Final Agenda

10:00 a.m. EST **Virtual Rare Disease Day at NIH Overview**
Alice Chen Grady, M.D., Program Officer, Office of Rare Diseases Research (ORDR), National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH)

10:10 a.m. **Welcoming Remarks**
Joni L. Rutter, Ph.D., Acting Director, NCATS, NIH
James K. Gilman, M.D., CEO, Clinical Center, NIH

10:20 a.m. **Caregiver Resilience — Mind, Body and Spirit**
Margaret Bevans, Ph.D., RN, FAAN, Director, Office of Research Nursing, Office of the Clinical Director, National Heart, Lung, and Blood Institute (NHLBI), NIH

10:35 a.m. **NIH Acting Director Remarks**
Lawrence A. Tabak, D.D.S., Ph.D., Acting Director, NIH

10:50 a.m. **Congressional Remarks**
Sen. Roger F. Wicker (R-MS), Senate Co-Chair, Rare Disease Congressional Caucus
Rep. G.K. Butterfield (D-NC), House Co-Chair, Rare Disease Congressional Caucus
Rep. Gus M. Bilirakis (R-FL), House Co-Chair, Rare Disease Congressional Caucus

11:00 a.m. **Session 1: Diversity in Rare Diseases Research and Equity of Care**
Due to the smaller patient population sizes involved, equitable and representative engagement for all patients with a rare disease is a key ingredient for successful and impactful research. This panel will address diverse and inclusive best practices, challenges and perspectives presented by research stakeholders (clinicians, researchers, advocates, etc.) to elevate all patients in the rare diseases community.
Moderator: *Vence L. Bonham Jr., J.D.*, Acting Deputy Director, National Human Genome Research Institute (NHGRI), NIH
Panelists:

- *Alexandra "Xan" C.H. Nowakowski, Ph.D., M.P.H.*, Assistant Professor, Geriatrics/Behavioral Sciences and Social Medicine and Vice Chair, Council on Diversity and Inclusion, Florida State University College of Medicine
- *Michelle Takemoto, M.S., CGC*, Genetic Counselor, Hawaii Department of Health Genetics Program; Genetics Project Specialist, Western States Regional Genetics Network; Co-Founder, Minority Genetic Professionals Network
- *Aisha T. Langford, Ph.D., M.P.H.*, Assistant Professor, Division of Comparative Effectiveness and Decision Science, Department of Population Health, NYU Langone Health

- *Nicole Kressin, RN, M.S.N.*, Co-Chair, Diversity Committee, Rare Diseases Clinical Research Network (RDCRN); Chair, Diversity Committee and Regulatory Manager, Inherited Neuropathy Consortium, RDCRN; Clinical Research Specialist, Department of Neurology, University of Iowa Health Care
- *Tracy M. King, M.D., M.P.H.*, Co-Chair, Diversity Committee, RDCRN; Medical Officer, Intellectual and Developmental Disabilities Branch, *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), NIH

12:00 p.m. **Break**

- Virtual Exhibits and Scientific Posters
- Art Exhibition
- Networking

12:30 p.m. **Session 2: Progress in the Development of Individualized Therapeutic Approaches and Personalized Medicine**

Advances in precision medicine, and, in particular, "N-of-1" trials of antisense oligonucleotides, have potentially broad implications for many rare genetic diseases. Panelists will discuss ongoing efforts toward standardizing and increasing the efficiency of this approach in multiple diseases.

Moderator: *Philip John (P.J.) Brooks, Ph.D.*, Acting Director, ORDR, NCATS, NIH

Panelists:

- *Hobart Rogers, Pharm.D., Ph.D.*, Reviewer, Division of Translational and Precision Medicine, Office of Clinical Pharmacology, Center for Drug Evaluation and Research, U.S. Food and Drug Administration
- *Mehmet Kuzu*, Parent of a Rare Disease Patient
- *Scott Demarest, M.D., M.S.C.S.*, Assistant Professor, Departments of Pediatrics and Neurology, University of Colorado School of Medicine; Clinical Director, Precision Medicine, Children's Hospital Colorado
- *Adeline L. Vanderver, M.D.*, Principal Investigator, Global Leukodystrophy Initiative Clinical Trials Network, RDCRN; Professor of Neurology, Perelman School of Medicine, University of Pennsylvania; Program Director, Leukodystrophy Center of Excellence, Children's Hospital of Philadelphia

1:30 p.m. **Break**

- Virtual Exhibits and Scientific Posters
- Art Exhibition
- Networking

2:20 p.m. **NCATS ORDR Update**
P.J. Brooks, Ph.D., Acting Director, ORDR, NCATS, NIH
Ainslie Tisdale, Program Analyst, ORDR, NCATS, NIH

2:35 p.m. **The National Economic Burden of Rare Disease Study**
Annie Kennedy, Chief, Policy and Advocacy, EveryLife Foundation for Rare Diseases

2:40 p.m. **Genetic and Rare Diseases (GARD) Information Center Update**
Eric W.K. Sid, M.D., M.H.A., Program Officer, ORDR, NCATS, NIH

2:50 p.m. **Session 3: Successful Clinical Trial Enrollment with True Advocacy Collaboration During Challenging Times**

This is a success story from the rare diseases patient community, where collaboration with advocacy and industry led to successful clinical trial enrollment. Discussion will include how successful strategies were implemented to support protocol design, patient enrollment and engagement to support a Phase 1/2 trial for pulmonary sarcoidosis, despite being faced with a challenging pandemic environment. This panel will discuss lessons learned and advice for clinical trial recruitment successes in the future.

