Since 2003, Genodermatoses in Mediterranean gets together stakeholders of the management of severe genodermatoses from Mediterranean and Middle-East countries. The aim of this project is to improve the health care and social care of patients with severe genodermatoses. This project has been initiated by Fondation René Touraine thanks to the support of one of its founding members: Laboratoires Pierre Fabre. Since 2008, this project is developing on the European level thanks to a co-funding from the European Union, in the framework of the Public Health Programme through the Together Against Genodermatoses grant (TAG n° 2007 335).

Together Against Genodermatoses is a European Reference Network for Rare Diseases pilot project

Centres of Expertise and European Reference Networks correspond to the needs expressed by rare disease patients. Patients ask for a better flow of scarce information and for better organisation of patient-centred care. This care must include social aspects as well as medical, and both need to be integrated at all levels. This care has to be improved for all patients throughout the EU, to address the concern for equity expressed by rare disease patients.
> Read the Eurordis Position Paper

European Reference Networks are a mechanism for cooperation between Member States to pool expertise and share knowledge in highly specialized medical fields with high European added value, particularly in the field of rare diseases and rare cancers (including pediatric cancers). The general principle of the European reference networks is based on the mobility of the expertise rather than the mobility of patients. These networks should benefit to healthcare professionals and patients in all Member States.

The specific objectives of the European networks of reference are

improve access to diagnosis and treatment for diseases requiring a particular concentration of resources and expertise (highly specialized healthcare)

contribute to the dissemination of expertise and the development of best practices

strengthen research, epidemiological surveillance, training and information

The Commission, in close collaboration with Member States, identifies and develops ERNs. They define the criteria and evaluate the specific conditions to be fulfiled by the European reference networks. They aslo make sure that ERN comply with these criteria/conditions and ensure their monitoring and external evaluation.
> European Commission - Public Health website

Eurordis : Rare Together / Communities of patients on the Internet

In December 2005, Eurordis has launched a new project to build communities of patients on the Internet. The aim of these communities is to reduce isolation of people with rare diseases, their families and those who care for them.

> Ichthyoses Community of Patients on the Internet

Life with Epidermolysis Bullosa: a new book provides guidelines on diagnostics, treatment and care
From Orphanews

This new book is the first to utilise evidence-based data derived from the world´s largest cohort of inherited EB patients, the American EB Registry. Clinical manifestations, diagnostic considerations,

pain management, and therapeutic approaches are all discussed, along with a chapter devoted to gene therapy in hereditary EB, which has been recently successfully performed within a localised skin site on a single EB patient as a proof-of-principle test.
Title: Life with Epidermolysis Bullosa: Etiology, Diagnosis, Multidisciplinary Care and Therapy
Authors: Fine, Jo-David; Hintner, Helmut -Eds
Publisher: Springer, 2009
ISBN: 978-3-211-7927

News from countries

Bulgaria: the management of Epidermolysis bullosa (EB) patients and genodermatoses activities

From Dr. Ivelina Yordanova, MD, PhD, Dermatologist, Assistant Prof. in the Department of Dermatology and Venereology, Faculty of Medicine, Medical University Pleven, Bulgaria. Vice-President of Association Epidermolysis Bullosa Bulgaria. Consultant in dermatology at ICRDOD
From Prof. Dimitar Gospodinov MD, PhD, Head of the Department of Dermatology and Venereology, Faculty of Medicine, Medical University Pleven, Bulgaria. Dean of the Faculty of Medicine, Medical University Pleven, Bulgaria.

Association Epidermolysis Bullosa Bulgaria (AEBB)
In Bulgaria, the people suffering from Epidermolysis Bullosa Hereditaria are united in Association Epidermolysis Bullosa Bulgaria (AEBB) which has been created and legally registered under the Bulgarian law in 1994, in the Department of Dermatology and Venereology at Medical University Pleven, Bulgaria by Prof. Mariana Traschlieva. AEBB is a voluntary, non-governmental charity organization with ideal object, with the only purpose to make Epidermolysis Bullosa (EB) patients' life better. AEBB is a member of the network of organizations for EB in the world – DEBRA (Dystrophic Epidermolysis Bullosa Research Associations) International since 1992.
> Learn more on AEBB

Epidemiological study on Epidermolysis Bullosa in Bulgaria
Till now, the epidemiological study on Epidermolysis Bullosa in Bulgaria for 39 years period (1970-2009) over the distribution of the disease in the country have discovered nearly 100 patients with different forms of Epidermolysis Bullosa. All of Epidermolysis Bullosa patients have been diagnosed by clinical criteria and electron microscopy examination. The patients aged from 20 to 29 are the majority. The highest percentage is that of EBS (59%), followed by EBD (37%) and JEB (4%).
The prevalence of :

all types of EB in Bulgaria (general population 7,973,671) up to 31 December 2007 is 8.6 cases/per million inhabitants
EBS – 5,3/mill
EBJ –0,1/mill
EBD – 3,1/mill

