Newborn Programs 101

NCSL: MCH Fellow Meeting

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state and local public health labs, state environmental and agricultural labs

Environmental Health

Emergency Preparedness

Informatics

Newborn Screening

Food Safety

Infectious Disease
Newborn Screening

1. Blood screen
   - Heel stick

2. Hearing screen
   - Hearing screen

3. Heart screen
   - Pulse oximetry
The Newborn Screening Story
How One Simple Test Changed Lives, Science, and Health in America
Public Health Laboratory NBS Program

Virtually all of the roughly four million babies born in the US each year receive newborn screening!
HEEL STICK
Within 48 hours of birth and before a baby leaves the hospital, a healthcare provider pricks the baby's heel to get a few drops of blood. The blood drops are placed and dried on a special paper.
A few drops of blood on filter paper are divided into many specimens so newborn screening labs can perform the multiple tests needed.

**SHIPPING AND TESTING**
Within 24 hours of the heel stick, the paper with blood drops should be sent to a newborn screening lab for testing.

**LAB RESULTS**
Laboratory results should be shared with the baby’s provider within five days of birth for time-critical conditions, within seven for all others.
FOLLOW UP

All newborn screening results should be reported to the baby’s provider within seven days of birth.
Positive screen results require further testing and immediate follow up.

NEGATIVE SCREEN

- Provider is notified.
- Provider should follow up with the baby’s family.
If parents don’t hear about results, call and ask the provider.

POSITIVE SCREEN

- Provider is notified.
- Provider follows up with baby’s family for further testing.
Diagnostic tests must be done immediately to confirm results and intervention should begin as soon as possible.
What disorders are tested for?

Metabolic Disorders

Organic acid conditions
- Propionic acidemia
- Methylmalonic acidemia (methylmalonyl-CoA mutase)
- Methylmalonic acidemia (cobalamin disorders)
- Isovaleric acidemia (IVA)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-Hydroxy-3-methylglutaric aciduria
- Holocarboxylase synthase deficiency
- 6-Ketothiolase deficiency
- Glutaric acidemia type I (GA I)

Fatty acid oxidation disorders
- Carnitine uptake defect/carnitine transport defect
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Very long-chain acyl-CoA dehydrogenase deficiency
- Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency
- Trifunctional protein deficiency

Amino acid disorders
- Argininosuccinic aciduria
- Citrulinemia, type I
- Maple syrup urine disease
- Homocystinuria

Endocrine Disorders
- Primary congenital hypothyroidism
- Congenital adrenal hyperplasia (CAH)
- Tyrosinemia, type I

Hemoglobin Disorders
- S,S disease (Sickle cell anemia)
- S, β-thalassemia
- S,C disease

Other Disorders
- Biotinidase deficiency
- Critical congenital heart disease (CCHD)
- Cystic fibrosis
- Classic galactosemia
- Hearing loss
- Severe combined immunodeficiencies (SCID)
ADDING A DISORDER &
THE ROLE OF THE ADVISORY COMMITTEE
RUSP
Recommended Uniformed Screening Panel

A multi-disciplinary group or individual submits a nomination package to ACHDNC to consider adding a disorder to the RUSP.

ACHDNC reviews the package that describes the condition and treatment options. It must contain evidence-based information, including:
1. Validation of laboratory screening test
2. Availability and accuracy of a diagnostic test
3. A population-based pilot study.

Based on this information, ACHDNC assesses:
1. The evidence of potential net benefit of screening
2. The ability of states to screen for the disorder
3. The availability of effective treatment.

Based on the recommendation of ACHDNC, the Secretary of the US Department of Health and Human Services approves or denies the recommendation. If approved, the disorder is placed on RUSP.

Once on RUSP, state advisory committees and administrative bodies review the evidence and make a recommendation for addition to its state testing program based on current laws, funding, and the availability of tests and treatment.
Which states and territories have a newborn screening advisory committee?

- Advisory Committee Present
- Advisory Committee Not Present
Which state and territory newborn screening advisory committees are mandatory?

