



## Harmonization of NBS Efforts in the Heartland

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Update from 2009 Spring NBS  
Workshop held in Lincoln, NE

Julie Miller, Program Manager  
Nebraska Newborn Screening & Genetics



## Newborn Screening Workshop

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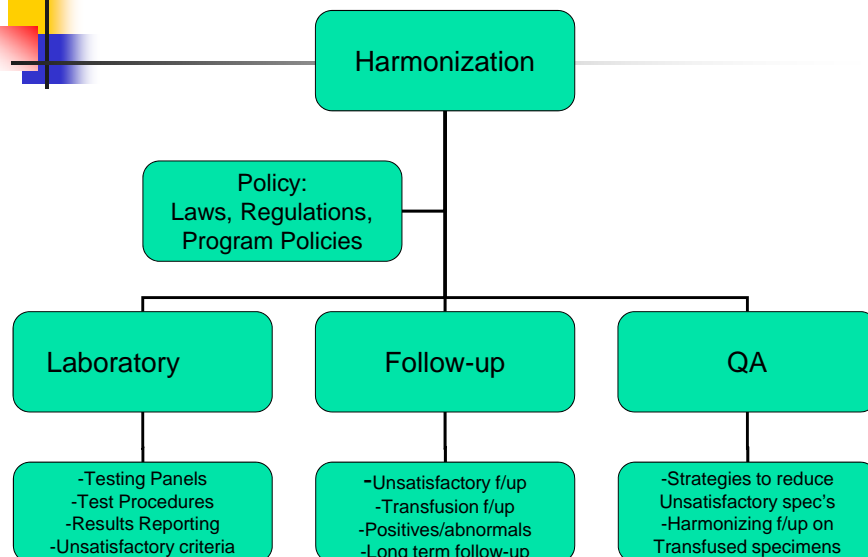
- April 30, May 1, 2009 in Lincoln
  
- Attended by all 8 States: 3 Arkansas, 6 Iowa, 4 Kansas, 4 Missouri, 3 Nebraska + 3 contracted lab/corporation, 1 North Dakota, 2 Oklahoma, and 1 South Dakota
  - from Newborn Screening laboratories and follow-up as well as advocates and program management.
  
- Heartland staff support: Shona Whitehead

## Meeting theme

### “Harmonization” also included:

- Guest speakers
  - John Colombo, M.D. Pediatric Pulmonologist, and Director of Nebraska’s Accredited Cystic Fibrosis Center
    - Cystic Fibrosis, new challenges in diagnosis and treatment associated with NBS
  - Amy Brower, Ph.D.
    - Cystic Fibrosis, molecular technology update, what the technology does and doesn’t tell us
- State Updates on Newborn Screening systems, ACMG panel adoption, staffing, fees, lab and follow-up resources

## Heartland Inter-state HARMONIZATION of NBS Systems for Emergency Back-up





## Harmonization Activities

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- Iowa/Missouri Backup Testing Project
  - (Stan and Patrick will report on)
- Incorporating follow-up (Short & long term) into emergency preparedness
  - Intra-State and/or Inter-State
  - Prioritization of which activities must be done, which activities will be on hold (care coordination & tx. high priority, research data collection lower as less direct impact on families)



## Harmonization Activities (cont'd)

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- Emergency back-up systems (f/up)
  - Interstate harmonization benefit:
    - Filter paper data elements collected
    - Laboratory result reporting
      - /language/format/content
    - Procedures for f/up on transfused babies
    - Procedures for f/up on unsatisfactory specimens

# Filter paper data elements collected

(Ongoing project: Amy Brower, Patrick Hopkins, Julie Miller)

Data Element	Arkansas	Iowa	Kansas	Missouri	Nebraska	North Dakota	Oklahoma	South Dakota
infant's last name & first name	baby's	x	x	baby's	newborn's	x	x	x
middle name					x			
sex m/f	x	x	x	x	x	x	x	x
birth date & birth time	x	x	x	x	x	x	x	x
first screen	x	x	initial	initial form	x	x	x	x
repeat screen	x	x	x	q	x	x	x	x
previous lab #							x	
change in name for 2nd specimen	x							
infant in NICU		x	x			x		x
multiple birth	twin	y/n	yes only	x		y/n	x	y/n
birth order	a or b	1, 2, 3, etc	write in box	enter a-f, single -		1, 2, 3, etc	a-h	1, 2, 3, etc

# Laboratory result reporting /language/format/content

The screenshot shows a laboratory report from Hygienic Laboratory, The University of Iowa. The report is titled "Newborn Screening Report" and includes the following information:

