NewSTEPs

A Program of the Association of Public Health Laboratories™

National Newborn Screening Meeting on New Disorders

Pompe, MPS I and X-ALD

Meeting Summary and Notes

June 22-23, 2017

Bethesda, MD

This meeting was supported by the Health Resources and Services Administration (HRSA) under Cooperative Agreement # UG9MC30369 New Disorders Implementation Project. Meeting presentations are solely the responsibility 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.

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ACKNOWLEDGEMENTS

The Association of Public Health Laboratories would like to recognize the National Newborn Screening Meeting on New Disorders Planning Committee members who generously donated their time and input for the development of the meeting agenda.

Mei Baker, MD, FACMG, Wisconsin State Laboratory of Hygiene
Michele Caggana, ScD, FACMG, New York State Department of Health
Patrick Hopkins, Missouri Newborn Screening Laboratory
Rasoul Koupaei, PhD, California Department of Health
Amelia Mumford, Baby's First Test
Kimberly Piper, RN, BS, CPH, CPHG, Iowa Department of Public Health
Anthony Steyermark, PhD, Minnesota Department of Health
Staff Liaison: **Kshea Hale**, MPH, Association of Public Health Laboratories
Staff Evaluator: **Yvonne Kellar-Guenther**, PhD, Colorado School of Public Health

We also would like to thank our presenters and speakers who shared their experiences.

Mei Baker, MD, FACMG, Wisconsin State Laboratory of Hygiene
Natasha Bonhomme, Baby's First Test
Amy Brower, PhD, American College of Medical Genetics and Genomics
Michele Caggana, ScD, FACMG, New York Department of Health
Lisa Feuchtbaum, DrPH, MPH, California Department of Public Health
Amy Gaviglio, MS, CGC, Minnesota Department of Health
Celia Hagen, MPH, Association of Public Health Laboratories
Patrick Hopkins, Missouri Department of Health
Priya Kishnani, MD, Duke University Medical Center
Adrienne Manning, Connecticut Department of Public Health
Amelia Mumford, Baby's First Test
Claudia Nash, MS, Illinois Department of Health
Paul Orchard, MD, University of Minnesota
Kimberly Piper, RN, BS, CPH, CPHG, Iowa Department of Public Health
Rong Shao, MD, Illinois Department of Public Health
Marci Sontag, PhD, Colorado School of Public Health
Anthony Steyermark, PhD, Minnesota Department of Health
Chester Whitley, PhD, MD, University of Minnesota

BACKGROUND

Pompe was added to the Recommended Uniform Screening Panel (RUSP) in March 2015, and X-Linked Adrenoleukodystrophy (X-ALD) and Mucopolysaccharidosis type I (MPS I) were added in February 2016. State newborn screening programs pursuing universal implementation of these three new disorders encounter laboratory, staffing, clinical follow-up, personnel, equipment, education and legislative challenges, and various solutions have been developed by the programs.

MEETING PURPOSE

The purpose of the National Newborn Screening Meeting on New Disorders was to convene newborn screening personnel to discuss newborn screening for new disorders added to the RUSP, as well as pertinent partners and stakeholders who have experience with implementing new disorders. The implementation process includes all stages, beginning with the decision to screen through final implementation and universal screening.

MEETING OBJECTIVES

1. Discuss current status of newborn screening for Pompe, MPS I and X-ALD.
2. Discuss policy, laboratory, follow-up and education needs, barriers and solutions for newborn screening.
3. Provide state experiences in implementing newborn screening.
4. Identify the questions that need to be considered prior to considering implementation for new disorder newborn screening.

STATE OF NEW DISORDERS NEWBORN SCREENING

Figures 1, 2 and 3 depict the states who are offering universal newborn screening for Pompe, MPS I and X-ALD, respectively, in the United States as of July 2017.

Pompe is currently universally screened for in five states- Illinois, Kentucky Missouri, New York and Pennsylvania.

MPS I is currently universally screened for in four states- Illinois, Kentucky, Missouri and Pennsylvania. New York is offering MPS I newborn screening to select populations.

X-ALD is currently universally screened for in five states- California, Connecticut, Minnesota, New York and Pennsylvania

