Agenda
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. Gallin1, 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 Collins2, M.D., Ph.D. — Director, NIH

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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.
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2:00 p.m. Bench-to-Bedside Lecture #2
Genetics of Inherited Paragangliomas and Gastric Stromal Tumors
Constantine A. Stratakis, M.D., D.M.Sci. — 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