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13th International Postgraduate Course on
Lysosomal Storage Diseases

Nierstein (Mainz), Germany
June 1 - 6, 2014



Arrangement by Prof. Michael Beck,
Pediatric Department
Johannes Gutenberg-University of Mainz, Germany

Supported by an educational grant from
Shire HGT, a business unit of Shire plc
IMS is providing logistic support for the course.



13th International Postgraduate Course on Lysosomal Storage Disorders (LSDs)

Applicants should be physicians with some years of clinical experience, should be seeing patients and have an interest in LSDs.

Applicants with active research projects will have priority.

The number of participants will be limited to approximately twenty five.

The faculty consists of experienced lecturers in different specialities.

The following topics will be addressed during the course:

- Cell biology of lysosomes
- Epidemiology
- Genetics
- Diagnosis
- Fabry Disease
- Gaucher Disease
- MPS syndromes

- Ceroid-ipofuscinoses
- Other LSDs
- Patient Organisations

A good command of the English language is necessary for active participation.

Questions regarding the scientific content of the course should be addressed to:

Professor Michael Beck
Pediatric Department, Johannes Gutenberg-
University of Mainz
michael.beck@unimedizin-mainz.de

Questions regarding logistics can be addressed to:

IMS GmbH, Sabine Michels
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FACULTY

Prof. Michael Beck
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Medical Genetics Service,
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Dr. Eugen Mengel
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Dr. Nicole Muschol
Children's Hospital
University of Hamburg,
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Prof. Ans van der Ploeg
Center for Lysosomal and Metabolic
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Prof. Paul Saftig
Biochemical Institute,
Christian-Albrechts-Universität Kiel,
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Prof. Konrad Sandhoff
LIMES, c/o Kekulé-Institut f.
Organische Chemie und Biochemie
Rheinische Friedrich-Wilhelms-
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Prof. M. Scarpa, MD, PhD
Dept. of Paediatrics,
University of Padova, ITALY

Dr. Marie T. Vanier
Laboratoire Fondation Gillet Mérieux,
Centre Hospitalier Lyon-Sud,
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PD Catharina Whybra
Pediatric Department,
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Johannes Gutenberg-University of
Mainz, GERMANY

13th International Postgraduate Course on Lysosomal Storage Diseases Nierstein (Mainz) June 1 - 6, 2014

Sunday, June 1

Arrivals – all participants and lecturers will be met at the Frankfurt airport. Dinner will be arranged at the hotel.

Monday, June 2

08.15 - 8.45 Course introduction
M. Beck
General overview of LSDs

08.45 - 09.30 Cell biology of lysosomes
V. Gieselmann

09.30 - 10.15 Pathophysiology in LSDs
A. van der Ploeg

10.15 - 10.30 Coffee break

10.30 - 11.15 Genetics of LSDs
A. Gal

11.15 - 12.00 Defects of the lysosomal membrane
P. Saftig

12.00 - 13.15 LUNCH

13.15 - 14.15 Ceroid-Lipofuscinosis
H.H. Goebel

14.15 - 15.15 The blood brain barrier in LSDs
D. Begley

15.15 - 15.30 Coffee break

15.30 - 17.45 Group work: Case reports
The next day all cases will be presented

18.30 – Welcome Dinner

Tuesday, June 3

08.30 - 10.00 Sphingolipids, sphingolipidoses
and the lipid phase problem
K. Sandhoff

10.00 - 11.00 Fabry disease
N. Karabul

11.00 - 11.15 Coffee break

11.15 - 12.15 Gaucher disease (non-neuronopathic form):
Clinical manifestation and treatment
E. Mengel

12.15 - 13.15 LUNCH

13.15 - 14.30 Sphingolipidoses
M. Vanier

14.30 - 15.15 Stem cell transplantation in lysosomal storage
disorders
N. Muschol

15.15 - 15.45 Coffee break

15.45 - 17.30 Presentation and discussion of
case reports from group work
5 min for each case !

17.30 - 17.45 Coffee break

17.45 - 19.30 Case reports cont.

20.00 Dinner

Wednesday, June 4

08.30 - 09.15 CNS manifestation of Gaucher disease
E. Mengel

9.15 - 10.00 Leukodystrophies
M. Vanier

10.00 - 10.15 Coffee break

10.15 - 11.15 Neurodegeneration in LSDs -
Treatment today and tomorrow
M. Scarpa

11.15 - 12.00 Pompe disease
A. van der Ploeg

12.00 - 12.45 Pre- and postnatal presentation of LSDs
C. Whybra

13.00 Lunch
Afternoon at free disposal
(Optional participation at excursion)

Thursday, June 5

08.30 - 10.00 Mucopolysaccharidoses (MPS) and
glycoproteinosis
R. Giugliani

10.00 - 10.15 Coffee break

10.15 - 11.30 Treatment of mucopolysaccharidoses
J. Hennermann

11.30 - 12.15 Acid lipase deficiency - Wolman's disease
and Cholesterol Ester Storage Disorder
M. Beck

12.30 - 13.45 LUNCH

13.45 - 14.30 Principles for laboratory diagnosis in LSDs
M. Vanier, M. Beck

14.30 - 15.00 Newborn screening
R. Giugliani

15.00 - 15.45 Cardiac manifestations in LSDs
C. Kampmann

15.45 - 16.00 Coffee break

16.00 - 17.30 Role of patient support groups in LSDs
C. Lavery

19.00 - FAREWELL DINNER

Friday, June 6

Departures
Optional visit to Villa Metabolica/Department of Pediatrics in
Mainz (09.00-11.00) before departure

Application form

13th International Postgraduate Course on
Lysosomal Storage Diseases
June 1 – 6, 2014

Name

Position

Department

Hospital

Hospital address (for correspondence)

Phone

Fax

Email (please print)

Short description of your education/experience

Years of training after medical school

Clinical research

Laboratory research

Career intentions

Please give a short description of why you would like to attend this course

Please give a brief outline of what your five minutes case report will be about

The application form should be sent – **not later than May 1, 2014** – to:

IMS GmbH · Sabine Michels
Langenbeckstraße 1, D-55131 Mainz,
Tel. +49 6131/17-6552, Fax +49 6131/17-6608
ims@um-mainz.de

Applicants will be informed by Professor Beck about the outcome before May 11, 2014.