Figure 1: Pompe Newborn Screening Status in the United States as of July 2017

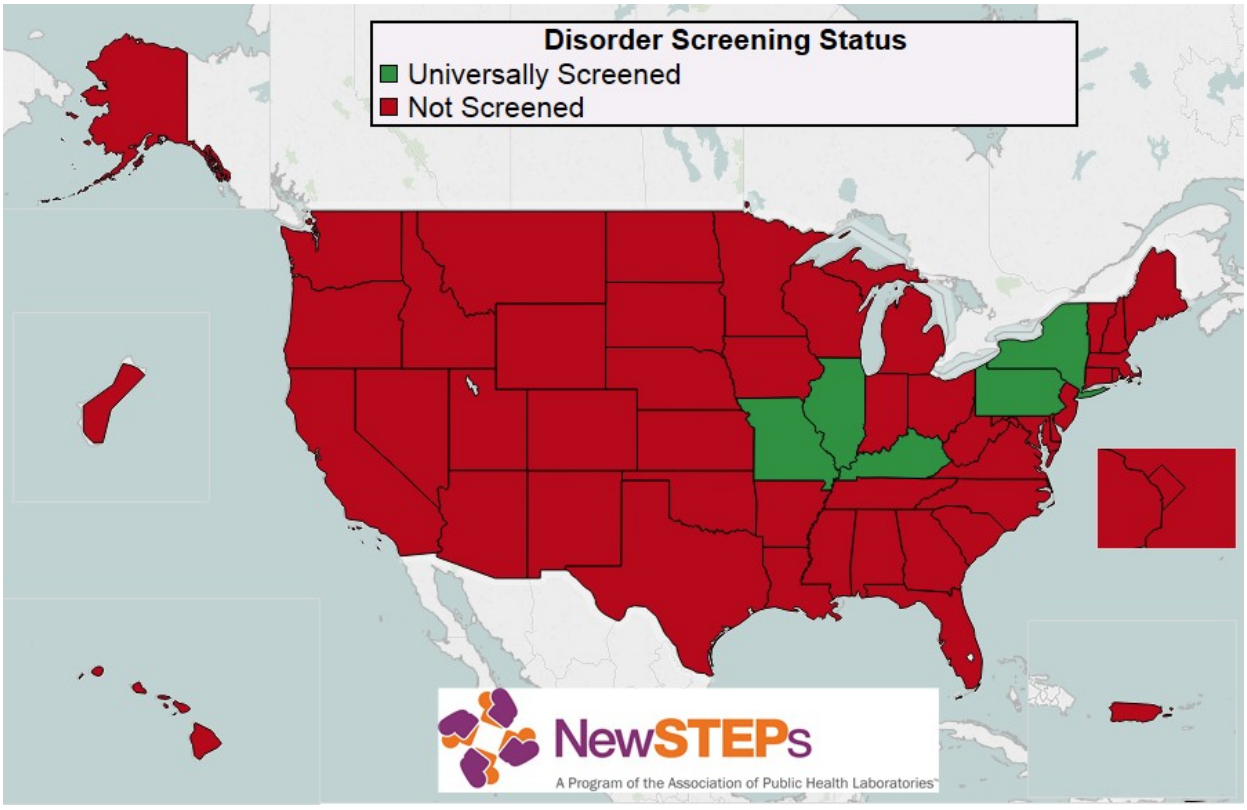


Figure 2: MPS I Newborn Screening Status in the United States as of July 2017

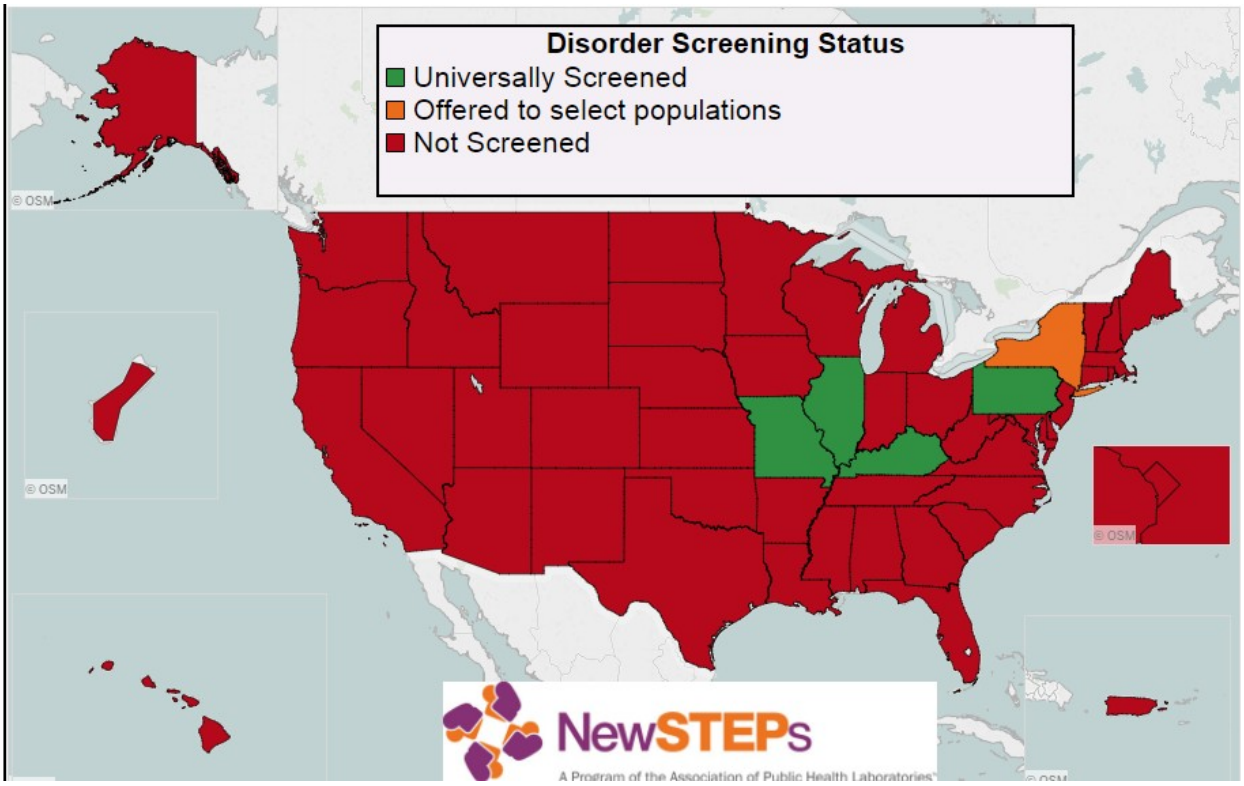
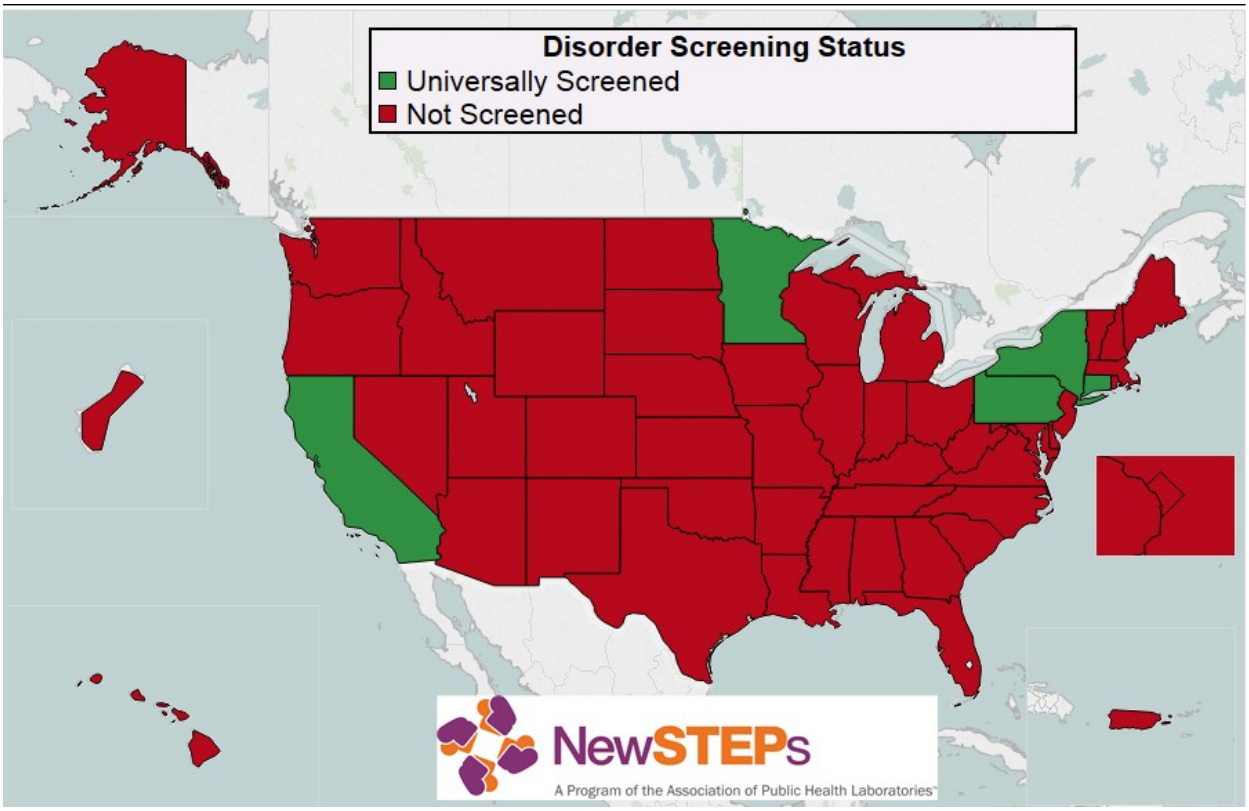


