RARE DISEASE DAY at NIH

Feb. 27, 2026 | #RDDNIH

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Preliminary Event Agenda

As of 12/22/2025

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

9:00 a.m. **Rare Disease Day at NIH Overview**
Meera Shah, M.P.H., Health Specialist, Division of Rare Diseases Research Innovation (DRDRI), National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH)

9:10 a.m. **Welcoming Remarks**
Joni L. Rutter, Ph.D., Director, NCATS, NIH

9:30 a.m. **NCATS DRDRI Update**
Philip John (P.J.) Brooks, Ph.D., Acting Director, DRDRI, NCATS, NIH

9:45 a.m. **Beyond the Diagnosis Portrait Unveiling**
Patricia Weltin, CEO/Founder, Beyond the Diagnosis
Lucas Kolasa, Artist, Beyond the Diagnosis
Aisha Campbell, M.B.A./H.C.M., Director, Resident Services and Family Programs, The Children's Inn at NIH

10:00 a.m. **Session 1: 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. Presenters will discuss two case studies of currently ongoing clinical trials. This session also will discuss challenges for innovative trial designs.
Moderator: Elena Schwartz, Ph.D., Program Officer, Coordinating Center for Clinical Trials, National Cancer Institute (NCI), NIH
Panelists:

- Malcolm A. Smith, M.D., Ph.D., Associate Branch Chief for Pediatric Oncology, Clinical Investigations Branch, NCI, NIH
- Troy Torgerson, M.D., Ph.D., Director of Experimental Immunology, Allen Institute for Immunology

10:30 a.m. **Break**

- Exhibits and Scientific Posters
- Art Exhibition
- Networking

10:50 a.m. **Rare Story: Corrective Genome Editing to Treat Hereditary Connective Tissue Disorder — A Patient–Scientist Perspective**
Lauren Testa, Ph.D. Candidate, Cell and Molecular Biology (Gene Therapy and Vaccines), University of Pennsylvania Perelman School of Medicine

11:10 a.m. **Rare Story: The Full-Time Job of Living Rare — Advocacy, Resilience and the Fight for Care**
Lindsay Guentzel, Multimedia Journalist, Storyteller, Advocate

11:30 a.m. **Session 2: Artificial Intelligence — Clinical 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 research and clinical applications relevant to the rare diseases community. Presenters will then delve into two case studies that demonstrate AI use in rare diseases research.

Moderator: Benjamin Soloman, M.D., Clinical Director, National Human Genome Research Institute

Panelists:

- Nara Sobreira, M.D., Ph.D., Associate Professor in the McKusick-Nathans Department of Genetic Medicine and Associate Professor of Pediatrics, Johns Hopkins University
- Vivek Rudrapatna, M.D., Ph.D., Assistant Professor in the Division of Gastroenterology and Director of the Center for Real-World Evidence, University of California, San Francisco
- Tala Fakhouri, Ph.D., M.P.H., Vice President Consulting: AI & Digital Policy, Real-World Research, Parexel

12:30 p.m. **Lunch (on your own)**

- Exhibits and Scientific Posters
- Art Exhibition
- Networking

2:20 p.m. **Rare Story: Conquering the Unconquerable — Turning Hope Into Progress Through Research**

Jake Juip, Rare Disease Advocate and College Student

2:40 p.m. **Rare Research: How Collaboration Becomes Cures — A Juvenile Myositis and CAR T Story**

3:00 p.m. **Rare Story**

Missy Ramirez

3:20 p.m. **Break**

- Exhibits and Scientific Posters
- Art Exhibition
- Networking

3:40 p.m. **Session 3: Gene Therapy — Recent Approvals, Success Stories and Patient Perspectives**

Gene therapy continues to transform the treatment landscape for rare diseases, offering new hope to patients and families. This session will highlight two recent cases of gene therapy advancement and NCATS' involvement: the approval of a therapy for aromatic L-amino acid decarboxylase (AADC) deficiency and the development of a personalized CRISPR gene-editing therapy for severe carbamoyl phosphate synthetase 1 (CPS1) deficiency. The session also will include a broader discussion of patient considerations and decision-making factors when pursuing gene therapy.

Moderator: Philip John (P.J.) Brooks, Ph.D., Acting Director, DRDRI, NCATS, NIH

Panelists:

- Elizabeth A. Ottinger, Ph.D., Director, Therapeutic Development Branch, Division of Preclinical Innovation, NCATS, NIH
- Jodi A. Cook, Ph.D., President and CEO, Skylark Bio
- Rebecca Ahrens-Nicklas, M.D., Ph.D., Assistant Professor of Pediatrics, Division of Human Genetics, Children's Hospital of Philadelphia and the University of Pennsylvania
- Kiran Musunuru, M.D., Ph.D., M.P.H., M.L., M.R.A., Director, Genetic and Epigenetic Origins of Disease Program, Cardiovascular Institute, Scientific Director, Center for Inherited Cardiovascular Disease, University of Pennsylvania Perelman School of Medicine

4:40 p.m. **Closing Remarks**
Joni L. Rutter, Ph.D., Director, NCATS, NIH

5:00 p.m. **Adjournment**
Posters and Exhibits Close