Newborn Screening: Current Status of State Newborn Screening Programs

Newborn Screen Positive Infant ACTion Project
Learning Session 2
February 12, 2011

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University of Texas Health Science Center at San Antonio
Newborn Screening and Genetics Resource Center
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I have no relevant financial relationships with the manufacturers of any commercial products and/or provider of commercial services discussed in this CME activity. I do not intend to discuss an unapproved/investigative use of a commercial product/device in this presentation.
Newborn Screening is MORE than a blood test!
Newborn Screening is MORE than a hearing test!
Newborn Screening is a SYSTEM!
Newborn Screening Education for Parents

Important Information for Parents about the Newborn Screening Test

More Information About Supplemental Newborn Screening

Newborn Screening Tests & Your Baby

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California Department of Health Services
Newborn Screening Program
Genetics, Cleft Lip Branch
2001 Berkshire Way, Annex 4
Berkeley, CA 94704
CLSI Newborn Screening Collection
The Newborn Screening Follow-up Process (CLSI I/LA 27-A)

- **Report "In Range"**
  - **Screening Test Result**
    - "In Range" (Including No Test)
    - "Invalid"
      - Follow-up for New Screening
        - Lost
  - "Out of Range"
    - Carrier
      - Facilitate Counseling
    - Results not Definitive
      - Facilitate Confirmatory Testing
        - Not Affected
          - Close Case
        - Confirmatory Testing
          - Is Not Definitive
            - Monitor Clinical Observation
              - Lost
            - Definitive Diagnosis
              - • Facilitate Intervention/Management
                • Facilitate Appropriate Services
                • Maintain in Case Registry
              - • Periodically Monitor Care Coordination
                • Periodically Monitor Functional Outcomes
                • Evaluate Long-term Data
                - Definitive Diagnosis
  - **Results Appear Definitive**
    - Facilitate Confirmatory Testing
      - Not Affected
      - Close Case
      - Lost

**Key**
- Action or Outcome
- Lost to Follow-up Possible
- Process End Point
Newborn Screening: The Role of the Obstetrician

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The history of newborn screening is relatively short, spanning only 40 years since the pioneering work by Guthrie. It was he who first realized that a blood sample could be taken from a newborn, absorbed and dried onto standardized filter paper, transported to a central testing laboratory, and then analyzed for biochemical indicators of inborn disorders of metabolism such as phenylketonuria (PKU). Because inborn metabolic errors are relatively rare (PKU in U.S. populations was shown to be about 1:15,000 at the time), it took the efforts of parents lobbying in behalf of the health of their newborns to convince health policy makers of the value of this type of population screening. In 1965, the American Academy of Pediatrics Committee on the Fetus and Newborn finally recommended a newborn screening blood test for PKU for all newborns "no sooner than 24 hours after onset of milk feeding and previous to discharge." Within a few years, most states in the United States and many other countries in the industrialized world were performing newborn screening for these "rare" disorders. As screening developed, it was inevitable that automated testing and data handling systems would evolve, and that new procedures would improve disease detectability in newborns. Indeed, by the end of the 1970s, automated sample preparation and improved testing sensitivity and specificity had led to expansion of many screening programs to include dried blood spot screening for congenital hypothyroidism (worldwide incidence of about 1:3,500, except in iodine-deficient areas, where it is much more prevalent). Today newborn screening is perhaps the best example of a successful and ongoing preventive public health population-screening program. Definitions of newborn screening have traditionally been limited to biochemical

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Quality Improvement Reviews
(Since 1987)

Limited Review (8 incl PA, AL, KS)
1 Review (27)
2 Reviews (7)
Average Number of Newborn Screening Conditions Required in US Programs 1990-2010
U.S. Newborn Screening
Conditions Required – Feb 1, 2011
(Conditions available as an option to a selected population are not counted – Must be universally required)
Laboratory Service Delivery Models
States Using Contract Screening Laboratories (Public)
Laboratory Service Delivery Models
States Using Contract Screening Laboratories
(Commercial/Non-profit)
Laboratory Service Delivery Models
States Using Contract Screening Laboratories
(Public and/or Commercial/Non-profit)
U.S. Newborn Screening Fees – 2011
(Ascending Amount with Number of Mandated Disorders Overlayed and Normalized)
U.S. Newborn Screening Fees – 2011
(Ascending Amount with Number of Mandated Disorders Overlayed and Normalized)
U.S. Newborn Screening

Conditions Not On SACHDNC List

Toxoplasmosis (MA, NH)

G6PD [DC; PA (part)]

HIV and Krabbe (NY)

LSDs – Pompe, Gaucher, Fabry (pilot)

Note: Additional MS/MS disorders also included in CA, CT, IL, ME, MD, MA, MS, NB, ND, PA, SD, TN
~ 54% Newborn Pop. Stored for ≥18 yrs.

