RARE DISEASE DAY at NIH
March 1, 2021 | #RDDNIH
ncats.nih.gov/rdd

Final Agenda

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

Pius A. Aiyelawo, M.P.A., FACHE, Chief Operating Officer, Clinical Center, NIH

10:55 a.m. **Congressional Remarks from the United States (U.S.) Senate**
Sen. Roger F. Wicker (R-MS), Senate Co-Chair, Rare Disease Congressional Caucus

11:00 a.m. **NIH Director Remarks**
Francis S. Collins, M.D., Ph.D., Director, NIH

11:10 a.m. **Congressional Remarks from the U.S. House of Representatives**
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

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

11:35 a.m. **Rare Story #2: Industry and Patient Advocacy Collaborations — Making It a Win-Win**
Moderator:
- Shazia Ahmad, Senior Director, Patient and Physician Services, United BioSource LLC (UBC); Rare Disease Thought Leader, Patient and Stakeholder Engagement Expert

Panelists:
- Nadia Bodkin, Pharm.D., M.S., Rare Disease Patient Advocate, Rare Advocacy Movement (RAM)
- Christopher U. Missling, M.S., Ph.D., M.B.A., President and Chief Executive Officer (CEO), Anavex Life Sciences Corp.

11:50 a.m. **Rare Story #3: Hope in Gene Therapy**
Introduction:
- Cynthia J. Tifft, M.D., Ph.D., Deputy Clinical Director, Office of the Clinical Director, National Human Genome Research Institute (NHGRI), NIH

Speakers:
- Niclas Flysjö, Father of three young rare disease patients with GM1 gangliosidosis, Advocate
- Jessica Flysjö, Mother of three young rare disease patients with GM1 gangliosidosis, Advocate

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:

- Thomas W. Ferkol, M.D., Co-Principal Investigator, Genetic Disorders of Mucociliary Clearance Consortium (GDMCC), RDCRN; Alexis Hartmann Professor of Pediatrics, Professor of Cell Biology and Physiology, Washington University School of Medicine in St. Louis
- 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

12:45 p.m. **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, M.B.A., Co-Chair, RDCRN Coalition of Patient Advocacy Groups (CPAG) Steering Committee; Executive Director, American Porphyria Foundation

Panelists:

- Lisa Kosak, Rare Disease Patient, Alpha-1 Antitrypsin Deficiency
- Robert A. (Sandy) Sandhaus, M.D., Ph.D., FCCP, Professor of Medicine, Division of Pulmonary, Critical Care and Sleep Medicine, National Jewish Health; Medical Director, Alpha-1 Foundation, AlphaNet, and AlphaNet Canada
- Tracy Hart, Chair, RDCRN-CPAG Steering Committee; CEO, Osteogenesis Imperfecta Foundation
- Rachel L. Sher, J.D., M.P.H., Vice President, Policy and Regulatory Affairs, National Organization for Rare Disorders (NORD)

1:45 p.m. **Break**

- Virtual Exhibits and Scientific Posters
- Art Exhibition and Videos
- Networking

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

Rare diseases patients, families and advocates are well aware of how difficult it can be to find, understand and use health information to inform decisions and actions for themselves and others. This is a concept known as health literacy. Often, the burden for gaining access and understanding health information is placed on individual patients and families. Improved health literacy in a rare disease community can be an influential factor in promoting access to appropriate medical care, ensuring adequate patient quality of life and laying the groundwork for successful clinical trials. In this session, panelists will discuss how to elevate health literacy and why it is important to integrate health literacy raising activities into the rare diseases community through communication and partnerships. Panelists include patients, patient organizations and clinicians/researchers.

Moderator: Christen Sandoval, M.S., CHES, Public Health Specialist, Office of Communications and Public Liaison, Office of the Director (OD), NIH

Panelists:

- Eric W.K. Sid, M.D., M.H.A., Program Officer, ORDR, NCATS, NIH
- Laura M. Koehly, Ph.D., Senior Investigator, Social and Behavioral Research Branch, NHGRI, NIH
- Luke Rosen, M.S., Founder and Board Chair, KIF1A.ORG
- Tracy Dixon-Salazar, Ph.D., Executive Director, Lennox-Gastaut Syndrome (LGS) Foundation

3:10 p.m. **Rare Story #4: Novel Rare Diseases Course for Undergraduate Freshmen — Successes and Lessons Learned**

Reena Kartha, M.S., Ph.D., Assistant Professor, Associate Director of Translational Pharmacology, Center for Orphan Drug Research, University of Minnesota

3:20 p.m. **Rare Story #5: Partnership for Educating Health Care Providers on Rare Diseases**

Michelle Snyder, M.S., CGC, Rare Diseases Project Group Lead, Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

3:25 p.m. **Rare Story #6: CRISPR Genome Editing — Rewriting the Future of Health**

Introduction:

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

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

3:40 p.m. **Rare Story #7: The Platform Vector Gene Therapy (PaVe-GT) Program — Meet the Investigators**

Introduction:

- Anne R. Pariser, M.D., Director, ORDR, NCATS, NIH

Speakers:

- Carsten G. Bönnemann, M.D., Senior Investigator, Neuromuscular and Neurogenetic Disorders of Childhood Section, National Institute of Neurological Disorders and Stroke (NINDS), NIH
- Charles P. Venditti, M.D., Ph.D., Senior Investigator, Organic Acid Research Section, 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 Sandler, Ph.D., Executive Director, My Pediatric and Adult Rare Tumor (MyPART) Network, Pediatric Oncology Branch, Center for Cancer Research, NCI, NIH

Panelists:

- Lennie Woods, Co-Founder and Executive Director, Sara's Cure
- Jim Palma, Executive Director, TargetCancer Foundation
- Jeanne Whiting, Co-Founder, Desmoid Tumor Research Foundation
- Timothy Babich, Co-Founder and Director, the *RUNX1* Research Program
- Josh Sommer, Co-Founder and Executive Director, Chordoma Foundation
- 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**