ESGLD
2015 Course
for Graduate Students and Young Post-docs

TIGEM, Pozzuoli, Naples

Wednesday, September 30th

Cell biology of lysosomes

13.00-13.45
Biogenesis of Lysosomes **(Diego Medina)**
Autophagocytosis, Aps, LC, TOR, TFEB

13.45-14.30
Biosynthesis of lysosomal enzymes **(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.

14.30-15.15
Endocytotic delivery route to lysosomes **(Judith Klumperman)**
Endocytosis, pathway from CCV>EE >LE >Lysosome, APs, Rabs, ESCRT, Corvet, HOPS, MVBs, role of Ubiquitinilation.

15.15-16.00
Lysosomal membrane proteins (**Markus Damme and Paul Saftig**)
LAMPs, LIMP, Proton pump, transporters, sialin, cobalamin, cystine, etc. and respective diseases, possibly also NPC1 mucolipin and chloride channels.

16.00-16.30 Coffee Break

16.30-17.15
Lysosomes and cell death **(Paul Saftig and Thomas Braulke)**

17.15-18.00
Transporters and Ion Channels
(Bruno Gasnier and Tobias Stauber)

18.00-18.45
Autophagy **(Carmine Settembre)**
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. Methods: how to monitor autophagy. Trick and tips.

19.00-21.00 *Dinner*

Thursday, October 1st

Biology of lysosomal storage diseases

8.30-9.15
Molecular pathogenesis of LSD **(Emyr Lloyd Evans)**
´ *Focus on lysosomal ion homeostasis.*

9.15-10.00
Lipid degradation and lipid storage diseases **(Tim Cox)**
"The lysosome and sphingolipid metabolism": Historical and emerging rôles; synthesis and degradation of sphingolipids (simple and complex); enzymes, activators and cofactors; sphingolipidoses. Translating biology through exceptions - from human mutants to mechanistic understanding – informative comorbidities.

10.00-10.45
Glycosaminoglycan degradation and mucopolysaccaridoses **(Giancarlo Parenti)**
Heparan-, Dermatan-, Chondroitin-, Keratansulfates degradation pathways. Mucopolysaccharidoses, from biochemical defects to phenotype(s).

10.45-11.15 *Coffee Break*

11.15-12.00
Neuronal Ceroid Lipofuscinosis **(Angela Shultz)**

Diseases and underlying protein defects.

12.00-12.45
Gaucher disease **(Hans Aerts)**
Genetics, biochemistry, types of disease and molecular basis, therapy.

12.45-13.30
Therapy of lysosomal storage diseases **(Brian Bigger)**
ERT in Gaucher and other diseases, 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 applications, gene therapy, hematopoietic stem cell based and direct application of viral vectors.

13.30-14.30 *Lunch*

14.30-17.30 **JOURNAL CLUB**

17.30-18.00 *Coffee Break*