

# Meeting Agenda

24<sup>TH</sup> ESGLD  
Workshop and Graduate Course  
7-11 September 2022  
Lancaster UK



European Study Group  
on Lysosomal Diseases



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## **GRADUATE COURSE PROGRAMME**

### **DAY 1 - Wednesday 7<sup>th</sup> September 2022**

**12:30 – 14:00 Registration and Lunch – Lancaster University Management School Atrium**

**ALL Graduate course lectures will take place in Management School Lecture Theatre 14**

**14:00 – 14:15**

Introductory remarks (Brian Bigger)

**14:15 – 15:00**

Lysosome Biogenesis/Biosynthesis of enzymes (Thomas Braulke)

**15:00 – 15:45**

Autophagy (David Rubinsztein)

**15:45 – 16:30**

HOPS complex (Jan van der Beek)

**16:30 – 17:00**

Coffee Break and Refreshments (Management School Atrium)

**17:00 – 17:45**

Channels and Transporters (Bruno Gasnier)

**17:45 – 18:30**

Lipid degradation and lipid storage diseases (Hans Aerts)

**18:30 – 19:15**

Lysosomal Leukodystrophies (Volkmar Gieselmann)

**19:15 – 19:30**

Designation of flash-talk discussion leaders (Management School Atrium)

**20:00**

Dinner (Marketplace) **Students will need to prepare their flash talks over dinner.**

## **GRADUATE COURSE PROGRAMME**

### **DAY 2 - Thursday 8<sup>th</sup> September 2022**

**Breakfast 07:00 – 08:15 (Marketplace)**

**All Graduate course lectures will take place in Management School Lecture Theatre 14**

**08:30 – 09:15**

Misfolding in lysosomal diseases (Mia Horowitz)

**09:15 – 10:00**

Neuronal Ceroid Lipofuscinosis (Angela Schulz)

**10:00 – 10:45**

Mucopolysaccharidoses (Nicole Muschol)

**10:45 – 11:15**

Coffee break and Refreshments (Management School Atrium)

**11:15 – 12:00**

Lysosomal Membrane proteins (Markus Damme)

**12:00 – 12:45**

Current therapies for LSDs (Simon Jones)

**12:45 – 13:30**

Novel Therapies for LSDs (Brian Bigger)

**13:30 – 14:30**

Lunch (Management School Atrium)

**14:30 – 17:00 - FLASH TALKS BY GRADUATE STUDENTS AND POSTDOCS**

**(Management School Lecture Theatre 14)**

Group exercise. Graduate course students will be asked to prepare 1-3 slides (4-6 minute talk) on a current scientific topic over dinner on 7/09/2022. Flash talks will be delivered during this session.

## **SCIENTIFIC PROGRAMME**

**DAY 1 - Thursday 8<sup>th</sup> September 2022**

**14:00 – 18:00**

Registration (Management School Atrium)

**All sessions will take place in Management School Lecture Theatre 18**

**18:00 – 18:15**

Opening ceremony – Welcome address

**18:15 – 18:30**

Flash talk awards

**18:30 – 19:30**

**KEYNOTE CONFERENCE 1 (45 minutes talk + 15 minutes questions)**

**Overview of Gene Therapy for Lysosomal Diseases (Simon Jones)**

Chairs: Volkmar Gieselmann, Nicole Muschol

**20:00**

Get together and dinner (Barkers)

## SCIENTIFIC PROGRAMME

DAY 2 - Friday 9<sup>th</sup> September 2022

Breakfast 07:00 – 08:15 (Marketplace or Hotel)

### SESSION 1: Lysosome function

All sessions will take place in Management School Lecture Theatre 18

Chairs: Markus Damme, Bruno Gasnier

**08:30 – 09:30 - KEYNOTE CONFERENCE 2 (45 minutes talk + 15 minutes questions)**

**LYSET - a key protein in lysosome biogenesis (Prof. Sabrina Jabs)**

**09:30 – 09:50 – O01** Multiple lysosomal proteases knock-out models reveal new insights into their role in lysosomal proteolysis, autophagy, and mTORC1 activity (F. Bleibaum)

