DEBRA international CONGRESS 2008 Mechelen, Belgium

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CV's and Abstracts

Gunnar Naulaers

Prof Dr Gunnar Naulaers
Born 13-01-1965 in Leuven. Married with Hilde Boonants and father of three children : Rob, Astrid and Karel.
Paediatrician and neonatologist. Head of the neonatal intensive care unit.
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"Epidermolysis bullosa in Leuven : 1998-2008"

When a baby with epidermolysis bullosa was transferred to the neonatal care unit in december 1997, a protocol for neonatal treatment was made. Since than the unit developed a special interest in the care for babies with epidermolysis bullosa. A treatment protocol was made with also special attention for pain relief in babies, for which the Leuven neonatal pain scale was used. As the babies grew older, there was an increased need for follow-up. Therefore a multidisciplinary consultation was started, incorporating different specialisms. Two EB-nurses (0,2 FTE), supported by DEBRA, coordinate the care and follow-up. As children with EB were admitted in the hospital, different protocols and procedures were written in order to give standard care. The importance of psychological support became increasingly more important and the child psychiatry unit and the hospital school were asked for support. This gave us a new view and approach to the care of children with epidermolysis bullosa. Over the last 10 years, different people from different specialisms became personally involved in the care of EB, resulting in a better and more personalized care.

Joseph Schoenaers

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Mouth and dental problems: "Release of mouth contracture"

Abstract: Schoenaers J., Verhaert S.

Epidermolysis bullosa comprises a group of rare genetic disorders of the skin and mucosal surfaces, which are characterized by easy blistering after minimal friction.

The recurrent blistering of the oral mucosa can lead to fibrotic scarring and subsequent microstomia, ankyloglossia and difficult dental hygiene.

We present 2 case reports, up to 9 years of follow-up, where we ameliorated patient's comfort and mouth opening by performing a vestibuloplastic surgery with split thickness skingrafts.

Susanne Krämer

Susanne Krämer is a dentist from Chile. She has been working and researching about EB since she was a student at the School of Dentistry at the Universidad de Chile, where she studied the oral manifestations of different subtypes of EB. Currently she leads the dental team in Debra Chile at a distance because she is doing a Master of Science in Special Needs Dentistry at the Eastman Dental Institute in London and she is researching about different therapeutic approaches for increasing mouth opening in patients with EB.
Susanne Krämer,Debra Chile - susiks@yahoo.com

Mouth and dental problems: "Experience in Chile"

This presentation will include:

1. Identification of the main oral manifestations of the major EB types.
2. Oral health related Quality of Life data reflecting patients concerns.
3. Current dental treatment protocol - Debra Chile.
4. Overview on literature available and ongoing research in EB and Dentistry.

Marcel Jonkman

The career of Marcel F. Jonkman, born in 1957 in Hengelo (the Netherlands), was mainly on the University of Groningen. He graduated in 1984 as MD and finished his Ph.D.-thesis on artificial skin and epidermal wound healing in 1989 in the Department of Medical Electron Microscopy (prof. dr. I. Molenaar). From 1988-1992 he followed a dermatological tranining in the University Hospital (prof. dr. J.P. Nater, joined the faculty as assistant-professor in 1993 and was promoted to associate professor in 1997. In 1996 he visited the lab of dr J. Uitto Jefferson Medical College, Philadelphia as visiting professor. In 1999 he raised the Centre for Blistering Disorders in the dermatological department in Groningen. In 2002 he was appointed as full Professor of Dermatology with special regard to bullous diseases. In 2003 he was appointed as chair of the department of Dermatology in Groningen. In 2004 he was appointed as residents Programme director.
Dr Jonkman is founder of the Dutch Society of Experimental Dermatology. In the University Hospital Groningen he runs special clinics in bullous autoimmune diseases, epidermolysis bullosa, and genodermatoses. He is married and has three children.

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Research: "Revertant mosaicism in EB is not rare"

Some patients with junctional EB notice normal skin patches that never blister and feel strong. These patches may be caused by a kind of natural gene therapy caused by a kind of back mutation in the DNA during embryological development that restores the function of the specific EB gene, a phenomenon called revertant mosaicism. The skin daughter cells derived from the reverted skin stem cell produce the normal protein, and therefore have gained normal function.

