

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.

OPT-OUT	<input checked="" type="checkbox"/>
OPT-IN	<input type="checkbox"/>

SPOT SMA in CO: *Parent Education*

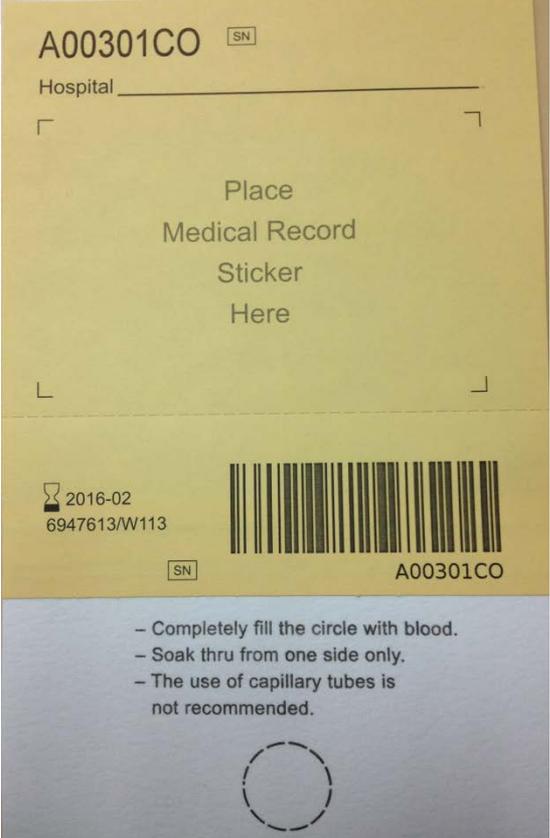
- Brochures dispersed in post-partum
- Some hospitals disperse prenatally
- Study Website
 - <http://www.spotsmaco.org/>

Important Information
for Parents about the
"SPOT SMA Study"



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



A00301CO SN

Hospital _____

Place
Medical Record
Sticker
Here

2016-02
6947613/W113

SN

A00301CO

- Completely fill the circle with blood.
- Soak thru from one side only.
- The use of capillary tubes is not recommended.