The distribution of EBS patients by subtypes indicates that EBS Weber-Cockaine (EBS WC) has the highest percentage, followed by EBS Koebner (EBS). We diagnosed 5 patients affected by JEB (Herlitz 4, non Herlitz -1). All the 4 patients with JEB Herlitz died before they were one years of age. In the EBD group almost 1/3 of the patients have dominant dystrophic EB subtype Cockaine-Touraine and another 1/3 has recessive DEB subtype Hallopeau-Siemens. 74 % of the patients with EBD demonstrate severe mucous membrane affection. 70 % of them had severe muscle-skeletal deformations.

The life of EB patients in Bulgaria

The most severe forms of the disease lead to early death, because of unfavourable conditions of bringing up, lack of medicaments and insufficient training of medical staff. Parents take care of their affected EB children. 30 of the sufferers are heavy invalids while the others, earn their own means of living. All patients and especially these with EBD need psychological and financial assistance. The arrangement of orthopaedic, dentologic, dermatologic and psychiatric services for them is essential. We are working for more precise diagnosis of the disease and for better possibilities of treatment and rehabilitation.

Collaborations at the European level

At present we collaborate with Dr. Cristina Has and Prof. Lenna Bruckner-Tuderman from the EB Center in Freiburg, Germany, where we do the immunofluorescence analysis of the skin samples (Antigen mapping) and DNA mutagen analysis of some of the affected EB Bulgarian families.

A book for patients and general practitioners about the Epidermolysis Bullosa Management

Following our research, we wrote and distribute among the patients with EB and General practitioners in Bulgaria a book on the medical management of the disease. The focus of the book is: Diagnosis and classification of the disease; Genetic consultation; Medical care for skin and mucous membranes; Nutrition in the different types EB and different ages; Diagnostic algorithm for EB; Algorithm for wound treatment; Prevention of the muscleskeletal and joint deformations by physiotherapy.

The Information Centre for Rare Diseases and Orphan Drugs (ICRDOD)

Information Centre for Rare Diseases and Orphan Drugs (ICRDOD) is a free educational and information service in Bulgarian and English languages, providing personalized replies to requests from patients with rare diseases, in particular with genodermatoses, their families and medical professionals. Any medical or health information, provided and hosted on this site is given by medically trained and qualified professionals. The information provided on this site is designed to support, not replace, the relationship that exists between a patient/site visitor and his/her doctor. ICRDOD is a project and activity of the Bulgarian Association for Promotion of Education and Science (BAPES), a non-government non-profit organization, registered under the Bulgarian law in town Plovdiv on legal persons with non-profit purposes in 2003. The President of ICRDOD is Prof. Rumen Stefanov, MD, PhD. For the realization of this website, BAPES utilizes its own funding, received from private donors. The director and all consultants of ICRDOD are contributing on volunteer basis.

One of the most important activities of ICRDOD is opening of Medical Centre "RareDis" on 15 May 2009. It's a logical extension of the ICRDOD activities, because after having received information about their disease, people need continuous medical cares and follow up. A main accent in the new centre's

activities is the team work in assuring physical, psychological and social rehabilitation, as well as training parents in providing qualified and adequate daily health care of their children with rare diseases. This centre would be of substantial national and regional importance, an innovative model for all European countries. Medical Centre "RareDis" co-operates closely with ICRDOD, scientific societies and patient associations in a number of joint projects. > Learn more about ICRDOD

National Alliance of People With Rare Diseases
During the last years the situation of the people with rare diseases is getting better with the help of some European initiatives. In almost all European countries there are patient associations which strengthen the social position and the influence of the people with rare diseases, create contacts between people, concerned by the same disorder, spread information about diagnosis, health care, treatment and lifestyle of people with rare diseases. That's why the National Alliance of People with Rare Diseases in Bulgaria has enormous importance for all. The main purpose of the National Alliance is to be a link between the people with rare diseases and the representatives of the social and healthcare system on the other side. The Alliance works for the protection of one of the principal human rights – the right of timely and equal medical care. The organisation also lobbies for the assumption of adequate laws in the field of the protection of the rights of the peopl e with rare diseases. It was created in April 2007. Association Epidermolysis Bullosa Bulgaria is one of the founder members of the Alliance.

