DEBRA the Netherlands is very proud to offer you this congress-special with the summaries of the International DEBRA Congress October 2011 in Groningen. Of course we did our very best to construct correct reports of all the lectures, forums and presentations, held during this congress, although English is not our native language.

But we also produced a digital version of this special, to be found at:
www.debracongress2011.com, or:
www.debra-international.org

This version contains a lot of possibilities for you, which a printed version can't always offer. For example:
-You have the possibilities to send this digital version to other people with a special interest in EB.
-You can select and print the abstracts which are important in keeping up your own archives
-You can select the summary that offers a lot of relevant information for your medical specialists or other assistances you see on a regular base as an EB-patient.
-You can exchange information with the presenters of the lectures; a lot of them can be contacted through their email-address.

For our Spanish speaking colleagues.
DEBRA Nederland is also very glad to draw your attention to a PDF file in a Spanish version. DEBRA Nederland made this decision after hearing the critical remarks during the congress; not everyone is capable of speaking English fluently or trying to absorb all the interesting information the English speaking lectures gave. We hope you will profit from these translations.

You can find it at:
www.debracongress2011.com, or:
www.debra-international.org

International debra congresspecial

Organised by Debra International
UMC Groningen department dermathology
Wenckebach intsitut
Debra Nederland

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debra

Epidermolysis Bullosa

www.debra.nl

PREFATORY NOTE

Finally it is happening; it is Thursday 27 October 2011, and the Fountain patio at the University Medical Centre in Groningen became packed with the participants of this International Debra-congress. For all the members of the Executive Committee of DEBRA the Netherlands, preparing this congress was an important moment. Now it will be clear if all the efforts and preparations will be successful; and I think they were in the following days.

Almost 200 participants from 27 countries were entirely enthusiastic about the scientific, social and psycho-social lectures offered within the daily programs. Apart from the sessions, there was enough time and possibility to exchange information en thoughts, to share experiences, which made these days an unforgettable happening for many DEBRA members .

The presence of a little market with all kinds of ointments, special bandages and gauzes made it possible for those present to get informed on the subject of the newest developments in the daily treatment of EB.

What also was very special during this congress, was the declaration of the universal rights of people with EB. Concluding we can say that we can look back at very special and successful days, thanks to the efforts of a lot of co-workers.

By this I want to give special attention to a few of them; Frank Houben, the great instigator of this congress, Malte Westheide, our treasurer, taking care of all our financial tasks, en especially Harry Gubbels, from the Wenckebach Institute in Groningen. He and his team took perfect care of all the organizing and practical jobs concerning this congress.

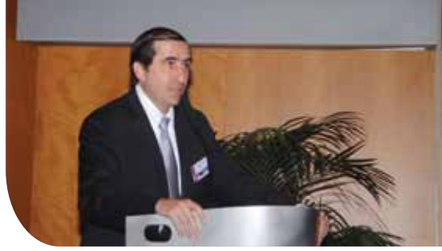
Apart from his help we also got a lot of support from John Dart, who, by means of his great experience in organizing congresses like this, could give us a lot of advice. Besides that we want to thank also all the chairmen, the interpreters, minutes-writers and speakers, with special attention for Dr. Marcel Jonkman,who took the initiative to give young scientists the opportunity to do a presentation.

The congress was able to get organized with the help of sponsors, especially our sponsor Molnlycke .This Company financed also the last international DEBRA congress held in the Netherlands in 1999.

We are also thankful to the VSB-fund who made it possible for Dutch DEBRA-members to visit this congress for a reduced fee.

We hope with this Congress-special to express our thanks to all of the participants who helped to make this congress successful.

Ank ten Siethoff-Bijkerk, Chairman DEBRA the Netherlands.



F. Palisson

Francis Pallison, MD, CHILI

An introduction to best practical EB-guidelines.

History :
During the International EB meeting in Belgium 2008, the President of DEBRA International, Mr. Graham Marsden, defined: "One of the best form to improve the quality of life in EB patients is to develop best clinical practice guidelines in every aspect that they need".

Starting to develop this initiative there was first a need to think about the protocol concerning the development of these guidelines. These thoughts led tot the following outcome; " We want to resolve specific clinical problems in the best way, based on a systematic and careful review of all the literature published in the field. This is reviewed by an international committee of experts and a consensus of re-comendations is generated, all approved by an international group of EB patients/parents. Finally, these are re-checked by the group of experts and published for world wide use."

Of course this kind of approach caused problems :
- It was a huge amount of work.
- It had to be on an international level, multi-centered, involving not only a group of experts, (because that could be a partial view) but also EB-patients and their care-takers.
- There are many items concerning EB that are not published and even not studied yet.
- There is new evidence coming, so there is a constant need to re-check the guidelines periodically
- It takes almost 2 years to develop a guidline; it is expensive.

But the advantages were and are:
- In this way you can standarise the best care treatments and recomendations for EB
- It provides teaching opportunities for professionals and patients /relatives about the clinical problem
- This approach stimulates the interaction between different specialists from different EB-centers, helps to share experience and define new protocols for Clinical Research.
- It creates the possibility to receive the feedback from the leaders of the different EB-groups, working on these topics on their current recommendations.
- Welcomes the comments from a patient perspective to integrate those comments into the draft guidelines.
- Stimulates all the persons concerned to ask all type of questions to define future guidelines.

The next presentations will be based on (the development of) the following guidelines:
- Dental care
  (Is published ; see presentation of Susanne Krämer)
- Pain Management
  (Is going to be developed ; see K. Goldschneider)
- Occupational Therapy
  (Will be published in 2012 : see F. Prinz)
- Cancer in EB
  (Is going to be developed : see J. Mellerio)

More information about the guidelines; www.debra-international.org

Susanne Krämer, DENTIST, SANTIAGO, CHILE

Best clinical practice guidelines in dental care in EB

EB is a rare disease with multiple oral manifestations, which require a special approach from the dental point of view. Due to its low prevalence, most of the dentists don't know about the disease. The scientific literature is scarce, making it difficult for dentists with no experience on EB to know how to approach a patient with EB in a safe manner with all the special care this group of patients need.

To develop the "Best Clinical Practice Guidelines Dental Care for Patients with Epidermolysis Bullosa" a group of health care professionals with vast experience in EB was gathered. It became necessary to gather experts from different centres around the world to discuss about the different treatment alternatives. A standard methodology based on a systematic review of the evidence was followed.

The aim of the guideline is to provide the users with information on the special cares people with EB require before, during and after dental treatment. These guidelines can be applied to all patients diagnosed with Epidermolysis Bullosa. As such, the guideline considers information for all four major types of EB: EB Simplex, Junctional EB, Dystrophic EB and Kindler Syndrome. The users are expected to be Specialists in Paediatric Dentistry, Special Care Dentistry, Orthodontics, Oral and Maxillofacial Surgery, Rehabilitation and General Dental Practitioners, Dental hygienists, Paediatricians, Dermatologists, Dietitians, parents and those living with Inherited Epidermolysis Bullosa.

These guidelines contain a chapter on general information on dental care of patients with EB, followed by a chapter explaining the precautions that should be taken into account when treating patients with each subtype of EB, as well as recommendations for dental treatment. The appendix includes a glossary, general information on EB, a description of its oral manifestations and an information sheet with oral exercises.

Kenneth Goldschneider, MD,

Pain management in EB; developing a practical guide

Inherited Epidermolysis Bullosa (EB) comprises a group of rare disorders, generally thought of as skin diseases. However, EB has multi-system effects and patients present with a number of both acute and chronic pain care needs.

Effects on quality of life are substantial. Due to its low prevalence, expertise in pain care for patients with this disease is often restricted to a few specialized care centres. Even then, evidence-based pain care is limited by a near absence of scientific literature specific to EB. This set of guidelines was requested by DEBRA International, to help standardize the approach to pain care for patients with EB in all parts of the world. Consequently, a group of clinical pain care experts from a few countries have come together to lend their experience to the limited scientific literature to create these guidelines. In order to generate useful clinical guidelines, an approach was taken that combined extrapolation from related literature and expert consensus.

The present guidelines on pain care for patients with EB are based on a review and synthesis of the available literature, guided by expert consensus and thoughtful application of theory. The guidelines are divided into three topics: acute pain, chronic and recurrent pain, and special topics. The acute pain section focuses on postoperative pain. The chronic and recurrent pain section includes dressing changes, baths, skin pain and joint and other body pains. Special topics include pain care in infants with EB, psychological approaches to pain care, and palliative pain care. While not pain per se, itching is a major source of discomfort and is also discussed. For each topic, research questions are presented to act as prompts for thinking about the next steps in producing evidence to support updated guidelines.

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Florian Prinz and Hedwig Weiss,
DEBRA- OOSTENRIJK TEAM, (ERGO) THERAPEUTEN

Occupational Therapy in Epidermolysis bullosa

A holistic concept for intervention

from infancy to adult

Epidermolysis bullosa (EB) is the collective term for a heterogenous group of genetic skin disorders characterised by the formation of blisters and erosions on skin and mucous membranes.

This book deals with early childhood motor and perceptual development, a particular challenge for children with extremely fragile skin. Going into functional treatment, it addresses rehabilitation of hands and feet in EB patients with fusions or contractures.

The authors are occupational therapists with long-term experience in the management of EB patients. In an extraordinary holistic approach, they present a combination of paediatric and functional areas of treatment.

Occupational therapy tools and interventions can help alleviate a number of accompanying complications and symptoms of EB, enhance self-care, and improve quality of life in general.

Due to its practical approach, this book not only serves as a unique guide for doctors and health care professionals but is also comprehensible and useful for patients.

De Donderdagsprekers

From the Contents:

Occupational Therapy in Epidermolysis bullosa

- Epidermolysis bullosa (EB) – The Condition
- Motor Development in earlychildhood
- Treatment of the tactile, vestibularand proprioceptive perception
- Development ofhandfunctions
- Graphomotor skills
- Independence in every-daylifeand the provisionofassistivedevices
- Client-directed therapy using the COSA
- Rehabilitation of the hand
- Rehabilitation of the foot

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A holisticconceptforintervention from infancyto adult

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AFTERNOON-SESSION

Squamous Cell Carcinoma

(SCC)

Introduced by:
DR. FRANCIS PALLISON

Generally speaking SCC is discovered in healthy people as well as in RDEB-Patients. But there is a sad difference: People with RDEB die from SCC at an early age; 90, 1% of RDEB-Patients die after they reached the age of 55. And getting older, this risk of dying is even much larger. The second problem is that SCC is hard to diagnose; a lot of RDEB-patients have so many ulcers and wounds at their hands, feet and elbows that it is hard to distinguish them from SCC.

