Review of Newborn Screening Timeliness

Presented to the Advisory Committee on Heritable Disorders in Newborns and Children

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Goal

To review timeliness activities undertaken by newborn screening programs to achieve the established timeliness goals, and to assess progress in meeting the timeliness goals.

a) What are states doing to meet the ACHDNC's timeliness goals for newborn screening
b) What progress has been made by newborn screening programs toward meeting newborn screening timeliness goals
c) What strategies are effective, and what challenges and barriers to meeting timeliness goals

Background

2006 - ACMG recommendations for timeliness of newborn screening

- all specimens should be received at the NBS laboratory within 3 days after being collected from the newborn
- newborn screening results should be reported within 2 days of receipt, and within 5 days after being collected from the newborn.
- 2013 ACHDNC meeting – discussion of an investigation of newborn screening practices across the country, identifying delays in processing, transport, and reporting, some of which were related to poor health outcomes.

2014 Timeliness Workgroup Review

- ACHDNC requested a comprehensive investigation of timeliness activities, NBS system and stakeholders, and recommendations
- Timeliness Workgroup – conducted focus groups and surveyed 51 NBS programs
- In 2014, few programs (≤7) met any timeliness goal for 95% of specimens
- No programs reported specimen receipt at laboratory within 24 hours of collection

1. 2014 State Survey on NBS timeliness practices, Jan 1-May 31 2014* (n=51 NBS programs).

Year	# States Reporting (of 51 surveyed)	Median % of NBS specimens meeting benchmark	# (%) of NBS Programs meeting Committee's goal of 95% specimens
Recommendation 1. NBS specimens should be collected within 24-48 hours after birth (n=43)			
2014*	N=43	82.2% (range 11.0 - 98.3)	~5† (11.6%)
Recommendation 2. NBS specimens should be received at state NBS laboratories within 24 hours of collection.			
2014*	N=31	25.0% (range 0.6 – 80.8)	0 (0%)
Recommendation 3. NBS results should be reported out for time-critical conditions within 5 days of life			
2014*	N=17††	75.8%‡ (range 0.0 – 99.0)	2 (11.8%)
Recommendation 4. Time from specimen receipt at the state's NBS laboratory to reporting out normal and out-of-range results from first specimens, for all disorders, should be within 5 days.			
2014*	N=22	81.9% (range 0.0 – 100.0)	7 (31.8%)

†Estimated from figures presented in Kelm et al. 2015.
†† of 37 NBS programs which differentiate time-critical conditions
‡of 17 of the 37 states provided data on NBS results

