National Research Centre (Prof. Ghada El Kamah).

Financial and social support for epidermolysis bullosa patients

Mrs Sadat, Yasmin's mother, an epidermolysis bullosa patient, has set up several actions to help people with epidermolysis bullosa in Egypt.
Mrs El Sadat presented these actions at the 2006, 2007 and 2008 working sessions.
She works with Prof. El-Darouti to develop a severe genodermatoses patients association.

ZOOM>>> The experience of Yasmin: fighting against Epidermolysis Bullosa in Egypt

Yasmin is 13 years old. She is the youngest artist in Egypt. Her second painting exhibition took place on March 21-29 2009.
Being a recessive dystrophic epidermolysis bullosa patient, she raises the awareness of general public throug medias, her friends, her drawings, flyers. Yasmin has sold 350 calendars and gave the earnings to the epidermolysis bullosa kids from the Al Kasr El Aini hospital.
Learn more: www.yasminwitheb.net -You-tube: yasmin el samra yasminelsamra@hotmail.com - Face book

France

Smail Hadj-Rabia, Christine Bodemer, MAGEC, Necker-Enfants Malades, Paris, France ; Salah Ferkal, Clinical Research Docteur, Neurofibromatosis Centre of Reference, Henri Mondor Hospital

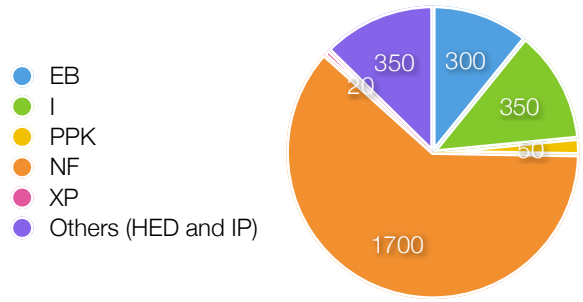
The first national plan for rare diseases (2005-2008) enabled the development of a national network organised around "centres of reference" and "centres of competence". In the framework of the first national plan for rare diseases (2005-2008), centres of reference gather university hospital teams highly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

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

CEMARA : a national database

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

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



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

5 centres of reference for rare dermatological diseases

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

6 centres of competence for genodermatoses

26 centres of competence specifically for neurofibromatosis

Costs of the disease for the patients

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

- Epidermolysis Bullosa: 1000 euros/month
- Ichthyosis: cream reimbursed by the National Health Service
- Palmoplantar keratoderma: pedicure reimbursed by the National Health Service
- Neurofibromatosis: learning disabilities covered

- Xeroderma Pigmentosum: sunscreen reimbursed by the National Health Service
- Anhidrotic Ectodermal Dysplasia: dental care reimbursed by the National Health Service under some conditions
- Incontinentia Pigmenti: reimbursed by the National Health Service under some conditions

Projects at MAGEC to improve patients' health care

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

• Epidermolysis Bullosa
The projects focus on:

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

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

• Neurofibromatosis (children)
Projects concern learning difficulties and their prevention.

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

• Anhidrotic Ectodermal Dysplasia
The writting of a National Protocol of Diagnosis and Care is in process.

• Incontinentia Pigmenti
Projects focus on the neurologic aspects of the disease.

Patients' organisations in France are well developed

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

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

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

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

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

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

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

• Xeroderma Pigmentosum

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

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

Greece

Adriana Mavrou, Prevention of Genetic Disorders, Department of Medical Genetics, Athens University School of Medicine
Alexandra Katsarou, Vassiliki Vosynioti, Anna Tagka, Angelika Roussaki, J. Bassoukas, Nicholas Stavrianeas, D. Sotiriadis, Andreas Katsambas, Greek network of Genodermatoses

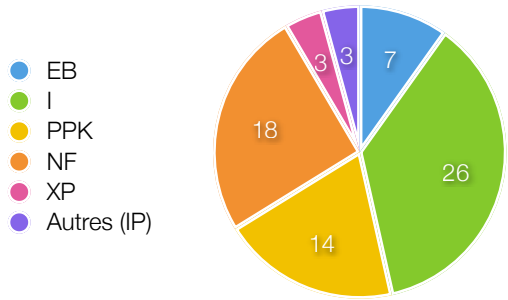
The development of an online epidemiological database

The advantages of this online database are numerous. It is an internet application: a browser is the only tool to install in each centre. For the connection, the user has an internet adress and a password. Data are kept in the same place and can be easily managed to avoid registring the same patient twice. This database is evolutive. Hospitals can access the database at the same time, which speeds up the data capture. Saving is automatic. Data are available as soon as they are registered, which enables reports on the base of the most recently registered data.

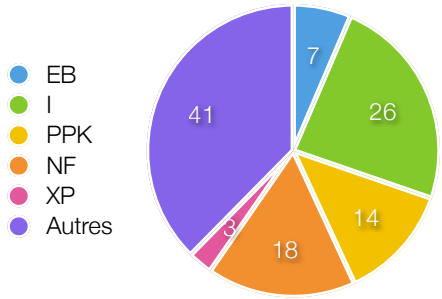
2008-2009 data from Athens and Larisa Hospitals

Disease	Number of cases
Ichthyosis	26
Epidermolysis Bullosa	7
Neurofibromatosis	18
Palmoplantar Keratoderma	14
Xeroderma Pigmentosum	3
Others*	41
Total	**109**

* Other Genodermatoses	Number of cases
Darier + Hailey-Hailey	17
Porokeratosis	13
Tuberous Sclerosis	4
Incontinentia Pigmenti	3
Ehlers-Danlos	1
Genotrichosis	3



Number of cases of epidermolysis bullosa (EB), icthyosis (I), palmoplantar keratoderma (PPK), neurofibromatosis (NF), xeroderma pigmentosum (XP) and incotinentia pigmenti (IP).



Number of cases of epidermolysis bullosa (EB), icthyosis (I), palmoplantar keratoderma (PPK), neurofibromatosis (NF), xeroderma pigmentosum (XP) and other genodermatoses.

Patients' needs

Patients' needs are: the health care by public health services, access to drugs, medical devices and cosmetics, development of centres of expertise for a better treatment, access to better care and genetic counseling, a better knowledge of the disease, of the evolution of the disease especially for children and information on new treatments. The biggest health insurance of Greece covers 10 boxes of emollient cream by month for ichthyosis patients.

Two centers of expertise

A centre of expertise for ichthyosis and plamoplantar keratoderma has been created in Athens, as well as a centre for neurofibromatosis in Larisa.

Italy

M. El Hachem, Ospedale Pediatrico Bambino Gesù, Rome ; G. Tadini, Università di Milano - Sezione, Milan ; G. Zambruno, Istituto Dermopatico dell'Immacolata - IRCCS, Rome

The health care of rare diseases in Italy is organised around the Istituto Superiore di Sanità with regional and interregional centres of reference.

The Italian legislation in the field of rare diseases is directed by the Decree n° 279 of 18/05/2001. A disease is said to be rare if, according to the requirements of the European Union, its prevalence is less than 1:2.000. The list established in Italy includes 284 rare diseases and syndroms (estimated around 5.000). This decree plans on creating a national network of regional and interregional centres of reference for diagnosis and health care of rare diseases. This decree also defines the role of the Istituto Superiore di Sanità in the field of rare diseases. This decree was supposed to be updated every 3 years, which has not been made for now.

• Regional centres of reference

Regional centres of reference are in charge of:

- Establishing the diagnosis, including biochemical and molecular, of specific diseases and communicating a certificate.
- Providing adapted care and follow up to the patients, also through a collaboration with general practitioners.
- Keeping patients' files and communicating each new case to the national register.

The regional requirements for the recognition of centres are not homogeneous. There is no follow up of the activities of centres and they don't benefit from any specific financing.

Every Italian region has spotted a regional centre of reference for rare diseases and has initiated a register for rare diseases. The list of the centres of reference is available on the website of the National Centre for Rare Diseases (http://www.iss.it/cnmr/), and for some regions on their regional website (as for example the website of the region of Emilia Romagna - http://www.saluter.it/malattierare/). The first epidemiological data should be available at the National Centre for Rare Diseases during the fall of 2009.

• Interregional centres of reference

Interregional centres of reference are in charge of:

- Coordinating the activities of regional centres and making sure the recommendations for diagnosis and therapeutic care, when available, are followed,
- Supporting general practitioner on rare diseases and available drugs,
- Participating in health personnel training and prevention of rare diseases,
- Spreading information and raising general public awareness on rare diseases and having links with patients' organisations,

To date, no interregional genodermatoses centre has been identified.

• The national centre for rare diseases of the Istituto Superiore di Sanità (ISS)

The national centre for rare diseases aims at:

- Coordinating epidemiological, clinical and research activities in the field of rare diseases,
- Gathering data of national registers for rare diseases and organising them into a national register,
- Establishing recommendations for the health care of rare diseases,
- Coordinating and granting research projects.

