20TH ESGLD Workshop

Pozzuoli (Naples), Italy

1 - 4 October, 2015

Scientific Programme

Day 1 – THURSDAY, October 1st

18.00	Introductory Remarks - **Graciana Diez Roux**
18:15	Introduction to Keynote Lecturer - **Andrea Ballabio**
18.30	Keynote Lecture "**Surprise Speaker**" **Don't Miss it!** 
19.30	Transfer from TIGEM to Hotel Gli Dei
20.00	Dinner at Hotel Gli Dei
	Transfer to the Hotels after dinner

Day 2 – FRIDAY, October 2nd

8.00		Transfer from the Hotels to TIGEM
8.30 - 9.50		Scientific session ***Biology of Lysosomes*** Chairs: **Bruno Gasnier**, **Volkmar Gieselman**
O - 6	**De Pace R**	Subunit interactions of mucolipidosis II and III-related hexameric GlcNAc-1-phosphotransferase complex
O - 10	**Kissing S**	The V-ATPase: In vivo analysis of its contribution to (auto-)phagosome fusion and mTORC1 signalling
O - 12	**Lloyd-Evans E**	The mucolipidosis type IV disease associated protein TRPML1 is a late endosomal pH regulated Ca2+ channel whose function is altered in Alzheimer's disease but not Niemann-Pick C
O - 13	**Makrypidi G**	Dysfunction of Cln3 protein impairs lysosomal and endocytic homeostasis
9.50 - 10.20		Coffee break
10.20 - 11.20		Scientific session ***Biology of Lysosomes*** Chairs: **Paul Saftig**, **Hans Aerts**
O - 15	**Massa Lopez D**	Characterization of MFSD1 - a new lysosomal membrane protein - and its physiological role in the mouse
O - 16	**Medina D**	Lysosomal calcium signaling regulates autophagy via calcineurin and TFEB
O - 18	**Pan X**	Desialylation of brain gangliosides by endosomal neuraminidases 3 and 4 is essential for neuronal function
11.20 - 12.00		Invited Speaker **Antony Galione** University of Oxford, UK ***"Two-pore channels and lysosomal calcium release"*** introduced by **Thomas Braulke**
12.00 - 13.30		Lunch
13.30 - 14.30		ESGLD Business meeting for members only

14.30 - 16.30	**POSTER SESSION I** Biology of Lysosomes, Disease Mechanisms, Animal Models
16.30 - 18.10	Scientific session ***Biology of Lysosomes*** Chairs: **Eeva-Liisa Eskelinen**, **Judith Klumperman**
O - 25 Schröder B	The role of the lysosomal intramembrane protease SPPL2a in the regulation of immune cell signaling
O - 28 Staiano L	New roles of OCRL in lysosomal function and autophagy
O - 29 Thelen M	Changes in the lysosomal proteome after MG132 treatment
O - 30 Verdon Q	Identification and biological significance of a lysosomal glutamine tranporter
O - 21 Polishchuk RS	Lysosomal system as a hub for copper sensing and homeostasis in health and disease
18.10 - 18.50	Invited Speaker **Timothy Cox** University of Cambridge, UK ***''Gaucher disease: an exceptional source of treasure''*** introduced by **Tony Futerman**
19.30	**GALA Dinner** at Villa Di Livia
	Transfer to the Hotels after dinner

Day 3 – SATURDAY, October 3rd

8.00	Transfer from the Hotels to TIGEM
8.30 - 9.10	Invited Speaker **David Rubinsztein** University of Cambridge, UK ***"Autophagy and neurodegeneration"*** introduced by **Carmine Settembre**
9.10 - 11.10	Coffee break and **POSTER SESSION II** Diagnosis, Therapy
11.10 - 12.30	Scientific session ***Disease Mechanisms and Animal Models*** Chairs: **Jonathan Cooper**, **Giancarlo Parenti**
O - 7 Horowitz M	The contribution of mutant gba alleles to the development of Parkinson's disease in carriers of Gaucher disease mutations
O - 14 Markmann S	Craniofacial and oral anomalies in mucolipidosis II knock-in mice
O - 19 Parker H	Mucopolysaccharidosis IIIA Storage Substrate Drives an Innate Immune Neuro-inflammatory Response
O - 22 Pshezhetsky A	Synaptic Defects in Mucopolysaccharidosis IIIC Mouse
12.30 - 14.00	Lunch
14.00 - 17.30	Excursions

