Date: August 10, 2007

Program

First World Conference on Ichthyosis
August 31 – September 2, 2007

Münster, Germany

Organized by
Network for Ichthyoses and related keratinization disorders (NIRK)
together with
Selbsthilfe Ichthyose e.V.
and
EU-Coordination Action GENESKIN

Contacts: H. Traupe, Münster, Email: traupeh@mednet.uni-muenster.de
B. Willis, Münster, Email: brigitte.willis@ukmuenster.de
Barbara Kleinow, Email: abcr.kleinow@t-online.de
Geske Wehr, Email: diewehrs@t-online.de

Location:

Lecture Hall
Department of Dermatology
University Hospital
Von Esmarch-Str. 58
48149 Münster
Germany

Friday, August 31, 2007

8:30 **Opening of the conference**

Welcome and greetings by

Prof. E. Schlatter, Dean for Research

Prof. T. Luger, Head of Department of Dermatology

B. Kleinow, Selbsthilfe Ichthyose e.V.

Prof. H. Traupe, Speaker of NIRK

Workshop on clinical diversity and diagnostic standardization

Chair: E. Sprecher and H. Traupe

9:00 D. Metze, Münster

Histopathology of ichthyoses: Clues for diagnostic standardization

9:20 I. Hausser, Heidelberg

Ultrastructural characterization of lamellar ichthyosis: A tool for diagnostic standardization

9:35 H. Verst, Münster

The data base behind the NIRK register: a secure tool for genotype/phenotype analysis

9:50 V. Oji, Münster

Classification of congenital ichthyosis

10:10 M. Raghunath, Singapore

Congenital Ichthyosis in South East Asia

10.25 **Panel discussion: How do we name autosomal recessive nonbullous congenital ichthyosis?**

Panel: E. Sprecher, P. Steijlen, H. Shimizu, A. Vahlquist, H. Traupe

10:40 **Coffee break**

Keratinization disorders and keratins

Chair: P. Steijlen, A. Vahlquist

11:15 I. Hausser, Heidelberg

Ultrastructure of keratin disorders: What do they have in common?

11:25 M. Arin, Köln

Recent advances in keratin disorders

11:45 E. Sprecher, Haifa

Naegeli-Franceschetti-Jadassohn Sydrome: a Keratin Disease

12:05 P.M. Steijlen, Maastricht

Epidermolytic palmoplantar keratoderma with “tono tubular” keratin

12:25 **Panel discussion: Epidermolytic keratinization disorders: how should we call them?**

Panel: P. Steijlen, E. Sprecher, D. Metze, I. Haußer

12:40 **Lunch break**

Molecular advances in epidermal differentiation

Chair: K.H. Grzeschik, D. Kelsell

14:30 D. Kelsell, London

Role of connexin isoforms for epidermal differentiation and wound healing

14:50 L. Bruckner-Tuderman, Freiburg

Role of kindlin in human disease and keratinocyte motility

15:10 M. Guerrin, Toulouse

Granular keratinocytes transcriptome: Identification and characterisation of new differentiation markers

15:30 K.H. Grzeschik, Marburg

Molecular basis of focal dermal hypoplasia

15:50 **Coffee break**

17:15 **Bus transfer for the reception and dinner**

GOP Restaurant and Vaudeville, Münster

Saturday, September 1, 2007

Recent advances in gene mapping and in lipid genes

Chair: J. Fischer, H.C. Hennies

9:00 J. Fischer, Paris
Mapping genes for nonbullous autosomal recessive congenital ichthyosis: What we know today

9:20 H.C. Hennies, Köln
Functional understanding of mutations in congenital ichthyosis

9:40 P. Krieg, Heidelberg
12R – Lipoxygenase Deficiency impairs Skin Barrier Function

10:00 G. Schmitz, Regensburg
Apolipoprotein E and lipid traffic within keratinocytes

10:20 H. Shimizu, Sapporo
What can we learn from Harlequin ichthyosis?

10:40 E. O'Toole, London
In vitro models for harlequin ichthyosis

10:55 **Coffee break**

11:20 R. Happle, Marburg
The CHILD syndrome revisited: the clinical perspective

11:35 A. König, Marburg
Functional understanding of NSDHL mutations

European and international perspective

Chair: R. Happle, H. Shimizu

11:50 G. Zambruno, Rome EU coordination action GENESKIN
Purpose, structure and achievements of GENESKIN

12:10 I. Zwoch, Bonn
Orphan diseases and the European Union – what patients and scientists may expect

12:30 M. Schmuth, San Francisco
Structur and aims of Foundation for Ichthyosis and Related Skin Types

12.45 **Lunch break**

Joint workshop together with Selbsthilfe Ichthyose e.V. and European network of self support groups for ichthyosis

Therapy of ichthyosis: a challenge in daily practice

Chair: G. Wehr, M. Arin

14:00 A. Vahlquist, Uppsala
Introduction to the topic: therapy of ichthyosis/general principles and substances

14:20 M.L. Preil, Bad Salzschlirf
Management of ichthyosis: The TOMESA experience

14:40 A.M. van Steensel, Maastricht
Our experience with RAMBAs in treatment of congenital ichthyosis

14:55 A. Vahlquist, Uppsala
Results of an ongoing study with Liarozol for lamellar ichthyosis

Topical treatment/the patient perspective

Chair: G. Wehr, F. Minelli

15:10 G. Wehr, Kürten
The experience from Germany

15:25 J. Devidts, Belgium
The experience from Belgium

15:40 F. Minelli, Italy
The experience from Italy with special focus on the scalps

16:10 M. Sandström/M. Olsson, Sveden
What can be done for palms and soles

16.25 **Panel discussion and questions for experts**

Chair: G. Wehr, F. Minelle, J. Devidts, A.M. van Steensel, M.L. Preil, A. Vahlquist, H. Traupe

16:45 **Coffee break**

Experimental therapies

Chair: J.A. McGrath, D. Roop

17:15 M. Braun-Falco, Freiburg
Gene therapy for keratinization disorders: what is the current state?

17:35 J. Chen, D. Roop, Denver
Oligonucleotide therapy for keratin disorders

17:55 H. Traupe, Münster
Enzyme replacement therapy of lamellar ichthyosis: the current state

18:15 J. A. McGrath, London
Cell therapy approaches: the example of Epidermolysis bullosa

18:35 End of program for the day
Evening at individual disposal

Sunday, September 2, 2007

Proteases and keratinization disorders

Chair: A. Taïeb and P. Hachem

9:00 P. Hachem, Brussels
Importance of serine proteases for epidermal differentiation

9:20 A. Taïeb, Bordeaux
Insights into Pathogenesis of Ichthyosis in Trichothiodystrophy Syndromes

9:40 A. Hovnanian, Toulouse
Towards functional understanding of Netherton syndrome

10:00 A. Ishida-Yamamoto,
Distinct intracellular transport for different epidermal lamellar body molecules

10:20 **Coffee break**

Ichthyoses and the cornified envelope

Chair: M. Paulsson

10:45 M. Schmuth, San Francisco
How do abnormalities in brick constituents cause barrier abnormalities?

11:05 S. Weidinger, München
Genetics of epithelial barrier integrity in atopic diseases

11:25 M. Paulsson, Köln
Transglutaminase-3 deficient mice: a subtle skin phenotype

11:45 WK Jacyk, Pretoria
Bathing suit ichthyosis, the South African experience

12:05 K. Aufenvenne, Münster
Towards functional understanding of bathing suit ichthyosis

12:25 B. Ahvazi, Bethesda
Modelling of transglutaminase-1 and transglutaminase-3: what can we predict?

End of the conference