• The Italian Agency of Drugs: Agenzia Italiana del Farmaco

L'Agenzia Italiana del Farmaco is in charge of promoting investments in the field of research and development and to guarantee, through the simplification of drugs registration procedures, a fast access to innovating medical products and to orphan drugs for rare diseases.

• The reimbursment of rare diseases patients' care

The national care system reimburses:

- All the costs of the examinations performed to establish a diagnosis including molecular and antenatal diagnosis,

- The costs of laboratory examinations, consultations with specialists and hospitalizations
- The costs of drugs, medical devices and in some regions emollients, antiseptics, (nutritional supplements), sun screens, dressings and bandages.

Transportation is not reimbursed.

• National recommendations for health care of rare diseases patients

The national centre for rare diseases of the Istituto Superiore di Sanità is in charge of establishing recommendations for diagnosis, follow up and treatment of rare diseases. These recommendations are prepared by a group of experts following the Delphi method.

Genodermatoses recognised by the health system in Italy

• The genodermatoses targeted by the partners of the TAG project managed by the Italian health system

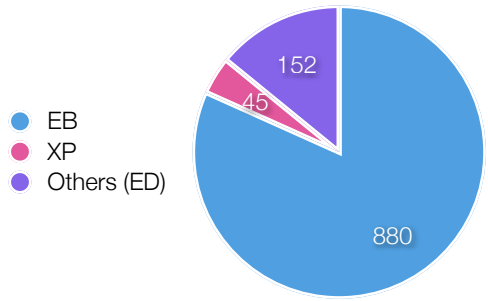
The following genodermatoses, targeted by the partners of the TAG project, are managed by the Italian national care system:

- Congenital ichthyosis (including X-linked ichthyosis, Netherton Syndrome, all the types of Lamellar Ichtyosis as well as congenital nonbullous ichtyosiform erythroderma, Harlequin syndrome, ichthyosis hystrix)
- Epidermolytic hyperkeratosis
- Progressive symmetric erythrokeratodermia
- Erythrokeratodermia variabilis
- KID Syndrome
- Sjögren-Larsson Syndrome
- Refsum Disease
- Conradi-Hunermann Syndrome
- Inherited Epidermolysis bullosa
- Xeroderma pigmentosum
- Incontinentia pigmenti
- Neurofibromatosis

• The genodermatoses targeted by the partners of the TAG project not managed by the Italian care system

Hypohydrotic Ectodermal Dysplasia and Palmoplantar Keratoderma patients cannot get free diagnosis and care (only Epidermolytic Hyperkeratose (including Epidermolytic Palmoplantar Keratoderma) and Erythrokeratoderma are managed). There is no centre of reference officially recognised and there is no data collected for these diseases.

Italian registers for Epidermolysis bullosa, Ectodermal dysplasia and Xeroderma Pigmentosum



Number of cases of Epidermolysis Bullosa (EB), Xeroderma Pigmentosum (XP) and Ectodermal Dysplasia patients (others - ED)

• The Italian register for epidermolysis bullosa and the Italian register for ectodermal dysplasia

Gianluca Tadini (Centre for Inherited Cutaneous Diseases at the Institute of Dermatological Sciences of Milano University) is in charge of the national register for inherited epidermolysis bullosa and the national register of ectodermal dysplasia. These registers have been updated in 2009.

• The Italian register for epidermolysis bullosa: 880 cases

Period	Number of cases
1991-2002	706
2003-2007	116
2008-2009	58
Total	**880**

Type	Frequency	%
Epidermolytic Epidermolysis Bullosa	258	29,3
Junctional Epidermolysis Bullosa	82	9,3
Dermolytic Epidermolysis Bullosa	524	59,5
Epidermolysis Bullosa (others)	16	1,9
Total	880	100

Italian register of epidermolysis bulllosa 1991-2009 (880 cases)

• The Italian register for ectodermal dysplasia: 152 cases

152 cases are registered including 40% confirmed by molecular diagnosis.

• The Italian register for xeroderma pigmentosum : 45 cases

Kleijer WJ, Laugel V, Berneburg M, Nardo T., Fawcett H, Gratchev A, Jaspers NGJ, Sarasin A, Stefanini M, Lehmann AR (2008) DNA Repair, 7: 744-750. Incidence of DNA repair deficiency disorders in Western-Europe: Xeroderma pigmentosum, Cockayne syndrome and Trichothiodystrophy

The incidence of Xeroderma Pigmentosum in Western Europe has been established for the first time a 2,3 on one million of viable children

Group Xeroderma Pigmentosum (XP)	Italy 30 cases	Western Europe 89 cases including the 30 Italian patients
XP-A	17 %	15,7 %
XP-B	-	-
XP-C	43 %	60,7 %
XP-D	10 %	11,2 %
XP-E	27 %	2,2 %
XP-F	3 %	4,5 %
XP-G	-	5,6 %
XP-variant	14,6 %	10,6 %

Xeroderma Pigmentosum: frequency of the NER defective complementation groups

Year of birth	Number of patients		
	Total	Autochtonic	Non autochtonic
<1940	3	3	-
1940-1959	7	7	-
1960-1969	7	7	-
1970-1979	9	8	1
1980-1989	12	11	1
1990-1999	6	5	1

Distribution in time of xeroderma pigmentosum patients living and diagnosed in Italy

Regional centres for genodermatoses health care

- Inherited Epidermolysis bullosa: 35 recognised centres
- Congenital Ichthyosis: 43 centres
- Neurofibromatosis : 62 centres
- Xeroderma pigmentosum : 31 centres
- Incontinentia pigmenti : 38 centres

Recommendations for diagnosis of inherited epidermolysis bullosa

A panel of 18 experts (dermatologists, pediatricians, geneticists, molecular biologists, ethicists and a patients' organisation's representative) has worked for 2 years on the recommendations for diagnosis of inherited epidermolysis bullosa.

The multidisciplinary health care of children affected by epidermolysis bullosa, ichthyosis, neurofibromatosis and tuberous sclerosis at the Ospedale Pediatrico Bambino Gesù, Rome

The health care of these patients is organised around a "case manager".

• Therapeutic and multidisciplinary health care of inherited epidermolysis bullosa

The case-manager is the dermatologist. The health care begins during the antenatal period. Later organ complications are infections, nutrition problems, oesophagus stenosis, dental problems, anesthesia and organ surgery: pylorus stenosis, hands, etc... In 2009, 12 patients were followed in day hospital. 45 patients were hospitalized in 2008. The hospital registered 362 admissions, 208 hospitalisations and 154 day hospital for main diagnosis and treatement of for secondary diagnosis.

Admission of EB patients	Number of admissions
Day hospital	154
Hospitalizations	208
Total	362

Number of admissions of EB patients at the Ospedale Pediatrico Bambino Gesù

Year	Number of EB patients in day hospital
2007	4
2008	11
2009	12

Number of EB patients in day hospital at the Ospedale Pediatrico Bambino Gesù

Departement	Number of EB hospitalized patients
Dermatology	19
Pediatrics	4
Intensive Care	3
Digestive Surgery	10
General Surgery	1
Infectious diseases	5
Neurology	1
Genetics	1
Plastic Surgery	1
Total	**45**

Number of EB patients hospitalisés in 2008 in Ospedale Pediatrico Bambino Gesù

• Multidisciplinary health care for neurofibromatosis patients

The case-manager is Cristina Digilio from the Department of Genetics of the Ospedale Pediatrico Bambino Gesù, Rome. She follows 20 patients per month. Involved specialists are the dermatologist, ophtalmologist, neurologist, psychologist, neurosurgeon, general surgeon, plastic surgeon, orthopedist, ENT, endocrinologist and cardiologist. The examinations performed in the follow up of the neurofibromatosis patients are the abdominal ultrasound, the blood and urine tests, the urine vanillylmandelic acid level, the mesure of the blood pressure.

The development of a national network for genodermatoses

• The working group on rare diseases of the "Associazione Dermatologi Ospedalieri Italiani"

A national network for epidermolysis bullosa has been under development for the last few years. Links are already existing for other genodermatoses but they have to be developed. The creation of a working group on rare diseases within the organisation of Italian Hospital Dermatologists (ADOI-Associazione Dermatologi Ospedalieri Italiani) should facilitate the development of networks for genodermatoses. This group is made of 27 members.

• Conventions with the Ospedale Pediatrico Bambino Gesù

- A convention between the Istituto Dermopatico dell'Immacolata (IDI) and the Ospedale Pediatrico Bambino Gesù

Since 2000, an active and close collaboration has developed between the Istituto Dermopatico dell'Immacolata (IDI) of Roma to provide a global assistance to patients without having them travelling from one centre to another. This collaboration was formalized in 2004 by a convention.

