Preliminary Agenda  
as of 1/21/2020

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

9:00 a.m.  NCATS Office of Rare Diseases Research (ORDR) Update
Anne R. Pariser, M.D., Director, ORDR, NCATS, NIH

9:30 a.m.  Session 1: Shortening the Diagnostic Odyssey
Rare disease patients often spend many years searching for answers before receiving a diagnosis. Diagnosis remains a challenge, but advances in genomics, medical informatics and novel clinical approaches are helping to make progress toward quicker diagnoses. Panelists will share their experiences, as well as advances in shortening the diagnostic odyssey.

Moderator: Marshall Summar, M.D., Director, Rare Disease Institute; Chief, Division of Genetics and Metabolism, Children’s National Hospital; Chairman of the Board of Directors, National Organization for Rare Disorders (NORD)

Panelists:
- Lisa Deck, Founder and Director, Sisters@Heart
- Stephen Kingsmore, M.D., D.Sc., President and CEO, Rady Children's Institute for Genomic Medicine, Rady Children's Hospital–San Diego
- Michael R. Knowles, M.D., Professor, Division of Pulmonary Diseases and Critical Care Medicine, University of North Carolina School of Medicine
- Richard Moscicki, M.D., Chief Medical Officer and Executive Vice President of Science and Regulatory Advocacy, Pharmaceutical Research and Manufacturers of America (PhRMA)

10:30 a.m.  A Rare Story #1
Speakers:
- Amanda Ombrello, M.D., Staff Clinician, National Human Genome Research Institute, NIH
- Chip Chambers, M.D., Founder and President, DADA2 Foundation

10:45 a.m.  Networking Break
11:15 a.m.  **A Rare Story #2**  
*Introduction*: Stephanie Feinberg, Resident Services Operations Manager, The Children’s Inn at NIH  
*Speaker*: Noah Victoria, Rare Disease Patient

11:30 a.m.  **Session 2: Individualized Therapeutic Approaches and Personalized Medicine**  
*Advances in precision medicine have potentially broad implications for many rare genetic diseases. Panelists will discuss one patient’s journey toward individualized therapy, as well as challenges encountered with this approach.*  

*Moderator*: Philip John (P.J.) Brooks, Ph.D., Program Director, ORDR, NCATS, NIH  
*Panelists*:  
- Timothy W. Yu, M.D., Ph.D., Principal Investigator, The Yu Lab; Attending Physician, Division of Genetics and Genomics, Boston Children’s Hospital; Assistant Professor of Pediatrics, Harvard Medical School  
- Julia Vitarello, Founder and CEO, Mila’s Miracle Foundation  
- Patroula Smpokou, M.D., Clinical Team Leader, Division of Gastroenterology and Inborn Errors Products, Office of New Drugs, Center for Drug Evaluation and Research, U.S. Food and Drug Administration

12:30 p.m.  **Lunch (on your own)**  
*Available Activities*  
- Poster Session  
- Exhibit Tables  
- NIH Clinical Trial Resources  
- Tour of the NIH Clinical Center  
- Tour of the National Library of Medicine  
- Art Exhibition by Beyond the Diagnosis and Others

2:00 p.m.  **A Rare Story #3**  
*Introduction*: Elizabeth A. Ottinger, Ph.D., Senior Program Manager and Drug Development Team Lead, Therapeutics for Rare and Neglected Diseases (TRND) Program, Division of Pre-Clinical Innovation, NCATS, NIH  
*Speaker*: Neena Nizar, Ed.D., Founder and President, The Jansen’s Foundation

2:15 p.m.  **Session 3: Nontraditional Approaches to Improving Access for Rare Diseases**  
*For rare diseases, where resources and specialized expertise may be limited, nontraditional methods may help expand access. Learn about Project ECHO® (Extension for Community Healthcare Outcomes) and how this academic resource has been used by patient groups to expand access to knowledge.*  

*Moderator*: Kristen Wheeden, Executive Director, American Porphyria Foundation
Panelists:
- Sophie Lanzkron, M.D., M.H.S., Associate Professor of Medicine and Oncology, Johns Hopkins School of Medicine
- Teresa M. Kohlenberg, M.D., Phelan-McDermid Syndrome Foundation (PMSF), ECHO PMS Neuropsychiatric Consultation Group
- Laura L. Tosi, M.D., Director of the Bone Health Program, Children’s National Hospital
- Michael Stewart, Program Manager, Rare Bone Disease TeleECHO Clinic Series; Regional Program Services Manager, Osteogenesis Imperfecta Foundation

3:15 p.m. **Sharing Rare Stories**
**Introduction:** Marrah Lachowicz-Scroggins, Ph.D., Program Director, Division of Lung Diseases, National Heart, Lung, and Blood Institute, NIH  
**Speaker:** Mary Rose Kitlowski, Founder, Running On Air

**Introduction:** Elena Schwartz, Ph.D., Program Director, Center to Reduce Cancer Health Disparities, National Cancer Institute, NIH  
**Speaker:** Kurt R. Weiss, M.D., Director of the Musculoskeletal Oncology Laboratory, Assistant Professor of Orthopaedic Surgery, Division of Musculoskeletal Oncology, University of Pittsburgh School of Medicine

**Introduction:** Anne R. Pariser, M.D., Director, ORDR, NCATS, NIH  
**Speaker:** David Hysong, Founder and CEO, SHEPHERD Therapeutics

3:45 p.m. **Town Hall: Open Mic Q&A**  
**Moderator:** Joni L. Rutter, Ph.D., Deputy Director, NCATS, NIH  
**Panelists:**  
- NIH staff and researchers

4:15 p.m. **Closing Remarks**  
Christopher P. Austin, M.D., Director, NCATS, NIH

4:30 p.m. **Adjournment**

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*If interested in a walking tour of the National Library of Medicine on the NIH campus, please meet Tara Mowery at the Natcher lower level registration desk at 4:15 p.m.*