

## The Newborn Screening System: A theoretical framework

Michele A. Lloyd-Puryear, MD, PhD  
Health Resources and Services  
Administration



## Newborn Screening

- *Each year, more than 4 million U.S. infants are screened to detect conditions that threaten their life and long-term health.*
- *Newborn screening programs are located in state public health agencies.*



## What is Newborn Screening?

- *An **essential public health program** that prevents catastrophic health consequences.*
- *A **complex system** of testing, evaluation, and treatment.*

## Environmental Context (U.S.)

- Nation with culturally diverse population
- Advances in science - Human Genome Project
- Advances in Information technology
- Public health agency roles changing-less direct services
- Federal-state division of responsibility
- Health care delivery and financing
  - More children using publicly-subsidized health coverage

## Principles for Developing Newborn Screening Programs

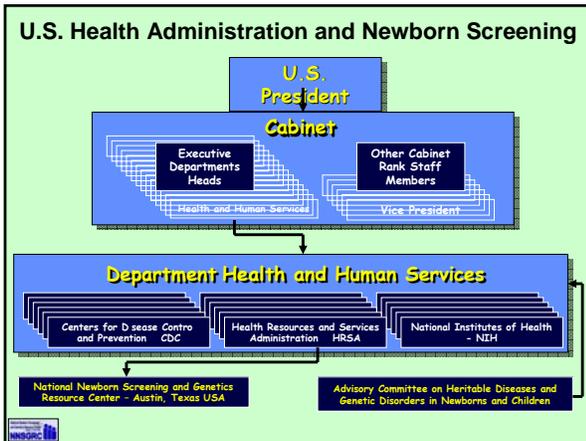
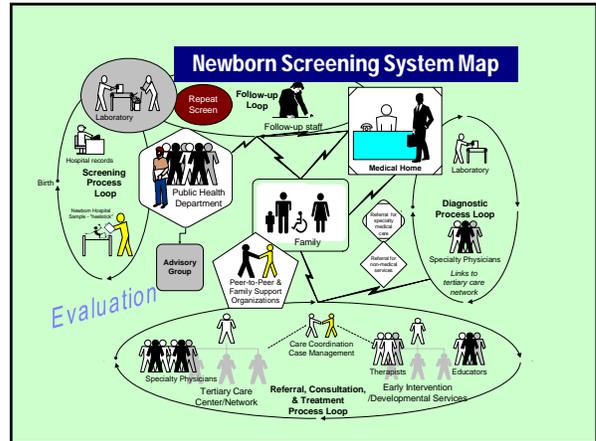
- Infants should benefit from/be protected by newborn screening programs
- Public health agencies should assume responsibility for oversight of newborn screening systems

## Principles for Developing Newborn Screening Programs

- Standards and guidelines for newborn screening should be consistently applied
- Uniformity benefits families, professionals, and public health agencies
- Newborn screening systems should link to a medical home

### Newborn Screening

- Family Members
- Primary Health Care Professionals
- Laboratory Personnel
- Administrative and Follow-up Personnel
- Specialty Care Centers
- Source(s) of Payment
- Policy Makers
- Manufacturers



### Components of Newborn Screening System

**Screening:**

- Newborn initial test
- Analysis of test
- Repeat, as necessary

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**Screening:**

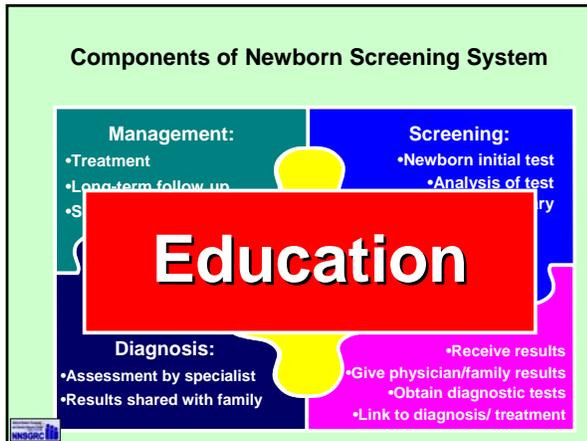
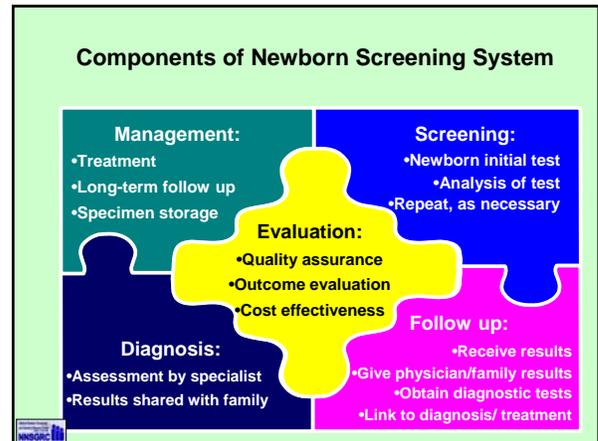
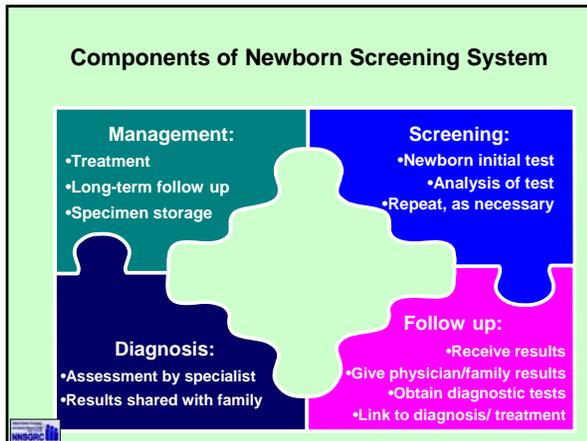
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**Diagnosis:**

- Assessment by specialist
- Results shared with family

**Follow up:**

- Receive results
- Give physician/family results
- Obtain diagnostic tests
- Link to diagnosis/ treatment



### Parent/Professional Education

**7 Things Parents Want to Know About Newborn Screening From Their Child's Health Professional**

**HRSA**

**Newborn Screening Disorders**

What parents want to know about newborn screening disorders.

