Newborn Screening: Tennessee Experience

Webinar
Why Does Everything in the Universe Rotate?
Pheromones: Profoundly Mysterious  Dust Devils

Discover
JULY 2003
SCIENCE, TECHNOLOGY, AND MEDICINE

Now the Genetic Testing Really Begins
It Starts With a Single Drop of Blood Taken From Each Newborn

And Ends When Scientists Predict Everyone's Physical and Mental Future

Human red blood cells. Magnification: 15,600x
NBS
50 Years of NBS

• European effort
  – Method to detect PKU (1934, Folling)
  – Diet to treat PKU (1953, Bickle)
• 1961 - Simple, inexpensive NBS for PKU, Dr. Robert Guthrie (Buffalo)
• 1963 - Mass screening methods for NBS, Dr. Mary Efron (Massachusetts General)
• 1973 - Screening method for congenital hypothyroidism, Dr. Jean Dussault (Canada)
• 1973 - Developed screening for sickle cell/hemoglobin disorders, Dr Michael Garrick (Buffalo)
• 1977 - Developed screening for CAH, Drs. Songya Pang and Maria New
• 1982 – Colorado, first state to screen for cystic fibrosis
• 1984 - Developed screening for biotinidase deficiency Dr. Barry Wolf (Richmond)
• 1990 – 1993 - Tandem mass spectrometry methods for NBS, Drs. Donald Chace, David Millington, and Ed Naylor
• 1990 – Hawaii first state to enact uniform newborn hearing screening
• 2005 – SACHDNC endorses uniform screening panel (29 conditions)
• 2010 – SACHDNC endorses addition of SCID to universal newborn screening panel
• 2012 – Critical congenital heart disease - addition to uniform screening panel

Basic scheme of an MS/MS analysis

3 mm Blood Spot

Deproteinization

Derivatization

Tandem Mass Spectrometer

Ion source

Collision cell

MS$_1$

MS$_2$

Detector

Interpretation

Mass spectrum

Computer
NEWBORN SCREENING
NBS
Tennessee NBS

• 1968 - PKU

• 1980 - Hypothyroidism

• 1988 - Hemoglobinopathies

• 1992 – Galactosemia

• 2001 - Hearing (voluntary, 2008 universal)

• 2003 - Biotinidase deficiency

• 2004 - TMS based disorders (amino acids, organic acids, fatty acid oxidation (all primary targets, >95% secondary targets)

• 2008 - Cystic fibrosis (IRT)

• 2013 - CCHD; (SCID pending)
NBS
Tennessee NBS

• Demographic information
  – Population – 6,346,000
  – Long state bordered by 8 other states
  – 8 Metro areas (pop. >100K) have ~80% pop
  – 95 counties
  – ~85,000 birth rate
  – Racial/ethnic component
    • Caucasians - ~76%
    • African-Americans - ~21%
    • Oriental, Pacific-Islanders - ~3%
    • (Hispanic - ~9%)
NBS
Tennessee NBS

• Department of Health
  – Laboratory Services
    • Newborn Screening
    • Others (Microbiology, Serology/Viral, Environmental, etc)
  – Maternal and Child Health Programs
    • Programs for
      – Infants Children and Adolescents
        » Newborn Screening
          • Follow-up Program
          • Genetics Advisory Committee
        » Newborn Hearing Screening
        » Children’s Special Services
        » Others
  – Women’s Health
  – Improving Services to Women and Children
NBS
Oversight

• Genetics Advisory Committee (GAC)
  – Advisory to Genetics and NBS section of TN Maternal and Child Health, and State Newborn Screening Lab (2 sessions/year)
  – Established in 1970’s
  – Representation from
    • Comprehensive Genetic Centers/Satellite Genetic Centers
    • Hematology/Endocrine/Pulmonary/Pediatric Cardiology Centers
    • Consumers
Let’s Do It Right the First Time
For Newborn Babies and Their Families
a program for the collection of bloodspots
for newborn screening

The National Laboratory Training Network (NLTN) is a training system sponsored by the Association of Public Health Laboratories (APHL) and the Centers for Disease Control and Prevention (CDC).
NBS
GAC Function

• Advise on policies concerning
  • Delivery of genetic services statewide
  • Education about genetic disorders/NBS
    – Health Care Providers
    – Consumers
  • Outreach to communities
  • Maintaining data on genetic disorders, and
  • NBS panel components
NBS
Tennessee NBS

• TN – NBS web site
  – [http://health.state.tn.us/MCH/NBS.shtml#1](http://health.state.tn.us/MCH/NBS.shtml#1)

• Site has:
  – Parent information (pamphlet link)
  – Provider information (pamphlet link)
  – Disorder information (separate for parents/providers)
  – Information for hospitals/providers
    • Blood collection
    • Hearing screening
    • CCHD screening
    • Unsatisfactory rate of specimens received/hospital
  – Secure Remote Viewer for providers (on-line)
  – Quarterly newsletter
  – Resource links (e.g. National Newborn Screening and Gentsics Resource Center, MOD, ACMG ACT Sheets, AAP, etc)
NBS

TN NBS – Unsatisfactory Specimens

• Unsatisfactory rate issues
  – Adopted goal of < or = 2% of submitted specimens
  – Source of:
    • Possible missed diagnoses
    • Delay in diagnosis
    • Additional expense

  – ‘Unsatisfactory’ definition
    • Currently 20 reasons for rejection/non-reporting of specimens
    • All specimens are run if possible
    • Report may only say “Unsatisfactory specimen” and reason, but no official report
    • Recommendation is to ‘Repeat’

• Sources of unsatisfactory specimens
  • Hospitals
  • Health Departments
  • Primary Providers
NBS

Tennessee NBS – Unsatisfactory Specimens

• State attempts to remedy by:
  – Provide educational opportunities
    • Working on hospitals with largest # unsats/and/or largest %
    • Positive reinforcement to hospitals/institutions meeting goal
    • Instructional DVD
    • Inform hospital of unsat rates and how compares to others
    • On-site education

• Shrinking funds to provide on-site education

• County Health Departments/providers offices challenged by lower numbers

• Genetic/specialty centers encouraged to provide additional expertise/education to
  – Hospitals
  – Health departments
  – Providers
NBS
Tennessee NBS – Unsatisfactory Specimens

• Therefore
  – Need for further educational efforts
  – Nurses primary patient caretaker in
    • hospitals
    • public health departments, and
    • provider sites
  – Nurses academic training gets less information about
    • NBS
    • Advances in NBS
    • Importance of NBS disorders