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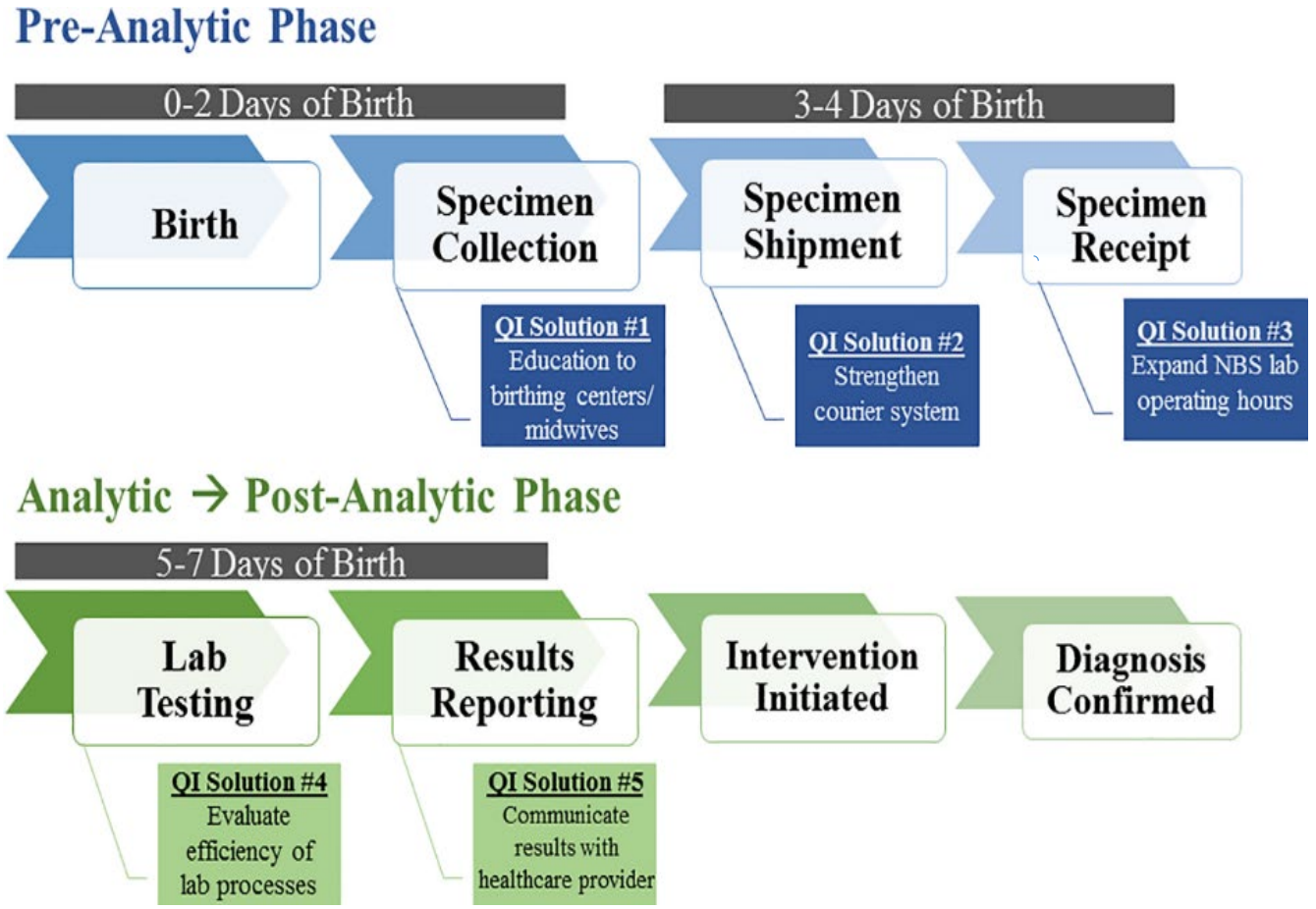
Newborn Screening Phases, with ACHDNC Recommended Timeliness Goals



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ACHDNC Recommendation: states meet NBS Timeliness Goals for ≥95% or more specimens

ACHDNC Newborn Screening Timeliness Goals

A. To achieve the goals of timely diagnosis and treatment of screened conditions and to avoid associated disability, morbidity and mortality, the following time frames should be achieved by NBS systems for the initial newborn screening specimen:

1. Presumptive positive results for time-critical conditions should be communicated immediately to the newborn's healthcare provider but no later than 5 days after birth.
2. Presumptive positive results for all other conditions should be communicated to the newborn's healthcare provider as soon as possible but no later than 7 days after birth.
3. All NBS tests should be completed within 7 days after birth with results reported to the healthcare provider as soon as possible.

B. In order to achieve the above goals:

1. Initial NBS specimens should be collected in the appropriate time frame for the newborn's condition but no later than 48 hours after birth, and
2. NBS specimens should be received at the laboratory as soon as possible; ideally within 24 hours of collection.

