NORD® National Organization for Rare Disorders

Rare Disease Scientific Symposium

APRIL 14-15 2026

Advancing Innovation in Rare Disease Research

Agenda subject to change.

* All times are ET

TUESDAY, APRIL 14, 2026

7:45AM	**CONFERENCE REGISTRATION AND CONTINENTAL BREAKFAST**
8:30AM	**NORD'S WELCOME & OPENING REMARKS** Pamela Gavin, CEO, **NORD**
8:45AM	**DAY 1 KEYNOTE ADDRESS** Tracy Beth Høeg, MD, PhD, Acting Director, **Center for Drug Evaluation and Research (CDER), FDA**

SECTION I: Clinical Research and Trial Design in Rare Disease

9:10AM	**BAYESIAN AND ADAPTIVE TRIAL DESIGNS FOR ULTRA-RARE POPULATIONS** Bayesian and adaptive approaches offer alternative ways to design and analyze studies when patient populations are extremely small. This session will frame how these methods differ from traditional trial designs and why they are increasingly considered in ultra-rare disease research, including their potential to support learning over time, reduce patient burden, and make more efficient use of limited data. **Moderator:** Edward Neilan, MD, PhD, Chief Medical and Scientific Officer, **NORD** **Speakers:** Yuan Ji, PhD, Professor of Biostatistics, **University of Chicago** Kelley Kidwell, Ph.D, Professor of Biostatistics, Interim Associate Dean of Faculty Affairs, **University of Michigan School of Public Health, a NORD Rare Disease Center of Excellence** Olivia Morgan, PhD, Statistician, **FDA**
10:00AM	**ALTERNATIVES TO PLACEBO CONTROLS AND RCTS IN RARE DISEASE TRIALS** Placebo-controlled trials are often impractical or ethically challenging in rare disease settings. This session will explore approaches that use alternative comparators, such as within-patient comparisons or external data sources, and discuss how these strategies can be used to generate interpretable evidence while addressing feasibility, ethical considerations, and regulatory expectations. **Moderator:** Amy Comstock Rick, JD, Director of Strategic Coalitions, **FDA Rare Disease Innovation Hub** **Speakers:** Oxana V. Crysler, MD, MHS, Clinical Associate Professor of Internal Medicine, **University of Michigan, Rogel Cancer Center, a NORD Rare Disease Center of Excellence** Wonyul Lee, PhD, Senior Statistical Reviewer, **CDER, FDA** David Margolin, MD, PhD, Vice President, Clinical Development, **uniQure, Inc.** Susana Zaph, PhD, Senior Director, Head of Quantitative Systems Pharmacology -US, R&D, **Sanofi**
10:50AM	**NETWORKING BREAK**

TUESDAY, APRIL 14, 2026 *(continued)*

11:10AM

FUNDING RARE DISEASE RESEARCH

Sustaining rare disease research requires funding models that account for small populations, long development timelines, and limited commercial incentives. This session will examine broad approaches to funding rare disease research and the roles that patient advocacy organizations, academic institutions, and industry partners can play across different stages of development.

Moderator: Tracey Sikora, Vice President of Research and Clinical Programs, **NORD**

Speakers:

Michael Hund, MBA, CEO, **EB Research Partnership**

Matthew S. McCoy, PhD, Assistant Professor of Medical Ethics and Health Policy, **University of Pennsylvania, a NORD Rare Disease Center of Excellence**

Nicole Paulk, PhD, CEO, Founder, President, **Siren Biotechnology**

12:00PM

NETWORKING LUNCH

1:30PM

INNOVATIVE CLINICAL TRIAL DESIGNS

Nontraditional trial designs are being used across therapeutic areas to address heterogeneity, small sample sizes, and evolving scientific understanding. This session will provide an overview of nontraditional clinical trial structures, such as umbrella, basket, and pragmatic trial designs, highlighting their applicability to rare disease research and their potential to improve efficiency, generalizability, and evidentiary yield.

Moderator: Annette Bakker, CEO, **Children's Tumor Foundation**

Speakers:

Jaishri Blakeley, MD, Professor of Neurology, Neurosurgery and Oncology, **The Johns Hopkins University, a NORD Rare Disease Center of Excellence**

Gwen Nichols, MD, EVP, Chief Medical Officer, **Blood Cancer United**

Amy Nicole Nayar, PharmD, MS, Vice President, Medical Affairs, **Lundbeck**

SECTION II: Drug Development: From Clinical Trials to Clinical Care

2:20PM

FDA REGULATORY INNOVATION: PLAUSIBLE MECHANISM

For many rare diseases, especially those with strong biological rationale but limited clinical data, regulatory approaches may rely on alternative forms of evidence. This session will provide context on how concepts such as plausible mechanism and platform-based development are considered within the regulatory framework for therapies targeting small populations.

Moderator: Julia Vitarello, Founder & CEO, **Mila's Miracle Foundation**

Speakers:

Teresa Buracchio, MD, Director, Office of Neuroscience, **CDER, FDA**

Michelle Mellion, MD, Chief Medical Officer, Clinical Development and Strategy Advisor; Former Chief Medical Officer, **EveryONE Medicines**

Lowell Schiller, JD, Nonresident Senior Scholar, **University of Southern California Schaeffer Center, a NORD Rare Disease Center of Excellence;** Former Principal Associate Commissioner for Policy, **FDA**

Judy Stecker, Founder, **Wheeler's Warriors;** Patient Advocate; Former Deputy Chief of Staff, **HHS**

Timothy Yu, MD, PhD, Associate Professor of Pediatrics, **Harvard Medical School, Boston Children's Hospital, a NORD Rare Disease Center of Excellence**

3:10PM

NETWORKING BREAK

3:30PM

ABANDONED, SHELVED, AND RESCUED THERAPIES FOR ULTRA-RARE DISEASES

Drug development programs for ultra-rare diseases may be discontinued for reasons unrelated to scientific validity, including resource constraints or shifting priorities. Presenters will describe case studies of therapies that were abandoned for commercial or logistical reasons and later revived through academic leadership, public-benefit models, or through repositioning.