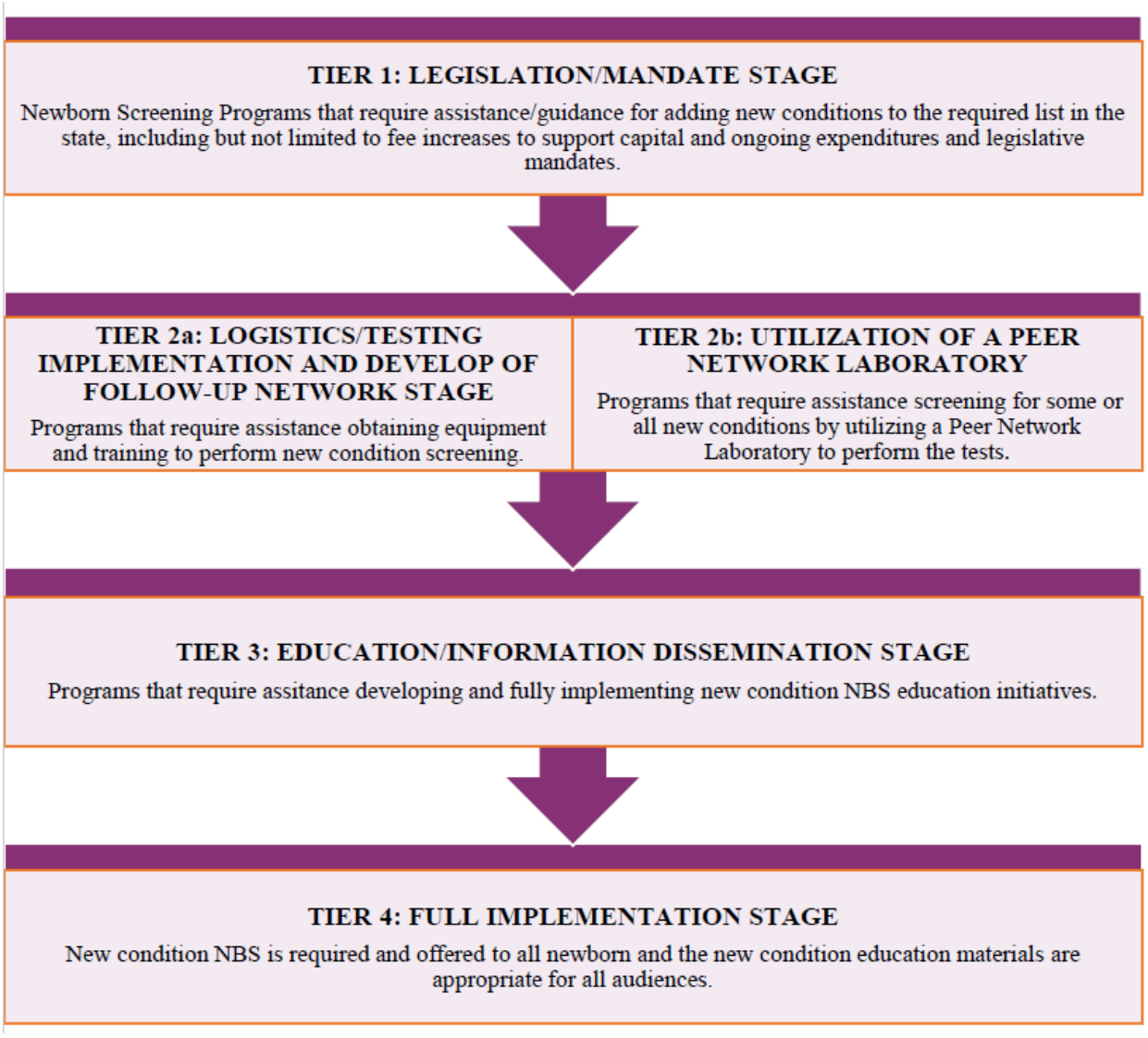
Figure 3: X-ALD Newborn Screening Status in the United States as of July 2017



READINESS TOOL RESULTS

In spring 2017, NewSTEPs issued a New Disorder Readiness Tool for completion to all states attending the New Disorders National Meeting. The purpose of this tool is to capture and track over time the resources, tools and activities required by newborn screening programs for implementation of a new disorder during all stages of implementation (Figure 4). The tool will help identify variations in readiness for population screening in each state and can be used to connect states to one another for experience sharing purposes. A detailed summary of the Readiness Tool results can be found linked here.

Figure 4: New Disorders Implementation Four-Tier Model



At the time of the meeting, 33 states had provided data for the Readiness Tool. About half the states had started or completed at least one activity in the Legislative/Mandate Phase. This dropped to slightly less than half who have engaged in at least one activity in lab preparation (See Figure 5) and about one third who engaged in at least one activity in follow-up (See Figure 6). Only a few states have demonstrated or initiated readiness for Information Technology activities, education activities, pilot testing, and statewide implementation. The Readiness Tool data did highlight the time involved in the steps can vary by as much as three years. The data also revealed that while many states begin at Phase 1 (Figure 4), the Legislative/Mandate Phase, some states have started by considering requirements for the follow-up program or education materials.

Figure 5: Laboratory Facility and Infrastructure Readiness within the United States

