Preliminary Program - Oral Communications

Thursday, September 26, 2013

12.00-18.00		*Registration*	
18.00-19.30		*Get together*	
19.30-19.45		**Welcome address and Opening**	**Paschke E**
19.30-20.30		**Key Note Lecture**	*Chair* ***Ballabio A***
		The Role of TREX1 in Regulation of Lysosomal Biogenesis and Autoimmunity	**Yan N**
20.30		Dinner	

Friday, September 27, 2013

7.30-8.30		*breakfast*	
08.30–10.30		*Scientific Session I* ***Cell Biology of Lysosomes***	*Chair:* ***NN***
8.30-8.50	O01	The role of HOPS complex components in organizing the endo-lysosomal system	**Klumperman J**
8.50-9.10	O02	Mannose 6-phosphate-independent transport mechanisms to lysosomes	**Markmann S**
9.10-9.30	O03	Proteolytic activation of the Golgi-resident GlcNAc-1-phosphotransferase	**Klünder S**
9.30-9.50	O04	Interactions between subunits of the Golgi-resident GlcNAc-1-phosphotransferase complex and analysis of mucolipidosis type II patient mutations	**De Pace R**
9.50-10.10	O05	Characterization of a novel lysosomal sulfatase - arylsulfatase K	**Lübke T**
10.10-10.30	O06	Late stages of EGF-Quantum dot endocytosis	**Belyaeva T**
10.30–11.00		*Break*	
11.00–12.40		*Scientific Session II* ***Cell Biology of Lysosomes***	*Chair:* ***NN***
11.00-11.20	O07	Lysosomal amino acid transporters	**Gasnier B**
11.20-11.40	O08	An extended proteome map of the lysosomal membrane reveals novel potential transporters.	**Verdon Q**
11.40-12.00	O09	Identification of high-affinity ligands of human sialin	**Anne C**
12.00-12.20	O10	Characterization of TMEM106B, a risk factor for FTLD, suggests a lysosomal function	**Lang CM**
12.20-12.40	O11	Emerging roles of a lysosomal intramembrane protease - how SPPL2a controls B cells by cleaving the invariant chain CD74	**Schröder B**
12.40–14.00		***Lunch & Posters***	
14.00-15.20		*Scientific Session III* ***Pathophysiology of Lysosomal Diseases***	*Chair NN*
14.00-14.20	O12	The origins of glucosylsphingosine	**Ferraz MJ**
14.20-14.40	O13	Alterations of the sphingolipidomic profile in a novel mouse model for Farber disease.	**Sabourdy F**

14.40-15.00	O14	Emerging role of microRNAs in CLN3 disease	**Makrypidi G**
15.00-15.20	O15	Analysis of the immune phenotype in CLN3 patients and *Cln3* $^{\Delta ex7/8}$ mice	**Schmidtke C**
15.20-15.50		*Break*	
15.50-17.30		*Scientific Session IV and Posters* ***Pathophysiology of Lysosomal Diseases***	*Chair* ***NN***
15.50-16.10	O16	Systematic approaches towards drafting the Neuronal ceroid lipofuscinosis interactome in the brain.	**Lalowski M**
16.10-16.30	O17	A functional role for excess and non-lysosomal heparan sulphate in Hurler disease	**Bigger B**
16.30-16.50	O18	Role of Oligodendrocyte Precursor Cells in Metachromatic Leukodystrophy	**Brysch K**
16.50-17.10	O20	Identification of a spontaneous novel mutation in the NPC2 gene in a cat affected by Niemann Pick C disease.	**Zampieri S**
17.10-19.30		***Attended Poster Session***	
17.15-18.15		***Business meeting***	
20.00		*Dinner*	