- Conventions between Debra and the Ospedale Pediatrico Bambino Gesù
In 2004, a convention was signed with DebRA Italia. In 2008, a new convention "In ospedale con Ebby" was signed to create a specific room for epidermolysis bullosa patients.

- A convention between Modena University and the Ospedale Pediatrico Bambino Gesù
In July 2008, a convention was signed with Modena University to develop gene therapy at the Ospedale Pediatrico Bambino Gesù.

Training on rare dermatological diseases and on epidermolysis bullosa

• Training on rare diseases
The next training session on rare dermatological diseases will take place in Roma, November 20-21st 2009.

• Training of specialists involved in the health care of inherited epidermolysis bullosa
The training of specialists involved in the health care of inherited epidermolysis bullosa is made thanks to a continuous collaboration between dermatologists and other specialists involved to illustrate on the job the specificity of this disease and avoid secondary complications.

Organisations for genodermatoses patients in Italy

Patients' organisations are very active and well developed:

• Organisation for epydermolysis bullosa patients
Epidermolysis bullosa are gathered within DEBRA Italy, www.debraitaliaonlus.org

• Organisation for ichthyosis patients
Ichthyosis patients are gathered within UNITI, www.ittiosi.it

• Organisations for neurofibromatosis patients
Neurofibromatosis patients are gathered within 5 organisations:

- ANANas, www.ananasonline.it
- ANF, www.neurofibromatosi.org
- IOCISONO, www.associazioneiocisono.it
- LINFA, www.associazionelinfa.it
- AMINF, www.associazioni.milano.it/itsos/aminf/index.html

• Organisation for anhidrotic ectordermal dysplasia
Anhidrotic ectodermal dysplasia patients are gathered within ASSOANDE, www.assoande.it

ZOOM >>> "In ospedale con Ebby": Debra Italy's project of equipping a room for epidermolysis bullosa patients in the Ospdale Pediatrico Bambino Gesù

Debra Italy supports epidermolysis bullosa patients through the following actions: communication of the diagnosis, organisation of consultations, participation in the development of specific recommendations, public authorities awareness raising to improve the help brought to patients, awareness raising in schools, organisation of holiday trips. Debra Italy is member of the Board of the Italian Federation of Rare Diseases - UNIAMO and of the EURORDIS board. Debra Italy signed in July 2008 a convention with the Ospedale Pediatrico Bambino Gesù: "In ospedale con Ebby". This convention will enable the equipping of a room for the epidermolysis bullosa patients who are hospitalized or coming to the day hospital. The construction is planned for 2009.

European collaborations: the *GENESKIN* network and experience

• Rare genetic skin diseases: advancing diagnosis, management and awareness through a European network

GENESKIN is an acronyme for the Rare genetic skin diseases: advancing diagnosis, management and awareness *through a European network* project. This action is coordinated by Prof. Giovanna Zambruno of the Istituto Dermopatico dell'Immacolata - IRCCS in Rome. It was financed by the European Commission for 3 years: July 1st 2005 – June 30 2008.

• GENESKIN gathered 32 clinical centres (27 organizations), research centres and patients' organisations from 12 European countries

• GENESKIN enabled the development of a European network for 5 groups of rare skin diseases

This network, through a website, helps raising the awareness on these diseases and spreading the knowledge. This network also participates in improving diagnosis and health care. It enabled ratifying new diagnosis tools, encouraging research on rare diseases. Through this network, training activities on specific groups of diseases have been organised as well as experts meetings on specific subjects (17 meetings in 8 countries gathering 665 participants in total). Finally, this network made possible the creation of a group of ethicists and the elaboration of an ethical document on genetic counseling for genodermatoses.

Libya

Prof. Benghazil, Dr. B. Zendah, Dr. K. Elhafi, Dr. S.Reyan, Department of Dermatology, Tripoli Medical Center

Prof. A. Elzurgani, Dr. N. Mormesh, Dr. A. Marsit, Dr. M. El-Hashemi, Department of Dermatology, Tripoli Central Hospital

Prof. G. Duweb, Dr. M. Bozgia, Dr. O. Bugrein, Department of Dermatology, Benghazi

Dr. A. Alahlafi, Department of Dermatology, El-Beida Hospital

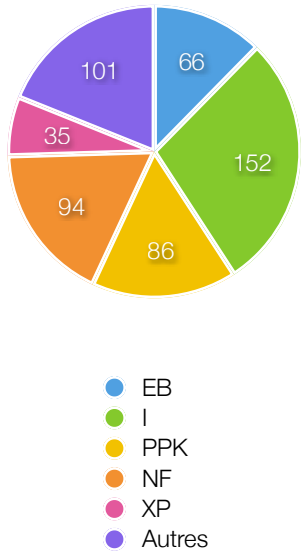
Dr. A. Lashab, Department of Dermatology, El-Khoms Hospital

Presented by Dr. Mohamed El Hashme / LGP TASK FORCE: GENO SITE.pub 4.mht

Almost 300 new cases in 2009

The collect of epidemiological data is done in a particular context. Libya's total area is 1,759,540 Km2 with a population of 6,039,000 inhabitants and a limited number of dermatologists. Data are collected in 3 regions: North West, South West and East. Data are not available in the South of the country. 560 cases have been registered: 220 at the Tripoli Central Hospital, 205 at the Tripoli Medical Centre and 135 at the Gamhoria Hospital.

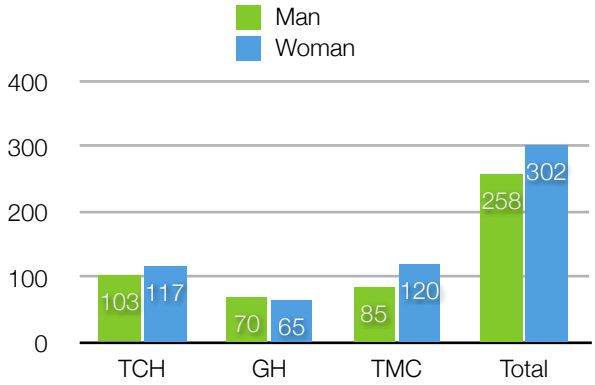
Disease / Syndrome	Tripoli Central Hospital	Triploli Medical Center	Gamhoria Hospital	Total
Ichthyosis	53	67	32	152
Neurofibomatosis	55	25	14	94
Palmoplantar Keratoderma	48	23	15	86
Xeroderma Pigmentosum	12	18	5	35
Epidermolysis Bullosa	23	23	20	66
Other Genodermatoses *	32	17	52	101
Total	**223**	**173**	**138**	**534**



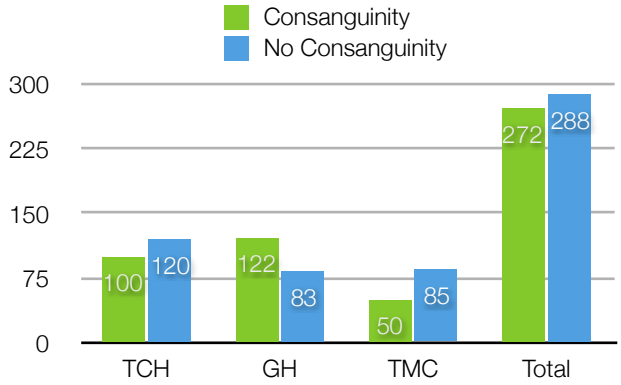
2009 Register: Number of cases of epidermolyses bullosa (EB), recessive icthyosis (I), palmoplantar keratoderma (PPK), neurofibromatosis (NF), xeroderma pigmentosum (XP) and other genodermatoses in Tripoli Central Hospital (TCH), Tripoli Medical Centre (TMC) et Gamhoria Hospital (GH)

* Other genodermatoses	Tripoli Central Hospital	Triploli Medical Center	Gamhoria Hospital	Total
Tuberous Sclerosis	10	2	3	15
Albinism	3	0	1	4
DD	4	5	20	29
Others	15	10	28	53

2009 Register: Number of cases other genodermatoses inTripoli Central Hospital (TCH), Tripoli Medical Centre (TMC) et Gamhoria Hospital (GH)

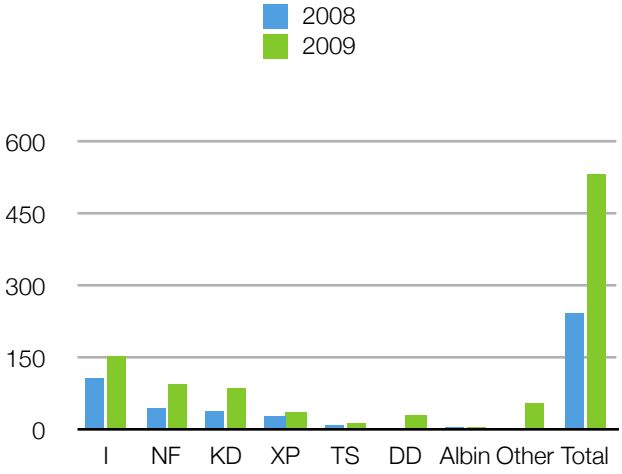


Gender Distribution in Tripoli Central Hospital (TCH), Tripoli Medical Centre (TMC) and au Gamhoria Hospital (GH) patients