Anne W. Lucky, M.D., volunteer professor of dermatology and Pediatrics University of Cincinnati College of medicine and co-director of the Cincinnati Children's Epidermolysis Bullosa, Cincinnati, Ohio, USA: A case history; a 20 year old girl with invasive and metastatic squamous cell carcinoma.

Before beginning her PowerPoint presentation, Dr Anne Lucky apologized on beforehand for the shocking images this presentation would show; especially the impression the images would made on the EB patients among the audience.

A 19 year old girl was presented with a non-healing, tender ulcer in the left shin in 2005. Although she was treated with silver dressings, the ulcer continued to enlarge. At surgery one found that the young girl had much more invasive and more extensive squamous cell carcinoma than was expected, and below the knee an amputation had to be performed. But after that, in more ulcers at the left upper thigh, which contained also staphylococcus and streptococcus, a biopsy showed that the thigh also was threatened by invasive SCC. Finally the young woman refused further evaluation and treatment; because therapies were not successful. In 2006 the patient was removed to a hospice, where she died peacefully but much too early, 14 months after the diagnosis with SCC.

Summary:
- Squamous cell carcinoma often occurs in the 3rd decade of life.
- It is rapidly progressive both to locally and to distant metastases, often at the time of presentation.
- It is poorly responsive to therapy, but there have been very few trials.
- Patients may choose to refuse further diagnostic and therapeutic intervention except for comfort care, especially pain management.

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Jemima Mellerio, OF PAEDIATRIC DERMATOLOGY,
Great Ormond Street Hospital for Children NHS Trust, London, UK

Best clinical Practice guidelines in cancer management and EB.

Especially RDEB-patients carry a great risk getting Squamous Cell Carcinoma (SCC). 80% of people with RDEB have, as they reached the age of 45, experience with this for them very aggressive skin cancer-variation. The malicious cells are mostly found in the skin of the hands, elbows, feet and legs. And with EB-patients, having a lot of blisters and ulcers, it is hard to distinguish skin-cancer from these "normal"wounds.

The rarity of this genetic disease, in combination with the way SCC goes off in RDEB-patients strengthens the need to develop a practical guideline for cancer-management. The guide is being constructed with the same method Dr. Francis Pallisin showed in his introduction in the morning session: "The development of the guide is based on a systematic and careful review of all the literature published in the field. This is reviewed by an international committee of experts and a consensus of recomendations is generated, all approved by an international group of EB patients/parents. Finally, these are re-checked by the group of experts and published for world wide use."

Dr Mellerio informed the audience of her recommendations : how to obtain to make a quick diagnose of SCC:

- Check every three months the skin of the RDEB patient, done by nurses and dermatologists with enough experience in this area .
- Akl the mediacal team, and also the patients, parents or partners at home, to take photo' s of wounds, ulcers and blisters , just tot see how the skin is behaving
- If the images give reason to be worried; take 4 mm biopsies
- Instruct the patient in a clear, discreet and and tactful way; help them in this way tot check their skin as careful as possible.
- If surgery is the only option left; choose dermatologists, surgeons, oncologists and pathologists to compose a team. After hat surgery histo-pathologists have to search for metastasis.
- The removed texture has to be searched by a pathologist with RDEB experience.
- Frequent checks of the lymphatic glands can be part of the treatment; but it is known that biopsies of these glands does not give any decisive answer on the subject of SCC.

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jemima.mellario@gstt.nhs.uk

There are still no succesful alternatives tot help in the struggle agianst SCC in RDEB-patients.
A few known experiments :

- Radio-therapy : unsure, not enough experience.
- Chemotherapy : the results do vary a lot.
- Tyrosine Kynase : is only used in the case of metastasis
- Aldare-cream : there is only one case-history of a patient using this cream. (Aldare-cream is used when skincells show deformities)
- Using Cetuximab : Only one case is known. (Cetuximab is a targeted therapy ; aims at slowing down cancer-cells.)

As long as there is no real cure or suitable therapies for healing SCC, removing the SCC-skin-fragments stays the best option.

Mark de Souza
PHD: RESEARCH

Lotus Tissue Repair Inc. and the development of a protein therapy.

There are currently no approved treatments to heal RDEB. But now Lotus Tissue Repair Inc. has announced the closing of a $26 million Series A financing led by Third Rock Ventures. Proceeds of the financing will be used to advance the company's proprietary recombinant collagen type VII (rC7) technology as the first and only protein replacement therapy in development for the treatment of RDEB, an orphan disease that causes devastating skin blisters, morbidity and early mortality.

Future applications of the company's technology include treatments for dermatologic conditions in which rC7 may play an important role in accelerating chronic wound healing, such as diabetic foot ulcers, venous stasis ulcers and similar conditions. The company thinks in 2 to 5 years to have found a successful therapy for RDEB.

The first experiments with human subjects, has been good: and showed also results on the long run.

The scientists and founders of this company Dr. Mei Chen and Dr David Woodley, are the initiators of this new therapy; now known as Recombinant Collagen VII technology. (Rc7)

They both are experts in the application of Rc7 as a protein replacement therapy. In short and simplified: the mutated protein Collagen VII is replaced by a none-mutated type of collagen VII.
Now Lotus Repair Tissue Inc. would love to have the cooperation of the members of DEBRA International.
The company-researchers discovered that after a critical survey, the international registered number RDEB-patents were substantial more than was known; 1: 250.000 RDEB patients in the USA (3 to 4 times higher than was officially registered.)
(In Europe there seem to be 1: 1.3000.000 people with RDEB, according to Mark de Souza.)

If all members of DEBRA International choose to participate in a new and updated RDEB- registration, it will soon be clear what the costs are in treatments of RDEB-Patients. This information can help to motivate the development of expensive therapies from a financial perspective; a situation in which both parties will get the most optimal success.
Mark de Souza asked the International DEBRA–members to give information to his company concerning the number of EB –patients, registered in their own country. He would like to have this information within 14 days.
All the members of DEBRA international were enthusiastic about this initiative and will be eagerly waiting for the results of this new therapy, developed with help of an enormous financial injection. They all hope that the research to a definitive cure for RDEB-patients will be successful.

More inquiries:
http://www.lotustissuerepair.com
lotus_info@lotustr.com

Joy van der Stel in dialogue with Yvonne Sellies,
COUNSELLOR DEBRA THE NETHERLANDS.

The power of my disability; the life story of an inspiring woman.

More Information:
www.empowermij.com
(with an English written variation)
Email: joy@empowermij.com

Counsellor Yvonne Sellies introduced Joy van der Stel, a young woman who turned to be spastic after birth; caused by anoxia. The parents of Joy reacted quite differently in relation to their daughter's disability: the mother was rather sceptical about the quality of Joy's future life. The father had different expectations; he was more convinced that Joy still would have a lot of possibilities. So Joy was really challenged by her dad to do as much as possible with her life.

But is being taken care of in general leading always positive?
Joy grew up at home till she was twelve. But after that she entered a boarding school for disabled children. And there, instead of doing as much as she could do herself, everything was taken out of her hands. Joy missed the challenges, and that made her feel more handicapped than she felt before... It was quite a frustrating dilemma in the 5 years living in that institution. At home she was considered a real human being, a real personality, but in the boarding school she was treated like a patient in the first place.
Joy missed in her supporters the belief in what she herself was able to do, in spite of the fact that Joy knew perfectly well her own barriers.
And she realized: if you stick to an institutional approach like this, you take the risk to get hospitalized in such a way that it can get very frustrating for your future development.

For all children and grownups with EB there are a lot of hampers to take, but also there are challenges to answer too. And why should you spoil the dreams of handicapped children if they want to be something in their future that is not very realistic?
Joy 's opinion is that everybody has his own right to his own dreams, and people as grownups know of course what they are able to do and what they want of their future.
If you get older some of your dreams will vanish, step by step you will get more realistic.

After the period in the boarding school Joy moved to Het Dorp (The Village), a community specially built for people with a physical disability, completely provided with all the facilities you need for your daily life.
A very protective surrounding, totally made fit for the life of handicapped people; the only inhabitants of this little village. Was it a friendly meant ghetto? Positive discrimination? (The village as it was does not exist anymore in the Netherlands. The integration of disabled people with a common community is preferred nowadays). Luckily Joy finally got an apartment of her own, and she is quite able to live there as independently as is possible. She is married and has a beautiful daughter.

Concluding advises of Joy:
Being mentally and psychologically strong is a must for diasabled people; sometimes you even need to have real challenges to change negative energy into positive feelings. And always hold on to your aims, targets and dreams.
If you have EB there are sure a lot of restrictions in your life you have to cope with, but also think of the possibilities you have; enjoy life!

After this presentation the audience was given the opportinity to buy a book, written by Joy:
"The power of my disability;
the life story of an inspiring woman".

Kim van Zijp
MOTHER OF A 9 YEARS OLD JESSE (JEB NON-HERLITZ)

Combining Parenthood with intensive Care.

August 19, 2002: Jesse is born; the first child of Kim. But soon the parents saw that something was wrong; blisters everywhere; Jesse was born with Junctional EB, non-herlitz, and that would mean a life of pain, itch and fear.

Kim had looked forward to the birth of Jesse, and really wanted to be a mother. But from the first day of his birth the world turned upside down, different rules applied.
A few days after the diagnose of Jesse, Kim had a consultation with a social worker, who told her to demand as much help as she could. But Kim ignored what she meant at that time. Yet it soon became clear how intense Jesse's needs and care actually were; day and night feeding, giving medication, caring and bandaging wounds, blisters and stinging ointments. And every action took a lot of energy, because the guilt caused in having to hurt their child while treating him was indescribable. And at that time Kim understood what the social worker had intended: this she could not endure any longer ,taking care of Jesse in this way would last for many years.

So finally Kim was convinced that she had no choice but to share the care and regular nursing had to be arranged. But Kim wanted to set up the care for Jesse by nurses according to her own very high standards, and some nurses could not cope with this. Finally she had to accept that other people took care of Jesse in their own way, and lowered her standards, because the disease causes the pain, not the caregivers. Kim really wanted to be a mother and not a nurse; she wanted the nurses to 'hurt' him so that she could read to him afterwards, make jokes, hugging him, could go out with him, and do what mothers do. And although she and her husband were the best caregivers, it was impossible for them to take on Jesse's care all by themselves. They wanted be parents first, wanted to console him and be there for him, and did not want to give EB a central place in their lives.

Soon after Jesse's birth the parents met Jany Fisher, director of a day-care centre with nursing care. Jany was a very skilled nurse, understood immediately the situation and saw what was needed. She took Jesse in for three days a week and she bathed and bandaged him. She gave Kim the confidence that she and her colleagues were just as good as Kim was. For Jesse it was also nice to spend time with other children. In the time that Jesse was in the centre, Kim could look for competent nurses who could take over the bathing on the other days. And that did not prove to be easy, because unfortunately, not every nurse is equally knowledgeable and involved. Think what it means investing in people when they do not appear to be suitable of reliable, at the expense of Jesse. What it means for your privacy to have nurses on the floor at odd hours. Or the soil on the floor, because of dirty shoes, or because a nurse commented on the mess in the house. Or because they pour out their heart to you because they are so sorry for Jesse.

