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Improving Children's Health through Newborn Screening

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Notable Date in Newborn Screening

- September 30, 2014

The 50th anniversary of the Children's Bureau recommendation for "the screening of all newborn infants for PKU [phenylketonuria] on a routine basis."

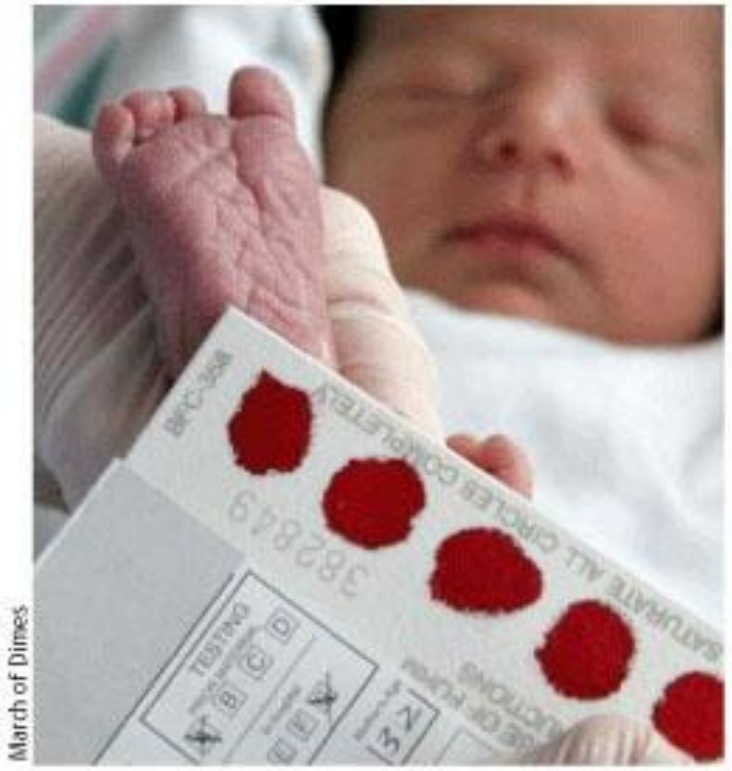
  - Celebrated by special symposia at AACC, ACMG, Newborn Screening Society and other annual meetings in July 2014

Newborn screening for metabolic, endocrine and other conditions

- A Public Health Program
- Aimed at identifying genetic conditions for which early intervention can prevent mortality, morbidity, and disabilities
- Performed, for the most part, by analysis of biochemical markers in blood spots collected on filter paper at birth
- Testing procedures have been designed by laboratorians to use the smallest possible blood sample
- Listed in top ten public health achievements by CDC

Background

- More than 4 million babies are born every year in the United States
- In the vast majority these are term pregnancy with no perinatal complications
- Newborn screening is the first laboratory blood test that most people encounter in their lives
- Awareness of parents is often limited, and most are unprepared to cope with an apparently abnormal result



Newborn screening and Early Intervention for PKU works

The HRSA/ACMG Uniform Panel (2006)

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executive summary

Michael S. Watson, PhD, Marie Y. Mann, MD, MPH, Michele A. Lloyd-Puryear, MD, PhD, Piero Rinaldo, MD, PhD, and R. Rodney Howell, MD, editors

The Maternal and Child Health Bureau commissioned the American College of Medical Genetics to outline a process for the standardization of outcomes and guidelines for state newborn screening programs and to define responsibilities for collecting and evaluating outcome data, including a recommended uniform panel of conditions to include in state newborn screening programs. The expert panel identified 29 conditions for which screening should be mandated. An additional 25 conditions were identified because they are part of the differential diagnosis of a condition in the core panel, they are clinically significant and revealed with screening technology but lack an efficacious treatment, or they represent incidental findings for which there is potential clinical significance. The process of identification is described, and recommendations are provided. ***Genet Med* 2006:8(5, Supplement): 1S–11S.**

Key Words: *Newborn screening, genetics, public health, congenital, metabolic disease*

Expansion of NBS Screening (2006)

- 29 primary conditions
  - 20 detected by MS/MS*
  - 3 Hemoglobinopathies (S/S, S/βThal, S/C)
  - 7 others (BIOT, CAH, CF, CH, GALT, HEAR)

- 25 secondary targets
  - 22 detected by MS/MS*
  - 1 Hemoglobinopathy (many variants counted as 1)
  - 2 others (GAL-epimerase, GAL-kinase)

*Driven by laboratory advances in mass spectrometry

Implemented in 99.8% of all US births today

Public Law 110-204
Newborn Screening Saves Lives Act of 2008

A BILL

To amend the Public Health Service Act to establish grant programs to provide for education and outreach on newborn screening and coordinated followup care once newborn screening has been conducted, to reauthorize programs under part A of title XI of such Act, and for other purposes.