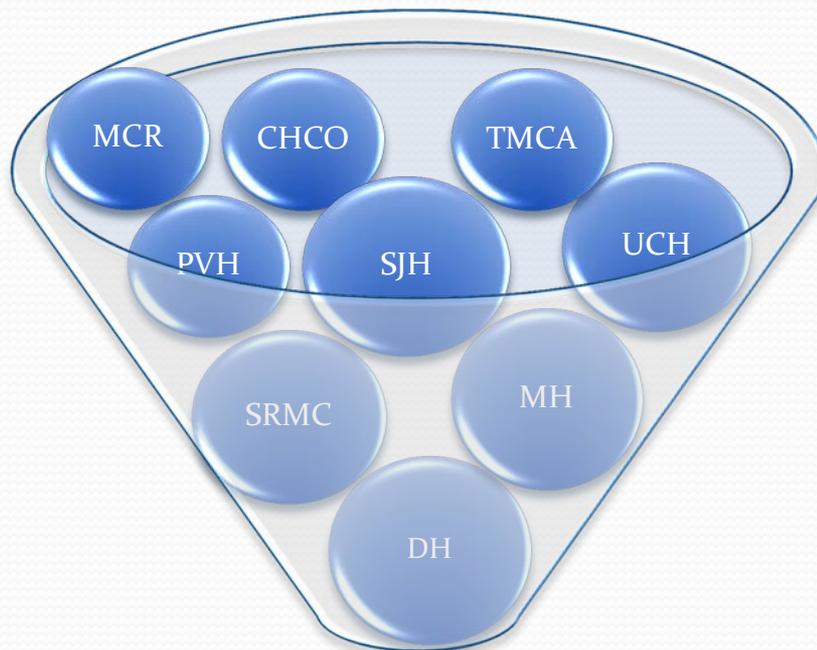
CDPHE Abstains from Project

- 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



COLORADO
Department of Public
Health & Environment

Actively Screening at 9 Hospitals

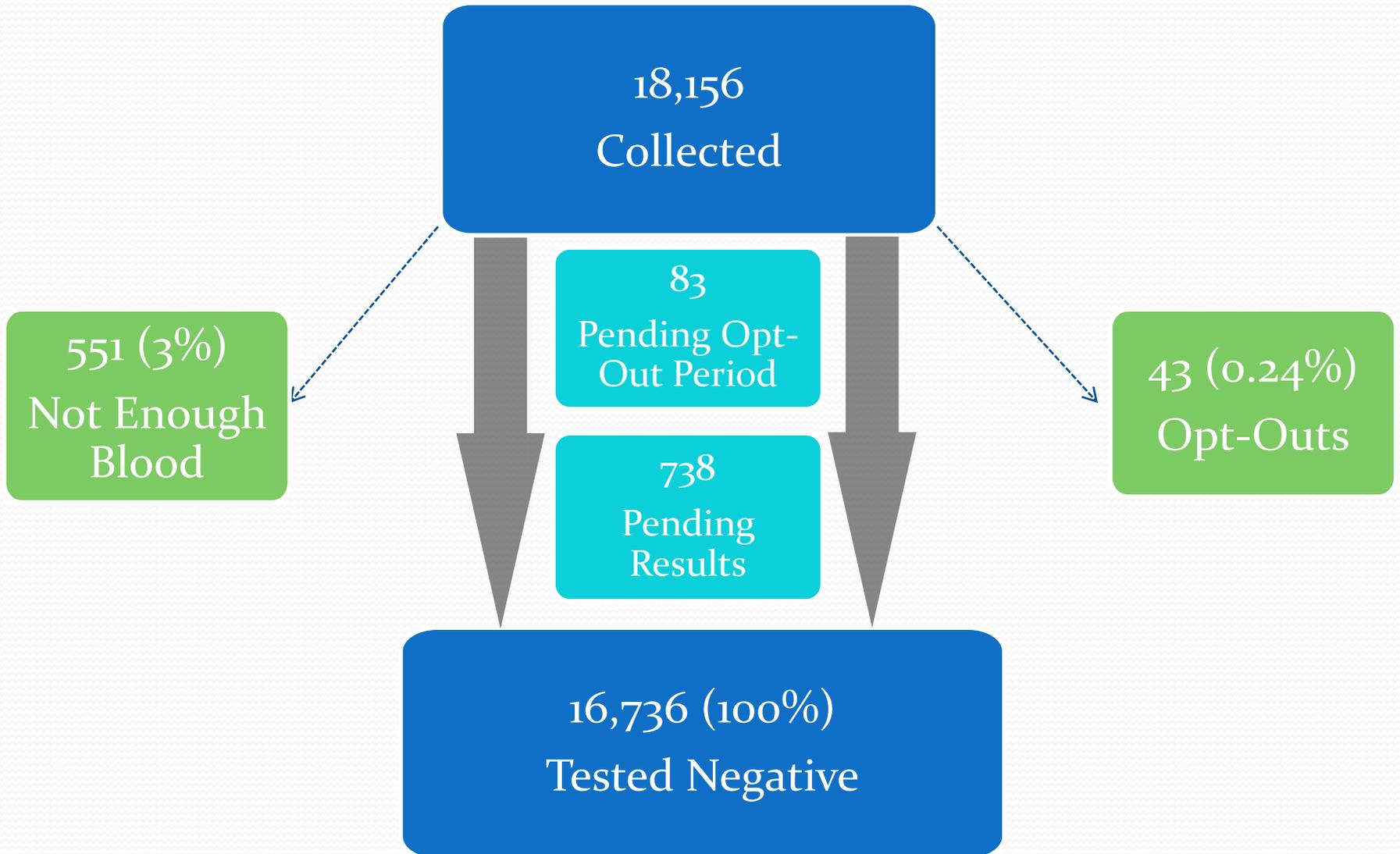


18,156

SMA Screening
Cards Collected

- **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

Screening Results as of 9/30/14



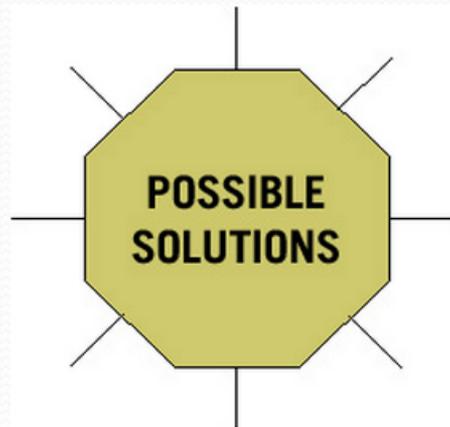
Mid-Study Challenge 1: Low Enrollment

- 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



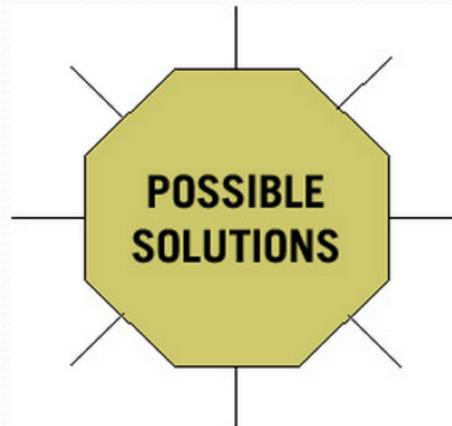
Mid-Study Challenge 2: No Positives

- 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

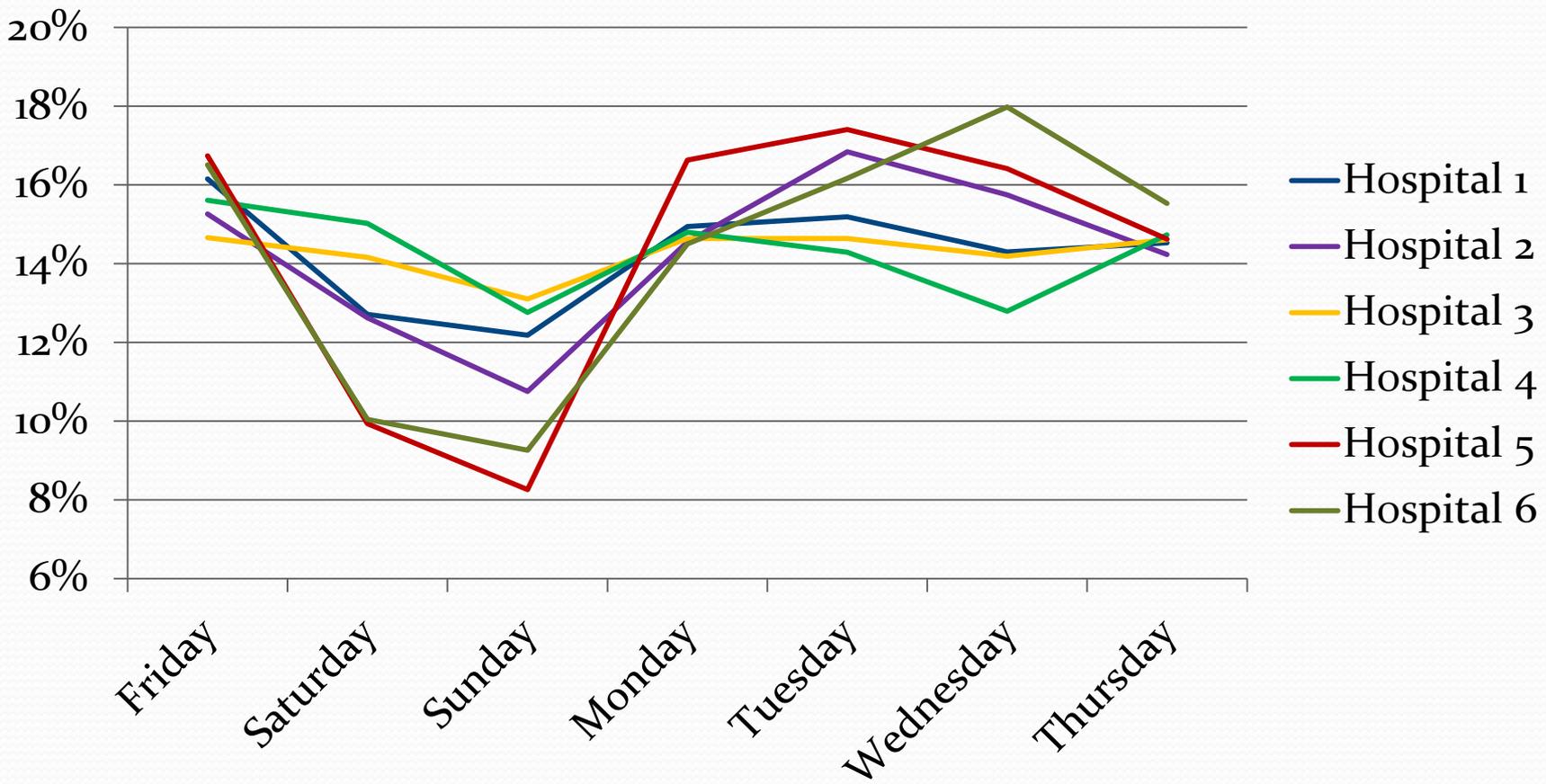


SMA cards collected/opt-outs do not match number of births

