**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[1], 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 Collins[2], M.D., Ph.D.* — Director, NIH

*[1]Rare Disease Day Ribbon presentation to Dr. Gallin by Nicole Boice, Global Genes Project.*
*[2]Rare 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**