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. Gallin, 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, 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 Program (TRND)**
Christopher P. Austin, M.D. — Director, NIH Center for Translational Therapeutics (NCTT)

11:30 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 Collins, M.D., Ph.D. — Director, NIH

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. — Acting Scientific Director, NICHD, NIH

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

Su Young Kim, M.D., Ph.D. — Assistant Clinical Investigator Pediatric Oncology Branch, National Cancer Institute (NCI), NIH

2:40 p.m. **Genetic Testing Registry**
Cathy Fomous, Ph.D. — Senior Health Policy Analyst, Office of Biotechnology Activities, NIH

3:00 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

3:25 p.m. **Break**

3:40 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

4:10 p.m. **Patient Group Representatives**
Peter Saltonstall — C.E.O., National Organization for Rare Diseases
Sharon Terry — C.E.O., Genetic Alliance

4:50 p.m. **FDA Office of Orphan Products Development**
Christine Mueller, D.O. — Office of Orphan Products Development (OOPD), Food and Drug Administration (FDA)

5:15 p.m. **Closing Remarks**