Moderator: Edward Neilan, MD, PhD, Chief Medical and Scientific Officer, **NORD**

Speakers:

Paul Ayoub, PhD, MBA, Founder & CEO, **Rarity PBC**

Terry Pirovolakis, Founder & CEO, **Elpida Therapeutics**

Matthew Porteus, MD, PhD, Professor of Pediatrics, **Stanford University, a NORD Rare Disease Center of Excellence**

4:20PM

INNOVATIONS IN DRUG REPURPOSING FOR RARE DISEASES

Drug repurposing seeks to identify new disease applications for existing therapies. This session will provide an overview of how repurposing approaches are being explored in rare diseases and why they may offer a pragmatic path to treatment development when traditional discovery and development models are not feasible.

Moderator: Tracey Sikora, Vice President of Research and Clinical Programs, **NORD**

Speakers:

Heather Stone, MPH, Health Science Policy Analyst, **FDA**

C. Lee Cohen, MD, MBA, Medical Officer, **CDER, FDA**

Sarah Fuchs, MD, MSCI, Medical Officer, **CDER, FDA**

Kasha Morris, M.Ed, Co-Founder, **TANGO2 Research Foundation**

Savannah Latimer, Clinical Research Assistant, Division of Emergency Medicine, **Children's National Hospital, a NORD Rare Disease Center of Excellence**

Maurice Leary, **Children's National Hospital, a NORD Rare Disease Center of Excellence**

Natasha Shur, MD, Medical Geneticist, Professor of Pediatrics, **Children's National Hospital, a NORD Rare Disease Center of Excellence**

5:10PM

PRESENTATION OF THE 2026 MEDICAL & SCIENTIFIC TRAILBLAZER RARE IMPACT AWARDS®

Susan A. Berry, MD
University of Minnesota, NORD Rare Disease Center of Excellence

Stephen Kingsmore, MD, DSc
Rady Children's Institute for Genomic Medicine, NORD Rare Disease Center of Excellence

5:45PM

NETWORKING RECEPTION

WEDNESDAY, APRIL 15, 2026

8:00AM **CONTINENTAL BREAKFAST**

9:00AM **DAY 1 RECAP AND OPENING REMARKS**

9:05AM **NIH KEYNOTE ADDRESS**

Annica Wayman, PhD, Deputy Director, **National Center for Advancing Translational Sciences, NIH**

SECTION III: Leveraging Data for Innovation

9:50AM **DATA FOR ENDPOINT SELECTION**

Selecting appropriate endpoints is a central challenge in rare disease research, particularly when clinical outcomes are heterogeneous or poorly characterized. This session will explore how different data sources can inform endpoint selection and support endpoints that are clinically meaningful, feasible to measure, and interpretable for regulators and other decision-makers.

Moderator: Craig Lipset, Co-Chair, **Digital Trials & Research Alliance;** Clinical Innovation, **Buffalo Initiative**

Speakers:

Gabrielle Conecker, MPH, Executive Director & Co-Founder, **Decoding Developmental Epilepsies**

Gerald F. Cox, MD, PhD, FACMG, Consultant, **Gerald Cox Rare Consulting, LLC;** Staff Physician in Genetics, **Boston Children's Hospital, a NORD Rare Disease Center of Excellence**

Amena Fine, MD, PhD, Assistant Professor of Neurology, **Kennedy Krieger Institute, a NORD Rare Disease Center of Excellence**

Amy Raymond, PhD, PMP, Therapeutic Strategy Lead, Rare Diseases and Cellular & Genetic Medicines, **Worldwide Clinical Trials**

10:50AM **NETWORKING BREAK**

11:10AM **REGISTRIES AND REAL-WORLD DATA AS DEVELOPMENT PLATFORMS**

Registries and real-world data sources are increasingly used to support multiple stages of rare disease research. This session will frame how these data assets can function as development platforms, informing natural history studies, clinical trial design, and evidence generation beyond traditional clinical trials.

Moderator: 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**

Speakers:

Amy Palmer Laster, PhD, Chief Scientific Officer, **Foundation Fighting Blindness**

Mayowa Azeez Osundiji, MD, PhD, Medical Geneticist, **Mayo Clinic**

Srilakshmi (Sri) Raj, PhD, Assistant Professor of Genetics, **Albert Einstein College of Medicine, a NORD Rare Disease Center of Excellence**

Theresa Strong, PhD, Director of Research Programs, **Foundation for Prader-Willi Research**

12:10PM **ACCESS AND COVERAGE: DATA AND CLINICAL DEVELOPMENT**

Decisions about coverage and access are influenced by the type and strength of evidence generated during and after clinical development. This session will examine how clinical and real-world data intersect with payer and coverage considerations, and why early alignment between development strategies and evidence needs is particularly important in rare diseases.

Moderator: Pamela Gavin, CEO, **NORD**

Speakers:

Ravi Pathak, PhD, MBA, Medical Director, Lysosomal Storage Disorders, US Medical, **Takeda**

Mark Trusheim, MSm, Senior Strategy Advisor, NEWDIGS, **Tufts Medical Center**

Margarita L. Valdez Martinez, Chief Advocacy Officer, **ASGCT**

Danny Yeh, PhD, Vice President, Rare Disease, **AESARA**

1:00PM **CLOSING REMARKS**

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