**09:50 – 10:10 – O02** The lysosomal enzyme trafficking factor LYSET enables nutrient generation from extracellular proteins (C. Pechincha)

**10:10 – 10:30 – O03** Phosphorylation-independent cytoplasmic retention of acetylated TFEB (A. Laudicina)

**10:30 – Coffee Break and Refreshments** (Management School Atrium)

**11:00 – 11:20 – O04** Role of the Cl<sup>-</sup>/H<sup>+</sup> exchanger CIC-7 in maintaining lysosomal morphology and function (S. Bose)

**11:20 – 11:40 – O05** Crosstalk of class 3 PI3K signaling and lysosomal membrane damage response in LSDs (E. Le Guillou)

**11:40 – 12:00 - O06** Direct control of lysosomal catabolic activity by mTORC1 through regulation of V-ATPase assembly (E. Ratto)

**12:00 – 13:00 - ESGLD General Assembly**

**(For ESGLD members only, Management School Lecture Theatre 18)**

**13:00 – 14:00 – Lunch Break** (Management School Atrium)

**(13:00 – 14:00 ESGLD Board Meeting – lunch in a separate room; Management School Ground Floor Meeting Room)**

**14:15 – 18:00 Networking** – be at meeting point at 14:15, coaches depart at 14:30.

**White Scar Cave coach will depart at 14:00.**

### SESSION 1 (continued): Lysosome Function

**(Management School Lecture Theatre 18)**

Chairs: Thomas Braulke, Mia Horowitz

**18:30 – 19:30 KEYNOTE CONFERENCE 3 (45 minutes talk + 15 minutes questions)**

**The interplay of autophagic processes and lysosomal homeostasis (Dr. Oliver Florey)**

**20:00 - Dinner** (Barkers)

## SCIENTIFIC PROGRAMME

DAY 3 - Saturday 10<sup>th</sup> September 2022

Breakfast 07:00 – 08:15 (Marketplace or Hotel)

### SESSION 2: Therapies

All sessions will take place in Management School Lecture Theatre 18

Chairs: Sandra Alves, Shaun Wood - TBC

- 08:30 – 08:50 - **O07** Improved enzyme replacement therapy of metachromatic leukodystrophy in mice using a superstable mutant of human arylsulfatase A (V. Gieselmann)
- 08:50 – 09:10 - **O08** A cell-based therapy for Pompe disease: transplantation of ex-vivo expanded muscle regenerative cells improves muscle function and lysosomal pathology (G. Schaaf)
- 09:10 – 09:30 - **O09** Development and validation of a GMP stem cell gene therapy manufacturing process for Mucopolysaccharidosis II (MPSII) (S. Ellison)
- 09:30 – 09:50 – **O10** A novel Adeno-associated viral gene therapy treatment protocol in sheep for use in MPSIIIC and other neurological disorders (B. Bigger)
- 09:50 – 10:10 - **O11** Fluoxetine treatment ameliorates the systemic pathology in a novel mouse model of Multiple Sulfatase Deficiency (MSD) (N.C. Sorrentino)
- 10:10 – **Coffee Break and Refreshments** (Management School Atrium)
- 10:40 – 11:00 - **O12** Brain targeting of GAA Promotes Full Correction of Murine Pompe Disease at a Clinically Relevant Dosage of Lentiviral Gene Therapy (F. Catalano)
- 11:00 – 11:20 - **O13** AAV-mediated homology-independent targeted integration leads to supraphysiological levels of lysosomal enzyme secretion from liver and correction of MPS VI in mice (F. Dell'Aquila)
- 11:20 – 11:40 - **O14** Ambroxol as a treatment for neurological manifestations in GD 2 (C. Aries)
- 11:40 – 12:00 - **O27** Endothelial dysfunction in Fabry disease in primary Endothelial Colony Forming Cells (S.Tokic)

