

15th International Postgraduate Course on **Lysosomal Storage Disorders**

Nierstein (Mainz), Germany
June 05 - June 10, 2016



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Shire HGT, a business unit of Shire plc
IMS is providing logistic support for the course.



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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 and pathophysiology of lysosomes
- ✓ Genetics
- ✓ Diagnosis and Treatment
- ✓ Fabry Disease
- ✓ Gaucher Disease

- ✓ MPS syndromes
- ✓ Pompe disease
- ✓ Leukodystrophies
- ✓ Other LSDs
- ✓ Patients' Organisations

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

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Questions regarding logistics can be addressed to:

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GERMANY

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Nierstein (Mainz) June 05 - June 10, 2016

Sunday, June 05

Arrival

Monday, June 06

- 9.00 – 9.10 Welcome and Introduction
Julia B. Hennermann
- 9.10 – 10.10 Cellular pathophysiology of LSDs
Volkmar Gieselmann
- 10.10 – 11.00 Defects of lysosomal membrane proteins
Paul Saftig
- 11.00 – 11.15 Coffee break
- 11.15 – 12.30 Genetic principles of LSDs: A practical
session on Mendelian genetics and pedigree
analysis
Andreas Gal
- 12.30 – 14.00 Lunch
- 14.00 – 15.00 Mucopolysaccharidoses and Mucopolidoses
Roberto Giugliani
- 15.00 – 15.30 Coffee break
- 15.30 – 18.00 Group work: Case reports
ALL
- 19.00 Welcome Dinner

Tuesday, June 07

- 9.00 – 10.00 Sphingolipids, sphingolipidoses and
the lipid phase problem
Konrad Sandhoff
- 10.00 – 10.45 Niemann Pick diseases
Marie T. Vanier
- 10.45 – 11.00 Coffee break
- 11.00 – 11.45 Gaucher disease
Eugen Mengel
- 11.45 – 12.30 Fabry disease
Laila Arash-Kaps
- 12.30 – 13.45 Lunch
- 13.45 – 14.30 Acid lipase deficiency
Simon Jones
- 14.30 – 15.15 Pompe disease
Ans van der Ploeg
- 15.15 – 15.45 Eye manifestations in LSDs
Susanne Pitz
- 15.45 – 16.00 Coffee break
- 16.00 – 17.30 Presentation and discussion of case reports
ALL
- 17.30 – 17.45 Coffee break
- 17.45 – 19.00 Presentation and discussion of case reports
ALL
- 20.00 Dinner

Wednesday, June 08

- 9.00 – 10.00 The blood brain barrier and LSDs
David J. Begley
- 10.00 – 11.00 Mechanisms of neurodegeneration in LSDs
Maurizio Scarpa
- 11.00 – 11.15 Coffee break
- 11.15 – 12.15 Neuronal Ceroid Lipofuscinoses
Alfred Kohlschütter
- 12.15 – 13.15 Leukodystrophies
Jutta Gärtner
- 13.15 Lunch
Free afternoon

Thursday, June 09

- 9.00 – 9.45 The effectiveness of ERT in different LSDs
Michael Beck
- 9.45 – 10.15 Stem cell transplantation in LSDs
Kurt Ullrich
- 10.15 – 11.00 New treatment options in LSDs
Julia B. Hennermann
- 11.00 – 11.15 Coffee break
- 11.15 – 12.00 Potential interface of lysosomes with
common diseases
Roberto Giugliani
- 12.00 – 12.30 Pre – and postnatal presentation of LSDs
Catharina Whybra-Trümpler
- 12.30 – 14.00 Lunch
- 14.00 – 15.00 Principles of laboratory diagnosis in LSDs
Marie T. Vanier
- 15.00 – 15.30 Neonatal screening for LSDs
Alberto Burlina
- 15.30 – 15.45 Coffee break
- 15.45 – 16.45 The role of patient support groups
Sophie Thomas
- 16.45 – 17.00 Summary
Julia B. Hennermann
- 20.00 Farewell Dinner

Friday, June 10

Departures

Optional visit of the Villa Metabolica/Department of Pediatric and
Adolescent Medicine in Mainz (09.00-11.00) before departure