Tentative Agenda  
As of Feb. 2, 2018

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

8:50 a.m. NCATS Office of Rare Diseases Research (ORDR) Update  
Petra Kaufmann, M.D., M.Sc., Director, ORDR, NCATS, NIH  
Anne Pariser, M.D., Deputy Director, ORDR, NCATS, NIH

9:00 a.m. Food and Drug Administration (FDA) Update  
Katherine Needleman, Ph.D., Director, Orphan Products Grants Program, Office of Orphan Products Development, FDA

9:15 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:
- 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
- Erica Thiel, Patient Representative

10:00 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 ORDR, NCATS, NIH

10:10 a.m. Break

10:30 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, 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

11:30 a.m. **Lunch (on your own)**
- Poster Session
- Exhibits
- NIH CC Tours
- Videos

1:00 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:**
- Maria Kefalas, Co-Founder, Calliope Joy Foundation
- Jeffrey D. Marrazzo, M.B.A., M.P.A., Co-Founder and CEO, Spark Therapeutics, Inc.
- Kristin Smedley, President, Curing Retinal Blindness Foundation
- Peter Marks, M.D., Ph.D., Director, Center for Biologics Evaluation and Research, FDA

1:45 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:
- Shira Strongin, Founder, Sick Chicks
- Maddie Shaw, Founder, Maddie’s Herd
- Taylor Kane, Founder, Young ALD (Adrenoleukodystrophy) Carriers; Founder, Remember the Girls
- Tej Neaz Powell, Patient Representative, The Children’s Inn at NIH

2:30 p.m.  **Student Athletes Use Their Platform to Make an Impact**
Robert M. Long, Director of Strategic Development, Uplifting Athletes

2:40 p.m.  Break

3:00 p.m.  **NIH Director Remarks**
Francis S. Collins, M.D., Ph.D., Director, NIH

3:15 p.m.  **Congressional Remarks**
Rep. Leonard Lance (R-NJ), House Co-Chair, Rare Disease Congressional Caucus

3:45 p.m.  **Summary and Highlights**
Petra Kaufmann, M.D., M.Sc., Director, ORDR, NCATS, NIH
Anne Pariser, M.D., Deputy Director, ORDR, NCATS, NIH

3:55 p.m.  **Closing Remarks**
Christopher P. Austin, M.D., Director, NCATS, NIH; Chair, IRDiRC
James K. Gilman, M.D., CEO, NIH CC

4:00 p.m.  Adjournment