Timeliness of Newborn Screening

January 17, 2014

Susan Tanksley, Ph.D. – Co-chair,
Laboratory Standards and Procedures
Subcommittee

Timeliness of Newborn Screening

- Newborn screening is a “simple” test used to identify many life-threatening illnesses before any symptoms begin.
- Based on public comment in Sept. 2013 DACHDNC Meeting, Laboratory Standards and Procedures Subcommittee was tasked with:
  - looking into issues related to the timely handling of samples
  - whether the Committee should make recommendations on this issue

Priorities for Lab Subcommittee

- **Priority A:** Review new enabling/disruptive technologies
  - SUAC Implementation Survey Update
- **Priority B:** Provide guidance for state NBS programs in making decisions about lab implementation, integration, follow-up (FU), and quality assurance (QA)
  - SCID Slide Deck – Update in May
  - Timeliness of newborn screening
- **Priority C:** Establish process for regular review and revision of the Recommended Uniform Screening Panel (RUSP)
  - No update at this time

Approach

- Gather background information
  - Work with CDC and APHL to collect data
    - APHL surveyed states
  - Review recommendations
    - ACMG 2005 report
    - CLSI guidelines
- Committee discussion
- Proposal for moving forward

Specimen Collection and Transport Within NBS Programs Survey: Preliminary Results

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Laboratory Sub-Committee
DACHDNC
January 16, 2014

APHL ASSOCIATION OF PUBLIC HEALTH LABORATORIES

Methods

- Created a web based survey (early December 2013)
- All state NBS laboratory directors, managers and follow-up coordinators were sent survey.
- Time frame for completing survey was from Dec. 19, 2013 – Jan. 6, 2014.
- 32 states responded to the survey to date.
  - Note: 6 other states are completing survey
- Quality assurance and data control checks were performed on data provided.

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Results

What does your state use to send specimens from birthing hospitals to the NBS laboratory? (n=32)

Service	Number of NBS Programs
Courier Services	18
Overnight Delivery Services	19
US Mail Services	20
Other	8

*Multiple responses are captured.

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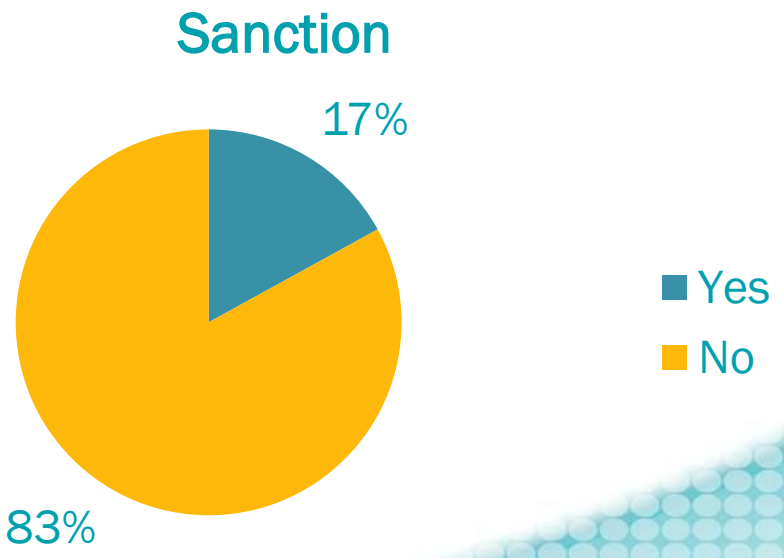
Results

Does your state have a recommended time period for when specimens should be received by the NBS programs from the hospitals? (n=31)

- 62% had a policy/practice/recommendation/law for when specimens are received.
- 19% had no policy/practice/recommendation/law.
- **19% indicated they had a policy/ practice/recommendation/law for when specimens should be *sent*.**

Results

Does your state have regulatory authority to fine or sanction hospitals that do not comply with state laws for sending samples in the state-specified time frame? (n=24)



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Does your NBS program keep a record of transit performance by hospital?

	Number of NBS Programs	Percentage of NBS Programs
Yes	31	97%
No	1	3%

Does your state NBS program review the transit performance times?

	Number of NBS Programs	Percentage of NBS Programs
Yes	29	91%
No	3	9%

Does your state NBS program provide feedback to birthing hospitals regarding transit times?

