# EHDI and Dried Bloodspot: We are all Newborn Screening!

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Health and Wellness for all Arizonans



#### ► 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 3<sup>rd</sup> birthday

#### Arizona's Screening Panel

#### ► 35 of 37 Conditions

- ► 33 Dried Bloodspot Tests
- 2 Point of Care tests
  - ► Hearing

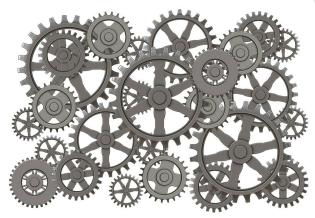
► CCHD

- 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

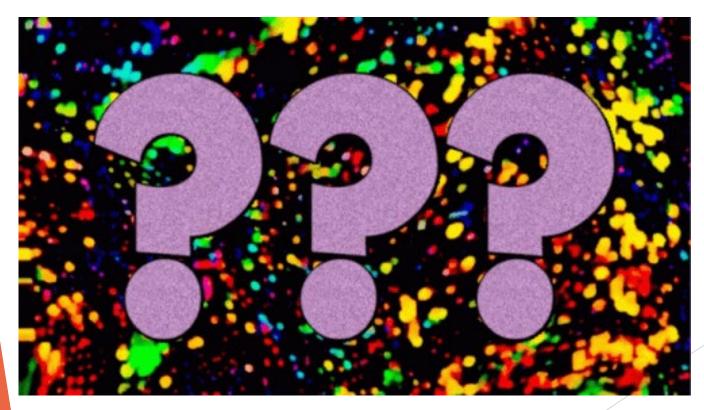
#### Take Home Ideas

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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