

EU FP6 GENESKIN
Individual Training Course
Epidermolysis Bullosa
Groningen

June 27-28, 2006



Coordinator: prof. dr M.F. Jonkman (UMC Groningen)
Programme organizer: Ir Marjon Pasmooij
Social organizer: Drs Marieke Bolling

Registration and information: Mrs Anneke Hoekstra
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Location: Onderwijscentrum (lokaal 15, 18 and Rode Zaal)
UMCG

Aim: Epidermolysis bullosa (EB) are genetic conditions that are characterized by blistering and erosions of the skin and mucous membranes after trauma mostly starting at birth. The past 15 years we have witnessed dramatic advances in our understanding of the components involved in cell-extracellular matrix and cell-cell attachment of keratinocytes that may cause EB. More than 20 phenotypes are recognized and so far 10 genes were identified.

In the Center for Blistering Diseases in the Netherlands, located at the University Medical Center in Groningen, more than 250 patients with EB from the Netherlands (and Belgium) were fully registered. From each patient (family) history and clinical presentation were meticulously recorded, skin biopsies of intact and bullous skin were taken for immunofluorescence and electron microscopy, and DNA was isolated from peripheral blood cells on a routine basis. Moreover, for scientific research techniques have been developed to analyse DNA and RNA from skin biopsies, and to visualise aberrant EB molecules from keratinocyte cell cultures.

This two-day training programme is ment for medical doctors and graduates who wish to know more of laboratory diagnostics and investigations in EB. The morning session of the first day will be plenary and focus on laboratory diagnosis. In the afternoon the participants will caroussel in small groups along 30 min workshops where specific laboratory techniques will be introduced and discussed. In the morning of the second day genetic analysis will be the focus, closed by a quiz with interactive cases to be solved by the participants. During the minisymposium in the afternoon the participants will be given more insight in the topic of 'natural gene therapy' and lessons we learn from this phenomenon for future cell and gene therapy. The scientific programme closes with the defense and official promotion ceremony of Marjon Pasmooij in the Academieggebouw in the historic center of Groningen.

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Programme day 1

TUESDAY JUNE 27, 2006. PATIENTS, PROTEINS AND TISSUES

09:00–09:00	Prof. Marcel Jonkman	Welcome
09:10–10:00	Prof. Marcel Jonkman	Clinical phenotypes and classification
10:00–10:15	Dr Marcelus de Jong	How to come to a perfect IF picture?
<i>10:15–10:30</i>		<i>Coffee</i>
10:30–10:50	Prof. Marcel Jonkman	Immunofluorescence antigen mapping
10:50–11:20	Prof. Marcel Jonkman	Electron microscopic analysis
11:20–11:40	Ir Marjon Pasmooij	Additional value of laser dissection microscopy and mRNA analysis
11:40–12:00	Dr Hendri Pas	Visualisation of aberrant EB molecules
<i>12:00–12:15</i>		<i>Photograph</i>
<i>12:15–13:30</i>		<i>Lunch</i>
13:30–16:30	Gonnie Meijer Janny Zuiderveen	Workshop: Immunofluorescence staining techniques
13:30–16:30	Prof. Marcel Jonkman	Workshop: Immunofluorescence antigen mapping
13:30–16:30	Tjasso Blokzijl Hans van Rossum	Workshop: Laser Microdissection Microscopy
13:30–16:30	Dr Gilles Diercks Hilda Keuning	Workshop: Electron microscopy of EB
<i>13:30–16:30</i>		<i>Thee</i>
13:30–16:30	Miranda Nijenhuis	Workshop: Cell culture of EB keratinocytes
13:30–16:30	Dr Hendri Pas Guus Kloosterhuis	Workshop: Immunoblot of EB keratinocyte extracts
16:30–17:30	Prof. Marcel Jonkman	Translational research: the quest from patient to gene

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Programme day 2

WEDNESDAY JUNE 28, 2006. GENES AND CODING

09:00–09:45 Dr Hans Scheffer DNA mutation detection and interpretation

09:45–10:15 *Coffee*

10:15–10:45 Prof. Irwin Mclean In silico mining

10:45–11:30 Drs Marieke Bolling EB cases quiz

11:30–12:30 *Lunch*

Minisymposium: Revertant mosaicism in human genetic diseases

12:30–13:00 Prof. Rudolf Happle Introduction and mechanisms

13:00–13:30 Ir Marjon Pasmooij Natural gene therapy: random or directed?

13:30–14:00 Prof. Hans Joenje Selection advantage of revertant cells

14:00–14:30 Prof. Irwin McLean RNAi therapy for keratin disorders - applying the lessons taught by revertant mutations

Promotion ceremony and social events

16:15–17:30 Dr Ir Pasmooij Defense of thesis
“Genotyping of unusual phenotypes in epidermolysis bullosa”

17:30–18:30 *Reception Marjon, Academiegebouw*

19:00–22:00 *Diner course participants*

22:00–24:00 *Party Marjon, Christiaans, Oude Kijk in 't Jatstraat 53*