RARE DISEASE DAY at NIH
Feb. 29, 2024 | #RDDNIH
ncats.nih.gov/rdd

Preliminary Agenda
As of 1/8/2024

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

9:00 a.m. **Rare Disease Day at NIH Overview**
Meera A. Shah, M.P.H., Program Analyst, Division of Rare Diseases Research Innovation (DRDRI), National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH)
Ainslie Tisdale, M.P.H., 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

9:40 a.m. **NCATS DRDRI Update**
Dominique C. Pichard, M.D., M.S., Director, DRDRI, NCATS, NIH

9:50 a.m. **Rare Story #1: Shortening the Diagnostic Odyssey—Project GIVE (Genetic Inclusion by Virtual Evaluation) for the Rio Grande Valley**
Seema R. Lalani, M.D., Professor, Department of Molecular and Human Genetics, Baylor College of Medicine

10:10 a.m. **Rare Story #2: A Typical Normal—Finding Our Stride Through Exceptional Times**
Jessica Swanson, M.A., BCBA, QBA, LBA, CAS, Rare Disease Caregiver; Co-Chair, Undiagnosed Diseases Network Participant Engagement and Empowerment Resource

10:25 a.m. **Break**
- Exhibits and Scientific Posters
- Art Exhibition and Films
- Networking

10:55 a.m. NIH Director Remarks
Monica M. Bertagnolli, M.D., Director, NIH

11:05 a.m. **Session 1: Artificial Intelligence (AI) and Its Potential Role in Rare Diseases**
Moderator: *Christine Cutillo, M.M.C.i.,* Health Data Scientist for AI Ethics, Office of Data Science Strategy, Office of the Director, NIH
Panelists:
- *Kimberly A. Moran, Ph.D., M.B.A., CDP,* Head, U.S. Rare Diseases, UBC, Inc.

- *Manisha Balwani, M.D., M.S., FACMG,* Professor and Chief, Division of Medical Genetics and Genomics, Department of Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai
- *Ewy A. Mathé, Ph.D.,* Director, Informatics Core, Division of Preclinical Innovation, NCATS, NIH

Q&A will follow the panelist presentations.

12:05 p.m. **Rare Story #3: Microphysiological Systems as Applied for Rare Diseases Therapeutic Development**

James J. Hickman, Ph.D., Professor, Chemistry, University of Central Florida; Co-Founder and Chief Scientific Officer, Hesperos, Inc.

12:25 p.m. **Rare Story #4: Repurposing With Purpose—From Discovery to Rapid Treatment of Bachmann-Bupp Syndrome**

André S. Bachmann, Ph.D., M.S., Professor and Associate Chair for Research, Scientific Director of the International Center for Polyamine Disorders, Department of Pediatrics and Human Development, College of Human Medicine, Michigan State University

Caleb P. Bupp, M.D., FACMG, Division Chief, Medical Genetics and Genomics, Corewell Health and Helen DeVos Children's Hospital; Assistant Professor, Department of Pediatrics and Human Development, College of Human Medicine, Michigan State University; Clinical Director, International Center for Polyamine Disorders

12:45 p.m. **Lunch (on your own)**

- Exhibits and Scientific Posters
- Art Exhibition and Films
- Networking

2:00 p.m. **Rare Story #5: Authenticity and Vulnerability in Storytelling**

Ashley Eakin, Rare Disease Patient, Advocate, Film Director, and Writer

2:20 p.m. **Rare Story #6: Wolfram Syndrome: Diagnosis, Clinical Protocols, and Advances in Gene Editing, Regenerative, and Molecular Targeted Therapies**

Fumihiko Urano, M.D., Ph.D., FACMG, Samuel E. Schechter Professor of Medicine, Director of Wolfram Syndrome and Related Disorders Clinic, Division of Endocrinology, Metabolism, and Lipid Research, Washington University School of Medicine in St. Louis

2:40 p.m. **Rare Story #7**

3:00 p.m. **Break**

- Exhibits and Scientific Posters

- Art Exhibition and Films
- Networking

3:30 p.m. **Rare Story #8**
Moderator: *Dominique C. Pichard, M.D., M.S.,* Director, DRDRI, NCATS, NIH
Speakers:
- *Eric P. Hoffman, Ph.D.,* Associate Dean of Research and Research Development, Professor of Pharmaceutical Sciences, School of Pharmacy and Pharmaceutical Sciences, Binghamton University State University of New York; President and CEO, ReveraGen BioPharma, Inc.
- *Sharon Hesterlee, Ph.D.,* Chief Research Officer, Muscular Dystrophy Association

3:45 p.m. **Session 2: Recent Approvals and Advancements in Gene Therapy**
Introduction and Moderator: *Philip John (P.J.) Brooks, Ph.D.,* Deputy Director, DRDRI, NCATS, NIH
Presentations:
- **Gene Therapy for Patients With Dystrophic Epidermolysis Bullosa**
  M. Peter Marinkovich, M.D., Associate Professor, Department of Dermatology, Stanford Medicine
- **Gene Therapy for Patients With Cerebral Adrenoleukodystrophy**
  Florian S. Eichler, M.D., Director, Center for Rare Neurological Diseases, Massachusetts General Hospital; Professor, Neurology, Harvard Medical School
- **Gene Therapies for Patients With Sickle Cell Disease**
  Q&A will follow each presentation.

4:45 p.m. **Closing Remarks**
Joni L. Rutter, Ph.D., Director, NCATS, NIH

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