Consanguinity in Tripoli Central Hospital (TCH), Tripoli Medical Centre (TMC) et au Gamhoria Hospital (GH) patients

Disease / Syndrome	2008	2009
Ichthyosis	105	152
Neurofibromatosis	43	94
Palmoplantar Keratoderma	37	86
Xeroderma Pigmentosum	27	35
Epidermolysis Bullosa	17	66
Tuberous Sclerosis	9	12
DD	0	29
Albinism	4	4
Others	0	53
Total	**242**	**531**



Cases in 2009 and 2008 in Tripoli Central Hospital, Tripoli Medical Centre et Gamhoria Hospital

Working groups focused on the epidemiological data collect and on the management of each disease

These groups gather regularly under the coordination of:

- Data Center Coordinator: Dr. Amal Merset, Tripoli Central Hospital
- East and South Data Center Coordinator: Dr. Mariam Bozgia, Gamhoria Hospital, Benghazi
- West Data Center Coordinator for Xeroderma Pigmentosum: Dr. Aisha Najat A. Mormesh, Tripoli Central Hospital
- Epidermolysis Bullosa Group Coordinator and Website: Dr. Abdelaziz H. Mohamed, Al-Bayda Hospital
- Ichthyosis Group Coordinator: Dr. Bashir Zendah, Tripoli Medical Centre
- Palmoplantar Keratoderma Group Coordinator : Dr. Reyan Elmaghrabi, Tripoli Medical Centre
- Neurofibromatosis Group Coordinator and Spokesperson: R. Omran Omar Bugrein Gamhoria H., Benghazi
- Xeroderma Pigmentosum Group Coordinator: Dr. Ali Saleim Lashab, Al-khoms Hospital
- Darier Disease Group Coordinator : Dr. Kafia Elhafi, Tripoli Central Hospital
- Tuberous Sclerosis Group Coordinator: Dr. Mohamed Elhashme, Tripoli Central Hospital, Paediatric Society
- International Coordinator: Prof. Gamal Duweb, Gamhoria Hospital, Benghazi

The development of a multidisciplinary approach

Specialists involved in the management of patients are pediatricians, pediatric surgeons, ophtalmologists, orthodontists, maxillofacial surgeons, pharmacists, urologists, neurologists, plastic surgeons.

Training and raising the awareness of care personnel, patients, families and general public

Training given to care personnel is made through lectures in Faculties of Medecine. Actions to educate patients are done with families. General public awareness raising is done through the media.

A regular supply of drugs and dressings

Drugs are free and with priority access. To ensure the regular supplying of drugs and dessings, the head of surveillance of drugs joined the Libyan genodermatoses group.

Charities

There is no severe genodermatoses patients' organisation. A social support is brought through the following organisations: Safe childhood, International African Arab Organization for Disabled Rights (IAAODR). Collaborations are viewed with Waatasimo, Shabab Libya Organization, Amal Orphan Association.

Access to diagnosis and genetic counseling difficulties

Antenatal diagnosis, diagnosis of specific cases and genetic counseling are difficult to develop because of the cultural context.

Morocco

Dr Abdellatif IDRISSI AZZOUZI, Service des maladies dermatologiques, DELM/Ministère de la Santé, Morocco

Dermatology care in Morocco

• 4 university hospital dermatology departments

There are 4 university hospital dermatology departments: the dermatology department of Rabat University Hospital with a genodermatoses consultation, the dermatology department of Casablanca University Hospital, the dermatology department of Fès University Hospital and the dermatology department of Marrakech University Hospital.

• 3 non governmental organisations focused on skin

The Moroccan Genodermatoses Organisation (AMDG) located in Rabat University Hospital, the Moroccan Psoriasis Organisation, the Gildy Organisation (Casablance University Hospital). These organisations are local and led by doctors.

• Access to care for the destitute

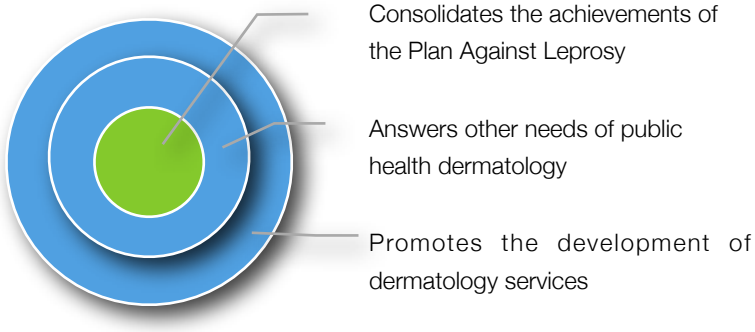
Access to care for severe genodermatoses patients is linked to the general issue that is the access to care in Morocco: access difficulties because of the geographical isolation, the financial difficulties, the cultural context. To remedy to that, a medical assistance system for the destitute (RAMED) has been introduced.

Genodermatoses are on the list of public health dermatology priorities set in the National Programme Against Leprosy and Dermatological Diseases (2008-2012)

• The National Programme Against Leprosy and Dermatological Diseases (2008-2012)

The major concern in Morocco was to include severe genodermatoses on the list of public health dermatology priorities. Two opportunities happened: the revision of the strategy of the Leprosy Programme (2006-2007) and the elaboration of an Action Plan of the Ministry of Health for 2008-2012. This plan consolidates the achievements of the Plan Against Leprosy in a decentralized framework, answers other needs of public health dermatology and promotes the development of dermatology services.

Strategy of the National Programme Against Leprosy and Dermatological Diseases



Plan of the Ministry of Health (2008-2012)

Goal	Action	Activity
1. Consolidates the achievements of the Plan Against Leprosy	1. Consolidates the achievements of the Plan Against Leprosy in a decentralized and adequatly integrated framework	1. Integration of « ORE » and « CPE » in the Plan Against Leprosy
		2. Revision of the mission and the status of the National Center on Leprology
		3. Organisation of regional meetings on the control of dermatological diseases and leprosy
		4. Organisation of training seminars on dermatology and leprology
		5. Reinforcement of ICT activities

Plan of the Ministry of Health (2008-2012)

Goal	Action	Activity
		6. Gathering of additional resources thanks to partnerships
2. Answers other needs of public health dermatology	232. Develoment of action plans specific to the new diseases targeted in the program.	7. Development of a Plan Against psoriasis
		8. Development of a national strategy against skin cancer
		9. Development of a plan against dermatoses linked to poverty and precariousness
		10. Development of a network dedicated to severe Genodermatoses
3. Promotes the development of dermatology services	231. Development of dermatology services and coordination between the different levels of health care delivery	11. Equiping dermatology departments
		12. Participation to the development of a provincial action plan to provide training courses in dermatology
		13. Support to the development of a reference/counter-reference system in dermatology and leprology
		14. Revision of the information system (development of national registers)
		15. Support to the development of a reference/counter-reference system in dermatology and leprology
	233 : Support to moving strategy	16. Support to medical caravan

• A budget estimated to 500 000 euros for the National Programme Against Leprosy and Dermatological Diseases for 2009-2012

Year	2009	2010	2011	2012
Budget *(dirham)*	62 500	1 390 000	1 480 000	1 580 000

The actions of the programme are equipping and updating dermatology departments, purchasing drugs, developing coordinatinon means and actions for information and Public Health Services personnel awareness raising.

• Epidemiological data collect in the framework of the National Programme Against Leprosy and Dermatological Diseases

Available epidemiological data are mainly series of retrospective cases. There is a few prospective studies. The development of national registers is one of the objectives of the national project to set up network dedicated to severe genodermatoses in the framework of the action plan of the Ministry of Health for 2008-2012.

