Newborn Screening in the Genomic Era: Setting a Research Agenda

5635 Fishers Lane, Terrace Level, Rockville, MD

December 13–14, 2010

Sponsored by:

Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)

National Human Genome Research Institute (NHGRI)

NIH Office of Rare Diseases Research (ORDR)

Monday, December 13th

8:00–8:30 a.m.  Registration and Refreshments

8:30–8:50 a.m.  Welcome from Institute Directors and Introductions
    Alan Guttmacher – NICHD
    Eric Green – NHGRI

8:50–9:10 a.m.  Introduction to Newborn Screening
    Piero Rinaldo – Mayo Clinic

9:10–9:30 a.m.  Predicting the Future: From Biochemistry to Sequence
    David Valle – Johns Hopkins University School of Medicine

9:30–9:50 a.m.  How Genetic Testing Prior to Birth Relates to Newborn Screening
    Arthur Beaudet – Baylor College of Medicine

9:50–10:20 a.m.  Discussion
    R. Rodney Howell – NICHD

10:20–10:30 a.m.  Break

10:30–11:20 a.m.  Thinking Outside the Box: Potential Technological Contenders for Use in Newborn Screening
    Chair – Jeff Schloss – NHGRI
    Targeted Sequencing and New Technologies
    Ronald Davis – Stanford Genome Technology Center
    Fitting Current Technologies for Newborn Screening
    Richard Gibbs – Baylor College of Medicine

11:20–11:50 a.m.  Discussion
    Jeff Schloss – NHGRI
11:50 a.m.–1:30 p.m.  Working Lunch – Technology Talks
  Chair – Jeff Schloss – NHGRI

  Digital Microfluidic Lab-on-a-chip for Multiplexing Tests in Newborn Screening
  Vamsee Pamula – Advanced Liquid Logic

  TR-FRET-based End-point PCR Utilizing Dried Blood Disc as a Sample Matrix
  Petri Huhtinen – PerkinElmer

  Targeted Genetic Screens Using Bead-based Multiplexed Molecular Assays
  Keld Sorensen – Luminex Corporation

  Nanopore-based DNA and Protein Analysis
  Andre Marziali – University of British Columbia

1:30–2:30 p.m.  Challenges of New Technologies in the Ethical, Legal, and Social Contexts
  Chair – Alan Fleischman – March of Dimes Foundation

  The Danish Newborn Screening Biobank: Establishment and Use
  Bent Nørgaard-Pedersen – Statens Serum Institut, Copenhagen

  Developing New Tests and Expansion: Legal, Social, and Ethical Issues
  Fred Lorey – California Department of Public Health

  Parental Engagement in Newborn Screening Technology
  Jeff Botkin – University of Utah School of Medicine

2:30–3:00 p.m.  Discussion
  Alan Fleischman – March of Dimes Foundation

3:00–3:05 p.m.  Charge to Breakout Groups
  David Valle – Johns Hopkins University School of Medicine and Piero
  Rinaldo – Mayo Clinic

3:05–3:15 p.m.  Break

3:15–5:15 p.m.  Breakout Groups

  Breakout Group I – Technology
  (Performance, specificity, sensitivity, cost, validation, bioinformatics, samples)
  Co-chairs – David Goldstein – Duke University School of Medicine
  and David Ledbetter – Geisinger Health System

  Breakout Group II – Education/Expectations
  (Healthcare providers, general public, researchers, return of results)
  Co-chairs – Bruce Korf – University of Alabama at Birmingham
  School of Medicine and Jeff Murray – University of Iowa
Breakout Group III – Scope and Timing
(What to screen for, when to screen for it)

Co-chairs – Geoff Duyk – TPG Biotech and Richard Weinshilboum – Mayo Clinic

5:15–5:30 p.m.  Wrap up Day 1 – David Valle – Johns Hopkins University School of Medicine and Piero Rinaldo – Mayo Clinic

5:30 p.m.  Adjourn

6:30 p.m.  Group Dinner

Rockville Hilton Hotel

Tuesday, December 14th

8:00–8:30 a.m.  Refreshments

8:30–10:00 a.m.  Presentations from Breakout Groups

Co-moderators – Susan Berry – University of Minnesota Medical School and Pearl O’Rourke – Partners HealthCare Systems

Breakout Group I
Breakout Group II
Breakout Group III

10:00–10:15 a.m.  Break

10:15–11:00 a.m.  Open Discussion: What Pieces of the Puzzle Are Missing?

Co-moderators – Susan Berry – University of Minnesota Medical School and Pearl O’Rourke – Partners HealthCare Systems

11:00–11:45 a.m.  Wrap-up Discussion: Priorities for Moving the Field Forward

Co-moderators – Michele Lloyd-Puryear – Health Resources and Services Administration and Wendy Chung – Columbia University Medical Center

11:45 a.m.–12:00 p.m.  Institutes’ Closing Comments

Alan Guttmacher – NICHD
Eric Green – NHGRI

12:00 p.m.  Adjourn