

# 2012 Rare Disease Day at National Institutes of Health (RDD@NIH)

Wednesday, February 29, 2012

Masur Auditorium • NIH • Bethesda, MD

## AGENDA

- 8:00 a.m. Registration** (Masur Auditorium Reception Area)
- 8:30 a.m. Welcoming Remarks**  
*Stephen C. Groft, Pharm.D.* — Director, Office of Rare Diseases Research (ORDR), National Center for Advancing Translational Sciences (NCATS), NIH
- 8:45 a.m. Director's Remarks**  
*Francis Collins, M.D., Ph.D.* — Director, NIH

## NEW TECHNOLOGIES

- 9:05 a.m. "Hypothesis-Generating Modes of Research Using Next-Generation Sequencing"**  
*Les Biesecker, M.D.* — National Human Genome Research Institute (NHGRI), NIH
- 9:35 a.m. "Stem Cells and Regenerative Medicine"**  
*Mahendra Rao, M.D., Ph.D.* — Director, NIH Center for Regenerative Medicine National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), NIH
- 10:00 a.m. "Uncovering the Molecular Basis of Rare Diseases"**  
*Richard Lifton, M.D., Ph.D.* — Sterling Professor of Genetics and Professor of Medicine (Nephrology); Chair, Department of Genetics, Yale University
- 10:25 a.m. Break**

## NEW RESEARCH PARADIGMS

- 10:40 a.m. "FDAs Role in Rare Disease Research and Collaboration on the Path to Product Development"**  
*Gayatri Rao, M.D., J.D.* — Acting Director, Office of Orphan Product Development (OOPD), Food and Drug Administration (FDA)
- 10:55 a.m. "NeuroNEXT – A network to accelerate drug development in neurology"**  
*Petra Kaufmann, M.D., M.Sc.* — Associate Director for Clinical Research, National Institute of Neurological Disorders and Stroke (NINDS), NIH
- 11:10 a.m. "NHLBI Centers for Accelerated Innovations / VITA: Vascular Interventions/Innovations and Therapeutic Advances"**  
*Jodi B. Black, Ph.D., M.MSc.* — Deputy Director, Division of Extramural Research Activities, National Heart, Lung, and Blood Institute (NHLBI), NIH  
*Zorina Galis, Ph.D.* — Branch Chief, Vascular Biology and Hypertension Branch, NHLBI

- 11:25 a.m. “Collaborative Development of HPBCD as a therapy for NPC1 disease”**  
*John McKew, Ph.D.* — Therapeutics for Rare and Neglected Diseases (TRND), NCATS, NIH  
*Denny Porter, M.D., Ph.D.* — National Institute of Child Health and Human Development (NICHD), NIH  
*Mark Kao, Ph.D.* — Johnson & Johnson
- 12:10 p.m. Lunch (on your own) and Poster Session (South Lobby)**  
Announcement – Uplifting Athletes’ 2012 Rare Disease Champion
- 1:25 p.m. Welcome to the Afternoon Session**  
*John I. Gallin, M.D.* — Director, Clinical Center, NIH
- 1:35 p.m. “Recombinant Immunotoxins for the Treatment of B cell Malignancies and Mesothelioma”** *Ira Pastan, M.D.* — NIH Distinguished Investigator, Co-Chief, Laboratory of Molecular Biology, National Cancer Institute (NCI), NIH
- 2:05 p.m. “Marfan Syndrome and Related Disorders: From Molecules to Medicines”** *Hal Dietz, M.D.* — Victor A. McKusick Professor of Medicine and Genetics Investigator, Howard Hughes Medical Institute Director, William S. Smilow Center for Marfan Syndrome Research, Institute of Genetic Medicine Departments of Pediatrics, Medicine, and Molecular Biology & Genetics Johns Hopkins University School of Medicine
- 2:35 p.m. Break**

## NEW RARE DISEASES

- 2:55 p.m. “Actin-interacting Protein 1 Dysfunction: A new immunodeficiency with abnormal cytoskeletal function”.**  
*Douglas B. Kuhns, Ph.D.* SAIC — National Institute of Allergy and Infectious Diseases (NIAID), NIH
- 3:15 p.m.** *Sue Swedo, M.D.* — Branch Chief, Pediatrics & Developmental Neurosciences Branch, National Institute of Mental Health, NIH
- 3:35 p.m. “Childhood fevers and rashes...not all are the same. Lessons from rare autoinflammatory diseases”**  
*Raphaella T. Goldbach-Mansky, M.D., M.H.S.* — NIAMS, NIH
- 3:55 p.m. “The NIH Undiagnosed Diseases Program: The Odyssey Continues”**  
*Cynthia Tiffit, M.D., Ph.D.* — Deputy Clinical Director, NHGRI, NIH
- 4:20 p.m. Patient Group Representatives**  
*Peter Saltostall* — C.E.O., National Organization for Rare Disorders  
*Sharon Terry* — C.E.O., Genetic Alliance
- 5:00 p.m. Community Forum**
- 5:30 p.m. Rare Disease Day at NIH activities end**