

Lysosomal Diseases
January 23-28, 2011
Hotel Galvez
Galveston, TX

Agenda

SUNDAY

4:00 pm - 8:00 pm	Arrival and Check-in (Office Closed 6:00 pm - 7:45 pm)
6:00 PM	Dinner
7:30 pm - 7:40 pm	Welcome / Introductory Comments by GRC Site Staff
7:40 pm - 9:30 pm	History and Current Status Chair: Tony Futerman (Weizmann Institute) Discussion Leader: Fran Platt (Oxford University)
7:40 pm - 8:05 pm	Chair's opening comments and overview
8:05 pm - 8:35 pm	Timothy M. Cox (Cambridge University) "Tay-Sachs disease - The Debt of Science to Medicine and the Ghetto"
8:35 pm - 8:45 pm	Discussion
8:40 pm - 9:10 pm	Elizabeth F. Neufeld (UCLA) "Evolving views of lysosomal storage"
9:10 pm - 9:20 pm	Discussion
9:20 pm - 9:30 pm	Closing comments and discussion
9:30 PM	Welcome Party

MONDAY

7:30 am - 8:30 am	Breakfast
8:30 AM	Group Photo
9:00 am - 12:30 pm	Basic Lysosomal Biology Chair: Fred Maxfield (Cornell University) Discussion Leader: Paul Pryor (University of York)
9:00 am - 9:10 am	Chair's overview
9:10 am - 9:35 am	J. Paul Luzio (University of Cambridge) "Lysosome fusion machinery"
9:35 am - 9:45 am	Discussion
9:45 am - 9:55 am	Qian Cai (NINDS, NIH) "Retrograde transport regulates neuronal autophagy-lysosomal function"
9:55 am - 10:00 am	Discussion
10:00 am - 10:30 am	Coffee Break
10:30 am - 10:55 am	Juan S. Bonifacino (NICHD, NIH) "Requirement of Retrograde Transport from Endosomes and the TGN for Lysosome Biogenesis and Function"
10:55 am - 11:05 am	Discussion
11:05 am - 11:15 am	Maaïke S. Pols (University Medical Center Utrecht, the Netherlands) "Implications of HOPS components hVps41 and hVPS39 in LAMP trafficking and lysosome function"
11:15 am - 11:20 am	Discussion
11:20 am - 11:45 am	Antony Galione (University of Oxford) "Lysosomes as NAADP-sensitive calcium stores"

11:45 am - 11:55 am	Discussion
11:55 am - 12:05 pm	Tobias Stauber (Leibniz-Institut für Molekulare Pharmakologie / Max-Delbrück-Centrum für Molekulare Medizin, Berlin) "Luminal chloride accumulation is required for lysosomal function"
12:05 pm - 12:10 pm	Discussion
12:10 pm - 12:30 pm	Closing comments and discussion
12:30 PM	Lunch
1:30 pm - 4:00 pm	Free Time
4:00 pm - 6:00 pm	Poster Session
6:00 PM	Dinner

Relationships to Other Diseases

	Chair: Raffi Schiffmann (Baylor College)
	Discussion Leader: Gregory A. Grabowski (Children's Hospital Medical Center, University of Cincinnati)
7:30 pm - 7:40 pm	Chair's overview
7:40 pm - 8:05 pm	Ellen Sidransky (NIH) "Mutations in Glucocerebrosidase as a Risk Factor for Parkinsonism"
8:05 pm - 8:15 pm	Discussion
8:15 pm - 8:25 pm	Matt Gegg (University College London) "The effect of GBA1 deficiency on mitochondria: implications for Parkinson's disease"
8:25 pm - 8:30 pm	Discussion
8:30 pm - 8:55 pm	Ralph Nixon (NYU Langone Medical Center / Nathan Kline Institute) "Lysosomal System Failure in Alzheimer's Disease"
8:55 pm - 9:05 pm	Discussion
9:05 pm - 9:15 pm	Silva Hecimovic (Rudjer Boskovic Institute, Zagreb, Croatia) "Endocytic traffic jam in Niemann Pick type C disease - a cause of increased Alzheimer's amyloid- β formation"
9:15 pm - 9:20 pm	Discussion
9:20 pm - 9:30 pm	Closing comments and discussion
9:30 PM	Social gathering and poster viewing

