

Newborn Screening Overview

Policy Roundtable

Cynthia M. Powell, MD

Associate Professor of Pediatrics and Genetics

Chief, Division of Pediatric Genetics and Metabolism

The University of North Carolina at Chapel Hill

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Newborn Screening

- Newborn Screening in the U.S. is a public health program aimed at the early identification of conditions for which early and timely intervention can prevent or reduce associated mortality and morbidity

Newborn Screening Task Force Report, Pediatrics 106: 383-427 (2000)



Untreated PKU

The Metabolic Basis of
Inherited Disease,
Stanbury et al . eds,
McGraw-Hill, 1966

Criteria for Effective Newborn Screening Programs

- Treatment is available.
- Early institution of treatment before symptoms manifest reduces or eliminates severity of illness.
- Routine observation or exam will not reveal disorder in newborn (a test is required).

Criteria for Effective Newborn Screening Programs

- A rapid and economical laboratory test is available that is highly sensitive (no false negatives) and reasonably specific (few false positives).
- The condition is frequent and serious enough to justify the expense of screening (screening is cost effective).
- The social infrastructure is in place to inform the newborn's parents and physicians of the results of the screening test, confirm the test results, and institute appropriate treatment and counseling.

NC Newborn Screen Tests

- 1965 - Phenylketonuria (PKU)**
- 1979 - Hypothyroidism**
- 1987 - Sickle cell disease (limited)**
- 1988 - Galactosemia**
- 1989 - Congenital adrenal hyperplasia**
- 1994 - Sickle cell disease and other hemoglobinopathies (universal)**

NC Newborn Screen Tests

**1997 - Tandem mass spectrometry pilot
(MS/MS)**

Amino acidopathies

Organic acidopathies

Fatty acid oxidation defects

MCAD deficiency

- Normal at birth
- Well when not fasting
- With fasting or illness
 - ◆ hypoglycemia
 - ◆ seizures
 - ◆ death
- Prior to screening and early diagnosis: 1/3 died with initial presentation, 1/3 left with severe neurological impairment (CP, MR)

NC Newborn Screen Tests

- 1999 - MS/MS newborn screening**
increasing the number of
disorders detectable by >30
- 2001 - Newborn hearing screening**
- 2004 - Biotinidase screening**

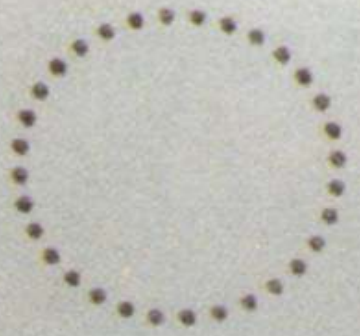
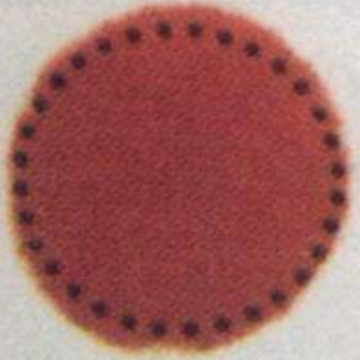
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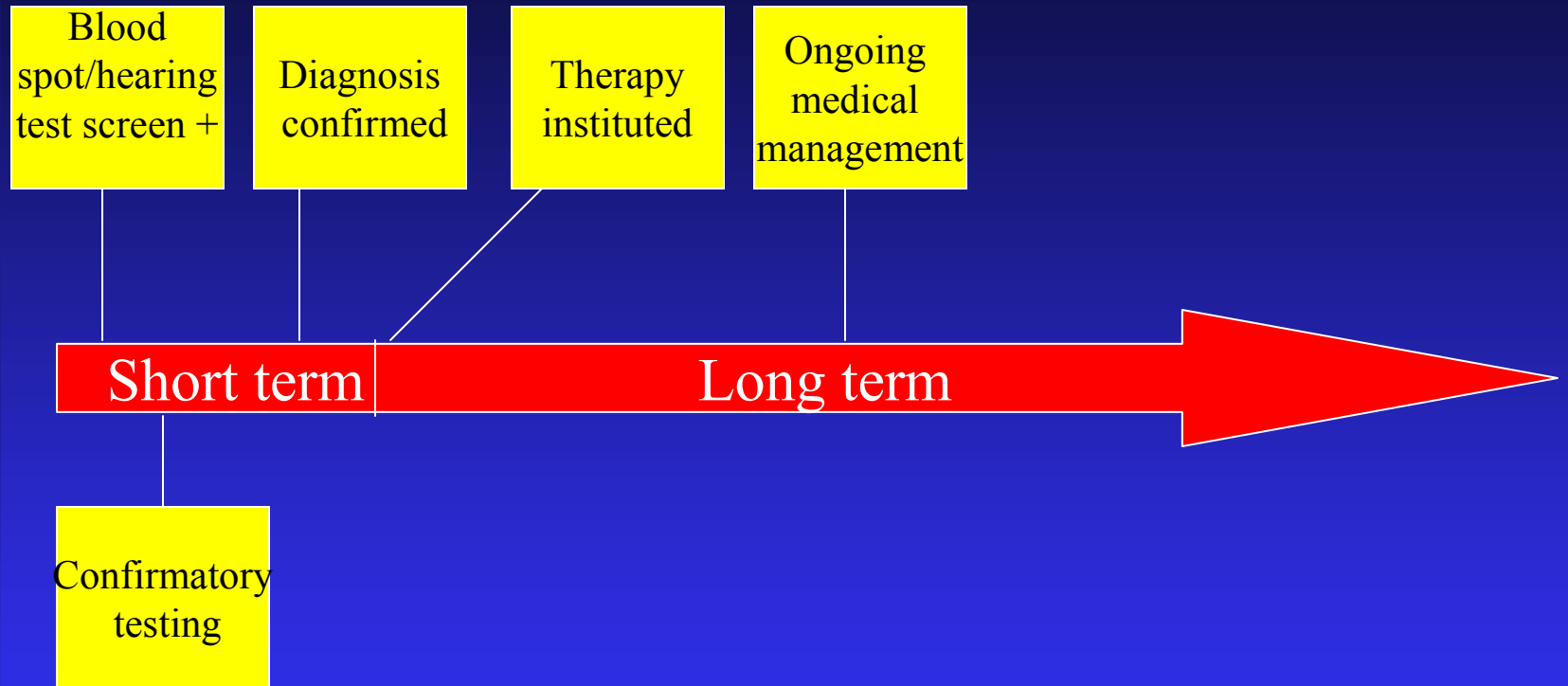
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Family Needs in Expanded Newborn Screening