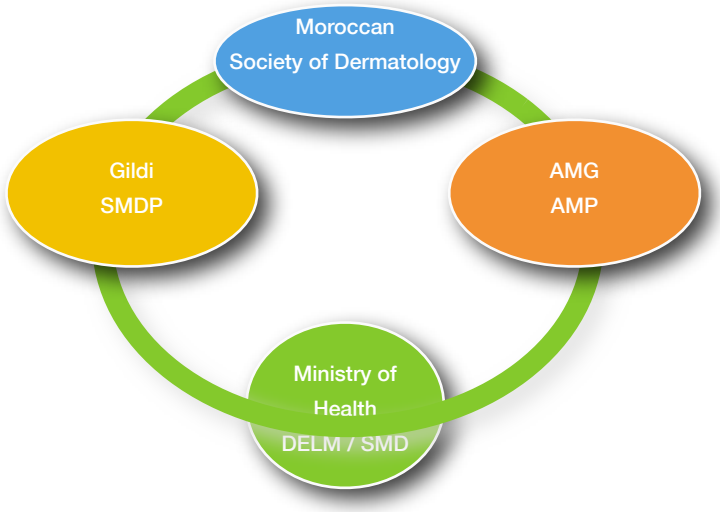
• The organisation of the severe genodermatoses care path in the framework of the National Programme Against Leprosy and Dermatological Diseases

The organisation of the care path in dermatology is one of the objectives of the National Programme Against Leprosy and Dermatological Diseases. There are two levels: the level 1 is to link general practitioners and dermatologists, and the level 2 is to link dermatologists to centres of reference with a multidisciplinary team. The identification and the official recognition of centres of reference for severe genodermatoses is a real concern. Identifying priority genodermatoses is necessary.

• A partnership between the Ministry of Health, the societies of dermatology and pediatric dermatology, the non governmental organisatons in the framework of the National Programme Against Leprosy and Dermatological Diseases

The partners of the programme of the Ministry of Health (Head of Epidemiology, Department of Dermatological Diseases) are the Moroccan Societies of Dermatology and Pediatric Dermatology (SMDP), Gildi, the Moroccan Genodermatoses Organisation (AMDG) and the Moroccan Psoriasis Organisation (AMP).

These partnerships need to be formalised by signing conventions between the Ministry of Health and non governmental organisations in order to elaborate specific action plans.



A partnership between the Ministry of Health, the societies of dermatology and pediatric dermatology (SMD and SMDP), the non governmental organisatons (AMP, AMG, Gildi) in the framework of the National Programme Against Leprosy and Dermatological Diseases

Suggestions to improve health care of severe genodermatoses patients

- Identify priority severe genodermatoses
- Define a specific health care strategy adapted to the context and the national resources
- Define a system of health care reference and a procedure handbook
- Involve patients and widen the geographical cover of organisations
- Review the statutes of the Moroccan Society of Dermatology and organize thematic working groups
- Precise research needs and their implication in public health
- Develop the international partnership (North-South and South-South)

Palestinian Territories

Spiro Tams MD, Associate Prof. of Dermatologie, Faculté de Médecine, Université Al- Quds, Jérusalem

The Israeli occupation hinders the functioning of the health system

Since June 1967, the West Bank and the Gaza Strip are under Israeli occupation. This occupation hinders the development of medical services. The Palestinian Authority, in charge of health, cannot answer the needs of the population: lack of resources and medical expertise, lack of equipments, financial difficulties, closing of checkpoints, restriction of the moves of civil and medical population. More specifically, according to the last report of WHO, the surrounding of Gaza hinders the functioning of health system, supply of medical equipment, training of health personnel and stops severe patients from receiving adapted treatments in time outside of Gaza Strip.

Two Societies of Dermatology in occupied Palestinian territories

In Palestinian society, dermatologists works mainly in private practice. There is no hospital with a dermatology department for a population of 4,2 millions of inhabitants. Recently, the Palestinian Ministry of Health nominated 7 dermatologists in the public sector and 1 in army.

• The organisation of dermatology in the West Bank

The West Bank's dermatologists are gathered within the Association of Dermatology. This association was created in 1996 with 9 dermatologists, it is a specialised group of the Medical Union of the West Bank. The Palestinian Association of Dermatology has 34 members, to date. The last 2 years, the new board of the Palestinian Association of Dermatology has initiated a new guidance for a better cooperation between dermatologists, a clear and responsible management of all the activities. Since January 2009, the association is called the Palestinian Society of Dermatology. Its first international congress of dermatology took place on March 27-30, 2009.

• The organisation of dermatology in the Gaza Strip

The Gaza Strip population has been under an Egyptian occupation before being under the Israeli occupation. So, their Medical Union could not be integrated in the Jordanian Medical Union.

So, there are two Societies of Dermatology: one for the West Bank et one for the Gaza Strip.

Consanguinity, one of the cause of the prevalence of autosomal recessive genodermatoses in the Palestinian society

• Traditional marriage and consanguinity

Traditional marriage, often between first cousins, has been considered for hundreds of years as normal -almost as a right- and without consequences on the health of children to be. This kind of marriage can be explained by the will of keeping goods and properties within the same family or as an easy and economic way of getting married in a close and conservative society, mainly rural.

• Autosomal recessive genodermatoses

Genodermatoses diagnosis is difficult without a dermatopathologist (biopsies are usually unconclusive or missread) and the DNA mapping is to expensive.

Epidermolysis bullosa, Ichthyosis, Palmoplantar Keratoderma, Neurofibromatosis, Xeroderma Pigmentosum, Ectodermal Dysplasia and Incontinentia Pigmenti are genodermatoses present in the Palestinian territories. There are also very rare autosomal recessive genodermatoses such as the papular atrichia, Naxos disease, autosomal recessive ectodermal dysplasia, the H-syndrom and unidentified diseases. The papular atrichia can be considered as a typically Palestinian genodermatoses: in a perimeter of 20 km^2, between Bethleem and Hebron, hundreds of people are affected. It must be the highest incidence in the world.

The recognition of genodermatoses, a public health issue

The recognition of autosomal recessive genodermatoses linked to the development of a primary health education and genetic counseling, could raise the population awareness on the health risks of in-family marriages.

Portugal

Marisa André, Isabel Cordeiro, Carolina Gouveia, Ana Medeira, Dept. of Dermatology, Dept. of Genetics, Pediatric Dermatology Outpatient Clinic, Hospital de Santa Maria, Lisbon, Portugal

Genodermatoses: from chronical diseases to rare diseases status

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

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

Epidemiological data collected in Lisbon and Porto

- Data collected in Lisbon

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

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

- Data collected in Porto

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

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

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

- The development of a Portuguese centre of reference

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

- The objectives of the Portuguese centre of reference

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

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

• The NF project of the Genetics Department

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

The development of a national network close to the patient

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

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

Awareness raising of medical staff and general public

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

Identification of patients' organisations

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

• Neurofibromatosis

The Portuguese Organisation for neurofibromatosis patients is the APNF.

• Epidermolysis bullosa

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

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

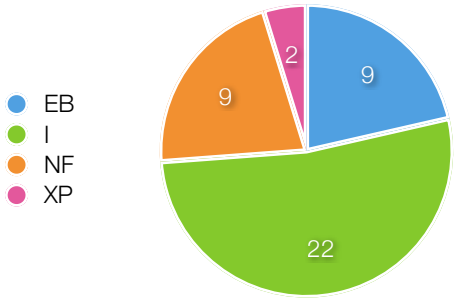
These collaborations and interactions with centres of expertise for genetic diseases would allow improving diagnosis (molecular diagnosis, electron microscopy, specific biochemical tests, immunology studies) and treatments. These collaborations would also offer a multidisciplinary approach to the patients.

Romania

Rodica Cosgarea, Dept. of Dermatology, University of Mdedicine and Pharmacy, Cluj-Napoca, Roumanie

Epidemiological data are available at a national level for epidermolysis bullosa and in the department of dermatology of the Faculty of Medecine and Pharmacy of Cluj-Napoca for other genodermatoses

The data available on a national level are those concerning the epidermolysis bullosa. According to the data available on the website of the Ministry of Health in Romania in 2008, 36 patients are officially recognized as epidermolysis bullosa patients. But in reality, they are aroung 50 epidermolysis bullosa patients. In Cluj-Napoca's department, 9 epidermolysis bullosa patients, 22 ichthyosis patients, 9 neurofibromatosis patients and 2 xeroderma pigmentosum patients are followed.



Maladie / Syndrome	Number of cases
Ichthyosis (I)	22
Epidermolysis Bullosa (EB)	9
Neurofibromatosis (NF)	9
Xeroderma Pigmentosum (XP)	2
Total	**42**

Number of cases of Epidermolysis Bullosa (EB), Icthyosis (I), Palmoplantar Keratoderma (PPK), Neurofibromatosis (NF), Xeroderma Pigmentosum (X) in the Department of Dermatology, University of Medicine and Pharmacy, Cluj-Napoca, Romania

The epidermolysis bullosa health care in the dermatology departments of Brasov and Cluj-Napoca

The multidisciplinary team is composed of a dermatologist, a pediatrician, a surgeon, a geneticist. Some skin care products are reimbursed by health insurance. Some specific dressings for the health care of epidermolysis bullosa (Mepilex, Mepitel) are available in the dermatology department of Cluj-Napoca. The regular follow up of patients includes a general checkup, lab examinations, skin and lesions examinationination, a nutritional checkup (anemia ; malnutrition ; lack of proteins, vitamins and minerals).