	Number of NBS Programs	Percentage of NBS Programs
Yes	30	94%
No	2	6%

Results

Type of feedback provided to hospitals regarding the timing of specimen receipt by NBS laboratory

- Report Cards
- Quality Improvement Tools
- Educational Materials
- Newsletters
- Feedback Meetings

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Laboratory Operating Hours

- 12 of 32 respondents (38%) indicated their labs are consistently closed on Saturdays and Sundays.
- 20 of 32 respondents(62%) reported their labs are open at least 6 days a week.
- 4 of 20 respondents only received specimens and do not do any other activity on Saturday or Sunday.
- 6 laboratories (of 12) are considering opening at least 6 days a week within the next 1-2 years.

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Follow-Up Operating Hours

- 17 of 28 (61%) are consistently closed on Saturdays and Sundays.
- 11 of 28 (39%) are open at least 6 days a week.
- 20 of 31 (65%) offer after hours paging/on call services on Saturdays or Sundays.

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Quality Indicators

QI 1. Percent of invalid dried blood spot specimens due to improper collection and/or transport

QI 2. Percent of dried blood spot specimens missing essential information

QI 3. Percent of eligible infants not receiving valid newborn screening test, stratified by dried blood spot or point of care test(s).

QI 4. Percent of loss to follow-up

QI 5. Time elapsed from birth to screening, follow-up testing, confirmed diagnosis

QI 6. Percent of out of range results

QI 7. Frequency of condition detected by newborn screening for each disorder

QI 8. Percent of missed cases (false negatives), stratified by disorder

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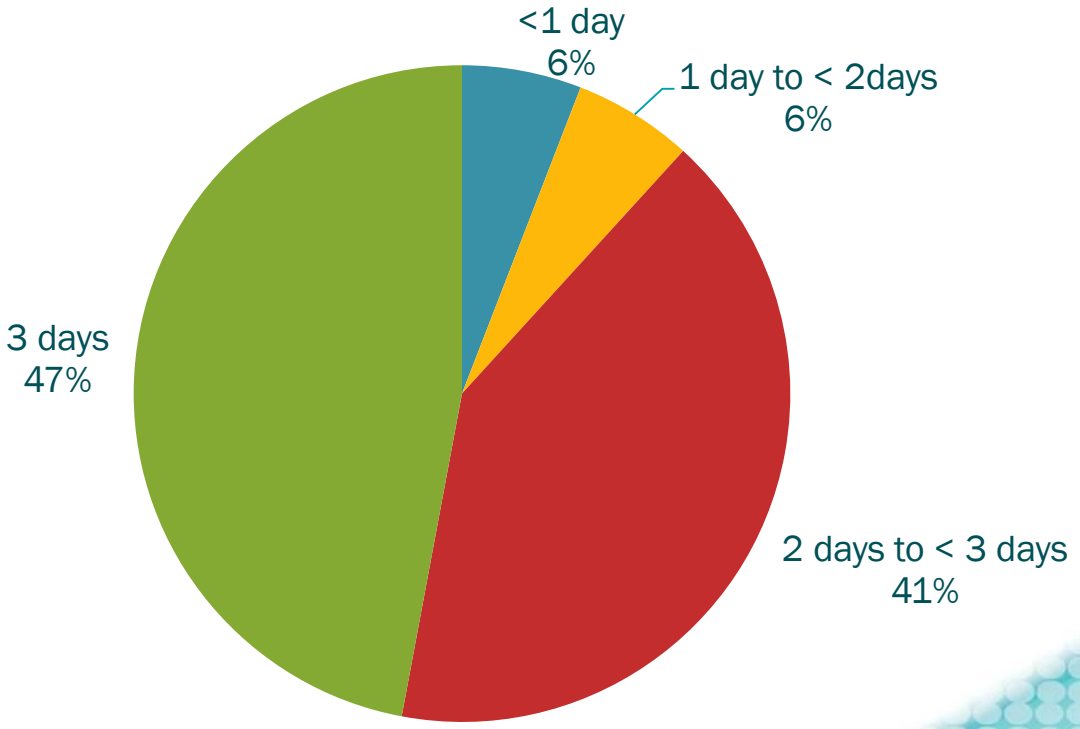
Results

All respondents noted that the time frame from birth to specimen collection was in the 24-48 hour time frame.

