



RARE DISEASE DAY at NIH

March 1, 2018 | #RDDNIH

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); Chair, International Rare Diseases Research Consortium (IRDiRC)
- 8:40 a.m. **NIH Director Remarks**
Francis S. Collins, M.D., Ph.D., Director, NIH
- Congressional Remarks**
Rep. Leonard Lance (R-NJ), House Co-Chair, Rare Disease Congressional Caucus
- 9:00 a.m. **NIH Clinical Center Chief Executive Officer Remarks**
James K. Gilman, M.D., Chief Executive Officer (CEO), NIH Clinical Center (CC)
- 9:10 a.m. **Food and Drug Administration (FDA) Update**
Katherine Needleman, Ph.D., Director, Orphan Products Grants Program, Office of Orphan Products Development, FDA
- 9:25 a.m. **Session 1: Gene Editing**
Panel members will discuss the groundbreaking work and collaborative approaches that led to the first-in-human gene editing clinical trial for Hunter Syndrome, collectively providing clinical research, patient and industry perspectives.
- Moderator: Chester B. Whitley, Ph.D., M.D., Professor, Gene Therapy Center, Department of Pediatrics and Experimental and Clinical Pharmacology, University of Minnesota; Principal Investigator, Lysosomal Disease Network
- Panelists:
- Erica Thiel, Patient Representative
 - R. Scott McIvor, Ph.D., Professor, Department of Genetics, Cell Biology and Development, University of Minnesota
 - Sandy Macrae, M.B., Ch.B., Ph.D., President and CEO, Sangamo Therapeutics
- 10:10 a.m. **Overview of the NIH Common Fund Somatic Cell Genome Editing Program**
Philip John (P.J.) Brooks, Ph.D., Program Director, Division of Clinical Innovation (DCI) and Office of Rare Diseases Research (ORDR), NCATS, NIH
- 10:20 a.m. Break



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10:45 a.m.

Session 2: Collaborating for Successful Research

Patient groups play an essential role in advancing rare disease research. Patient organization representatives will discuss how collaborations improve understanding of these disorders and help advance development of new therapies.

Moderator: Marshall Summar, M.D., Chairman of the Board of Directors, National Organization for Rare Disorders; Chief, Division of Genetics and Metabolism, Children's National Health System; Director, Rare Disease Institute

Panelists:

- Margie Frazier, Ph.D., LISW-S, Executive Director, Batten Disease Support and Research Association
- Theresa V. Strong, Ph.D., Co-Founder and Director of Research Programs, Foundation for Prader-Willi Research
- Salvatore La Rosa, Ph.D., Vice President, Research and Development, Children's Tumor Foundation
- Kathleen Brewer, Founder and President, Erdheim-Chester Disease (ECD) Global Alliance
- James O'Leary, M.B.A., Chief Innovation Officer, Genetic Alliance

11:45 a.m.

Lunch (*on your own*)

- Poster Session
- Exhibits
- NIH CC Tours
- Art Exhibition
- Videos

1:15 p.m.

Session 3: Gene Therapy

Gene therapy for rare diseases is moving forward rapidly. Panelists will share their perspectives on gene therapy scientific advances and research gaps, and on how to advance gene therapies for more rare diseases.

Moderator: P.J. Brooks, Ph.D., Program Director, DCI and ORDR, NCATS, NIH

Panelists:

- Peter Marks, M.D., Ph.D., Director, Center for Biologics Evaluation and Research, FDA
- Kristin Smedley, President, Curing Retinal Blindness Foundation
- Jeffrey D. Marrazzo, M.B.A., M.P.A., Co-Founder and CEO, Spark Therapeutics, Inc.
- Maria Kefalas, Co-Founder, Calliope Joy Foundation



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2:00 p.m.

Session 4: Engaging the Next Generation of the Rare Diseases Community

This panel focuses on the perspective of young adults in the rare diseases community. The panelists will share their individual experiences and discuss how their generation can support research, provide access to care, or advocate for change using the power of social media, digital communications and modern technology.

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

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

Panelists:

- Tej Neaz Powell, Patient Representative, The Children's Inn at NIH
- Shira Strongin, Founder, Sick Chicks; Youth Adult Advisory Council, The Children's Inn at NIH
- Taylor Kane, Founder, Young ALD (Adrenoleukodystrophy) Carriers and Remember the Girls
- Maddie Shaw, Founder, Maddie's Herd

2:45 p.m.

Student Athletes Use Their Platform to Make an Impact

Robert M. Long, Director of Strategic Development, Uplifting Athletes

2:55 p.m.

Break

3:25 p.m.

Congressional Remarks via Video

Sen. Amy Klobuchar (D-MN), Senate Co-Chair, Rare Disease Congressional Caucus

3:35 p.m.

NCATS ORDR Update

Anne Pariser, M.D., Director, ORDR, NCATS, NIH

3:50 p.m.

Closing Remarks

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

4:00 p.m.

Adjournment