Feasibility of a Population Based Newborn Screening Study for Spinal Muscular Atrophy in Colorado

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Spinal Muscular Atrophy (SMA)

- Autosomal recessive disorder
- Significant motor disability
- Respiratory and nutritional compromise
- About 1 in 10,000 babies have SMA
SPOT SMA Study

- Nominated for consideration on RUSP in 2008
  - SACHDNC Recommended Pilot Study
  - Pilot initiated by Kathy Swoboda, MD, principal investigator
- First of its kind DNA-based (melting point technology) population-based newborn screening pilot study
- Study sites:
  - Utah, Colorado, recently added Chicago
  - Each Site has their own recruitment model
SPOT SMA in CO: Opt-Out Model

- Rationale for opt-out
- Logistics
  - Parents have 2 weeks from baby’s date of birth
  - Can opt-out either in person, via phone, or online.
SPOT SMA in CO: Parent Education

- Brochures dispersed in post-partum
- Some hospitals disperse prenatally

Study Website
- [http://www.spotsmaco.org/](http://www.spotsmaco.org/)
SPOT SMA in CO: SMA Screening Card

- 1 card for every baby
- Shipped or picked up once per week
- Stored for 2 weeks (until opt-out period expires), then shipped or destroyed
CDPHE Abstains from Project

- Colorado Department of Public Health and Environment

Obstacles:
1. Education and timing for opting out
2. Notification of positive results to families
3. Releasing the screening results – HIPAA issues
4. Non-CDPHE staff viewing PHI in the lab
5. LAB FTE
Actively Screening at 9 Hospitals

- **UCH** = University of Colorado Hospital
- **MH** = Memorial Health
- **SRMC** = Sky Ridge Medical Center
- **DH** = Denver Health
- **SJH** = Saint Joseph Hospital
- **TMCA** = The Medical Center of Aurora
- **CHCO** = Children’s Hospital of Colorado
- **PVH** = Poudre Valley Hospital
- **MCR** = Medical Center of the Rockies

18,156 SMA Screening Cards Collected
Screening Results as of 9/30/14

18,156 Collected

- 83 Pending Opt-Out Period
- 738 Pending Results
- 43 (0.24%) Opt-Outs

16,736 (100%) Tested Negative
Mid-Study Challenge 1: Low Enrollment

- Hindered by having to enroll each hospital one by one
- Establishing initial communication with hospitals
- Hospital System IRB approvals
- Individual Hospital Research/Contract Dept.
Possible Solutions: Low Enrollment

- Work with state newborn screening program
- Have all IRB systems cede to central IRB
- Find contacts at each hospital that have time and buy-in
- Have contracts/research departments present at initial meetings
Mid-Study Challenge 2: No Positives

- Incidence = 1 in 10,000
- Low enrollment numbers
- NICU Screening
  - Timing of NICU admission
Possible Solutions: No Positives

- Higher population density states for rare genetic disorders in combination with...
- States whose health departments are willing to participate
- Need to ensure all NICU admissions are screened
Challenge 3: Patient and Staff Education

- Low opt-out rate (.24%)
- Opt-out model not conducive to patient education
- Brochures in post-partum and website may not be enough
- More education by nurses?
- Lack of study staff presence at hospitals
SMA cards collected/opt-outs do not match number of births

<table>
<thead>
<tr>
<th>Facility</th>
<th>Total Live Births During Active Screening Period</th>
<th>Total Screening Cards Collected</th>
<th>Shortage in Screening Cards Collected</th>
<th>Percent of Shortage</th>
</tr>
</thead>
<tbody>
<tr>
<td>HOSPITAL 1</td>
<td>4955</td>
<td>4303</td>
<td>652</td>
<td>13.16%</td>
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<tr>
<td>HOSPITAL 2</td>
<td>4037</td>
<td>3415</td>
<td>622</td>
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<td>3674</td>
<td>53</td>
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<td>895</td>
<td>143</td>
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<td>HOSPITAL 6</td>
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<td>2033</td>
<td>267</td>
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<tr>
<td>HOSPITAL 7</td>
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<td>Just Started Screening</td>
<td>Just Started Screening</td>
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<td>HOSPITAL 8</td>
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<td>Just Started Screening</td>
<td>Just Started Screening</td>
<td>Just Started Screening</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td>19424</td>
<td>17294</td>
<td>2130</td>
<td><strong>10.97%</strong></td>
</tr>
</tbody>
</table>

True opt-out rate lies in the range of 0.24% - 10.97%
Babies born on the weekend are less likely to have an SMA blood spot
Possible Solutions: Patient and Staff Education

- Speak with each hospital and assess needs/problems
- Reeducation of nurse/lab staff
- Continued Education of nurse/lab staff
- Educational videos played in post-partum
- More face-to-face education
- Statewide implementation would allow education through additional mechanisms (hospitals, pediatricians, OB/GYNs)
After Speaking with our Gold Standard Site

- Nurses claim this work on Professional Clinical Advancement Program (ProCAP)
- Healthcare tech ("CHAMPION") does 80% of draws
- Clerical staff attaches SMA card to State card at admission
- Continuing education is important
  - Study staff should have a physical presence
  - Reinforce the importance of what they are doing
- Good communication with a central coordinator
Conclusion

- Pilot studies are needed in order to understand population based NBS for new disorders
- Public health departments may be wary of research studies
- Challenges of hospital based research may be hard to overcome
- Opt-out model may be feasible with appropriate education
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