- ### “Need-to-know” Information for Parents
- **All** babies are screened
  - Screening will **benefit** the baby
  - Testing is **safe**- not harmful
  - The baby may need to be **retested**
  - Parents will be **notified** if retesting is needed
  - Its important to **act quickly** if retesting is necessary
- Cost of screening and consent are not important*

### Parent/Professional Education

**Quick Reference to Newborn Screening Disorders**

**7 Things Parents Want to Know About Newborn Screening From Their Child's Health Professional**

1. All newborn babies are screened for the most common newborn screening disorders before they leave the hospital.
2. Babies with these disorders may look healthy at birth.
3. Not all newborn babies are screened for the most common newborn screening disorders.
4. To do the tests, a nurse will take a few drops of blood from your baby's heel.
5. Your baby's health professional will tell you how to get a copy of the test results. Call your health professional if you would like to talk about the results.
6. Some babies will need to be retested. If your baby needs to be retested, you will be notified. A nurse will explain to you what you need to do.
7. If you have further questions you can call the clinic or the State Health Department.

**Professional Education**  
**Basic Information for Physicians**

**7 Things Parents Want to Know About Newborn Screening from Their Child's Health Professional**

**The Health Professional's Guide for Brief Prenatal Discussion with Parents**

- 1) All newborn babies are required by the State to get tested for some rare disorders before they leave the hospital.
- 2) Babies with these disorders may look healthy at birth.
- 3) Serious problems can be prevented if we find out about the disorders right away.
- 4) To do the tests, a nurse will take a few drops of blood from your baby's heel.
- 5) Your baby's health professional and the hospital will get a copy of the test results. Call your baby's pediatrician if you would like to talk about the results.
- 6) Some babies will need to be retested. If your baby needs to be retested, you will be notified. It is very important to get retested quickly.
- 7) If you have further questions, you can call the clinic or the State Health Department.

**Professional Education**  
**Basic Information for Physicians**  
**ACMG**

**Newborn Screening ACT Sheet [Primary TSH test/Elevated TSH] Congenital Hypothyroidism**

*Differential Diagnosis:* Primary congenital hypothyroidism (CH), transient CH.

*Condition Description:* Lack of adequate thyroid hormone production.

*You Should Take the Following Actions:*

- Contact family IMMEDIATELY to inform them of the newborn screening test result.
- Consult pediatric endocrinologist, referred to endocrinologist if considered appropriate.
- Evaluate infant (see clinical considerations below).
- Initiate timely confirmatory/diagnostic testing as recommended by the specialist.
- Initiate treatment as recommended by consultant as soon as possible.
- Educate parents/caregivers that hormone replacement prevents mental retardation.
- Report findings to state newborn screening program.

*Diagnostic Evaluation:* Diagnostic tests should include serum free T4 and thyroid stimulating hormone (TSH); consultant may also recommend total T4 and T3 resin uptake. Test results include reduced free T4 and elevated TSH in primary hypothyroidism; if done, reduced total T4 and low or normal T3 resin uptake.

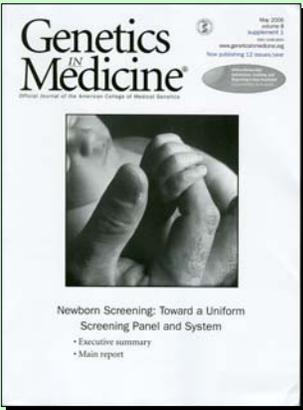
*Clinical Considerations:* Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy faces, large fontanelles, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or mental retardation and poor growth.

*Additional Information:*  
 Click on the name to take you to the website. Complete ERAs are listed in the Appendix.  
[New England Newborn Screening Program](#)  
[American Academy of Pediatrics](#)  
[Genetics Home Reference](#)

*Refer to local, state, regional and national:*  
[Laurie N. White Pediatric Endocrinology Society "Find a Doc"](#)  
 Contact local/regional University-affiliated medical center

**PRIMARY TSH TEST / ELEVATED TSH**  
**Congenital Hypothyroidism**

**ACMG Report on Newborn Screening**  
**May 2006**



**Genetics IN Medicine**  
 Official Journal of the American College of Medical Genetics

May 2006  
 Volume 8  
 Supplement 2  
 www.geneticsinmedicine.org  
 New publishing @ genetics.org

**Newborn Screening: Toward a Uniform Screening Panel and System**

- Executive summary
- Main report

**Contact Information**

Michele Lloyd-Puryear, M.D., PhD  
 Maternal and Child Health Bureau  
 Health Resources and Services Administration

[mpuryear@hrsa.gov](mailto:mpuryear@hrsa.gov)  
 301-443-1080



Newborn Screening is more than a test—it is a complex system of services

