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)

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

8:45 a.m. **NIH Director Remarks via Video**
Francis S. Collins, M.D., Ph.D., Director, NIH

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

9:20 a.m. **NCATS Resources to Empower the Rare Diseases Patient Community**
Eric W.K. Sid, M.D., M.H.A., Program Officer, 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 L. 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; Director, Moyamoya Foundation
- Stephen F. 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 A. 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., Associate Research Physician, Adult and Pediatric Rheumatologist, National Human Genome Research Institute (NHGRI), NIH
- Chip Chambers, M.D., Assistant Clinical Professor of Surgery, Vanderbilt University Medical Center; 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, Clinical Center, NIH

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 their journeys 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:
- Julia Vitarello, Founder and CEO, Mila's Miracle Foundation
- 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
- 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:
- NIH Clinical Trial Resources *[Room G1/G2]*
- Networking Room *[Room F1/F2]*
- Poster Session and Exhibit Tables *[Atrium, Room E1/E2, Lower Level Foyer]*
- Tour of the NIH Clinical Center *[Shuttles outside Main Entrance]*
- Tour of the National Library of Medicine *[Lower Level Foyer]*
- Art Exhibition by Beyond the Diagnosis, The Art of Living with a Rare Disease and Others *[Room C1/C2, Upper Lobby, Upper Walkway]*

2:00 p.m. **A Rare Story #3**
Introduction: Elizabeth A. Ottinger, Ph.D., Senior Program Manager, Therapeutics for Rare and Neglected Diseases (TRND) Program, Division of Pre-Clinical Innovation (DPI), 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, Rare Disease Advocate and Patient

Introduction: Elena Schwartz, Ph.D., Program Director, Center to Reduce Cancer Health Disparities, National Cancer Institute (NCI), NIH
Speaker: Kurt R. Weiss, M.D., Director of the Musculoskeletal Oncology Laboratory, Associate 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:

- Krishna (Balki) Balakrishnan, Ph.D., M.B.A., Senior Technology Transfer Manager, Office of Strategic Alliances, NCATS, NIH
- P.J. Brooks, Ph.D., Program Director, ORDR, NCATS, NIH
- Lea C. Cunningham, M.D., Medical Director, RUNX1 Program, NHGRI, NIH; Associate Research Physician, Immune Deficiency & Cellular Therapy Program (ID-CTP), NCI, NIH
- Nikita Curry, M.H.A., Supervisor, Office of Patient Recruitment, Clinical Center, NIH
- Matthew D. Hall, Ph.D., Acting Director, Early Translation Branch; Biology Group Leader, DPI, NCATS, NIH

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.***