Newborn Screening Guidelines for Premature and/or Sick Newborns

Proposed Guideline

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A document developed through the **consensus process** describing criteria for a general operating practice, procedure, or material for voluntary use. A guideline may be used as written or modified by the user to fit specific needs.
CLSI “Consensus” Process

- The CLSI has formal criteria for:
  - Authorization of the project
  - Development of “Proposed” level document
  - Publication of proposed level document for CLSI delegate vote and comments
  - Revision of documents in response to delegate / public comments creating an “approved” level guideline
  - Acceptance and publication of a document as a consensus approved guideline
CLSI “Consensus” Process

Subcommittee working group developing the document consists of representatives from three different constituencies:

1. Government agencies
2. Professional societies
3. Industry representatives

International community representation also sought to help assure the global applicability of the document.
Subcommittee working group members

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• Currently in the “Proposed” stage
  – Proposed-level documents can **NOT** and should **NOT** be considered final
  – Proposed-level documents are published for CLSI delegate review and comments
  – CLSI Proposed-level documents are available for purchase
    (Price depends on CLSI organizational membership status)
Guiding Principles

Guiding principle in order to ID those at higher risk who need more testing:

• Use infant’s limited blood resources efficiently by collecting at times which will give the most reliable results with the least number of specimens

• Obtain specimens in time to identify those conditions with the shortest “screening windows” (time when we have the greatest chance of detecting condition before symptoms or permanent damage occurs)

• Minimize stress for medically fragile newborns
Goals

• Assure rapid consistent & complete blood spot & hearing screening, follow-up, diagnosis & treatment

• Minimize risk of missed or delayed diagnosis

• Optimize timing / minimize # specimens/hearing screens

• Streamline follow-up

• Define essential elements of QA

• Provide education on effects of NICU/SCBU treatments on screening

• Identify areas needing further research
Primary Considerations

• Effects of maternal conditions and treatment on the NBS results

• Prematurity, low birth weight or illness

• Treatment given in the NICU (SCBU)
General Recommendations

• **SCREEN UPON ADMISSION**
  – Advantage:
    • Reliable for hemoglobins, GALT & biotinidase enzymes
    • Provides baseline amino acids and acylcarnitines
    • May be more likely to detect FAO’s due to stress
    • Increases chance screening specimen will be drawn on every SCBU newborn
  – Disadvantage:
    • Increased false positive and negative results for TSH/17OHP and IRT
      – (alternative solutions are recommended for dealing with this)
General Recommendations

• **SCREEN AT 48-72 HOUR (IF FIRST ONE <24°)**
  
  – Advantages:
    • Should be reliable for CH (unless TSH primary), CAH, most aminoacidopathies (unless parenteral nutrition)
      – (90% of first specimen abnormalities disappear)
  
  – Disadvantages:
    • Carnitine disorders may be masked due to parenteral nutrition
    • Will not identify CH with delayed rise in TSH
General Recommendations

- **RESCREEN AT 28 DAYS OR UPON DISCHARGE only for infants < 33 6/7 weeks gestation or < 2000g**
  - This “qualifier” in recommendation because: 3\textsuperscript{rd} screen may not be necessary for the many admitted infants 34-40+ weeks gestation with shorter NICU stays and fewer interfering interventions during their admission.

Advantages:
- For especially small premature babies, thyroid function may have matured to expected newborn levels
- May resolve (for most) any previous abnormal results such as multiple amino acids or carnitine elevations
- Especially beneficial for the very low birth weight babies likely to have had more interventions that interfere with NBS results

- Disadvantages:
- Cost of additional screening may be an issue?
Birth of premature, LBW or sick newborn.

TRANSFER/ADMISSION to NICU or special care baby unit (SCBU)

UPON ADMISSION
COLLECT NEWBORN SCREENING DRIED BLOOD SPOT SPECIMEN
(interpret abnormal results for CH, CAH and CF with caution as many of these may normalize on a repeat screen)

48-72 HOURS OF LIFE
COLLECT REPEAT DBS FILTER PAPER SPECIMEN
on infants initially tested at <24 hours of age or any infant with abnormal results on first screen; Re-screen complete panel on all

AT TWENTY-EIGHT (28) DAYS OF LIFE
COLLECT REPEAT DBS SPECIMEN
(For infants < 33 6/7 weeks gestation or < 2000 grams)
(If discharge is before 28 days of life, collect a specimen upon discharge)
Re-screen thyroid on all; Depending on earlier results, repeat all tests previously outside normal limits / reference ranges.
General Recommendations

Hearing Screening

• All NICU babies admitted for greater than 5 days are to have auditory brainstem response (ABR) included as part of their screen so that neural HL will not be missed.
• NICU infants are to be screened when considered medically stable and prior to discharge. The goal for screening is by one month of age but this may not be possible for very preterm infants or critically ill infants on ventilators.
Public Comments!

- All comments received from the CLSI delegates and other experts who obtained the proposed-level document will be addressed and included as an appendix of the “approved-level” document.
- To obtain a copy of the proposed level document contact CLSI at www.clsi.org.
- Proposed level documents are open for 60-day voting and commenting period (December 2008-February 2009).
ACKNOWLEDGEMENTS

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• Gdlns. Committee Working Group Members
• Guidelines Committee Advisors
Any Questions?

Photo courtesy of Judi Tuerck, RN, MSN who would love to help me respond to your questions! 😊