17.30 - 19.10		Scientific session ***Disease Mechanisms and Animal Models*** Chairs: **Generoso Andria**, **Pietro Strisciuglio**
O - 27	**Sikora J**	Acid ceramidase deficiency (Farber disease) – behavioral, biochemical and cellular CNS pathologies in Asah1P361R/P361R mice
O - 20	**Platt N**	Differential Inflammasome Dysregulation in Lysosomal Storage Diseases
O - 23	**Raimo S**	A physiological role of TFEB in LSDs
O - 3	**Colaco A**	Tangier Disease and Niemann-Pick type C disease: mechanistic convergence and shared therapeutic targets
O - 11	**Klumperman J**	Mutations in Vps41, encoding a regulator of lysosomal fusion events, cause a Parkinson-like phenotype and reduction in cellular LAMP levels
19.10		Transfer from TIGEM to Hotel Gli Dei
20.00		Dinner at Hotel Gli Dei
		Transfer to the Hotels after dinner

Day 4 – SUNDAY, October 4th

8.00		Transfer from the Hotels to TIGEM
8.30 - 9.30		Scientific session ***Disease Mechanisms and Animal Models*** Chairs: **Angela Schultz**, **Paola Di Natale**
O - 2	**Brandestein L**	Lysosomal dysfunction and impaired autophagy in a novel mouse model deficient for the lysosomal membrane protein Cln7
O - 5	**Damme M**	Pld3 – A new lysosomal protein implicated in Alzheimer's disease
O - 24	**Sambri I**	Lysosomal dysfunction disrupts presynaptic maintenance in neurodegenerative diseases through a α-synuclein- and CSPα-dependent pathway
9.30 - 10.10		Invited Speaker **Marja Jäättelä** Cell Death and Metabolism, Danish Cancer Society Research Center, DK ***"Sphingomyelin regulates autophagosome biogenesis and lysosomal membrane stability"*** introduced by **Frances Platt**
10.10 - 10.40		Coffee break
10.40 - 12.40		Scientific session ***Therapy*** Chairs: **Brian Bigger**, **Alberto Auricchio**
O - 1	**Bartolomeo R**	Identification of molecular targets for the treatment of the skeletal phenotype in Lysosomal Storage Disorders
O - 26	**Serafini M**	Neonatal bone marrow transplantation prevents bone pathology in a mouse model of mucopolysaccharidosis type I
O - 4	**Cooper J**	Defining spinal cord neuropathology in a mouse model of Infantile Neuronal Ceroid Lipofuscinosis (INCL) and assessing the efficacy of intrathecal Enzyme Replacement Therapy (ERT)
O - 8	**Gatto F**	AAV-mediated TFEB overexpression alleviates skeletal muscle pathology and motor impairment in a Pompe disease mouse model

O - 9	**Kallemeijn W**	In vivo studies on retaining glycosidases with fluorescent activity-based probes
O - 17	**Mitchell N**	Viral-mediated gene therapy prevents disease development in ovine models of neuronal ceroid lipofuscinosis