- “The experience gained over the past 30 years with screening for PKU...shows that the period elapsing between the first alarm signal and the actual onset of care remains permanently engraved in the parents’ memory, and that blunders committed in word or deed will never be forgotten” Farriaux et al. (2003) J. Inherit. Metab. Dis 26:729-744

Follow-Up in Newborn Screening





Smith's Recognizable
Patterns of Human
Malformation, 4th ed. 1988
W.B. Saunders

Maternal PKU – mother with PKU not on diet during pregnancies – 3 children with maternal PKU effects including microcephaly, mental retardation and congenital heart defects

Family Needs in Expanded Newborn Screening

- Public health benefits and improved outcomes through NBS programs will only be realized if there are adequate resources for long-term follow-up care and management
- “Newborn screening is only as effective as the care it prompts” Susan Berry, MD, University of Minnesota

How do states pay for newborn screening and follow-up?

- Report by GAO in 2003: FY 2001 > \$120 million spent to screen 4 million newborns in U.S. (avg \$30/infant)
- Fees (66%)
- Medicaid (10%)
- Title V Maternal and Child Health Services Block Grants (5%)
- State general revenue funding (19%)

State	Fee	Program Components Covered by Fee
Alabama	\$139.33	Laboratory
Delaware	\$78.00	Lab, program admin/f-up, medical genetics consultant
Iowa	\$77.00	Lab, program admin/follow-up, medical consultants, metabolic formula, short-term and long-term follow-up, courier, developmental fund
North Carolina	\$14.00	Laboratory
Rhode Island	\$110.00	Lab, program admin/follow-up, specialty formulas
Wisconsin	\$69.50	Lab, program admin/follow-up, treatment, genetic counseling
Reference:	http://www2.uthscsa.edu/nnsis/	Data from the National Newborn Screening Information System

Comparison of Fees Charged for Newborn Screening in the U.S. in 2008

- 5 states charge no fee
- Of states charging fee: \$14.00 - \$139.33
- Program components covered by fee:
 - ◆ 10 states laboratory only
 - ◆ 31 states include program admin/follow-up
 - ◆ 17 states include treatment
 - ◆ 9 states include genetic counseling and genetic services
 - ◆ 6 states use fee to cover special formulas

State Handling of Newborn Screening Fees

- Fees deposited into general fund
 - ◆ Newborn screening program competes with other state programs for budget appropriation
- Fees deposited directly into special budget category and/or restrict use of fees to the support of genetic or newborn screening programs
 - ◆ Budgets linked to fee levels

Future directions

- Lysosomal storage diseases
 - ◆ Krabbe disease
 - ◆ 1:100,000 births
 - ◆ Infantile form – death by age 2
 - ◆ New York State: Newborn screening began in 2006
 - ◆ The only effective treatment is stem cell transplant

