Expanding Newborn Screening: The Minnesota Model

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Organization and Guidance

- MN Statutes 2003, Chapter 144.125-128 and MN Rule 4615.0300 – 4615.0760
- Mandated newborn screening advisory committee
- Health Commissioner oversight
- Guidance from WHO, AAP, APHL, NNSGRC and ACMG
**Minnesota Timeline**

<table>
<thead>
<tr>
<th>Year</th>
<th>Disorder</th>
<th>Incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>1965</td>
<td>Phenylketonuria</td>
<td>1:13,500 (~5 per year)</td>
</tr>
<tr>
<td>1967</td>
<td>Congenital Hypothyroidism</td>
<td>1:2,500 (~23 per year)</td>
</tr>
<tr>
<td>1974</td>
<td>Galactosemia</td>
<td>1:60,000 (~1 per year)</td>
</tr>
<tr>
<td>1988</td>
<td>Sickle Cell Anemia (+ others)</td>
<td>1:3,800 (~17 per year)</td>
</tr>
<tr>
<td>1993</td>
<td>Congenital Adrenal Hyperplasia</td>
<td>1:17,000 (~4 per year)</td>
</tr>
</tbody>
</table>

>1,300 newborns diagnosed in MN
2001: A Year of Changes

Expansion begins

Congenital Hearing Loss
- Voluntary screening
- 108 / 111 birthing centers
- 98% infants screened

Tandem Mass Spectrometry (MS/MS)
- 204,000 infants screened
- 90 confirmed positives
- Aggregate incidence ~ 1:2,300
Clinical Impact of Expanded Screening Using MS/MS

<table>
<thead>
<tr>
<th>Year</th>
<th>2001</th>
<th>2002</th>
<th>2003</th>
</tr>
</thead>
<tbody>
<tr>
<td>Screened</td>
<td>39,974*</td>
<td>69,821</td>
<td>71,147</td>
</tr>
<tr>
<td>Abnormal</td>
<td>247</td>
<td>277</td>
<td>172</td>
</tr>
<tr>
<td>True Positives</td>
<td>11</td>
<td>28</td>
<td>46</td>
</tr>
<tr>
<td>False Positives</td>
<td>236</td>
<td>249</td>
<td>126</td>
</tr>
</tbody>
</table>

* 7 months 1:3,634 1:2,493 1:1,547
Challenges

- Minimal funding
- Sustainability of expanded screening
- Limited outcome tracking and follow-up
- Limited support for primary care, infant and family
- LACK OF COMMUNICATION

HOW DO WE FIX THIS?
The Minnesota Model
What is the Minnesota Model?

A multi-component newborn screening program based on COORDINATION and COMMUNICATION between all parties involved with the goal to expand and enhance the delivery of patient care, laboratory tests, and family services.
Coordination & Communication
(The Minnesota Model)

• Notification of abnormal results
• Verification
• Reporting
• Referral of true positives
• Follow-up
Notification
• Phone
• Fax
• E-mail

Screening Laboratory

Biochemical Genetics Laboratory

Primary Care Provider

Metabolic Clinic
Verification of Abnormal Results

Screening Laboratory

Biochemical Genetics Laboratory

Primary Care Provider

Metabolic Clinic

1. Discourage repeat submission and testing of blood spots.
2. Prevent "blind" referrals to specialists of false positives.
3. Activate rapid confirmatory testing.

Activate rapid confirmatory testing.
Reporting of Diagnostic Testing

Screening Laboratory

Biochemical Genetics Laboratory

Primary Care Provider

Metabolic Clinic
Referral of TRUE Positives

Screening Laboratory

Biochemical Genetics Laboratory

Primary Care Provider

Metabolic Clinic

Confirmatory testing is already DONE
SHORT Term Follow-Up

Screening Laboratory

Biochemical Genetics Laboratory

Primary Care Provider

Metabolic Clinic

BENEFITS
Accurate statistics
Monitoring of trends
Learning from experience
Education
<table>
<thead>
<tr>
<th>Established Conditions</th>
<th>Screening</th>
<th>Confirmatory</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cong. Hypothyroidism</td>
<td>S,FU</td>
<td>-</td>
</tr>
<tr>
<td>Galactosemia (GALT)</td>
<td>S,FU</td>
<td>-</td>
</tr>
<tr>
<td>Sickle Cell Anemia</td>
<td>S,FU</td>
<td>-</td>
</tr>
<tr>
<td>21-Hydroxylase Def (CAH)</td>
<td>S,FU</td>
<td>2T</td>
</tr>
<tr>
<td>MS/MS (AA, OA, FAO)</td>
<td>FU</td>
<td>S,C</td>
</tr>
</tbody>
</table>

- **S**: Screening
- **FU**: Follow-up
- **FU**: Follow-up
- **2T**: 2nd tier test in DBS
Joint Newborn Screening Program in Minnesota (2004-2005)

Future Additions

<table>
<thead>
<tr>
<th>Condition</th>
<th>Development</th>
<th>Pilot Study</th>
<th>Screening</th>
<th>Follow-up</th>
<th>Confirmatory</th>
</tr>
</thead>
<tbody>
<tr>
<td>Biotinidase Deficiency</td>
<td>D,S,FU</td>
<td>C</td>
<td>?</td>
<td>?</td>
<td></td>
</tr>
<tr>
<td>G6PD Deficiency</td>
<td>D,S,FU</td>
<td>-</td>
<td>?</td>
<td>?</td>
<td></td>
</tr>
<tr>
<td>Wilson Disease</td>
<td>S,FU</td>
<td>D,P</td>
<td>?</td>
<td>?</td>
<td></td>
</tr>
<tr>
<td>Others</td>
<td>?</td>
<td>?</td>
<td>?</td>
<td>?</td>
<td></td>
</tr>
</tbody>
</table>

D     Development  S     Screening  C     Confirmatory
P     Pilot Study  FU     Follow-up
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