TIMELINESS OF NEWBORN SCREENING: SUGGESTED RECOMMENDATIONS FROM DACHDNC LABORATORY STANDARDS AND PROCEDURES SUBCOMMITTEE

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TIMELINESS - BACKGROUND

- In order to effectively reduce disability, morbidity and mortality, NBS must occur before onset of symptoms.
- NBS panels have changed and include time-critical conditions
  - Conditions that may manifest with acute symptoms in the first days of life
  - Require immediate treatment to reduce risk of morbidity and mortality

TIMELINESS - BACKGROUND

- DACHDNC Laboratory Standards and Procedures Subcommittee tasked with investigating timeliness of newborn screening in the United States (September 2013)
  - Public comment at DACHDNC meeting
  - States surveyed on current practices
  - Guidelines/literature were reviewed

- Media raises the issue nationally to the general public (November 2013)

TIMELINESS - BACKGROUND

January 2014: 4 recommendations made by DACHDNC

1. Initial NBS specimens should be collected at 24 to 48 hours of life.

2. NBS specimens should be received at the laboratory within 24 hours of collection.

3. Newborn screen results for time-critical conditions should be available within 5 days of life.

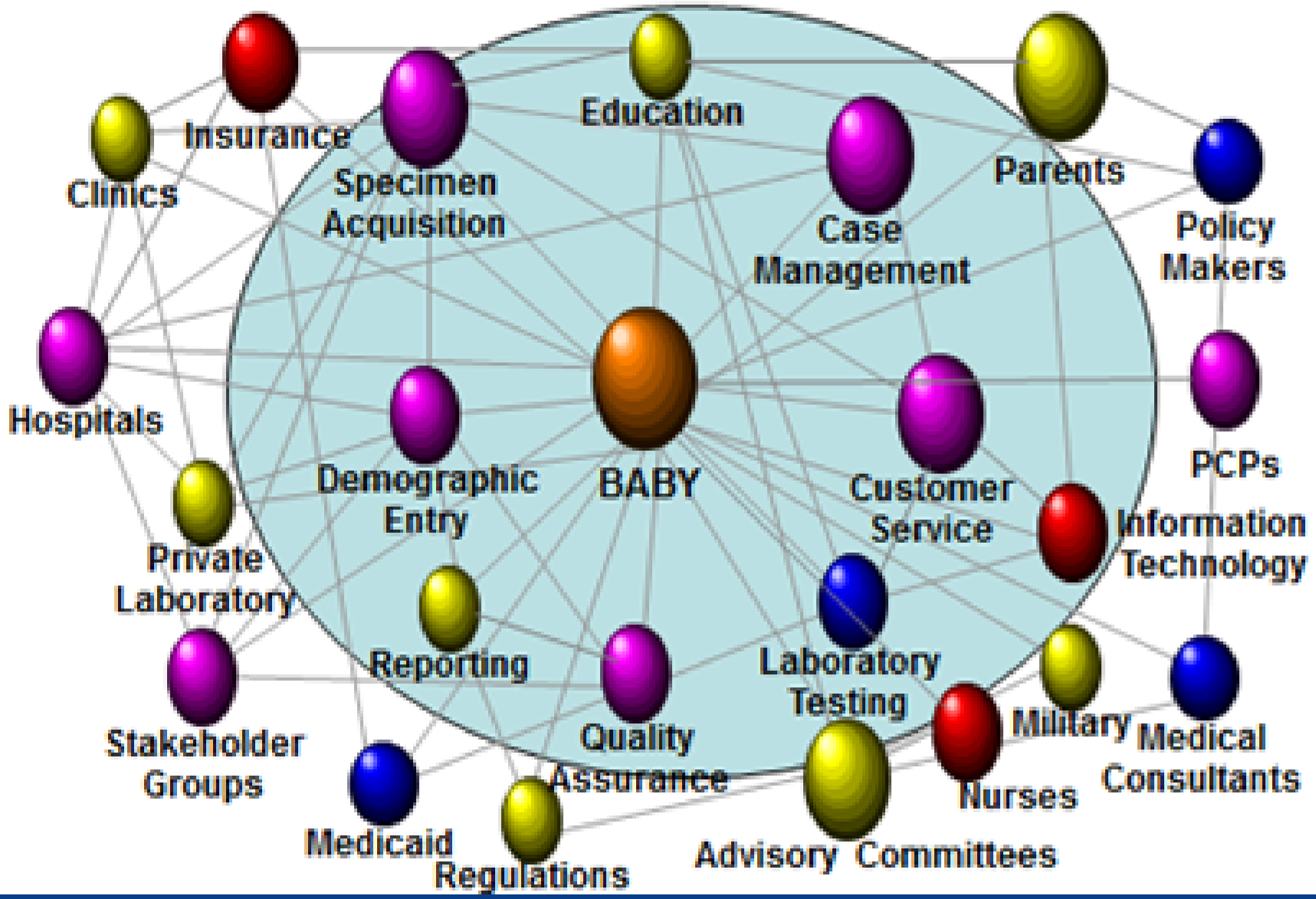
4. All NBS results should be available within 5 days of collection.

