

EHDI and Dried Bloodspot: We are all Newborn Screening!

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ARIZONA DEPARTMENT
OF HEALTH SERVICES

Health and Wellness for all Arizonans

Disclosures

- ▶ We have no disclosures

Introductions

- ▶ Fran Altmaier
 - ▶ Manager for the newborn screening follow up program
 - ▶ Short term follow up for hearing and bloodspot and Long term follow up
 - ▶ Degree in Social Work
 - ▶ Started in hearing follow up
- ▶ Melissa Dominguez
 - ▶ Senior follow up Specialist
 - ▶ Degree in Healthcare Compliance
 - ▶ Started in Bloodspot follow up
 - ▶ Promoted to senior specialist and transitioning to hearing and bloodspot follow up

Learning Objectives

- ▶ Review the history and background of Newborn Screening
- ▶ Learn how similar Hearing screening Follow-up and DBS screening Follow-up really are
- ▶ Explore data system collaboration for locating families
- ▶ Understand the importance of Newborn Screening follow up collaboration

What is Newborn Screening?

- ▶ A state based, public health service designed to screen and treat newborns for serious health conditions
- ▶ RUSP- National Recommended Uniform Screening Panel (established in 2010 with 29 core conditions)
 - ▶ 2012 Added Point of Care Newborn Screening
- ▶ Strict nomination and voting process by the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)
 - ▶ Best outcomes when treated prior to symptoms
 - ▶ All have treatments available
 - ▶ Some as simple as a diet change or pill, others as complex as Bone Marrow transplant, Enzyme Replacement therapy, Gene Therapy or support for language acquisition
 - ▶ 37 core conditions currently on the recommended core panel-
 - ▶ HEARING is one of them!

History of Newborn Screening



- ▶ Every state has a newborn screening program
 - ▶ All are independent from each other
 - ▶ Not all states screen for all conditions
- ▶ 2023 celebrated 60 years of newborn screening
- ▶ Newborn Screening started by Dr Robert Guthrie
 - ▶ A parent trying to find the cause of his sons cognitive impairment
 - ▶ He was as physician and microbiologist
 - ▶ Developed an inhibition assay to test for PKU
- ▶ Developed the filter paper test using the same inhibition assay only spotted on the filter paper
 - ▶ Tested his theory by testing residents at a state institution and identified 23 of the residents had untreated PKU

Expansion of the Guthrie Test

- ▶ Connected with 29 states that were interested in piloting this test on newborns
- ▶ Used a sheltered workshop to assemble the test kits
- ▶ Within 2 years 400,000 infants tested and 39 cases diagnosed
- ▶ By 1966 newborn screening (for PKU) was mandatory in most states



Advancements in Testing and Additional Conditions

- ▶ 1980's started looking at mutation analysis
- ▶ 1990's development of Tandem Mass Spectroscopy
 - ▶ Made the inhibition test obsolete
 - ▶ High throughput method
 - ▶ Dr Guthrie Died in 1995
- ▶ Universal Newborn Hearing Screening
 - ▶ In 1999 federal legislation passed to begin funding
 - ▶ Prior to this, less than 10% of all newborns received a hearing screening
 - ▶ 47% of children were not diagnosed until after their 3rd birthday

Arizona's Screening Panel



- ▶ 35 of 37 Conditions

- ▶ 33 Dried Bloodspot Tests

- ▶ 2 Point of Care tests

- ▶ Hearing

- ▶ CCHD

- ▶ RUSP aligned state- will add any new condition within 2 years of addition to the RUSP

- Every baby has 2 bloodspot screens completed (24 hrs of life and again between 5-10 days of age)

Components of NBS System

- ▶ Point of Care tests at Hospital
- ▶ Laboratory
- ▶ Demographics
- ▶ Education
- ▶ Follow Up
 - ▶ Short Term follow up
 - ▶ Dried Bloodspot and Hearing
 - ▶ Long Term Follow Up
 - ▶ All NBS conditions
- ▶ Newborn Screening Video



NBS Follow Up Collaboration

- ▶ Collaboration helps with locating babies needing follow up services, providing notifications to providers and sharing valuable data system resources.
 - ▶ Follow Up Specialists assigned to specific disorders
 - ▶ Metabolic, Hearing, Endocrine, LSD's, Enzyme, Pulmonary, Blood, others
 - ▶ Shared access to all data systems
 - ▶ Ability to locate information
 - ▶ Cross training
 - ▶ Team collaboration

Sharing Similar Workflows and Processes

- ▶ Hearing and DBS follow up have very similar workflows
 - ▶ Review screening results
 - ▶ Locate PCP and report results and request referral
 - ▶ Fax instructions to PCP
 - ▶ Contact parents to review and assist family with next steps (unique to hearing in AZ)
 - ▶ Follow up on status of diagnostic test results
 - ▶ Referral to Parent support and Early Intervention if needed
 - ▶ Will use this process established for hearing to build for bloodspot long term follow up



Universally Shared Data Systems

- ▶ Hearing and Bloodspot have different data systems that work hand in hand
 - ▶ Hi-Track and Neometrics(NATUS)
 - ▶ All are referenced regardless of condition
 - ▶ Hearing data system has Vital records merge
 - ▶ Bloodspot system has follow up PCP information
 - ▶ Notes in one system may help a follow up specialist in another system
 - ▶ Health Information Exchange
 - ▶ Both Hearing and Bloodspot use this resource
 - ▶ Able to access all health records
 - ▶ Updated demographic contact information
 - ▶ Able to see if seen in specialty clinics like audiology or Diagnostic labs for DBS
 - ▶ Immunizations data system- ASIIS
 - ▶ Able to utilize to locate PCP information
 - ▶ Dried bloodspot follow up and hearing follow up are more alike than different

Take Home Ideas

- ▶ If not already connected, reach out to your states bloodspot team
 - ▶ Schedule regular meetings
 - ▶ share ideas
- ▶ Discuss ways to collaborate on follow up initiatives
- ▶ Share resources
 - ▶ Phone lists
 - ▶ processes
- ▶ Explore sharing access to data systems
- ▶ Remember.....

We are all newborn screening!

Questions?



THANK YOU

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