Saturday, September 28, 2013

7.30-8.30		*breakfast*	
8.30–10.30		*Scientific Session V:* ***Lysosomes and Neurodegenerative or Common Diseases***	*Chair NN*
8.30-8.50	O21	A role for GBA2 activity in neuropathology in Niemann-Pick type C	**Marques F**
8.50-9.10	O22	Loss of function of a-synuclein in neurodegenerative lysosomal storage disorders	**Sambri I**
9.10-9.30	O23	The Connection between ERAD, UPR, Gaucher Disease and Parkinson's Disease	**Horowitz M**
9.30-9.50	O24	Disruption of central monoamine/pterin metabolism in lysosomal storage disorders; A mechanistic link to Parkinson's Disease ?	**Heales SJR**
9.50-10.10	O25	Lack of the lysosomal membrane protein, NCU-G1, expression results in liver fibrosis.	**Kong XY**
10.10-10.30	O26	Regulation of insulin signaling by sialylation: can sialidase deficiency contribute to diabetes?	**Pshezhetsky AV**
10.30–11.00		*Break*	
11.00–12.40		*Scientific Session VI* ***New Methods for Diagnosis and Treatment of Lysosomal Storage Diseases***	*Chair NN*
11.00-11.20	O27	An efficient toolkit to detect sequence variations of 283 genes involved in lysosomal-autophagic pathways.	**Ballabio A**
11.20-11.40	O28	Visualization of active glucocerebrosidases in brain	**Herrera Moro Chao D**

11.40-12.00	O29	Substrate reduction therapy of metachromatic leukodystrophy: Development of a high-throughput screening assay for identification of cerebroside sulfotransferase inhibitors.	**Zech I**
12.00-12.20	O30	The MorquioBetter Project: An International Patient Database For Morquio B Disease and Late-Onset GM-1 Gangliosidosis	**Stockler S**
12.20-13.30		***Lunch and Posters***	
13.30-18.30		***Excursion***	
19 00		***Get-Together***	
19.30		***Symposium Dinner***	

Sunday, September 29,2013

7.30-8.50		*breakfast*	
8.50–10.20		*Scientific Session VII* ***Animal Models of Lysosomal Storage Diseases***	*Chair NN*
8.50-9.10	O31	Characterization of murine Arylsulfatase G and Arylsulfatase G knockout mice	**Damme M**
9.10-9.30	O32	Gene disruption of M*fsd8 /Cln7* in mice provides the first animal model for CLN7 disease	**Brandenstein L**
90.30-9.50	O33	Reversal of storage pathology in CLN5- deficient sheep neural cultures with lentiviral gene therapy	**Palmer DN**
9.50 - 10.10	O34	Myeloid Driven Stem Cell Gene Therapy Corrects a Mouse Model of Mucopolysaccharidosis IIIA	**Bigger BW**
10.10–10.40		*Break*	
10.40- 12.10		*Scientific Session VIII* ***Treatment of Lysosomal Storage Diseases***	*Chair NN*
10.40-11.00	O35	Enzyme therapy of metachromatic leukodystrophy: uptake rates for arylsulfatase A by mouse brain cells depend on the donor and acceptor cell type	**Matzner U**
11.00-11.20	O36	Intravenous administration of an AAV9-Hexb vector prolongs lifespan and improves phenotype in Sandhoff mice	**Niemir N**
11.20-11.40	O37	Combined pharmacological chaperone therapy and enzyme replacement therapy in patients with Pompe disease	**Parenti G**
11.40-12.00	O38	10 years therapy with Agalsidase A: Prevention of renal insuffiency in men with timely initiation of ERT	**Arash-Kaps L.**
12.00- 12.30		Young Scientist's award	**ESGLD Board**
12.30-12.50		20th ESGLD Workshop	**NN**
12.50-13.00		Closing remarks	**Paschke E**
13.00 - 15.00		Lunch and departure	

