Highlights from the 2nd Nordic Epidermolysis Bullosa Symposium in Stockholm, April 2005

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Three years ago the first Nordic Epidermolysis Bullosa (EB) meeting took place in Oslo. It was arranged by the DEBRA Norway in collaboration with the Nordic patient organization for EB and health professionals involved in all aspects of the care of EB patients. The meeting was a great success and it was decided to continue this acitivity on a 3-year-interval basis. Thus in April this year a second EB meeting took place in Stockholm and gathered about 170 participants mainly from Scandinavia but also from other European countries (and even from Costa Rica and Chile!).

Among the participants were dermatologists, plastic surgeons, ophthalmologists, pediatricians, dentists & oral surgeons, dermatologic & pediatric nurses, dieticians, psychologists, social workers, and occupational & physical therapists. The program comprised plenary lectures, workshops and a guided interview of 3 patients with different forms of EB (see issue No. 2, 2005 of Forum for abstracts).

The topics of the opening lectures ranged from the history of EB in Sweden to current understanding and classification of EB into the simplex, junctional and dystrophic subtypes. The historic role played by the Swedish paediatrician Gillis Herlitz, who described 7 cases of congenital lethal EB in 1932, was high-lighted (Fig. 1). Sixty years later, Herlitz junctional EB was shown to be due to a deficiency of laminin 5. Due to a genetic founder effect in northern Scandinavia, a relative high incidence of Herlitz EB is seen in Sweden and Norway (Table I). We were told by a paediatrician from Umeå University hospital that on average one newborn child per year with Herlitz EB is treated in their intensive care unit. Unfortunately, the prognosis is pessima; the medium survival time is only 7–10 months with no child surviving more than 3 years.

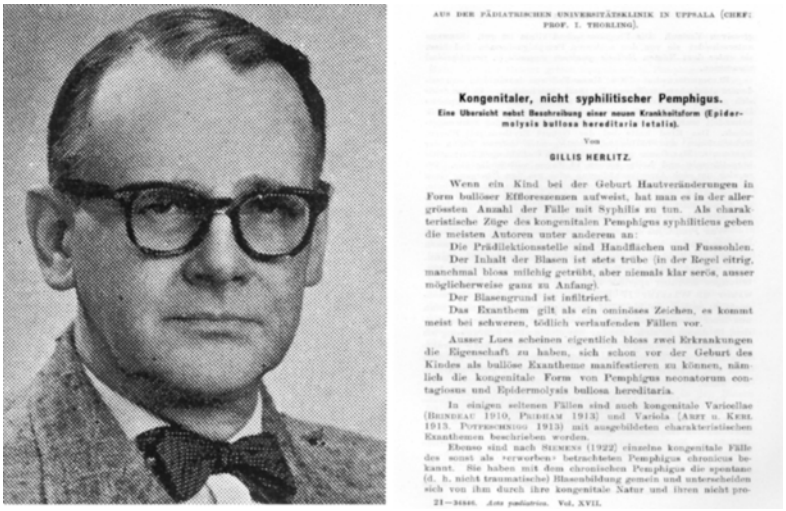

Fig. 1 Gillis Herlitz (1903-82) and the front page of his famous publication in Acta Paediatrica from 1932.

Recessive dystrophic EB (RDEB), the other very severe form of the disease, is due to collagen 7 deficiency. This genetic trait is less common than that of Herlitz EB but since the patients usually survive into adulthood, the prevalence of RDEB is higher. However the severely crippled

No. of EB families in the Nordic countries
(Kero, Acta D-V 1984; Gedde-Dahl, Scand Univ Books 1971 and Acta D-V 2002)

	(Total)	EBS	JEB	D-DEB	R-DEB
Sweden	(93)	13	53	9	18
Norway	(80)	30	27	13	10
Finland	(37)	24	3	8	2
Denmark	(19)	9	4	3	3
Total	(229)	76	87	33	33

Table I: *Estimated number of families affected with epidermolysis bullosa (EB) in the Nordic countries. EBS: EB simplex; JEB: Junctionl (Herlitz) EB; D-DEB: dominant dystrophic EB; R-DEB: recessive dystrophic EB. Blue colour denotes EB subtypes with distinct geographic distribution.*

