



NEWBORN SCREENING

Kimberly Noble Piper, RN, BS, CPH, CPHG
Iowa Department of Public Health,
Center for Congenital and Inherited Disorders

WHAT NEWBORN SCREENING IS...

- **A medical screening test-**
 - **Similar to a colonoscopy or mammogram – it can tell you if you might be at risk of having a condition (there is an increased chance you could have a condition) - like if you have a polyp or lump – but you can only tell if you have a condition after further testing (like a biopsy)**
- **A population-based screen – meaning all individuals of a certain population, in this case Iowa newborns, are screened**
- **One of the most successful public health initiatives of the 20th century**
- **A system of care – not just a lab test. Parents, health care providers, hospitals, labs, and newborn screening staff all have a part to play to make sure the system works in the best interest of the babies**

WHAT NEWBORN SCREENING ISN'T...

- **It is not a diagnostic test – it will not tell us that a baby has a condition for sure**
- **If it is a late-onset type of condition, newborn screening cannot tell when, if, or how bad the symptoms of the condition may occur**

THREE IOWA NEWBORN SCREENING PROGRAMS



HEARING SCREENING PROGRAM

Early Hearing
Detection and
Intervention program
(EHDI)

HEART DISEASE SCREENING PROGRAM

Critical Congenital
Heart Disease
Screening Program
(CCHD)

NEWBORN BLOOD SPOT SCREENING PROGRAM

Referred to as the
Iowa Newborn
Screening Program

IOWA NEWBORN SCREENING PROGRAM

- **The Iowa Department of Public Health is required by law to screen all Iowa newborns for certain congenital and inherited disorders.**
- **Currently screen for over 40 conditions**
- **National recommendations to add 4 more conditions to states' screening panel – Pompe` disease, Mucopolysaccardosis Type I (MPSI), X-linked Adrenoleukodystrophy, and Spinal Muscular Atrophy (SMA)**

NEWBORN SCREENING PROCESS

PARENT EDUCATION

- **Every parent is to have received information about the newborn screening before the blood is collected**
- **Responsibility of hospital staff or midwife to review information with parents**
- **If hospital birth, often the NBS information is given to the mother after delivery in a packet of other information the mother is expected to read**
- **Baby's blood is drawn without the parents even knowing**
- **Issues - hospital staff is busy, hospital staff may not understand NBS or be able to adequately answer parent's questions, mother is exhausted and uncomfortable**

BLOOD COLLECTION

- **Baby's heel is pricked with a small lancet**
- **Information is filled out on the form about the mother and baby**
- **Drops of blood are placed on a special filter paper collection form**
- **Collection form is dried for at least three hours and sent to laboratory for processing (if out-of-hospital birth, blood spots are dried for three hours before sending to the State Hygienic Laboratory)**

SPECIMEN IS SENT TO THE STATE HYGIENIC LABORATORY

- **A courier service picks up the specimens from each birthing hospital or birth provider every day (or when they schedule a pick-up) and delivers them to the SHL newborn screening lab in Ankeny, IA by 9:30 every night.**
- **The courier operates 365 days a year.**

SPECIMEN TESTING

- **The State Hygienic Laboratory Newborn Screening Program operates 365 days/year with a day shift and a night shift.**
- **Specimens are logged in as soon as the courier delivers them.**
- **Demographic information is entered into the data system.**
- **Specimens are reviewed to make sure they are good enough to be accurately tested (enough blood on the spots, blood has not clotted, collection form is not damaged, etc.)**
- **Specimens are prepared and placed in the testing machines to begin the processing. Processing for almost all conditions runs overnight.**

REPORTING SCREENING TEST RESULTS

- **Test results are considered normal or abnormal (positive) based on pre-set levels of detection of the substance they are testing for each condition. These pre-set levels are called “cut-offs.” Each type of test has a cut-off value that indicates whether the baby may be at risk for that condition. Cut-off values are set in order to find the most true, or real, cases without having a lot of false, or not real, cases. It can be a delicate balancing act.**
- **Since this is a screening test (not a diagnostic test), we say that a baby with an abnormal test result is “presumptive positive,” or is presumed to have the condition, but further testing is needed to tell for sure.**

REPORTING SCREENING RESULTS

- **Presumptive positive (abnormal) results are reported to the newborn screening short-term follow-up (STFU) staff as soon as the results are double-checked to make sure they are correct.**
 - **STFU staff will call the baby's health care provider to discuss the abnormal result and recommend next steps (usually repeating the screening)**
- **Some conditions are time-critical, meaning the baby needs treatment immediately in order to prevent death. STFU staff contact the baby's doctor immediately and talk to them about what to do for the baby until the baby can receive diagnostic testing. The baby's doctor will immediately call the parents to let them know what to do.**
- **Most babies have a normal screening result. Normal newborn screening reports are sent to the birthing hospital, and the hospital sends them to the baby's health care provider.**
- **The baby's health care provider will share the report with the parents.**

SHORT-TERM FOLLOW-UP (STFU)

- **Short-term Follow-up staff at the University of Iowa Stead Family Department of Pediatrics coordinate the follow-up of any baby with an abnormal result. The STFU staff work 365 days/year.**
- **STFU staff make sure every baby with a presumptive positive test result gets follow-up testing and gets access to appropriate services.**
- **STFU staff stay with a newborn with a presumptive positive screen until they receive a diagnosis and are getting specialty care, or until the newborn is determined to not have the condition.**

LONG-TERM FOLLOW-UP

- **Once a baby is diagnosed as having one of the conditions discovered through newborn screening, they are sent on to receive care from specialists.**
- **The Iowa Newborn Screening Program is trying to figure out how to best work with the specialty care providers to monitor how those babies with a diagnosis are doing years down the road. This information will help us to determine if newborn screening for the condition has had a positive impact on the life of the child.**

THANK YOU!