Results

What is the median time (in days) from SPECIMEN COLLECTION TO RECEIPT BY LAB for your state? (initial samples)(n=17)?



* Excluded states where median was not provided.

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Survey Limitations

- Time frame for response was short and during holidays.
- Some data, including time quality indicator data was incomplete.
- Some questions may be left to interpretation.
- Quality Indicator definitions could have been included for more clarity.

ACMG Standards for NBS and their Oversight

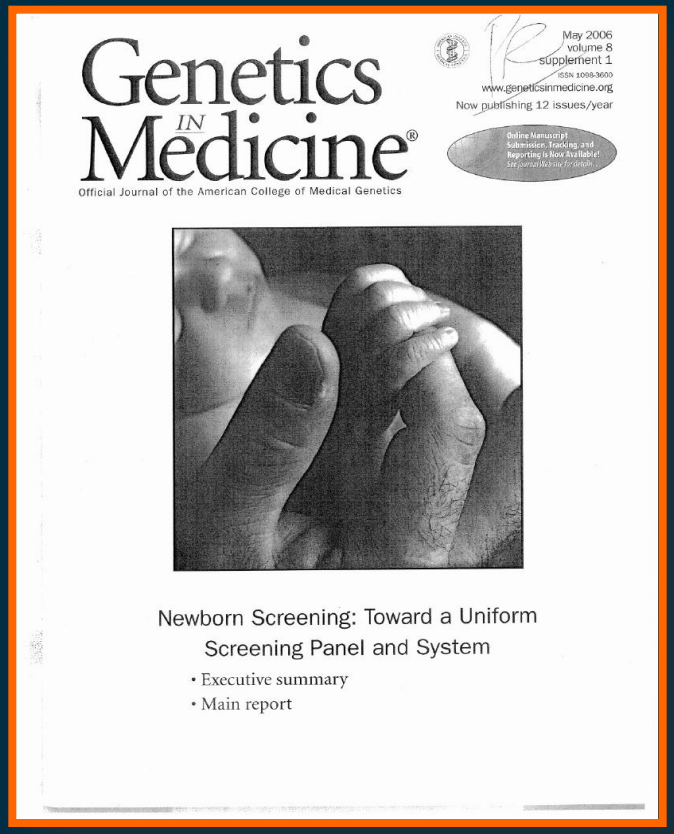
Michael S. Watson, MS, PhD, FACMG
ACMG

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

Standardization of Newborn Screening in the United States

- In 2001, Maternal and Child Health Bureau (HRSA) charged **American College of Medical Genetics**
  - To evaluate the scientific and medical information related to **screening for specific conditions** .
  - To make recommendations based on this evidence
- Expert group convened in December 2002
  - >70 physicians, scientists, consumers, **state laboratorians, lawyers, ethicists, and others**
  - Results reviewed by an independent newborn screening external review group
- Newborn Screening: Toward a Uniform Screening Panel and System (report published in 2006)

Screening: Towards a Uniform Screening Panel and System



- Section I – A Uniform NBS Panel
- Section II – The System
  - Program evaluation
  - Cost-effectiveness analysis
  - Information gaps and a research agenda
  - Future Needs

Program Standards 2

- Language and terminology should be standardized in order to better compare performance among programs.
- Turnaround time in reporting screen-negative results should be improved.
  - At a minimum, all results from the initial screening test (some states perform a second test later) should be available less than 5 days after the blood sampling for the first post-hospital discharge visit to be of use in this clinical visit and to facilitate awareness of lifelong screening. Most results should be available within two days of the specimen arriving in the laboratory, and specimens should arrive in the laboratories within three days of collection.

Program Standards 4

- Hospitals and JCAHO have significant roles to play, and standards need to be developed to improve quality, minimize errors, and facilitate tracking of newborns requiring active participation in testing follow-up.
- All newborn screening laboratories should be CLIA-certified and should participate in CDC and CAP/ACMG proficiency testing programs or other equivalent programs as applicable.
- All states should have an active system-wide newborn screening QA and total quality management program.