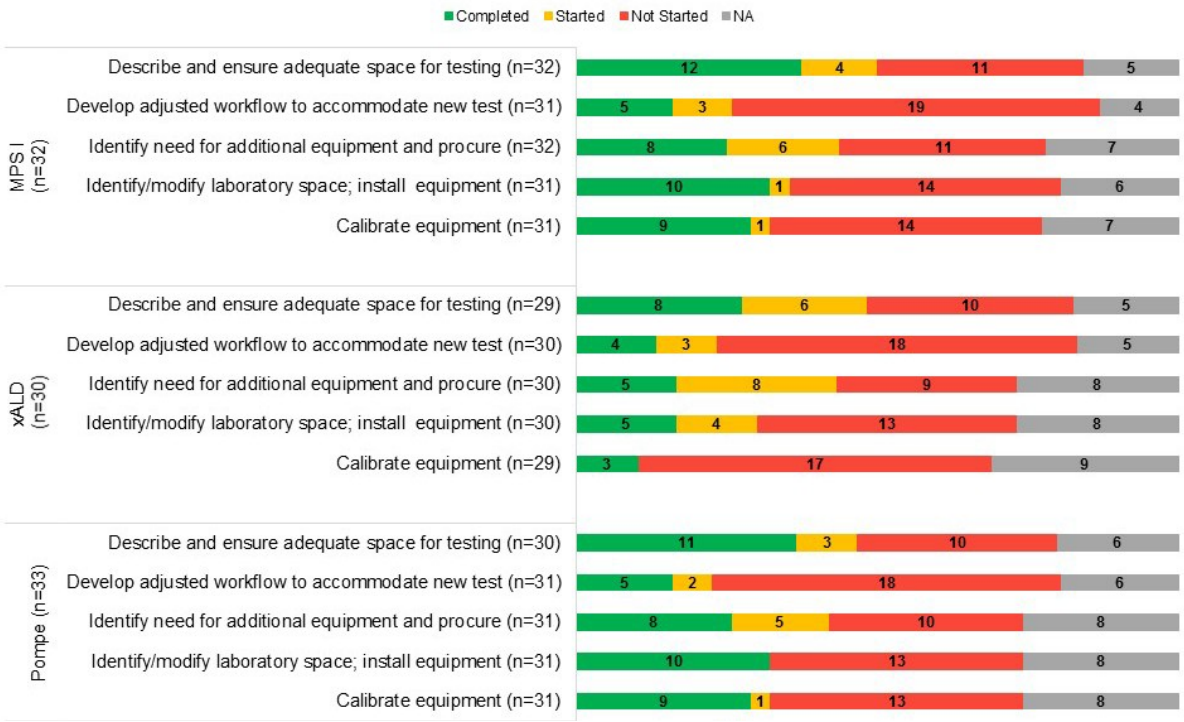
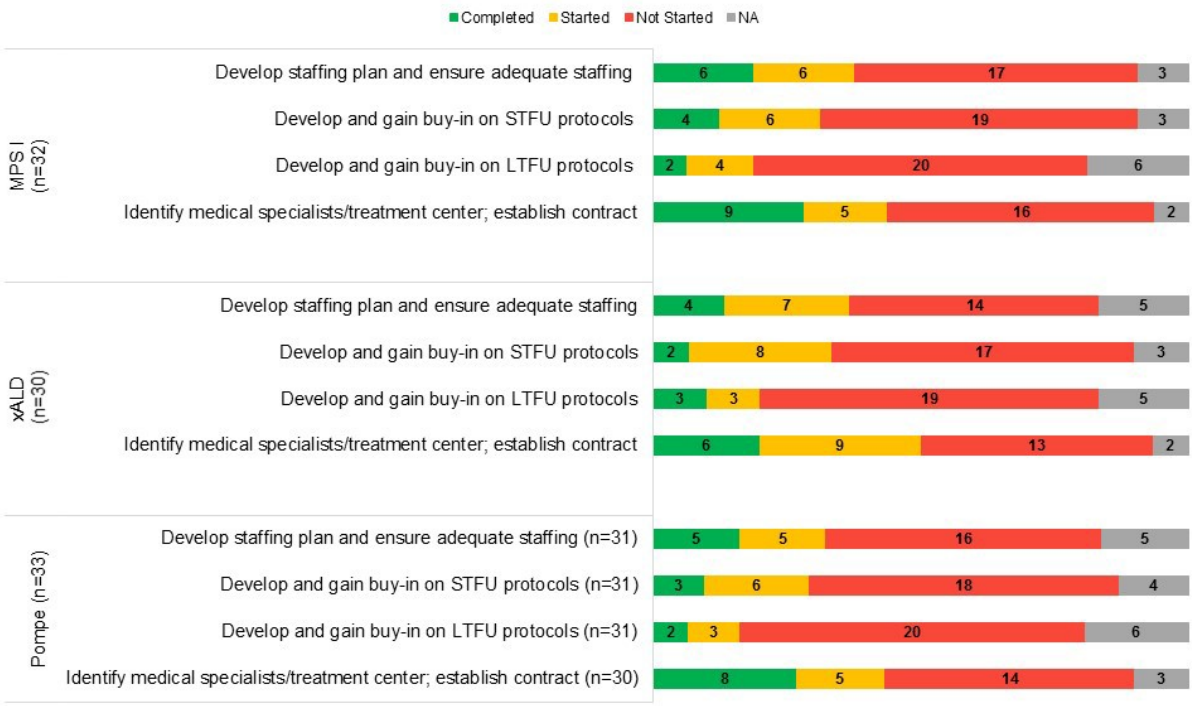


Figure 6: Follow-Up Readiness within the United States



MEETING SUMMARIES

The purpose of this meeting was for newborn screening program personnel to: (1) learn from those who have implemented one of the three new disorders, (2) learn from clinical experts on the three new disorders, (3) identify what questions the programs should ask when they consider implementation of newborn screening for Pompe, X-ALD, and MPS I, and (4) to ask each other questions and share ideas.

The cross-cutting considerations highlighted and summarized below are stratified into various sections, but there is overlap between sections. NewSTEPs.org will continue to collect and share practices with the community to address the considerations identified below. In subsequent sections of this report, presentations provided by laboratory, follow-up and clinical experts address questions on a state-by-state basis and may serve as practices to consider. Please reach out to Kshea.Hale@aphl.org if you would like additional information from a particular newborn screening program or more detailed information regarding any of the considerations highlighted below.

Cross-Cutting Considerations

This section includes the questions and considerations that were brought up during the presentations and interactive discussions, organized by thematic category.

Cross-cutting laboratory considerations for implementing new disorder newborn screening

- How many screening tiers are required for the laboratory algorithm for each disorder?
  - Possible solutions:
    - First tier only
      - Is it acceptable to begin screening by enzyme analysis in the absence of a second tier solution to distinguish pseudo-deficiencies, late onset, etc.?
        - Some programs are choosing this option.
    - First tier reflex to second tier (biochemical and/or molecular)
      - FDA approved assays versus Laboratory Developed Tests versus Homebrew assays
      - Biochemical
        - Advantage: fast, inexpensive
        - Disadvantage: ongoing assay maintenance
      - Molecular
        - Should pseudo-deficiencies be reflexed in NBS laboratory vs. testing in the clinical lab?
        - Newborn screening programs are currently utilizing various options in implementing second tier screens for the new disorders.
        - See presentations in Meeting Presentations section below regarding laboratory implementation practices.

- Who performs second tier screens?
    - Possible solutions:
        - In-house
            - Considerations: cost, space, turn-around time, trained staff.
        - One versus two screen states: do algorithms change?
            - Is second tier screening conducted on first screen or on the second screen?
            - As more two screen states implement new disorder newborn screening, the algorithms will become available.
        - Outsource to Peer Network Resource Center, Regional Laboratory or Commercial Laboratory
            - Considerations: Turn-around time and mechanisms in place to report results.
- Establishing cut-offs
    - Considerations include seasonal changes, humidity, age, birthweight, prematurity, assay type, instrument used.
    - Fixed versus floating cut-offs
        - Percent of daily mean versus percent of daily median
    - Are there analytic values that can help distinguish early onset variants (prior to availability of genotype results)
    - Tools for establishing cut-offs
        - Peer data
        - CLIR
        - Percent of daily means/medians
- Staffing Needs
    - Contingent on size of state, population and methods used
    - Full Time staff versus contractor mechanism
- Screening algorithms will vary depending on state and disorder. NewSTEPs will collate these algorithms on www.newsteps.org Resource Library in the coming months.

