AGENDA – DAY 1
Monday, December 7, 2009

7:30 A.M.  REGISTRATION & CONTINENTAL BREAKFAST

8:00 A.M.  Introduction: Overview of the Challenges of Research into Central Nervous System (CNS) Manifestations of Metabolic Diseases
Marc Patterson, M.D. — Mayo Clinic

8:30 A.M.  Patient Perspective
Cynthia Le Mons — National Urea Cycle Disorders Foundation (NUCDF)

8:45 A.M.  Translational Medicine and Rare Disease Research

Effectiveness of interventions in inborn errors of metabolism diseases affecting the CNS
R. Rodney Howell, M.D. — University of Miami, National Institute of Child Health and Human Development (NICHD), National Institutes of Health (NIH)

Brain-directed treatment in Menkes disease
Steven Kaler, M.D. — NICHD, NIH

Is your treatment ready for clinical trials?
Anne Pariser, M.D. — Center for Drug Evaluation and Research (CDER), Food and Drug Administration (FDA)

Panel Discussion
Moderator: Christopher Austin, M.D. — National Human Genome Research Institute (NHGRI), NIH
Marc Walton, M.D., Ph.D. — Office of Translational Science, FDA
Amy Rosenberg, M.D. — Division of Therapeutic Proteins, FDA
Lois Freed, Ph.D. — Division of Neurology Products, FDA
Cynthia Le Mons — NUCDF
Steven Kaler, M.D. — NICHD, NIH

11:00 A.M.  Abstract Presentations & WORKING LUNCH

• Differences in Adaptive Language Functioning in Hurler Syndrome Before and After HCT: A Qualitative Comparison of Treatments and Risk Factors
  Julie B. Eisengart, Ph.D. — University of Minnesota

• Intrathecal Enzyme Replacement for Cognitive Decline in Mucopolysaccharidosis I
  Agnes Chen, M.D. — Los Angeles Biomedical Research Institute at Harbor-UCLA Medical Center
Abstract Presentations (continued)

- White Matter Abnormalities and Cognitive Deficits in Partial OTCD
  Bennett Gertz, M.D. — Department of Neurology, Children’s National Medical Center

12:30 P.M. Assessing Outcomes in CNS Part 1 – Clinical Assessments

**Pediatric movement disorders: Outcome measures**
Jonathan Mink, M.D., Ph.D. — University of Rochester School of Medicine and Dentistry

**Urea cycle disorders: Short- and long-term neurocognitive development**
Lauren Krivitzky, Ph.D. — Children’s National Medical Center

Cognitive and behavioral testing methods
Kendra Bjoraker, Ph.D., L.P. — University of Minnesota

**Issues in longitudinal studies of brain structure and function in small samples**
Elsa Shapiro, Ph.D. — University of Minnesota Medical School

Panel Discussion
Moderator: Marc Patterson, M.D. — Mayo Clinic
Phil Sheridan, M.D. — Division of Neurology Products, FDA
Lauren Krivitzky, Ph.D. — Children’s National Medical Center
Jonathan Mink, M.D., Ph.D. — University of Rochester School of Medicine and Dentistry
Kendra Bjoraker, Ph.D., L.P. — University of Minnesota
Elsa Shapiro, Ph.D. — University of Minnesota Medical School

2:20 P.M. Questions & BREAK

2:35 P.M. Assessing Outcomes in CNS Part 2 – Neuroimaging and Delivery

**Neuroimaging in inborn errors of metabolism**
James Provenzale, M.D. — Duke University School of Medicine, Emory University School of Medicine

**Neuroimaging findings in inborn errors: Survey of findings on standard imaging with emphasis on change over time**
Andrea Grigman, M.D. — Children’s National Medical Center

**CNS magnetic resonance imaging (MRI) spectroscopy in inborn errors of metabolism**
Peter Barker, D.Phil. — Johns Hopkins University School of Medicine

**Tissue specificity/CNS delivery**
Beverly Davidson, Ph.D. — University of Iowa Carver College of Medicine
Assessing Outcomes in CNS Part 2 – Neuroimaging and Delivery (continued)

Panel Discussion
Moderator: Alan P. Koretsky, Ph.D. — National Institute of Neurological Disorders and Stroke (NINDS), NIH  
James Provenzale, M.D. — Duke University School of Medicine, Emory University School of Medicine  
Andrea Gropman, M.D. — Children’s National Medical Center  
Peter Barker, D. Phil. — Johns Hopkins University School of Medicine  
Beverly Davidson, Ph.D. — University of Iowa Carver College of Medicine  
Steven Kaler, M.D. — NICHD, NIH  
Marc Walton, M.D., Ph.D. — Office of Translational Science, FDA

4:30 P.M. Wrap-up
Marc Patterson, M.D. — Mayo Clinic

5:00 P.M. ADJOURN
AGENDA — DAY 2
Tuesday, December 8, 2009

7:30 A.M.  REGISTRATION & CONTINENTAL BREAKFAST

8:00 A.M.  Introduction
Marc Patterson, M.D. — Mayo Clinic

8:15 A.M.  Ethics – Case Presentations and Discussion
Sara Goldkind, M.D., M.A. — Office of the Commissioner, FDA
Robert Nelson, M.D., M.Div., Ph.D. — Office of Pediatric Therapeutics, Office of the Commissioner, FDA

10:15 A.M.  BREAK

10:30 A.M.  Executing Clinical Trials in Rare Diseases: Novel Approaches

Intrathecal and intravenous enzyme replacement therapy combined with hematopoietic stem cell transplantation for Hurler syndrome
Paul J. Orchard, M.D. — University of Minnesota

The story of Lorenzo’s oil for X-ALD: What is known now and any lessons we can take from the history of its study
Gerald Raymond, M.D. — Kennedy Krieger Institute, Johns Hopkins University

Challenges of designing and executing clinical trials in mitochondrial diseases
Bruce H. Cohen, M.D. — Cleveland Clinic

Brain-directed stem cell therapy for neuronal ceroid lipofuscinosis
Robert Steiner, M.D. — Oregon Health & Science University

Panel Discussion
Moderator: Patricia Dickson, M.D. — Harbor-UCLA Medical Center
Paul Orchard, M.D. — University of Minnesota
Robert Steiner, M.D. — Oregon Health & Science University
Gerald Raymond, M.D. — Kennedy Krieger Institute, Johns Hopkins University
Bruce Cohen, M.D. — Cleveland Clinic

12:30 P.M.  Challenges and Future Directions in Rare Disease Research (WORKING LUNCH)
Emil Kakkis, M.D., Ph.D. — Kakkis EveryLife Foundation

The Need for Global Partnerships in Rare Diseases Research
Stephen Groft, Pharm.D. — Office of Rare Diseases Research (ORDR), NIH
Challenges and Future Directions in Rare Disease Research (continued)

Panel Discussion
Moderator: Elizabeth McNeil, M.D. — Office of Orphan Products Development, FDA
Emil Kakkis, M.D., Ph.D. — Kakkis EveryLife Foundation
Stephen Groft, Pharm.D. — ORDR, NIH

3:00 P.M. ADJOURN
Marc Patterson, M.D. — Mayo Clinic