Together Against Genodermatoses

N°5

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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 Rene 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).

2010 working session programme

The TAG meeting will be held in Rome on October 22nd and 23rd.

The goal of the meeting is to define the best available strategy for the management of each disease, in each country, focusing on patients' daily life: health care, nursing care, social care will be taken into consideration.

As usual, the coordinators of each disease working group will present the results achieved by their own group.
Moreover, the following topics will be discussed and developed:

- summary of collected data in each country;
- progress in collecting epidemiological data;
- assessment of the needs of patients and the costs for patients;
- development of centers of expertise;
- creation of associations of patients;
- review of the management strategies for genodermatoses;
- development of a national and international network.

During this meeting some innovations will be introduced: a transversal working group has been created to focus on the development of the network of laboratories for diagnosis and genetic counseling.

Moreover, experts of genodermatoses, who are not members of the working groups, will participate in the Meeting, and deliver lectures on recent advances in clinics and research of some diseases in their own countries.

Finally, an inter-religious debate will take place during the final round table on bioethics.

We are waiting for you to share knowledge coming from the different countries, and to improve our culture and our skills, with the ultimate goal of providing advanced care to children and their families.

Learn more on www.tag-eu.org

Together Against Genodermatoses 2010

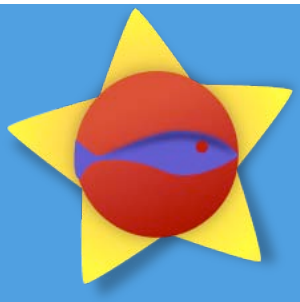
The 2010 meeting will be held in Rome, on 22-24 October 2010, in Ospedale Pediatrico Bambino Gesù, in the framework of the TAG project.
The TAG project (TAG N° 2007 335) has received funding from the European Union, in the framework of the Public Health Programme.

Executive Agency for Health and Consumers

Bambino Gesù OSPEDALE PEDIATRICO

National Plans or strategies on Rare Diseases adopted by the Member States

An update on the website of the European Commissions about the national plan or strategies on Rare Diseases adopted by Member States.
Learn more on
http://ec.europa.eu/health/rare_diseases/national_plans/detailed/index_en.htm#national



Our new logo

Thank you very much for your answers and your suggestions regarding our new logo. Please, feel free, to use it for your presentations.

The Rammal Award 2009

The Rammal Award 2009 rewards Fondation René Touraine for the development of the Genodermatoses in Mediterranean Network

Thanks to the involvement of every partner of the Genodermatoses in Mediterranean network, the Rammal Award 2009 rewards Fondation René Touraine.

The Rammal Award, created in memory of the great Lebanese physicist Rammal Rammal (1951-1991), is awarded each year to an personality (or an organization) who or which has elevated the level of scientific exchanges in Mediterranean area.

Learn more about the The Rammal Award Session and Prize Giving Ceremony at ESOF2010 in Torino

News from countries

Croatia: the management of Epidermolysis Bullosa

This contribution is mostly authored and based on the experience and activities of DEBRA Croatia, specially its President, Mrs. Vlasta Zmazek.

However, we would also like to point out the activities of our Dermatovenereological communityin the Genodermatoses and Mediterranean project during the last 5 years (Prof. Skerlev, V. Zmazek)

Concerning the 26 years experience (Mrs. Vlasta Zmazek) of living with child with EB and being in contact with more than 100 families from all over the world, we hope that these guidelines should be useful.

Debra Croatia is 24 hours service for its members

What are the needs of family with EB child?

- Information about disease & doctors
- Information about other families with same problem – to share experience in day-to-day problem
- Information on social rights and assistance in achieving those rights
- Encouraging in "normal life" – by sharing experience and social practice and/or by psychological workshops, by creative workshops.....
- Public awareness & lobbing

After 14 years of work Debra Croatia managed to provide the following services:

- Information about centres & doctors
- Additional trainings for specialists

News from the countries / Croatia

From Prof. Mihael Skerlev, MD, PhD, President of the Croatian Society of Dermato-Venereology, Dept of Dermatology, Zagreb University Hospital and Medical School, Croatia

From Mrs. Vlasta Zmazek, President DEBRA Croatia
http://www.debra-croatia.com/

- Education for parents (specially for newborn babies)
- Link between specialist & families collaboration is necessary because there is a need of work with families to realise how their sharing experience with doctors is precious and for easier and quicker appointements, treatments and check ups. -
- Advocacy for reimbursement of daily care costs (bandages, eg. mepitel, mepilex, needles,

We managed to assure (by advocacy) free of charge certain quantities of bandages, Monlycke products and antibiotic creams.