Still Jesse' s parents were and are of course responsible for his care, like managing the nursing, financing, supplies, supervision and so on. As a parent of an EB child, there are still the injuries, feeds and stools, clothing, school. You cannot outsource being a parent of an EB-child and that is already stressful enough.

Kim kept going and held on, for nine years now. It was a lonely battle, in Kim's opinion also with the world. She came across a lot of misunderstanding. Stop work and take care of Jesse at home seems the obvious solution, but not for Kim. Unluckily, there are few who understand. Fortunately, Kim's husband and she agreed, they hold on to each other. So does their immediate family; without their support and those of highly competent nurses they would not have outlived all the problems.

That is how Kim finally achieved space to refuel. She could be a mother, work again, and be Kim again. They could afford to go on a holiday, and even look forward to a next pregnancy; with twin-sisters as a result!

But life still is not easy; for instance setting limits is a central theme in pedagogy. Every parent will recognize the boundless demands of children and their efforts to set limits and keep them. Children need boundaries to learn to set limits themselves. But with an EB- child it is different: this child is bordered by pain. A more suitable topic for EB parents is how help your child across boundaries.

Jesse is nine years now, has an average weight and length compared with healthy boys. It's priceless how Jesse, after all the problems related to feeding and stools, can now enjoy oysters, an English breakfast and feast on a nice chicken leg. Food means so much: it is growing, enjoying it together and it is pain.

To conclude the presentation Kim suggested the following device to the audience:
'What you yourself can do, you have to do, but not at the expense of your own wellbeing. Because you can´t take care of others if you can´t take care of yourself....'

Humphrey Hanley
NEW ZEALAND

Living with EB

Having REB;
what will be the key to a life that is as healthy and especially as happy as possible, in spite of having EB?

In any case it is important to search for a right balance between the misery EB causes, and the effort to try to reach as much independency as you can get.

For newborn babies with EB, constantly having pain and daily being bothered by the treatment of their wounds and blisters, life can be a real torture. The general opinion in New-Zealand is that parents of young EB-children should not be directly involved in the changing of gauzes and bandages every day.

It was the reason that an institution named Family Care (originally set up for asthmatic patients) got involved in the treatment of Humphrey. The parents did no longer participate in the changing of gauzes, and in this way got a better and more normal relationship with their son. This approach had a positive effect on Humphrey; he did well, grew into a reasonably mobile teenager and became more and more independent. Of course his peer group took his skin disease into account, but in general their way of dealing with Humphrey was quite common.

"Try to live as happily as you can, even if you have EB."

Nowadays Humphrey has a girlfriend, and managed to get a bachelors degree. He's having a job now in a big power-company. Working there asked for a few adaption's, like a PC with a headset, and being able to have flexible working-hours. In this way it was made easier to integrate treatments in his daily working life.

Humphrey, is spite of EB, still aspires to function as normal as possible .He took too much risks now and then, but also learned to handle this ; like participating in sport; horse driving, diving, water boarding, skiing and cricket. Taking a part in all those sports became possible for him, although sometimes the sport-outfits had to be adapted with extra protection material.

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Matija Zmazek, CROATIA

Have every confidence in your own possibilities.

Every youngster, and of course also young people having EB, ask themselves the same questions about the meaning of their life; What are my dreams to come true, what are the aims I want to fulfill in my own life?

Matija wanted to find his own answers to these questions, to find a real solution for the social-emotional problems EB causes. He thinks that you can look at EB as something that just belongs to him, is a part of him. And in this way he wants to be received and looked at by family and friends with an open heart and mind, without being too much reminded of his EB condition. There is much more to life than being constantly focused at and being aware of the deformities in the appearance of a people with EB. It is very important for Mattija to struggle for independency; looking for your own choices, making your own decisions, facing challenges yourself.

Mattija gives another valuable advice to people with a medical profession, parents and friends: Have more confidence in people with EB; have enough trust in the possibilities these people have, despite their condition.

If you want to get in contact with Mattija:
vlasta@debra-croatia.com

The discussion after the presentation of Humprey, Joy and Mattija was above all based on the following remarks and questions:

- Children with EB, and it does not matter which variation they have, should visit a " normal " school , to get really integrated into the daily life of children of their own age. Sport lessons or other activities bearing risks for EB can always be avoided or making fit for these children. Give enough information; but be aware of the fact that not all the teachers are willing to give enough attention to extra care.
- No matter what disability you have, trying to reach an utter independency is very important. Try to participate in all the common activities other people do, even if it can bring you some disappointments.
- How can you, despite the encouraging stories of the 3 presenters' this afternoon, hold on to a positive way of living, if you hear all the sad stories about dying of SCC?
- This knowledge forms is an enormous dark cloud hanging above your head, but it can also turn into a challenge to get the best out of your life as long as you can.
- For parents of young children it can be very hard to agree upon putting barriers between taking good care and overprotection.
- Should parents get involved in the changing of gauzes and bandages? Is it better to leave that to professional caretakers?
- For the next international DEBRA-meeting, it is strongly advisable to give again a lot of attention to the quality of the life of children and grown-ups with EB.

M.D. Marcel Jonkman

The presentation of research results by young scientists.

Directly involved with the organization of this DEBRA International meeting and supervisor of the EB-Centre in the Netherlands, M.D. Marcel Jonkman took an interesting initiative.

Since 1990 a lot of scientists, involved with research and EB, are still working in this area. And without being negative about " old boys network " there is also a new generation of researchers with their own questions. To give these young scientists an opportunity to present themselves, he invited them to tell the public of the results of their research or visions in relation to EB. But he also made them the following condition; try to translate your scientific way of putting things into a clear and transparent lecture for a broad audience!

To make it even more exiting; the public could, after these morning sessions vote for the best presentation!

Dimitra Kiritsi MD
DEPARTMENT OF DERMATOLOGY,
UNIVERSITY MEDICAL CENTER FREIBURG, GERMANY

Bijzondere EB-vormen

A few examples of selected cases of a special type of EB:

A variation of JEB was discovered with an 83 years old woman; (a late onset).
Her hair and fingernails were healthy looking, but her toenails were dystrophic, and she suffered from occasional localized blistering. Two of her sisters showed the same deformity.

The clinical spectrum of Kindler syndrome (KS):
KS is a fourth variation of EB newly discovered and named in 2008; it is also caused by a collagen mutation; leading to blistering on the skin of the hands, gums and intestines. It can occur at a later age and can be a late diagnosis of KS. But it is difficult to diagnose; is it a 'normal 'DEB or KS?

Lethal congenital epidermolysis bullosa (LECEB):

A homozygous nonsense mutation in the *JUP* gene leads to absence of plakoglobin *in situ*.

APSS
A deformity, showing a lot of resemblance with EB; it demonstrates itself also by deformities in the outer skin.

Summary
- Late onset and very mild phenotypes of JEB and KS exist; diagnosis of hereditary EB at an advanced age.
- LECEB: congenital, extensive epidermolysis, complete absence of hair and onycholysis; caused by loss-of-function mutations in the JUP gene
- APSS: an important differential diagnosis for EBS

Peter van den Akker

Phenotype prediction in dystrophic epidermolysis bullosa

Dystrophic epidermolysis bullosa (DEB) results from mutations (= errors) in the gene (= genetic code) for type VII collagen. This gene is called COL7A1. Type VII collagen is needed to bind the two layers of the skin together. A gene can be compared to a long barcode on an article in the supermarket where each bar represents one DNA molecule. If something is wrong with one of the bars, the scanner at checkout cannot understand the barcode. If something is wrong with one of the DNA molecules (= a mutation), the cells of the body cannot understand the gene. In case of COL7A1: type VII collagen will not be made at all or only in dysfunctioning form and as a result someone gets DEB. Everybody carries two copies of COL7A1: one from the mother and one from the father. DEB can be inherited in two ways: dominantly (DDEB) or recessively (RDEB). Dominant means that a mutation in one of the two copies of COL7A1 is enough to get DDEB. Recessive means that someone needs a mutation in both copies of COL7A1 to get RDEB. The nature of the mutations that cause DDEB and RDEB is different.

RDEB is usually more severe than DDEB, but there are many subtypes of both. The typical features and disease course of a subtype are collectively called "the phenotype". Newborns with different phenotypes can have a similar presentation. This makes it difficult to determine the exact phenotype in newborns and thus to accurately predict the future course of disease, while this is so important to parents.

In our research, we try to get more insights into the complex processes that collectively determine the phenotype, in order to be able to better predict that phenotype. DNA analysis is of great help here, as mutations are the best predictors of the phenotype. In order to make the information on all COL7A1 mutations and DEB patients ever identified available and easily accessible to everyone, we have built the International Dystrophic Epidermolysis Bullosa Patient Registry (www.deb-registry.org). Mutations found in a patient can be checked online for free against the registry to see what is known about the mutations, which is a great help in clinical practice.

In the peculiar "RDEB-inversa" phenotype, blisters are usually restricted to the trunk and the body folds (axillae, groins, gluteal fold), but there can be severe swallowing difficulties due to esophageal blistering and narrowing. Patients with RDEB-inversa therefore need specific check-ups. We have found that RDEB-inversa is caused by specific mutations in COL7A1, which enables us to correctly predict this phenotype already in newborns with DEB.

Another good predictor of the phenotype is the amount of type VII collagen in a skin biopsy. We have found that RDEB patients who have a very low amount of type VII collagen in their skin do not get the most severe "RDEB-severe generalized" phenotype. They do develop fusion of the fingers and toes (= pseudosyndactyly), but it starts only after the age of 7 years and is not as severe as in "RDEB-severe generalized". We named this phenotype "RDEB-severe generalized with late-onset pseudosyndactyly".

MMP1 is a protein that degrades type VII collagen. Several groups have studied whether MMP1 influences the phenotype in DEB. However, the results were contradictory. We have also studied MMP1, but we could not confirm that MMP1 influences the phenotype in DEB.

Altogether, our work helps to better predict the phenotype and thus the future in children with DEB, which is so important to the parents and the child.

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M.C. Bolling
RESIDENT IN DERMATOLOGY; PHD STUDENT AT UMCG

Cardiocutaneous syndromes

Skin and heart have more in common than it seems at first sight. Both tissues are subject to shear mechanical stress and the intercellular structures that need to provide strength yet also flexibility, the desmosomes, show remarkable resemblance in skin and heart. Mutations in genes encoding desmosomal proteins shared by skin and heart cause (desmosomal) cardiocutaneous syndromes. The most well known are Carvajal syndrome (mutations in desmoplakin, gene: DSP) and Naxos disease (mutations in plakoglobin, gene: JUP) clinically characterized by skin fragility, palmoplantar keratoderma, woolly hair and cardiac disease leading to early demise. The skin abnormalities are present from birth or early age, and may therefore provide important clues for the presence of cardiac disease. With early recognition, regular follow-up, medication, and eventually an intra-cardiac devise, lifespan can be prolonged significantly.