- Patient Information:** Name: [Redacted], Sex: [Redacted], Birth Date: [Redacted], Birth Time: [Redacted], Birth Place: [Redacted], Birth Weight: [Redacted], Birth Length: [Redacted], Birth Head Circumference: [Redacted].
- Screening Results:**
  - Phenylalanine: [Redacted]
  - Galactose-1-epimerase: [Redacted]
  - Glucose-6-phosphate dehydrogenase: [Redacted]
  - Glucuronosyltransferase: [Redacted]
  - Hexosaminidase B: [Redacted]
  - Hexosaminidase A: [Redacted]
  - Hexosaminidase C: [Redacted]
  - Hexosaminidase D: [Redacted]
  - Hexosaminidase E: [Redacted]
  - Hexosaminidase F: [Redacted]
  - Hexosaminidase G: [Redacted]
  - Hexosaminidase H: [Redacted]
  - Hexosaminidase I: [Redacted]
  - Hexosaminidase J: [Redacted]
  - Hexosaminidase K: [Redacted]
  - Hexosaminidase L: [Redacted]
  - Hexosaminidase M: [Redacted]
  - Hexosaminidase N: [Redacted]
  - Hexosaminidase O: [Redacted]
  - Hexosaminidase P: [Redacted]
  - Hexosaminidase Q: [Redacted]
  - Hexosaminidase R: [Redacted]
  - Hexosaminidase S: [Redacted]
  - Hexosaminidase T: [Redacted]
  - Hexosaminidase U: [Redacted]
  - Hexosaminidase V: [Redacted]
  - Hexosaminidase W: [Redacted]
  - Hexosaminidase X: [Redacted]
  - Hexosaminidase Y: [Redacted]
  - Hexosaminidase Z: [Redacted]



## Procedures for Follow-up/Repeat testing on transfused babies

(Krystal Baumert & Karen Eveans leads)

- Wide variance in State practices
  - Leads have developed strategic plan to help move towards more harmonized practices.
- Each State represented on work group is currently conducting written survey/phone clarification
  - # transfusions,
  - # whole blood, packed RBC's, platelets, fresh frozen plasma,
  - Requirements (CMV negative, leuko depleted, irradiated, etc.)
  - Special circumstances: ECMO/ECLS, Exchange transfusions, Intra-uterine transfusions



## Procedures related to unsatisfactory specimens

(Two projects both with two project co-leads)

- Special considerations:
  - Laboratory criteria for determining which specimens are unsatisfactory
  - Regulatory agencies' impact on ability to harmonize (e.g. CLIA inspectors interpretation/application of guidelines differ per CLIA region)
  - Laboratory procedure for testing or not testing, reporting or not reporting
  - Follow-up procedure for repeat specimen collection & testing



## Unsatisfactory Specimens (cont'd)

- States' strategies for reducing unsatisfactory specimens
  - Distribution of CLSI Standards and training video on blood-spot collection
  - Web-site links/training
  - Newsletters
  - On-site QA visits
  - QA reports to hospitals
  - Impact of "rejection practices" vs. test part or all of the panel



## Unsatisfactory Specimens

Leads: Dennis Freer, PhD PerkinElmer Genetics; Marcia Valbracht, Iowa

- Innovative Proposed Project:
  - State replication/collaboration of PerkinElmer lab investigation into test results/values on specimens deemed unsatisfactory due to overspotting, heavily applied or layered blood spots.
  - Can algorithms be developed to safely screen these blood spots for some conditions
  - Representative from each state lab participating in workgroup & study



## Unsatisfactory Specimens

- Develop urgency/panic list of screen results that might be detected on an “unsatisfactory” specimen that should still be reported out immediately for urgent follow-up.
  - Work group of one representative from each state. (Lead:      )



## Recognition of Contributors

Annette Arnold (AK)	Julie Raburn-Miller (MO)
Michael Elliott (AK)	Sharmini Rogers (MO)
Leslie Himstedt (AK)	Barbara Schweitzer (ND)
Elizabeth McDonald (IA)	Krystal Baumert (NE)
Kim Turner (IA)	Karen Eveans (NE)
Stan Berberich (IA)	Julie Miller (NE)
Esther Blanchard (IA)	Tonya McCallister (OK)
Lauren Fridley (IA)	Sharon Vaz (OK)
Marcia Valbracht (IA)	Lucy Fossen (SD)
Jamey Kendall (KS)	Amy Brower Ph.D- molecular speaker
Colleen Peterson (KS)	John Colombo, MD- CF speaker
Laura Ross (KS)	Dennis Freer PhD (PerkinElmer)
Linda Williams (KS)	Janet Perkins (PerkinElmer)
Darla Eiken (MO)	William Slimak (Perkin Elmer)
Patrick Hopkins (MO)	Shona Whitehead (Heartland Staff)