

# The 2nd Nordic symposium on Epidermolysis Bullosa 24–26 April, 2005, Stockholm, Sweden (list of abstracts)

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FC9: Surgical management of the hand in Dystrophic EB. *Vokurkova J & Buckova H.*  
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FC12: Alterations of oral mucosa in hereditary EB (poster). *Sadler E et al*  
FC13: Treatment of sweat-induced blistering in mechanobullous diseases. *Swartling C, Vahlquist A*  
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FC16: Post-operative hand therapy in RDEB. *Formsma, SA*  
FC17: Body mass index is a reliable tool to optimize length growth. *Herpertz, I*  
FC18: In severe EB, what is desirable weight status. *Hayenes L, Denyer J*



## Organizing committee

Malin Netz, project leader, DEBRA Sweden/Nordic  
Kristina Gustafsson-Bonnier, socialworker/organizer  
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## Scientific committee

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Gabor Koranyi, ophthalmic surgeon  
Carl-Fredrik Wahlgren, MD, Assoc.Prof.  
Bitte Ahlborg, DDS, Senior Consultant  
Gerd Wohlin, occupational therapist  
Christina Eklund, dietitian

## PLENARY LECTURES

### PL:1

#### **A SHORT HISTORY OF EPIDERMOLYSIS BULLOSA IN SWEDEN.**

*Anders Vahlquist*

Department of Medical Sciences, Genodermatosis Centre, Akademiska sjukhuset, SE-751 85 Uppsala, Sweden

The Swedish pediatrician Gillis Herlitz (1902–1982) will be remembered for describing 8 cases of lethal EB already in 1935 (*Acta Paediatr* 17, 315–371). Later on, Dr. Tobias Gedde-Dahl, Norway, has helped many Swedish physicians to correctly classify new cases of EB. According to his data, collected in 1965–95 and reported at the Nordic EB conference in Oslo in 2002 (*Acta Derm Ven* 82, 238), the cumulative number of Swedish EB families is: EBS 14, JEB 46 and DEB 25 (9 dominant/16 recessive).

In a study from 2001, initiated by the late Dr. Sven Wittboldt of DEBRA-Sweden, we asked 200 Pediatric and Dermatology units all over the country about the number of EB-patients recorded in their files. A total of 80 patients were identified: EBS 39, JEB 5 (subtype not specified) and DEB 24 (only 4 recessive). However, the true prevalence of EB in Sweden is probably in the order of 150–200 cases (1:50 000), because EBS in particular is likely to be underreported. These figures do not include patients with bullous ichthyosis or pachonychia congenita who frequently have foot blisters indistinguishable from EBS (about 20 patients identified in Sweden; *Acta Derm Ven, Suppl.* 213, 34–37, 2003).

All the abovementioned genodermatoses are at the focus of a new EU-project (GENESKIN) which officially starts in June 2005 (Nordic participant: Uppsala University). A network of scientists and health staff with a special interest in EB will hopefully help to improve the diagnostic accuracy, genetic counselling and care of EB patients.

### PL:2

#### **CLASSIFICATION AND DIAGNOSIS OF EB**

*Robin A J Eady*

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Various classifications have been proposed for EB, ever since the disorder was first described over a century ago. Recently, as many as 26 subtypes of EB have been suggested. With the explosion in our knowledge of the causative genes underlying all the major and most of the more minor forms of EB, it has become evident that the division of EB into so many subtypes is not always supported by elucidation of the genotype (mutant gene or type of mutation underlying the disorder). There is therefore a strong case for reverting to a simpler classification of EB, yet retaining the 3 main types of EB, namely EB simplex, junctional EB and dystrophic EB (EB dystrophica). In other words, the classification will still depend on the depth in the skin where the primary split or blister occurs. In EB

simplex, this is at the level of the epidermis; in junctional EB it is within the dermo-epidermal junction (DEJ) and ultrastructurally at the level of the lamina lucida; and in dystrophic EB it is also at the DEJ, but slightly lower and beneath the lamina densa. With either simple or more complicated classifications, the placing of certain subtypes of EB may still be a challenge. For example, the form of EB associated with pyloric atresia is conventionally grouped under junctional EB, yet, in a high proportion of patients, the primary level of skin cleavage is found to be intraepidermal. Do these cases therefore have EB simplex rather than junctional EB? We also need to consider whether we should be more inclusive and incorporate certain genetic skin fragility disorders not normally recognized as EB, such as Kindler syndrome, Shabbir syndrome and skin fragility with ectodermal dysplasia (plakophilin deficiency), within the EB 'family'. Inclusion would benefit the patients, especially infants, from the increasing levels of specialist care that are now available for those with EB. Since the main diagnostic test for EB still relies on the provision and microscopic analysis of an appropriate skin biopsy, these methods will be briefly discussed.

### PL:3

#### **PATHOGENESIS AND PROGNOSIS OF EB**

*Leena Bruckner-Tuderman*

Department of Dermatology, University of Freiburg, Freiburg, Germany

The term epidermolysis bullosa (EB) refers to a clinically, genetically and biologically heterogeneous group of inherited disorders characterized by blistering of the skin and certain other epithelia. The blisters are a manifestation of a separation between the epidermis and the dermis along the basement membrane and usually result from minor trauma. This characteristic is common to the whole group, although the severity of expression ranges from mild blistering to extensive bulla formation, erosions, scarring, mutilation and lethal outcome. The group contains many distinct subtypes, and defects in 10 different genes are known to lead to the clinical abnormalities. Enormous progress has been made in the last years in understanding the genetic basis of EB and, as a consequence, some old concepts about this disease have been modified. The new, simplified classification is based on categorization according to the precise level of blister formation, the molecular background and clinical presentation.