We discovered mutations in another desmosomal protein, plectin (gene: PLEC1), in a patient with skin fragility resembling epidermolysis bullosa simplex (EBS, skin blistering with a level of blister formation low in the epidermis), mild muscular dystrophy from ~age 30, and a cardiomyopathy. This adds plectin to the proteins involved in cardiocutaneous syndromes. Currently a group of patients with arrhythmogenic right ventricular cardiomyopathy (ARVC), a pure cardiac, autosomal dominant disease characterized by heart rhythm disturbances and cardiomyopathy leading to sudden death or heart failure at an early age, which is in most cases caused by dominant mutations in desmosomal proteins, is investigated for mutations in PLEC1 encoding plectin. As mentioned above, early recognition of potential cardiac disease within families (especially in case of autosomal dominance) can be lifesaving.

We have also investigated a group of patients with pure EBS in which no mutations were found in the currently known genes involved in EBS (KRT5 encoding keratin 5, and KRT14 encoding keratin 14) for mutations in PLEC1. In 4 out of 16 patients dominant PLEC1 missense mutations (one amino acid change in the protein) were found, making PLEC1 a gene to consider when KRT5/KRT14 mutation analysis fails to reveal a mutation.

Saturo Shinkuma
OF DERMATOLOGY, HOKKAIDO UNIVERSITY GRADUATE
SCHOOL OF MEDICINE, SAPPORO, JAPAN.

Proteine therapie in DEB.

New treatments of EB can be divided in three alternatives:
- Gene therapy
- Cell-therapy
- Protein-therapy

This last named therapy stimulates mutant cells by an injection with protein to manipulate themselves in healthy and productive cells. This therapy is promising to get successful; the therapy attacks only those cells showing mutations.
This therapy is used also in the treatment of diabetes , but, as it seems now; the therapy can also be of great advantage in the treatment of DEB.

Protein plays an important part in the formation of the skin. In a lab in Sapporo a protein type Collagen VII is produced, and afterwards injected in mice with mutations in the domain of this protein. These experiments, in 2009, were successful.

But there are still some problems:
- The dosage of the exact amount of this protein: exactly how much protein has to be injected? Of course this has to be different for young children comparing to grown-ups.
- What will be the right frequency of the injections?

Saturo Skinkuma haalde veel motivatie voor zijn opleiding tot arts uit het feit dat hij zelf geboren is met EB. Hij adviseerde ook dat EB- patiënten ondanks hun aangetaste lichaam vooral van zichzelf moeten houden, en van het leven te genieten, ook al ondervinden zij veel problemen.

De jonge onderzoekers

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Functional correction of RDEB in vitro and in vivo

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Dystrophic Epidermolysis bullosa (DEB) is caused by inherited alterations in the collagen 7 gene that produces collagen 7 protein. Collagen 7 protein is the major component of structures called anchoring fibrils that hold the layers of the skin together. People with DEB have a reduced number of anchoring fibrils or completely lack these structures in their skin. Therefore minor mechanical trauma leads to the characteristic blistering of the skin of DEB patients.

The aim of my research project is to develop a gene therapy for people suffering from DEB in order to cure the skin disease. Using a special gene therapy technique, called SMaRT, we want to repair the altered collagen 7 gene in skin cells so that the cells recover their ability to produce a normal functional collagen 7 protein for skin adhesion. During the last years this method was established in our laboratory.
To repair a gene of interest, we first have to generate a repair molecule which brings in the normal copy of the gene portion to be replaced in the cells. Furthermore, the repair molecule has to contain a recognition site that ensures the identification of the target gene within a cell. We have constructed a repair molecule, which accounts for the precise binding to the collagen 7 gene and subsequent replacement of the altered part by a correct copy. We have already successfully introduced this repair molecule into skin cells of an RDEB affected person in the laboratory. As a consequence, the treated cells regained their ability to produce collagen 7 protein. Further, we have constructed an artificial skin using the repaired patient cells, which showed a normal coherence of the skin layers due to restoration of collagen 7 production.

As a next step we wanted to verify if the correction of the collagen 7 gene lasts over a longer period of time. For this purpose we grafted the cultivated skin which was made with the SMaRT corrected cells onto a so-called nude mouse.
This mouse completely lack an immune system, thus no graft rejection was expected. Five weeks later the human skin was analyzed according to its cohesiveness. No blistering was observed and collagen 7 protein was still present between the skin layers after 5 weeks, which indicates a long-term effect of the gene correction.

Based on these results, our long-term goal is to develop an ex vivo gene therapy for DEB patients. For that reason cells are removed from the patient, grown in the laboratory and then treated with the repair molecule. Cells corrected by the repair molecule are then cultivated to generate a thin skin layer, which is grafted back onto non-healing wounds of the patient's skin. In the natural environment of the skin, the transplanted cells should develop into normal skin cells reproducing the lacking protein. As a consequence, the wounds of the treated skin areas will be closed and further the new skin will show the properties of healthy skin.

M.M. (Magda) van der Kooi-Pol
DEPARTMENT OF MEDICAL MICROBIOLOGY, UMCG GRONINGEN

High genetic diversity of Staphylococcus aureus strains colonizing patients with epidermolysis bullosa

Patients with the genetic blistering disease epidermolysis bullosa (EB) frequently suffer from chronic wounds that become colonized by bacteria, such as the opportunistic pathogen Staphylococcus aureus.

To determine the S. aureus colonization and transmission rates in EB patients, swabs were collected from the anterior nares, throats and wounds of 52 EB patients. Swabs were also collected from the nares and throats of 13 healthcare workers who occasionally meet the sampled EB patients. All EB patients with chronic wounds and 75 % of the patients without chronic wounds were colonized with S. aureus. In contrast, 39 % of the sampled healthcare workers were colonized with S. aureus. Multiple-locus Variable Number of Tandem Repeats Analysis (MLVA) and spa typing revealed a high degree of genetic diversity of 184 collected S. aureus isolates. Self-transmission of S. aureus in individual EB patients with chronic wounds was shown to occur frequently, whereas transmission of S. aureus between EB patients appeared to be rare. There was no evidence for S. aureus transmission between EB patients and healthcare workers. Instead, the present results show that the S. aureus population structure in the sampled EB patients mirrors the general S. aureus population structure in the Netherlands, and that the colonization of EB patients is not biased towards particular S. aureus lineages.

The authors wish to thank the anonymous patients with EB from the Dutch Epidermolysis Bullosa Registry and healthcare workers from the Department of Dermatology at the UMCG for their participation in the present study.

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Revertant cell therapy

Revertant mosaicism
Revertant cell therapy is a novel therapeutic possibility in patients with epidermolysis bullosa (EB) and revertant mosaicism (also called "natural gene therapy") - a phenomenon of a second genetic event, i.e. a back mutation/reversion, which restores function of the disease-causing gene. Revertant mosaicism has been described for a number of diseases of blood, muscle, liver and skin. In the skin it was first demonstrated for the junctional type of EB. In a patient with mutations in the type XVII collagen (COL17A1) gene – healthy skin patches were present next to affected skin. It was also described later for other types of EB.

Concept of revertant cell therapy
Presence of revertant (=healthy) skin patches has a great value for EB patients. These are however limitations in area and control over the placement of the patches on one's body. Because of it, the idea of revertant cell therapy was born: to expand revertant patches by transplantation of the patient's own skin cells (=keratinocytes) in a form of engineered skin grafts.

An elegant approach would be to process a small skin biopsy (about 1cm2), isolate skin cells, and culture them in a laboratory setting. Stem cells of the skin, present in such a biopsy, would proliferate and expand, and skin grafts of an area much greater than the original biopsy (theoretically up to few m2) could be produced. Those revertant skin grafts could then be transplanted back to the patient and provide long-term improvement.

Clinical experience with revertant cell therapy
First experiments with revertant keratinocytes showed a possible problem with mutant (=unhealthy) cells overgrowing healthy cells during the graft production. The produced skin graft consisted of only 3% revertant cells; the other 97% being patient's mutant cells. Such a low percentage of revertant cells is not sufficient to maintain a healthy skin patch. To investigate this problem we have used an animal model and analysed every step of revertant cell therapy: cell proliferation, graft production, transplantation and long-term survival. Unfortunately, we have observed a gradual disappearance of type COL17A1 revertant cells during the whole process (from 40 to 3 %). The reason for this disadvantage of revertant cells during the whole process is yet to be established.

Currently we are assessing possibilities of revertant cell therapy for revertant patients with dystrophic EB. In the preliminary studies we see a higher rate of revertant cell survival than in previous experiments.

Future and chances for successful therapy
There are a few possible explanations why the revertant cell therapy has not yet been successful: the small number of revertant stem cells, growth disadvantage outside the patient's body or unwanted influence of culture techniques on revertant cells. We are currently looking into possible cell isolation and selection methods to enrich our grafts with revertant cells.

In the near future the need for enrichment might be replaced by revertant cell therapy based on induced pluripotent cells (iPS). Those cells, induced from patient's revertant cells, could be differentiated back into keratinocytes and used to produce multiple skin grafts, or into bone marrow cells for systemic therapy. With these new possibilities we are eager to continue our experiments.

M.G. Pasmooij
UMCG

Revertant mosaicism

What is revertant mosaicism?
Revertant mosaicism refers to the presence of healthy skin areas, where no blistering formation occurs, surrounded by affected skin. The blister formation in the affected skin areas is caused by mutations in the gene that is involved in the origin of epidermolysis bullosa. In the healthy skin areas without blistering an additional mutation has occurred in the disease-causing gene. This additional mutation corrects the first mutation, and subsequently leads to healthy skin. This process is called revertant mosaicism: a mosaic pattern in the skin as a result of a back mutation (= reversion). Sometimes this phenomenon is also referred to as "natural gene therapy", as the disease-causing mutation is "naturally" corrected.

Revertant mosaicism in all subtype of EB
The first patient with revertant mosaicism and EB was described in 1997. The 28-year-old patient with junctional EB had several revertant areas that did not blister on her hands, arms and ankle. In every distinct patch a different correcting mutation had occurred in the COL17A1 gene. At that time revertant mosaicism was thought to be rare. However, in 15 years, patients have been identified with revertant mosaicism in all four main subtypes of EB, i.e. EB simplex, junctional EB, dystrophic EB and Kindler syndrome. Since 2010, three patients have been described with dystrophic EB. Further, during the 41st annual ESDR meeting in Barcelona, the first patient with Kindler syndrome and revertant mosaicism was shown.