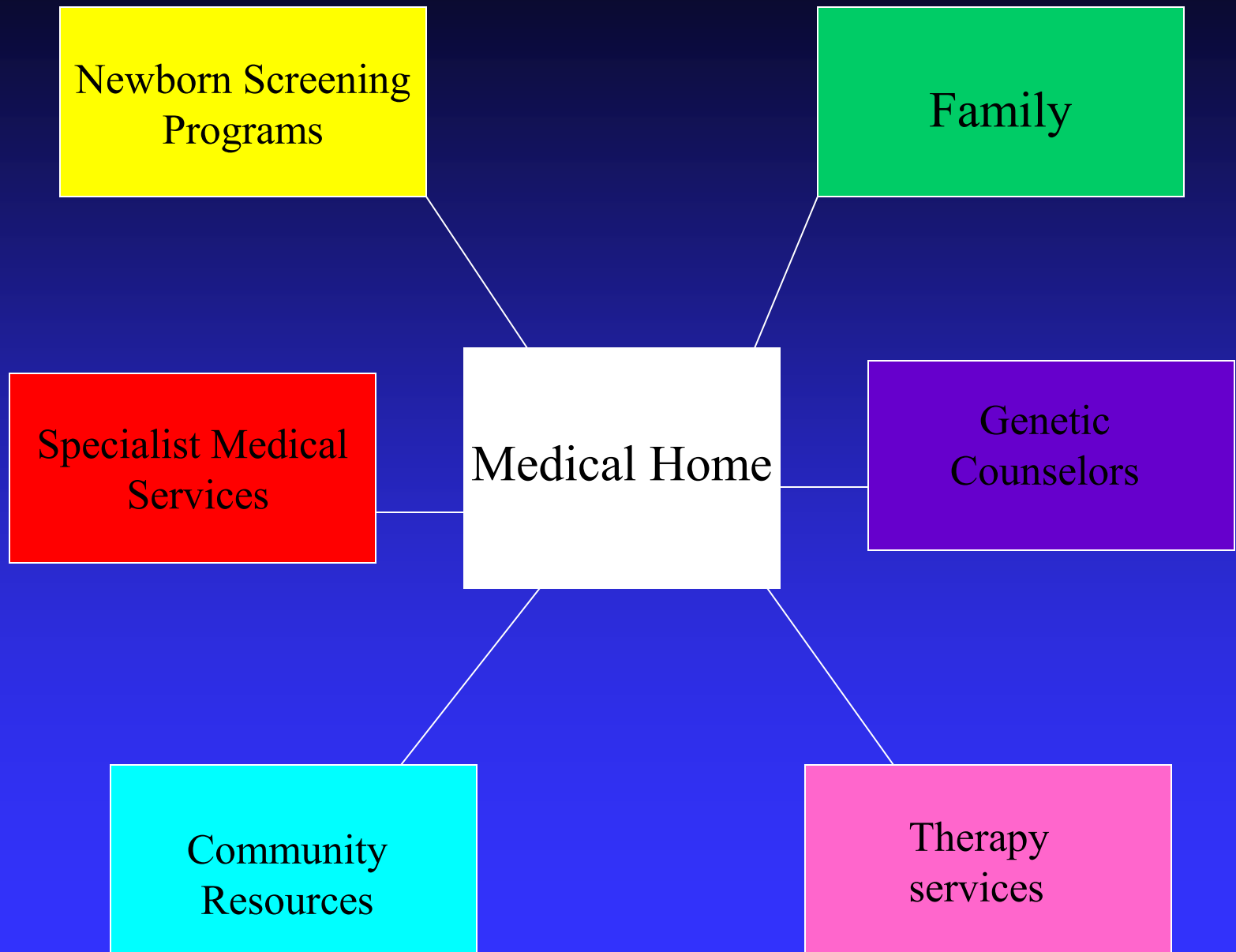


Future Directions

- Cystic fibrosis screening
- Additional conditions under consideration for adding to screening panels: SCID, lysosomal storage diseases, fragile X syndrome.
- Criteria are expanding (“treatable”)
- Ethical debates about identifying unaffected “carriers”, or conditions that may not present until adulthood

Needs in Expanded Newborn Screening

- Who will provide follow-up medical services and genetic counseling?
- Current needs
 - ◆ Clinical and biochemical geneticists
 - ◆ Pediatric subspecialists
 - ◆ Genetic counselors
 - ◆ Metabolic dieticians
 - ◆ Audiologists/Otolaryngologists



Needs in Expanded Newborn Screening

- Future needs
 - ◆ Stem cell transplant teams
 - ◆ Infusion centers for enzyme therapy
 - ◆ Gene therapy centers
 - ◆ Developmental therapists
- Consideration of health care resources
- Evidence-based analysis of cost-effectiveness, treatment efficacy, natural history
- Newborn Screening Translational Research Network Coordinating Center (funded through NICHD)

The Newborn Screening Saves Lives Act was signed into law by the President on April 24, 2008. This legislation establishes **grant programs**, which are to be awarded to eligible entities to **provide education in congenital, genetic, and metabolic disorders; and training in newborn screening technologies.** Grant programs are also to be used to **coordinate follow-up care.** In addition to grant programs, this bill increases consumer awareness and knowledge of family support services, research, and other resources in newborn screening; improves laboratory quality standards; develops a national contingency plan if a public health situation arises; and establishes a central online clearinghouse. Finally, the bill renews the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, and expands and coordinates research, particularly on conditions that could be added to the panel in the future. **The legislation authorizes \$44.5 million in fiscal 2008 to fund the bill's various programs, with the amount increasing each year through 2012.**