Preliminary Agenda as of 02/12/2025

February 27, 2025

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

9:00 a.m. **Registration and Exhibit Booth/Poster Viewing**

10:00 a.m. **FDA-NIH Rare Disease Day 1 Overview**

10:10 a.m. **Welcoming Remarks: Day 1**
Joni L. Rutter, Ph.D., Director, National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH)

10:20 a.m. **NIH Acting Director Remarks**
Matthew J. Memoli, M.D., M.S., Acting Director, NIH

10:30 a.m. **FDA Acting Commissioner Remarks**
Sara Brenner, M.D., M.P.H., Acting Commissioner, U.S. Food and Drug Administration (FDA)

10:40 a.m. **FDA Update**
Amy Comstock Rick, J.D., Director of Strategic Coalitions, Rare Disease Innovation Hub, FDA

10:50 a.m. **NCATS Division of Rare Diseases Research Innovation (DRDRI) Update**
Dominique C. Pichard, M.D., M.S., Director, DRDRI, NCATS, NIH

11:00 a.m. **Session 1: Artificial Intelligence — Clinical and Regulatory Applications for Rare Diseases**
Artificial intelligence (AI) is revolutionizing how rare diseases challenges are addressed in research and regulation. This session introduces AI principles and tools and discusses their growing impact within the rare diseases ecosystem and limitations of use. Attendees will gain foundational knowledge and learn about AI use across drug development and relevant applications for the rare diseases community. Presenters will then delve into two case studies that demonstrate AI use in rare diseases research.
Moderator: Annie Saha, Associate Director for Strategic Initiatives, Digital Health Center of Excellence, FDA
Panelists:

- Tala Fakhouri, Ph.D., M.P.H., Associate Director for Data Science and AI Policy, FDA
- Nara Sobreira, M.D., Ph.D., Associate Professor in the McKusick-Nathans Department of Genetic Medicine, Associate Professor of Pediatrics, Johns Hopkins University

- Vivek Rudrapatna, M.D., Ph.D., Co-director, UCSF Center for Real World Evidence; Gastroenterologist, UCSF Health

12:00 p.m. **Lunch (on your own)**

- Exhibits and Scientific Posters
- Art Exhibition
- Networking

1:50 p.m. **Rare Resource: Getting a Genetic Test**
Julie Sapp, Sc.M., CGC, Genetic Counselor, Precision Genomics Section, National Human Genome Research Institute, NIH

2:05 p.m. **Rare Story: Corrective Genome Editing to Treat Hereditary Connective Tissue Disorders — A Patient-Scientist Perspective**
Lauren Testa, Ph.D. Candidate, Cell and Molecular Biology (Gene Therapy and Vaccines), University of Pennsylvania School of Medicine

2:20 p.m. **Break**

- Exhibits and Scientific Posters
- Art Exhibition
- Networking

2:45 p.m. **Session 2: Gene Therapy — Recent Approvals, Success Stories and Patient Perspectives**
Gene therapy continues to transform the treatment landscape for rare diseases, offering hope to patients and families. This session will explore recent FDA gene therapy approvals and NCATS' involvement with a recent gene therapy approval for aromatic l-amino acid decarboxylase (AADC) deficiency. Attendees also will engage in a thoughtful discussion about patient considerations when pursuing gene therapy.

Moderators:

- Philip John (P.J.) Brooks, Ph.D., Deputy Director, DRDRI, NCATS, NIH
- Peter Marks, M.D., Ph.D., Director, Center for Biologics Evaluation and Research (CBER), FDA

Panelists:

- Nicole Verdun, M.D., Super Office Director, Office of Therapeutic Products, CBER, FDA
- Elizabeth A. Ottinger, Ph.D., Director, Therapeutics Development Branch, Division of Preclinical Innovation, NCATS, NIH
- Jodi A. Cook, Ph.D., President, Chief Executive Officer (CEO), Skylark Bio, Inc.
- Kenneth Hobby, President, Cure SMA

3:45 p.m. **Overview Remarks: Day 1**
Joni L. Rutter, Ph.D., Director, NCATS, NIH