- Support for creams and other items for daily care which are not covered by health insurance (by financing those items)
- Apartments – Debra has two bedroom apartments for families while they are visiting doctors & hospitals in Zagreb
- Raising public awareness – by organizing all kind of events (concerts, exhibitions...) or appearing in media - especially on TV programs and radio. The most efficient are campaigns with special humanitarian phone number (to collect financial support for association)
- Socialization – we try to encourage people with EB to participate in "normal life" – for example: to go to regular school (not to have teacher at home – which is also a possibility), to go for holidays (in spite of difficulties to organize everything required), to drive a car and to be independent to the maximal extent of possible. Apartment provided has a great role in socialization – our members (teenagers or older) can use it just for weekend in town, socializing with friends, participating in workshops (creative), visiting cinema or concert.
- Meeting with school teachers and social workers of the family are considered to be very important, as well. Nurse & social workers who know and understand the problem are crucial support for family (Debra is financing such a nurse and her travel to families).

Social rights depend on each country legislative. There are certain categories Debra Croatia would recommended:

- monthly allowance for child with disability
- right for a parent to stay at home with child up to age of 7
- right for a parent to work half time and get paid full time (this is something Mrs. Zmazek would personally very much recommend)
- right to be so called "nursing parent" and get paid (solution for unemployed parents)
- to get twice or three times a week a patronage nurse and physical therapist (exercise).

"After all the most efficient and encouraging for families are meeting with other families to share experience and to learn how much can be done in quality of life. Quality of life for a child with EB definitely depends on the family ability to cope with the situation and on understanding of the whole society."

The development of an EB network in Croatia

- involvement of our Dermatovenereological community
- raise the importance of this issue in the eyes of the other medical specialists involved in the EB management.
- founding of the Referral centre for EB in our University Department,
- the multidisciplinary EB Committee of the Ministry of Health and Social Welfare of Republic of Croatia (co-ordinated by Dr Slobodna Murat-Sušić, dermatovenereologist and paediatrician specially dedicated to EB)

The active involvement of Croatian EB stakeholders in many international acitvities:

- Active involvement of DEBRA Croatia in many international activities
- Organization of the scientific regional symposia on EB,
- Participation in the *Genodermatoses in Mediterranean* network during the last 5 years (starting with M.Skerlev from the part of Croatia and including other important "key persons" afterwards, eg. Vlasta Zmazek)
- Formal and official decision of the Ministry of Health and Social Welfare of Republic of Croatia to become one of the patrons of the *Genodermatoses in Mediterranean* Project.

"We are enthusiastic and happy because of that what has been achieved, however, we are, at the same time, aware of the fact that there is still a lot to be done..."

News from countries

Czech Republic: the management of Epidermolysis bullosa patients and genodermatoses activities

The Department of Pediatric Dermatology (DPD) Faculty Hospital Brno was founded in Children´s hospital with complex child care in 1953.

DPD has focused on severe genodermatoses such as Epidermolysis bullosa congenita, Ichtyosis, Neurofibromatosis, Incontinentia pigmenti Bloch-Sulzberger, Ectodermal dysplasia, Tuberous Sclerosis for a long time. We collected about three hundred patient´s .

Our department is in close contact with the Centre of Molecular Biology and Gene Therapy .../...

.../...and this cooperation enables finding of affected mutated gene in EB patients and Neurofibromatosis.

EBCentrum

The EB Centre Czech Republic, http://ebcentrum.debra-cz.org founded by Hana Bucková MD in 2001, specialised in providing medical care and advice for patients with Epidermolysis bullosa and their families (children and adults).

There is an interdisciplinary team of dermatologist, gastroenterologist (children and adult), haematologist, ophthalmologist, anaesthesiologist, stomatologist, plastic surgeon, mollecular and clinical geneticists, pathologist, nutrition specialist, social worker, psychologist, and specialist in radiodiagnostics in the EB Centre.

A special ´EB Patient Clinical Day takes place once a year in the EB Centre and all specialists are focused on patients with the most severe symptoms at the same time.

We also provide complex care for EB patients with squamous cell carcinoma EB patients.

A National EB register

We have a National EB register of all EB patients from the Czech Republic. There are 153 EB patients of all three major types in EB centre, 39% dystrophic EB, 7% junctional EB, 54% simplex EB (1972-2009). Incidence of EB is 1:50 000 – 1: 70 000 patients. Diagnosis of EB is based on clinical symptoms, electron microscopy, antigen mapping and molecular genetic examination of blood. In molecular analysis of Junctional EB patients we cooperate at the European level with EB Hauz Salzburg. Prenatal diagnostics and preimplantation genetic diagnosis may be offered to patients once a mutated alela has been found. Six pregnant women have undergone prenatal diagnoses already.

