WHAT’S NEW IN NEWBORN SCREENING?

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What is Newborn Screening?

- Newborn screening is a simple blood test used to identify many life-threatening genetic illnesses before any symptoms begin.
- An essential public health program that prevents catastrophic health consequences through early detection, diagnosis and treatment.
- A complex system of testing, evaluation, and treatment that is dependent upon the dedication of persons working within the system.
History of Newborn Screening

• Guthrie developed filter paper test for PKU.
• Identified newborns with PKU whose diet could be modified thus preventing mental retardation.
PHENYLKETONURIA
IN
PUBLIC HEALTH LAW

§ 2500 a

"It shall be the duty of (1) the administrative officer or other person in charge of each institution caring for infants twenty-eight days or less of age and (2) the person required ... to register the birth of a child, to cause to have administered to every such infant or child in its or his care a test for phenylketonuria in accordance with rules or regulations prescribed by the commissioner. ...

§ 2. This act shall take effect January first, nineteen hundred sixty-five."
History of Newborn Screening

1970’s
- Other filter paper tests became available.
  - (e.g. CH, CAH and Sickle cell)
- Technology improved, allowing program expansion

1980’s
- Programs became computerized.
- Expansion continued, including DNA studies.

1990’s
- DNA tests used as second tier.
- Application of tandem mass spectrometry (MS/MS) to allow simultaneous detection of multiple disorders.

2000’s
- DNA tests used for primary screening (SCID)
GAO Report on Newborn Screening Programs prepared at the request of Sen. Dodd (CT) and Sen. DeWine (OH)
Newborn Screening Program
Differences Noted in GAO Report

- 51 States (incl. D.C.) had laws allowing or mandating NBS.
- Three programs required consent for NBS.
- Some did not allow dissent for any reason.
- Disparity in conditions screened.
  - Ranged from 4 to 36 with avg. of 8 conditions
- Eight programs did not charge a fee.
- The amount of Medicaid reimbursement varied widely with about one-third of all births Medicaid.
Recommendations

• Mandate screening for all core panel conditions.
• Mandate reporting of all secondary target conditions and reporting of any abnormal results.
• Maximize the use of multiplex technologies.
• Consider the range of benefits realized by newborn screening beyond infant’s mortality and morbidity.
• Served as the impetus to quickly expand NBS nationally.
Newborn Screening: Toward a Uniform Screening Panel and System

Contents

EXECUTIVE SUMMARY .................................................................................................................. 1s
MAIN REPORT ............................................................................................................................... 12s
ABSTRACT ................................................................................................................................. 12s
INTRODUCTION ......................................................................................................................... 14s

SECTION 1
Developing a Uniform Screening Panel
Advises the Secretary of Health and Human Services regarding the most appropriate application of universal newborn screening tests, technologies, policies, guidelines and standards for effectively reducing morbidity and mortality in newborns and children having, or at risk for, heritable disorders.
Newborn Biochemical Screening in New Jersey

Newborn Screening System
Key Components of Newborn Screening

• Education (throughout the process)
• Screening, including specimen collection and testing
• Follow-up and result reporting
• Diagnostic confirmation
• Management
• Program evaluation and CQI
Texas Newborn Screening Laboratory

8 plates are distributed to 5 areas to test for 29 disorders.

Hemoglobinopathy Screening:
One test is used to identify:
Sickle Cell Anemia
Sickle Hemoglobin C Disease
Sickle/Beta Thalassemia Disease
Other hemoglobinopathy diseases and traits

Galactosemia & Biotinidase Screening:
Two tests are used to identify:
Galactosemia
Biotinidase Deficiency

Endocrine & Cystic Fibrosis Screening:
Three tests are used to identify:
Congenital Hypothyroidism
Congenital Adrenal Hyperplasia
Cystic Fibrosis

SCID Screening:
One molecular test is used to identify:
Severe Combined Immunodeficiency

Tandem Mass Spectrometry Screening:
One test is used to identify:
6 amino acid disorders (e.g. PKU)
5 fatty acid disorders (e.g. MCAD)
9 organic acid disorders (e.g. glutaric acidemia type 1)
Sample Follow-Up Flow

Abnormal Report Received via Lab Merge
- Letters generated to PMD and Mom
- Info verified, chart opened, mail sent
- Follow up in 3 weeks

Retest Received?

YES

Retest Normal
- Case Closed

NO

Retest Repeat Borderline
- BD or CH only
- Follow Double Borderline Category

Retest Presumptive
- CAH or CF any abnormal result
- BD or CH presumptive level result
- Follow Presumptive Category

NO

Retest Received?

NO

No Retest Received
- Send certified reminders to PMD and Mom
- Follow up in 3 weeks

Cert Card Signed by Mom and in Chart
- Case Closed

Cert Card not in Chart
- Send final certified letters to PMD and Mom
- Case Closed
<table>
<thead>
<tr>
<th>Year Screening Began</th>
<th>Program to Date Data from 1994 to December: 2010</th>
<th># of Babies with Confirmed Classic Disease</th>
<th># of Babies with Variant Disease or Carrier Status</th>
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Challenges
CHAPTER 175

AN ACT concerning screening for certain disorders in newborn infants, designated as Emma’s Law, and supplementing Title 26 of the Revised Statutes.

BE IT ENACTED by the Senate and General Assembly of the State of New Jersey:

C.26:2-111.5 Testing of newborns for certain lysosomal storage disorders required.

1. a. All infants born in this State shall be tested for the lysosomal storage disorders known as Krabbe, Pompe, Gaucher, Fabry, and Niemann-Pick diseases within six months following the occurrence of all of the following:
Newborn Screening and Genetics Program at APHL
QUESTIONS