Joint Commission (Formerly Joint Commission for the Accreditation of Hospital Organizations – JCAHO)

- First contacted JCAHO in December 2003
- Conference call with JCAHO February 4, 2004
  - Major issues
    - Role of hospital in getting samples to screening labs
    - Whether the hospital has a role in tracking down patients for follow-up
    - Highlighted alignment of NBS standards with critical reporting priorities of JCAHO
    - Needed an analysis of established legal liabilities associated with NBS activities

Next Steps

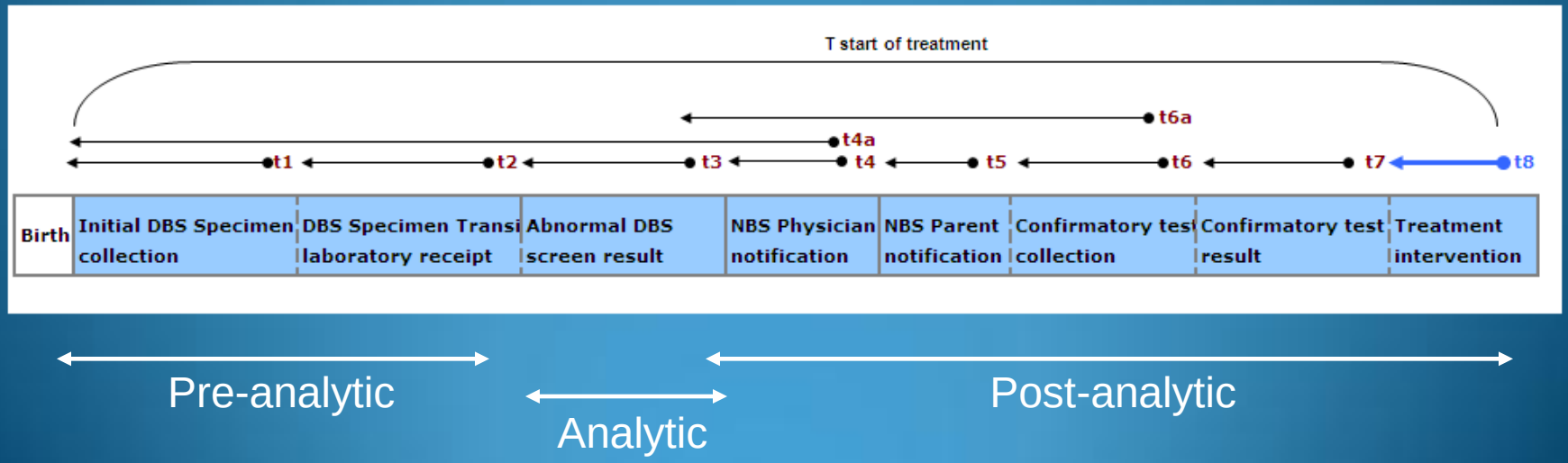
- States are in an awkward position in enforcing standards against the hospitals on which they rely for program delivery
  - Prefer to provide education and training to resolve problems
  - Transparency compromised by lack of public awareness of hospital performance
- Re-engage Joint Commission on development of standards for hospitals

Newborn Screening is a System

- Key Components of Newborn Screening
  - Education (throughout the process)
  - Screening, including specimen collection and testing
  - Follow-up and result reporting
  - Diagnostic confirmation
  - Management
  - Program evaluation and CQI

What are the issues & where are they occurring?

- Measuring each step allows you to pinpoint where a breakdown has occurred
  - Time of collection
  - Transit time (from collection to receipt in lab)
  - Time to result/report (from collection to screen results)
  - Time to treatment (from birth to treatment)