Preliminary Program - Posters

Cell Biology of Lysosomes		
P01	LIMP-2 and Acid β-glucosidase (GCase) trafficking.	**Malini E**
P02	Heptahelical protein PQLC2 is a lysosomal cationic amino acid exporter underlying the action of cysteamine in cystinosis therapy	**Leray X**
P03	Transport of lysosomal enzymes in Congenital Disorders of Glycosylation	**Floßdorf T**
P04	Lysosomes, cellular regulation of autophagy and its interaction with degenerative cellular programs	**Pupyshev AV**
P05	Heptahelical protein PQLC2 is a lysosomal cationic amino acid exporter underlying the action of cysteamine in cystinosis therapy	**Leray X**
P06	Characterization of the mammalian CORVET complex	**Jonker C**
P07	Is the Bovine lysosomal phospholipase B-like protein an amidase?	**Heikinheimo P**
Pathophysiology of Lysosomal Diseases		
P08	Molecular analyses of genes involved in mannose 6-phosphate independent trafficking	**Alves S**
P09	Molecular characterization of Mucolipidosis types II and III: Genotype-phenotype correlations and prenatal diagnosis.	**Sabourdy F**
P10	Poloxamer 407-induced lipidosis in mice and involvement of lysosomes	**Korolenko TA**
P11	Characterization of TATA-less HGSNAT promotor	**Richtrová E**
P12	Molecular characterization of new GALNS genetic alterations in Morquio A patients.	**Tonin R**
P13	Lymphocytes in Mucopolysaccharidosis type VI patients	**Macedo MF**
P14	Understanding neuronal dysfunction in MPS VII using human iPSC-derived cells	**Creyssels S**
P15	Behavioral, neurochemical, and physiological abnormalities in the male Fabry knockout mice.	**Rodrigues LG**
P16	Generation of induced pluripotent stem cells derived from a mouse model of Fabry disease and their differentiation into selected cell types	**Rybová J**
P17	Functional characterization of the common c.-32-13T>G mutation of GAA gene: identification of potential therapeutic agents.	**Dardis A**
P18	Point mutations in the CTSA gene causing Galactosialidosis	**Caciotti A**
P19	Genotyping of 31 patients with Metachromatic Leukodystrophy: Eight new pathologic mutations in Arylsulfatase A gene characterized	**Böhringer J**
P20	Sphingolipidoses patients present disease specific imbalances in iNKT cells.	**Macedo MF**
P21	G_{M2} -gangliosidosis in British Jacob sheep	**Jackson M**
P22	Niemann-Pick type C disease – a model to study the role of cholesterol in the pathogenesis of Alzheimer's disease	**Hecimovic S**
P23	Antisense-induced Exon Skipping for the correction of mis-spliced mRNAs in Fabry disease.	**Ferri L**
P24	Identification of lysosomal cathepsins in Gaucher spleen	**Oussoren SV**

