



## 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