Rare Disease Scientific Symposium

NORD® National Organization for Rare Disorders

Advancing Innovation in Rare Disease Research

Agenda subject to change. * All times are ET

MONDAY, JUNE 2, 2025

8 a.m.	**Check-in and Continental Breakfast**
9 a.m.	**NORD's Welcome & Opening Remarks**

Innovative Study Designs and Methods for Rare Disease Research

Section Chair: P.J. Brooks, PhD, Deputy Dir, DRDRI, NCATS, **National Institutes of Health**

9:10 a.m. — **Leveraging Innovative Trial Designs and Technologies to Accelerate Research**

In this session, speakers will explore cutting-edge clinical trial strategies — such as basket trials, platform trials, and gene therapy platforms — that offer scalable, efficient approaches that may apply to multiple rare disease states. Presenters will share real-world examples and discuss the impact of these designs on accelerating research and regulatory pathways.

SPEAKERS:

Lisa Forbes Satter, MD, Associate Professor of Pediatrics, Section of Immunology, Allergy and Rheumatology, **Baylor College of Medicine and Texas Children's Hospital, a NORD Rare Disease Center of Excellence**

Kiran Musunuru, MD, PhD, MPH, ML, MRA, Professor of Cardiovascular Medicine, Genetics, and Pediatrics, Perelman School of Medicine at the University of Pennsylvania, **a NORD Rare Disease Center of Excellence**

Scott Plotkin, MD, PhD, Giovanni Armenise Endowed Professor of Neurology, Harvard Medical School, Chief, Division of Neuro-Oncology, **Mass General Brigham, Harvard Medical School, a NORD Rare Disease Center of Excellence**

Kevin S. Thorneloe, PhD, Senior Medical Director, **Pharming Healthcare**

10:10 a.m. — **Approaches to Address Challenges of Small Trial Enrollment Numbers and Disease Heterogeneity**

This session will highlight statistical, operational, and regulatory approaches for overcoming barriers related to small populations and phenotypic variability in rare disease trials. Experts from FDA, academia, and industry will share strategies and experiences to inform trial design across rare diseases.

SPEAKERS:

Kerry Jo Lee, MD, Associate Director of Rare Diseases and Lead, Accelerating Rare Disease Cures (ARC) Program, **Center for Drug Evaluation and Research, U.S. Food and Drug Administration (FDA)**

Rebecca Rothwell Chiu, PhD, Supervisory Mathematical Statistician, **Center for Drug Evaluation and Research, FDA**

John Scott, PhD, Director, Division of Biostatistics, **Center for Biologics Evaluation and Research (CBER), FDA**

Nicole Verdun, MD, Super Office Director, Office of Therapeutic Products, **CBER**, **FDA**

11:15 a.m.	**Networking Break**

MONDAY, JUNE 2, 2025 (continued)

11:30 a.m.

Global Trials: Successes and Challenges

Experts will discuss the complexities of conducting international rare disease trials, from regulatory harmonization to operational logistics. Presenters will provide insights into successful global collaborations and lessons learned from multinational trial designs.

SPEAKERS:

Nancy Bolous, MD, MA, MSc, Senior Research Scientist, **St. Jude Children's Research Hospital**

Maurizio Scarpa, MD, PhD, Director, **European Reference Network for Hereditary Metabolic Diseases (MetabERN)**

Anabela Marçal, PharmD, EMA Liaison Official to US FDA, **European Medicines Agency**

12:30 p.m.

Networking Lunch

1:35 p.m.

On the Ground Perspectives: Rare Disease Clinical Trial Principal Investigators

Clinical trialists will share firsthand experiences designing and running rare disease trials. This session offers a candid look at successes and obstacles in real-world trial execution.

SPEAKERS:

Patricia Musolino, MD, PhD, Associate Professor of Neurology, **Harvard Medical School,** Neurocritical Care and Vascular Neurologist, **Massachusetts General Hospital / Harvard Medical School, Harvard Medical School, a NORD Rare Disease Center of Excellence**

Jerry Vockley, MD, PhD, Chief of Genetic and Genomic Medicine, Director of Center for Rare Disease Therapy, **University of Pittsburgh Medical Center, a NORD Rare Disease Center of Excellence**

Eva Morava-Kozicz, MD, PhD, Professor of Genetics and Genomic Sciences, **Icahn School of Medicine at Mount Sinai, a NORD Rare Disease Center of Excellence**

Integrating Patient-Centric and Real-World Data to Advance Rare Disease Understanding

Section Chair: Mark Skinner, JD, President and CEO, Institute for Policy Advancement Ltd.

2:30 p.m.

Approaches to Collecting and Managing Patient-Sourced Rare Disease Data

This session will cover methodologies and tools for collecting structured and unstructured data directly from patients and caregivers. Speakers will highlight community-engaged research models, data standardization, and best practices in longitudinal data collection.

SPEAKERS:

John Concato, MD, MPH, Adjunct Professor, **Yale School of Medicine, a NORD Rare Disease Center of Excellence**

Sydney Martinez, PhD, MPH, Associate Professor in Epidemiology, **University of Oklahoma Health Sciences Center**

Sheri Schully, PhD, Deputy Chief Medical and Scientific Officer, **NIH | All of Us Research Program**

3:25 p.m.

Networking Break

3:50 p.m.