NewSTEPs 360 – CoIIN Project (n=28)

QI Timeliness Measure	Time Frame	N	2016		2017		2018	
			Median %	IQR	Median %	IQR	Median %	IQR
Birth to specimen collection	48 Hours	25	95.1%	88.1% - 97.4%	96.4%	90.8% - 97.8%	97.0%	92.4% - 98.3%
Specimen collection to receipt at lab	1 Day	19	40.0%	28.6% - 52.5%	39.4	30.4% - 56.4%	41.8%	28.9% - 56.5%
Specimen collection to receipt at lab	2 Days	19	74.3%	67.8% - 86.6%	79.6%	69.9% - 88.7%	80.9%	70.3% - 88.45%
Receipt to Reporting Results								
Presumptive positive* for time-critical disorders	2 Days	16	65.5%	38.0% - 89.9%	69.7%	50.2% - 88.4%	75.8%	50.5% - 90.4%
Presumptive positive* for non-time-critical disorders	4 Days	15	80.2%	56.9%-93.9%	90.0%	72.4% -95.1%	93.5%	67.3% - 96.3%
All (normal and presumptive positive results)	4 Days	19	90.3%	69.1% - 98.8%	90.8%	83.0% - 99.2%	94.2%	88.3% - 99.3%
Birth to Reporting Results								
Presumptive positive* for time-critical disorders	5 Days	16	48.9%	25.8% - 73.8%	48.8%	34.3% - 71.5%	63.5%	42.5%-71.0%
Presumptive positive* for non-time-critical disorders	7 Days	15	64.4%	57.8% - 77.9%	75.9%	67.1% - 86.0%	80.9%	68.0% - 90.7%
All (normal and presumptive positive results)	7 Days	18	88.9%	68.8% - 96%	87.6%	78.4% - 95.7%	89.5%	84.8% - 98.2%

* Presumptive positive indicates with high probability that the infant may have the disorder; however confirmatory diagnostic testing is required.

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By 2018, among 25 of 28 CoIIN participating programs, median % of specimens meeting timeliness goals for 95% of specimens increased, showing progress. Birth to specimen collections within birthing centers are approaching benchmark goals. However, few programs have met the ACHDNC's timeliness goals for 95% of specimens.

Effective Strategies

- **Birth to Collection time** – work with birthing hospitals and staff to educate and train, conduct quality improvement to develop timely and efficient workflows, and monitor
- **Collection to Receipt at Lab** –
  - Expanding operating hours improves timeliness
  - External laboratories improves timeliness

Days/week Labs Open	Med % of time-critical specimen results reported out within 5 days (2016-2018)
7	>80%
6	<65%
5	<50%

- **Time to report out of results** (from lab receipt, from collection). Involves communication and coordination across multiple stakeholder entities. Enhance IT and electronic data capture and transmission systems to facilitate 24/7 access to reports. Major change required. Also facilitates other workflow and NBS reporting systems (e.g., follow up).
- **Distinction between time-critical and non-time-critical disorders**. Not all states distinguish these. Critical outcomes of timeliness depend on meeting goals for time-critical disorders.
- **NewSTEPs data repository and quality indicators tracking** has facilitated monitoring infrastructure – common definitions, technical support, regular reporting.

Challenges

- Incomplete data reports re: timeliness metrics. Only ~26% to 35% (14-35) programs report timeliness metrics.
- Difficult to track individual program progress over time.
- Programs continue to operate only 5 days/week. Six programs report both courier and laboratories operating 5 days/week. Timeliness metrics for these state programs are not reported.
- State variability in challenges and facilitators creates additional obstacles.

Recommendations

- Revisit Timeliness Goals and timeline for achieving them (was 2017). Other metrics considered (e.g., 48 hours receipt by lab) may be more sensitive to progress.
- Current Goals focus on laboratory processing, less on goals for newborn screening and clinical impact. Additional tracking indicators that align with goals for timeliness of NBS -- Time-critical disorders and time-to-diagnosis and to intervention/treatment – provide useful information on critical outcomes for timeliness.
- Ensure that ALL state programs are making progress toward goals.
- Continue quality improvement practices for specimen transport and post-analytic/reporting phases.