The dermo-epidermal junction zone, the site of attachment of the epidermis to the dermis, is pathologically altered in EB. Three major categories are defined: EB simplex, in which the separation occurs within basal keratinocytes, is caused by mutations in the genes for keratin 5/14, or plectin. In junctional EB, the cleavage occurs along the basement membrane, and mutations in at least 6 genes are involved, including LAMA3, LAMB3 and LAMC2 encoding laminin 5, COL17A1 for collagen XVII, and ITGA6 and ITGB4 for integrin  $\alpha 6\beta 4$ . Dystrophic EB, with separation below the basement membrane, is caused by mutations in the collagen VII gene, COL7A1. A large number of mutations are associated with a surprisingly broad spectrum of EB phenotypes.

Not only definition of mutations but also cell biological, protein chemical and suprastructural studies of the mutated molecules have shed light on the molecular pathomechanisms in EB and provided a basis for both prognostication and development of novel therapeutic strategies. International collaborative efforts have resulted in successful gene transfer into EB keratinocytes and in stable expression of correctly folded proteins by these cells in vitro. Although the clinical application of such therapies for EB may still be years away, the rapid development of new technologies holds promise for individually designed and biologically valid curative treatments.

#### PL:4

##### **MULTIPROFESSIONAL CARE AND CASE MANAGEMENT**

*J.C. Duipmans, M.S., and M.F. Jonkman, M.D.*  
Center for Blistering Diseases, University Medical Center Groningen, the Netherlands

Epidermolysis bullosa is a complex genetic disorder affecting more than only skin. The complications associated with EB are plentiful and include pain, itch, dental and eye problems, airway obstruction, dysphagia, oesophageal strictures, gastro-oesophageal reflux, constipation, anaemia, scarring and contractures, and limitations in mobility. Furthermore, the complex problems in EB conditions more or less serious developmental and psychosocial problems (affective, social, and occupational) and disturbances in the integration of the personality, both for patients with EB and their care takers.

Research in patients with a chronic condition and/or their parents demonstrates that the most important needs of patients/parents are being informed, being acknowledged, being partners in care, and recognized by professionals.

For all the needs and problems patients with EB need specialized care from many disciplines. An interdisciplinary professional team is the standard approach for complex diseases like EB. This includes multidisciplinary out-patient clinics and hospital admissions, as well multidisciplinary care when the patient is at home.

Medical specialists are experts in their field. The main problem is that experts cannot prioritise the patient's needs and wishes beyond their professional field. For that the patients need a case holder. In many patients one of the parents functions as case holder. However, the parent has no formal relation with the professionals. For patients with a chronic disease it is important to have also case manager in the hospital, who both can focus on the individual and the group of patients.

Our choice is to use a nurse practitioner as case manager working from the hospital. In addition DEBRA NL supports a social worker who works as a case manager from the home. The nurse practitioner and social worker do take a central position, with the parent/patient, in the network of professionals.

The nurse practitioner (typically female) is fitted for the job as case manager because of the generalistic and easy approachable and accessible role of her function. Besides her knowledge of options in care and cure in EB, of pa-

tients needs, and with competence in care for patients with EB, she also gives excellent advice to specialist during hospital procedures. She is able to make use of an accessible network of professionals. Furthermore, she also facilitates the collaborative relationship between medical professionals and administrators of the multidisciplinary EB-team. From the patients view, the nurse practitioner creates a relationship with the patients that emphasizes reciprocal exchange and mutual decision making

Together with the patient/parents the nurse practitioner lists and prioritizes the problems and coordinates and communicates the needed care by referring to the relevant professionals. In this way the collaborative practice of the EB-team provides superior patient care by combining the unique expertise of all professionals; it maximizes effective and efficient care.

It results in high levelled, demand driven, coordinated and collaborative care for EB-patients with complex problems and the collaborative multiprofessional care of the EB-team, including the social worker, meets the needs of the patients: optimal treatment, information, acknowledgement, and partnership in care.

#### PL:5

##### **MY ROLE AS A SOCIAL WORKER IN THE HOSPITAL SETTING**

*Janice Carrera (social worker)*  
DEBRA International Support (Magnolia Cottage School Rd, Saltwood Hythe, CT21 4QB,UK

The role of the Social Worker in the hospital setting is somewhat different to colleagues who work in the Community as there is rarely the opportunity to visit clients in their own homes due to distances. In a Specialist Centre such as St Thomas' Hospital London patients are treated from all over Europe and the world so not only is it difficult to assess living conditions but cultural and language differences become of prime importance. The Social Worker must act as an advocate on behalf of the patient. There is a need to be flexible, to advise and liaise, enable, promote and encourage disciplines to work together.

Communication with the patient is central to all care. Often we pay only 'lip service' to this concept and continue to talk 'about' the patient instead of 'with' him/her. We tell and advise with the very best of intentions, instead of listening to what the persons needs are. We often decide for him on his behalf since he/she is in 'no fit state to decide for him/herself'. This is often, we tell ourselves to protect the person, but it is often to protect ourselves.

On contacting the patient the Social Worker must find out whether or not their help is wanted. The official offer of help must be specific (not just "let me know if there is anything I can do"):

Become informed about the medical situation and offer to be at the clinic when the patient visits.

- Ask the patient to think about the questions that they want answered, help them by writing down their questions and put them in order of importance as time may be limited.

- During the appointment don't speak on behalf of the patient unless asked.
- Listen to the information the doctor gives, as the patient may want part of it repeated at a later date.

One of the greatest services a Hospital Social Worker can provide is to listen to the patient's fears and allow them to talk, often a service that the doctors do not have the time or training to deal with:

- We need to understand the patient's attitude to treatment:
- Expectations - desired outcome - level of motivation
- Anxieties - depression - fears of failure etc
- Quality of interaction with parents, siblings and relatives
- Parental attitudes and behaviours towards sexuality
- Ethnic and religious influences

Patients must be encouraged to visit the hospital on a regular basis for their skin to be checked for suspicious areas. This can be painful and frightening for them and this is where the Social Worker can be of great help in befriending, supporting and encouraging regular visits.

When a diagnosis of cancer is made it is often relayed to the patient or carer by a junior doctor who has not had the training in such matters. It would be preferable if the Social Worker were involved perhaps with the Specialist Nurse or Psychologist when an appropriate 'time and place' can be chosen to give such news.

