Newborn Screening

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Outline

- Introduction
- Principles of newborn screening
- Logisitics of newborn screening program
- The basics of inborn errors of metabolism
- Review of metabolic disorders
- Case study
- The good, bad, and ugly of newborn screening

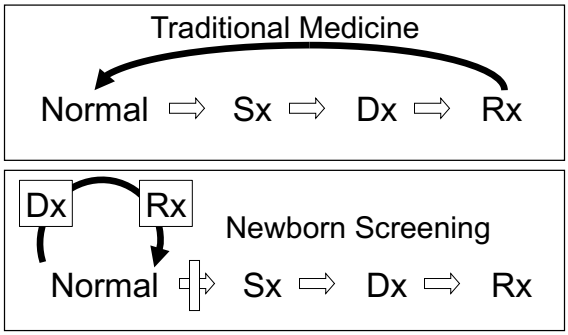
My Normal Spiel

- Who am I?
- Why do you think you are here?
- Why you are here
- Who gets a newborn screen (NBS)?
- Why do we NBS?
- When/how is NBS done?
- ***"Is my baby okay?"***

Impact of NBS on Families

- Fear and anxiety
- Waiting time for test results
- Genetic counseling

Newborn Screening vs. Traditional Medicine



Goals of Newborn Screening (NBS)

- Diagnose metabolic disorders before acute symptoms appear, thereby decreasing damage to the infant and its development
- Distinguish individuals who probably have treatable disorder from those who do not have the disorder ("screening")
- Educate the health care team and parents in the diagnosis and management of these disorders

Limitations of Screening Program

- Screening tests are not diagnostic
  - False positives
  - Screen for metabolites, not genes
- In many cases, the full clinical spectrum of disease is unknown
- Cannot always differentiate milder variants from severe variants based on biochemical parameters
- Does not diagnose all metabolic diseases or other genetic diseases

Selection Criteria for Disorders Screened in Program

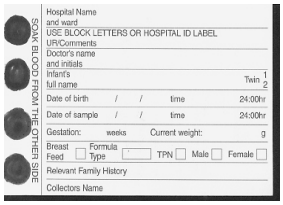
- Relatively "high" incidence
- If untreated, severe morbidity or mortality
- Treatment available
- Treatment offers clear and immediate benefit to affected individual
- Good assay
  - Accurate (sensitive, specific, high positive predictive value)
  - Reproducible
  - Inexpensive
  - Cost effective (per positive result, per diagnosis)

History of NBS in the United States

- 1959 Guthrie's filter paper test for PKU
- 1961: Voluntary Screening
- Parents pressed for testing through organized lobbying
- 1963: Legislatures began to pass mandatory NBS laws

History of NBS in the United States

- 1970s: Other tests became available on filter paper
- 1980s-1990s: added higher incidence disorders (CH) & disorders that result in death if untreated (CAH, galactosemia)



The Evolution of TN NBS

- January 2004: MS/MS screening begins for MCADD, MSUD, and homocystinuria
- April 1, 2004: MS/MS screening for additional 14 biochemical markers and 28 disorders begins
- Summer 2004: analytes for all 57+ disorders reported
- Spring 2008: Cystic Fibrosis (CF) screening begins by immunoreactive trypsinogen (IRT)
- January 1, 2013: Critical Congenital Heart Disease
- 2016: Severe Combined Immunodeficiency (SCID)
- 2017: Lysosomal Storage Disorders (LSDs)
- 2018: X-Linked Adrenoleukodystrophy (X-ALD)

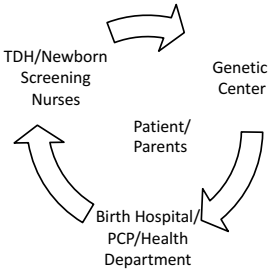
Current Disorders Screened

- Galactosemia
- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- Hemoglobinopathies
- Cystic Fibrosis
- Severe Combined Immunodeficiency (SCID)
- Organic Acid Disorders
- Fatty Acid Oxidation Disorders
- Amino Acid Disorders
- Biotinidase Deficiency
- Critical Congenital Heart Disease
- Lysosomal Storage Disorders
- Hearing Screening

Process of Screening

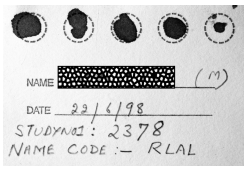
- Birth hospital collects specimen
- Tennessee Department of Health (TDH) Lab analyzes specimen, orders repeat for unsatisfactory specimens
- TDH Newborn Screening nurse reports abnormal to genetic center and PCP/Health Department by phone and fax; normal to PCP only by mail – computer system to check results
- Genetic center contacts PCP/Health Department regarding follow-up: see pt, repeat screen, send pt to VUMC
- Multiple interactions between all these groups at any time
- Genetics always happy to call parents, providers at any time

Circle of Care



Sample Collection

- Heelstick after 24 hours of age but before 7 days, within 48 hours if possible
- Specific process requiring community education
- 5 drops of blood can test for 60+ disorders!



Selected Errors in Sample Collection

- Cards not filled out properly and completely
- Contamination of blood spots
- Inadequate blood on spots
- Cards not air dried
- Not delivered to laboratory promptly
- Obtained too early (<24 hours)

Reports By State

https://www.newsteps.org/

- **Screened Conditions Report** - Report of screened condition counts
- **Conditions By Query Report** - Query for screened condition details
- **NBS Fees Report** - Provides information on the NBS fees each state NBS program is charging
- **DBS Retention Report** - Provides information on the dried blood spot specimen storage /retention times and storage conditions for each state NBS program
- **Courier System Report** - Provides information on the Courier system each state NBS program is using
- **LIMS System Report** - Provides information on the LIMS system each state NBS program is using
- **Data Retention Report** - Provides information on NBS data retention periods for each state NBS program

Inborn Errors of Metabolism / Metabolic Disorders

Normal Metabolism

Substrate → Product

↑

Enzyme

Inborn Errors of Metabolism

Substrate ▭ ⇉ Product

Inborn Errors of Metabolism

Substrate ▭ ⇉ Product

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