Curriculum ESGLD Graduate Course

Wednesday

Part I Cell biology of lysosomes

1. Biosynthesis of lysosomal enzymes 13.00-13.45 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,

2. Lysosomal membrane proteins 13.45-14.30 Paul Saftig

LAMPs, LIMP, Proton pump, transporters, sialin, cobalamin, cystine, etc and respective diseases, possibly also NPC1 mucolipin and chloride channels

Break 14.30-15.00

3. Delivery routes to lysosomes 15.00-15.45 Volkmar Gieselmann

Endocytosis, pathway from CCV>EE >LE >Lysosome, APs, Rabs, ESCRT, Corvet, HOPS, MVBs, role of Ubiquitinilation,

4. Biogenesis of Lysosomes 15.45-16.30 Andrea Ballabio
Autophagocytosis, Aps, LC, TOR, TFEB

16.30-18.00

Get together of participants and lecturers to coordinate presentation of papers in seminars

Dinner 18.00-19.00

Presentation of seminal papers (historic or current) by participants after 19.00 until ~ 21.00-22.00

Thursday

Part II Biology of lysosomal storage diseases

1. **Lipid degradation and lipid storage diseases 8.30-9.15 Tim Cox**

Ganglioside degradation, enzymes, prosaposin, GM2 activator, sphingolipidosis, NPA, NPB, NPC

2. **Glycosaminoglycan degradation and mucopolysaccaridosis 9.15-10.00 Eduard Paschke**

Heparan-, Dermatan-, Chondroitin-,Keratansulfates, degradation enzymes and respective diseases,

Break 10.00-10.30

3. **Neuronal Ceroid Lipofuscinosis 10.30-11.15 Anu Jalanko**

Diseases and underlying protein defects

4. **Therapy of lysosomal storage diseases 11.15-12.00 Brian Bigger**

ERT in Gaucher and other disease, differentiation Mannose receptor mediated and M6P mediated, problem in neurologic diseases,
Results of selected clinical trials,
Substrate reduction therapy, NBDNM, principle and selected clinical trials,
Molecular chaperones, examples and clinical appications
Gene therapy, Hematopoetic stem cell based and direct application of viral vectors

Lunch 12.00-13.00

5. **Molecular pathogenesis of LSD 13.00-13.45 Emyr Lloyd Evans**

´ Examples: role of autophagy, alterations in Ca++ signalling, alteration in other signalling pathways, lysolipids, alterations in trafficking,

6. **Selected diseases: Gaucher disease 13.45-14.30 Hans Aerts**

Genetics, Biochemistry, types of disease and molecular basis, Therapy,

Break 14.30-14.45

14.45 -16.15 Presentation of selected papers by participants.

16.15-16.30 break

16.30-18.00 Presentation of selected papers by participants