Revertant mosaicism occurs in all patients with generalised JEB-nH
Mutations in the type XVII collagen gene (COL17A1) result in non-Herlitz junctional EB (JEB-nH). The incidence of revertant mosaicism was identified in a cohort of 14 patients with JEB-nH caused by COL17A1 mutations. Ten patients had generalised blistering, whereas four patients had localised blistering. Revertant mosaicism was confirmed in the DNA in 6 out of 10 generalized JEB-nH patients. The clinically healthy areas manifested as patches of homogeneously pigmented skin. EB nevi can also be hyperpigmented, though EB nevi can be distinguished from the revertant skin patches, as EB nevi are often irregularly pigmented plaques that may give rise to small satellite nevi surrounding the primary nevus, whereas the revertant areas are homogenously pigmented macules without satellites. Further, photo-material and clinical history of the other four generalized JEB-nH patients demonstrated that each patient has revertant skin areas. In contrast, revertant mosaicism was not detected in the four localised JEB-nH patients.

Possibilities for therapy
Revertant mosaicism offers opportunities for cell therapies in which the patient's own naturally corrected cells are used as a source. The revertant skin cells might be used for obtaining a skin graft of naturally corrected cells to treated affected skin. Another exciting possibility is combining natural gene therapy with induced pluripotent stem (iPS) cell technology. Patient-specific skin cells from revertant mosaic patches could be used as source for patient-specific iPS cells. These iPS cells can then be differentiated back to skin cells and provide an essentially unlimited number of patient-specific cells for grafting. Alternatively, iPS cells may be differentiated into both hematopoietic and mesenchymal stem cells, which can home into blistered areas of the skin following bone marrow transplantation.

Closing of the saturday-morning session

And the winner is….

Those present listened to and looked with attention at the (Powerpoint) presentations of these 8 young scientists. But, as Marcel Jonkman already announced at the beginning of the morning-session, it was time to vote now for the best and most transparent presentation.

The voting turned out to be exciting; two of the scientists; Peter van den Akker, who held a witty lecture, and Satoru Shinkma, who made an impression with his personal statement, finished close together.
But finally Peter van den Akker was the winner!

Marcel Jonkman thanked the young people, and the public had reasons to thank Marcel Jonkman for this very useful initiative. It is very sensible to involve young scientists in an international meeting like this, including the condition that they had to bring their story in a clear way. And that is also a big challenge for the older generation….

Marcel Jonkmans and Peter van den Akker

Prof.Marcel Jonkman
DEPARTMENT OF DERMATOLOGY, UMCG

An update of the results: International EB-research.

In circles of scientists there is more and more attention for EB-research. Nowadays it is possible to get a reasonably quick diagnosis after birth, this in contrast to the situation 20 years ago. And also there is more known about the varieties concerning EB.

EB-simplex
New in EB-simplex is EB Suprabasal, caused by a shortage of plakoglobin. This variation is lethal, as is the case with the variation caused by desmoplakin; first discovered in the medical centre in Groningen; children died a few days after birth. Another newly found variation in EB-simplex is the lack of the molecule BGPA1 in the skin of hands and feet.

Junctional EB
As for this variation: in the case of EB-junctional also new research took place; for instance a late onset of junctional EB appearing on hands, feet and teeth. It is produced by recessive mutations in COL17A1.

Dystrophic EB
Future healing therapies in EB will make use of medicines-, protein-, gene- or cell therapy. There is yet no real cure for EB, but with each new therapy our knowledge is growing; and we are on our way to a real cure in the future.

A therapy with medicines:
At the university of Dublin there is an experiment going on with the method siRNA. This method is being used to suppress " wrong coded" gene expression; and removes " bad " molecules in the case of skin-diseases. This approach can be an option for the treatment of EB-Simplex.

Protein-therapy:
At the university of California an experiment with mice takes place. The mice are injected with human protein (Type VII Collagen)
This treatment was also spoken of by Marc de Souza, (See session on Thursday) and there is a possibility that with Lotus Tissue repair Inc. the research and use of this protein therapy will be sponsored.

Gen-therapy:
The European Community donates 5 million Euro to help the research of gene-therapy in the case of RDEB. These experiments will take place "at vivo."

Cell-therapy:
There is a possibility that a (mutant) type VII Collagen can be generated in the skin of EB-patients, in this way reducing blistering en stimulating wounds to heal in a quicker way.
The protein VII Collagen is produced by skin cells. In their turn these cells are being stimulated by fibroblasts; and these cells are easily to cultivate in labs.
Those fibroblasts can be injected in the cutis of RDEB-patients.
These injections with these not very immunological cells cause a kind of response of the outer skin-cells, putting them up to the production of more VII Collagen.
At the time being; the experiment is not finished yet; the fibroblasts disappear after two weeks.
But, the mutant type of VII Collagen is kept in the skin for several months; thus helping to heal their wounds.

Stem cell-therapy:
A pluripotent stem cell is a cell with a lot of regenerating qualities; being able to change into another cell type.
In Egypt, Chili and Minnesota (USA) experiments are developed with stem cells and EB; with reasonably satisfying results.
Bone marrow transplantation can be applied with EB-patients, but there is risk of infection. And also there are questions that have to be answered:

- Which cells have to be transported out of the marrow?
- What is the reason of the death of the children who underwent this treatment?

But there are small luminous points; in the past it was complicated to obtain suitable stem cells, but nowadays we can produce stem cells for all possible human organs; including the human skin. These cells are named: Induced pluripotent stem cells. (iPSCs)

Revertant mosaiscism:
A natural type of gentherapy (see; lecture of M.Pasmooij "morning session)
Revertant skin cells can function as body related source for this iPSC technology.

Conclusion: There are sure some luminous points to look forward for and hope for, but, besides the financial help of Lotus repair Inc, we will surely need at least 100 million dollars to develop all these therapies. They have to be of practical use and give accessible for all patients with EB.

Dr JW Frew
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Quality of Life Evaluation in Epidermolysis Bullosa

Quality of Life (QoL) is a complex concept which takes into account the physical, emotional, psychological and social aspects in which a disease may impact upon an individual. In individuals with Epidermolysis Bullosa (EB) it is especially important as it can help identify areas in which to focus treatment, help evaluate new treatments, compare the burden of disease across different types of EB and give accurate measurements of changes in QoL in EB across time.

Previous studies have used general dermatology QoL scales to measure QoL in EB which do not take into account specific problems encountered by individuals with EB. Such measurement tools can result in underestimating the QoL impact found in patients with EB.

Due to these problems with current QoL measurement tools, we developed an EB-specific Quality of Life scale (The QOLEB Questionnaire) which was statistically proven to be valid and reliable and allowed us to stratify quality of life into very mild, mild, moderate, severe and very severe QoL impairment, as well as allowing us to monitor QoL changes during the use of new treatments in clinical trials.

One such clinical trial was a double blind randomized placebo controlled trial of allergenic fibroblasts compared with injection of a placebo suspension solution. Our results showed dramatic wound healing with both fibroblast and placebo injections as well as increases in Collagen VII levels and overall improvements in QoL. 3 of the 5 patients in the trial showed QoL changes which were above the Minimal Clinically Important Difference (MCID) for the QOLEB Questionnaire which indicated the treatment made a significant difference to their overall Quality of Life.

Therefore, our QOLEB Questionnaire has a significant contribution in evaluating Quality of Life in EB and is also useful within clinical trials to show the effectiveness of new treatments in creating overall benefit in the QoL of patients with EB. In the future we hope to translate the QOLEB into a variety of different languages in order to help encourage the use of this questionnaire worldwide.

W.Y. Yuen

TERMINAL PHASE IN HERLITZ JEB: EXPERIENCES AND DATA

Junctional epidermolysis bullosa, type Herlitz (JEB-H) is a rare autosomal recessive disease characterized by extensive and devastating blistering of the skin and mucous membranes, leading to death in early childhood.

Our objective is to present the long-term follow-up of a cohort of JEB-H patients, to give guidelines for prognosis, treatment and care, and to identify the needs of parents who have lost their child to JEB-H. All JEB-H patients registered in the Dutch Epidermolysis Bullosa Registry between 1988 and 2011 were followed longitudinally by our EB team.

In total, we included 22 JEB-H patients over a 23-year period. Fourteen of their parents were willing to cooperate in semi-structured in-depth interviews. The average age at death of the patients was 5.8 months (range 0.5-32.6 months). The causes of death were, in order of frequency, failure to thrive, respiratory failure, pneumonia, dehydration, anemia, sepsis, and euthanasia. We cpuld not identify any predictors of livespan. Invasive treatments to extend life did not promote survival in our patients.

Parents indicated that they have the need (1) for a fast and correct referral to a specialized EB clinic, (2) to be informed as honestly as possible about the diagnosis and lethal prognosis, (3) to have a structured network of care givers in the palliative care, (4) to be involved in the care and the medical decisions involving their child, (5) to be informed about the end-of-life and to discuss euthanasia, (6) for guidance and to have remembrances of their child, and (7) for genetic counseling.

In conclusion, it is important to diagnose JEB-H as soon as possible after birth, so that the management can be shifted from life saving to comfort care. Furthermore, health care professionals should pay attention to the needs of the parents.

Saturday-afternoon 29 October

FINISHING MEETING

"The meaning of this session was also meant to show how valuable it is, to tell others of your ideas and solutions."

Brainstorming

Frank Houben, (DEBRA the Netherlands) one of the organizers of this congress, gave information about the ambitions of DEBRA International.

Because of the rarity of EB, it is one of these ambitions of DEBRA International to share ideas, solutions and knowledge among each other. To stimulate an exchange like that, all participants were asked to send their suggestions to Frank Houben.