Often the patient and/or carer want to discuss treatment, the stay in hospital and other practical arrangements in which the Social Worker can be of assistance. Patients and relatives have often been dissatisfied and confused by the information given and have felt it too difficult to seek clarification from the doctors.

Another very important role that a Hospital Social Worker undertakes is to advise and ensure that every EB patient has all the State Benefits and Allowances that they are entitled to, plus aids and adaptations to their home. This entails liaising with many multidisciplinary colleagues and official bodies within the Hospital & in the Community.

## PL:6

### **WITH FOCUS ON DAILY LIFE - A MEMBER STUDY**

*Elisabeth Wallenius, President of the Swedish Association of Rare Disorders*

As president of the Swedish Association of Rare Disorders, I have many times expressed that "rarity in itself" - the fact that a disorder is rare - is an additional disability. Within the association this has been the focus of our discussions rather than the problems with the various disabilities. A rare disorder makes the contacts with other people more difficult and complicates the contacts with those professionals whose duty it is to give care and support.

According to our members, they encounter the greatest problems in the contacts with care staff and the social insurance office. However, there have also been problems in the contacts with officials of the municipalities and in

school. Thus, the aim of the study was to establish the extent of the problems mentioned above, and to investigate the similarities and differences that exist between our different groups of disorders.

The study has been carried out through a questionnaire with mainly quantitative questions. The questionnaire was divided into two sections. One was concerned with how the family is affected and the other dealt with the problems that the person with the rare disorder encounters. Three of the questions were open questions, where members were asked to describe their situation with their own words. Members were also encouraged to write a story telling about their daily life and the problems they have to face. These stories make up the basis of the different case descriptions. The research material has been coded in such a way that the result can be applied to the entire group in the study, as well as to each individual disorder.

The results of the study not only reveal that most persons with a rare disability have very complex problems, but that the rarity as such is a particular problem. Consequently, the need for associations for various disorders has been expressed clearly in the study. The questionnaire has been answered by 37 (27 women and 10 men) of the DEBRA association's members. The results show that the problems in practical life are extensive and that plenty of time is needed for personal care. 50 % of those who have answered also declare that they have extra expenses of SEK 200 or more per month due to DEBRA.

In comparison to others with a rare disorder, many persons with a DEBRA diagnosis experience that being diagnosed facilitates contacts with specialised care. But still more people state that they have problems with the latter. Many have also experienced problems in the contact with other care givers and more than 50 % say that they have been exposed to malpractice on one or more occasions.

## PL:7

### **THERAPEUTIC SUMMARY: THE SKIN**

*Jackie Denyer, EB clinical nurse  
London*

This short presentation will highlight the danger of clinical diagnosis in the absence of supportive immunohistochemistry and electron microscopy findings.

It will raise the question "are skin lesions in infants a reliable indication of disease or prognosis?"

Factors such as mode of delivery and use of inappropriate dressing materials will be used to demonstrate factors responsible for severe skin loss, often suggesting severe disease.

A short case study will relate an unusual scenario with disturbing consequences.

## PL:9

### **THERAPEUTIC SUMMARY: MOUTH AND TEETH**

*Nina Skogedal, specialist in Pediatric Dentistry*  
TAKO-centre, Resource Centre for Oral Health in Rare  
Medical Conditions, Lovisenberg Diakonale Hospital, Oslo

Epidermolysis bullosa (EB) is characterized by blistering of the skin and mucosal membranes. EB exist in different forms, and the most severe affliction is seen in the dystrophic EB.

Oral health problems include vesicular and bullous eruptions of mucous membranes, scarring, restricted mouth-opening, impaired movement of lips and tongue and pain. Hypomineralized and hypoplastic teeth are recognized as features in some types of EB. Oral hygiene may be painful and difficult to carry out due to affected hands and oral blisters.

Dental treatment of the EB patient is a challenge due to microstomia, fragility of oral mucosa, obliteration of oral vestibulum, reduced tongue-mobility and hypersensitive teeth. Therefore, the highest priority in the EB-group as a whole, is prevention of oral disease. Counselling in proper oral hygiene routines should start before eruption of the first primary tooth. The preventive regime includes frequent professional cleaning and fluoride treatment as well as dietary advice.

The presentation will focus on implications complicating daily oral hygiene in these patients as well as precautions which need to be taken by dentist/dental hygienist to avoid unnecessary blisters and pain for the patient. Various methods available to make an oral hygiene programme easier will be presented. Furthermore, examples of dietary advice for caries prevention will be emphasized. Cooperation with other health care personnel is essential in order to give good oral care to the EB-patient.

## PL:10

### **FUTURE THERAPY: GENE THERAPY ?**

*Leena Bruckner-Tuderman*  
University of Freiburg, Germany

Due to the severe symptoms in some EB subtypes, a scientifically valid therapy is urgently needed. Different strategies using genetic or protein materials have been critically evaluated, with all of their positive and negative aspects. The conclusion is that the most promising therapeutic developments today are in the field of skin gene therapy, although clinical applications still may be years away. The skin, or keratinocyte, gene therapy is based on introduction of a normal gene into cells isolated from a skin biopsy of a person with EB. The "repaired" cells will be grown into a skin graft which can be transplanted onto a very fragile skin area. The graft should develop into stable adherent skin.

Many research centers work on the development of gene therapy protocols and several international networks have been formed in order to combine expertise and to

optimize progress. The expertise of dermatologists, cell and molecular biologists, virologists, and geneticists is needed to develop optimal gene vectors, cell cultures, product quality controls, transplantation techniques and clinical management. Many technical problems must yet be solved, and the prevailing opinion is that the efficacy of skin gene therapy must be proven in animal models before clinical trials can be considered. In addition, significant bureaucratic and legal hurdles must be overcome before treatment of individuals with EB can be tested.