Moderator: *Shazia Ahmad*, Senior Director and Head, Patient and Physician Services, United BioSource LLC

Panelists:

- *Sanjay S. Shukla, M.D., M.S.*, President and CEO, aTyr Pharma
- *Tricha Shivas, M.B.E.*, Chief Strategy Officer, Foundation for Sarcoidosis Research (FSR)
- *Erica Courtenay-Mann*, Member, FSR Women of Color Patient Advisory Committee

3:25 p.m. **Session 4: Our Journey with NIH — How One Rare Disease Organization Forged a Transformational Research Partnership**

In 2003, a small group of volunteer parents and grandparents created the Cure JM Foundation to address the fact that very little was understood about the devastating pediatric autoimmune disease juvenile myositis. Fast forward to 2022, and Cure JM is a force within the rare diseases community — funding research and clinical trials to find better treatments and a cure. Learn how the evolution of one small group has turned into strong relationships with the NIH and with clinicians and researchers throughout the United States and abroad.

Moderator: *James Minow*, Executive Director, Cure JM Foundation

Panelists:

- *Lisa Rider, M.D.*, Head and Senior Clinician, Environmental Autoimmunity Group, Clinical Research Branch, National Institute of Environmental Health Sciences (NIEHS), NIH

- *Kristine Alderfer*, President, Board of Directors, Cure JM Foundation; Mother of a Rare Disease Patient
- *Katherine Alderfer*, Student; Rare Disease Patient, Clinical Center, NIH

3:45 p.m. **Session 5: Natural History Data Collection for a Gene Therapy Clinical Trial**

This session will describe experiences with natural history data collection in the context of an ongoing National Institute of Neurological Disorders and Stroke (NINDS)-sponsored gene therapy clinical trial for aromatic l-amino acid decarboxylase (AADC) deficiency, a rare neurodevelopmental condition. The important role of natural history data and methods used in this study, including home visits and telemedicine, will be discussed from the perspectives of an investigator and a parent.

Speakers:

- *Toni S. Pearson, M.B.B.S.*, Associate Professor of Neurology, Washington University School of Medicine in St. Louis
- *Shillann Rodriguez-Pena*, Parent of a Child with AADC Deficiency

4:05 p.m. **Break**

- Virtual Exhibits and Scientific Posters
- Art Exhibition
- Networking

4:30 p.m. **Session 6: The Use of Telehealth During COVID-19**

With the COVID-19 pandemic continuing to affect our way of life, telehealth has become an integral part of the rare diseases community in multiple ways. In this session, learn how telehealth has revolutionized care for many rare disease patients, clinicians and researchers. Panelists will share their perspectives on the benefits and challenges to the quick adoption of this technology.

Moderator: *Tiffani Bailey Lash, Ph.D.*, Program Director, Digital Health and Point of Care Technologies, Division of Health Informatics Technologies (Informatics), National Institute of Biomedical Imaging and Bioengineering (NIBIB), NIH

Introduction: *Heidi Ross, M.P.H.*, Acting Vice President, Policy and Regulatory Affairs, National Organization for Rare Disorders

Panelists:

- *Jeanine M. D'Armiento, M.D., Ph.D.*, Professor of Medicine in Anesthesiology, Director of the Center for Lymphangioleiomyomatosis (LAM) and Rare Lung Disease, Columbia University Medical Center; Chair, Board of Directors, Alpha-1 Foundation
- *Kristin Grassi*, Rare Disease Patient
- *Andrea L. Gropman, M.D.*, Principal Investigator, Urea Cycle Disorders Consortium, RDCRN; Professor and Division Chief, Neurodevelopmental Pediatrics and Neurogenetics, Children's National Hospital

5:10 p.m. **Session 7: A Journey Down the Long and Winding Road of the Diagnostic Odyssey for Rare and Undiagnosed Conditions**

It takes a village to complete the comprehensive diagnosis of rare and undiagnosed conditions successfully. It requires a combined effort from patients, family members, caregivers, clinicians and researchers. This panel's participants will share their experiences from their diagnostic odyssey. The session also will provide direction to useful tools and resources.

Moderator: *Elena Schwartz, Ph.D.*, Program Director, Coordinating Center for Clinical Trials, National Cancer Institute (NCI), NIH

Introduction: *Cynthia J. Tifft, M.D., Ph.D.*, Deputy Clinical Director, Office of the Clinical Director, NHGRI, NIH; Director, Pediatric Undiagnosed Diseases Program, NIH

Panelists:

- *Troy Evans*, Rare Disease Patient
- *Erika Cox*, Mother of a Rare Disease Patient
- *Monique and Vivianne*, Rare Disease Patients, Mother and Daughter, Clinical Center, NIH
- *Terrance and Terran*, Father of a Rare Disease Patient and Rare Disease Patient, Clinical Center, NIH
- *Eric W.K. Sid, M.D., M.H.A.*, Program Officer, ORDR, NCATS, NIH

5:50 p.m. **Closing Remarks**

Joni L. Rutter, Ph.D., Acting Director, NCATS, NIH

6:00 p.m. **Adjournment**