Preliminary Agenda
As of 2/9/2024

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**
This session will begin with an introduction to Artificial Intelligence (AI) and how it can be used to benefit rare disease patients. Attendees will then learn about multiple case studies exemplifying the role AI has played in reducing the diagnostic odyssey, identifying rare disease patients for clinical trials, and building tools to repurpose

drugs. Presentations will be followed by a moderated Q&A with the panelists, paying particular attention to patient privacy and ethical concerns.

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: Gene Therapy Development in Industry: Past, Present, and Future**
Meredith Schultz, M.D., M.S., SVP, Clinical Development and Medical Affairs, Taysha Gene Therapies

3:00 p.m. **Break**
- Exhibits and Scientific Posters
- Art Exhibition and Films
- Networking

3:15 p.m. **Rare Story #8: Vamorolone: How a Public-Private Partnership Succeeded in Bringing a Rare Disease Drug to Market**
Moderator: *Dominique C. Pichard, M.D., M.S.,* Director, DRDRI, NCATS, NIH
Speakers:
- *Eric P. Hoffman, Ph.D.,* Professor of Pharmaceutical Sciences, Binghamton University - State University of New York; CEO, ReveraGen BioPharma, Inc.
- *Sharon Hesterlee, Ph.D.,* Chief Research Officer, Muscular Dystrophy Association

3:35 p.m. **Session 2: Recent Approvals and Advancements in Gene Therapy**
This session will begin with a brief introduction and overview of genetic therapies. Attendees will then hear from the director of FDA's Center for Biologics Evaluation Research - Office of Therapeutic Products on new initiatives and recent guidances related to gene therapies. Following these remarks, presenters will describe the path to approval for three recent approvals of genetic therapies for rare diseases, highlighting potential clinical impact, relevance for similar diseases, remaining challenges and concerns, as well as patient perspectives.
Introduction and Moderator: *Philip John (P.J.) Brooks, Ph.D.,* Deputy Director, DRDRI, NCATS, NIH
Presentations:
- **FDA Rare Disease Gene Therapy Initiatives**
  Nicole Verdun, M.D., Super Office Director, Office of Therapeutic Products, Center for Biologics Evaluation and Research (CBER), The U.S. Food and Drug Administration (FDA)
- **Gene Therapy for Patients With Dystrophic Epidermolysis Bullosa**
  M. Peter Marinkovich, M.D., Associate Professor, Department of Dermatology, Stanford Medicine
  Aaron Owens, Rare Disease Patient
- **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**
  Alexis K. Leonard, M.D., Assistant Member, Department of Hematology, St. Jude Children's Research Hospital
  Nasir Mason, Patient Perspective Advocate for Sickle Cell Disease

Q&A will follow each presentation.

4:50 p.m. **Closing Remarks**
Joni L. Rutter, Ph.D., Director, NCATS, NIH

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