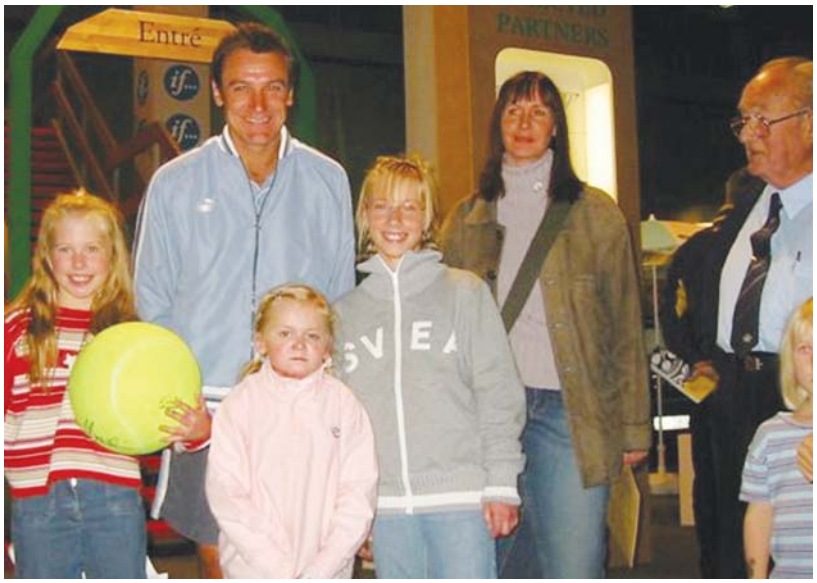

Fig. 2. Malin Netz (organizer) and Sven Wittbold (far right; chairman of DEBRA Sweden and initiator of the meeting who sadly died last year) standing together with children of DEBRA and Mats Wilander who kindly donated the benefits of a tennis show in Stockholm, December 2003, to the meeting.

RDEB patients usually succumb to skin cancer, infections and malnutrition while in their 30–40ies.

The simplex types of EB are essentially due to dominant mutations in keratin 5/14. The phenotype is usually milder and many patients are presumably misdiagnosed as "unspecific foot blisters". Yet, a proper diagnosis is helpful especially in cases with painful plantar hyperkeratosis and nail dystrophy, which can mimic pachonychia congenital and may respond to local botulinum toxin injections to prevent a sweat-induced deterioration.

Among the invited lecturers at the meeting were *Prof. Robin Eady* (London), *Prof. Leena Bruckner-Tuderman* (Freiburg) and *Prof Marcel Jonkman* (Groningen), all of whom are leaders of specialized centers for the diagnosis and treatment of blistering skin diseases in Europe (Figs. 3, 4). They all emphasized the advantage of a multiprofessional team in the treatment EB. A good example is the "The EB-Haus" (head: *Prof Johann Bauer*), which is a newly erected building in Salzburg made possible by private donations exquisitely designated for EB research and patient care. All these centres are part of a

Fig. 3. Prof Leena Bruckner-Tuderman (in red dress) talking to Finish colleagues during the meeting.

Fig. 4. Prof Marcel Jonkman (left) talking to Prof Johann Bauer from Salzburg.

nascent EU-network for diagnosis and therapy of genodermatoses (GENESKIN), headed in Rome by *Prof Gianna Zambruno* and with Uppsala University as the Scandinavian collaborator.

The focus of the meeting was on therapy of EB, with special emphasis on plastic surgery for reconstructing hands with syndactylia and for removing skin cancers (*Dr Bryan Mayou*, London), oral surgery & dentistry for prevention of mouth problems (*Drs Nina Skogedal*, Oslo and *Mats Jontell*, Gothenburg), and eye surgery for replacing scarring tissue in the conjungtivae to prevent visual problems (*Dr Gabor Koranyi*, Stockholm). Wound dressings and anaesthetic procedures in children with EB were highlighted as well as nutritional problems especially in RDEB patients. In addition to all these medical aspects, the psychological and social consequences of EB and various possibilities to interventions were also covered in lectures, posters and workshops.

Alas, despite recent progress in EB therapy, there is still no curative treatment in sight, but the latest development in gene therapy was reviewed by *Prof Bruckner-Tudermann*. In the two concluding talks, *Kris Aaseth* of DEBRA Norway first exemplified the importance of patient organisations and then *Dr Tobias Gedde-Dahl Jr*, the nestor of Scandinavian EB research, gave his personal views and memories in a lecture entitled "A life-time story of EB".

The organising committee consisted of *Malin Netz* (chairman), *Heidi Silseth, Carl-Fredrik Wahlgren, Gabor Koranyi, Gerd Wohlin, Bitte Ahlborg, Kristina Gustafsson-Bonnier, Christina Eklund, Birgitta Schiött and Anders Vahlquist*.

More information about EB can be found on the internet:
www.internationalebforum.org
www.debra-sweden.org
www.sos.se/smkh

Course in Skin Allergy and Inflammation

A postgraduate course in Skin Allergy and Inflammation has been held at the Department of Dermatology, Karolinska University Hospital, Solna, December 13–17, 2004. The course was focusing on the skin barrier, atopic eczema, contact eczema, psoriasis and the mast cell.

The course leaders were professor Magnus Lindberg (left), professor Klas Nordlind (middle) and assoc. professor Lena Lundeberg (right).