## PL:11

### **PEER COUNSELING, CO-OPERATION BETWEEN PATIENTS, PROFESSIONALS AND SOCIETY**

*Jan Kristen Aaseth,*  
DEBRA Norway

The first patient organizations were largely initiated and to some degree dominated by healthcare professionals. In the last 30 years, most patient organizations have been "taken over" by the members, and become organizations of the patients.

Patient organizations today have more members than political parties; this is a trend seen throughout Europe.

Due to the development in medicine, giving more conditions and illnesses a clear diagnosis, there has been a large increase in patient organizations, most within small and rare diagnoses. In many these diseases the healthcare services have limited knowledge of treatment and experience with such ailments.

Most patient organizations, large or small, have very similar primary objectives, i.e., to inform us what the members find most important.

Promote professional knowledge and the best treatment. Patient organizations have a lot of knowledge about their diagnoses, and who and where in the healthcare services key personnel are. Additionally, there is often an international network where contacts can be made with international experts. These are important resources that professional healthcare services should use and benefit from.

Provide services and support to people whose lives are affected by the diagnosis. Patient organizations give advice and information to their members, and by doing so they generate a feeling of security. This enables the patient to participate more actively in his own treatment. The most unique, and in my opinion the most important, resource the patient organizations have is their knowledge of what living with the diagnosis means. Based on this knowledge they can contribute with answers and comfort, and help there members to establish a new normal life, with there diagnose.

This is an experience no professional healthcare service will ever be able to attain.

## WORKSHOPS/FORUM/POSTERS (= Free Communications in order of submission)

### FC:1

#### EXPRESSION OF MATRILYSIN-1 (MMP-7) AND COLLAGENASE-3 (MMP-13) AND LOSS OF MMP-19 IN CUTANEOUS SQUAMOUS CELL CARCINOMA IN RECESSIVE DYSTROPHIC EPIDERMOLYSIS BULLOSA.

Atte Kivisaari<sup>1</sup>, J. McGrath<sup>2</sup>, J. W. Bauer<sup>3</sup>, F. Weber<sup>4</sup>, R. Königová<sup>5</sup>, D. Sawamura<sup>6</sup>, K. C. Sato-Matsumura<sup>6</sup>, H. Shimizu<sup>6</sup>, I. Hausser<sup>7</sup>, K. Sinemus<sup>8</sup>, W. Beckert<sup>9</sup>, M. Csikós<sup>10</sup> and V-M. Kähäri<sup>1</sup>.

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Matrix metalloproteinases (MMPs) are a family of zinc-dependent metalloendopeptidases collectively capable of degrading essentially all extracellular matrix components. Expression of MMP-7 and MMP-13 are associated with invasive phenotype of many malignant tumors including UV-induced squamous cell carcinomas of the head and neck in non EB-population. MMP-19 is induced in hyperplastic epithelium in wounds, but absent from invasive areas of squamous cell carcinoma.

Recessive dystrophic epidermolysis bullosa (RDEB) is one of the most severe hereditary mechano-bullous diseases, characterized by scarring blister formation, nail dystrophy, cutaneous contractures, mutilations of the hands and feet and oesophageal stenosis. Cutaneous squamous cell carcinoma (SCC) is a frequent complication in chronic ulcers in these patients. Although the cutaneous SCCs in RDEB are in general histologically well differentiated they are characterized by rapid invasion and development of metastases resulting in poor prognosis. Cutaneous SCC is one of the major causes of the premature death of RDEB patients.

In this study, we wanted to elucidate the role of extracellular matrix degrading enzyme matrilysin-1 (MMP-7), collagenase-3 (MMP-13) and MMP-19 in development and invasion of these carcinomas.

We have obtained formalin-fixed, paraffin embedded samples from 25 cutaneous SCCs and 3 lymph node metastases from 18 RDEB patients. Immunostaining of the sections was performed by the avidin-biotin peroxidase complex. 3-amino-9-ethylcarbazole (AEC) was used as chromogenic substrate and Mayer hematoxylin as a counterstain. We used mouse monoclonal antibodies to study localization of MMP-7 and MMP-13. For detection of MMP-19 we used rabbit polyclonal antibody. Negative control stainings were performed without primary antibody.

In tissue sections cytoplasmic staining for MMP-7 and MMP-13 was noted in tumor cells but not in stromal cells. MMP-7 expression was also detected in tumor cells in

lymph node metastases examined. MMP-7 was also expressed by exocrine epithelial cells in sweat glands as a positive control where as no staining was noted in normal epidermis or in negative control stainings. Staining for MMP-19 was detected in keratinocytes in hyperplastic epithelium, but not in malignant cells.

Expression of MMP-7 and MMP-13 is specifically induced in malignantly transformed keratinocytes in this aggressive subset of cutaneous SCCs suggesting a role for these MMPs in early cutaneous SCC development, invasion and metastasis in RDEB patients. Also the loss of expression of MMP-19 may have a role in malignant transformation of the keratinocytes.

These results indentify MMP-7 and MMP-13 as a potential therapeutic targets to inhibit growth and invasion of SCCs in patients with RDEB.

RDEB patients have abundantly chronic ulcers and non-malignant wounds which may resemble histologically well-differentiated SCCs. The loss of MMP-19 might help in making the differential diagnosis.

### FC:2

#### EB CENTRE IN CZECH REPUBLIC

Buková, H.<sup>1</sup>, Buek, J.<sup>2</sup>, Vokurková, J.<sup>1</sup>, Samohel, J.<sup>2</sup>, Brauner, R.<sup>1</sup>, Kozák, L.<sup>1</sup>, Fajkusová, L.<sup>1</sup>, JeYábková, B.<sup>1</sup>, Gaillyová, R.<sup>1</sup>, Buková, E.<sup>1</sup>, Procházková, D.<sup>1</sup>, Svobodová, N.<sup>1</sup>, Kittlerová, E.<sup>1</sup>

<sup>1</sup>University Hospital Brno, Czech Republic, <sup>2</sup>St. Ann's University Hospital Brno, Czech Republic

In 2001 The EB centre in Czech Republic was founded by the Department of Pediatric Dermatology, University Hospital Brno. Over the last ten years we register 89 EB patients (46% DEB, 10% JEB, 44% SEB).