DACHDNC MEETING – JANUARY 2014

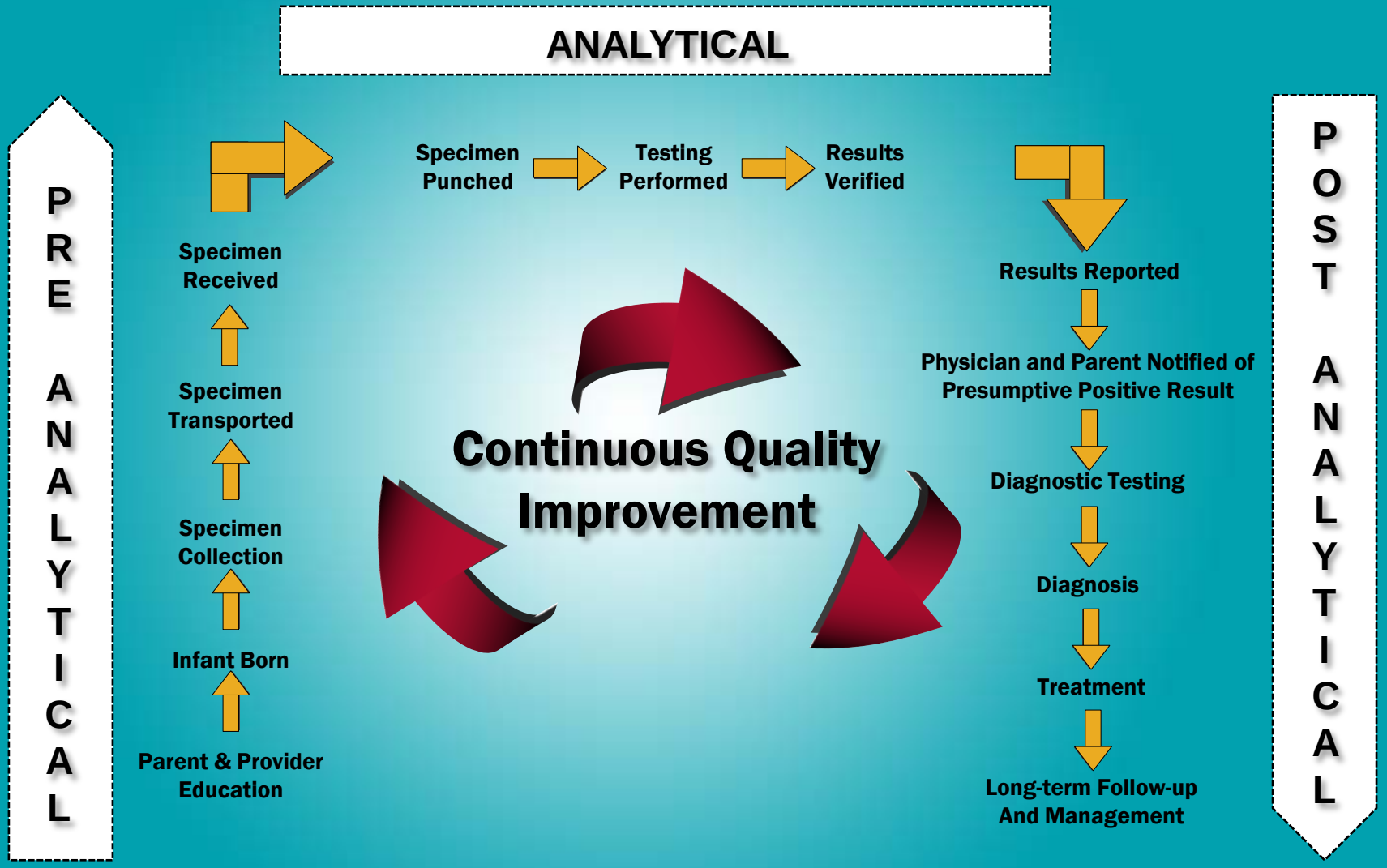
Subcommittee tasked to:

1. Outline the NBS system
2. Investigate existing gaps and barriers in NBS systems
3. Identify strategies to achieving these goals
4. Develop a list of critical conditions that require urgent follow-up
5. Review the recommendations in light of new technologies
6. Suggest revisions, if needed.

Newborn Screening System Partners



NBS System Process



TIMELINESS WORKGROUP

- Group of laboratory experts from the Laboratory Standards and Procedures Subcommittee:

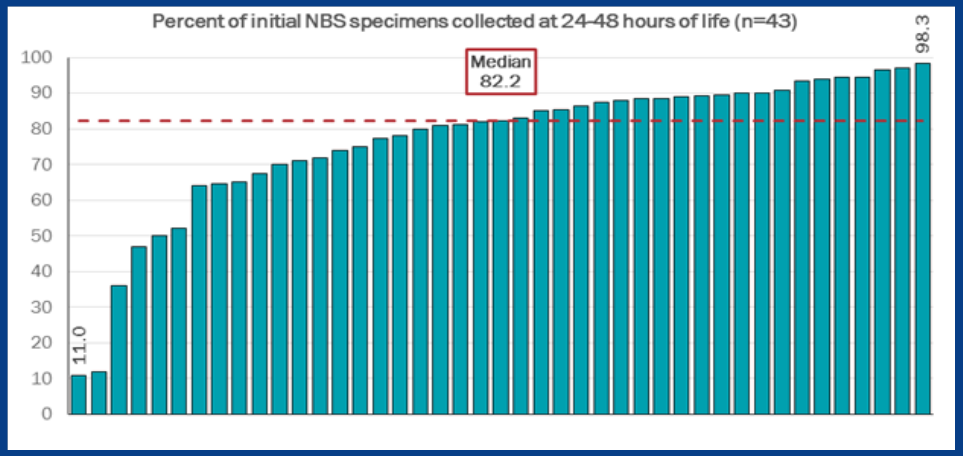
Stan Berberich	Dieter Matern	Michele Caggana
Mei Baker	George Dizikes	Bill Slimak
APHL	Debi Sarkar	Tina Turgel
Susan Tanksley	Kellie Kelm	