4:00 p.m. **Adjournment**

Preliminary Agenda as of 02/12/2025

February 28, 2025

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

9:00 a.m. **Registration and Exhibit Booth/Poster Viewing**

10:00 a.m. **FDA-NIH RDD Day 2 Overview**

10:10 a.m. **Welcoming Remarks: Day 2**
Hilary Marston, M.D., M.P.H., Chief Medical Officer, FDA

10:20 a.m. **Rare Resource: Brain and Tissue Donation — Why It Matters**
Tish Hevel, CEO, Founder, The Brain Donor Project

10:40 a.m. **Session 3: Developing Treatments Through Innovative Trial Designs for Rare Diseases**
Innovative clinical trial designs are paving the way for more efficient and effective drug development, especially for complex and rare diseases. This session will introduce the topic of innovative trial designs, and presenters will give an overview of two case studies of current ongoing innovative clinical trials. This session will also discuss challenges regarding innovative trial designs.
Moderator and Introduction: Kerry Jo Lee, M.D., Associate Director for Rare Diseases; Division of Rare Diseases and Medical Genetics; Office of Rare Diseases, Pediatrics, Urologic and Reproductive Medicine; Office of New Drugs; Center for Drug Evaluation and Research (CDER); FDA
Panelists:

- Rebecca (Becky) Chiu, Ph.D., Supervisory Mathematical Statistician, Division of Biometrics IV, Office of Biostatistics, CDER, FDA
- Troy Torgerson, M.D., Ph.D., Director of Experimental Immunology, Allen Institute for Immunology
- Malcolm A. Smith, M.D., Ph.D., Associate Branch Chief, Pediatrics, Clinical Investigations Branch, Cancer Therapy Evaluation Program, Division of Cancer Treatment and Diagnosis, National Cancer Institute, NIH
- Martha Donoghue, M.D., Associate Director of Pediatric Oncology and Rare Cancers, Oncology Center of Excellence, FDA

11:40 a.m. **Rare Story: Neonates and Rare Diseases — Why Early Diagnosis and Approved Treatments Are Imperative**
An Massaro, M.D., Supervisory Medical Officer, Neonatology and Rare Pediatric Disease Team, Office of Pediatric Therapeutics, Office of the Chief Medical Officer, Office of the Commissioner, FDA

12:00 p.m. **Lunch (on your own)**

- Exhibits and Scientific Posters
- Art Exhibition
- Networking

1:30 p.m. **Session 4: Ensuring Access Across the Rare Diseases Treatment Pipeline**
This session will explore the critical stages of the rare diseases treatment pipeline and discuss strategies for ensuring equitable and efficient access at different phases. Panelists will discuss innovative approaches for encouraging investment in rare diseases research, addressing barriers to clinical trial participation and treatment access, and exploring sustainable payment models for groundbreaking therapies.
Moderator and Introduction: Julie Tierney, J.D., Deputy Center Director, CBER, FDA
Panelists:

- Adam Hartman, M.D., Program Director, Division of Clinical Research, National Institute of Neurological Disorders and Stroke, NIH
- Jenifer Waldrop, M.S., Executive Director, Rare Disease Diversity Coalition, Black Women's Health Imperative
- Jack Rollins, M.P.H., Director of Federal Policy, National Association of Medicaid Directors

2:30 p.m. **Rare Story: The Full-Time Job of Living Rare — Advocacy, Resilience and the Fight for Care**
Lindsay Guentzel, Multimedia Journalist, Storyteller, Advocate

2:45 p.m. **Break**

- Exhibits and Scientific Posters
- Art Exhibition
- Networking

3:15 p.m. **Rare Story: Conquering the Unconquerable — Turning Hope Into Progress Through Research**
Jake Juip, Rare Disease Advocate and College Student

3:30 p.m. **Rare Resource: RAREly Told Stories — How to Share Your Rare Disease Experience**
Daniel DeFabio, Menkes Disease Dad, Director of Community Engagement and Education at Global Genes, Co-founder of The Disorder Channel

3:45 p.m. **Closing Remarks**
Joni L. Rutter, Ph.D., Director, NCATS, NIH

4:00 p.m. **Adjournment**