



RARE DISEASE DAY at NIH

Feb. 28, 2023 | #RDDNIH
ncats.nih.gov/rdd

Final Agenda

- 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
- 9:20 a.m. **NIH Leadership Remarks via Video**
Lawrence A. Tabak, D.D.S., Ph.D., Performing the Duties of the NIH Director
- 9:30 a.m. **Rare Disease Congressional Caucus Remarks**
Sen. Roger F. Wicker (R-MS), Senate Co-Chair [video]
Rep. Gus M. Bilirakis (R-FL), House Co-Chair [video]
Rep. Doris O. Matsui (D-CA), House Co-Chair [invited; video]
Sen. Amy J. Klobuchar (D-MN), Senate Co-Chair [invited]
- 9:50 a.m. **Session 1: NCATS Rare Diseases Programs and Resources**
- **Overview**
Philip John (P.J.) Brooks, Ph.D., Acting Director, DRDRI, NCATS, NIH
 - **Genetic and Rare Diseases (GARD) Information Center**
Eric W.K. Sid, M.D., M.H.A., Program Officer, DRDRI, NCATS, NIH
 - **RARE-SOURCE: Integrated Bioinformatics Resource for Rare Diseases**
Elizabeth A. Ottinger, Ph.D., Acting Director, Therapeutic Development Branch (TDB), Division of Preclinical Innovation (DPI), NCATS, NIH
Uma Mudunuri, M.S., Director, Advanced Biomedical Computational Science, Frederick National Laboratory for Cancer Research
 - **Rare Disease Alert System**
Qian Zhu, Ph.D., Team Lead, Rare Diseases Translational Research, Informatics Core, DPI, NCATS, NIH
 - **Platform Vector Gene Therapy (PaVe-GT) Program**
Richa Madan Lomash, Ph.D., Scientific Project Manager, TDB, DPI, NCATS, NIH
- Q&A will follow the presentations.*



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- 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](#)
- 11:40 a.m. **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 cancer or other 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:
- *Hilary Gan, M.A.*, Director, Hospital Programs and Services, Teen Cancer America, Inc.
 - *Alison Silberman*, CEO, Stupid Cancer, Inc.
 - *Lori Wiener, Ph.D., DCSW, LCSW-C*, Senior Associate Scientist, Pediatric Oncology Branch, Center for Cancer Research, NCI, NIH
- 12:20 p.m. **Rare Story #1: Becoming the Captain — My Experience Transitioning From Pediatric to Adult Health Care**
Abbey Hauser, Young Adult Rare Disease Advocate
- 12:35 p.m. **Rare Story #2: Care Transition Challenges for Young Adults With Rare Diseases — A Patient and Provider Perspective**
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



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12:50 p.m.

Lunch ([on your own](#))

- [Exhibits and Scientific Posters](#)
- [Art Exhibition](#)
- [Networking](#)

1:30 p.m.

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

Genetic technologies and gene-targeted therapies hold immense promise for the diagnosis and treatment of rare diseases, but they also have the potential to become a new frontier for inequity and health disparities. In addition to the common drivers of inequity and health disparities found in other fields, a number of social, economic and technical issues are unique to or disproportionately affect gene-targeted therapies. The speakers on this panel will discuss obstacles to realizing the benefits of gene-targeted technologies in an equitable way across diverse groups and the efforts required to prevent inequity and health disparities in this relatively new field.

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
- *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
- *Mario A. Estevez*, Rare Disease Caregiver; Board Member and International Ambassador, Project Alive; Organizer and Trainer, Racial Equity Institute

2:30 p.m.

Rare Story #3: Where There Is Research, There Is Hope

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 — The Power of Your Patient Community

Pangkong Fox, Ph.D., Science Engagement Director, CACNA1A Foundation, Inc.; Rare Disease Caregiver



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- 3:25 p.m. **Rare Story #5: Creating Your Natural History Study**
Nasha Fitter, M.B.A., Rare Disease Caregiver; Co-Founder and CEO, FOXG1 Research Foundation; Vice President, Real-World Evidence and Ciitizen Platform, Invitae
- 3:40 p.m. **Rare Story #6: Scientific and Patient Journey to the First U.S. Food and Drug Administration–Approved Drugs for Two Rare Eosinophilic Diseases**
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 life cycle 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 (UBC)
Panelists:
 - *Sharon J. King*, Manager, Advocacy and Community Engagement, Aldevron
 - *Sandra Abrevaya, J.D.*, Rare Disease Caregiver; Co-Founder, I AM ALS
 - *Brian Wallach, J.D.*, Rare Disease Patient; Co-Founder, I AM ALS
 - *Kristen Wheeden, M.B.A.*, Rare Disease Caregiver; CPAG Representative, Porphyrias Consortium, RDCRN; President, United Porphyrias Association
 - *Jennifer Beck*, Erythropoietic Protoporphyrin Patient and Advocate
- 4:45 p.m. **Closing Remarks**
Joni L. Rutter, Ph.D., Director, NCATS, NIH
- 5:00 p.m. **Adjournment**