To date we discovered in 33% COL17A1 mutant nH-JEB patients reverse mutations in skin cells. Moreover, in a series of three LAMB3 mutant nH-JEB patients, revertant mosaicism was revealed in 66%. Rigorous physical examination revealed multiple revertant skin patches within each individual that had been repaired by different in vivo reversions (second-site mutation, gene conversion or a real back point mutation) located near the germline mutation. The number of reversion events was therefore more than one in each individual. The highest number of different reversions observed in a single patient sofar is four. In all patients the clinical reversion was underlined by enhanced expression of protein.

We conclude that reversion is overlooked easily and happens more often than expected. Our data offer for a growing number of cases prospects for EB treatment. The possibility of applying revertant cell therapy in mosaic EB using autologous naturally corrected keratinocytes bypasses the recombinant gene correction phase. If succesfull it will lead to quick and easy replacement of diseased skin with minimal risks.

Anja Diem

Physician since 1993, working in the field of EB since 2003 at the Department of Dermatology in Salzburg (head of department: Prof. Helmut Hintner). Since November 2005 Manager of the outpatient clinic at the eb-house Austria, first contact person for all medical problems associated with EB, working together with a great team of doctors, nurses and many other professionals of the General Hospital Salzburg and Paracelsus Private Medical University.
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"To treat or not to treat ?"

This presentation is about the many treatment decisions that have to be made in a life with EB and in a life as a health care worker for people with EB (doctors, nurses, dieticians,…). It's about the difficulties to decide what is "the best" to do – and about the question, who is the one who decides what it means: "the best". Treatment of EB requires – much depending on the EB-type - many therapeutical measures, not only wound care but also nutrition therapy, physiotherapy, oral hygiene, medication, psychological measures and more. All these measures need their time and all of them are certainly important. But to try to do everything that is required is hardly possible. There would be nearly no time left for the opportunity to experience the many things that life has to offer and, even with many disease-associated limitations, to be able to lead to a happy and fruitful life. Health care workers and people with EB and their families need to prioritize accordingly, they need open eyes and ears to catch critical moments, but they also must be encouraged to accept imperfectness. Treatment decisions have to be made, treatment must be as simple as possible and nevertheless effective. Some measures must be put off, but of course we have to avoid harmful consequences. In EB – especially in the more severe forms – this is challenging and tricky. It is not possible to discuss all recurrent problems at full length, but some examples will show the dimension of the difficulties we have to face, but also some approaches for making it easier to make good and manageable treatment decisions.

Peter Adriaenssens

Prof. dr. Peter Adriaenssens is a child and adolescent psychiatrist. In 1987 the University Hospital of the Catholic University at Leuven (Belgium) took him on to start up the Confidential Center for Child Abuse & Neglect. He has been the centre's director since 1995. Between 1993 and 2000 he was also Head of the Department of Child and Adolescent Psychiatry at the University Hospital Gasthuisberg, Leuven, where he is now Head of Liaison & Forensic Child Psychiatry. Since 1994 he has been professor of Child Psychiatry attached to the Medical Faculty of the Catholic University at Leuven. He is family therapist and family therapy trainer at the Center for Marital, Family and Sex Therapy (KU Leuven).
He publishes regularly and leads workshops and training sessions on social child psychiatry, child abuse, family therapy and trauma.

"How do families cope with a child with EB?"

EB can be considered as a traumatic experience in early childhood, characterized by chronic developmentally adverse traumatic events, occurring within the child's caregiving system. Research demonstrated how this kind of complex trauma impaires the regulatory capacities of children in the affective, cognitive, relational, behavioural field. It dysregulates the biological stress system. Understanding the underlying neuropsychiatric deficits and their behavioural manifestations is essential because of the broad implications for the development of these children, and their daily quality of life. We recognise similar mechanisms in the parents. We discuss the diagnosis and clinical findings, and implications for family guidance and support of the child with EB.

Elizabeth Pillay

Liz Pillay is currently EB Nurse Consultant and Team Leader for the Adult Nursing Team DebRA UK – Time in Post 14 years.
She has extensive experience in hospital and community nursing over 30 years. She is a qualified Nurse, Midwife and Health Visitor with additional qualifications in wound management and counseling.

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Kate Moss

is a psychoanalytic psychotherapist working within the adult EB service at St. Thomas' Hospital in London.
Since 2006 she has been developing 'talking treatments' for those people with EB who may find them helpful.
Outside the hospital, she works with adults in the general population who seek psychotherapy.

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Emma Dures

is a health psychology researcher from the University of the West of England, Bristol (UK). She is currently working on a 3 year DebRA funded project to explore the psychological and social impact of EB on the daily lives of adults with the condition, their family members, and the professionals involved in their treatment and care. It is hoped that the research will lead to recommendations for support materials and interventions.

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