Cross-cutting follow-up considerations for implementing new disorder newborn screening

- Where does short term follow-up end and extended short term follow-up begin and transition to long term follow-up?
- How to address the shortage of a sub-specialist workforce?
- Identification of treatment centers and access to clinical intervention
- Education of follow-up workforce
    - Heighten knowledge of molecular terminology as well as how to convey information to primary care providers
    - Just-in-time information for primary care providers
- Mutation reporting strategies
    - Variants of unknown significance versus known disease causing mutations
- Early versus late onset tracking and communicating

- Reporting strategy for incidental findings

Cross-cutting reporting considerations for implementing new disorder newborn screening

- Should reporting results on newborn screening reports include sequencing results?
- Are both normal and abnormal screens reported?
- How are various forms of disease reported? Mutation reporting?
- Electronic reporting codes for new disorders
- Integration of ordering and reporting in Laboratory Information Management Systems (LIMS)

Cross-cutting clinical considerations for implementing new disorder newborn screening

- Who incurs the cost of therapy? Insurance issues? Does Medicaid cover treatment?
- Are drugs/therapies readily available in clinical centers?
- Is there universal access to therapy?
- Which disorders require time critical treatment?
- Understanding genotype/phenotype correlations
- Pre-symptomatic monitoring
- Clinician support for increased workload of presumptive and false positives

Cross-cutting policy considerations for implementing new disorder newborn screening

- How to communicate with stakeholders when adding new disorders to state screening panels?
    - How to work with advocates and legislators
    - How to work with other newborn screening stakeholders
- Coordinating addition of new disorders with cost analysis and fee increase activities
    - How to overcome the draw of a personal story for decision makers
    - How to realistically assess costs and develop budgets
    - How to successfully negotiate a fee increase
    - How to identify how many new staff are required to screen/follow-up newborns for a new disorder

Cross-cutting education considerations for implementing new disorder newborn screening

- Education of parents and providers is imperative, particularly when there is a lack of general knowledge about very rare disorders
- Community engagement
- Accuracy versus accessibility in information
- Advance education versus just-in-time education
- Leveraging the resources of advocacy organizations

Meeting Presentation and Summaries

The presentations during the first day of the national meeting focused on providing insight on considerations and practices around implementation of newborn screening for Pompe, MPS I, and X-ALD. For each disorder there was a presentation from a laboratory that was screening for the condition, a program performing follow-up for the condition, and a clinical specialist to provide insight on the manifestation and progression of the disease. Below are the highlights from those presentations as well as links to the speaker's PowerPoint presentations, when available.

Pompe Disease

Pompe was the first disorder presented during the national meeting, with the panel offering two distinct laboratory perspectives- from a program mandated to screen and a program screening voluntarily. These are summarized below.

Laboratory Presentation (Mandated Screening): ***Patrick Hopkins, Missouri Department of Health***

- PowerPoint slides linked here
- Summary notes:
  - Missouri initiated Pompe newborn screening by mandate; being the first state to universally offer newborn screening for Pompe.
  - Utilize the digital microfluidics platform
  - Challenges: new platform, shifts in workload/staff, development of cut-offs, impact of humidity on enzymes
  - Benefits: multiplexing multiple lysosomal disorders helped to detect compromised samples.

Laboratory Presentation (Voluntary Screening): ***Michele Caggana, ScD, FACMG, New York Department of Health***

- PowerPoint slides linked here
- Summary notes:
  - New York initiated Pompe screening via a consented pilot in 2012
  - 93% uptake of pilot
  - October 1, 2014: universal screening began in New York for Pompe

Follow-Up Presentation: ***Sharmini Rogers (delivered by Patrick Hopkins on Ms. Rogers' behalf), Missouri Department of Health***

- PowerPoint slides linked here
- Summary notes:
  - Missouri convened a Lysosomal Storage Disorders (LSDs) taskforce

- Considerations: funding and staff, data system changes, guidelines for follow-up, required confirmatory tests, education for parents and providers, exploration of long term follow-up.

Clinical Considerations for Pompe Disease: ***Priya Kishnani, MD, Duke University Medical Center***

- Summary notes:
  - Pompe is a deficiency of the GAA enzyme and is multi-systemic in its presentation with a single continuum of disease. Muscle damage manifests with clinical variability.
  - The rate of clinical deterioration is faster in the infantile onset of the disease.
  - Pulmonary and neurological presentation are possible.
  - In the infantile form there is also cardiac and genetic presentation.
  - Later onset of the disease can manifest with a wide clinical spectrum. Pompe can appear as non-classical infantile at one year of age. This is not an adult-only disorder in its late onset form.
  - Follow-up of patients detected by newborn screening is necessary to quantitate progression of disease.
  - DNA analysis allows for confirmation of disease as well as informs disease management.
  - Early initiation of treatment is critical. Treatment can include chemotherapy and Enzyme Replacement Therapy (ERT).
  - Prevalence ranges from 1 in 9,000 to 1 in 24,00 (including later onset forms)

Mucopolysaccharidosis Type I

As with Pompe, there was a presentation on screening for MPS I from a laboratory viewpoint, short-term follow-up perspective as well as clinical specialist explanation of diagnosis and progression.

Laboratory Presentation: ***Rong Shao, MD, Illinois Department of Public Health***

- PowerPoint slides linked here
- Summary notes:
  - Key decision points: test methodology that enabled multiplexing, availability of instrumentation and reagents, ease of hiring additional staff.
  - Changes required to prepare for screening: instrument purchase, laboratory construction, hiring and training of three additional laboratory technicians, initiation of Saturday shifts due to 17 hour incubation period, integration of test receipt and ordering in Laboratory Information Management Systems (LIMS).
  - Cut-off ranges identified following analysis of ~12k de-identified random samples for enzyme activity distribution.

Follow-Up Presentation: ***Claudia Nash, MS, Illinois Department of Health***

- PowerPoint Slides linked here
- Summary notes:
  - Added screening for 5 lysosomal storage disorders in June 2015.
  - Administrative code change defined criteria for designation of specialists and increased the newborn screening fee.
  - The program hired two full time follow-up staff to support the additional testing.
  - 7 hospital systems are designated referral centers.
  - A multi-disciplinary lysosomal storage disorders subcommittee comprised of staff from all referral centers meets monthly to provide input on what diagnostic and long term follow-up data to collect and to establish standardized clinical diagnostic protocols.
  - Follow-up and reporting protocols are similar to other newborn screening disorders. Results are reported to the primary care physician by phone, fax or email.
  - Data elements required changes to the PerkinElmer database, the development of a consent form, and determination of the diagnostic information to collect.
  - Educational resources, including a physician fact sheet were developed.
  - Expect the unexpected.
  - Insurance issues included lengthy delays in molecular testing approval (four to eight weeks) as well as denial of coverage in some cases.
  - There were challenges associated with case categorization as well, including considerations around variants of unknown significance and pseudo-deficiencies.