The needs for epidermolysis bullosa are: dressings, surgery, medical treatments, nutritional supplement, social and psychological support. According to the informations available on the website of the Ministry of Health in Romania, the average cost of these needs is assessed to 1000 RON = 238€ per month (2008).

The Degeb project: the development of a centre for molecular diagnosis of epidermolysis bullosa

The Dermatology Department of the Faculty of Medecine of Cluj-Napoca has initiated the Degeb project in 2006, www.degeb.org. This project aimed at developing molecular diagnosis of epidermolysis bullosa in Romania with the development of new technics for research, diagnosis, treatment and prevention of epidermolysis bullosa and developing a national register on genodermatoses.

This project consisted in taking sample of patients and families. This project enabled the achievement of the antigene mapping for the identification of different types of epidermolysis bullosa and the molecular analysis for mutations identification.

Mini Debra, the organisation for epidermolysis bullosa patients in Romania

Mini Debra, www.minidebra.ro, gathers epidermolysis bullosa patients and their families. It aims at promoting the knowledge of epidermolysis bullosa. It is also a link between patients, doctors, international organisations and sponsors. There is also a Debra organisation in Brasov which gathers health professionals.

International collaborations for training, diagnosis and treatments

These collaborations are in the framework of training exchange programmes. They are also developing in the diagnosis field with the centre of reference for diagnosis of Prof. Leena Bruckner-Tuderman from the dermatology department of Freiburg in Germany.

The Romanian government can finance treatements abroad for the severe diseases that cannot be treated in Romania (OG 28/2003): http://sas.mmssf.ro/compendiumLegislativ.php?id=142).

Slovenia

Mateja Dolenc-Voljč , Vlasta Dragoš , Vesna Tlaker-Žunter, Marko Potočnik, Faculty of Medicine of Ljubljana, Department of Dermatology and Veneorology, Zaloška 2, Ljubljana, Slovénia

Four dermatology departments providing a multidisciplinary approach

Slovenia has a population of 2 millions of inhabitants with 4 dermato-venerology departments including 2 university hospitals. All the departments have developed a multidisciplinary approach.

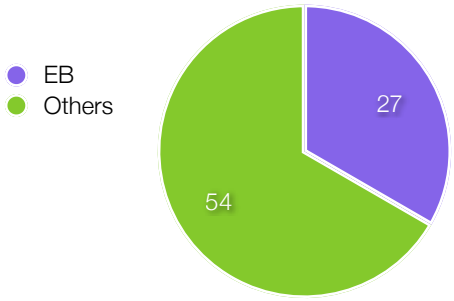
Available data for epidermolysis bullosa and Darier disease

54 cases of Darier disease have been spotted and 27 cases of epidermolysis bullosa. Among the 27 cases of epidermolysis bullosa:

- dystrophic epidermolysis bullosa: 5 patients including 3 children
- junctional epidermolysis bullosa: 2 patients
- simplex epidermolysis bullosa: 20 children in 4 families,
- the exact number of adult patients is unknown and for some of them the disease is slightly expressed.

Some cases of congenital ichthyosis, congenital erythroderma ichthyosiforms and neurofibromatosis were recently spotted.

For the other genodermatoses (palmoplantar keratoderma, vulgaris ichthyosis, anhidrotic ectodermal dysplasia, incontinentia pigmenti) the number of cases is unknown.



Epidermolysis bullosa	Number of cases
Dystrophic Epidermolysis Bullosa	5
Junctional Epidermolysis Bullosa	2
Epidermolysis Bullosa Simplex	20
Total	27

Number of cases of Epidermolysis Bullosa (EB) and Darier disease (Others).

The dermato-venerology department of Ljubljana: a specialised centre for the health care of genodermatoses children and the health care of epidermolysis bullosa adults and children

• All the children affected by genodermatoses are followed in the dermato-venerology department of Ljubljana

- The health care of genodermatoses children (except epidermolysis bullosa children)

All the children affected by genodermatoses are followed in the dermato-venerology department of Ljubljana in cooperation with pediatricians and other specialists. Parents are trained to answer the needs of children and provide adapted care. Cooperation with social workers is well developed.

- The health care of epidermolysis bullosa children

Children are regularly followed (at leats 3 times a year). They are hospitalized at least once a year for a thorough examination. Skin lesion dressings are regularly renewed at home by trained nurses. Since 1993, diagnosis is confirmed for all children on the histologic level and through electron microscopy.

• All the epidermolysis bullosa patients are followed in the dermato-venerology department of Ljubljana

- A multidisciplinary team

In 2009, a multidisciplinary team has been formed for the health care of epidermolysis bullosa. This team works thanks to the collaboration between dermatologists, pediatricians, geneticists, ophtalmologists, stomatologists, plastic surgeons, internists, psychologists. This team aims also at increasing the knowledge of epidermolysis bullosa among the specialists and improving the possibilities of treatment for patients.

- The health care of epidermolysis bullosa children
See above.

- The health care of epidermolysis bullosa adults
All the dystrophic and junctional epidermolysis bullosa patients are hospitalized once a year for thorough examination.

Other dermatology departments manage adults affected by genodermatoses other than epidermolysis bullosa

Adults patients with genodermatoses other than epidermolysis bullosa are followed in departments other than Ljubljana's. Regular check ups are done by dermatologists. Their health care is multidisciplinary, following the existing recommendations, but adapted to the local context. Home care is provided by nurses.

The costs of the disease are covered by insurance companies with an exception for dressings, creams and topic treatments

Debra Slovenia : an organisation for epidermolysis bullosa patients in Slovenia

Debra Slovenia was created in 2005. This organisation gathers epidermolysis bullosa patients and their relatives. It facilitates meeting of patients and sharing experience. Debra Slovenia also brings support for social integration of patients. It has a role of education, training and information to patients and their families and health professionals: education and information on news in the field of skin care and treatments for patients. Debra Slovenia also provides specific dressings for epidermolysis bullosa care. Debra Slovenia raises general public awareness. It also raises funds to answer the needs of patients.

European collaborations to favour exchange between patients, improve diagnosis and training of health professionals

• Cooperation between Debra Slovenia and Debra Croatia
Debra Slovenia and Debra Croatia have organised joint meetings.

• Contacts with the Department of Dermatology and Molecular Dermatology of Fribourg in Germany
These contacts aim at providing genetic analysis for dystrophic and junctional epidermolysis bullosa patients.

• Training health professionals in United-Kingdom and Croatia
Two dermatologists, two nurses and a pediatrician participated in a symposium on epidermolysis bullosa in United-Kingdom and in Croatia in 2009.

The projects of the dermato-venerology department of Ljubljana to improve the health care of genodermatoses patients in Slovenia

- Prepare a national register on genodermatoses in Slovenia
- Organise a national network between the dermatology departments for genodermatoses
- Reinforce collaborations in Europe
- Improve diagnosis and treatment
- Train young dermatologists and other health professionals

Tunisia

The Tunisian Genodermatoses group was represented by Monia Kharfi, Dept. of Dermatology, Charles Nicolle Hospital, Tunis, Tunisia

Seven university hospital dermatology departments in Tunisia

There are 7 university hospital dermatology departments in Tunisia: four in Tunis, one in Sousse, one in Monastir, one in Sfax.

Genodermatoses are recognized as a handicap, a public health priority in Tunisia

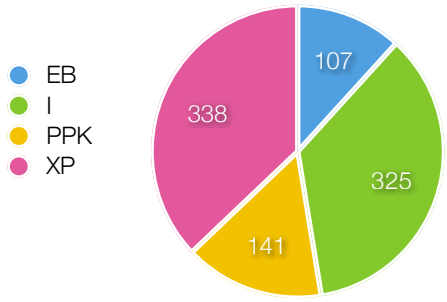
In 2006, the law widens the definition of handicap. Genodermatoses are recognized as disabling diseases. Genodermatoses patients have the same rights as disabled people: a «handicap card», free care in public health structures, free drugs available in hospitals (such as the Neotigason), free access to transportation (urban and 50% off inter-urban).

Data available in 4 university hospital dermatology departments and a mapping for xeroderma pigmentosum

There is no national prevalence established for the different genodermatoses in Tunisia. Presented data are from studies and reports of the 4 Tunisian university hospital dermatology departments: Charles Nicolle Hospital, Military Hospital, Sousse Hospital and Sfax Hospital.

A xeroderma pigmentosum mapping was made by the Habib Thameur Hospital (Tunis) team. It spotted 338 cases. The number of affected children is estimated to more than 800 cases. The highest concentration of patients is in the North-West where its frequence is sometimes above1/100.