From Hana Bučková, MD, PhD, Head of Department of Pediatric Dermatology, Faculty Hospital Brno and Faculty of Medicine Masaryk University Brno, Haed of Society of Pediatric Dermatology of Czech Society of Dermatology and Venerology, Member of Czech Medical Association of J. E. Purkyne, Coordinator of EB Centre and Consultant of DebRA Czech Republic, Consultant of Incontinentia Pigmenti Association (IPi)

News from countries

France: MAGEC, the expertise center for genetic skin diseases in Necker Children Hospital

MAGEC is the National Expertise Centre for Genetic Skin Diseases. Prof. Bodemer (Dept. of Dermatology - Hôpital Necker Enfants Malades – Paris - France) is the coordinator of MAGEC.

This centre unites activities of 3 departments of Assistance Publique - Hôpitaux de Paris:

- Dept. of Dermatology - Hôpital Necker - Enfants Malades (Prof de PROST) for care of children and adults with genetic skin diseases (head: Prof Bodemer).
- Dept. of Dermatology - Hôpital Avicenne (Prof LAROCHE) for care of adults with genetic skin diseases (head: Prof Caux).
- Dept. of Dermatology - Hôpital Saint-Louis (Pro BAGOT) for care of adults with genetic skin diseases (head: Dr Blanchet-Bardon).

MAGEC team is committed to improve health care and social support for patients with genodermatoses and works for an early management of patients with genetic skin diseases thanks to:

- Development of a network and of interactions between health professionals,
- Recommendations for health care for health professionals and patients,
- Partnership with support groups and families,
- Registries.

MAGEC website

www.magec-eu offers practical information about genodermatoses and the MAGEC.

« Tem'Peau », innovation in therapeutic education

To improve daily life of young patients and their families, the « Tem'Peau » group has been created. Based on patient needs, this group discusses and develops actions for patients and families.

Tem'Peau is a multidisciplinary team trained in:

- Management of rare diseases
- Impact of rare diseases on the daily life of patients
- Therapeutic education .../...

News from Countries / France

Tem'Peau team works on a voluntary basis.

Following the success of the 2007 therapeutic education weekend, another one is organized on March 19-21, 2010.

The 2010 Tem'Peau weekend objectives are to:

- Build links and promote mutual support
- Organize therapeutic education session
- Gather children, families and carers for a weekend and share daily life as well as care practice.

1...2...3 Tem'Peau is a new therapeutic education tool for patients with ichthyoses and their families. 1...2...3 Tem'Peau tool will be used for the first time during the 2010 weekend.

News from the countries / France

From C. Bodemer, Dept. of Dermatology - Hôpital Necker Enfants Malades – Paris - France

News from the associations

News from the Associations Czech Republic

From Hana Bučková, MD, PhD Consultant of DebRA Czech Republic

DebRA Czech Republic was founded in 2004 by the EB Centre, www.debra-cz.org.

DebRA is non-profit organisation for EB patients and their families. Social support, educational and informational activities are the main DebRA activities provided by three employees. DebRA is part of the Children´s Hospital too and works together with the EB Centre. A week´s curative stay for EB families and DebRA conference are annually organised by DebRA CR. Employees have a close relationship with EB families (internet, telephone).

DebRA ČR

This organisation is funded by Government grants, donors and sponsors. Thanks to this sponsorship we ensure financial support for EB patients and their families.
DebRA CR cooperates with DebRA International and DebRA CR organised the DEBRA International Congress 2009 10. - 13.9.2009 in Prague.

Incontinentia pigmenti (IP) organisation IPi

IPi organisation was founded in January 2010 by parents of children affected by IP to improve communication among IP families and serve as a psychosocial support for IP patients and their parents.

The aim of these communities is to reduce isolation of people with rare diseases and their families.

France: the French Association on Ectodermal Dysplasia

AFDE, the French Association on Ectodermal Dysplasia has been cretated in 2000 to support patients and families.

The AFDE missions are:
- inform patients and families
- encourage social integration
- improve health care including oral and dental care
- encourage exchanges with health professionals, media, patient associations
- support research on ectodermal dysplasia

AFDE supports the online international registry hosted by the National Foundation for Ectodermal Dysplasias. The purpose of this registry is to consolidate information on those affected by ectodermal dysplasia into a single data repository.

To learn more on AFDE, you may visit their website: http://afde.net.

Information about ectodermal dysplasia is available in English on the Ectodermal Dysplasia website, www.ectodermaldysplasia.org

News from the Associations / France

Oliva Khalifa, Presdident of the French Association on Ectodermal Dysplasia (AFDE)

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