- Red: Mandatory
- Blue: Voluntary
- Gray: Data Not Provided

Map of the United States and territories showing states and territories with mandatory newborn screening advisory committees.
Advisory Committee

• Who is on them?
  – Geneticists
  – OBGYN
  – Pediatrician
  – Consumer, family member of a consumer
  – Ethicists
  – Hospital Association
Considerations

• **Sufficient evidence** of the effectiveness of screening for the condition?

• Will the child and family **benefit from early detection**?

• Is **funding** available to support all costs of implementation?
  – including parental education, follow-up, diagnosis, treatment and management, and program evaluation
  – Staffing and administrative capacity to support
For a disorder to be added to a state NBS panel, what should it have?

- A valid laboratory screening test
- Availability and accuracy of a diagnostic test
- Evidence of potential net benefit of screening
- The ability of states to screen for the disorder
- The availability of an effective treatment
Unintended Consequences

- False positives due to a low quality screening test
- Lack of a coordinated follow-up system
- Unanswered questions for parents of children found to have a questionable positive screens
- Lack of adherence to follow-up
- Untimely diagnostic work-up and delayed treatment
Newborn Screening Grows Up

It started with one test, in one state. But technological advancements, increased awareness, and federal action in recent years have spurred sharp growth in the average number of conditions states include in their newborn screening programs.
Private genetic companies versus state NBS programs?
FUNDING
How is the Newborn Screening Program currently funded?

- NBS Fee: 43
- General Funds: 10
- Title V: 7
- Insurance: 2
- Agency funds as needed: 1
- Federal Funds: 1
- Grant Funded: 1
- State-funded fee fund: 1
Newborn Screening is MORE than a series of tests!

NBS Fee

funds

Screening
NBS begins with a small heel prick to collect a few drops of blood on filter paper cards within first 24-48 hours of life. Samples are transported to the public health laboratory for screening.

Testing
Negative (normal) results - Provider is notified and parents are informed of results at baby’s first wellness visit; no further action is needed.

Out-of-range (abnormal) results - Provider is notified and sometimes requests a retest or further testing. Diagnostic tests must be done immediately to confirm results.

Education

Follow-up

Case management
Why is there a range in the fee nation-wide?

1. A state’s annual birth rate
   - Volume of samples
   - Appropriate staffing
   - Coordination of services

2. 1 screen vs 2 screen state
   Double the specimens

3. The # of disorders tested on a NBS state panel

4. Where testing is performed:
   - In state
   - Regional PHL
   - Commercial

NBS Fee Range: $50 - $150
Why such a range in the fee nation-wide?

Variability in services….
How is the NBS fee collected?

**Indirect:** Upfront & Pre-Sold

* NBS Programs that directly bill insurance have more difficulty with reimbursement than those that primarily deal with birthing facilities.
Direct Billing Hurdles

Administrative Burden

• Increased staffing needed
  – Outstanding claims to be addressed
  – Medical billing expertise
  – Constant turn-over

• Denials & Resubmissions

• Insurance company needs differ
  – Patient information: incomplete, not consistent
  – Billing codes/reimbursement
  – Negotiation contracts
No Fee
The NBS Fee is NOT always dedicated for NBS activities! It can be placed into other “funds” and used for other state programs.
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.
SPINAL MUSCULAR DYSTROPHY (SMA)
Public Health Impact

Barrier to implementing screening for SMA

- Access to cost-benefit analysis of SMA
- Cost of kits, supplies, treatment
- Lack of genetic counselors and specialists to perform appropriate follow-up
- Lack of funding
  - Delay in the collection of revenue and Medicaid reimbursement
  - Meet the demand of current testing results and follow-up services while expanding to new disorders
- Cost, access, outcomes of treatment - Spinraza
- Having spending authority
FEDERAL ROLE IN NBS
Newborn Screening Saves Lives Act

The Newborn Screening Quality Assurance Program

The only comprehensive quality assurance program using the dried-blood spots

Laboratory Services Provided by NSQAP

1. Filter paper evaluation
2. DBS reference and quality control materials
3. Proficiency Testing
4. Internet reporting site for laboratories
5. Follow-up of False negative results
6. Training, consultation, network resources
Contact

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