TIMELINESS OF NEWBORN SCREENING: RECOMMENDATIONS FROM ADVISORY COMMITTEE ON HERITABLE DISORDERS IN NEWBORNS AND CHILDREN

Susan Tanksley, PhD

May 19, 2015

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 may manifest with acute symptoms in the first days of life
  - Require immediate treatment to reduce risk of morbidity and mortality

TIMELINESS - BACKGROUND

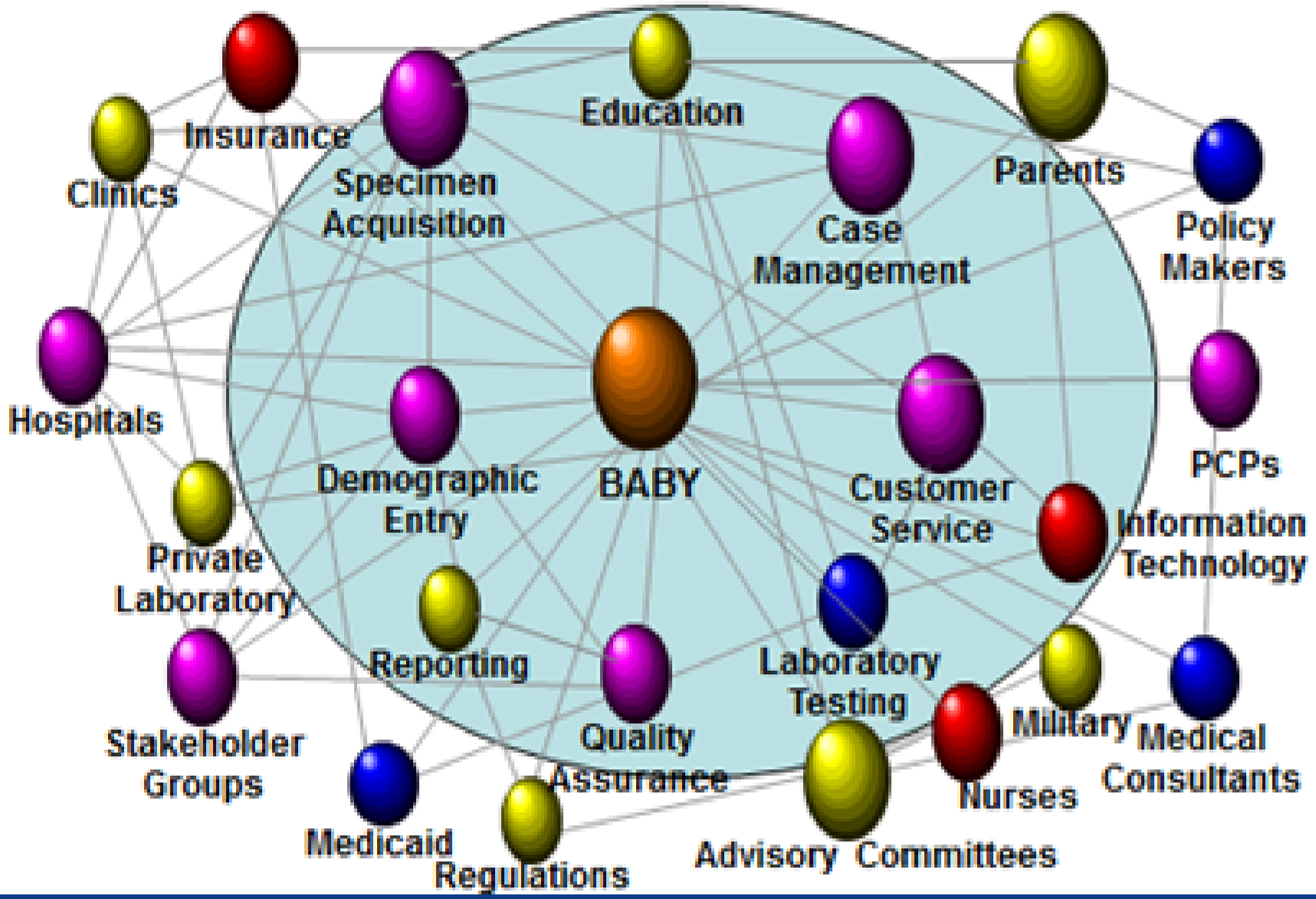
- DACHDNC Laboratory Standards and Procedures Subcommittee tasked with investigating timeliness of newborn screening in the United States (September 2013)
  - Public comment at DACHDNC meeting
  - States surveyed on current practices
  - Guidelines/literature were reviewed
- Media raises the issue nationally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

