RARE DISEASE DAY at NIH
Feb. 28, 2019 | #RDDNIH

Tentative Agenda
As of Feb. 12, 2019

7:30 a.m. Registration and Poster/Exhibit Booth Setup

8:30 a.m. **Welcome**
Christopher P. Austin, M.D., Director, National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH); Former Chair, International Rare Diseases Research Consortium (IRDiRC)

James K. Gilman, M.D., Chief Executive Officer (CEO), Clinical Center (CC), NIH

8:55 a.m. **NCATS Office of Rare Diseases Research (ORDR) Update**
Anne R. Pariser, M.D., Director, ORDR, NCATS, NIH

9:10 a.m. **Zebbie Award Presentation**
Anne R. Pariser, M.D., Director, ORDR, NCATS, NIH

9:20 a.m. **Beyond the Diagnosis Unveiling: Portrait by Jota Leal**
Patricia Weltin, CEO and Founder, Beyond the Diagnosis

9:30 a.m. **Session 1: The Collective Research Model with the NIH Rare Diseases Clinical Research Network (RDCRN)**
Panel members will discuss the importance of collaborative research approaches for rare diseases. A diverse panel will share case studies and illustrative examples from the RDCRN, which integrates academic investigators, patient groups, trainees, NIH scientific staff and others to accelerate rare diseases research.

Moderator: Tiina K. Urv, Ph.D., Program Director, ORDR, NCATS, NIH

Panelists:

- Michael E. Shy, M.D., Principal Investigator (PI), Inherited Neuropathies Consortium; Director, Division of Neuromuscular Disease and Division of Neurogenetics, Department of Neurology, University of Iowa
- Mustafa Sahin, M.D., Ph.D., PI, Developmental Synaptopathies Consortium (DSC); Director, Translational Neuroscience Center, Boston Children's Hospital
- Steven L. Roberds, Ph.D., Coalition of Patient Advocacy Groups (CPAG) Representative, DSC; Chief Scientific Officer, Tuberous Sclerosis Alliance
- Seema S. Aceves, M.D., Ph.D., PI, Consortium of Eosinophilic Gastrointestinal Disease Researchers (CEGIR); Professor of Pediatrics and Medicine, University of California, San Diego; Director, Eosinophilic Gastrointestinal Disorders Clinic, Rady Children's Hospital, San Diego
- Ellyn Kodroff, CPAG Representative, CEGIR [Campaign Urging Research for Eosinophilic Disease (CURED), American Partnership for Eosinophilic Disorders, Eosinophilic Family Coalition]; President and Founder, CURED

10:30 a.m. Networking Break

10:45 a.m. **NIH Director Remarks**
Francis S. Collins, M.D., Ph.D., Director, NIH

11:00 a.m. **Session 2: The Power of Patients — Harnessing Quality Registries to Understand Your Rare Disease**
An essential step in improving the diagnosis and treatment of a rare disease is to establish an understanding of the disease's natural history. Natural history studies often provide foundational information for clinical research and drug development programs, and these studies often rely on active partnerships with patient organizations. Panel members will discuss their experiences in developing registries to understand rare diseases and in using natural history data to advance care for rare disease patients.

Moderator: Eric W.K. Sid, M.D., M.H.A., Presidential Management Fellow, ORDR, NCATS, NIH

Panelists:

- Forbes Denny Porter, M.D., Ph.D., Clinical Director and Senior Investigator, Division of Intramural Research, *Eunice Kennedy Shriver* National Institute of Child Health and Human Development, NIH
- Emily Milligan, M.P.H., Executive Director, Barth Syndrome Foundation
- Jeanine D'Armiento, M.D., Ph.D., Professor of Medicine in Anesthesiology, Director of the Center for LAM and Rare Lung Disease, Columbia University Medical Center; Chair, Board of Directors, Alpha-1-Foundation
- Janet Maynard, M.D., M.H.S., Acting Director, Office of Orphan Products Development, Food and Drug Administration

Noon Lunch *(on your own)*

- Networking Rooms
- Demonstration of the NCATS Toolkit and Rare Diseases Registry (RaDaR) Program
- Poster Session and Exhibits
- NIH CC Tours
- Beyond the Diagnosis Art Exhibition
- Videos:
    - Personal Rare Disease Stories
    - Messages from Global Rare Disease Leaders
    - Rare Diseases Are Not Rare! Prize Challenge Exhibition

1:30 p.m. **Congressional Remarks**
Rep. G. K. Butterfield (D-NC), Co-Chair, Rare Disease Congressional Caucus

1:45 p.m. **Session 3: Success Factors for Rare Cancer Research — Building Strong Foundations**
Panelists will discuss the similarities and differences in the challenges that researchers face in rare cancers as compared to other rare diseases. Experts from NIH's National Cancer Institute (NCI) and leading rare cancer advocates will describe their efforts to coordinate research both nationally and internationally, as well as different models of success for rare cancer advocacy and the role of social media in building rare cancer communities and recruiting patients for studies.

Moderator: Abby B. Sandler, Ph.D., Executive Director, My Pediatric and Adult Rare Tumor (MyPART) Network, Center for Cancer Research (CCR), NCI

Panelists:

- Karlyne M. Reilly, Ph.D., Director, Rare Tumor Initiative, CCR, NCI
- Jack J. Welch, M.D., Ph.D., Medical Officer, International Rare Cancers Initiative, Center for Global Health, NCI
- Denise Reinke, M.S., N.P., M.B.A., President and CEO, Sarcoma Alliance for Research through Collaboration
- Jim Palma, Executive Director, TargetCancer Foundation
- Corrie Painter, Ph.D., Associate Director, Count Me In; Associate Director of Operations and Scientific Outreach, Broad Institute of Massachusetts Institute of Technology and Harvard University

2:45 p.m. **Session 4: No Disease Left Behind, No Patient Left Behind**

New technologies such as gene editing and genome therapy have potentially broad implications for many rare diseases. However, for clinical trials, there are practical challenges that need to be addressed to ensure that these technologies are accessible to all patients that might benefit from them. Panelists, including scientists, patients and other stakeholders, will discuss these issues.

Opening Remarks: Philip John (P.J.) Brooks, Ph.D., Program Director, ORDR, NCATS, NIH

Moderator: Jonathan Jackson, Ph.D., Center Director, Community Access, Recruitment, and Engagement (CARE) Research Center, Massachusetts General Hospital (MGH)

Panelists:

- John F. Tisdale, M.D., Chief, Cellular and Molecular Therapeutics Branch, National Heart, Lung, and Blood Institute, NIH
- Tesha F. Samuels, Rare Disease Patient
- Helen Hemley, Program Manager, CARE Research Center, MGH
- Mandy Mansaray, RN, M.A., Program Coordinator, Clinical Research Volunteer Program, Office of Patient Recruitment, CC, NIH
- Miguel Negrete, Parent of a Rare Disease Patient

3:45 p.m. **The Children's Inn at NIH: A Place Like Home**

Jennie Lucca, M.S.W., CEO, The Children's Inn at NIH

3:55 p.m. **Closing Remarks**

Christopher P. Austin, M.D., Director, NCATS, NIH; Former Chair, IRDiRC

4:00 p.m. Adjournment

If interested in a walking tour of the National Library of Medicine at NIH, please meet Tara Mowery at the lower level registration desk at 4:00 p.m.