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), NIH  
Panelists:  
• 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
  o 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
  o Stephen Kingsmore, MBChB, BAO, B.Sc., D.Sc., President and CEO, RCIGM
  o Annemieke Aartsma-Rus, Ph.D., Professor of Translational Genetics, Department of Human Genetics, Leiden University Medical Center
• Scaling Artificial Intelligence
  o Thomas Defay, Ph.D., Deputy Head, Diagnostics Strategy and Operations, Alexion Pharmaceuticals, Inc.
• Scaling Treatment Protocols
  o James Wilson M.D., Ph.D., Director, Gene Therapy Program, University of Pennsylvania’s Perelman School of Medicine
  o Philip John (P.J.) Brooks, Ph.D., Program Director, ORDR, NCATS, NIH
  o 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
  o 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