A few of these ideas are reproduced here:

Opening a door
People with EB and reduced hand ability have difficulties to open a door with a key that have to be turned. But there are possibilities to use a remote control to get inside.
Sabine.daby@googlemail.com

Bandages
Countries with no real (financial) ways to supply EB-patients with the needed bandages and gauzes can perhaps be helped by using plastic foil: it can be a cheap and easy to obtain way of healing (not infected) wounds
Lauradesena@hotmail.com

Special gloves
There are special gloves to protect the delicate hands of EB-patients. The gloves, have a resemblance with the gloves cyclists use, but the seams are flattened and soft. The gloves are available in different sizes, colours and, on demand, in thinner and delicate material. The gloves protect the hands of children and grown-ups doing their daily activities. Hands in gloves give less opportunity to get embarrassing remarks than bandaged hands mostly do.
Sabine.daby@googlemail.com

Holidays in Marbella.
DEBRA Espagna can offer a chance to have a holiday in Spain for EB-Patients and their partners/family, in a special accommodated apartment in Marbella.
info@debra.es

Instead of Bandages
Also an idea coming from Spain; a suit of tender and soft material, used instead of bandages.It reduces the hours of being treated and stimulates the independency of the patients.
info@debra.es

Cannabis and pain-management.
VBor Verkroost (RDEB) informed the audience about his moderate use of medical Cannabis. It reduces pain, en stimulates appetite. It is a much more pleasant and a less benumbing experience than the use of opium based medication. For people who absolutely don't want to smoke (and of course in this case it concerns grown-ups) there is now a choice to use a vaporizer (an electronic kind of water pipe) In the Netherlands several experiments with this kind of inhaling Cannabis are happening now. Arno Hazekamp, pharmaceutical researcher at the University of Leiden and since 5 years studying the use of Cannabis as a medicine, states: "This vaporizer forms a bridge between this medicine and the patient",
florisleeuwenberg@gmail.com
Info@1-up.nl (Bor)
Have a look at: www.vaporizerblog. (English-spoken blog)

To prevent unnecessary rubbing
An idea of Frank Houben; he benefits a lot from this idea: he wears two pair of thin socks to prevent rubbing, and so causing blisters while walking.
Frank@frankhouben.nl

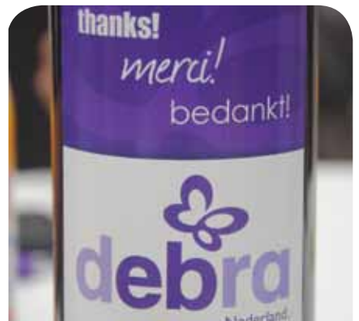
Stabilizing the opening of the mouth
Problems in oral treatments.
In the case of anesthetization, but also according to taken care of by the dentist, the opening of the mouth, because of EB getting too small, can become quite a problem. With the assistance of a dental technician a special device was developed, suitable to be used quite independently by children.
sabine.daby@googlemail.com

The application of Apligraf after surgery
Apligraf on wounds of persons affected by EBD has repeatedly proven to support rapid healing of the treated areas. This observation applies to acute wounds following surgery, as well as chronic wounds with poor healing tendency. On addition to this, the application of Apligraf on the wounds immediately following hand surgery leads to significant reduction of pain in the treated area. (Apligraf® Organogenesis, MA) is a bi-layered bio-engineered skin substitute and was the first engineered skin US Food and Drug Administration (FDA)-approved to promote the healing of ulcers that have failed standard wound care. Apligraf provides both cells and matrix for the nonhealing wound. Its exact mechanism of action is not known, but it is known to produce cytokines and growth factors similar to healthy human skin.
kristin.kernland@insel.ch

The meaning of this session was also meant to show how valuable it is, to tell others of your ideas and solutions. DEBRA International is THE international platform to give us the opportunity to share knowledge and ideas. Frank Houben hopes that DEBRA International can create a way of showing all the ideas in, f. e. a website. In this way it is easily accessible for all the DEBRA' s and can be a source of inspiration for everyone.

The session is also an appeal to all the countries here assembled to share all the knowledge and make it available for all of us.

"We share a mission, so let's share also all our knowledge to fulfil this mission.".



Saturday-afternoon 29 October

FINISHING MEETING

Mail suggestions, remarks and ideas to:
Jimmy Fearan: Jimmy@debraireland.org or
Graham Marsden: graham.marsden@95jermynstreet.com

The new Executive Committee of DEBRA International.

Rainer Riedl	(Austria)	President
Francis Palisson	(Chile)	Deputy President
Michael Fitzpatrick	(Australia)	Treasurer
Gena Brumitt	(Canada)	Deputy Treasurer
Jimmy Fearon	(Ireland)	Secretary
Vlasta Zmazek	(Croatia)	Deputy Secretary
Rick Gallagher	(USA)	
Robin Hood	(UK)	
Evanina Makow	(Spain)	
Polona Zakosek	(Slovenia)	

Internal auditors:
Shoaib Gopalani (Canada)
Graham Marsden (UK)

Graham Marsden (DEBRA UK) gave some information about the ideas within the committee.
There are now 40 countries member of DEBRA International. Thanks to this international institution a lot can be attained. Also the way of communicate with each other is a lot easier thanks to the possibilities of email, tweets, video-conferencing etc. That can be of great help to give assistance to new national DEBRA groups.

There is more and more attention from medical professionals and the pharmaceutical industry for new therapies like DNA and protein-therapy.

But fundraising won't be easy, because of the almost world-wide economical crisis.

Important is that all members of DEBRA International participate in the future of DEBRA International. They too have to give ideas, suggestions and answer questions like:
What do they expect of DEBRA International?

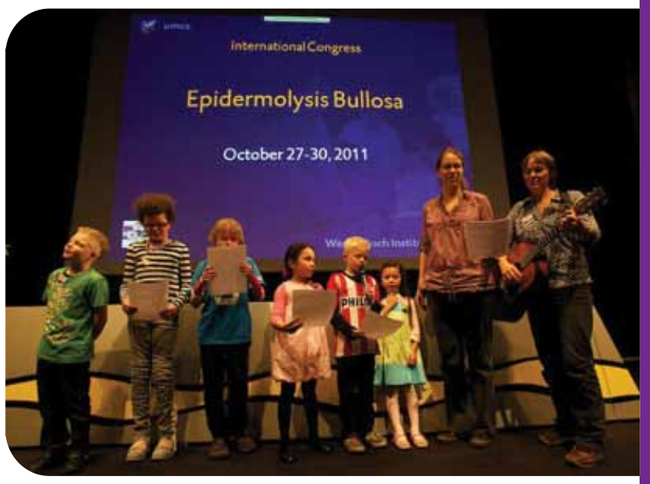
NOBODY IS PERFECT!

Several Dutch Congress participants took their children with them: and that was no problem; DEBRA the Netherlands organized a special nursery for them.

That is the reason that we did not see much of them while this congress took place.

So it was a nice surprise when those children wanted to say goodbye to us by singing a song together:

Nobody is perfect!
We all got sometimes something to worry about,
there can be something wrong with us,
with our health, our body, looks or mind,
but that does not really matter,
Because we all are little brothers and sisters,
and we really do love each other



Petra Bleeker
EXECUTIVE COMMITTEE DEBRA THE NETHERLANDS

Dinner in the St.Martin's church

To finish off a wonderful day of this international congress we were expected to have a diner in the St.Martin's church at the marketplace in Groningen. The church was constructed in the year 1245, but the building of this church was preceded by a long history; which you could immediately experience when you entered this church.
If you, together with this emotion, take also into account be welcomed by organ sounds and the beautiful way in which the tables were set, than you can imagine what I meant.
We first were offered a drink to give the opportunity to talk to each other about the congress.
In general a feeling of togetherness and solidarity was dominant; language was no barrier during this informal part of the evening.

After being seated at the tables and listening to a word of welcome by Ank ten Siethoff, Chairman of DBRA the Netherlands; we could listen to Tom Haze. Tom himself has EB, unluckily he did not get through the qualifying rounds in the contest; "Holland got talent", but we are convinced that Tom is a really talented singer and guitar player!

Diner was going on when the organ-player, Eeuwe Zijlstra, informed us by men as of an inspiring little lecture about his organ. The organ of this St Martin's church is the Rolls Royce amongst all the church-organs, it is a very large, really majestic and its sound is unapproachable!
And especially our EB-kids had their lucky chance this evening: accompanied by the organ-player they could have a thorough look at this organ, en were allowed to use some organ-registers; a tremendous experience!
And this was not the only surprise for the audience; because also national folk-songs were to be heard and recognized …

For us a perfect ending of a successful congress-day!

Yvonne Westheide-Sellies
CONSULTANT DEBRA NETHERLANDS

Depression and EB

Ilda Mollema
Social worker University Hospital UMCG Groningen

The forum was introduced with a PowerPoint presentation and a YouTube movie about depression:
(Kijk op: http://www.youtube.com/watch?v=H02MTQovphI&feature=results_main&playnext=1&list=PL5ABC990D17E22382)

Everyone experiences "blue moods": temporary sadness, irritability, loss of energy, mild feelings of "what's the point?" But depression may be defined as a more serious and prolonged case of the blues with the following features (symptoms):
- Cognitive disturbance: negative evaluations of self (I'm a failure. I am no good. Why should anyone like me? I'm so mediocre. There's no hope for things working out. I feel bad, and nothing will change.
- Mood disturbance: deep sadness, hopelessness, aloneness, rejection (no one cares), indifference, suicidal desires.
- Behavioural disturbance: decrease in activity, limited self-initiated activities, little activity for the sake of fun, in attendance to responsibility: reduced sociability appetite loss, reduced sexual interest, sleep problems. Sometimes depression will take the form of no decrease in activity or responsibilities, but there is indifference in all the person does; no sense of vitality about life.
- Physical problems: chest pains, joint stiffness, headaches, dizziness.

Why do people experience depression?
The person has experienced some form of loss, insult, injury, or disillusionment and responds to it with self-pity rather than acceptance.
Depression is the result responding to a "down period". People feel "the blues" from time to time either because of "own mistakes" in their life (guilt) or because of unpleasant events (sickness, loss of job, death of a loved one, etc.). Depression is the deep, pervasive sadness, which results from responding to these events by letting responsibilities and interests slide. The pressure from being behind, the guilt from avoiding responsibility and the loss of good feelings from doing fewer "fun things" results in less motivation to live responsibly. The downward spiral produces depression.

People become depressed when they
- fail to reach a goal, which they believe they must reach to establish their worth.
- They perceive that continuing efforts to reach that goal may never be successful.
- They therefore quit trying to work toward a goal.

With EB it may be:
- I am never going to be well. Therefore my life is worth very little.
- It doesn't matter how hard I work on myself, I will not get out of this.
- I quit trying. Why bother with an education, being friendly, doing stuff, etc.

When is it time to intervene?
• When there are mood swings – up for weeks or months, then down for weeks or months; then into the cycle again.
• Depression is severe to the point where you can't get a relationship going – sits blankly and stares, responds to questions with no emotion in monosyllables, seem unable to make any decision.

You should consider that depression is serious enough to warrant medical attention if at least five of the following eight signs are present:

1. Poor appetite/weight loss
OR increased appetite/weight gain
2. Sleep difficulty.
3. Major loss of energy, tiredness.
4. Marked slowness of movement
OR inappropriate agtation (jumpy, fidgety).
5. Loss of interest in usual activities.
6. Feelings of self-reproach, excessive inappropriate guilt.
7. Complaints or evidence of diminished ability to think or concentrate.
8. Recurrent suicidal thoughts or wishes.