Clinical Considerations for MPS I: ***Chester Whitley, PhD, MD, University of Minnesota***

- Summary notes:
  - Establishment of the Lysosomal Storage Disease Network with National Institutes of Health (NIH) resources.
  - MPS Type I is a lysosomal a-L-iduronidase enzyme metabolic defect.
  - Disorders should be referred to as lysosomal disorders as opposed to lysosomal storage disorders due to the fact that all of the enzyme activity is not limited to the storage component of the lysosome.
  - MPS I may present with orthopedic constraints including in the spine, upper and lower extremities.
  - The path to diagnosis typically involved clinical suspicion followed by an urgent referral followed by a definitive diagnosis by substrate assay (urine GAG) or enzyme assay (gold standard) and DNA testing.
  - Newborn screening has been a powerful, disruptive shift in the diagnostic paradigm, enabling earlier detection and treatment. For every month that a treatment is delayed, a child's IQ drops by 1.6 points.
  - Hematopoietic stem cell therapy is a standard-of-care (bone marrow, umbilical cord blood), and addition of enzyme replacement therapy is effective in improving cognitive functioning.

X-Linked Adrenoleukodystrophy

The final set of presentations day one focused on X-ALD.

Laboratory Presentation: ***Adrienne Manning, Connecticut Department of Public Health***

- PowerPoint slides linked here
- Summary Notes:
    - Parent advocacy groups served as a driving factor in the statute to screen for new disorders
    - Implementation timeline: bill introduced in July 2013 and live for screening on July 1, 2016.
    - Methodology: HPLC MS/MS, using CDC assay followed by diagnostic tests at Kennedy Krieger and Baylor laboratories.
    - Screening results since October 2015: 15 screen positives, 9 confirmed, 2 siblings identified, 1 Zellweger.

Follow-Up Presentation: ***Lisa Feuchtbaum, DrPH, MPH, California Department of Public Health***

- Summary Notes:
    - 500,000 newborns screened annually.
    - Three tier approach: FIA MS/MS measuring C26 followed by LC MS/MS followed by sequencing at Greenwood Genetics Center.
    - Challenge: new disorders new newborn screening performed at central laboratory whereas routine newborn screening performed across five contract laboratories.
    - All X-ALD second tier screen positives appear as “headline cases” where follow-up coordinators contact the primary care physician who then refers to a preferred metabolic center (115 in state).
    - Dynamic definition for short term follow-up with X-ALD screening, requiring education, newsletters, announcements and interpretation of uncertain molecular findings, diagnoses dependent on the ability to follow-up on children and timeframes.
    - Timeliness changes when sequencing is introduced.
    - Large number of unknowns associated with sequencing results. Guidelines for care are introduced, with a standardized approach to resolving ALD cases in the computer system, coupled with monthly meetings with specialists and routine data review by central lab for consistency.
    - Extended long term follow-up occurs up to age 21.

Clinical Considerations for X-ALD Presentation: ***Paul Orchard, MD, University of Minnesota***

- PowerPoint slides linked here
- Summary Notes:
    - X-ALD caused by defect in ABDC1 gene resulting in the inability to transport fatty acids into peroxisome.
    - Phenotypes: childhood cerebral, adolescent cerebral, adult cerebral, Adrenomyloneuropathy/spinal cord disease
    - Adrenal insufficiency in ALD exacerbated by stress (prevalence 80%).
    - Newborn screening for ALD is critical, resulting in decrease in deaths from adrenal insufficiency, reduction in lifelong disability, identification of family members, ongoing monitoring of disease, identification of X-ALD before demyelination occurs.

Technology Considerations

The planning committee for the national meeting requested that there be an opportunity for newborn screening programs to have a Question and Answer session with vendors who create testing platforms that can be used to screen for Pompe, MPS I and X-ALD. The vendors were asked to address the following questions:

1. What do you have to offer for assisting with screening for these three new disorders?
2. How do we know it works well in a newborn screening system?
3. Are you seeking FDA approval for your assays? What is the estimated timeline?
4. How much does it cost per test via reagent rental or versus buying the instruments?
5. What is the footprint, Turn-Around Time and throughput?

The following notes include the information provided during the short (oral) vendor presentations and the answers the vendor representatives provided when responding to audience questions.

- Summary notes:
  - Baebies
    - Seeker platform, FDA approved for lysosomal storage disorders: MPS I, Pompe, Gaucher, Fabry
    - No daily maintenance required
    - Same day referral; timely instrument runs
    - Plug and play system; allows for cross-training
    - $1 per test per baby
    - No false negatives reported to date; false positives consistent with other disorders.
  - PerkinElmer
    - Expanded amino acid acylcarnitine panel (including X-ALD and additional analytes for urea cycle disorders).
    - Throughput: 2 minutes per sample
    - Currently in late clinical validation phase, with submission to FDA expected early next year.
    - Pipeline product for lysosomal storage disorders: 6-plex assay for Neimann Pick, Krabbe, Gaucher, Fabry, MPS I and Pompe.
    - 18 hour incubation in aqueous buffer
    - Same instrument being used for neobase assay
  - Illumina
    - Sequencing/genomics solution (sequencing by synthesis)
    - Targeted panel, 3.5 day turn-around time.
    - Long term goal of seeking FDA approval for broad panel
    - Targeted panel for lysosomal storage disorders
    - Illumina can help build customized, targeted panels

Short- and Long- Term Follow-Up Considerations

The objective of this session was to encourage programs to consider the spectrum between short- and long-term follow-up particularly with the screening of the newest disorders added to the RUSP.

Short Term Follow-Up Considerations: ***Amy Gaviglio, MS, CGC, Minnesota Department of Health***

Long Term Follow-Up Considerations: ***Anthony Steyermark, PhD, Minnesota Department of Health***

- Summary presentations linked here and here
- Summary notes:
    - Long Term Follow-Up
        - Families are contacted by the LTFU program a month after diagnosis.
        - Nursing assessments occur at one month, 1 year and 4 year intervals.
        - Engaged with legislature and council of health plan to identify gaps in coverage for medical formulas/foods; Identifying if health insurance is meeting needs.
        - Language barriers contribute to late diagnosis; as well as transportation barriers and insurance status.
        - Minnesota is developing follow-up protocols for late onset forms of disorders.
        - Program engages families to assess what their long term follow-up needs are.
    - Short Term Follow-Up
        - Pre-analytical considerations: improving information dissemination prior to screening
        - Analytical considerations: understanding of testing approach
        - Post-analytical considerations: parents report being under-informed and overwhelmed.
        - The greater the understanding of the patient, the greater the recall of the patient, the greater the satisfaction of the patient and all of this will lead to better adherence over time.
        - Considerations in confirming a case: nice to know versus need to know information; capturing discrete fields and utilization of web portals; electronic reporting requirements.
        - Continuous quality improvement is critical in follow-up.

