



# Gene-Targeted Therapies: Early Diagnosis and Equitable Delivery

June 3 from 12-4:30 p.m. EDT (The Who, What, and When)  
June 10 from 12-4:30 p.m. EDT (Infrastructure and Mechanics)  
June 17 from 12-4:30 p.m. EDT (Regulatory and Equitable Access)

## JUNE 3 — DAY 1: The Who, What, and When

- 12:00 p.m. ET **Welcome Remarks**  
*Joni L. Rutter, Ph.D.*, Acting Director, National Center for Advancing Translational Science (NCATS), National Institutes of Health (NIH)
- 12:10 p.m. **Introduction**  
*Robert Green, M.D., MPH*, Professor of Medicine, Harvard
- 12:20 p.m. **Gene-Targeted Therapies for All Who Need Them: Tremendous Opportunities, Tremendous Challenges**  
*Christopher P. Austin, M.D.*, CEO-Partner, Flagship Pioneering
- 12:40 p.m. **Milasen: The First Truly Individualized Medicine and Its Impact on the Future of Drug Development**  
*Julia Vitarello*, Mila's mom & Founder and CEO, Mila's Miracle Foundation
- 1:00 p.m. **Current State of Treatments**  
This session will give an overview of three gene-targeted therapeutic platforms: gene therapy, oligonucleotide therapy, and somatic genome editing  
  
Moderator:  
  - *Philip John (P.J.) Brooks, Ph.D.*, Program Director, Office of Rare Diseases Research (ORDR), NCATS, NIHPanelists:  
  - *Timothy Yu, M.D., Ph.D.*, Neurogeneticist, Boston Children's Hospital and Harvard Medical School
  - *James Wilson M.D., Ph.D.*, Director, Gene Therapy Program, University of Pennsylvania's Perelman School of Medicine
  - *David R. Liu, Ph.D.*, Director, Merkin Institute of Transformative Technologies in Healthcare
- 1:45 p.m. **Break**
- 2:00 p.m. **Working Group 1 Presentations**  
Introduction  
*Priya Kishnani, M.D., MBBS*, Chief, Medical Genetics Division, Duke University Medical Center (DUMC)  
*Melissa Wasserstein, M.D.*, Professor of Pediatrics and Genetics, Albert Einstein College of Medicine

**Who** are the individuals that could benefit from gene-targeted therapies – now and in the future?

- *Melissa Parisi, M.D., Ph.D.*, Chief, Intellectual and Developmental Disabilities Branch (IDDB), Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), NIH

**What** novel approaches are needed to enable development of gene-targeted therapies for all genetic rare diseases – now and in the future?

#### Technology

- *Huda Zoghbi, M.D.*, Professor, Baylor College of Medicine, Director, Jan and Dan Duncan Neurological Research Institute, Investigator, Howard Hughes Medical Institute
- *Steven Gray, Ph.D.*, Associate Professor, Department of Pediatrics, University of Texas Southwestern Medical Center

#### Methods

- *Julie Lekstrom Himes, M.D.*, Vice President and Head of Clinical Sciences, Hematology, Takeda Pharmaceutical
- *Erika Fullwood Augustine, M.D., M.S.*, Chief Science Officer, Director of the Clinical Trials Unit, Kennedy Krieger Institute

#### Best Practices

- *Jennifer Cohen, M.D.*, Assistant Professor of Pediatrics, Division of Medical Genetics, Duke University

**When** is the optimal time to identify individuals who could benefit from gene-targeted therapies?

- *Michele Caggana, ScD, FACMG*, Director, Newborn Screening, Wadsworth Center
- *Joseph Gleeson, M.D.*, Rady Professor, University of California San Diego Neurosciences, Rady Children’s Institute for Genomic Medicine
- *Tippi MacKenzie, M.D.*, Professor of Surgery, University of California, San Francisco
- *Amy Brower, Ph.D.*, Medical Geneticist, American College of Medical Genetics and Genomics (ACMG)
- *Nancy Mendelsohn, M.D.*, Chief Medical Officer, Complex Health Solutions, UnitedHealth Group
- *Priya Kishnani, M.D., MBBS*, Chief, Medical Genetics Division, Duke University Medical Center (DUMC)

#### Education, Engagement and Partnerships

- *Lex Cowsert, Ph.D.*, Director, Research Development, National PKU Alliance (NPKUA)
- *Amy Brower, Ph.D.*, Medical Geneticist, American College of Medical Genetics and Genomics (ACMG)
- *Louis P. Garrison, Ph.D.*, Professor Emeritus; Comparative Health Outcomes, Policy, and Economics (CHOICE) Institute; Department of Pharmacy; University of Washington

3:40 p.m.

**Questions and Open Discussion**

Moderator: *Philip John (P.J.) Brooks, Ph.D.*, Program Director, Office of Rare Diseases Research (ORDR), NCATS, NIH

*To submit questions or comments:*

1. *Email ORDR@nih.gov*
2. *Send Live Feedback via Videocast*
3. *Tweet: #NIHGTTmtg*

4:20 p.m.

**Closing Remarks**

- *Huda Zoghbi, M.D.*, Professor, Baylor College of Medicine, Director, Jan and Dan Duncan Neurological Research Institute, Investigator, Howard Hughes Medical Institute
- *Steven Gray, Ph.D.*, Associate Professor, Department of Pediatrics, University of Texas Southwestern Medical Center

4:30 p.m.

**Adjourn**