12:00 – 13:30 – **Lunch Break** (Management School Atrium)

### SESSION 2 (continued): Therapies

(Management School Lecture Theatre 18)

Chairs: Volkmar Gieselmann, Nicole Muschol

13:30 – 14:30 – **KEYNOTE CONFERENCE 4 (45 minutes talk + 15 minutes questions)**

**Transporting biotherapeutics across the blood-brain-barrier**

**A case example: Brain penetrant enzyme replacement therapy for MPSII (Dr. Annie Arguello)**

- 14:30 – 14:50 - **O16** Developing an iPSC-based neural gene therapy for Sanfilippo syndrome (O. Mandolfo)
- 14:50 – 15:10 - **O17** Drug repurposing approach in Mucopolysaccharidosis type IIIA: the case of Fluoxetine (A. Capuozzo)
- 15:10– 15:40 – **Coffee Break and Refreshments** (Management School Atrium)
- 15:40 – 16:00 - **O18** Improving IDS.fusion peptides for BBB-crossing in Mucopolysaccharidosis type II (R. Searle)

- 16:00 – 16:20 – **O19** Fusion of RVG and gh625 peptide sequences to Iduronate-2-Sulfatase for the Treatment of Mucopolysaccharidosis Type II (S.Wood)
- 16:20 – 16:40 – **O20** An attenuated form of MPS plus due to deficiency of VPS33A and impaired HOPS complex (E. Pavlova)
- 16:40 – 17:00 – **O21** A precision medicine tool for high utilization and quality of individual treatment trials with immunomodulatory drugs in Mucopolysaccharidoses (A. M. Wiesinger)

**17:00 – 19:00 – Attended Poster Session with Refreshments (Management School Atrium 2)**

**20:00 – GALA Dinner Party (Private Dining Room)**

## **SCIENTIFIC PROGRAMME**

**DAY 4 - Sunday 11<sup>th</sup> September 2022**

**Breakfast 07:00 – 08:30** (Marketplace or Hotel)

### **SESSION 3: Lysosomal Pathology**

**All sessions will take place in Management School Lecture Theatre 18**

Chairs: Stuart Ellison, Francisca Coutinho - TBC

- 08:50 – 09:10 - O22** Secondary deficiency of neuraminidase 1 and its contribution to the CNS pathophysiology in neurological mucopolysaccharidoses (A. Pshezhetsky)
- 09:10 – 09:30 - O23** Investigating the role of inflammation in MPS IIIA neuropathology (O. Mandolfo)
- 09:30 – 09:50 - O24** Mice lacking TRPML1 present kidney disease (G. Grieco)
- 09:50- 10:20 – Coffee Break and Refreshments** (Management School Atrium)
- 10:20 – 10:40 - O25** Using Schizosaccharomyces pombe to understand the pathogenesis of CLN3 disease (J. Clemente-Ramos)
- 10:40 – 11:00 - O26** Haplotypic and MicroRNA Profiling in Niemann-Pick type C Portuguese Patients (M. Encarnação)
- 11:00 – 11:20 - O15** Brain-targeted lentiviral haematopoietic stem cell gene therapy for severe Mucopolysaccharidosis Type I: peptide tag optimisation for more efficient delivery across the blood-brain-barrier. (T. Andreou)

**11:20 – 11:30 -** (Scientific committee to discuss prizes)

**11:30 -** Prizes and Photos

**11:45 -** Closing Remarks

**12:00 Lunch Box and Departure** (Management School Atrium)

**12:30 – Coach Transfers to Manchester Airport /Lancaster Railway Station.**

The coach to Manchester Airport will depart at 12:30 from Meeting Point (ETA 14:00)

The shuttle bus to Lancaster Railway Station will depart at 12:30 from Meeting Point.

\*Alcoholic Beverages and networking activities are funded outside of the scientific programme.