Policy Considerations

This presentation discussed the role that APHL can play in assisting states in navigating Phase 1 – Legislative/Mandate Phase (Figure 4). The presentation also allowed for discussion amongst meeting attendees about the impact of adding new conditions to their newborn screening program day-to-day work.

Kimberly Piper, RN, BS, CPH, CPHG, Iowa Department of Public Health

Celia Hagen, MPH, Association of Public Health Laboratories

- Summary notes:
    - The Association of Public Health Laboratories is exploring state needs focused on the addition of new disorders, whether mandatory of voluntary.
    - APHL can provide assistance to programs faced with legislative barriers.
    - APHL can help connect states to NBS partners, help garner support from state health officials and provide education to parent advocacy groups.
    - Resources:
        - APHL Legal and Legislative Issues in Newborn Screening workgroup: discuss NBS legal and legislative topics.
        - Legislation tracking: database monitoring newly introduced NBS legislation is being proactively tracked by APHL.

Education Considerations

This final presentation from Baby's First Test staff focused on the importance of education for families, providers and the general public. Baby's First Test is partnering with APHL to convene new disorder specific task forces. Results of data presented from these task forces are summarized below.

Natasha Bonhomme, Baby's First Test

Amelia Mumford, Baby's First Test

- Summary notes:
    - Awareness versus education versus training versus engagement: all of these are unique
        - Awareness = exposure to info
        - Education = imparting knowledge and tools
        - Training = imparting "how to" /process knowledge
        - Engagement = bi-directional process of collaboration
    - Education is a right and necessity
    - May communicate life-saving info
    - Helps prepare families for what is ahead
    - Encourages families to be proactive
    - Spreads from individual to friends, family and community
    - Supports families in feeling empowered, informed and active participants in child's care

- Key factors in how people access information:
    - Medicaid covers about 50% of births in the country
    - Millennial parents have $200B spending power
    - 87% of adults are online
    - 64% pregnant women access info from smartphone
- NBS is becoming increasingly complex; more rare disorders, ability to detect carriers, etc.
- Only 12% of adults have proficient health literacy, almost 10% of the United States population is considered limited English proficient.
- Baby's First Test facilitates 3 specific condition workgroups (X-ALD, MPS I, Pompe) with the following core activities: to aggregate existing educational materials, to organize and create materials for families and providers and to produce a template sheet/materials
- Unifying themes resulting from workgroups:
    - Lack of condition-specific knowledge (providers)
        - Many medical professionals have not received sufficient education/training on these disorders
        - New challenges for clinicians accustomed to seeing older children
        - Bottom line, providers need educational support
    - Disjointed care team
        - Shifting multidisciplinary care team
        - Access to specialists, travel
        - Education is ideally a team effort, but communication can be lacking
    - Families as condition experts
        - Provide continuity
        - "Inverse" flow of information: families educate providers
        - Parents learn to be effective advocates – but is this an additional burden?
    - Individuality of the disorders
        - Different presentation and progression between patients; "no child is textbook"
        - Condition continuum/spectrum; nuances can be lost in rigid classical definitions
        - Temper expectations and assumptions
    - Value of connection with other families
        - Message – "you're not alone"
        - Convey uniqueness and variability of experience of living with condition
        - Education through personal stories
        - Establish community and emotional support system
    - Simplicity, clarify, depth of language

        - Many terms refer to same condition; lack of consistency can lead to confusion
        - Overuse of acronyms or classifiers without context
        - Accuracy vs accessibility (what is the content – how is it conveyed – how is it tailored to who's receiving it)
- Role of the web and social media
    - Significant online presence of advocacy groups
    - Support groups and discussion boards
    - Double-edged sword of internet searches (too much information, information may be scary, but provider may not be informed/educated about condition)
- What, how much, when
    - Well in advance vs just in time (varies from state to state, whether state reports pseudo-deficiency, etc.)
    - Range of information-seeking behaviors and preferences
    - Education is an ongoing process
    - Access vs "data dump"
- Key role of advocacy organizations
    - Wealth of information already available through advocacy groups
    - Synthesize research, news, family testimonials, policy activities, clinical/drug trials, insurance coverage, treatment updates, etc.
    - Partnership building

APPENDICES

Peer Network Resource Centers

As program needs for new disorder screening evolve, NewSTEPs can help evaluate those needs and offer support that will help you achieve your goals. A part of the support network is Peer Network Resource Centers, with the offerings by each detailed below. For more information please contact Kshea Hale at Kshea.Hale@aphl.org

Summary of Services Offered by Missouri

- Technical assistance for Lysosomal Storage Disorder (LSD) testing method validation and screening implementation
- Education for LSD screening and follow-up implementation
- First Tier Testing* (for emergency/continuity of operations purposes)
- Second Tier Testing* using Digital Microfluidics for four LSDs (Pompe, MPS I, Fabry, and Gaucher) to assist in method validation, pilot and implementation phases

Summary of Services Offered by New York

- Technical assistance for Pompe, MPS I and X-ALD
- Education for Pompe, MPS I and X-ALD
- Pompe, X-ALD and MPS I next-generation sequencing (currently under development)
- First Tier Testing* using tandem mass spectrometry (MS/MS) for Pompe, MPS I and X-ALD
- Second Tier Testing* using DNA sequence analysis for the LSDs and HPLC MS/MS for X-ALD
- Third Tier Testing* using DNA sequencing analysis for X-ALD

Summary of Services Offered by Wisconsin

- Technical assistance for Pompe
- Second Tier Testing* for Pompe, MPS I and X-ALD using DNA Sequencing Analysis
- Pompe newborn screening Educational materials for parents
- Pompe newborn screening materials for primary care providers

*Please note that all programs who choose to utilize the PNRCs to perform screening for their programs will need to work directly with the PNRC to establish payment and specimen transport mechanisms.