Be it enacted by the Senate and House of Representatives of the United States of America in Congress assembled,

SECTION 1. SHORT TITLE.

This Act may be cited as the "Newborn Screening Saves Lives Act of 2008".

SEC. 2. IMPROVED NEWBORN AND CHILD SCREENING FOR HERITABLE DISORDER.

Public Law 110-204
Newborn Screening Saves Lives Act of 2008

- This statute amends the Public Health Service Act to facilitate the creation of federal guidelines on newborn screening

  −To assist state newborn screening programs in meeting federal guidance

  − To establish grant programs to provide for education and outreach on newborn screening and follow-up care once newborn screening has been conducted

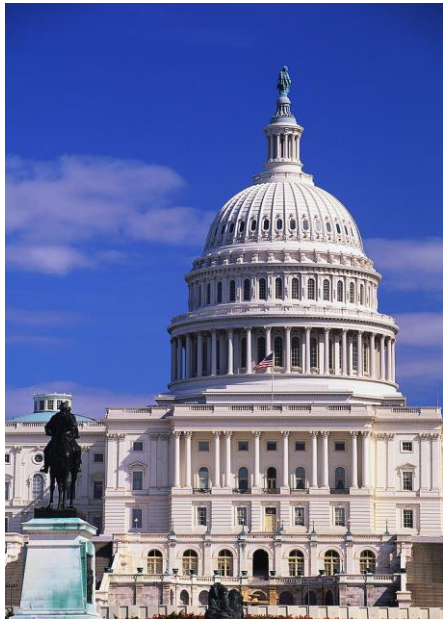
  − To reauthorize programs under Part A of Title XI of the Act

Uniform Screening Panel (2014)

- 31 primary conditions
  - 20 detected by MS/MS (AA, FAO, OA)
  - 3 Hemoglobinopathies (S/S, S/βThal, S/C)
  - 9 others (BIOT, CAH, CF, CH, GALT, HEAR, SCID, CCHD)

- 26 secondary targets
  - 22 detected by MS/MS (AA, FAO, OA)
  - 1 Hemoglobinopathy (many variants counted as 1)
  - 3 others (GAL-epimerase, GAL-kinase, other T-cell def.)

Progress



- The United States has achieved one of the most advanced and uniform implementation of newborn screening programs worldwide

- Approximately 10,000 newborns every year are spared severe morbidity and mortality, and related health care costs associated with late diagnosis

- Federal agencies (CDC), States, academic institutions, professional organizations including AACC, and the private sector are developing partnerships to develop, validate, and implement new tests to diagnose other conditions as new effective therapies become available

Continuing Challenges

- Still discrepancies in number of screening tests between states
- Approximately 1,000 newborns face death or disability that could have been prevented with absolute uniformity
- Reduce the timeline for reviewing new conditions, delays result in missed opportunities
- Improve follow-up care for newborns testing positive for condition detected through newborn screening. Not all follow up is uniform despite guidelines from ACMG and AACC (through NACB)

Congressional Response

- Representatives Lucille Roybal Allard (D-CA) & Rep. Mike Simpson (R-ID) introduced H.R. 1281, the Newborn Screening Saves Lives Reauthorization Act, in the House

- Senators Kay Hagan (D-NC) & Senator Orrin Hatch (R-UT), introduced companion legislation, S. 1417 in the Senate

Newborn Screening Bill Would:

- Reauthorize HRSA grants to expand and improve screening programs, educate parents and health care providers
- Improve follow-up care for infants with a condition detected through newborn screening
- Reauthorize federal advisory program to consider new conditions
- Reauthorize CDC quality assurance program

Newborn Screening Bill Would Also:

- Provide additional money for developing new treatments though NIH Hunter Kelly Newborn Screening Program

- Shorten the time for adding new conditions to the Recommended Uniform Screening Panel

Current Status

- Senate passed S.1417 in January
- House passed amended H.R.1281 in June
- Awaiting final Senate approval of compromise legislation

Call for Action

- Have your Members contact House and Senate leaders and urge them finalize this critical life-saving legislation when Congress returns after the elections

Thank you for your attention