



Rare Disease Day

28 February 2014

Patients and Researchers, Partners for Life!

Agenda

8:30 a.m. First Morning Session

- **Opening Remarks**
John I. Gallin, M.D. — Director, Clinical Center, National Institutes of Health (NIH)
- **Welcoming Remarks**
Francis S. Collins, M.D., Ph.D. — Director, NIH
- **The Accomplishments and Goals of the Rare Disease Caucus**
Congressman Leonard Lance — Representative (R-NJ 7th District)
- **National Center for Advancing Translational Sciences (NCATS) Director Remarks**
Christopher Austin, M.D. — Director, NCATS, NIH
- **Common Devices for Rare Diseases**
William Heetderks, M.D., Ph.D. — Director, Extramural Science Program, National Institute of Biomedical Imaging and Bioengineering (NIBIB), NIH
- **Radiology Imaging of Rare Diseases: New Frontiers**
Ronald M. Summers, M.D., Ph.D. — Senior Investigator and Staff Radiologist Chief, Clinical Image Processing Service Chief, Imaging Biomarkers and Computer-Aided Diagnosis Laboratory Radiology and Imaging Sciences, Clinical Center, NIH
- **An Open Notebook: A Reporter's Perspective on Covering Rare Diseases and the NIH Cyclodextrin Trial**
Amy Dockser Marcus — Reporter, *Wall Street Journal*
- **Introducing the Undiagnosed Diseases Network**
Anastasia Wise, Ph.D. — Program Director, National Human Genome Research Institute (NHGRI), NIH

10:15 a.m. Morning Break

10:30 a.m. Second Morning Session

- **Introduction and Short Talk on Intramural Research**
Michael Gottesman, M.D. — Deputy Director for Intramural Research, NIH
- **Brain-Directed Adeno-Associated Virus (AAV) Gene Therapy for Neurometabolic Rare Diseases**
Stephen G. Kaler, M.D. — Senior Investigator, Molecular Medicine Program, *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), NIH
- **A Recurrent Mutation Causing Ablepharon Macrostomia Syndrome**
Taylor Davis — Post-Baccalaureate Intramural Research Training Award (IRTA) Fellow, Undiagnosed Diseases Program (UDP), NHGRI, NIH
- **Development of Effective Therapies for Neurofibromatosis Type 1 Related Tumors: Preclinical and Clinical Trials**
Brigitte Widemann, M.D. — Senior Investigator, National Cancer Institute (NCI), NIH
- **Targeting the Metabolic Basis of Kidney Cancer**
W. Marston Linehan, M.D. — Chief, Urologic Surgery, NCI, NIH





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12:00 p.m. **Lunch and Poster Session, Clinical Center Tours**

1:30 p.m. **First Afternoon Session**

- **NICHD/NHGRI Newborn Sequencing Program**
Tiina Urv, Ph.D. — Program Director, Intellectual and Developmental Disabilities Branch (IDDB), NICHD, NIH
- **Clinical Center Sequencing Program**
Dan Kastner, M.D., Ph.D. — Scientific Director, Division of Intramural Research, NHGRI, NIH
- **The Diploid Alignment Pipeline: Using Individual, Parental, and Population Information to Improve Sequence Alignment**
Elise Flynn — Post-Baccalaureate IRTA Fellow, UDP, NHGRI, NIH
- **UDPICS: Translational Research Information System Developed by UDP**
Murat Sincan, M.D. — Research Fellow, NHGRI, NIH
- **Uplifting Athletes Update**
Scott Shirley — Executive Director, Uplifting Athletes
- **Next Steps for Newly Diagnosed**
Tamandra Morgan, M.S. — Genetic and Rare Diseases Information Center (GARD), Office of Rare Diseases Research (ORDR), NCATS, NIH

3:00 p.m. **Afternoon Break**

3:15 p.m. **Second Afternoon Session**

- **Biopharmaceutical Pipeline and Orphan Diseases**
Genia Long — Senior Advisor, Analysis Group, Inc.
- **Rare Diseases and the Biopharmaceutical Industry**
William W. Chin, M.D. — Executive Vice President, Science and Regulatory Affairs, The Pharmaceutical Research Manufacturers of America (PhRMA)
- **Food and Drug Administration (FDA) Updates**
Gayatri Rao, M.D., J.D. — Director, Office of Orphan Product Development (OOPD), FDA
- **Genetic Alliance Updates**
James O'Leary, M.B.A. — Chief Innovation Officer, Genetic Alliance
- **National Organization for Rare Disorders (NORD) Updates**
Diane Dorman — Vice President of Public Policy, NORD

4:45 p.m. **Wrap Up**

5:00 p.m. **Adjourn**

