Gene Sequencing in Public Health
Newborn Screening Meeting

Funding for this conference was made possible in part by the US Centers for Disease Control and Prevention. The views expressed in written conference materials and by speakers and moderators do not necessarily reflect the official policies of the Department of Health and Human Services, nor does the mention of trade names, commercial practices or organizations imply endorsement by the US Government.

This project was also supported by the Health Resources and Services Administration (HRSA) under grant # U22MC24078 Heritable Disorders $950,000. This information or content and conclusions are those of the authors and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the US Government.

February 16 - 17, 2017
The Westin Buckhead Atlanta
Day 1 Recap
The purpose of this meeting is to convene pertinent stakeholders to discuss the current status of gene sequencing in newborn screening, and identify barriers and solutions for the successful incorporation of gene sequencing into newborn screening.
Gene Sequencing Timeline in Routine Newborn Screening

- **2004**: Hemoglobinopathies, Texas begins HBB sequencing, 3rd tier test
- **2005**: Krabbe, NY begins GALC sequencing, 2nd tier
- **2006**: Advent of Next Gen Sequencing
- **2007**: X-ALD, NY begins ABCD1 sequencing, 3rd tier
- **2008-2012**: Cystic Fibrosis
- **2013**: NY begins CFTR sequencing, Next Gen Sequencing
- **2014**: CA begins ABCD1 sequencing, 3rd tier
- **2015**: NY begins GAA sequencing, 2nd tier
- **2016**: MPS 1 (IDUA), NY: 2nd tier
- **2017**: Krabbe, Pompe & MPS 1 (GALC, GAA & IDUA), NJ: 2nd tier
- **2018**: Cystic Fibrosis, WI begins CFTR Next Gen Sequencing, NJ: 2nd tier
- **2019**: X-ALD, CA begins ABCD1 sequencing, 3rd tier
- **2020**: APHL Creates Molecular Subcommittee for NBS
- **2021**: Pompe, MPS 1, X-ALD (GAA, IDUA, & ABCD1), New Eng: 2nd tier
- **2022**: Pompe & MPS 1 (GAA, IDUA), MN: 2nd tier
- **2023**: Pompe (GAA), WI: 2nd tier
- **2024**: VLCAD (ACADVL), TX: 2nd tier
- **2025**: Cystic Fibrosis Next Gen Seq, NY: 3rd tier

**Analysis. Answers. Action.**

www.aphl.org
Day 1 Recap

Session 1: Current Status of Gene Sequencing in Newborn Screening (NBS)
- Introduction and Background of Gene Sequencing in NBS
- Current Second Tier and Future Applications of Gene Sequencing in NBS
- Mutation Panel vs. Sequencing Data Pros and Cons
- Case Studies with Molecular Analysis

Session Two: What Does the NBS Program Need to Make this Happen?
- Instrumentation and Informatics Requirements
- Implementation Considerations for an Emerging NBS Technology
- Program Planning
- Laboratory and Follow-Up Barriers to Incorporating Complex Molecular Testing into the NBS Workflow