Italiy: training on the multidisciplinary approach to Epidermolysis Bullosa and Inherited Icthyosis

From Maya El Hachem, Ospedale Pediatrico Bambino Gesu, Rome, Italy
and Giovanna Zambruno, Istituto Dermopatico dell'Immacolata, IRCCS, Rome, Italy

Theoretical and practical training course on the multidisciplinary approach to EB
One theoretical and practical training course on the multidisciplinary approach to EB was held on March 5th and 6th at Bambino Gesù Hospital. The course illustrated the epidemiology, the new classification, the cutaneous and systemic involvement and complications, the psychological and quality of life aspects in this disease group. The practical session focused on multidisciplinary management, nursing and diagnostic procedures. The number of participants was 37. The official language was Italian and registration was free.

Theoretical and practical training course on the multidisciplinary approach to inherited ichthyoses
One theoretical and practical training course on the multidisciplinary approach to inherited ichthyoses will be held on June 18th and 19th, at Istituto Dermopatico Dell' Immacolata Hospital. The course will illustrate the epidemiology, the new classification, the cutaneous involvement and complications in non-syndromic ichthyosis, dermatological and systemic aspects of syndromic ichthyosis, the psychological and quality of life aspects and the treatment. A presentation will be dedicated to palmo-plantar kertoderma and the last session will be devoted to clinical case presentation on non-syndromic and syndromic ichthyoses. The official language is Italian and the registration is free (lunch and coffees will be provided).

Libya
From Mohamed Elhashme, Libyan Genodermatosis Task Force, EB coordinator

Since the participation of the Lybian Genodermatoses Task Force in the 2009 Together Against Genodermatoses working session in Greece, our three genodermatoses clinics continued their activities providing the usual care to all genodermatoses patients and new patients were added to our registry. Contact and communication between the group members became stronger more frequent through the Libyan Dermatology Association. A Libyan geneticist will be added to our task force members.
A two days course on genetics and genetic skin diseases was held in Tripoli Central Hospital on 16 and 17 January 2010 organised by the Libyan Genodermatoses Task Force and the Libyan Dermatology Association and sponsored by the Libyan Board of Medical Specialities. Dr. Sonia Abdelhak, a geneticist, from Insitut Pasteur in Tunisia was the invited speaker. Dr. Othman Mohamed a Libyan geneticist participated in the introductory part of the course while Dr. Mohamed Elhashme talked about the Libyan Genodermatoses data base.
In March 2010, a sientific meeting on genodermatoses will be held at Ibn Sina Hospital in Sirt.
A formal letter with the requirements of Libyan EB patients was sent to the Ministry of Health and we are working on that to secure continuous supply of wound care products and special clothes for these patients.
The fourth Libyan Dermatology Conference will be held by the end of this year and genodermatoses will get enough coverage.

Turkey

From Seniz Ergin, MD, Seniz ERGİN,MD, Pamukkale University, Faculty of Medicine, Denizli, Turkey
The accurate prevalence of genodermatoses is unknown in Turkey. Genodermatoses are not included in Mandatory Notification of Disease group unlike cancer so data is missing on this subject. However, there are regional data gathered and published from the hospitals' patient records. The occurance rate of genodermatoses was reported as 0.43% by Kokcam et al., as 0.37% by Baysal et al. and as %0.33 by Koc et al., in 1994, 1997 and 2006 respectively. Ichthyoses, palmoplantar keratodermas, neurofibromatosis, epidermolysis bullosa and Darier's disease were the most prevalent forms. Diagnostic procedures such as immunofluorescence mapping and electron microscobic tecniques for epidermolysis bullosa group are inadequate. Furthermore there are no active social support groups for genodermatoses in Turkey. There is much to be done on this issue that remain untouched.

News from the associations of patients

Association Epidermolysis Bullosa Bulgaria (AEBB)

In Bulgaria the people suffering from Epidermolysis bullosa hereditaria are united in Association Epidermolysis Bullosa Bulgaria (AEBB) which has been created and legally registered under the Bulgarian law in 1994, in the Department of Dermatology and Venereology at Medical University Pleven, Bulgaria by Prof. Mariana Traschlieva. AEBB is a voluntary, non-governmental charity organization with ideal object, with the only purpose to make Epidermolysis Bullosa (EB) patients'life better. AEBB is a member of the network of organizations for EB in the world – DEBRA (Dystrophic Epidermolysis Bullosa Research Associations) International since 1992.
The main tasks of AEBB are:

- to create a National Register and information Bank for the patients with EB in Bulgaria
- to support daily necessities of the patients with EB (medicines, dressings and food)
- to prepare information materials for physicians, nurses, patients with EB and their relatives
- to establish personal contacts between the patients and consolidate the international links with the organizations of EB individuals in Europe
- to create favorable conditions for raising, education, professional orientation of the patients with EB

AEBB has organized information activity:

- to favor informal communications among EB patients and medical specialists
- to publish bulletins about the association's activity
- to prepare information concerning different problems related with the disease
- to inform the public about the problems of the EB patients

to stimulate and organize charity to collect funds

to help the patients with EB to feel adequate members of society, to facilitate their life, to improve their physical and emotional status by joining them in to different and proper activities

Since 2005, we have established contact with Information CENTRE for RARE DISEASES AND ORPHAN DRUGS in Plovdiv, Bulgaria. In 2007, we become a member of National Aliance of the people with rare disease in Bulgaria.
Five workshops of AEBB have been organized in Bulgaria: in 2000, 2004, 2005, 2006 and 2009. The last meeting has been conducted with assistance of DEBRA house Austria (D-r Gabriela Pohla Gubo) and DEBRA Croatia (Mrs. Vlasta Zmazek).
> Visit the AEBB website

Debra Spain

Your help can give them wings...