The centre follows patients from the neonatal period until adulthood. The complex care is based on the work of a multidisciplinary team of specialists coordinated by the dermatologist. The team contains of dermatologist, plastic surgeon, geneticist, histopathologist, gastroenterologist, pediatrician, ophthalmologist, physiotherapist, dentist, nurse specialist, social worker, psychologist and dietician.

The diagnose is based on the clinical features, histological examination including imunohistochemistry and electron microscopy and molecular genetic analyse. From autumn 2004 the DNA analyse of the dystrophic form has been introduced by our Centre of molecular genetics.

The dystrophic, junctional and generalised simplex forms visit the centre regularly 3 or 4 times per year. The most serious cases and complication are solved by a special committee gathering 3-4 times per year. The wound care is performed by special trained nurses. In May 2004 was founded DebRa CR, who is a member of DebRA International.

The aims of the EB Centre in Czech Republic are complex care of EB patients development and improvement of the molecular genetic analyses introduction of prenatal diagnostic foundation of the patients register closer international cooperation on the EB field

### FC:3

#### **FIRST EXPERIENCES WITH MUTATION ANALYSIS OF DYSTROPHIC FORM OF EPIDERMOLYSIS BULLOSA IN THE CZECH REPUBLIC**

*B. JeYábková, L. Fajkusová, L. Kozák, H. Buková*  
Center of Molecular Biology and Gene Therapy, University Hospital Brno, the Czech Republic, Department of Pediatric Dermatology, University Hospital Brno, the Czech Republic

Dystrophic epidermolysis bullosa (DEB) is a clinically heterogeneous skin disorder, characterized by abnormal anchoring fibrils at the dermal-epidermal basement membrane zone. DEB has been linked to the COL7A1 gene at chromosome 3p21 which encodes collagen VII, the major component of the anchoring fibrils. The gene consists of 118 separate exons. DEB is transmitted in autosomal recessive (RDEB) or dominant (DDEB) fashion.

We investigated 13 patients, two with DDEB and 11 with RDEB. We designed 87 primer pairs corresponding to flanking intronic sequences which allow PCR amplification of all 118 exons directly from genomic DNA. The first step of mutation analysis of each patient is sequencing of exon 73 (over 10% of all COL7A1 mutations have been found in it), and exon 74, 75. Others exons are examined by denaturing high performance liquid chromatography (DHPLC) and if the result is positive, the PCR products are sequenced. The exons with high probability of occurrence of some mutation are analysed using DHPLC in the next step (see mutation map on pages: <http://archive.uwcm.ac.uk/uwcm/mg/search/128750.html>). Four mutations in exon 73 were identified: the substitution G2049E in three patients and the insertion 2027insC in one patient and the substitution R2063W in exon 74. These are recurrent mutations which have been reported previously. We would like to analyse all exons and flanking intronic sequences of each patient and his relatives by this procedure. Our intention is also introduction of DNA diagnostic of EB simplex and junctional EB.

The present diagnostics is based on clinical manifestations, immunohistochemical analysis and electron microscopy. We would like to extend a spectrum of methods used in EB diagnostics in the Czech Republic by analysis of the genes linked to the individual types of EB. Identification of mutations in affected families has important implications for genetic counselling - the detection of carriers, assessment of the mode of inheritance and early prenatal diagnosis. It is also the first step towards gene therapy.

### FC:4

#### **SOMATIC REPRESSION IN JUNCTIONAL PRETIBIAL EPIDERMOLYSIS BULLOSA (PEB) IS REVERSIBLE**

*Laimer M, Bauer JW, Klausegger A, Koller J, Pohla-Gubo G, Muss W\*, Sadler E, Emberger M, Lanschuetzer CM, Hametner R, Wally V, Oender K, and Hintner H*  
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Pretibial epidermolysis bullosa (PEB) is a genetic mechanobullous disease with an anatomically restricted phenotypic expression, the reason of which is unknown. We describe a patient suffering from the typical clinical symptoms of PEB that is principally a subtype of dystrophic epidermolysis bullosa (DEB). Ultrastructural, immunohistochemical and laminin-5 gene analyses, however, were consistent with junctional PEB. This notion was further substantiated by the DNA sequence analysis indicating compound heterozygosity for the missense mutation C290S and the nonsense mutation R635X in the LAMB3 gene. Skin grafting was successfully performed as a therapeutic approach with a favourable long-term outcome. Remarkably, at the graft donor sites the patient developed reproducibly prolonged blistering of up to 6 months suggesting activation of a formerly repressed blistering pheno-/genotype. Therefore we conclude that the morphologic characteristics of PEB are ubiquitously inducible if provoked by mechanical and/or inflammatory stimuli (Koebner phenomenon). It is likely that this somatic repression is a mechanism operative in other patients with localised disease expression.

### FC:5

#### **"EB-HAUS": THE AUSTRIAN CENTRE FOR EPIDERMOLYSIS BULLOSA**

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The various cutaneous and extra-cutaneous problems of Epidermolysis bullosa (EB) require interdisciplinary care and extensive coordination of all who are involved. Thus, the decision was made to provide the necessary infrastructure by building a small centre of excellence for EB: the "*eb-haus*" in Salzburg.

To reach this objective in times of low-budgets in health care systems the Department of Dermatology at the General Hospital Salzburg has initiated a unique co-operation with the support group *debra-austria* ([HYPERLINK "http://www.debra-austria.org"](http://www.debra-austria.org) [www.debra-austria.org](http://www.debra-austria.org)) which raises funds for the building and supports the research in an outstanding and very successful way.