DACHDNC MEETING – JANUARY 2014

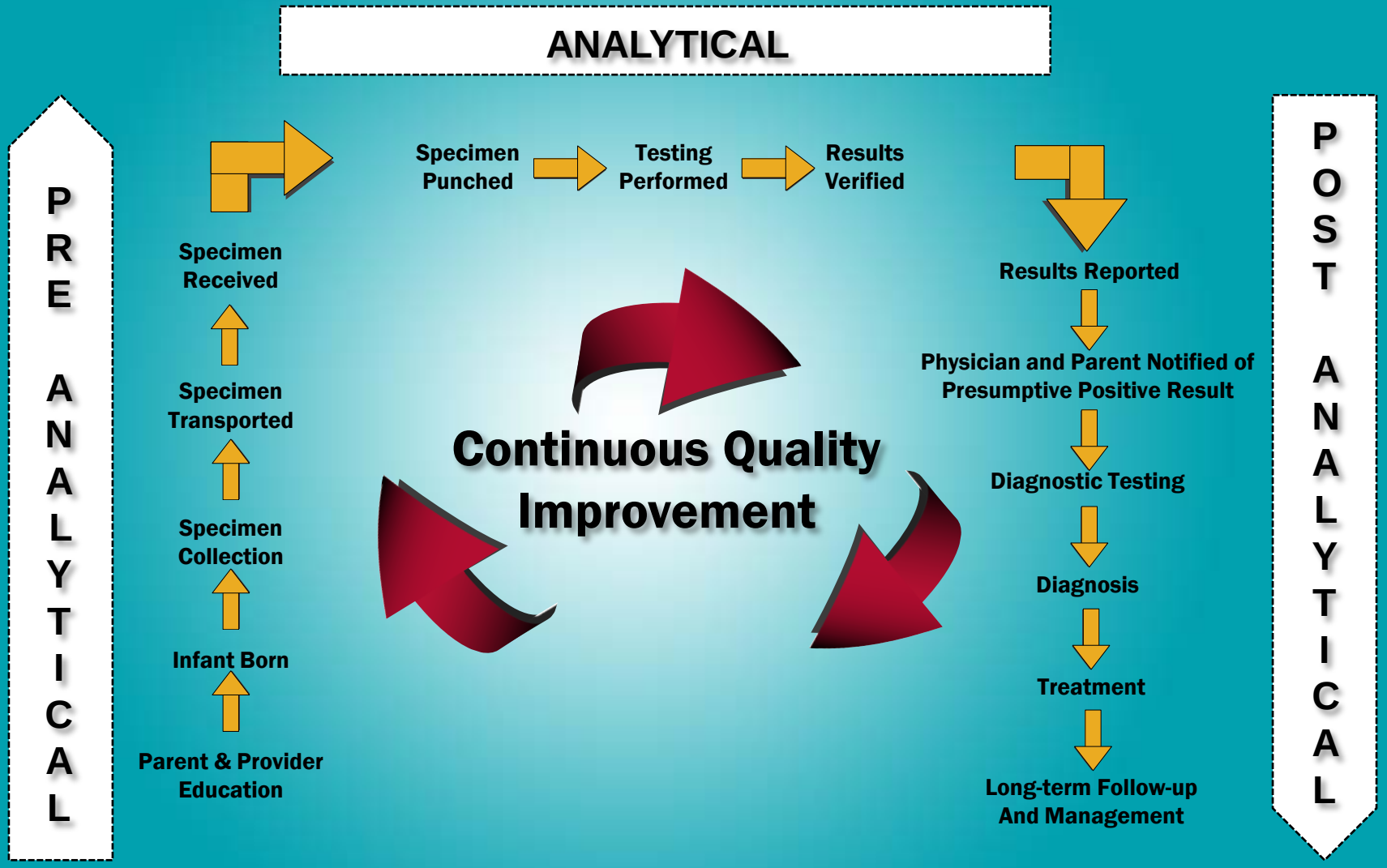
Subcommittee developed 4 draft recommendations and tasked Lab Standards and Procedures Subcommittee to:

1. Outline the NBS system
2. Investigate existing gaps and barriers in NBS systems
3. Identify strategies to achieving the 4 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



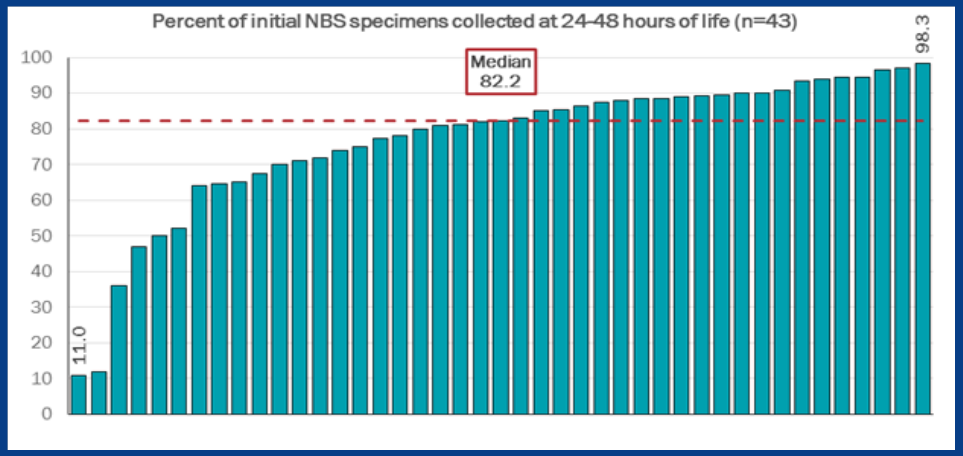
NBS System Process

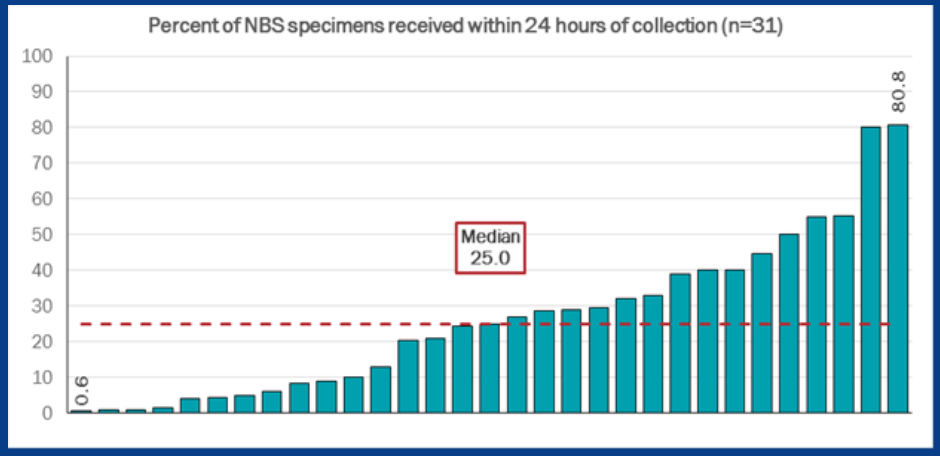


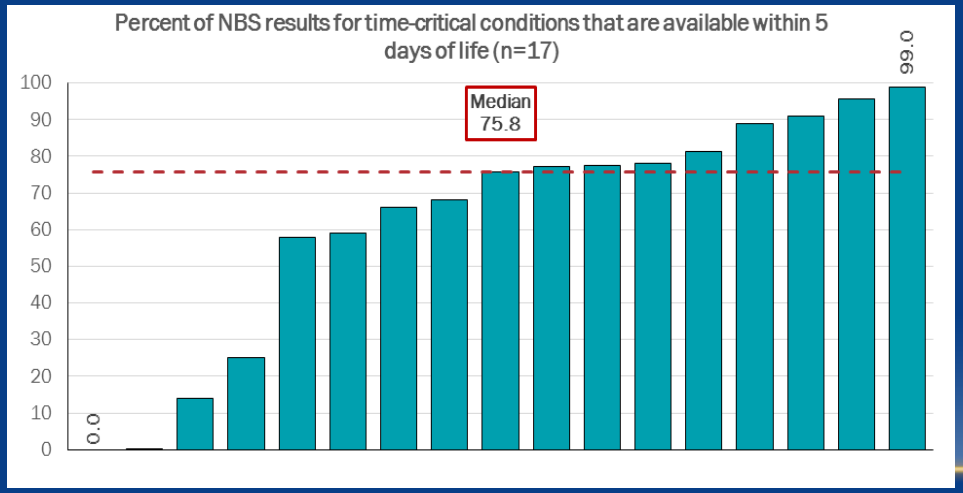
TIME-CRITICAL DISORDERS

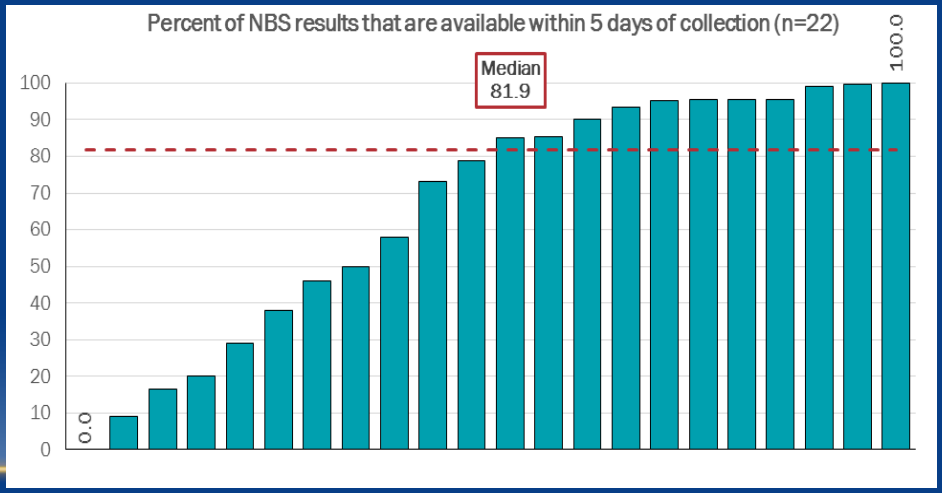
Organic Acid Conditions	Fatty Acid Oxidation Disorders
Propionic acidemia (PROP)	Medium chain acyl-CoA-dehydrogenase deficiency (MCAD)
Methylmalonic acidemia (methylmalonyl-CoA mutase) (MUT)	Very Long chain acyl-CoA dehydrogenase deficiency (VLCAD)
Isovaleric acidemia (IVA)	Long chain L-3-hydroxyacyl-CoA 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)	

NEWBORN SCREENING TIMELINESS – SURVEY REPORT:
STATUS OF DRAFT 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
- Monitor performance and provide 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 systems for the initial newborn screening specimen:

