Preliminary Agenda
as of 1/11/2021

10:30 a.m. EST  Virtual Rare Disease Day at the National Institutes of Health (NIH) Overview
Alice Chen Grady, M.D., Program Officer, Office of Rare Diseases Research (ORDR), National Center for Advancing Translational Sciences (NCATS), NIH

10:40 a.m.  Welcome Remarks
Christopher P. Austin, M.D., Director, NCATS, NIH
James K. Gilman, M.D., Chief Executive Officer (CEO), Clinical Center, NIH

11:20 a.m.  Rare Story #1: From Diagnosis to Destiny
Speaker:
• Jacob Thompson, aka TEN20, Rare Disease Patient, Advocate, Artist, Author, Inspirational Speaker

Rare Story #2: Industry and Patient Advocacy Collaborations — Making It a Win-Win
Speakers:
• Shazia Ahmad, Director, Patient and Physician Services, United BioSource LLC (UBC); Rare Disease Thought Leader, Patient and Stakeholder Engagement
• Nadia Bodkin, Pharm.D., M.S., Rare Disease Patient Advocate, Rare Advocacy Movement (RAM)
• Christopher U. Missling, Ph.D., President and CEO, Anavex Life Sciences Corp.; Member of RAM

12:05 p.m.  Break

12:20 p.m.  NCATS ORDR Update
Anne R. Pariser, M.D., Director, ORDR, NCATS, NIH

12:30 p.m.  Rare Diseases Clinical Research Network (RDCRN): Research Survey on Impacts of COVID-19
Speakers:
• Marc E. Rothenberg, M.D., Ph.D., Principal Investigator, Consortium of Eosinophilic Gastrointestinal Disease Researchers (CEGIR), RDCRN; Director, Division of Allergy and Immunology, Cincinnati Children’s Hospital Medical Center (CCHMC); Director, Cincinnati Center for Eosinophilic Disorders, CCHMC; Professor, University of Cincinnati Department of Pediatrics
Session 1: How Care and Research Have Changed During COVID-19

The COVID-19 pandemic brought about uncertainty for all, especially for the rare diseases community where health care expertise and resources are already scarce. In this session, panelists will discuss how the rare diseases community navigated this challenge, including how health care and research in the rare diseases community changed amid the COVID-19 pandemic. Panelists include patients, patient organizations and researchers.

Moderator: Kristen Wheeden, Co-Chair, RDCRN Coalition of Patient Advocacy Groups (CPAG) Steering Committee; Executive Director, American Porphyria Foundation

Panelists:
- Robert A. (Sandy) Sandhaus, M.D., Ph.D., Division of Pulmonary, Critical Care and Sleep Medicine, National Jewish Health
- Tracy Hart, Chair, RDCRN-CPAG Steering Committee; CEO, Osteogenesis Imperfecta Foundation
- Rachel L. Sher, J.D., M.P.H., Vice President of Regulatory and Government Affairs, National Organization for Rare Disorders (NORD)

1:40 p.m. Lunch Break
- Virtual Posters and Exhibits
- Art Exhibition and Videos

2:10 p.m. Session 2: Using Health Literacy to Elevate the Rare Diseases Community and Enhance Research

Moderator: TBD

Panelists: TBD

3:10 p.m. Rare Story #3: How Genomics Education Helps Health Care Providers Help Patients

The Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)
Rare Story #4
Speaker:
- Jennifer A. Doudna, Ph.D., Nobel Laureate, The Nobel Prize in Chemistry 2020; Professor of Chemistry, Professor of Biochemistry and Molecular Biology, University of California, Berkeley

Rare Story #5: “Meet the NIH Investigator” — A Virtual Tour
Speakers:
- Carsten G. Bönnemann, M.D., Senior Investigator, National Institute of Neurological Disorders and Stroke (NINDS), NIH
- Charles P. Venditti, M.D., Ph.D., Senior Investigator, National Human Genome Research Institute (NHGRI), NIH

3:55 p.m.  Break

4:10 p.m.  Session 3: From Kitchen Tables to Changing Paradigms — Advocacy as a Driving Force in Advancing Progress in Rare Diseases Research
Advocacy is playing an increasingly important role in accelerating progress in understanding and treating rare diseases. Many rare disease advocacy organizations got their start with one or two patients or caregivers who had a goal of finding treatments and have grown their efforts into organizations that are changing the rate at which rare diseases research is progressing. In this session, panelists will discuss their experiences and lessons learned in establishing advocacy foundations focused on various rare cancers or on running clinical trials while working closely with advocates. Exciting recent progress toward identifying new treatments will be highlighted.

Moderator: Abby B. Sandler, Ph.D., Executive Director, My Pediatric and Adult Rare Tumor (MyPART) Network, Pediatric Oncology Branch, Center for Cancer Research, National Cancer Institute (NCI), NIH

Panelists:
- Jeanne Whiting or Marlene Portnoy, Co-Founder, Desmoid Tumor Research Foundation
- Laneen (Lennie) Woods, Co-Founder and Executive Director, Sara’s Cure
- Jim Palma, Executive Director, TargetCancer Foundation
- Josh Sommer, Co-Founder and Executive Director, Chordoma Foundation
- Timothy Babich, Co-Founder and Director, the RUNX1 Research Program
- Eric J. Sherman, M.D., Medical Oncologist, Memorial Sloan Kettering Cancer Center
5:20 p.m.  **Closing Remarks**  
Christopher P. Austin, M.D., Director, NCATS, NIH

5:30 p.m.  **Adjournment**