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.

For bloodspot disorders, screen every infant twice

1ST 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 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.

Timing of 2nd screen—the second screen should be collected between 5-10 days of life. Statewide the average is 15.8 days. Please do not wait for the 2 week follow-up visit to order the screen.

Goal of NBS: To identify newborns with certain rare diseases or hearing loss and help get them needed treatment.

• Several disorders can kill or impair development in the first week or two of life if not detected and treated.
• Only some affected infants will be symptomatic in the first weeks of life.
• Infants with hypothyroidism lose significant IQ points if thyroid function is not normalized by 2 weeks of age.
• Significant hearing loss is present in 1 to 3 out of every 1,000 newborn infants and doubles by school age.

Most babies discharge before lab results are known—ask for a copy of the report; read the discharge summary.

If collecting in your office, ship bloodspot specimens promptly to the State Lab.

• Send specimens within 24 hours of collection
• Don’t use the mail—use courier service (we pay for it!)
• Specimens should be received within 3 days but can be analyzed up to 14 days after collection.
• Babies up to one year of age can be tested

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

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

Meconium ileus is a cardinal sign of CF in babies; don’t rely on a normal newborn screen result to rule out disease.

The Newborn Screening program contracts with sub-specialists; they provide technical/clinical expertise to the state and to YOU. Keep their contact information handy and use it!