The "*eb-haus*" consists essentially of three parts:

- A centre for therapy on an out-patient base with an EB-physician, an EB-nurse, co-operating with a team of experts of various medical disciplines.
- A research department for basic science, genetic research and clinical trials.
- An academy which facilitates the intense interchange of knowledge and experience between scientists, physicians, nursing staff, patients and their relatives.

This centre of excellence will concentrate all strategies to face the difficulties of this at present incurable, severely disabling and sometimes life threatening disease. The resulting efficiency will be of great benefit to all EB suffer-

ers in Austria - and also Europe-wide - and considerably improve their quality of life.

## FC:6

### **CASE REPORT: JUNCTIONAL EPIDERMOLYSIS BULLOSA (EB) WITH ECTHYMA GANGRENOSUM**

*A.Diem, J.W.Bauer, I. Walser\*, G.Pohla-Gubo and H.Hintner,*

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We report on a girl with junctional EB of the Herlitz type with Ecthyma gangrenosum. A few days after hospitalisation because of fever she developed skin lesions on her neck, shoulders and back with dark red colour, a central haemorrhagic blister and subsequent necrotic skin lesions. *Pseudomonas aeruginosa* was found in tissue and blood culture. Despite intravenous antibiotic therapy she died of *Pseudomonas sepsis*.

Skin infection with *Pseudomonas aeruginosa* is very common in junctional EB of the Herlitz type, but this progressive course of the infection is rare. We will discuss possible underlying mechanisms and treatment options.

## FC:7

### **ESOPHAGEAL DILATION AND PEG -TUBES IN CHILDREN WITH EPIDERMOLYSIS BULLOSA**

*J. Bauer, T.M. Boemers, \*Anja Diem*

Clinic of Pediatric Surgery, SALK, \*Clinic of Dermatology, SALK, Salzburg, Austria

Six pediatric patients with dysphagia due to esophageal stenosis were treated by esophageal dilation in our clinic. The onset of dysphagia generally occurred between the sixth and the seventh year of age.

The repeated esophageal dilation relieved dysphagia and improved the nutritional status in all patients.

Unfortunately, esophageal strictures and developing microstomias made necessary to create PEG-Tubes in three of our patients.

## FC:8

### **RELEASE OF PSEUDOSYNDACTYLY OF HANDS IN CHILDREN WITH EB?**

*Barbara Ludwikowski, Dept. of Paediatric surgery Paracelsus Private Medical University Salzburg- Austria*

Children with recessive dystrophic epidermolysis bullosa and development of pseudosyndactyly of the hands are presented. Surgical intervention is commonly recommended and performed. Unfortunately the need for repeated surgery is necessary. Musculoskeletal complications are

described, if no intervention is performed and early interventions are recommended (*J Hand Surg [Br]. 2005 Feb; 30 1: 14-22*).

We want to discuss the indication for surgical interventions. A benefit of this surgery should be for the children and families in their daily life. We reinvestigated our patients (clinical, x-ray and questionnaire) to get more information about the outcome and questioned early surgery.

## FC:9

### **SURGICAL MANAGEMENT OF THE HAND IN DYSTROPHIC EPIDERMOLYSIS BULLOSA**

*J.Vokurkova,M.D., Ph.D., H.Buckova, M.D., Ph.D. Debra Brno, Czech Republic*

Surgical correction of hand deformities and treatment of spinaliomas of DEB patients has a long history on Plastic surgery department in Brno, Czech republic. Now, all patients are registered in the Center for Epidermolysis Bullosa (EB) in Brno which is based on the multidisciplinary team cooperation - DEBRA,CZ.

In the last ten years our center gathered 89 patients with EB, 46% of them had DEB. Eleven out of 14 patients with acral form of DEB have undertaken 56 hand surgeries. The mean age was 16 years (range 4-48 years). Extensive surgical procedure with separation of the fused web spaces and releasing flexion contractures was performed. The raw areas were covered by split skin grafts harvested on the thigh. The splitting of the hands was continuous for 3 months and then fingers were webbed daily with splitting during night.

Assessment of the results of surgery was measured by extension and flexion deficit and the adduction deformity of the thumb. The recurrence of pseudosyndactyly was observed each 3 months.

Clinically major improvement was observed in the patient's grasp and the ability to pick up objects. The young patients without secondary joint disease achieved better results with surgery than patients with long standing uncorrected hand deformity. The better long term splittage the better functional results.

## FC:10

### **AMNIOTIC MEMBRANE IN OCULAR SURGERY FOR EB.**

*Gabor Koranyi, MD, Ophthalmologist St. Eriks Eye Hospital, Stockholm, Sweden*

Symblepharon, adherens between the eyelid and the eyeball is not uncommon in dystrophic EB. It is progressive, obstructs vision and eye movements. Material and methods: A 5 years old patient with a major symblepharon in the left eye was presented for surgery. The lesion affected more than half of the cornea. At the surgery a minor symblepharon was found also in the right eye. The eyes were operated with the use of amniotic membrane.



Results: After 5 years follow up, the right eye is still without symblepharon, while the surgery on the left eye had to be repeated several times with still unsatisfying result. Conclusion: Early surgery of symblepharon could prevent further development of this sight threatening condition.

## FC:11

### **ANAESTHESIA IN EB-PATIENTS.**

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Patients with epidermolysis bullosa need careful preoperative screening regarding infection, anaemia and electrolytes in order to correct any disturbances before the planned procedure.

In the operating theatre special steps must be taken to ensure protection of the skin and airways. A general principle is to avoid all kinds of tape and adhesives. Intravenous cannulas must be fixed with gauze and bandage, and stitches might also be used. Gentle airway manipulation is of utmost importance. If tracheal intubation is necessary, great care must be taken when introducing the laryngoscope and endotracheal tube in order to avoid blistering of the mucosa or damage to the skin.

The monitoring systems anaesthetists routinely use, are electrocardiography (ECG), pulse oxymetry, blood pressure and capnography.

ECG monitoring with adhesive pads must be avoided. The pulse oxymetry probe if possible to use, need to change position so as not to harm the skin. Blood pressure monitoring with cuffs must be placed over soft gauze.