P25	SCARB2 mutations as modifiers in Gaucher disease: the wrong enzyme at the wrong place	**Coutinho MF**
P26	Characterization of NPC1 expression on mRNA and protein level in a cohort of Niemann-Pick type C disease patients.	**Majer F**
P27	Glucocerebrosidase Activities and Gaucher disease, Potential neuronal glial differences.	**Burke DG**
P28	The Molecular Basis Underlying the Association between Gaucher disease and Parkinson's disease	**Maor G**
P29	Lysosomal enzyme activities in different brain regions from subjects affected by PD, PDD and DLB	**Paciotti S**
Diagnosis of Lysosomal Storage Diseases		
P30	Stability of lysosomal enzymes in human cerebrospinal fluid	**Paciotti S**
P31	Glycosaminoglycan levels for newborn screening in MPS I, MPS II and MPS III	**van Vlies N**
P32	An algorithm to predict phenotypic severity in mucopolysaccharidosis type I in the first month of life	**SDK Kingma**
P33	Cytogenomic evaluation of chromosome rearrangements by array-CGH in the IDS gene region in MPS II patients	**Giugliani R**
P34	Overview of the first 1,000 patients identified by the MPS BRAZIL NETWORK	**Giugliani R**
P35	Proteomic Analysis Reveals Possible Mechanisms for Cognitive Impairment in Mucopolysaccharidosis type I mice	**Giugliani R**
P36	New assays for measurement of X-inactivation skewing in patients with Fabry disease and other X-linked disorders	**Musalkova D**
P37	Spatial detection of Gb3Cer isotypes by MS imaging in mouse model of Fabry disease	**R. Dobrovolný**
P38	A systematic review on screening for Fabry disease: prevalence of individuals with genetic variants of unknown significance.	**van der Tol L**
P39	Fabry disease: report and limits of laboratory diagnosis in 47 families	**Piraud M**
P40	Consensus recommendation on Fabry disease diagnosis in adult patients with left ventricular hypertrophy	**Smid BE**
P41	Chitotriosidase activity in mice of different strains and in specific pathogen free (SPF) mice, effect of chitin	**Korolenko TA**
P42	Biomarker responses to eliglustat, an investigational oral substrate reduction therapy for Gaucher Disease type 1	**Peterschmitt MJ**
P43	Acid lipase deficiency: pitfalls along the way to diagnosis	**Burda P**
P44	The development of a Cathepsin D assay for the diagnosis of CLN10	**Burke DG**
P45	Flow cytometry allows detection of extremely rare populations of LAMP2 deficient peripheral white blood cells - implications for diagnostics of Danon disease.	**Majer F**
P46	Inherited NeuRoMetabolic Diseases Information Network (InNerMeD-I-network)	**D'Avanzo F**
P47	NPC BRAZIL NETWORK: Overview of the 2010-2012 period (first 3 years of operation)	**Giugliani R**
P48	Early severe pulmonary involvement in a patient with Niemann-Pick disease type C due to a mutation in the NPC2 gene: description of a new case.	**Ciana G**

P49	High throughput screening of microRNAs that regulate TFEB, a master controller of the lysosomal-autophagic pathway	**Davidson NP**

Clinical Symptoms and Treatment of Lysosomal Storage Diseases

P50	Ultrasound Examination Reveals Typical Alterations In Joints of Mucopolysaccharidosis IV (M. Morquio) Patients	**Karabul N**
P51	Mucolipidosis: update and challenges	**Selmi R**
P52	Niemann-Pick type B disease: a radiological classification of pulmonary involvement	**Brondani G**
P53	Neuroinflammation is a new therapeutic target in mouse models of metachromatic leukodystrophy	**Stein A**
P54	Long term follow up of the Mainz cohort of GD type 3 patients	**Mengel E**
P55	Biphosphonate treatment in a patient affected by MPS IVA with osteoporotic phenotype	**Di Natale P**
P56	Hematopoietic stem cell transplantation (HSCT) in different lysosomal storage disorders: A swiss experience	**Link B**
P57	Neonatal bone marrow transplantation strategy for the cure of Hurler disease skeletal phenotype	**Pievani A**
P58	Long term outcome of haematopoietic stem cell transplantation in MPSI-H patients	**Boncimino A**
P59	Sap B deficiency and successful bone marrow transplantation: biochemical vs clinical insight	**Kuchař L**
P60	Treatment effect of Coenzyme Q10 and of an antioxidant cocktail in fibroblasts of patients with Sanfilippo Disease	**Gort L**
P61	Enzyme replacement and substrate reduction therapy for Gaucher disease- a Cochrane systematic review	**Deroma L**
P62	Efficacy of long-term velaglucerase alfa on haematological and visceral parameters in treatment-naïve patients with type 1 Gaucher disease	**Zimran A**
P63	A Nanoparticle-based approach for drug delivery to the brain in Lysosomal Storage Disorders	**Salvalaio M**
P64	Six-months enzyme replacement therapy's efficacy in a non-classic infantile Pompe disease form: our experience in a 2-year-old affected child	**Scelsa V**
P65	Evaluation of stop-codon readthrough therapy in several lysosomal storage diseases	**Gómez-Grau M**
P66	Corrective GUSB transfer to the canine mucopolysaccharidosis VII brain	**Kremer EJ**
P67	Neural Progenitors isolated from multiple sulfatase deficiency as a model to study neurodegeneration and validate pharmacological approaches in Lysosomal Storage Disorders	**Medina DL**