EB is a serious disability. People suffering from this condition require intensive daily care from relatives or professional carers. They loose the use of their hands and feet due to the extent of the blistering and the retraction of the skin in these areas. As well as physical suffering, those affected by EB also face a number of social and medical obstacles.

Adequate treatment is very difficult to find due to the rarity of the disease. There are still only a very small number of medical consultants and reference centres available, and these are by no means multi-disciplinary, even though EB affects many aspects of a patients health.

A lack of coordination at treatment centres and training of social services staff, means that patients are often poorly or inadequately treated. Due to administrative separation of the health system between the 18 regions in Spain (Comunidades Autónomas), the economical and medical support received is not equal to all persons affected by EB.

The lack of support adds to the isolation of families as they find themselves in a situation where one person is required to become a dedicated carer and the financial burden is significant.

DEBRA Spain's Mission
DEBRA Spain, founded in 1993, is a non profit-making organization set-up to provide support and to improve the quality of life of those affected by EB. The charity works to provide the following:

Emotional and medical support to families and carers, including information, guidance and training

Specialized nurses who travel throughout Spain visiting families in their own homes or at hospital

Specialized psychologist and social worker who guide relatives and patients through the different resources which are available to improve their quality of life

Information and advice on all aspects of the disease, including the coordination with doctors and health services to ensure patients receive the correct treatment

Support for research programmes for a possible cure or treatments

Media campaigns to promote awareness & fund-raising through events and charity shops

A number of yearly national meetings to bring affected people together, as well as conferences for multi-disciplinary professionals

A link to DEBRA in all other countries

Continuous work to obtain private and public grants

DEBRA Spain's current projects

Respite Home in Marbella: A place where patients can come and stay for a few days of respite. Throughout their stay they are supported by a medical team, including a psychologist, nurses, a physiotherapist and a social worker, all provided by DEBRA Spain.

Accommodation for families in Madrid: A place to stay free of charge in Madrid for those families who need to take their children for specialist treatment at the reference Hospitals of Niño Jesus and La Paz.

Chimeric skin project: Clinical trial with chimeric skin grafts, involving 11 patients, finalising end

Second skin project: To produce a whole body suit that will work as a substitute for bandages, significantly reducing the time it takes for the daily healing procedures.

Set-up of a national reference centre, through the organisation of EB-days, networks of specialised doctors, etc

> Visit Debra Spain

Xeroderma Pigmentosum association in Algeria

www.xpalgerie.org: a new website
This website is dedicated to inform patients, families, specialists and increase general public and government awareness. www.xpalgerie.org provides information about the disease, the diagnostic, a list of contacts in Algeria.
> Visit www.xpalgerie.org
> Contact www.xpalgerie.org

Xeroderma Pigmentosum association in Tunisia

Tunisia: Children of the Moon!
An article by Kamel Bouaouina with the Tunisian Support Associationfor Xeroderma Pigmentosum Children published in Le Temps, a Tunisian newspaper in French.
This article includes:
- Interview of Dr Mohamed Zghal, Department of Dermatoloy, Hôpital Habib Thameur, Tunis, Tunisia
- Interview of Lamia Hakim, a young Xeroderma Pigmentosum Patient
- Interview of Noomen Hakim, President of the Tunisian Support Association for Xeroderma Pigmentosum Children

A new website for the Tunisian Support Associationfor Xeroderma Pigmentosum Children

Hayett Prize to the Tunisian Support Association to Xeroderma Pigmentosum Children
Hayett, a Tunisian insurance company, has created a contest to contribute to the efforts of Tunisian state in the field of child protection through support to cultural, educational and social works.
The prize (5 000 dinars - about 2645 euros) for the best educational and social work was ex aequo awarded to Association d'Aide aux Enfants de la Lune (Tunisian Support Association to Xeroderma Pigmentosum Children).

News from the working groups

To learn more about the working groups activities, please ask your login and your password and visit the private access working group website

Together Against Genodermatoses 2010

The 2010 meeting will be held in Rome, on 22-24 October 2010, in Ospedale Pediatrico Bambino Gesù, in the framework of the TAG project