TUESDAY

7:30 am - 8:30 am	Breakfast
9:00 am - 12:30 pm	Pathogenic Cascades
	Chair: Steve Walkley (Albert Einstein College of Medicine)
	Discussion Leader: Fran Platt (Oxford University)
9:00 am - 9:10 am	Chair's overview
9:10 am - 9:35 am	Andrea Ballabio (Telethon Institute and Baylor College of Medicine) "Modulation of cellular clearance in lysosomal storage diseases"
9:35 am - 9:45 am	Discussion
9:45 am - 9:55 am	Matthew C. Micsenyi (Albert Einstein College of Medicine) "Autophagy dysfunction in lysosomal disease"
9:55 am - 10:00 am	Discussion
10:00 am - 10:30 am	Coffee Break
10:30 am - 10:55 am	Jon Cooper (King's College London) "The NCLs: a different sort of LSD?"
10:55 am - 11:05 am	Discussion
11:05 am - 11:15 am	Alessandro Fraldi (Telethon Institute of Genetics and Medicine, Naples) "Lysosomal fusion and SNARE function are impaired by cholesterol accumulation in lysosomal storage diseases"
11:15 am - 11:20 am	Discussion

11:20 am - 11:45 am	Alessandra d'Azzo (St. Jude Children's Research Hospital) "Role of NEU1-regulated lysosomal exocytosis in lysosomal storage diseases"
11:45 am - 11:55 am	Discussion
11:55 am - 12:05 pm	Heather Flanagan-Steet (University of Georgia, Athens) "Zebrafish Model of ML-II Provides Insight into the Pathogenesis of Bone and Cartilage Defects"
12:05 pm - 12:10 pm	Discussion
12:10 pm - 12:30 pm	Closing comments and discussion
12:30 PM	Lunch
1:30 pm - 4:00 pm	Free Time
4:00 pm - 6:00 pm	Poster Session
6:00 PM	Dinner
7:30 pm - 9:30 pm	Biomarkers Chair: John Hopwood (University of Adelaide) Discussion Leader: Cynthia Tift (NIH)
7:30 pm - 7:40 pm	Chair's overview
7:40 pm - 8:05 pm	Daniel Ory (Washington University School of Medicine) "Lipid Biomarkers for Niemann-Pick C Disease"
8:05 pm - 8:15 pm	Discussion
8:15 pm - 8:25 pm	William C. Lamanna (University of California, San Diego) "Secondary storage of Dermatan sulfate in Sanfilippo Disease"
8:25 pm - 8:30 pm	Discussion
8:30 pm - 8:55 pm	Johannes Aerts (Academic Medical Center, Amsterdam) " <i>In situ</i> visualization of active glucocerebrosidase molecules: applications for diagnosis and research"
8:55 pm - 9:10 pm	Discussion
9:10 pm - 9:15 pm	Zoltan Lukacs (Hamburg University Medical Center, Hamburg, Germany) "High-risk population screening for Pompe disease using dried blood specimens - Does heterozygosity confer a risk of muscle disease?"
9:15 pm - 9:20 pm	Discussion
9:20 pm - 9:30 pm	Closing comments and discussion
9:30 PM	Social gathering and poster viewing

WEDNESDAY

7:30 am - 8:30 am	Breakfast
9:00 am - 12:30 pm	Defective Lysosomal Transmembrane Proteins Chair: Bruno Gasnier (Paris Descartes University) Discussion Leader: Thomas Braulke (Hamburg University)
9:00 am - 9:10 am	Chair's overview
9:00 am - 9:35 am	Susan A. Slaughaupt (Massachusetts General Hospital) "Mucopolidosis Type IV - Using Phenotype to Guide Functional Studies"
9:35 am - 9:45 am	Discussion
9:45 am - 9:55 am	Rosa Puertollano (NHLBI, NIH) "Mucolin-1 Regulates the Activity of the LPTM Family of Lysosomal Transporters: New Clues For Understanding Mucopolidosis type IV"
9:55 am - 10:00 am	Discussion
10:00 am - 10:30 am	Coffee Break
10:30 am - 10:55 am	Yiannis A. Ioannou (Mount Sinai School of Medicine) "Niemann-Pick C1 and L1 Proteins: Opposing ends with similar functions?"
10:55 am - 11:05 am	Discussion
11:05 am - 11:15 am	Haoting Xu (University of Michigan)

