Newborn Screening Essentials

Goal of NBS: To identify newborns with certain *rare* and *common* disorders, hearing loss and *critical* congenital heart defects (CCHDs) and help get them needed treatment.

- Only *some* affected infants will be symptomatic in the first weeks of life.
- Significant hearing loss is present in 1 to 3 out of every 1,000 newborn infants and doubles by school age.

1. Since this is a screening test, the possibility of a false positive or negative result *must* be considered. The test may need to be repeated and diagnosis confirmed or ruled out by additional diagnostic studies. Remain watchful for signs or symptoms of these disorders.

2. Every infant should have **2** bloodspot screens, **1** pulse ox screen, and at least **1** hearing screen.
   1ST BLOODSPOT SCREEN: Collect between 24 - 36 hours of life **OR** prior to transfusion/intervention, **OR** prior to discharge. Although less than ideal, it's *never* too early
   2ND BLOODSPOT SCREEN: Collect between 5 - 10 days of age **OR** at first doctor visit if unknown screening status (however, there must be at least 3 days between screens). Although less than ideal, infants up to 1 yr. of age can be screened.
   HEARING AND PULSE OXIMETRY: Screen at about 24 hours of life.

3. Timing and Collection—the second bloodspot screen should be collected between 5-10 days of life and if required, the second hearing screen should be completed before 30 days.

4. Most babies discharge before lab results are known—make sure parents know if more testing is required.

5. The **Blue Immunization card** should have the hearing screening results on the back and bloodspot and CCHD results on the inside right corner.

6. Don't forget about Hearing Screening
   - 1 – Hearing screen no later than 1 month
   - 3 – Diagnose no later than 3 months
   - 6 – Early intervention as soon as diagnosed

7. NICU Babies require follow-up
   - Infants in the Neonatal Intensive Care Unit (NICU) for 5 days or more are more likely to have false positive and false negative results; remain vigilant
   - If a NICU graduate fails a hearing screen, refer to a pediatric audiologist for diagnostic testing immediately
   - Infants with risk factors for late onset or progressive hearing loss should have routine hearing testing

8. Convey the importance of newborn screening and consequences of *delayed identification* of affected infants
   - there is no way of predicting affected babies
   - most are identified with no family history
   - most affected babies "**look normal"** at birth
   - permanent damage can be done before symptoms appear and some babies can die
   - risk of a disorder is much more likely than winning the lottery

9. Follow EPDST periodicity guidelines for scheduling appointments