Gene-Targeted Therapies: 
Early Diagnosis and Equitable Delivery

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, NIH

Panelists:
• 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, Takada 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: #NIHGTattg

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**