Considerations for adding conditions to state newborn screening panels

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About APHL

• **Vision:**
  – A healthier world through quality laboratory systems.

• **Mission:**
  – Shape national and global health outcomes by promoting the value and contributions of public health laboratories and continuously improving the public health laboratory system and practice.
What is a public health laboratory?
A public health laboratory...

- Is located in every state, territory and the District of Columbia
- Monitor and detect threats
  - Food Safety
  - Emergency Response
  - Environmental Health
  - Newborn Screening
Population – Based Risk Assessment

Tests must be universally available and timely

Credit: Dr. Michele Caggana
Basics of Newborn Screening

- Three screens
  - Heel stick (dried blood spot)
  - Hearing loss
  - Critical congenital heart disease
- Conditions screened for are rare
- Newborn is usually asymptomatic
Newborn Screening Process

Pre-analytic Phase
- Birth
- Dried Blood Spot Collection, Point-of-care Tests
- Dried Blood Spot Shipment
- Arrival/Receipt at Lab
- Data Entry

Analytic → Post-analytic Phase
- Lab Processing/Testing
- Report Results
- Diagnosis confirmed, Intervention initiated
Components of a Newborn Screening System
Basics of Newborn Screening

- Advisory Committee on Heritable Conditions in Newborns and Children (ACHDNC)
  - Recommended Uniform Screening Panel (RUSP)

Evidence Review → ACHDNC Vote → Secretary of HHS Acceptance → Addition to State Screening Panels

Engagement of Advocacy Groups → Not a mandate → State by state discretion; State Advisory Committee makes the final decision
Basics of Newborn Screening
• State-based and mandated public health programs
What goes into adding a disorder to a state’s NBS panel?

Ongoing internal communication – weekly or biweekly

**Phase 1**
- Obtain approval
- Determine test method
- ID staffing needs
- Develop budget

**Phase 2**
- Obtain equipment
- Perform validation study/studies
- Define follow-up algorithms

**Phase 3**
- Integrate testing into current workflow
- Develop fact sheets, follow-up letters
- ID follow-up data needs

**Phase 4**
- Build and test cutoffs and logic into laboratory information management system
- Issue press release
- Notify health care providers

**Go Live and Post-go Live**
Steps to adding disorders to state NBS panels: there’s more to it
Average time to implementation

- Pompe: average based on 9 states that implemented between 2015 – 2019
- MPS-I: average based on 10 states that implemented between 2016 – 2019
- X-ALD: average based on 7 states that implemented between 2016 – 2019
Barriers to implementation

- Availability of a validated screening test
- Ensuring availability and readiness of specialists
- Long-term follow-up for carriers (depending on the disorder)
- Laboratory information management system (LIMS) incorporation
- Receiving support for a fee increase
- Developing follow-up algorithms and processes with a new group of specialists (depending on the disorder)
What’s Next?

• Federal: appropriations/authorization
  – House-passed 2021 appropriation - $21.8 (+$4m) for HRSA; $20.2m (+$2m) for CDC
  – House-passed authorization – H.R. 2507
  – Senate ?

• States
  – Considering addition of new disorders
A Premier NBS Community

NewSTEPs is a national newborn screening resource center designed to provide data, technical assistance and training to newborn screening programs and assist states with quality improvement initiatives.

What does a profile get you?
A NewSTEPs login permits access to the NewSTEPs data repository (data access restricted according to user role), and allows users to save NewSTEPs resources in a centralized location.

Sign up for the NewSTEPs CoLiABorate Community to always stay connected.
Questions?