



# Newborn Screening Programs

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# Introduction

Newborn screening is a critical public health service, identifying conditions that can affect a child's long-term health.

Early detection, diagnosis, and interventions facilitated by newborn screening and follow-up services can prevent death or disability and assist children to reach their full potential.

- Heelstick
- Hearing
- CCHD

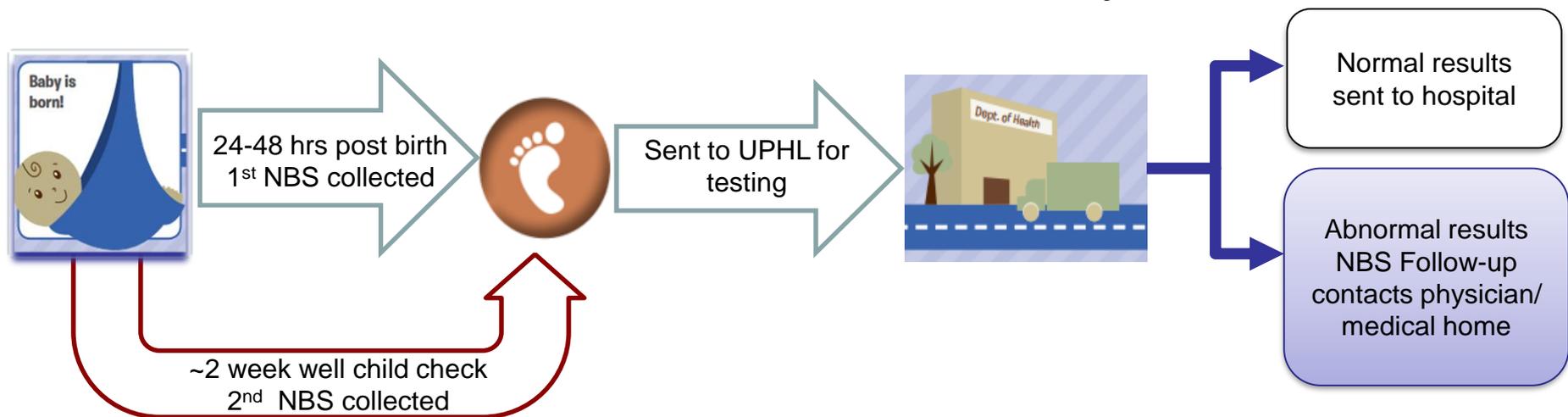


# Heelstick: Story of Baby Max



# Heelstick Testing Process

- Two screen method
  - First obtained between 24 – 48 hours age
  - Second obtained between 7 and 28 days



# The Heelstick Team



- **Laboratory**
  - Performs testing on blood samples
  - Reports normal results to the baby's doctor
  - Reports "abnormal" results to the Follow-Up staff
- **Follow-up**
  - Provides education regarding collection, timing and transportation of samples
  - Works with families and doctors to coordinate confirmatory testing
  - Works with hospitals and pediatrician's office to recollect samples that are unable to be tested

# Federal Recommendation Process



- Recommended Uniform Screening Panel (RUSP)
- Health & Human Services Advisory Committee on Heritable Disorders in Newborns and Children
  - Advises the Secretary of Health and Human services on appropriate newborn screening tests, technologies, policies, guidelines and standards
  - Committee recommends every newborn screening program include the disorders on the RUSP
  - **RUSP currently includes 34 core disorders and 26 secondary disorders**

# LFA Recommendations



## Kit Fee

- This year, the kit fee analysis will address UDOH indirect costs.
- The fee analysis will also take into account the addition of new disorders.

## Advisory Committee

- The committee recommends the addition of disorders to the Executive Director after review of 18 criteria such as treatment and laboratory testing.
- The committee is made up of experts in pediatrics, genetics, laboratory science, rare diseases, etc.

# Disorders Under Consideration

- Spinal Muscular Atrophy (SMA), type 1
  - **100% curability** if treated within 3 weeks of life
  - Should detect 5-6 cases per year
- X-Linked Adrenoleukodystrophy (X-ALD):
  - Treatment through Bone Marrow Transplant
  - Should detect 3 cases per year
- Mucopolysaccharidosis type I (MPSI)
  - Treatment through Bone Marrow Transplant or enzyme replacement
  - Should detect 1 case per year
- Pompe (Glycogen Storage disease, Type II)
  - Treatment through Bone Marrow Transplant
  - Should detect 2 case per year

# Initial Cost Estimates

## Preliminary cost analysis:

- SMA: Start screening July 2018 for **\$4/baby**
- X-ALD: Start screening July 2019 for **\$8/ baby**
- MPSI/Pompe: Start screening July 2019 for **\$8/baby**

Will finalize with full cost analysis

# Newborn Screening Programs



- Early Hearing Detection and Intervention (EHDI)
  - Provide newborn hearing screening oversight to assure access to early:
    - Screening
    - Identification
    - Intervention

- **Critical Congenital Heart Defects (CCHD)**
  - Safety net for all babies born in Utah
    - Educating health care providers,
    - Improving the screening process,
    - Statewide screening and data collection system
  - How
    - Increase pulse oximetry testing
    - Pulse oximetry screening reported on birth certificate
    - Infants failing shall have provider perform complete clinical evaluation, which may include a cardiac echocardiogram

Thank you