Disease	Habib Thameur	Charles Nicolle	Militaire	Sousse	Sfax	Total
Epidermolysis Bullosa	/	38	14	15	40	107
Ichthyosis	/	148	17	60	100	325
Palmoplantar Keratoderma	/	49	2	40	50	141
Xeroderma Pigmentosum	338	/	/	/	/	338
Total	**338**	**235**	**33**	**115**	**190**	**911**



Number of cases of Epidermolysis Bullosa (EB), Icthyosis (I), Palmoplantar Keratoderma (PPK) and Xeroderma Pigmentosum (XP)

The assessment of the costs of the health care for epidermolysis bullosa (1.100 euros per patient and per year), ichthyosis (2.200 euros per patient and per year), palmoplantar keratoderma (1.150 euros per patient and per year)

• Epidermolysis bullosa

For daily care, sterile compresses, sofra-tule, gauzes, cleansing products, antiseptics, emollients, local antibiotics and antibiotic and vitamin treatments are used. The costs of the health care of an epidermolysis patient are assessed to 2000 Tunisian dirhams for a year (1.100 euros). The cost of biological checkups and potential explorations must be added to these expenses.

• Ichthyosis

Acitretine (Neotigason*), emollients, antimycotics and antihistamines are used for the health care of ichthyosis patients. The costs of the health care of an ichthyosis patient are assessed to 4.000 Tunisian dirhams for a year (2.200 euros). During the antenatal period, the cost of care in specialised centre within the different departments of neonatalogy of Tunis, Sousse and Sfax and biological checkups and potential explorations must be added to these expenses,

• Palmoplantar Keratoderma

Acitretine (Neotigason*), emollients, antimycotics and antihistamines are used for the health care of a palmoplantar keratoderma patient. They are assessed to 2.100 Tunisian dirhams for a year (1.150 eurost. The cost of biological checkups and potential explorations must be added to these expenses.

A centre of expertise for the health care of xeroderma pigmentosum patients at the Habib Thameur Hospital (Tunis) and pediatric dermatology specialised consultations in 4 university hospitals

Xeroderma pigmentosum patients are managed, for most of them, in the centre of expertise of the Habib Thameur Hospital. Charles Nicolle Hospital (90% genodermatoses), the Children Hospital of Tunis, the Military Hospital have pediatric dermatology specialised consultations. La Rabta Hospital also opened a dermatology consultation in the spring of 2009.

Three medical genetics departments

There are 3 genetics departments: one in Tunis, one in Sousse, one in Sfax.

Three molecular biology laboratories

There are 3 molecular biology laboratories in Tunis: one at the Pasteur Institute, one at the Faculty of Medecine and one in the private sector.

Improve the health care of epidermolysis bullosa, ichthyosis and palmoplantar keratoderma: education of families and access to care devices and sun screen

Within the different medical teams, the interest in the health care of genodermatoses patients has increased. Specific actions have been developed in each department, among other the patient recruitment.

• The education of families

The doctors develop actions favouring education of families through explanations of the disease, education to the health care of children and help to home care.

- Epidermolysis bullosa: education and training of families

In each dermatology department, doctors and care personnel develop actions favouring education of patients and their families. Epidermolysis bullosa health care training is organised within the dermatology department of Charles Nicolle Hospital. Families are trained to provide adapted care to epidermolysis bullosa newborns. A trained nurse gives this training in day hospital, in a dedicated room. Parents are accompanied the first weeks to evaluate the quality of the care provided. A psychological management is also available for patients and families.

- Ichthyosis : education and training of families

During the antenatal period, health care is provided in the different neonatalogy departments of the university hospitals of Tunis, Sousse, Monastir and Sfax in collaboration with the dermatologist. Families are trained to provide adapated care to ichthyosis newborns. A trained nurse gives this training in day hospital, in a dedicated room. Parents are accompanied the first weeks to evaluate the quality of the care provided. After that, home care is organised and managed with regular checkups in day hospital consultation.

- Palmoplantar keratoderma

The health care of palmoplantar keratoderma patients is symptomatic treatment made at home. Patients are regularly seen in consultations. The treatment of frequent fungic secondary infections is made through antifungal.

Reinforced collaborations between dermatology, genetics and molecular biology departments

Collaboration has been reinforced between the dermatology departments of Tunis, the genetics department of Charles Nicolle Hospital and molecular biology department of the Pasteur Institute of Tunis.

The development of severe genodermatoses patients' organisations

• Creation of the organisation for Help to xeroderma pigmentosum children in 2008
Recently, and with the help of the French xeroderma pigmentosum organisation , an organisation for xeroderma pigmentosum patients has been developed. Website: www.xeroderma pigmentosum-tunisie.org.tn

• Development of an organisation for epidermolysis bullosa patients in Sfax
• Development of a genodermatoses organisation in Tunis

Collaborations with French teams

Several teams have contacts and collaborations with foreign teams, mainly French.

The objective of the Tunisian genodermatoses group: pool and structure the initiatives on the national level to optimize efforts

Génodermatoses et Méditerranée enabled the reinforcement of individual initiatives in the field of training, research and care. These initiatives participate in improving the health care of genodermatoses. This action also helped raising decisionmakers, health professionals and families awareness.

Yemen

Dr. Abdul Rahim AL-Samie, Dr. Yasin Al- Qubati, Dr. Aiman Al Shami, Dept. of Dermatology, Taiz Hospital

An underdeveloped health system

The Yemen population is mainly urban (80%). 43% have access to drinking water. The health situation is bad with underdeveloped infrastructures. The health expenses per person are assessed to 16$. Between 58 and 63% of the population have access to primary health care. The infant mortality (rate) is of 102/1000.

Initiatives for the improvment of the health care of genodermatoses patients are hindered by bureaucracy constraints. The issue is not managed by the Ministry of Health nor by non governmental organisations.

Data collected by the dermatology department of Taiz Hospital

The data collect on a national level is hindered by the lack of information system. The most frequently encountered genodermatoses are xeroderma pigmentosum, ichthyosis, epidermolysis bullosa, palmoplantar keratoderma, neurofibromatosis and acrodermatitis enteropathica. Presented data have been collected at the Hospital of Taiz. For each patient the following data have been collected: name, registration date, gender, age, address (government,district, town), phone number, concerned pathology (xeroderma pigmentosum, epidermolysis bullosa, palmoplantar keratoderma, ichthyosis) and family history.

Number of cases with Epidermolysis bullosa, Ichtyosis and Palmoplantar keratoderma

Severe genodermatoses patients have a free of charge health care

The objectives to improve genodermatoses patients health care in Yemen

- Train health professionals for diagnosis
- Train health professionals for the collect of severe genodermatoses data
- Include severe genodermatoses health care in a more general context of improvment of the health situation and development of a project related to regional and international initiatives.

The seven working groups

The specificity of *Génodermatoses et Méditerranée* and *Together Against Genodermatoses* is to help patients and their families in their daily life. Working groups have been organised in this perspective.

Six working groups by diseases have been created
The six working groups are focused on the diseases or groups of diseases targeted by the partners of *Génodermatoses et Méditerranée* and *Together Against Genodermatoses*:

- Epidermolysis Bullosa,
- Ichthyosis
- Palmoplantar Keratoderma
- Neurofibromatosis
- Xeroderma Pigmentosum
- Other genodermatoses (Anhidrotic Ectodermal Dysplasia, Incontinentia Pigmenti).

The aim of these groups is to define the best strategy for the health care of each disease in a extremely concrete way: health, nursing and social care.

A transversal group has been created
This transversal group "Transsectional approach of genodermatoses for a preventive synergistic action" has been introduced to discuss the development of a network of laboratories. One of the task of this network is to help patients and families to access antenatal diagnosis.

A double coordination to favour international and European exchanges
Each group is led by 2 coordinators (a coordinator from a European Union Member State in the framework of the *Together Against Genodermatoses* project and a coordinator from a non member country in the framework of *Génodermatoses et Méditerranée*). The list of the working groups participants is available on the projects' websites.

A 3 year programme
Each disease working group is in charge of defining the best strategy of health care of each disease. These recommendations must be adapted to the social and economic realities of each country to best answer the needs of patients. To do so, each working group will gather the following collected data in each country: epidemiological data, needs of patients and costs of the disease for the patients, centres of expertise, patients' organisations, recommendations for the health care of patients, national network, European and international collaborations.

First step: evaluate the prevalence of the disease
The collect of epidemiological data is of the utmost importance to evaluate the needs on a national level. They enable establishing the prevalence of the disease in each country and negociating with health authorities a budget for patients but also for research. The collect of these data can lead to a publication.

The epidermolysis bullosa working group
• Collect the epidemiological data
The first objective of this working group is to collect the epidemiological data with the identification of a coordinator in charge of collecting the data in each dermatology centre. These epidemiological data are of the utmost importance to determine the incidence of the disease in each country and to negociate with health authorities a budget for patients but also for research. The collect of these data can lead to a publication.

