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 Retrovirology, **Baylor College of Medicine and Texas Children's Hospital, a NORD® Rare Disease Center of Excellence**

Kiran Musunuru, MD, PhD, MPH, ML, Director of the Genetic and Epigenetic Origins of Disease Program, **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**

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 the U.S. Food and Drug Administration (FDA), academia, and industry will share strategies and experiences to inform trial design across rare diseases.

SPEAKERS:

Presenters TBA

11 a.m. — Networking Break

MONDAY, JUNE 2, 2025 (continued)

11:25 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, Coordinating Director, **European Reference Network for Hereditary Metabolic Diseases (MetabERN)**

Gopa Raychaudhuri, MD, PhD, Associate Director of Special Programs, **CBER, FDA**

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, Pediatric Neurology, **Massachusetts General Hospital, 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-Centered 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:

Sydney Martinez, PhD, MPH, Associate Professor in Epidemiology, **University of Oklahoma Health Sciences Center, a NORD® Rare Disease Center of Excellence**

Elizabeth Regan, MD, PhD, Associate Professor, **National Jewish Health**

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

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

3:30 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:

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

Janet Woodcock, MD, Former Director, Center for Drug Evaluation and Research, **FDA**

Minjee Park, Associate Director, Real World Evidence, **Alira Health**

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

4:50 p.m.	**Integrating AI into Rare Disease Data Collection and Clinical Use** Artificial intelligence (AI) is increasingly reshaping rare disease research. This session will explore current applications of machine learning and natural language processing in clinical and research settings. SPEAKERS: Steven Bedrick, PhD, Associate Professor of Medical Informatics and Clinical Epidemiology, **Oregon Health & Science University** Additional speakers TBA
5:50 p.m.	**Closing Remarks**
6:15 p.m.	**Networking Reception** *Special recognition for NORD Medical/Scientific Rare Impact Award® Honoree

TUESDAY, JUNE 3, 2025

8:15 a.m.	**Continental Breakfast**
9:00 a.m.	**Day 1 Recap and Opening Remarks**

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**

9:10 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 of the Rare and Orphan Disease Programs, **Critical Path Institute, Rare Disease Cures Accelerator – Data Analytics Platform (RDCA-DAP®)** Nara Lygia De Macena Sobreira, MD, Associate Professor of Genetic Medicine, **Johns Hopkins School of Medicine, a NORD® Rare Disease Center of Excellence** Melissa Haendel, PhD, FACMI, Department of Genetics, **University of North Carolina at Chapel Hill; Monarch Initiative, a NORD® Rare Disease Center of Excellence**
10:10 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 Strategies and Management, **BridgeBio** Christopher Sibley, MD, Senior Medical Director, Clinical Development, **Ascendis Pharma** Fallon Schultz, MSW, LCSW, Co-Founder and CEO, **International FPIES Association (IFPIES)** Christal Delagrammatikas, PhD, Director of Research, **Malan Syndrome Research, Overgrowth Syndrome Alliance**
10:45 a.m.	**Networking Break**

TUESDAY, JUNE 3, 2025 (continued)

11:05 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, Coordinating Director, **European Reference Network for Hereditary Metabolic Diseases (MetabERN)** Pramod Mistry, MBBS, PhD, MA, MD, Professor of Medicine (Digestive Diseases) and of Pediatrics (Gastroenterology), **Yale School of Medicine** Annette Bakker, PhD, Chief Executive Officer, **Children's Tumor Foundation**
11:50 a.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**
12:45 p.m.	**Closing Remarks**

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