Preliminary Agenda
As of 12/13/2022

8:00 a.m. EST  Registration and Exhibit Booth/Poster Setup

9:00 a.m.  Rare Disease Day at NIH Overview
Alice Chen Grady, M.D., Program Officer, Division of Rare Diseases Research Innovation (DRDRI), National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH)
Ainslie Tisdale, Program Analyst, DRDRI, NCATS, NIH

9:10 a.m.  Welcoming Remarks
Joni L. Rutter, Ph.D., Director, NCATS, NIH
James K. Gilman, M.D., CEO, Clinical Center, NIH

9:50 a.m.  Session 1: NCATS Rare Diseases Programs and Resources
Presentations:
- **DRDRI Update**
  Philip John (P.J.) Brooks, Ph.D., Acting Director, DRDRI, NCATS, NIH
- **Genetic and Rare Diseases (GARD) Information Center Update**
  Eric W.K. Sid, M.D., M.H.A., Program Officer, DRDRI, NCATS, NIH
- **Rare Disease Alert System**
  Qian Zhu, Ph.D., Team Lead, Rare Disease Translational Research, Informatics Core, Division of Preclinical Innovation (DPI), NCATS, NIH

Q&A will follow the presentations.

10:50 a.m.  The NIH Clinical Center’s Resources for Clinical Trials
Nikita Curry, M.H.A., Supervisor, Office of Patient Recruitment, Clinical Center, NIH

11:05 a.m.  The Children’s Inn and Resources for Patients and Families
Aisha Dubose Campbell, Director, Resident Services and Family Programming, The Children’s Inn at NIH

11:20 a.m.  Break
- Exhibits and Scientific Posters
- Art Exhibition
- Networking
Session 2: Meeting the Needs of Adolescent and Young Adult (AYA) Rare Disease Patients

AYAs with rare diseases face unique challenges, especially if they are diagnosed during their adolescent years. Adolescents with rare diseases, including cancer, do not neatly fit into either pediatric or adult care settings. This session will highlight work being done by AYA cancer advocacy groups and researchers as a model to improve the experience for adolescents facing rare diseases.

Moderator: Abby 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:
- Lori Wiener, Ph.D., DCSW, LCSW-C, Senior Associate Scientist, Pediatric Oncology Branch, Center for Cancer Research, NCI, NIH
- Hilary Gan, Director, Hospital Programs and Services, Teen Cancer America, Inc.
- Alison Silberman, CEO, Stupid Cancer, Inc.

Rare Story #1

Abbey Hauser, Young Adult Rare Disease Advocate

Rare Story #2

Cary O. Harding, M.D., Principal Investigator, Phenylalanine Families and Researchers Exploring Evidence (PHEFREE) Consortium, Rare Diseases Clinical Research Network (RDCRN); Professor, Molecular and Medical Genetics, School of Medicine, Oregon Health & Science University

Brittany M. Holmes, M.S.N., APRN, FNP-BC, Rare Disease Patient; Advisor, PHEFREE Consortium, RDCRN; Metabolism Nurse Practitioner, Division of Genetics and Genomics, Boston Children’s Hospital

Lunch (on your own)

- Exhibits and Scientific Posters
- Art Exhibition
- Networking

Session 3: Genetics, Gene-Targeted Therapies, and Diversity, Equity, Inclusion, and Accessibility

Moderator: Deanna Portero, Management Analyst, DRDRI, NCATS, NIH

Panelists:
- Tshaka J. Cunningham, Ph.D., Executive Director, Faith-Based Genetic Research Institute; Program Director, Biomedical Sciences Program, Future Kings; Co-Founder and Chief Scientific Officer, Polaris Genomics
• Mario A. Estevez, Parent of a Rare Disease Patient; Board Member, Project Alive; Organizer and Trainer, The Racial Equity Institute, LLC
• Shengdar Q. Tsai, Ph.D., Associate Member, Department of Hematology, St. Jude Children’s Research Hospital; Co-Chair, Steering Committee, NIH Somatic Cell Genome Editing Program

2:30 p.m. Rare Story #3
Marcela A. Ferrada, M.D., Staff Clinician, Vasculitis Translational Research Program, National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIH
Michael Linn, Director and Vice Chair, Relapsing Polychondritis Foundation

2:50 p.m. Break
• Exhibits and Scientific Posters
• Art Exhibition
• Networking

3:10 p.m. Rare Story #4: Building Your Preclinical Toolbox
Pangkong Fox, Ph.D., Science Engagement Director, CACNA1A Foundation, Inc.

3:25 p.m. Rare Story #5
Nasha Fitter, M.B.A., Co-Founder and CEO, FOXG1 Research Foundation; Vice President, Real-World Evidence and Ciitizen Platform, Invitae

3:40 p.m. Rare Story #6
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
Ellyn Kodroff, Coalition of Patient Advocacy Groups (CPAG) Representative, CEGIR, RDCRN; President and Founder, Campaign Urging Research for Eosinophilic Disease (CURED)

4:00 p.m. Session 4: Rare Diseases Therapeutics and the Role of Advocacy and Industry Collaborations
This session will explore the spectrum of advocacy–industry collaborations across the lifecycle of therapeutic development and share how patient advocacy can play an important role in research efforts. Examples include such efforts as spurring new therapeutic development, improving the efficiency of clinical development, increasing engagement and participation in clinical trials, informing the design of clinical trials, and assessing the meaningfulness of clinical outcomes for regulatory purposes. Panelists from patient advocacy and industry
will encourage participation in research and inspire hope for future treatments in more rare diseases.

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

**Panelists:**
- *Sharon J. King*, Manager, Advocacy and Community Engagement, Guanosine Monophosphate (GMP) Nucleic Acids Business Unit, Aldevron
- *Kristen Wheeden, M.B.A.*, Rare Disease Caregiver; CPAG Representative, Porphyrias Consortium, RDCRN; President, United Porphyrias Association
- *Jennifer Beck*, Erythropoietic Protoporphyrria Patient and Advocate

4:45 p.m. **Closing Remarks**

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

5:00 p.m. **Adjournment**