

17th Postgraduate Course on Lysosomal Storage Disorders

Nierstein (Mainz), Germany
May 30 – June 02, 2018



Programme

Wednesday, 30 May 2018

- 13.00 – 14.00 Arrival/Lunch-Snack
- 14.00 – 14.15 Welcome and introduction
Julia B. Hennermann
- 14.15 – 15.00 LSDs – A clinical overview
Michael Beck
- 15.00 – 16.00 Cellular pathophysiology of LSDs
Volkmar Gieselmann
- 16.00 – 16.30 *Coffee break*
- 16.30 – 17.15 Defects of lysosomal membrane proteins
Paul Saftig
- 17.15 – 17.45 Neonatal screening for LSDs
Alberto Burlina
- 17.45 – 18.30 Principles of laboratory diagnosis in LSDs
Marie T. Vanier
- 19.30 *Welcome Dinner*

Thursday, 31 May 2018

- 8.30 – 9.30 Mucopolysaccharidoses and Mucolipidoses
Roberto Giugliani
- 9.30 – 10.15 Leukodystrophies
Jutta Gärtner
- 10.15 – 10.45 *Coffee break*
- 10.45 – 11.30 Sphingolipids and Gangliosidoses
Konrad Sandhoff
- 11.30 – 12.15 Fabry disease
Dominique Germain
- 12.15 – 13.00 Gaucher disease
Eugen Mengel

Thursday, 31 May 2018

- 13.00 – 14.00 *Lunch*
- 14.00 – 16.00 Workshops/Discussion case reports
All
- 16.00 – 16.30 *Coffee break*
- 16.30 – 17.15 Niemann Pick diseases
Marie T. Vanier
- 17.15 – 18.00 Pompe disease
Ans van der Ploeg
- 18.00 – 18.30 Eye manifestations in selected LSDs
Susanne Pitz
- 19.30 *Meet the Expert – Dinner*

Friday, 1 June 2018

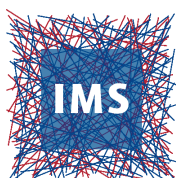
- 8.30 – 9.00 Mechanisms of neurodegeneration in LSDs
Kurt Ullrich
- 9.00 – 09.45 The blood brain barrier and LSDs
David J. Begley
- 09.45 – 10.15 *Coffee break*
- 10.15 – 11.00 Neuronal Ceroid Lipofuscinoses
Angela Schulz
- 11.00 – 11.30 Mannosidosis and selected Oligosaccharidoses
Julia B. Hennermann
- 11.30 – 12.00 Acid lipase deficiency
Simon Jones
- 12.00 – 13.00 *Lunch*
- 13.00 – 13.45 Enzyme replacement therapy in LSDs
Anna Tytki-Szymańska
- 13.45 – 14.30 Stem cell transplantation in LSDs
Rob Wynn

- 14.30 – 15.00 New treatment options in LSDs
Julia B. Hennermann
- Network afternoon*

20.00 *Farewell Dinner*

Saturday, 2 Jun 2018

- 8.30 – 9.00 Prenatal presentation of LSDs
Roseline Froissart
- 9.00 – 9.30 Radiological presentation of Mucopolysaccharidoses and Mucolipidoses
Esmee Oussoren
- 9.30 – 10.00 Orthopedic management of LSDs
Blanka Link
- 10.00 – 10.30 *Coffee break*
- 10.30 – 11.30 Genetic principles of LSDs: A practical session on Mendelian genetics and pedigree analysis
Andreas Gal
- 11.30 – 12.00 The role of patient support groups
Deborah Cavell
- 12.30 – 13.20 Presentation of Workshop results
All
- 13.20 – 13.30 Summary and Farewell
Julia B. Hennermann
- 13.30 *Lunch-Snack / Departure*



Gesellschaft für akademischen
Forschungstransfer mbH

Application to Sabine Michels

ims@um-mainz.de | www.ims-gmbh-mainz.de
Deadline April 30th, 2018



Shire Human Genetic Therapies
is a business unit of Sire plc



BEST WESTERN Wein – und Parkhotel Nierstein

An der Kaiserlinde 1 · 55283 Nierstein

Telefon: +49 6133/508 0 · Telefax: +49 6133/508 333

Info@weinhotel.bestwestern.de

www.weinhotel.bestwestern.de