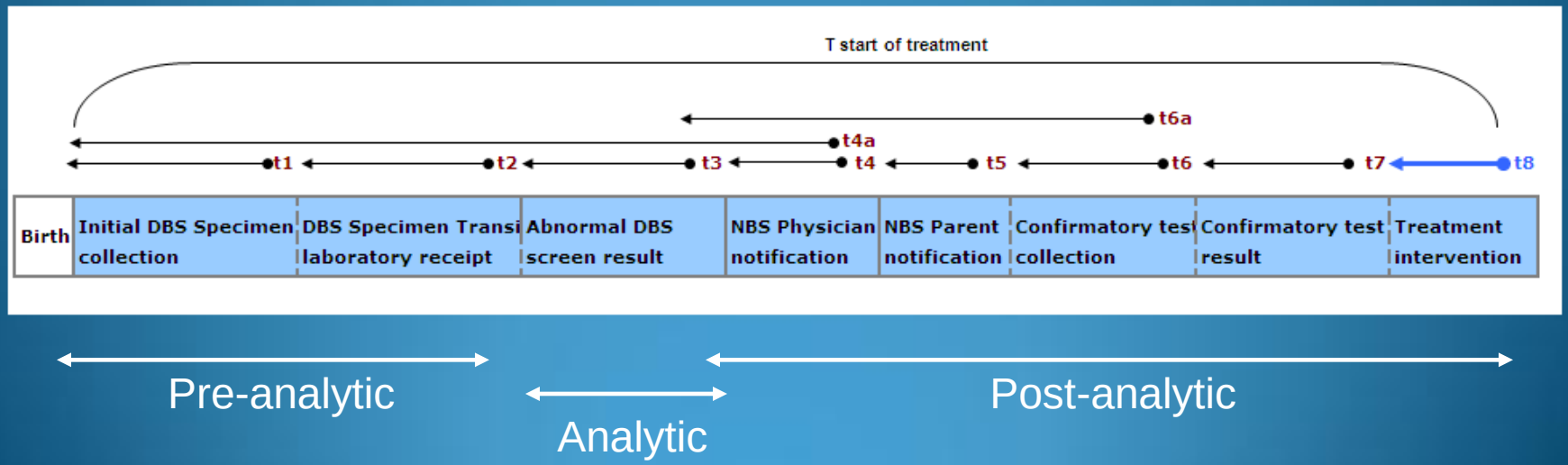


2005 Report "Newborn Screening: Toward a Uniform Screening Panel and System"

- Specimen transport addressed briefly on page 80.
- "It is suggested that specimens be transported by courier services that allow for receipt at the testing laboratories within 24 hours."
- "Some conditions can be life threatening (e.g., MSUD, CAH, GALT, organic acidurias, fatty acid oxidation disorders, urea cycle disorders) within a few days after birth, so it is desirable to initiate specimen processing within 24 hours of specimen receipt in the laboratory, with a 5-day turnaround time between birth and the availability of the test results."

Recommended Timeframes – ACMG Report

- Time of collection: 24-48 hrs
- Transit time: receive at lab within 24 hrs of collection
- Time to result for critical results: within 5 days of life
- Time to result for all results: within 5 days of collection



Key Points of Subcommittee Discussion

- Quality of samples is critical to timely results
- Access to data is key
  - Important to have raw data to be able to see outliers
- Education is needed throughout the healthcare system
  - Knowledge of time sensitive nature of newborn screening may not reach all levels (e.g. send out from lab)
- March of Dimes is convening a consortium to discuss the issues
  - Consolidation of efforts
- Need to review the appendix of the ACMG report

Key Points of Subcommittee Discussion

- State examples - Iowa gathered data and adjusted staffing and testing based on data
  - Prioritization of tests
  - 7 day/week testing & reporting
- Need to gather and share best practices
- Since 2005, test platforms have been added/changed and new conditions have been added.
  - Re-examine recommendations
    - Do they still make sense?
    - Should they be changed?
    - Are there other needed recommendations?

Next Steps:

Proposal for DACHDNC Discussion and Potential Recommendations

Kellie Kelm, Ph.D. – Chair,
Laboratory Standards and Procedures Subcommittee

Proposal for DACHDNC discussion and potential recommendations

1. Re-affirm the recommendations in the 2005 Report and urge states to work towards meeting the recommended timeframes.
   - Continue to work with stakeholders and members to gather timeliness data & best practices.
2. Give special consideration to new technologies and conditions added since 2005 and determine whether recommendations need to be updated or clarified.
3. Try to coordinate independent efforts to address the timeliness of newborn screening issue.
   - Avoid duplication of efforts.

Questions?

Proposal for DACHDNC discussion and potential recommendations

1. Re-affirm the recommendations in the 2005 Report and urge states to work towards meeting the recommended timeframes.

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  - Time to result for critical results: within 5 days of life
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Proposal for DACHDNC discussion and potential recommendations

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    Avoid duplication of efforts.