12.40 - 13.10 Closing remarks - **Andrea Ballabio**

13.10 - 13.30 Young Scientist's Award

13.30 - 15.00 Lunch and Departure

POSTER SESSION I - Biology of Lysosomes, Disease Mechanisms, Animal Models

Presenting Author	*Title*	*Poster N.*
Bembi	Is peripheral neuropathy a constitutive element of the non-neurological phenotype of Gaucher disease?	P-3
Buttgereit	Analysis of the accelerating mutation S228F in H+/Cl- exchanger ClC-7	P-5
Cachon-Gonzalez	Ripk3 deficiency does not modify the natural course of murine Globoid cell leukodystrophy or Sandhoff disease	P-6
Catarzi	Gaucher disease: a new silent mutation causing the complete exon 4 skipping of GBA gene	P-7
Ceccarini	Cerebrospinal fluid lysosomal enzymes in Parkinson's Disease	P-8
Clark	Lipid-based vectors for RNA interference affect lysosomal function and alter the expression of the lysosomal NPC1 protein	P-9
Clark	Lysosomal involvement in the pathogenesis of Huntington's disease	P-10
Dardis	TDP-43 pathology in Niemann pick type C	P-13
De Pasquale	Cardiac disease in the murine model of mucopolysaccharidosis IIIB	P-17
De Risi	Autism-like behavioral symptoms are associated to striatal dopamine system dysregulation in Mucopolysaccharidosis type III-A	P-73
Dobrovolny	MPS II in the three-year-old girl. Biochemical and molecular findings and cell biology study, Generation of iPSC model	P-18
Dobrovolny	Crispr/Cas9 Generation Of Ipsc Model Of Fabry And Schindler Disease	P-19
Eskelinen	LAMP1 and LAMP2 influence the intracellular lipid homeostasis	P-21
Hetmańczyk	Gene expression study in Niemann-Pick type C fibroblasts: preliminary results of a pilot study	P-29
Huebecker	Nervous system involvement in the Fabry mouse	P-31
Krambeck	Mannose 6-phosphate-independent transport of lysosomal enzymes in liver cells	P-34
Kuchar	Elevated plasma lyso-sphingomyelin as a biomarker for Niemann-Pick A/B disease	P-35
Kuo	Activity-based profiling of GH31 alpha-glucosidases	P-36
Kytidou	Production of active human lysosomal galactosidases in Nicotiana benthamiana plants	P-37
Lecommandeur	Consequences of RIP3 deficiency on the lipid profile of a mouse model of Krabbe disease	P-38
Levade	Blood and plasma ceramide levels in mice and patients with Farber disease	P-39
Liao	Evaluating the Role of G Protein PEG Encapsulation of Enzyme in a MPS I Mouse Model	P-40
Lloyd-Evans	Characterisation of the NPC1 protein as a lipid and heavy metal transporter using bioinformatics and mutation analysis	P-41
Macedo	The two tales of glycosphingolipids in Natural Killer T lymphocyte activation	P-42

Presenting Author	*Title*	*Poster N.*
Maguire	NPC1, a Lysosomal Zn2+ Transporting RND Permease	P-44
Maguire	Generation of a Nieman-Pick type C1 zebrafish colony for the purpouses of phenotyping and drug-screening	P-45
Musalkova	Niemann-Pick disease type C: complex analysis of the effect of mutations in NPC1 gene on the mRNA and protein levels	P-47
Newman	Impaired Fc-gamma and complement receptor mediated phagocytosis in Niemann-Pick disease Type C (NPC) macrophages	P-48
Nicoli	Impaired Drug Metabolism in Niemann-Pick Disease type C1 Mice	P-49
Ossouren	Lysosomal cathepsins and glucocerebrosidase in Gaucher disease	P-51
Palmer	The relevance of the storage of subunit c of ATP synthase in different forms of Batten disease (NCLs)	P-52
Parenti	Analysis of circulating and tissue-specific micro-RNAs in Pompe disease	P-53
Pupyshev	Osmotic behavior of lysosomes as an index of autophagy in cellular lipid overloading and experimental treatment of early senescence	P-55
Schuster	Stable isotope labelling with amino acids in cell culture (SILAC): A proteomics approach to compare the cell-surface interactomes of alpha-mannosidase and arylsulfatase A	P-59
Soria	Hyperammonemia induces hepatic autophagy and enhancement of autophagic flux improves clearance of ammonia in vivo	P-63
Tasegian	Origin of lysosomal α-mannosidase in cerebrospional fluid	P-64
van Meel	The lysosomal targeting of acid alpha-glucosidase	P-69
von Kleist	Identification and characterization of lysosomal calcium transporters	P-71
Wolf	Targeted disruption of lysosomal α-fucosidase leads to a severe type of fucosidosis in mice	P-72