Steps in counselling with a depressed person:
1. Briefly empathize with his/her feelings.
2. Get a history of the depression
3. Zero in on the emotional response to the loss or experience that triggered the depression.
4. Identify wrong goal based on wrong tape (what they are telling themselves).
5. Insist on appropriate behaviour

José Duipmans
EB-NURSING SPECIALIST, UMCG

Nursing Forum

Introduction:
Jose welcomed the participants; after making acquaintance with each other, everyone could exchange email addresses. In this way it is easier to get in contact with each other, and share experiences (possible through the international EB-nurses forum too). A PowerPoint presentation about wound healing was shown as introduction to the more practical part of this meeting.

EB and adolescents:
Jennie Hon (EB-nurse specialist UK) organizes once a year an outing for EB-youngsters between 16 and 19 years old. The reactions during this week are very positive; the youngsters love to be together and share their stories. It became clear that there is an information-gap between the knowledge about EB-babies and children and the information about EB youngsters growing up. So the idea was developed to compose a guideline for this target-group with not only practical advices but also experiences concerning sexuality and sexual relations. By means of a questionnaire youngsters can (anonymous) tell what's bothering them.

Wound care:
All participants got a copy of Jackie Denyers article: Wound-management for children with EB "and a copy of: "Principles of wound care in patients with EB", (Irene Corrales, Alan Arbuckle e. o). And then it was time to get into the practical part of this forum. There was a possibility to use 2 Lotus "patients" for this practice with artificial wounds at difficult to reach parts of their body. The forum-participants were divided in little groups; groups who bandaged the wound and others who watched and asked questions.

Looking back at this practical part everyone had the opinion that it was a highly teachable and significant training and experience. It was also very evident that there were a lot of differences between the countries; in some countries there is almost no experience in wound caring, or even worse, there is a total lack of the material that is needed in this case. In Australia a video is produced with examples of wound dressing: Louise Stevens, BrightkSky Australia (National Epidermolysis Bullosa Dressing Scheme) http://www.ebdressings.com.au/Educational_Support_and_Resources/Video.aspx

Jose Duipmans is planning to collect all the experiences of this forum and eventually publish them in a guideline for wound caring. This idea can be a topic in the Nurse Forum of DEBRA International.

The future of the Nurses Forum:
On behalf of Heidi Silseth (Norway) Jose asked the audience if there still is a necessity to continue this Forum. There is not much happening at this moment within this forum. Yet all the participants had the opinion that the underlying idea for this forum is a good one and can be very useful. But the way the forum is organized and is accessible could be better. It would also be convenient if the different fora about nursing and EB could take place on different dates; it would create the possibility to visit all these different meetings.

The impact of EB on nurses:
Everybody agreed; taking care of patients with EB is very emotional, stressful, nerve breaking and costs a lot of mental energy. Getting a burn-out is one of the risks in this profession. The empathy your direct management can show to you is very important, and psychological support can be necessary too: how to survive under all this pressure. Besides that it is, being a professional yourself, sometimes complicated to talk about your stress or asking for help. But you should; you can't cope with it at your own. Jose Duipmans will suggest to John Dart if the impact on nurses of taking care of EB-patients can be put down for consideration at t he next International Debra congress in 2012

Inquiries: j.c.duipmans@derm.umcg.nl.
More information about 'Woundmanagement for children with EB': jackie.denyer@debra.org.uk

Forum: NUTRITION AND EB

Rosie Jones SPECIALIST DIETITIAN IN EB, BIRMINGHAM CHILDREN'S HOSPITAL, UK

Nutritional challenges in a baby with Herlitz Junctional EB

Rosie Jones describes for the audience a case of problems with nutrition in a social deprived family, where a little girl was born with JEB. The physical condition of the child was severe; anemia, inflammations, constipation and a lot of blisters in the oral cavity.

The dental care was neglected, brushing teeth was no option. On top of this the girl was a little deaf and staying behind in her mental and physical development. In the overcrowded household one could not expect that the mother of the family could spend a lot of energy and time to improve the child's condition.

Still dieticians and other specialists and assistants tried to improve the quality of the girls' life; energy-drinks and special nutrition-supplements were prescribed, accompanied with practical advices and encouraging support for the whole family.

The girl did become speech training, and was offered help with her eat- and swallow problems. But despite this approach the situation stayed difficult; the family did not cooperate very well.

But the decision to let her have a gastrostomy turned out to be a disaster; the wound, caused by the introduction of the tube into the stomach, would not heal, and the tube had to be removed. The parents refused a second gastrostomy.

But, although the situation seemed to be hopeless the family assistants suggested a special adapted form of gastrostomy; another failure was of course out of the question. And this time, with much more precautions like special anti bacterial gauzes to take care of the little stomach wound, this approach succeeded. De then 4-year old girl could go home, where she also got some extra nutrition.

She was visibly getting better, had a lot more energy, started to move and walk again, and was being educated at a special school for children with hearing disabilities.

Meanwhile another baby with JEB was born in this family; but luckily the parents agreed with gastrostomy when the baby was 13 months old.

rosie.jones@loch.nhs.uk
dawn.jarvis@debra.org.uk

Susanne Marie Krämer, LECTURER IN PAEDIATRIC DENTISTRY, UNIVERSITY OF CHILE, SANTIAGO. DENTIST FOR DEBRA CHILE

Nutrition and Dental Health in EB, a team approach

Preventing dental caries is a common goal for patients, parents, dentist and dieticians. The presentation started by discussing the multiple risk factors involved in the development of caries and analyzing the factors that can and cannot be addressed in people with EB. Some caries risk assessment tools were presented and hints on how to brush the teeth in a gentle but effective manner were presented. Other preventative strategies such as topical fluoride, fissure sealants and chlorhexidine were also discussed.

Carcinogenic potential of food was discussed and the AAPD (American Association of Paediatric Dentistry) recommendations were presented:
1. Breast-feeding of infants to ensure the best possible health and developmental and psychosocial outcomes, with cessation of ad libitum breast-feeding as the first primary tooth begins to erupt and other dietary carbohydrates are introduced;
2. Educating about the association between frequent consumption of carbohydrates and caries;
3. Educating the public about other health risks associated with excess consumption of simple carbohydrates, fat, saturated fat, and sodium.

It was concluded that a dietary caries-prevention programme should be instigated at an early age and that dentists and nutritionists have to collaborate on an appropriate program.

More information: susiks@yahoo.com

Groepsbijeenkomst:
VOEDING & DIEET BIJ EB-PATIENTEN

Lynne Hubbard DIËTISTE ST.THOMAS HOSPITAL, LONDEN.

Gastrostomy bij volwassen patiënten met EB.

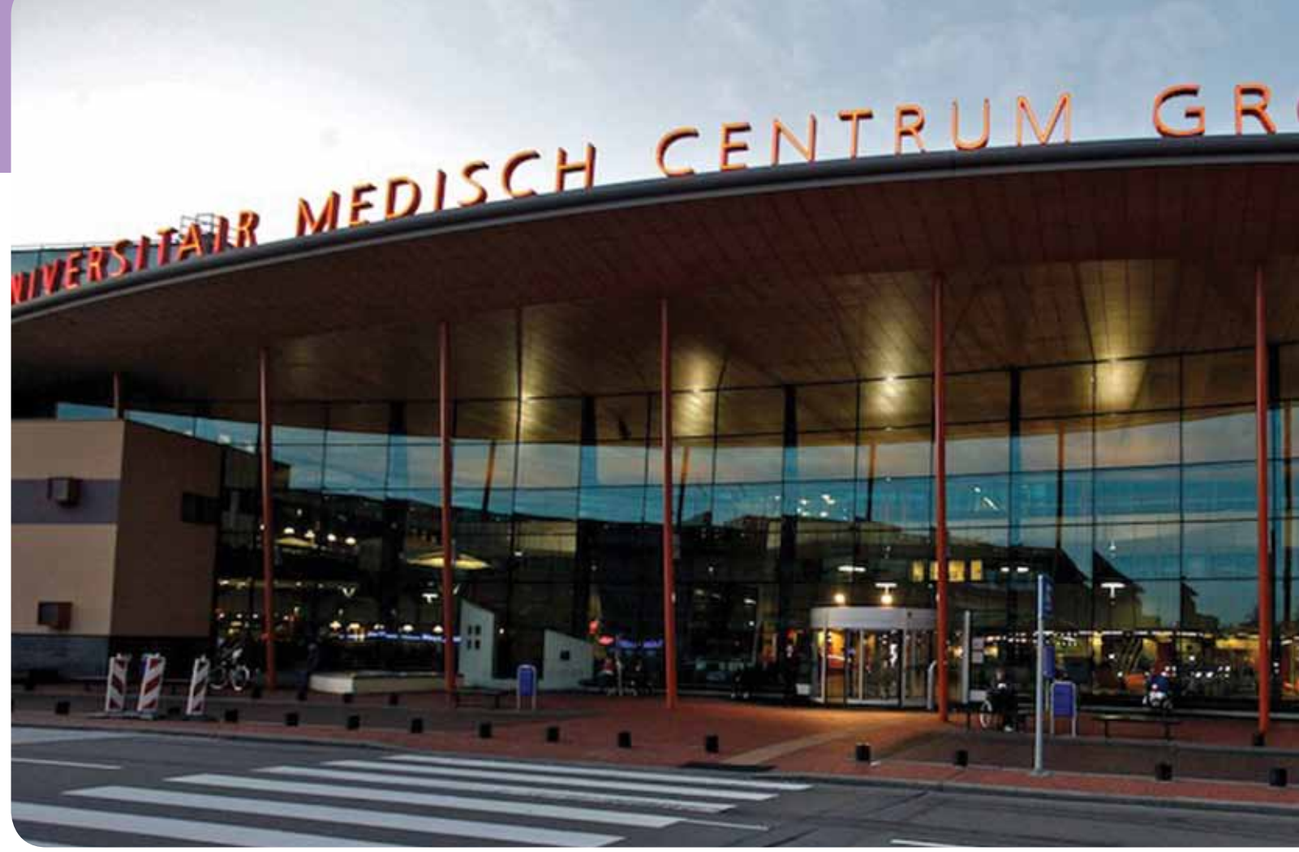
How are grown-up patients coping with gastrostomy on the long term?

A lot of them have problems swallowing food, and medication that has to be taken orally. And in that case gastrostomy stays a steady and safe solution. In this way chronic patients get enough vitamins, proteins, fibres and minerals.
Yet it was important to make an analysis of the effects of this approach on the long run.

More inquiries:
Lynne.Hubbard@gstt.nhs.uk

Therefore data of 12 patients were collected, based on growth and weight; these patients had an experience with gastrostomy from 10-19 years. The analysis showed that with 58% of the patients gastrostomy had a positive influence according to growth, and with 50% it had a positive effect on their weight.