Meeting Participants

Name	State/Organization	Email Address
STATE NEWBORN SCREENING PROGRAM REPPRESENTATIVES		
Rasoul Koupaei	California	Rasoul.Koupaei@cdph.ca.gov
Lisa Feuchtbaum	California	Lisa.Feuchtbaum@cdph.ca.gov
Bonita Taffe	Florida	Bonita.Taffe@flhealth.gov
Emily Reeves	Florida	Emily.Reeves@flhealth.gov
Dusty Edwards	Florida	Dusty.Edwards@flhealth.gov
Carol Johnson	Iowa	carol-johnson@uiowa.edu
Kimberly Noble Piper	Iowa	Kimberly.piper@idph.iowa.gov
Sarah VanGorp	Iowa	Sarah.VanGorp@idph.iowa.gov
Kerri-Lynn Lockwood	Michigan	lockwoodk@michigan.gov
Mike Sarzynski	Michigan	SarzynskiM@michigan.gov
Mary Seerterlin	Michigan	SeeterlinM@michigan.gov
Amy Gaviglio	Minnesota	amy.gaviglio@state.mn.us
Tony Steyermark	Minnesota	tony.steyermark@state.mn.us
Krystal Baumert	Nebraska	Krystal.Baumert@nebraska.gov
Karen Eveans	Nebraska	Karen.Eveans@nebraska.gov
Julie Luedtke	Nebraska	Julie.Luedtke@nebraska.gov
Suzanne Canuso	New Jersey	Suzanne.canuso@doh.nj.gov
Donna McCourt	New Jersey	donna.mccourt@doh.nj.gov
Dr. Kristin Clinard	North Carolina	kclinard@email.unc.edu
Dr. Joe Muenzer	North Carolina	muenzer@med.unc.edu
Hari Patel	North Carolina	hari.patel@dhhs.nc.gov
Rosemary Hage	Ohio	Rosemary.Hage@odh.ohio.gov
Sharon Linard	Ohio	Sharon.Linard@odh.ohio.gov
M. Christine Dorley	Tennessee	M.Christine.Dorley@tn.gov
Ashley Porter	Tennessee	Ashley.M.Porter@tn.gov
George Dizikes	Tennessee	george.dizikes@tn.gov
Katlyn Le	Texas	Katlyn.Le@dshs.texas.gov
D'Andra Luna	Texas	Dandra.Luna@dshs.texas.gov
Felipe Rocha	Texas	Felipe.Rocha@dshs.texas.gov
Tracy Klug	Missouri	Tracy.Klug@health.mo.gov
Patrick V. Hopkins	Missouri	Patrick.Hopkins@health.mo.gov
Michele Caggana	New York	michele.caggana@health.ny.gov
Mei W. Baker	Wisconsin	mei.baker@slh.wisc.edu
Dr. Josh Hyman	Wisconsin	jrhyman@wisc.edu
Leslie Himstedt	Arkansas	Leslie.Himstedt@arkansas.gov
Kostas Petritis	Arizona	Kostas.Petritis@azdhs.gov
Darren Michael	Colorado	darren.michael@state.co.us

Adrienne Manning	Connecticut	Adrienne.Manning@ct.gov
Marie Burlette	Connecticut	Marie.A.Burlette@ct.gov
Arthur Hagar	Georgia	Arthur.hagar@dph.ga.gov
Angela Wittenauer	Georgia	alwitte@emory.edu
Sylvia Mann	Hawaii	sylvia@hawaiigenetics.org
Frances Shigemasa	Hawaii	Frances.Shigemasa@doh.hawaii.gov
Claudia Nash	Illinois	claudia.nash@illinois.gov
Rong Shao	Illinois	Rong.Shao@Illinois.gov
Megan Griffie	Indiana	MGriffie@isdh.in.gov
Shirley Helms	Maine	shirley.helms@maine.gov
Wendy Smith	Maine	smithw@mmc.org
Adam Coleman	Maryland	Adam.Coleman@maryland.gov
Johnna Watson	Maryland	Johnna.Watson@Maryland.gov
Roger Eaton	Massachusetts	roger.eaton@umassmed.edu
Inderneel Sahai	Massachusetts	Inderneel.sahai@umassmed.edu
Natalye Jones	Mississippi	Natalye.Jones@msdh.ms.gov
Alyce Stewart	Mississippi	Alyce.Stewart@msdh.ms.gov
Linda L. Kincaid	New Hampshire	Linda.Kincaid@dhhs.nh.gov
Katie Bentz	North Dakota	kbentz@nd.gov
Lisa Caton	Oklahoma	LisaRC@health.ok.gov
Tonya McCallister	Oklahoma	TonyaJ@health.ok.gov
Christianne Biggs	Oregon	Christianne.biggs@state.or.us
Kim Billow	Pennsylvania	kbillow@pa.gov
PJ Borandi	Perkin Elmer (PA)	PJ.Borandi@PERKINELMER.COM
Sulay Rivera-Sánchez	Puerto Rico	sulay.rivera@upr.edu
Tanya Spells	South Carolina	spellsty@dhec.sc.gov
Sandi Hall	South Carolina	Hallss@dhec.sc.gov
Kim Hart	Utah	kimhart@utah.gov
Cindy Ingham	Vermont	Cindy.Ingham@vermont.gov
Willie Andrews	Virginia	willie.andrews@dgs.virginia.gov
Jennifer Macdonald	Virginia	Jennifer.Macdonald@vdh.virginia.gov
Lani Culley	Washington	lani.culley@doh.wa.gov
Bill Hoffman	Washington	Bill.Hoffman@DOH.WA.GOV
PARTNERS		
Natasha Bonhomme	Genetic Alliance	nbonhomme@geneticalliance.org
Amelia Mulford	Genetic Alliance	amulford@geneticalliance.org
Amy Brower	ACMG	abrower@acmg.net
Joanne Adelburg	ACMG	jadelberg@acmg.net
Helena Dessie	ACMG	hdessie@acmg.net
Megan Lyon	ACMG	mlyon@acmg.net
Michael Watson	ACMG	mwatson@acmg.net

STAFF		
Oluwafunke Akinsola	APHL	Oluwafunke.Akinsola@aphl.org
Sari Edelman	APHL	sari.edelman@aphl.org
Erin Darby	APHL	erin.darby@aphl.org
Kshea Hale	APHL	kshea.hale@aphl.org
Celia Hagan	APHL	celia.hagan@aphl.org
Jelili Ojodu	APHL	Jelili.Ojodu@aphl.org
Laura Russell	APHL	laura.russell@aphl.org
Ruthanne Sheller	APHL	ruthanne.sheller@aphl.org
Sikha Singh	APHL	sikha.singh@aphl.org
Careema Yusuf	APHL	careema.yusuf@aphl.org
Guisou Zarbalian	APHL	guisou.zarbalian@aphl.org
Yvonne Kellar-Guenther	University of Colorado	Yvonne.Kellar-Guenther@ucdenver.edu
Sarah McKasson	University of Colorado	sarah.mckasson@ucdenver.edu
Joshua Miller	University of Colorado	Joshua.I.Miller@ucdenver.edu
Marci Sontag	University of Colorado	Marci.Sontag@ucdenver.edu
CLINICAL EXPERTS		
Priya Kishnani	Duke University	priya.kishnani@duke.edu
Chester Whitley	University of MN Twin Cities	whitley@umn.edu
Paul Orchard	University of MN Twin Cities	orcha001@umn.edu
VENDORS		
Sricharan Bandhakavi	Perkin Elmer	sricharan.bandhakavi@perkinelmer.com
Rongcong Wu	Perkin Elmer	Rongcong.wu@perkinelmer.com
LeeAnn Dennewitz	Perkin Elmer	leeann.dennewitz@perkinelmer.com
Manjula Chelliserry	Illumina	mchelliserry@illumina.com
Dan Heard	Illumina	dheard@illumina.com
Jon Washburn	Baebies	JWashburn@baebies.com
Candice Brannen	Baebies	cbrannen@baebies.com
Vamsee Pamula	Baebies	VPamula@baebies.com