



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 10—DAY 2: Infrastructure and Mechanics

- 12:00 p.m. ET **Welcome Remarks**
Walter Koroshetz, M.D., Director, National Institute of Neurological Disorders and Stroke (NINDS), National Institutes of Health (NIH)
- 12:10 p.m. **Day 2 Overview**
Tippi MacKenzie, M.D., Professor of Surgery, University of California, San Francisco
- 12:20 p.m. **Current State of Patient Identification**
This session will provide an overview of several current pathways through which individuals with rare genetic disorders are being identified
Moderator:
 - *Tiina K. Urv, Ph.D.*, Program Director, Office of Rare Diseases Research (ORDR), National Center for Advancing Translational Science (NCATS), NIHPanelists:
 - *Teresa Sparks, M.D.*, Perinatologist, *University of California, San Francisco*
 - *Stephen Kingsmore, MBChB, BAO, B.Sc., D.Sc.*, President and CEO, Rady Children's Institute for Genomic Medicine (RCIGM)
 - *Don Bailey, Ph.D.*, Director, Center for Newborn Screening, Ethics, and Disability Studies, RTI International
 - *Robert Green, M.D., MPH*, Professor of Medicine, Harvard
- 1:20 p.m. **Patient 007: Rapid Diagnosis and Gene Therapy of an Infant with rare Severe Combined Immune Deficiency**
 - *Stephen Kingsmore, MBChB, BAO, B.Sc., D.Sc.*, President and CEO, RCIGM
 - *Christina Kettler*, Mother of Fitz Kettler
 - *Daniel Kettler*, Father of Fitz Kettler
 - *Fitz Kettler*, Genomic Trailblazer, Gene Therapy Clinical Trial Participant
- 1:40 p.m. **Break**
- 2:00 p.m. **Working Group 2 Presentations**
Introduction
Thomas Defay, Ph.D., Deputy Head, Diagnostics Strategy and Operations, *Alexion Pharmaceuticals, Inc.*

- Scaling Newborn Screening
 - *Amy Gaviglio, MS, CGC*, Public Health Genetics Consultant, Centers for Disease Control and Prevention (CDC), Association of Public Health Laboratories (APHL), and Expecting Health
- Scaling Whole Genome Sequencing
 - *Stephen Kingsmore, MBChB, BAO, B.Sc., D.Sc.*, President and CEO, RCIGM
 - *Annemieke Aartsma-Rus, Ph.D.*, Professor of Translational Genetics, Department of Human Genetics, Leiden University Medical Center
- Scaling Artificial Intelligence
 - *Thomas Defay, Ph.D.*, Deputy Head, Diagnostics Strategy and Operations, *Alexion Pharmaceuticals, Inc.*
- Scaling Treatment Protocols
 - *James Wilson M.D., Ph.D.*, Director, Gene Therapy Program, University of Pennsylvania's Perelman School of Medicine
 - *Philip John (P.J.) Brooks, Ph.D.*, Program Director, ORDR, NCATS, NIH
 - *Gerard Vockley, M.D., Ph.D.*, Professor of Pediatrics; Cleveland Family Endowed Chair in Pediatric Research, University of Pittsburgh School of Medicine
- Financing/Incentives/Funding
 - *Andrew W. Lo, Ph.D.*, Charles E. and Susan T. Harris Professor, MIT Sloan School of Management

3:45 p.m.

Questions and Open Discussion

Moderator:

- *Tiina K. Urv, Ph.D.*, Program Director, 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

Tom Defay, Ph.D., Deputy Head, Diagnostics Strategy and Operations, Alexion Pharmaceuticals, Inc.

4:30 p.m.

Adjourn