POSTER SESSION II - Diagnosis, Therapy

Presenting Author	*Title*	*Poster N.*
Amico	Use of an MLPA-based approach for an initial and simultaneous detection of GBA deletions and recombinant alleles in patients affected by Gaucher Disease	P-1
Banfi	Lysoplex: An efficient toolkit to detect DNA sequence variations in the autophagy-lysosomal pathway	P-2
Blomqvist	Validation of an NGS-panel for routine diagnosis of lysosomal and peroxisomal disorders	P-4
Coll	Niemann-Pick type C diagnosis across 20 years. Analysis of the different analytical methods over time and what we have learned	P-11
Coutinho	Mucopolysaccharidoses in Tunisia: a molecular portrait of allelic heterogeneity and consanguinity	P-12
D'Avanzo	Inherited Neurometabolic Diseases Database from InNerMeD-I-network EU project	P-15
D'Avanzo	Search for genetic and epigenetic determinants in MPS VI clinical phenotype: NGS analysis of two monozygotic twins	P-16
Dardis	Novel approaches to rescue normal splicing of GAA transcripts carrying the common c.-32-13T>G mutation	P-14
Ellison	Pre-clinical workup of lentiviral mediated stem cell gene therapy for Mucopolysaccharidosis type IIIA	P-20
Ferla	Combination of low-dose gene therapy and monthly enzyme replacement therapy improves the phenotype of a mouse model of lysosomal storage disease	P-22
Ferri	The challenge of significance of new GLA gene variants: the importance of functional studies	P-23
Fog-Tonnesen	Hsp70-based therapies as clinical candidates for lysosomal storage diseases	P-24
Gieselmann	Effects if cuprizone induced demyelination in a mouse model of Metachromatic Leukodystrophy	P-25
Gleitz	Development of a lentiviral-based gene therapy for Mucopolysaccharidosis II	P-26
Haslett	The complex mechanism of therapeutic action of Miglustat in Niemann-Pick type C disease	P-27
Haslett	Cellular phenotypes of CLN3 disease present new opportunities for phenotypic screening	P-28
Hrebicek	Targeted re-sequencing in the diagnostic work-flow of neurodegenerative metabolic diseases with Mendelian inheritance – a pilot study	P-30
Kirkegaard	Development of Heat Shock Protein based therapies for Lysosomal Storage Diseases	P-32
Korolenko	Intracellular lipid storage syndrome (lipidosis) following prolonged treatment of poloxamer 407 in mice	P-33
Maguire	Use of Ca2+ modulators for the treatment of Niemann-Pick type C disease	P-43

Presenting Author	*Title*	*Poster N.*
Majer	White blood cells in Danon disease diagnostics: efficiency can be demonstrated in families affected by LAMP2 exon-copy number variations and somatic/germinal mosaicism	P-46
O'Leary	Development of an adeno-associated viral mediated gene therapy approach for Mucopolysaccharidosis IIIC	P-50
Piraud	Measurement of lysosphingolipids and their isoforms by lc-ms/ms in plasma, urine and amniotic fluid: application to screening of sphingolipidoses	P-54
Rigon	Enzyme-loaded nanoparticles: a potential therapy for the neurological compartment in Mucopolysaccharidosis type II	P-56
Rouviere	Intravenous AAV9-mediated gene transfer prevents pathology in neonatal Sandhoff mice	P-57
Schuster	Enzyme Replacement Therapy of Metachromatic Leukodystrophy: Nanoparticles fail to deliver Arylsulfatase A across the Blood-Brain Barrier	P-58
Schwartz	Bone Infiltration of Gaucher Disease Evaluated by Bone Marrow Burden Score: evolution after one year of treatment	P-60
Schwartz	Identification of Recombinant Alleles in GBA1 Gene in Patients with Neuronopathic and Non-neuronopathic Gaucher Disease	P-61
Schwartz	Mucolipidosis II and III Alfa/Beta in Brazil: update on the GNPTAB gene analysis	P-62
Tonin	GALNS mRNA aberrant transcripts due to atypical splice site and deep intronic mutations in Morquio A patients	P-65
Tschernutter	Towards a Metabolomic Assay of Monosialoganglioside Turnover in Normal and GM1 Gangliosidosis Fibroblasts with High Resolution Q-Exactive Orbitrap LC/MS	P-66
Turpault	Oral Eliglustat for Gaucher Disease Type 1: Dosing by CYP2D6 Metabolizer Status	P-67
Valenzano	Pharmacogenetics Can Identify Fabry Patients Eligible for Treatment with Migalastat, and Indicate that the Majority of Migalastat-amenable Patients Have Mutations Associated with Classic Disease	P-68
Vilageliu	EXTL2 and EXTL3 inhibition with siRNAs as a promising substrate reduction therapy for Sanfilippo C syndrome	P-70