~ 46% Newborn Pop. Stored for ≤3 yrs.
Arkansas

- Births (2008) 39,502
- Medicaid Births (2003) 51.7%
- Screens (2008) 39,826
- Screens (2009) 41,069
- No. Screens Required 1
- No. Disorders Required 30
- Fee $89.25
- Residual Storage 3 - 6 mo. (freezer space)
  -20 °C
Colorado

- Births (2008): 70,527
- Medicaid Births (2003): 37.3%
- Screens (2008): 134,702
- Screens (2009): 131,921
- No. Screens Required: 2 (for 4 only)
- No. Disorders Required: 45
- Fee: $85.00
- Residual Storage: 6 mo.
  Room temp.
Kentucky

- Births (2008) 56,621
- Medicaid Births (2003) 43.7%
- Screens (2008) 60,408
- Screens (2009) 60,136
- No. Screens Required 1
- No. Disorders Required 31
- Fee $53.50
- Residual Storage 6 mo.
  - 2 - 8 °C
Missouri

- Births (2008): 81,992
- Medicaid Births (2003): 45.4%
- Screens (2008): 90,713
- Screens (2009): 89,230
- No. Screens Required: 1
- No. Disorders Required: 52
- Fee: $65.00
- Residual Storage: 1 mo (zip lock bags) -30°C (5 yr. July 1, 2011)
- Births (2008): 252,360
- Medicaid Births (2003): 40.5%
- Screens (2008): 277,449
- Screens (2009): 273,915
- No. Screens Required: 1
- No. Disorders Required: 54
- Fee: no fee
- Residual Storage: 27 yr.
  4 °C
North Carolina

- Births (2008): 132,106
- Medicaid Births (2003): 47.9%
- Screens (2008): 130,703
- Screens (2009): no report
- No. Screens Required: 1
- No. Disorders Required: 42 (SCID Rec.)
- Fee: $19.00
- Issues: testing unsat specimens, fee for repeat to physician
- Residual Storage: 5 yr.
  Room temperature
Births (2008): 149,346
Medicaid Births (2003): 32.1%
Screens (2008): 151,583
Screens (2009): no report
No. Screens Required: 1
No. Disorders Required: 41 (SCID under review – will take about a year if adv. approved this month)
Fee: $55.16
Residual Storage: 2 yr.
Room Temperature
Pennsylvania

- Births (2008) 148,460
- Medicaid Births (2003) 31.0%
- Screens (2008) 148,460
- Screens (2009) 148,474
- No. Screens Required 1
- No. Disorders Required 33 (6) (DBS issue)
- Fee no fee (discussions concerning possible legislative change; OZ system)
- Residual Storage 8 mo. (w/ desiccant) -20°C
Texas

- Births (2008): 412,127
- Medicaid Births (2001*): 47.6%
- Screens (2008): 795,974
- Screens (2009): 789,467
- No. Screens Required: 2
- No. Disorders Required: 51 (7 not impl; 14 likely to be detected)
- Fee: $34.50
- Residual Storage: 25 yr. (under review)
  
  No information
Utah

- Births (2008) 56,787
- Medicaid Births (2003) 30.2%
- Screens (2008) 111,915
- Screens (2009) 108,870
- No. Screens Required 2
- No. Disorders Required 47
- Fee $93.00
- Residual Storage 2 yr. (7 day room temp)
  -20 °C
Virginia

- Births (2008): 104,990
- Medicaid Births (2003): 27.6%
- Screens (2008): 113,922
- No. Screens Required: 1
- No. Disorders Required: 30
- Fee: $53.00
- Residual Storage: 6 mo (positives 10 yr.) room temperature
## Summary – Basic Program Information