The position of the patient on the operating table is important. The operating table should be soft and comfortable and the armrests must be equipped with atraumatic padding.

All personnel taking part in the preoperative, operative and postoperative procedures should be properly informed about the skin problems of EB patients in advance, in order to avoid any additional blistering.

## FC:12

### **ALTERATIONS OF ORAL MUCOSA IN HEREDITARY EPIDERMOLYSIS BULLOSA**

*Elke Sadler, Anja Diem, Helmut Hintner*

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The appearance and structure of perioral and intraoral epithelia differ markedly from one anatomic site to another, reflecting unique functions of different oral tissues under the variety of stresses that they endure. Oral mucosa is variable and frequently affected in hereditary epidermolysis bullosa (EB). The most severe and most frequent alterations are found in patients with recessive dystrophic EB (RDEB) subtypes. These patients show

intraoral disease activity in 92,3-100%. Mucosa of dominant dystrophic EB (DDEB) subtypes is affected in 81,1%, of junctional EB-Herlitz (JEB-Herlitz) in 83,3%, of JEB-non-Herlitz in 91,6% and of EB simplex in 34,7-58,6%. Most abnormal oral findings in EB patients are oral blisters and erosions, chronic gingivitis and oral milia (DEB 49-54%), occurring most often on the palatal mucosa. In RDEB chronic blisters and erosions can lead to perioral and intraoral scarring resulting in lack of lingual papillae, lack of palatal rugae, ankyloglossia, obliteration of the vestibulum oris and microstomia. Additional, continuous oral blistering, which results in oral scarring, ankyloglossia, and vestibular obliteration, may produce tissue changes with malignant potential. Because squamous cell carcinomas have been reported to occur in perioral and intraoral (tongue) sites in patients with generalized RDEB, periodically oral examinations of patients are important throughout life. Therapy of oral mucosal alterations is limited and consists of prophylactic strategies as soft diet, sucralfate and dental care.

## FC: 13

### **TREATMENT OF SWEAT-INDUCED BLISTERING OF THE FEET**

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Plantar sweating at high ambient temperatures increases the blistering in patients with EB simplex (EBS), which is due to KRT 5 or 14 mutations, as well as in pachyonychia congenita (PC), which is due to KRT 6, 16 or 17 mutations. The latter condition is also associated with painful, focal hyperkeratosis on the soles. Treatment of plantar sweating is notoriously difficult, but local injection of botulinum toxin (btx) can produce longstanding anhidrosis in patients with primary essential plantar hyperhidrosis without skin disease (C.Swartling, Thesis 2002). The anaesthesia required for the plantar injections is however a problem.

Here we report 5 patients with EBS and PC who had great walking problems, especially in summer time, due to painful blistering of the soles. They were treated with intracutaneous plantar injections of btx type A (Dysport, 100 units/ml, Ipsen, Slough, UK) after prior intravenous regional anaesthesia of the foot with a low tourniquet and 25 ml prilocain (5 mg/ml). Within one week all 3 PC patients experienced dryness and a remarkable relief of pain at plantar pressure sites. The effect duration was between 6 weeks and 6 months. Repeated injections over a 2 year period confirmed the good results with no side-effects or tachyphylaxis noted. Two patients with EBS were treated in the same way with unilateral btx injections. However, despite obvious anhidrosis on the treated side in both cases, the intensity of blistering and pressure-induced pain only marginally benefited from the therapy. It remains to be seen whether PC and EBS are truly different in this respect or if subsets of other patients with EBS may also show an excellent response to btx therapy.

## FC: 14

### **PHENOTYPE AND MOLECULAR MECHANISM ASSOCIATED TO GLYCINE SUBSTITUTION MUTATION IN THE LARGEST COLLAGENOUS DOMAIN OF COLLAGEN XVII**

*Laura Väisänen, Cristina Has, Marja-Leena Tuomi, Leena Bruckner-Tuderman, and Kaisa Tasanen.*  
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Mutations in the collagen XVII gene, COL17A1, are associated with junctional epidermolysis bullosa. Most COL17A1 mutations lead to a premature termination codon (PTC), while only a few mutations result in amino acid substitutions or deletions. We describe here a novel glycine substitution, G612R. A transition c.1834G>A causing a Gly to Arg substitution at the amino acid position 612 (G612R) was found from an 8-year-old Finnish girl, who had rather severe blistering at birth already. Generalized moderate blistering continues, and she also has nail dystrophy, dental problems, poorly healing wounds and partial alopecia of the scalp. Immunohistochemical staining with basement membrane zone antibodies showed junctional blistering with reduced collagen XVII staining. Her 22-year-old brother has had blisters since the neonatal period. His disease is milder than his sister's, but he also has partial alopecia and tooth and nail dystrophy. Based on clinical data and electron microscopic analysis in the early 1980s, he and also his sister were thought to have epidermolysis bullosa simplex. Their case demonstrates very clearly the importance of immunofluorescence antigenic mapping and mutation analysis for correct diagnosis of epidermolysis bullosa.

In order to investigate the molecular pathomechanisms of this glycine substitution, G612R was introduced into recombinant collagen XVII. The mutated collagen was expressed by transfection in COS-7 cells and its thermal stability was assessed using trypsin digestions at incremental temperatures. G612R significantly destabilized the ectodomain of collagen XVII, which manifested as 16°C lower T<sub>m</sub> (midpoint of the helix-to-coil transition) in trypsin assay. Thus, glycine substitution interferes with tight folding of the triple helix and renders the ectodomain of collagen XVII sensitive to non-specific degradation.

## FC: 15

### **THE IMPORTANCE OF INFORMATION AND PREVENTION OF PAIN IN CHILDREN UNDERGOING PAINFUL PROCEDURES**

*Ingrid Bäckmark, RN*  
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According to Swedish laws, all patients, including children and their parents, have the right to get correct information before undergoing examinations or procedures. This information should be given in a positive way on a level that the child can understand, according to the child's age and level of development.