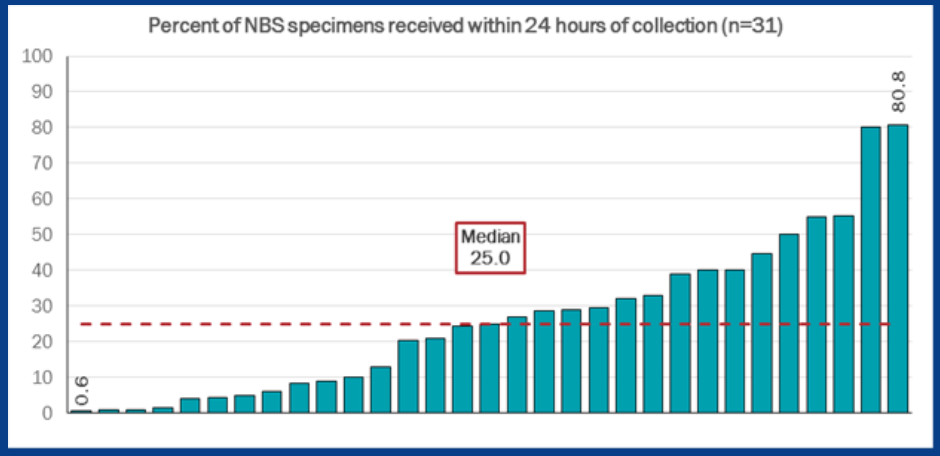
- Internal discussions
- Discussions with clinical experts
- Developed a Discussion Guide to gather information on state's compliance with recommendations, gaps/barriers and strategies for improvement
  - In-person meetings
  - Webinars & conference calls
- APHL fielded survey & developed: Newborn Screening Timeliness – Survey Report
  - Current status, gaps, barriers & best practices