<table>
<thead>
<tr>
<th>State</th>
<th>Births</th>
<th>Medicaid Births</th>
<th>Screens 2008</th>
<th>Screens 2009</th>
<th>Screens</th>
<th>Tests</th>
<th>Fee</th>
<th>Storage</th>
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<tbody>
<tr>
<td>AR</td>
<td>39,503</td>
<td>51.7%</td>
<td>39,826</td>
<td>41,069</td>
<td>1</td>
<td>30</td>
<td>$89.25</td>
<td>3-6 mo</td>
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<tr>
<td>CO</td>
<td>70,527</td>
<td>37.3%</td>
<td>134,702</td>
<td>131,921</td>
<td>2</td>
<td>45</td>
<td>(2) $85.00</td>
<td>6 mo</td>
</tr>
<tr>
<td>KY</td>
<td>56,621</td>
<td>43.7%</td>
<td>60,408</td>
<td>60,136</td>
<td>1</td>
<td>31</td>
<td>$53.50</td>
<td>6 mo</td>
</tr>
<tr>
<td>MO</td>
<td>81,992</td>
<td>45.4%</td>
<td>90,713</td>
<td>89,230</td>
<td>1</td>
<td>52</td>
<td>$65.00</td>
<td>1 mo</td>
</tr>
<tr>
<td>NY</td>
<td>252,360</td>
<td>40.5%</td>
<td>277,449</td>
<td>273,915</td>
<td>1</td>
<td>54</td>
<td>None</td>
<td>27 yr</td>
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<tr>
<td>NC</td>
<td>132,106</td>
<td>47.9%</td>
<td>130,703</td>
<td>No report</td>
<td>1</td>
<td>42</td>
<td>$19.00</td>
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<td>OH</td>
<td>149,346</td>
<td>32.1%</td>
<td>151,583</td>
<td>No report</td>
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<td>41</td>
<td>$55.16</td>
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<tr>
<td>PA</td>
<td>148,460</td>
<td>31.0%</td>
<td>148,474</td>
<td>145,367</td>
<td>1</td>
<td>33</td>
<td>None</td>
<td>8 mo</td>
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<tr>
<td>TX</td>
<td>412,127</td>
<td>*47.6%</td>
<td>795,974</td>
<td>789,467</td>
<td>2</td>
<td>*51</td>
<td>(1) $34.50</td>
<td>25 yr</td>
</tr>
<tr>
<td>UT</td>
<td>56,787</td>
<td>32.2%</td>
<td>111,915</td>
<td>108,870</td>
<td>2</td>
<td>47</td>
<td>(2) $75.00</td>
<td>2 yr</td>
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<tr>
<td>VA</td>
<td>104,990</td>
<td>27.6%</td>
<td>113,922</td>
<td>108,656</td>
<td>1</td>
<td>30</td>
<td>$53.00</td>
<td>6 mo</td>
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</table>

*Note: The table includes the number of births, Medicaid births, screens in 2008 and 2009, screens, tests, fees, and storage periods for each state.*
## Summary – Case Finding Information (2009)

<table>
<thead>
<tr>
<th>State</th>
<th>CAH</th>
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<th>CH</th>
<th>BIO</th>
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<td>1</td>
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<td>NY</td>
<td>11*</td>
<td>6*</td>
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<td>13*</td>
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<tr>
<td>NC</td>
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<td>2</td>
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<td>4</td>
</tr>
<tr>
<td>TX</td>
<td>23</td>
<td>3</td>
<td>11</td>
<td>4</td>
</tr>
<tr>
<td>UT</td>
<td>3</td>
<td>1</td>
<td>0</td>
<td>1</td>
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<tr>
<td>VA</td>
<td>5</td>
<td>2</td>
<td>0</td>
<td>2</td>
</tr>
</tbody>
</table>

* Total, Not divided by type
## Summary – Case Finding Information (2009)

<table>
<thead>
<tr>
<th>State</th>
<th>Hgb</th>
<th>PKU</th>
<th>CF</th>
<th>MS/MS Groupings</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>All</td>
<td>S,S</td>
<td>Classical</td>
<td>Variant</td>
</tr>
<tr>
<td>AR</td>
<td>22</td>
<td>11</td>
<td>1</td>
<td>0</td>
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<tr>
<td>CO</td>
<td>14</td>
<td>9</td>
<td>2</td>
<td>0</td>
</tr>
<tr>
<td>KY</td>
<td>0</td>
<td></td>
<td>4</td>
<td>0</td>
</tr>
<tr>
<td>MO</td>
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<tr>
<td>VA</td>
<td>80</td>
<td>46</td>
<td>5</td>
<td>12</td>
</tr>
</tbody>
</table>

* Incomplete year
Welcome to the National Newborn Screening Information System database

The database is hosted by the National Newborn Screening and Genetics Resource Center (NNSGRC) and is designed to provide a secure, Internet-based, real-time, information collection and reporting system for capturing state and territorial newborn screening information. The system uses existing reporting requirements specified in the former National Newborn Screening Annual Report.

States Where Reported % Unsat > 1 for 2010

Days Since Last Update
(have we heard from you lately?)

Graph showing days since last update for different states.
Current Issues

- 12 hr. vs. 24 hr. vs. 48 hr. for unsatisfactory specimens
- Required single screen vs. required two screens
- Financing – fees, Medicaid
- Best protocol for CF screening – IRT/DNA vs. IRT/IRT (carrier detection issues)
- Whether to mandate all conditions on the ACMG panel (detection and liability issues)
- Long-term follow-up responsibility
- Whether to universally mandate hearing screening
- National data reporting
Thank You for Your Attention!
Thank You!!

http://genes-r-us.uthscsa.edu
http://www2.uthscsa.edu/nnsis/
http://www.marchofdimes.com/peristats/