Of course gastrostomy has his problems; for instance; a leaking stoma, or painful experiences when the tube is taken in.
To prevent this problems
- One has tried to use better endoscopic methods related to the placing of the stoma
- to attend to the whole process as good as possible by giving advice to the patients, who are based on practical experience.

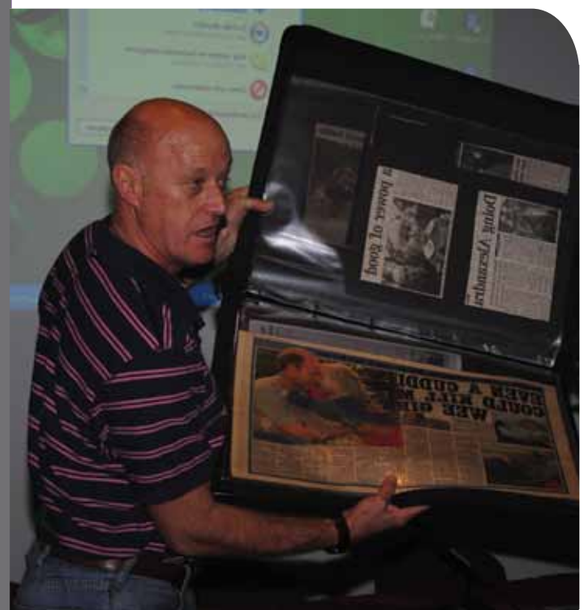


Forum Fundraising

A short summary of the ideas; not only to inform, but also to inspire other National DEBRA's to invent their own initiatives in the case of fundraising.

DEBRA Chile
Skiing
They had a campaign with banners, posters and e-mails: skiing for EB; three ski-games; search for butterflies in the snow! The output was about 6000 euro's, but the campaign stimulated people to become a DEBRA-member.
Another event organized by DEBRA Chile was the Family Race; EB-Families getting acquainted with snow. Skiing in Chile is mostly reserved to the upper ten; so financially spoken it's an interesting target group.

Robin Hood Fundraisers forum



It was the first time DEBRA Chile organized an event like this. All these campaigns distributed tot more awareness with

DEBRA Canada
An international EB Awareness week
The public relations for this event were taken care of by Photo-posters of EB children and grown-ups in English and French. There was a possibility to download a PDF file of these posters. 56 countries were involved in this event.
This initiative took place for the first time in 2005 with a letter to the American Congress; which in 2006 resulted in organizing a national EB Awareness week in the USA. Now this initiative has grown into a worldwide campaign; the international EB Awareness Week. Important for the P.R were the press releases, social media (1200 Facebook connections) and Metro news. The web shop offered specially printed cups, shirts etc.

The devices, used for this event were:
"Walk a mile in my shoes for a while."
"The only limitation is your creation."

DERBRA Spain
Special charity shops
With the support of DEBRA UK DEBRA Spain started with special charity shops; there are 7 of these shops now in Spain. The shops are taken care of by paid managers, but the other workers are all volunteers.
The shops depend on donations and the output of flea-markets and fancy-fairs. The advantage of these shops, comparing with other shops, are the permanent low prizes. The costs of the average shop with the best output (100.000 euro's) is about 35.000 euro's a year. That leaves 65.000 euro's as a profit for DEBRA Spain

DEBRA Croatia
A public sale
DEBRA Croatia organized a public sale of the costumes and clothing's, belonging to national well known celebrities. They sold these clothing via Internet. It resulted in a lot more awareness of EB and free publicity.

DEBRA Czech Republic
This DEBRA member used also the national celebrities; 12 of those well known ladies and gentlemen showed how butterfly-kids experience their daily surroundings . With those Photographs' expositions were set up, and calendars and diaries produced with support of several sponsors.
The costs of the design of these products were 14.000 euro's, but the output will be 30.000 euro's.
It can be a good idea to produce such a calendar on an international scale.

DEBRA Ireland
A lottery
DEBRA Ireland hired a professional telemarketing company to sell lottery-tickets by phone. The costs were higher than the output, but agreed was that no costs would have to be paid, if there was an output anyway. So finally DEBRA Ireland could profit; 200.000 euro's.

DEBRA UK
Emotional blackmail…
Robin Hood of DEBRA UK showed the audience a clear and moving little movie of his daughter, who died of EB. He's always ready to show this little DVD documentary in a little mobile DVD-player; "Can I have only 4 minutes of your time?" That is his opening speech for a talk about fundraising. He sticks to emotional blackmail, as he puts it with a smile, "keep on going and aim for the stars, never miss an opportunity".

DEBRA Australia
The Cannonball Charity Ride 2011
DEBRA Australia organized The Cannonball Charity Ride in 2011; a rally who takes you through the deserts of Australia. (Also organized in the years before)
25 drivers participate in this rally; their device was; roosting for a cause!
The output was 34.000 Aus. Dollars.

DEBRA New Zealand
Charity year
The New Zealand Ballet declared the year 2011 as EB Charity target.

USA/Internationaal
The Harley Davidson company is going to use DEBRA Badges on their clothing.

Other remarks: is there a possibility to have an international forum for fundraising related to the website of DEBRA International?

Brainstorming session, directed by Graham Marsden (UK)

THE END of the congress

The congress came to creative break-up; collecting the ideas and suggestions of the congress-participants before turning home. Directed by Graham Marsden, a lot of advices were gathered for the Future of DEBRA International. Remarks and suggestions:

Guidelines:
Guidelines, like the now published best practice guide for dental care in EB and occupational therapy are a very good initiative, asking for more; guidelines for other aspects related to the care for EB.Still, it is complicated to ask physicians, nurses and other EB-experts for their experience and co-operation, if in the countries where they do their work, very few EB-patients are registered.

International contacts, thanks to new communication-technology, is growing and getting more intensified. It also strengthens the need to have more best clinical practice guidelines, but those initiatives costs a lot of time, energy and money. (The guideline dental care: 15.000 euro)
It can be a possibility for DEBRA International to undertake fundraising for these guidelines. DEBRA UK has a fund to finance guidelines like this. Belgium and Australia were also prepared to give financial support.

A best clinical practice guideline related to nutrition and EB and EB and cancer is getting prepared. A nice side-effect while composing guidelines is the integration of the knowledge of all experts and the findings coming out of literature-research.

Ideas for other guidelines:
- Eye-care.
- Psychological- and social aspects in the life of EB-patients
- Quality of life and EB.

Other subjects:
Another very important issue is the co-operation of the DEBRA-members here present in the data-base, developed by Lotus Tissue Repair INC. Of course a reliable data-base is also convenient according to commercial aims and targets, but, being able to show reliable data, the chance to get more funds, is growing. Research to reach a definite therapy for EB will ask for a lot of money, and therefore reacting positive at the offer of this company and deciding to participate will sure be a situation in which both parties can gain. If the data-base is completed, there are more possibilities to attract scientists and researchers.

Results and findings from research as presented in the case of revertant mosaicism should be made accessible for everyone.

Smaller countries like f.e. Columbia are getting support of DEBRA International; this institution paid for hotel and tickets of the Colombian members.Maybe DEBRA International can help some other small countries too in supporting the organization of workshops in these countries, and inviting experienced DEBRA-members to manage these meetings.

A country like Romania also needs a lot of help according to the start and developing of a DEBRA- Romania. DEBRA International has a fund to support initiatives like this if accompanied with a good proposal. A team of EB-experts could travel to Romania to give suggestions and advices. This seems also to be a good idea for small countries like Colombia.

For the Japanese, Spanish, Italian, Portuguese and French delegates handling all the presentations in the English language is not an easy job. Besides Chinese and English, Spanish is worldwide the third on the list of the most spoken languages. Hopefully there is a possibility next congress to hire some interpreters to help all the Spanish speaking members.

The lack of money is the reason for DEBRA Japan not being able to translate the best clinical practice guidelines in the Japanese language; maybe DEBRA International can give some financial support to solve this problem. In Japan a special DEBRA- Asia EB congress will be organized. The question is of DEBRA International can participate here too.

Sometimes the lack of money is not always the main problem. There is also a great need for practical help and advice from people who really can offer help and support. It should be possible that the larger DEBRA's share their experience with the small or just starting DEBRA's.

Would it be useful to ask members to function as a kind of regional managers in continents as Africa and Asia. Both continents are very under-represented in international EB-meetings.

Can it be a good suggestion to ask grownup EB-Patients to lobby for more co-operation and sharing information?

The situation for people with EB in South-America is, comparing to a lot of western countries, quite different. This continent encloses a lot of pour countries, and is frequently dependent of the surgical expertise of other c countries.

would be convenient if all the here present DEBRA's would approach their own sponsors and ask them to give some financial support for these kind of international meetings.

The next congress should invite EB –youngsters and grown-ups like Humphrey Hanly, Mattija Zmazek and Bor Verkroost to tell more about the psychological and social aspects of EB; What about guilt? Do EB-youngsters and their parents feel guilty to each other? (They do, as is known) But why?

The online EB-Community does not function at all. So the best way to communicate with each other remains; e-mail, tweets, hyves etc

Composing this Congress-special, it became known, that the next international DEBRA meeting will take place in Toronto; September 13-16.
(Canada)

And after that…we hope that you all had a nice journey home!

Rainer Riedl

PERSONAL WORD:

Rainer Riedl

newly chosen president
of DEBRA International.

Dear colleagues,

It is an honour to have been elected President of DEBRA International and I come into the role full of enthusiasm and plans. As many of you know, my interest in EB is personal and it is now 18 years since my wonderful daughter Lena received her EB diagnosis. Since the early days of her life I have worked very hard to improve the lives of all those living with EB and have big plans to continue doing so.

There are several areas that I would like to give particular focus to during my presidency and they include the following:

• I am keen to convince all DEBRA groups to use a common branding and logo to ensure that we are seen as a strong and professional, worldwide network.
• I have a desire to see the DEBRA International website fulfil its potential and will be encouraging all members to use it as a portal and to provide us with information for it. I would also like to see more groups engaged in social networking channels of communication and, in particular, participating in the DEBRA International Face book page.
• A particular passion of mine is research into EB and I would like to see more of the national groups engaged, one way or another, in research. DEBRA Austria has a strong track record of supporting research and we invest €500K per annum in projects that have been deemed excellent by the DEBRA International Medical and Scientific Advisory Panel (MSAP). In a recent exciting development, we have provided €3M for large, collaborative projects that will focus on developing a treatment for EB. Successful projects will be decided on in the coming spring and we look forward to sharing that news with you.

I welcome any suggestions you might have to develop DEBRA International as an organisation and very much look forward to working with you all.

Best wishes for 2012,

Rainer Riedl

DEBRA Austria
www.debra-international.org

NEXT INTERNATIONAL DEBRA MEETING

13 - 16 SEPTEMBER 2012

TORONTO, CANADA

Sponsoring

MÖLNLYCKE HEALTH CARE

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