• Make a state of the art for the health care of epidermolysis bullosa
Participants also decided to present a state of the art of the health care of epidermolysis bullosa patients in their country (collect of epidemiological data, evaluation of the needs of patients and families, assessment of the costs of the disease, development of centres of expertise for patients and families, development of patients' organisations, strategy for genodermatoses health care, development of a national network, development of collaborations of European and international levels).

The ichthyosis working group

- Define the ichthyosis to be included in the project

The first objective of this working group is to define the forms of ichthyosis to be included in the project. The coordinator will collect the opinion of the different participants and summarise the situation. The group participants will collect available information they will address to the coordinator. The coordinator will make one summary that he will send to all the members to get it validated.

- Validate and introduce a software collecting data

The group members plan the development of a software collecting epidemiological data to be used by all the participants, on the model of the Greek software in order to establish databases in each country (3 years).

- Evaluate the needs of patients

The evaluation of the patients' needs will be made by every participant. Each of them will evaluated the patients' needs thanks to a survey made with the patients and their parents. Patients and parents could list their needs in the education, genetic counseling, moral and financial support fields.

- Assess the costs of the disease

Assessment will be made in each country judging from the cost of drugs, cosmetics, checkups and hospitalization per patient and per year, during and after the antenatal period.

- Train the care personnel

In each department, a nurse or a nurse team will be in charge of the health care of ichthyosis (genodermatoses in general). Nurses and care personnel will be trained by doctors of the department or through courses in collaboration with the dermatology department of Necker Hospital.

- Develop health care strategy

The coordinator collects the existing recommendations and elaborates recommendations for diagnosis, treatment and prevention. These recommendations will be sent to the group members for validation.

- Develop social support and the relationship with organisations

The group pointed out the importance of getting in touch with existing organisations and –when these organisations don't exist yet- support their creation. Raising general public awareness can be done through medias, posters, commercials...

- Develop relationships with public health authorities to train doctors and care personnel

Training doctors and care personnel, in the framwork of public health facilities, is crucial. This training will be developed through post-graduate education and meetings for the training of doctors and care personnel.

The palmoplantar keratoderma working group

Non communicated

The neurofibromatosis working group

- Establish the incidence and lead epidemiological studies of the disease
- Manage the disease on childen and adults according to an approach coordinated between specialists with the development of recommendations for examination and analysis to be done on patients
- Identify the multiplex families
- Develop neurofibromatosis websites to facilitate exchanges between the group members
- Validate case reports and observations

The Xeroderma Pigmentosum working group

- Collect the epidemiological data

One person will be in charge, in each country, of the collect of the compulsory data (patient's number, birth date, gender, country) and optional data (city, gravity -severe, average, variant, handicap, age at the first consultation, number of carrier family members, consanguinity, first name -3 first letters-, name -3 first letters).

- Evaluate the needs of patients

This evaluation will be made on the model of the one made in France and Tunisia.

- Develop trainings with public health authorities

One person will be in charge of being in touch with the national programmes as for example in Tunisia, with the Head of Primary Health Care of the Ministry of Public Health.

- Develop health care strategies

These strategies could be developed on the model of those introduced in France (national protocol for diagnosis and care), Tunisia and Egypt.

- Develop social support and relationship with patients' organisations

Raising patients awareness on the necessity of developing organisations for their health care is a real issue. One of the group's project is to present the basic means enabling the development of organisations for xeroderma pigmentosum patients. The number of patients is particularly high in the Mediterranean countries, so the creation of an organisation or federation at the Mediterranean level is in debate.

- Develop relationships with public health structures

These relationships have to be developed in the framework of the training of care personnel but also in the wider framework of health authorities awareness-raising on the severity of this pathology and on the necessity of introducing structures adapted to the patients.

The other genodermatoses working group
Non communicated

The transversal working group
The transversal working group has been created in 2009. Indeed, evaluating the prevalence of rare and severe genodermatoses is necessary to better answer the needs of the patients. The expertise acquired through the *Génodermatoses et Méditerranée* and *TAG* networks pointed out the clinical and genetic heterogeneousness of genodermatoses: the existence of a hot spot of mutation, of recurring mutation, of funding effect, consanguinity in the types of transmission. The objective of this group is to lean on this knowledge and on the one of other European networks to develop tools for a quick diagnosis and a good correlation cost/performance.

The first objective of the group is to list the available molecular data concerning the gene and its mutation for each disease (hot spot of the mutation, recurring mutation, funding effect, « private mutation », country). The second objective is to identify centres of molecular diagnosi: carrier diagnosis, antenatal and post natal diagnosis.

Our objectives for 2010

A better care

Define recommendations for the health and social care

At the 2010 working session, each working group will present their advances to develop recommendations for the care of epidermolysis bullosa, ichthyosis, palmoplantar keratoderma, neurofibromatosis, xeroderma pigmentosum, anhidrotic ectodermal dysplasia and incontinentia pigmenti patients.

A better orientation

Identify in each country the resources available

One of the missions of each partner of the project is to identify in his own country the resources available for the health care of severe genodermatoses patients: consultations, centres of expertise, diagnosis tests, genetic counseling, registers, patients' organisations.

Sharing information

Spread information in French and English
A newsletter on the activities in process in each country will be published, in French and English. The websites www.tag-eu.org and www.genodermatoses-et-mediterranee.org will be developed, updated and fully translated in English and in French.

Extend our experience on a Mediterranean, European and international level

Associate new partners to the project
The strenght and originality of *Génodermatoses et Méditerranée* and *Together Against Genodermatoses* is that these projects gather Mediterranean, Middle-Eastern and Member States countries. The widening of the project is really important because *Together Against Genodermatoses* has been chosen as a pilot project for the European networks of reference.

Together for a better care

Gather the partners for the annual working session
The 2010 working session will take place in Rome in Italy, on October 22-24. It will be organised by Ospedale Pediatrico Bambino Gesù. The aims of this meeting are to define the best strategies for the health care of each disease in each country, with a particular attention to the patients' daily life. Health, nurse and social care will be taken into account.

The coordinators of each group of diseases will present the results of their groups. The following points will be broached:

- Summary of the data gathered in each country,
- Advances in the collect of epidemiological data,
- Assessment of the patients' needs and of the costs of the disease for the patient,
- Development of centres of expertise,
- Creation and development of patients' organisations,
- Strategies for the health care of genodermatoses,
- Development of a national and international network.

During this session, advances will be presented:

- A transversal working group has been created to develop, among other things, a network of laboratories for diagnosis and genetic counseling,
- Experts in genodermatoses, who are not members of working groups, will participate in the working session and will present the late advances in the care and research field for some diseases in their country,
- An inter-religious debate will take place during the round table on bioethics.

Annexe

Participants in the 2009 meeting

First Name	Last Name	Country
Bakar	Bouadjar	Algeria
Mihael	Skerlev	Croatia
Vlasta	Zmazek	Croatia
Emad	El-Gamal	Egypt
Ghada	El-Kamah	Egypt
Adel Botros	Zaghloul	Egypt
Amin	Sharobeem	Egypt
Smaïl	Hadj-Rabia	France
Marie	Guillou	France
Louis	Dubertret	France
Françoise	Séris	France
Salah	Ferkal	France
Alexandra	Katsarou	Greece
Dimitrios	Sotiriadis	Greece
Anna	Tagka	Greece
Vasiliki	Vosynioti	Greece
Ioannis	Bassoukas	Greece
Adriana	Mavrou	Greece
Angelika	Roussaki	Greece
Maya	El-Hachem	Italy
Giovanna	Zambruno	Italy
Mariam	Bozghia	Libya
Mohamed	El Hashme	Libya
Bashir	Zendah	Libya
Ali	Lashhab	Libya
Najat	Murmesh	Libya
Abdelatiff Idrissi	Azzouzi	Morocco
Spiro	Tams	Palestinian Territories
Isabel	Cordeiro	Portugal
Marisa	Andre	Portugal
Rodica	Cosgarea	Romania
Monia	Kharfi	Tunisia
Sonia	Abdelhak	Tunisia
Mohammed	Zghal	Tunisia
Noomen	Hakim	Tunisia
Mohamed	Denguezli	Tunisia
Yasin	Al-Qubati	Yemen
Abdul Rahim	Al-Samie	Yemen
Aiman	Al-Sahmi	Yemen

Fondation René Touraine
Hôpital Saint Louis, Pavillon Bazin, 1 avenue Claude Vellefaux, 75010 Paris France
T: 33 (0)1 53 72 20 80 - 33 (0)6 32 38 63 08 F: 33 (0)1 53 72 20 61 Email: marie.guillou@fondation-r-touraine.org
www.genodermatoses-et-mediterranee.org - www.tag-eu.org