Facility	Total Live Births During Active Screening Period	Total Screening Cards Collected	Shortage in Screening Cards Collected	Percent of Shortage
HOSPITAL 1	4955	4303	652	13.16%
HOSPITAL 2	4037	3415	622	15.41%
HOSPITAL 3	3727	3674	53	1.42%
HOSPITAL 4	3367	2974	393	11.67%
HOSPITAL 5	1038	895	143	13.78%
HOSPITAL 6	2300	2033	267	11.61%
HOSPITAL 7	Just Started Screening	Just Started Screening	Just Started Screening	Just Started Screening
HOSPITAL 8	Just Started Screening	Just Started Screening	Just Started Screening	Just Started Screening
Total	19424	17294	2130	10.97%

True opt-out rate lies in the range of 0.24% - 10.97%

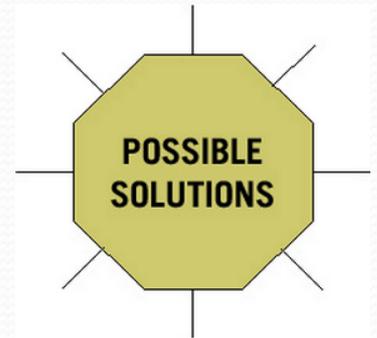
Babies born on the weekend are less likely to have an SMA blood spot



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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