The State of Newborn Screening Systems

Marci Sontag, PhD

www.newsteps.org

NewSTEPS is funded by Cooperative Agreement # U22MC24078 from the Health Resources and Services Administration (HRSA).
“The only constant in newborn screening is the baby”

• Harry Hannon, PhD
Newborn Screening Systems in the United States

• Fifty years of newborn screening
• One disorder in 1963, dozens in 2014
Deadly Delays

The nation's newborn screening programs depend on speed and science to save babies from rare diseases. But thousands of hospitals fall short, deadly delays are ignored and failures are hidden from public view — while babies and their families suffer.

Published Nov. 16, 2013

Delays at hospitals across country undermine newborn screening programs

Interactive: See how states compare and search for problem hospitals

Colton Hidde: Testing delay puts newborn's life in jeopardy

NewSTEPs
A Program of the Association of Public Health Laboratories®
The state of newborn screening systems is strong
What do we know about our newborn screening systems in the U.S.?

- 51 newborn screening programs
- 36 newborn screening labs
- Geographically diverse
  - 663,000 Square Miles to 1,212 square miles
Number of Annual Births in the U.S.
Number of Annual Births in the U.S.

Median – 52,200
Number of Annual Births in the U.S.

50% of states
21,000 – 87,000

States (each bar is one state)
Number of Annual Births in the U.S.
Birth rates vary between states

• The number of live births per 1,000 population varies
• May point to different needs

http://kff.org/other/state-indicator/birth-rate-per-1000/#map
NBS Fees in the United States

- In some states newborn screening fees are only a part of the funding source.
- How are fees used?
  - Lab tests
  - Follow-up
  - Clinical contractors
  - IT services
  - Courier Services
  - Administration
  - Education

<table>
<thead>
<tr>
<th>Description</th>
<th>Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Median</td>
<td>$75.00</td>
</tr>
<tr>
<td>25th percentile</td>
<td>$53.25</td>
</tr>
<tr>
<td>75th percentile</td>
<td>$101.50</td>
</tr>
</tbody>
</table>
Operating hours on Saturdays

Saturday Hours

- Closed
- All day
- Typical hours
- Limited hours

N = 32
Storage of Dried Blood Spots

- Storage of dried blood spots varies in state newborn screening programs
  - Recent legal events have changed the landscape
  - Changes are continuing
Adding new disorders

• Requirements for adding conditions vary by state
  – Legislative action
  – Board of Health/Advisory Committee

• Many states are faced with new screens added through legislative actions
Over 90% of infants are born in states with CCHD Screening Mandates

www.newsteps.org
Over 65% of infants in the U.S. are born in states with SCID mandates
New Conditions Under Consideration

• Decision pending from Secretary of Health Human Services for Pompe
  – Screened in 7 states (under mandates, pilots)
• Secretary’s Discretionary Advisory Committee for Heritable Disorders in Newborns and Children Currently Reviewing:
  – Mucopolysaccharidosis I (MPS-1)
    • Screened/pilots in 4 states
  – X-linked Adrenoleukodystrophy (ALD)
    • Screened/pilots in 6 states
Screened Conditions Not on the Recommended Uniform Screening Panel

<table>
<thead>
<tr>
<th>Condition</th>
<th>Number of States</th>
</tr>
</thead>
<tbody>
<tr>
<td>CPS I</td>
<td>6</td>
</tr>
<tr>
<td>EME</td>
<td>5</td>
</tr>
<tr>
<td>Gaucher</td>
<td>4</td>
</tr>
<tr>
<td>HHH</td>
<td>7</td>
</tr>
<tr>
<td>Krabbe</td>
<td>3</td>
</tr>
<tr>
<td>NKH</td>
<td>4</td>
</tr>
</tbody>
</table>

- **CPS I**: Condition is currently being pilot tested
- **EME**: Universally offered but not yet required
- **Gaucher**: Likely to be detected (and reported)
- **HHH**: Universally required
- **Krabbe**: Universally required
- **NKH**: Universally required
Screened Conditions Not on the Recommended Uniform Screening Panel

- **Screened/Pilots in only one program**
  - Formiminoglutamic acidemia - FIGLU
  - Hyperornithinemia with Gyrate Deficiency - Hyper ORN
  - Mucopolysaccharidosis II - MPS II
  - Prolinemia Type I/ Type II - PRO

- Offered to select populations, or by request
- Condition is currently being pilot tested
- Universally offered but not yet required
- Likely to be detected (and reported)
- Universally required
Making improvements in NBS
March Of Dimes Honors Arizona With First-Ever Newborn Screening Award
Arizona Health Director Honored for Reforms to Avoid Deadly Delays
CHANDLER, ARIZONA — Thursday, September 18, 2014

Arizona turned its newborn screening program into a model other states can follow in just a few short months, earning it the first-ever Newborn Screening Quality Award from the March of Dimes.

The March of Dimes presented Will Humble, M.P.H., Arizona’s Department of Health Services director, with the award at a ceremony held here today. Mr. Humble established a policy of full transparency for the length of time it takes Arizona hospitals to send newborn blood samples to the lab for analysis, and set a target of having 95 percent of samples screened within 72 hours.

“When hospitals hold onto blood samples for a few days, or a lab is closed on the weekend, this can lead to deadly delays for newborns,” said Edward McCabe, MD, March of Dimes chief medical officer, who presented Mr. Humble the award today. “But under Will Humble’s leadership, Arizona has put in place a process that is a model for other states to follow.”

The March of Dimes established the new awards in honor of Dr. Robert Guthrie, known as “the father of newborn screening” for developing the first mass screening test for babies 51 years ago. The awards recognize leadership in establishing culture of safety as a way to avoid those deadly delays in states’ newborn screening process. A Milwaukee Journal Sentinel investigative series highlighted the problem by telling the personal stories of babies who developed intellectual delays when their blood samples were not timely analyzed.

“Our goal is to save lives and prevent devastating disabilities for babies, and the cooperation and quick work of Arizona’s hospital officials and our state Department
Partnering with Colorado Hospital Association, laboratory and nursing staff: “NBS process improvement group”

Mapped out processes of from prenatal care to follow-up

Implementing best practices
Timeliness

Texas

2009 33% of first screen specimens received at greater than 4 days

April 2014 3.7% of first screen specimens received at greater than 4 days
South Carolina

Rapid improvement cycle partnership Hospital Association and Lab: Phased in Saturday and Holiday Testing and follow-up testing

47% shipped within 24 hours of collection at baseline

65% after one month shipped within 24 hours of collection at baseline
State of Newborn Screening in the U.S.

- The state of newborn screening is strong
- We are facing a dynamic future
- The one constant, now and in the future, is the baby
Acknowledgments

• U.S. NBS Programs
• NBS Community
• NewSTEPs Committees

Funding Source
  – HRSA (CoAg# U22MC24078)