



## RARE DISEASE DAY at NIH

Feb. 28, 2019 | #RDDNIH

### Final Agenda

- 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



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10:45 a.m.

### NIH Director Remarks

Francis S. Collins, M.D., Ph.D., Director, NIH

11:00 a.m.

### Session 2: There is Power in Numbers — Harnessing Patient Data through Registries

*An essential step in improving the diagnosis and treatment of a rare disease is collecting enough research information, such as through a natural history study, to enable a thorough understanding of that disease and its effects on a patient. Patient organizations play a critical role in this process by including their communities in registries, helping in recruitment for research studies, and acting as stewards for this data. Panel members will discuss what organizations need to gather information from within their community and how best to use patient experience data to support and advance the care of 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
- Janet Maynard, M.D., M.H.S., Director, Office of Orphan Products Development, Food and Drug Administration
- 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

Noon

#### Lunch (*on your own*)

- Networking Rooms [*Rooms A/B, G1/G2*]
- Share your insights on NCATS' rare disease-related websites [*Room D*]:
  - [Rare Diseases Registry Program \(RaDaR\)](#)
  - [NCATS Toolkit for Patient-Focused Therapy Development](#)
  - [Genetic and Rare Diseases Information Center \(GARD\)](#)
- Rare Diseases Are Not Rare! Prize Challenge Exhibition [*Room A/B*]
- Beyond the Diagnosis Art Exhibition [*Room C1/C2*]
- Videos:
  - Personal Rare Disease Stories [*Room C1/C2*]
  - Messages from Global Rare Disease Leaders [*Room G1/G2*]
- Poster Session and Exhibits [*Atrium, Room E1/E2, Lower Level Foyer*]
- NIH CC Tours [*shuttle outside Natcher Front Lobby*]

1:30 p.m.

### Session 3: Success Factors for Rare Cancer Research — Building Strong Foundations

*Panelists will discuss the similarities and differences in the challenges that advocates and 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.*



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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; Co-Chair, Rare Cancer Coalition, National Organization for Rare Disorders
- 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

2:30 p.m. Networking Break

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 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 D. 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.\*\**