European Study Group on Lysosomal Diseases

ESGLD

24th ESGLD Workshop and Graduate course

Lancaster, UK

7–11 September 2022

Graduate course programme

7th – 8th September 2022

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7th September	Day 1 of Graduate Course
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
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
19:30	Dinner

8th September	Day 2 of Graduate Course
07:00 – 08:30	Breakfast
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
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
14:30 – 17:00	Flash talks by graduate students and postdocs

ESGLD2022 Graduate course curriculum

- **Lysosome Biogenesis/Biosynthesis of enzymes and accessory proteins (Thomas Braulke)**
  Rough ER, N-Glycosylation, Processing, Generation M6P, Phosphotransferase and uncoupling enzyme, sorting from Golgi to lysosome, partial secretion, Proteolytic processing of lysosomal enzymes, Mucolipidosis Type II and III. Structure of 46kda and 300 kDa M6P receptors, IGFII/M6P/GlcNac-P-Man binding domains.

- **Autophagy (David Rubinsztein)**
  Regulation of autophagy, selective vs non selective forms of autophagy, termination of autophagy, physiological roles of autophagy during tissues development and maintenance, pharmacological modulation of autophagy.

- **HOPS complex (Jan Van Der Beek)**
  The role of HOPS in endolysosomal biogenesis and fusion, structure of HOPS complex, HOPS subunits, regulation and additional functions. Endocytosis, pathway from CCV>EE >LE >Lysosome, APs, Rabs, ESCRT, Corvet, HOPS, MVBs, role of Ubiquitinilation.

- **Channels and Transporters (Bruno Gasnier)**
  Lysosomal ion channels and small-molecule transporters, and their integration in cell metabolism and signalling.

- **Lipid degradation and lipid storage diseases (Hans Aerts)**
  Sphingolipid metabolism, Ganglioside degradation, enzymes, prosaposin, GM2 activator. Relevant biochemistry of major diseases Gaucher, TS, Fabry, lesser info on NPA, NPB, NPC, Sandhoff.

- **Lysosomal Leukodystrophies (Volkmar Gieselmann)**
  Disease pathology, animal models, current and new treatment options. Krabbe, MLD, MSD (relevant parts of Fabry, Gaucher GM gangliosidosis).

- **Misfolding in lysosomal diseases (Mia Horowitz)**
  misfolding in lysosomal diseases, its impact on LSDs, how can it be validated in animal models, relevant therapies – chaperones, and its importance in the association between a LSD and a neurological disease like Gaucher disease and Parkinson disease.

- **Neuronal Ceroid Lipofuscinosis (Angela Schulz)**
  Batten disease, disease pathology, animal models, current and new treatment options.

- **Mucopolysaccharidoses (Nicole Muschol)**
  Heparan-, Dermatan-, Chondroitin-, Keratan sulfate degradation pathways. Mucopolysaccharidoses, from biochemical defects to phenotype(s).

- **Lysosomal Membrane proteins (Markus Damme)**
  Lysosomal membrane proteins in general, LAMPs, LIMP, Proton pump, (storage) diseases caused by transporter deficiencies, NPC1, lysosomal membrane proteins and their interaction with cytosolic components.

- **Current therapies for LSDs (Simon Jones)**
  Overview of ERT, SRT and chaperone treatments approved and used for many lysosomal diseases. Overview of the use of HSCT in MPSI and beyond and HSC gene therapy in MLD.

- **Novel Therapies for LSDs (Brian Bigger)**
  Overview of therapies in development – What targets should we aim for, What are limitations of existing therapies, Routes of intervention, Brain targeted ERT, Gene therapy, next generation of brain and bone targeted HSC gene therapy, other novel therapies, Combination therapy.