Translating Patient Data into Research Insights

Patient-reported and real-world data can unlock new research directions when analyzed meaningfully. This session focuses on how such data can drive hypothesis generation, natural history modeling, and outcome measure development, with examples from academic, clinical, and industry perspectives.

SPEAKERS:

Elizabeth Regan, MD, PhD, Research Professor of Medicine, **National Jewish Health**

Mark Skinner, JD, President and CEO, **Institute for Policy Advancement Ltd**

Angela Waanders, MD, MPH, MS, Section Head, Neuro-Oncology, **Ann and Robert H. Lurie Children's Hospital of Chicago, a NORD Rare Disease Center of Excellence**

4:50 p.m.	**What's Next in Rare Disease Data Collection and Clinical Use** Artificial intelligence (AI) and novel approaches are increasingly reshaping rare disease research. This session will explore current applications of machine learning, natural language processing and data utilization in clinical and research settings. **SPEAKERS:** Steven Bedrick, PhD, Associate Professor of Medical Informatics and Clinical Epidemiology, **Oregon Health & Science University** Janet Woodcock, MD, Former Director, **Center for Drug Evaluation and Research (CDER), FDA** Manish Butte, MD, PhD, Professor and Division Chief, Immunology, Allergy, and Rheumatology, **University of California, Los Angeles, a NORD Rare Disease Center of Excellence**
5:50 p.m.	**Closing Remarks and Rare Impact Award Honoree Announcements** *Special recognition for NORD Medical/Scientific Rare Impact Award® Honoree
6:15 p.m.	**Networking Reception**

TUESDAY, JUNE 3, 2025

7:45 a.m.	**Continental Breakfast**
9:00 a.m.	**FDA Keynote Spotlight** **SPEAKER:** Vinay Prasad, MD, MPH, Director, **Center for Biologics Evaluation and Research (CBER), FDA**

Advancing Rare Disease Research Through Collaboration

Section Chair: Susan Berry, MD, Professor of Genetics and Metabolism, Department of Pediatrics, **University of Minnesota, a NORD® Rare Disease Center of Excellence**

9:45 a.m.	**Case Studies in Collaborative Data Use for Rare Disease Research** This session will present examples of how sharing and integrating data from multiple sources can lead to improved disease characterization, biomarker identification, and trial readiness. **SPEAKERS:** Collin Hovinga, PharmD, MS, FCCP, Vice President Rare Orphan and Pediatric Diseases, **Critical Path Institute** Nara Sobreira, MD, Associate Professor of Genetic Medicine, **Johns Hopkins University School of Medicine, a NORD Rare Disease Center of Excellence** Melissa Haendel, PhD, FACMI, Director, **Precision Health & Translational Informatics**; Sarah Graham Kenan Distinguished Professor, **University of North Carolina at Chapel Hill, a NORD Rare Disease Center of Excellence**
10:25 a.m.	**Multi-Stakeholder Collaboration in Research** Rare disease research moves ahead when multiple groups, including industry, regulators, patient advocacy organizations, and academia, come together to design and deliver impactful science. Speakers will provide case studies in partnership models, shared governance, and incorporating the patient voice into research design. **SPEAKERS:** Adora Ndu, PharmD, JD, Chief Regulatory Officer & EVP Portfolio Strategy and Management, **BridgeBio** Christopher Sibley, MD, Global Clinical Development Lead - Palopegteriparatide, **Ascendis Pharma** Fallon Schultz, MSW, LCSW, CAM, Co-Founder and CEO, **International FPIES Association (IFPIES)** Christal Delagrammatikas, PhD, Director of Research, **Malan Syndrome Foundation**
11:05 a.m.	**Networking Break**

TUESDAY, JUNE 3, 2025 (continued)

11:25 a.m.

ARPA-H Horizons: Addressing Diseases with Individualized Precision Genetic Medicines

SPEAKER:

Mimi Lee, MD, PhD, Program Manager, Health Science Futures, **Advanced Research Projects Agency for Health (ARPA-H)**

11:35 a.m.

Structuring and Sustaining Collaborative Rare Disease Research Networks

This session will examine the power of collaborative networks. Presenters will highlight infrastructure, funding strategies, and the role of shared data environments in sustaining long-term impact.

SPEAKERS:

Maurizio Scarpa, MD, PhD, Director, **European Reference Network for Hereditary Metabolic Diseases (MetabERN)**

Pramod Mistry, MD, PhD, FAASLD, Professor, Director, National Gaucher Disease Center, **Yale University School of Medicine, a NORD Rare Disease Center of Excellence**

Annette Bakker, PhD, Chief Executive Officer, **Children's Tumor Foundation**

12:10 p.m.

Successful Strategies for Earlier Pediatric Inclusion in Clinical Trials

Pediatric inclusion in rare disease trials remains a persistent challenge. Speakers will offer regulatory, ethical, and operational considerations for designing trials that safely and effectively include younger populations from the outset.

SPEAKERS:

Jennifer Cohen, MD, Assistant Professor of Pediatrics, Division of Medical Genetics, **Duke University School of Medicine, a NORD Rare Disease Center of Excellence**

Kristina AnHaack, Senior Global Project Head Lysosomal Storage Disorders and Neuromuscular Diseases, **Sanofi**

Stephen Rosenfeld, MD, MBA, Executive Director, **North Star Review Board**

12:55 p.m.

Closing Remarks

NORD® National Organization for Rare Disorders