DEPARTMENT OF HEALTH AND HUMAN SERVICES

Discretionary Advisory Committee on
Heritable Disorders in Newborns and Children
5600 Fishers Lane, Room 18W68
Rockville, Maryland 20857
www.hrsa.gov/heritabledisorderscommittee

April 16, 2015

The Honorable Sylvia Mathews Burwell
Secretary of Health and Human Services
200 Independence Avenue, S.W.
Washington, DC 20201

Dear Secretary Burwell:

During the past eighteen months, the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (Committee) has been reviewing current policies and practices relating to timeliness of newborn screening (NBS) in the United States. Prompted by a public comment during the September 2013 meeting, the Committee determined that it was necessary to address the concerns raised regarding the timely collection, receipt, and reporting out of the results of the initial newborn screening specimen.

NBS is a complex system that consists of six major components:

1. Education, including prenatal education
2. Screening, including specimen collection and testing
3. Follow-up, including results reporting
4. Diagnostic confirmation
5. Short term and long term management
6. Program evaluation and continuous quality improvement

Each component must not only function effectively independently but each component must align efficiently with all of the other activities to ensure the appropriate interventions are initiated within a timeframe which avoids harm and ultimately provides the best benefit to the newborn infant. To add to the complexity, each NBS program in the United States is a State run program so each program's activities may be conducted differently based on State legislation, policy and/or practice.

One critical aspect of the system is the timely collection, transport, laboratory testing and reporting of abnormal screening test results obtained from the initial newborn screening specimen. In 2005, the *Newborn Screening: Toward a Uniform Screening Panel and System Report*, made four recommendations on the timeliness of NBS (access at http://mchb.hrsa.gov/programs/newbornscreening/screeningreportpdf.pdf). The Committee felt that it was necessary to revisit these recommendations to determine if they held true in light of the incorporation of new technologies and an expanded RUSP. In addition, the Committee examined the current NBS system, in particular, the gaps and barriers which exist within the system for timely collection, transport, testing and reporting of abnormal screening test results.

The Committee also examined successful strategies within individual States to overcome gaps and barriers, and identified those critical conditions that require urgent follow-up care. The overall goal of these efforts is to reduce delays in reporting to minimize morbidity and mortality associated with conditions which require prompt intervention.

Many States were not meeting the initial 2005 recommendations.

- 82.2% of States (n=43) reported that initial NBS specimens were collected within 24-48 hours of life.
- Only 25% of States (n=31) reported that NBS specimens were received in the laboratory for analysis within 24 hours of collection.
- 75.8% of States (n=17) reported that NBS results for time-critical conditions were available within 5 days of life.
- 81.9% of States reported that all NBS results were available within 5 days of collection.

Gaps and barriers that impacted the ability to meet the initial goals included:

- Lack of awareness of urgency of NBS
- Lack of training/high turnover of staff performing dried blood spot (DBS) collection
- Batching of specimens 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/Laboratory
- 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 the Laboratory Information Management System (LIMS)

The Committee also examined strategies that many States implemented to improve timeliness of NBS including:

- Utilizing courier or overnight delivery services
- Expansion of NBS Program/Laboratory operating hours
- Providing educational activities to birthing facility staff, laboratory staff & parents
- Improving reporting and communications mechanisms
- Electronic ordering and resulting
- Establishing continuous quality improvement activities
- Batching by birthing facilities/submitters
- Decreasing time from receipt in the lab to reporting
- Improving data collection to allow for evaluation, performance monitoring and feedback

Based on our deliberations and findings, the Committee believes that the best NBS outcomes for newborn infants and their families will be achieved if stakeholders such as State NBS programs, birthing facilities and hospitals adopt the following practices and timelines:

A. To achieve the goals of timely diagnosis and treatment of screened conditions and to avoid associated disability, morbidity and mortality, the following timelines 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.

Although we are not seeking Secretarial action, the timelines mentioned above should be viewed as goals for the entire NBS system in order to achieve the best outcomes for infants identified through the NBS process with a potentially harmful or life-threatening condition. The Committee recommends State NBS programs monitor their progress in achieving the above goals and make the information readily available to the general public, hospitals, providers, and other stakeholders. As part of monitoring progress, the Committee encourages NBS programs to benchmark their progress with the goal of having 95% or more of newborns meeting the timeliness goals by 2017 and to report the information to a national data resource.

In addition, the Committee recognizes that States will need support in achieving these goals and encourages the development of a grant program to further assist States in overcoming barriers to meet the timeliness goals.

The entire NBS system process, from sample collection through transit to the laboratory, testing and reporting, needs to be time-effective to meet the recommendations. NBS systems can use the new goals for timeliness to achieve the best outcomes for all newborns and their families. We recognize the challenges that NBS programs, hospitals, parents, and other stakeholders face regarding the timeliness of NBS today and hope that the goals outlined above provide clarity and direction.

We will continue to support the Department's efforts to strengthen the newborn screening programs that play such an important role in improving the health of the Nation's children.

Sincerely yours,

Joseph A. Bocchini Jr., M.D.
Chairperson

Enclosure:
Timeliness of Newborn Screening: Recommendations from the Secretary's Discretionary Advisory Committee on Heritable Disorders in Newborns and Children

cc: Debi Sarkar, M.P.H.
Designated Federal Official
Health Resources and Services Administration