

**THE GLYCOPROTEINOSES: SECOND INTERNATIONAL WORKSHOP
ON ADVANCES IN PATHOGENESIS AND THERAPY**

**JULY 26-27, 2007
ANN ARBOR, MICHIGAN**

Scientific Conference Day 1

Morning Sessions, July 26

7:30-8:15 Continental Breakfast

8:15-8:30 Welcome and Introductions, Opening Remarks – Goals of the conference, progress since the last conference - Steven Walkley (with additional comments by officials from ISMRD, NINDS, ORD)

8:30-9:15 Plenary Address

- *Cross-Correction: Looking Back, Looking Forward* – **Elizabeth Neufeld**

Session 1: Animal Models, Pathogenesis, Experimental Therapies (Chair, Mark Haskins)

α -Mannosidosis

- 9:15-9:35 *Enzyme replacement therapy for α -Mannosidosis* - **Judith Blanz**
- 9:35-9:55 *α -Mannosidosis in the Guinea Pig – Pathophysiological, behavioral, and treatment issues* - **John Hopwood**

10:00-10:15 Break

α -Mannosidosis (continued)

- 10:15-10:35 *Strategies for translating gene therapy in the brain from rodents to clinical use* - **John Wolfe**
- 10:35-10:55 *Quantitative Imaging of gray matter disease associated with feline α -mannosidosis* – **Charles Vite**

β -Mannosidosis

- 10:55-11:15 *β -Mannosidosis mice – A model for the human disease?* – **Karen Friderici**

Fucosidosis

- 11:15-11:30 *Intrathecal enzyme infusion therapy in canine fucosidosis* – **Gauthami Kondagari**

11:30-12:00 General Discussion

12:00-1:30 Lunch

Afternoon Session, July 26

1:30-2:15 **Special Lecture:** *The Glycoproteinoses: Limited Progress in Molecular Insight still Surpasses Present-day Treatment Efficacy* – **Jules Leroy**

Session 2: Animal Models, Pathogenesis, Experimental Therapies (Chair, John Wolfe)

Galactosialidosis and Sialidosis

- 2:15-2:35 *Molecular pathogenesis of sialidosis and galactosialidosis: Lessons from the animal models* - **Allesandra d'Azzo**

Mucopolipidosis types II and III

- 2:35-2:55 *I-Cell disease (Mucopolipidosis II) in the cat* – **Mark Haskins**

2:55-3:15 Break

- 3:15-3:30 *Exploring the pathogenesis of mucopolipidosis II in zebra fish* – **Richard Street**
- 3:30-3:50 *Molecular analysis of the N-acetylglucosamine-1-phosphotransferase in ML II* – **Thomas Braulke**
- 3:50-4:10 *Molecular analysis of the N-acetylglucosamine-1-phosphotransferase in ML III* – **Stephan Tiede**

4:15-5:00 General Discussion

7:00 ISMRD Reception (all invited – Scientists, Clinicians, Families)

Scientific-Family Conference Day 2

(Joint sessions between Scientific and Family conferences)

7:30-8:00 Continental Breakfast

8:00 -11:00 Session 3: Clinical Issues (Chair, Marc Patterson)

- 8:00-8:20 *Sugar highs – Clinical consequences of glycoprotein accumulation and the need for biomarkers* – **Marc Patterson**
- 8:20-8:40 *The natural course and complications of alpha-mannosidosis* – **Dag Malm**
- 8:40-9:00 *European HUE-MAN Consortium – Therapy update for α -Mannosidosis* – **Christian Friis**
- 9:00-9:20 *Lessons from ML II and ML III - Why Natural History Studies Matter* – **Sara Cathey**
- 9:20-9:40 *Experience with Treatment of Glycoproteinoses and Related Conditions: Markers in and Clinical Outcomes* – **Chester Whitley**
- 9:40-10:00 *Umbilical cord blood transplantation to treat lysosomal storage diseases* – **Joanne Kurtzberg**

Break 10:00-10:15

- 10:15-10:35 *The use of Bone Marrow Transplantation (BMT) for treatment of α -mannosidosis* – **Charles Peters**
- 10:35-10:55 *Can we use iduronidase and/or arylsulfatase B to treat MLII-MLIII by enzyme replacement therapy?* – **Michael Vellard**

11:00 -12:00 Session 4: Scientific Conference Wrap-Up – (Elizabeth Neufeld, Chair)

- *Pathophysiology issues summary* – **Steven Walkley**
- *Treatment issues summary*– **Marc Patterson**
- *Where Do We Go From Here* Panel Discussion (**Elizabeth Neufeld, chair, with John Hopwood, Jules Leroy, Mark Haskins, Alessandra d’Azzo, and Chester Whitley**)

Afternoon Session, July 27 (Family Conference-All Invited)

12:00-1:00 “Working Lunch” – **Welcome to the ISMRD Family Conference**

12:45-1:00 *ISMRD Current and Future Focus* – John Forman, President to introduce FDA.

1:00-1:20 *FDA presentation on Orphan Rare Disease Drug Development – Global Impact*

1:20-1:40 *Relevance of Newborn Screening, Global Perspectives*

1:40-1:45 Question/Answer Session

1:45-2:15 *Therapeutic/Treatment Panel Presentation w/Question & Answer Session*

Dag Malm, PhD Norway – HUEMAN Project, ERT
Charles Peters, PhD U.S./J. Kurtzburg, PhD, U.S.
Jules LeRoy, Prof, PhD Belgium – BMT

2:15-2:30 **Afternoon Break**

2:30-3:15 *Panel Discussion: Therapies, the Family Perspective*, **Marc Patterson** moderator pros/cons of family panel on Enzyme Replacement Therapy and Bone Marrow Transplants
 Question & Answer Included

3:15-3:35 *Surgical & Anesthetic Approach with Affected Patients of Storage Diseases*

3:35-3:45 Question/Answer

3:45-3:50 *Closing of General Session* – Wrap Up/Announcements for Saturday Walk Run

4:00-5:00 **Family Focus Groups**—Disease Specific discussions to include Moderators