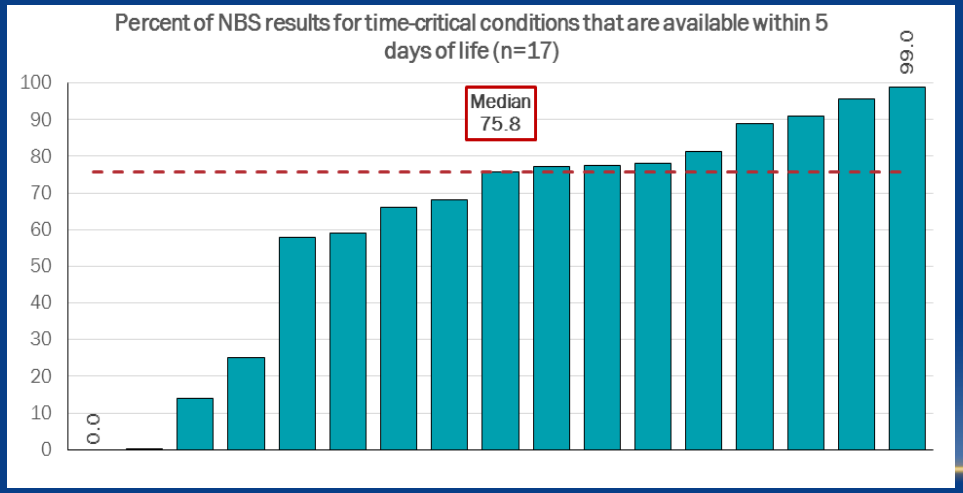
TIME-CRITICAL DISORDERS

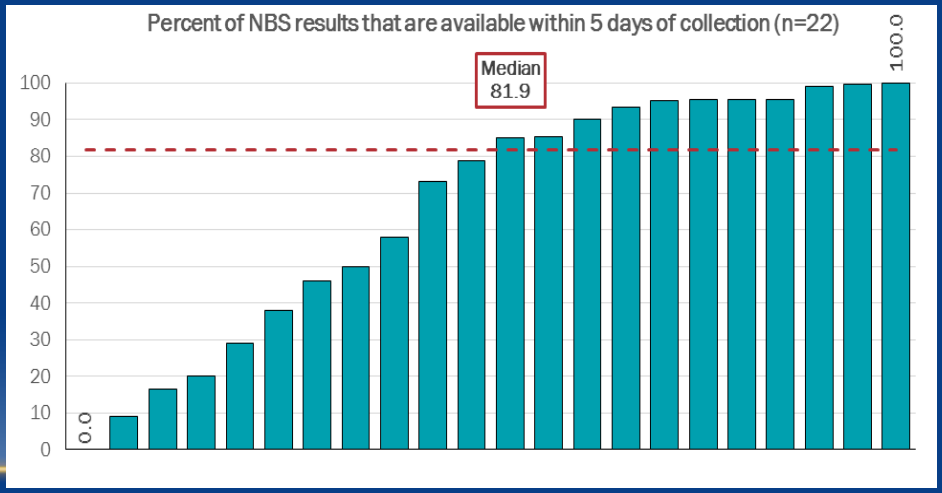
Organic Acid Conditions	Fatty Acid Oxidation Disorders
Propionic acidemia (PROP)	Medium chain acyl-CoA-dehydrogenase deficiency (MCAD)
Methylmalonic acidemia (methylmalonyl-CoA mutase) (MUT)	Very Long chain acyl-CoA dehydrogenase deficiency (VLCAD)
Isovaleric acidemia (IVA)	Long chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
3-Hydroxy-3-methyglutaric aciduria (HMG)	Trifunctional protein deficiency (TFP)
Holocarboxylase synthase deficiency (MCD)	
β-Ketothiolase deficiency (BKT)	
Glutaric Aciduria, Type 1 (GA1)	
Amino Acid Disorders	**Other**
Argininosuccinic aciduria (ASA)	Classic galactosemia (GALT)
Citrullinemia type 1 (CIT)	Congenital adrenal hyperplasia (CAH)
Maple syrup urine disease (MSUD)	

STATUS OF RECOMMENDATIONS









Data for Jan-May 2014

Many states were not meeting the recommendations.

GAPS/BARRIERS THAT IMPACT ABILITY TO MEET GOALS

- Lack of awareness of urgency of NBS
- Lack of training/High turnover of staff performing DBS collection
- Batching by birthing facilities
- Geographic distance from birthing facility to NBS laboratory
- Lack of availability of courier/overnight delivery services
- Operating hours of the courier
- Operating hours of the NBS Program
- Lengthy testing algorithms to avoid high false positive rate
- Lack of ability to collect complete data
- Inefficiencies in the system
  - Specimens collected in proper timeframe may not be dry & ready for courier pick up
  - Laboratory results ready but demographic information is not yet entered into LIMS

STRATEGIES FOR IMPROVEMENT

- Utilize courier or overnight delivery services
- Expansion of NBS program operating hours (laboratory & follow-up)
- Provide educational activities to birthing facility staff, laboratory staff & parents
- Improve reporting and communications mechanisms
  - Electronic ordering and resulting
- Focus on continuous quality improvement activities
  - Batching by birthing facilities/submitters
  - Decrease time from receipt in the lab to reporting
- Improve data collection to allow for evaluation
- Performance monitoring and feedback

SUGGESTED RECOMMENDATIONS FOR TIMELY NBS

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 programs:

    1. Presumptive positive results for time-critical conditions should be communicated immediately to the newborn's healthcare provider but no later than five days of life.
    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 seven days of life.
    3. All NBS tests should be completed within seven days of life.

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.

ISSUES NEEDING FURTHER INVESTIGATION

- Continue/expand collaboration with American Hospital Association and possibly the Joint Commission to work on collection and transport inefficiencies at hospitals
- Develop recommendations based on communication of NBS results, whether presumptive positive for or normal, to the family of the affected infant
- Continued need for improved standardization of reporting procedures/statements

MOVING FORWARD

Recommendations are GOALS for NBS systems to achieve the best outcomes for affected infants.

NBS is a system – The parts must work together to achieve the best outcomes

- To achieve goals:
  - Must remove gaps & mitigate barriers
  - Can follow examples of other states
  - Must have buy-in throughout the system
  - Must have funding
- Critical that as we work to improve timeliness that we achieve a balance and not negatively impact the NBS system.

ACKNOWLEDGEMENTS

- DACHDNC Timeliness Workgroup:
- APHL:

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  Careema Yusuf Jelili Ojodu

- Laboratory Standards and Procedures Subcommittee
- Society of Inherited Metabolic Disorders
- Clinical experts in endocrinology, hematology, and pulmonology