## Agenda

### SUNDAY

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

<table>
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<tr>
<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>7:30 am - 8:30 am</td>
<td>Breakfast</td>
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<tr>
<td>8:30 AM</td>
<td>Group Photo</td>
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<tr>
<td>9:00 am - 12:30 pm</td>
<td><strong>Basic Lysosomal Biology</strong></td>
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<td>Chair: <strong>Fred Maxfield</strong> <em>(Cornell University)</em></td>
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<td>Discussion Leader: <strong>Paul Pryor</strong> <em>(University of York)</em></td>
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<tr>
<td>9:00 am - 9:10 am</td>
<td>Chair's overview</td>
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<td>9:10 am - 9:35 am</td>
<td><strong>J. Paul Luzio</strong> <em>(University of Cambridge)</em></td>
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<td>&quot;Lysosome fusion machinery&quot;</td>
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<td>9:35 am - 9:45 am</td>
<td>Discussion</td>
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<tr>
<td>9:45 am - 9:55 am</td>
<td><strong>Qian Cai</strong> <em>(NINDS, NIH)</em></td>
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<td>&quot;Retrograde transport regulates neuronal autophagy-lysosomal function&quot;</td>
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<tr>
<td>9:55 am - 10:00 am</td>
<td>Discussion</td>
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<tr>
<td>10:00 am - 10:30 am</td>
<td>Coffee Break</td>
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<tr>
<td>10:30 am - 10:55 am</td>
<td><strong>Juan S. Bonifacino</strong> <em>(NICHD, NIH)</em></td>
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<td>&quot;Requirement of Retrograde Transport from Endosomes and the TGN for Lysosome Biogenesis and Function&quot;</td>
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<td>10:55 am - 11:05 am</td>
<td>Discussion</td>
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<td>11:05 am - 11:15 am</td>
<td><strong>Maaike S. Pols</strong> <em>(University Medical Center Utrecht, the Netherlands)</em></td>
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<td>&quot;Implications of HOPS components hVps41 and hVPS39 in LAMP trafficking and lysosome function&quot;</td>
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<td>11:15 am - 11:20 am</td>
<td>Discussion</td>
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<td>11:20 am - 11:45 am</td>
<td><strong>Antony Galione</strong> <em>(University of Oxford)</em></td>
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<td>&quot;Lysosomes as NAADP-sensitive calcium stores&quot;</td>
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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
7:30 pm - 9:30 pm  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
Alessandra d’Azzo (St. Jude Children’s Research Hospital)
"Role of NEU1-regulated lysosomal exocytosis in lysosomal storage diseases"

Heather Flanagan-Steet (University of Georgia, Athens)
"Zebrafish Model of ML-II Provides Insight into the Pathogenesis of Bone and Cartilage Defects"

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)
"In situ 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?"
"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 Business Meeting
(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)

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 Gieselman (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