	"Pairing Phosphoinositides with Calcium Ions in Endolysosomal Dynamics"
11:15 am - 11:20 am	Discussion
11:20 am - 11:45 am	Susan Cotman (Massachusetts General Hospital)
	"CLN3p Function and Dysfunction: Clues from Accurate Genetic Models of Juvenile Neuronal Ceroid Lipofuscinosis"
11:45 am - 11:55 am	Discussion
11:55 am - 12:05 pm	Pieter Steenhuis (University Medical Center Hamburg-Eppendorf, Hamburg)
	"Proteolytic cleavage and trafficking of the lysosomal storage disease-related membrane protein CLN7"
12:05 pm - 12:10 pm	Discussion
12:10 pm - 12:30 pm	Closing comments and discussion
12:30 PM	Lunch
1:30 pm - 4:00 pm	Free Time
4:00 pm - 6:00 pm	Poster Session
6:00 PM	Dinner
7:00 pm - 7:30 pm	<u>Business Meeting</u>
	(Nominations for the next Vice Chair; Fill out Conference Evaluation Forms; Discuss future Site & Scheduling preferences; Election of the next Vice Chair)
7:30 pm - 9:30 pm	Experimental Therapies
	Chair: Roscoe Brady (NIH)
	Discussion Leader: Bill Sly (St. Louis University School of Medicine)
7:30 pm - 7:40 pm	Chair's overview
7:40 pm - 8:05 pm	Beverly L. Davidson (University of Iowa)
	"Emerging therapies for lysosomal storage diseases affecting the brain"
8:05 pm - 8:15 pm	Discussion
8:15 pm - 8:40 pm	Jean Michel Heard (Pasteur Institute)
	"Correction of storage lesions and associated cellular disorders by gene therapy in mucopolysaccharidoses"
8:40 pm - 8:50 pm	Discussion
8:50 pm - 9:15 pm	Don Mahuran (University of Toronto)
	"Pharmacological chaperones for late onset GM2 gangliosidosis; from Pyrimethamine to more effective derivatives"
9:15 pm - 9:25 pm	Discussion
9:25 pm - 9:30 pm	Closing comments
9:30 PM	Social gathering and poster viewing

THURSDAY

7:30 am - 8:30 am	Breakfast
9:00 am - 10:45 am	Hot Topics
	Chair: Volkmar Gieselmann (University of Bonn)
	Discussion Leader: Marie Vanier (INSERM)
9:00 am - 9:20 am	Marco Henneke (Göttingen University)
	"RNASET2-deficient cystic leukoencephalopathy: a novel lysosomal storage disorder"
9:20 am - 9:25 am	Discussion
9:25 am - 9:45 am	Miquel Vila (Vall d'Hebron Research Institute)
	"Pathogenic lysosomal membrane permeabilization in Parkinson's disease"
9:45 am - 9:50 am	Discussion
9:50 am - 10:10 am	James Shayman (University of Michigan)
	"Lysosomal phospholipase A2 (PLA2G15) and autoimmunity"
10:10 am - 10:15 am	Discussion
10:15 am - 10:45 am	Coffee Break

10:45 am - 12:30 pm

Poster Talks

Nine short talks, each of 5 minutes in length, will be chosen at the meeting on the basis of those posters which generate the most interest and discussion during the poster sessions.

Discussion Leaders: **Maurizio Scarpa** (University of Padova), **Emyr Lloyd Evans** (University of Cardiff) and **Thierry Levade** (INSERM, Toulouse)
(Speakers TBA)

12:30 PM

Lunch

1:30 pm - 4:00 pm

Free Time

4:00 pm - 6:00 pm

Poster Session

6:00 PM

Dinner

7:30 pm - 9:30 pm

Clinical Trial Design and Outcomes

Chair: **Marc Patterson** (Mayo Clinic)

Discussion Leader: **Greg Pastores** (New York University Medical School)

7:30 pm - 7:40 pm

Chair's overview

7:40 pm - 8:05 pm

Ed Kaye (Genzyme Corp.)

"Clinical Drug Development in Rare Diseases: From Proof of Concept to Approval"

8:05 pm - 8:15 pm

Discussion

8:15 pm - 8:25 pm

Laura Segatori (Rice University, Houston, TX)

"Remodeling protein folding and Ca²⁺ homeostasis in Gaucher's disease patient-derived fibroblasts: a novel mechanism for mutant glucocerebrosidase proteostasis"

8:25 pm - 8:30 pm

Discussion

8:30 pm - 8:55 pm

Forbes D. Porter (National Institutes of Health)

"Biomarker development and clinical aspects of Niemann-Pick Disease, type C"

8:55 pm - 9:00 pm

Discussion

9:00 pm - 9:10 pm

Thomas Kirkegaard (Orphazyme)

"HSP70 and Lysosomal Storage Disorders: Novel Therapeutic Opportunities"

9:10 pm - 9:15 pm

Discussion

9:15 pm - 9:30 pm

Closing comments

9:30 PM

Social gathering and farewell party

FRIDAY

7:30 am - 8:30 am

Breakfast

9:00 AM

Departure