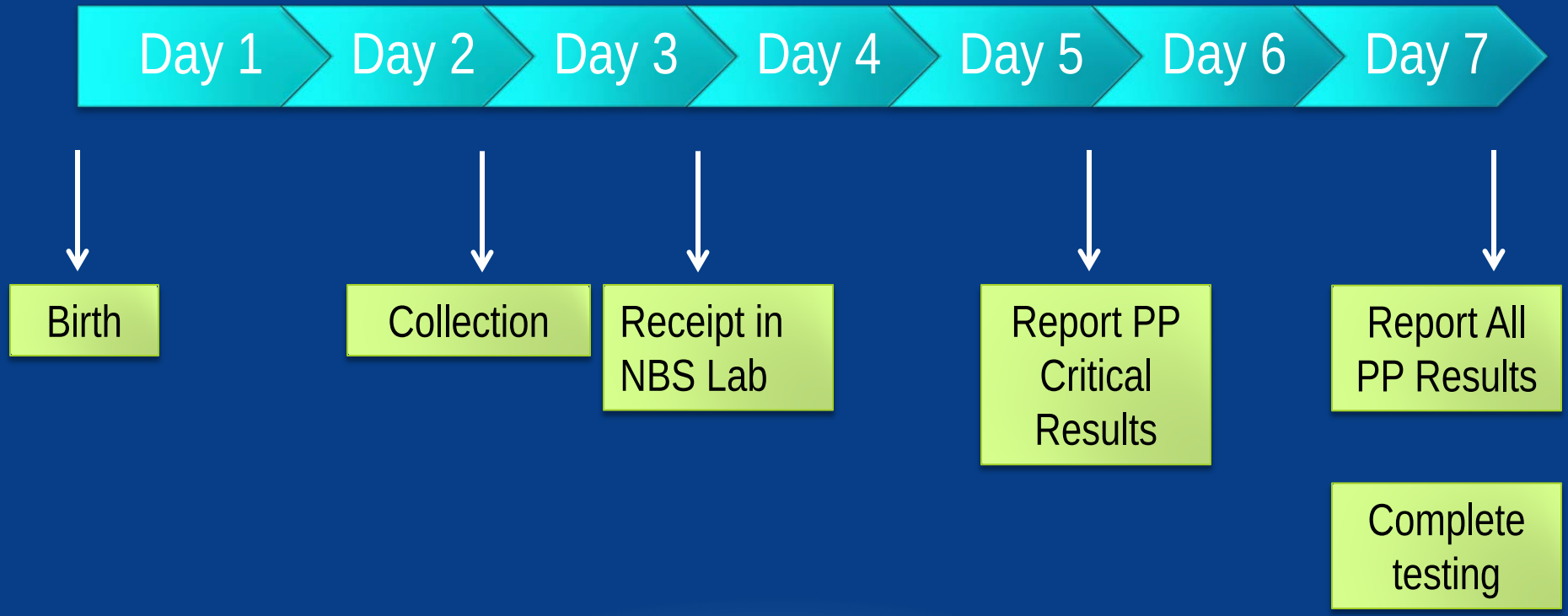
   1. Presumptive positive results for time-critical conditions should be communicated immediately to the newborn's healthcare provider but no later than five days of life.
   2. Presumptive positive results for all other conditions should be communicated to the newborn's healthcare provider as soon as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

The Committee encourages States to monitor their progress in achieving each recommendation and make the information readily available to providers and the general public.

RECOMMENDED TIMELINE



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

- Timeliness Workgroup:

Stan Berberich	Dieter Matern	Michele Caggana
Mei Baker	George Dizikes	Bill Slimak
Jelili Ojodu	Debi Sarkar	Tina Turgel
Guisou Piñeyro	Sikha Singh	Careema Yusuf
Susan Tanksley	Kellie Kelm	

- Advisory Committee on Heritable Disorders in Newborns and Children
- Laboratory Standards and Procedures Subcommittee
- Society of Inherited Metabolic Disorders
- Clinical experts in endocrinology, hematology, and pulmonology