Preliminary Agenda
As of 1/31/2022

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

10:25 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

10:40 a.m.  NIH Acting Director Remarks
Lawrence A. Tabak, D.D.S., Ph.D., Acting Director, NIH

11:10 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:

- Xan 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 M. Kressin, M.S.N., RN, Co-Chair, Diversity Committee and Regulatory Manager, Inherited Neuropathy Consortium, Rare Diseases Clinical Research Network (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

12:10 p.m.  
**Break**

• Virtual Exhibits and Scientific Posters
• Art Exhibition
• Networking

12:35 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:**

• 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

• Mehmet Kuzu, Parent of a Rare Disease Patient

• 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

• 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

1:35 p.m.  
**Break**

• Virtual Exhibits and Scientific Posters
• Art Exhibition
• Networking

2:30 p.m.  
**NCATS ORDR Update**

*P.J. Brooks, Ph.D., Acting Director, ORDR, NCATS, NIH
Ainslie Tisdale, Program Analyst, ORDR, NCATS, NIH*
2:40 p.m. **The National Economic Burden of Rare Disease Study**  
*Annie Kennedy*, Chief, Policy and Advocacy, EveryLife Foundation for Rare Diseases

2:45 p.m. **Genetic and Rare Diseases (GARD) Information Center Update**  
*Eric W.K. Sid, M.D., M.H.A.*, Program Officer, ORDR, NCATS, NIH

2:55 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:**  
- *Erica Courtenay-Mann*, Member, FSR Women of Color Patient Advisory Committee  
- *Sanjay S. Shukla, M.D., M.S.*, President and CEO, aTyr Pharma

3:30 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*, Mother of a Rare Disease Patient  
- *Katherine Alderfer*, Rare Disease Patient, Clinical Center, NIH
3:50 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

4:10 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

4:50 p.m. **Break**
- Virtual Exhibits and Scientific Posters
- Art Exhibition
- Networking
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
- Vivianne and Monique, Rare Disease Patients, Daughter and Mother, Clinical Center, NIH
- Terran and Terrance, Rare Disease Patient and Father of a 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**