Prevention of pain according to the procedure or surgery which is to be done should be started in a preventative way with local anaesthetics or different type of analgesia.

## FC: 16

### **POST-OPERATIVE HAND THERAPY IN RDEB**

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The aims of postoperative treatment of the hand in children with EB are to maintain optimal range of motion of the wrist, fingers and thumb and to delay recurrence of deformity in order to enlarge the possibilities of hand function. In literature two types of postoperative treatment programs are described: a program with static splinting and a program with dynamic splinting. The splints are aimed for abduction of the thumb and extension and abduction of the fingers.

In the University Medical Centre Groningen (UMCG) in the Netherlands the postoperative treatment is done by the occupational therapist/hand therapist and the rehabilitation physician, in close co-operation with the plastic surgeon. This treatment includes dynamic splinting, followed by static splinting in combination with exercises.

The dynamic splint is fabricated prior to surgery. The patient starts wearing the dynamic splint day and night after the first wound dressing, approximately ten days following surgery. The dynamic splint allows very early mobilisation of the hand to prevent recurrence of the deformities, but does not interfere with wound healing and dressings.

Approximately 21 days after surgery, depending on the progression of the healing of the wounds, a static splint is made for wearing during the night. The dynamic splint has to be worn several times a day in order to give well-balanced stretching of the wound and soft tissue of the hand. This splint is gradually cut back. The static splint should be worn at night as long as possible. Mullet (1998) wrote: "improved survival of children with RDEB seems to be accompanied by gradually decreasing severity, and relatively long-term stability may be achieved after the mid-20's". This might indicate that night splints should be worn at least until the mid-20's.

After approximately ten days following surgery, an exercise-programme is initiated in order to improve the joint mobility, prevent joint-contractures and prevent adhesions of the M. flexor digitorum profundus (fdp) and the M. flexor digitorum superficiales (fds). Initially, the wound and fragile skin should be protected against bumping and risk of wound infection. It is very important for the child to regain trust in using the hands in daily activities like playing, eating and writing. Regular exercising is phased out step by step. This depends on the function and the degree of usage of the operated hand by the child.

In addition to hand problems, the child with RDEB is at risk of a great deal of other physical problems. Surgery and the postoperative treatment of the hand are very time-consuming and have a great impact on the lives of both the child and their parents or caregivers. In the process, if and when surgery should take place the possibilities

and compliance of the child and his or her environment should be taken into consideration. The final result of the operation depends on the perseverance of both.

#### **FC: 17**

##### **BODY MASS INDEX IS A RELIABLE TOOL TO OPTIMIZE LENGTH GROWTH IN PATIENTS WITH SEVERE DYSTROPHIC EB**

*Irene C.E.M. Herpertz, Han M. Mandema, Jose C. Duipmans, Marcel F. Jonkman*  
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The incidence of length growth retardation in patients with severe dystrophic epidermolysis bullosa is high. It is generally accepted that an optimum height gain requires an adequate weight. Until now, the weight-for-height chart is a frequently used method to determine the ideal weight. However, weight for height can be normal in children with a growth delay. On the other hand the body mass index (BMI) is a generally accepted tool to identify overweight or obesity in patients. BMI is sex and age specific.

Therefore, we hypothesized that BMI standard deviation score (SDS) is a reliable tool to determine an ideal weight in children with severe dystrophic epidermolysis bullosa.

We performed, retrospectively, a survey on five patients (age 7-27) with severe dystrophic epidermolysis bullosa (3 RDEB-HS, 2 RDEB-inv). Weight and length of the patients and length of the parents was obtained. Standard deviation curves for BMI (weight/height<sup>2</sup>) for age (years) and height (cm) for age and the target height channel were constructed using Growth Analyser 3 (version 3.0) software.

Results: A BMI SDS below -1 leads to a progressively deviation from the previously defined growth channel.

Conclusion: In clinical practice the BMI can also be feasible to establish the ideal weight and, as such, create the ideal circumstances for an optimal length growth in patients with severe dystrophic epidermolysis bullosa.

We recommend the ideal BMI SDS 0, (minimum requirement -1/2 SDS and maximum requirement +1 SDS) to establish an optimal length growth in these patients.

#### **FC: 18**

##### **IN SEVERE EB, WHAT IS DESIRABLE WEIGHT STATUS TO PROMOTE BOTH MOBILITY AND OPTIMAL NUTRITION?**

*Lesley Haynes, dietician, and Jacqueline Denyer, EB specialist nurse*  
London

Maximising quality of life in severe EB is paramount and maintenance of optimal nutritional status and mobility are major challenges. Many factors influence, and are influenced, by both these issues and the forum will consider some of these, for example:

- # optimal weight gain when patients' heights are often compromised (use of growth and waist circumference charts), definition/assessment of overweight/underweight
- # effect of neonatal lower limb lesions and dressings on later weight-bearing
- # impact of pain relief on weight-bearing
- # promotion of wheelchair use
- # impact of steroids on excess weight gain
- # provision of adequate nutrition to promote immunity and wound healing whilst minimising tendency to accrue fat in preference to lean tissue

#### **FC: 19**

##### **NURSERY SCHOOL AND SCHOOL - TAILORING FOR EB-CHILDREN**

*Nina Braathen, occupational therapist, Norway*

The lecture will exemplify: 1) how the physiotherapist uses different positions and exercises to teach and maintain body control and good balance for the EB-client while at school, and 2) how the occupational therapist can work with the EB-family and school staff to tailor the environment in order to prevent sores and blisters to occur; i.e., what to look out for outside and inside buildings.

The need and contents of information to caretakers and other persons around the client will also be discussed. The main focus is to prepare for as good life quality as possible for the whole family. This includes:

1. A meaningful "every-day life" for the client
2. Independence to the greatest possible extent
3. Preventing sores, wounds and skin blisters by tailoring the environment
5. Planning for suitable transportation with mechanical aid (car, wheel chair and bicycle)
6. Providing examples for play-ideas and leisure time