RDD@NIH
Monday, February 28, 2011
Lipsett Amphitheater ● National Institutes of Health ● Bethesda, MD

Agenda

8:00 a.m.  Registration and Continental Breakfast
Lipsett Amphitheater Reception Area

8:30 a.m.  Welcoming Remarks
Stephen C. Groft, Pharm.D. — Director, Office of Rare Diseases Research (ORDR), National Institutes of Health (NIH)

8:45 a.m.  The Role of the NIH Clinical Center in the National Clinical Research Spectrum
John I. Gallin⁰, M.D. — Director, Clinical Center, NIH

9:30 a.m.  Clinical Center Resource: Biomedical Translational Research Information System (BTRIS)
James Cimino, M.D. — Chief, Clinical Center Laboratory for Informatics Development, NIH

10:10 a.m.  Undiagnosed Diseases Program: Will There Always be a Diagnostic Odyssey?
Cynthia Tifft, M.D., Ph.D. — Deputy Clinical Director, National Human Genome Research Institute (NHGRI), NIH

10:30 a.m.  Bench-to-Bedside Lecture #1
WAGR Syndrome: Clinical Characterization and Correlation with Genotype
Joan C. Han, M.D. — Assistant Clinical Investigator, Unit on Metabolism and Neuroendocrinology, National Institute of Child Health and Human Development (NICHD), NIH
Felicitas L. Lacbawan, M.D., F.C.A.P., F.A.C.M.G. — Clinical Professor and Director, Molecular Pathology, State University of New York Downstate Medical Center

11:10 a.m.  Therapeutics for Rare and Neglected Diseases (TRND) Program
Christopher P. Austin, M.D. — Director, NIH Center for Translational Therapeutics (NCTT)

11:30 a.m.  Genetic Testing Registry
Cathy Fomous, Ph.D. — Senior Health Policy Analyst, Office of Biotechnology Activities, NIH

11:50 a.m.  Lunch and Poster Session
The patio (1st floor, South/East atrium) is reserved

1:30 p.m.  Rare Diseases and Translational Science
Francis Collins⁰, M.D., Ph.D. — Director, NIH

¹Rare Disease Day Ribbon presentation to Dr. Gallin by Nicole Boice, Global Genes Project.
²Rare Disease Day Ribbon presentation to Dr. Collins by Nicole Boice, Global Genes Project, and Announcement of Uplifting Athletes’ Rare Disease Champion by Scott Shirley, Uplifting Athletes.
2:00 p.m.  Bench-to-Bedside Lecture #2
Genetics of Inherited Paragangliomas and Gastric Stromal Tumors
Constantine A. Stratakis, M.D., D.M.Sc. — Scientific Director, NICHD, NIH
Su Young Kim, M.D., Ph.D. — Assistant Clinical Investigator, Pediatric Oncology Branch, National Cancer Institute (NCI), NIH

2:40 p.m.  Rare Diseases Clinical Research Network
RDCRN: A Model for Successful Research in Rare Diseases
Jeffrey Krischer, Ph.D. — Professor, Department of Pediatrics, University of South Florida College of Medicine

The Natural History of a Rare Disease-Urea Cycle Disorder
Mark L. Batshaw, M.D. — Chief Academic Officer, Children’s National Medical Center; Professor and Chair, Department of Pediatrics, The George Washington University School of Medicine and Health Sciences

3:15 p.m.  Break

3:30 p.m.  Health Resources and Services Administration (HRSA) Newborn Screening: Service Infrastructure for Rare Disorders
Michele A. Lloyd-Puryear, M.D., Ph.D. — Chief, Genetic Services Branch, HRSA

NICHD Newborn Screening Translational Research Network: Resources for Rare Disease Research
Tiina Urv, Ph.D. — Program Director, NICHD, NIH

4:00 p.m.  Patient Group Representatives
Peter Saltonstall — C.E.O., National Organization for Rare Diseases
Sharon Terry — C.E.O., Genetic Alliance

4:40 p.m.  Orphans at FDA: The Fundamentals
Christine Mueller, D.O. — Office of Orphan Products Development (OOPD), Food and Drug Administration (FDA)

5:05 p.m.  Closing Remarks