

Newborn Screening: Tennessee Experience

Webinar

Why Does Everything in the Universe Rotate?

Pheromones: Profoundly Mysterious

Dust Devils

VOL. 24, NO. 7

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: 19,600x

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#DCR0063198737/3H23 FIB04
JIM A BROOKS 507034
P224
431 STONEWALL ST 0010
MEMPHIS TN 38112-5115



USA \$4.99

CAN \$6.50

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

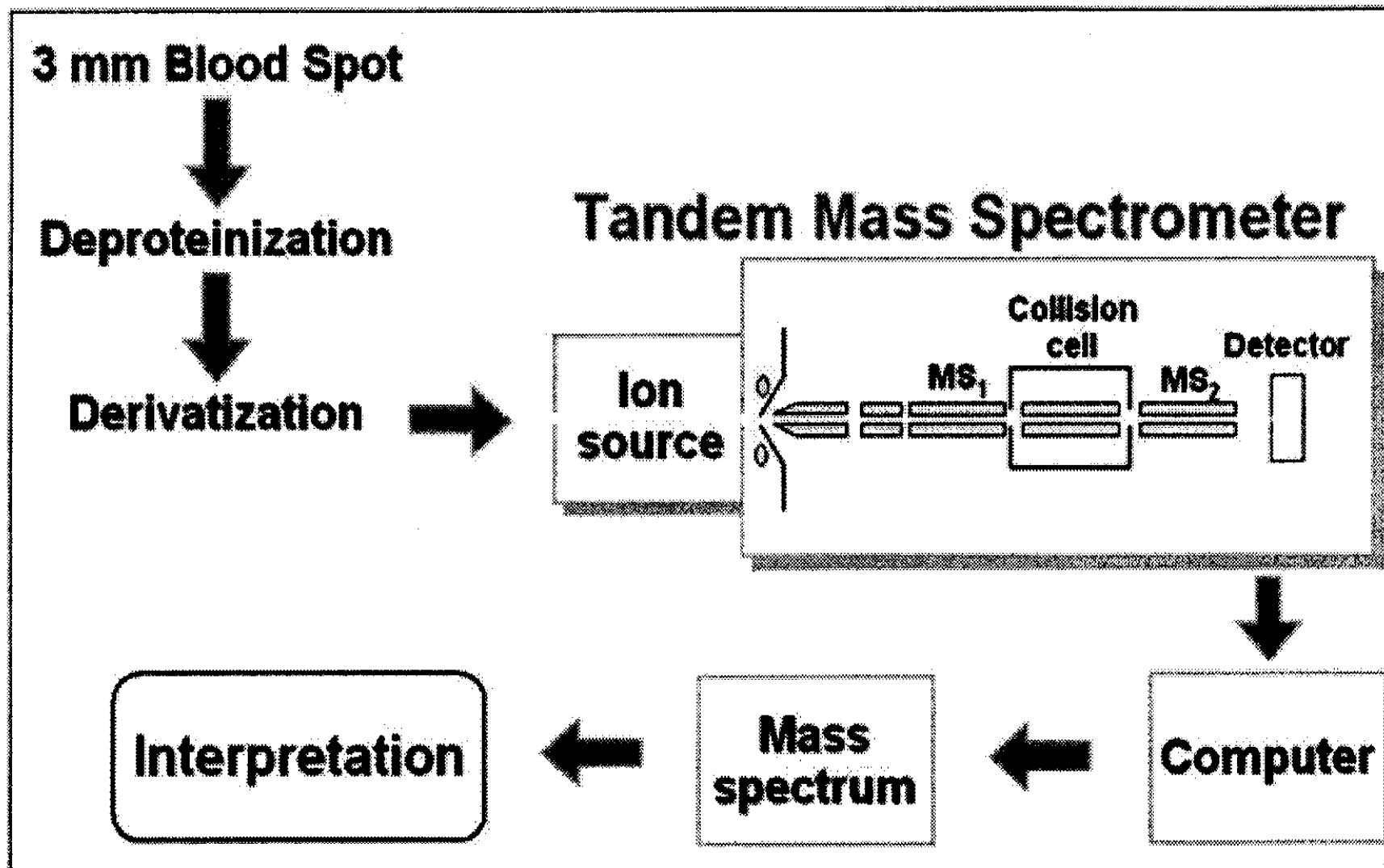
- 1982 – Colorado, first state to screen for cystic fibrosis
- 1984 - Developed screening for biotinidase deficiency Dr. Barry Wolf (Richmond)

- 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

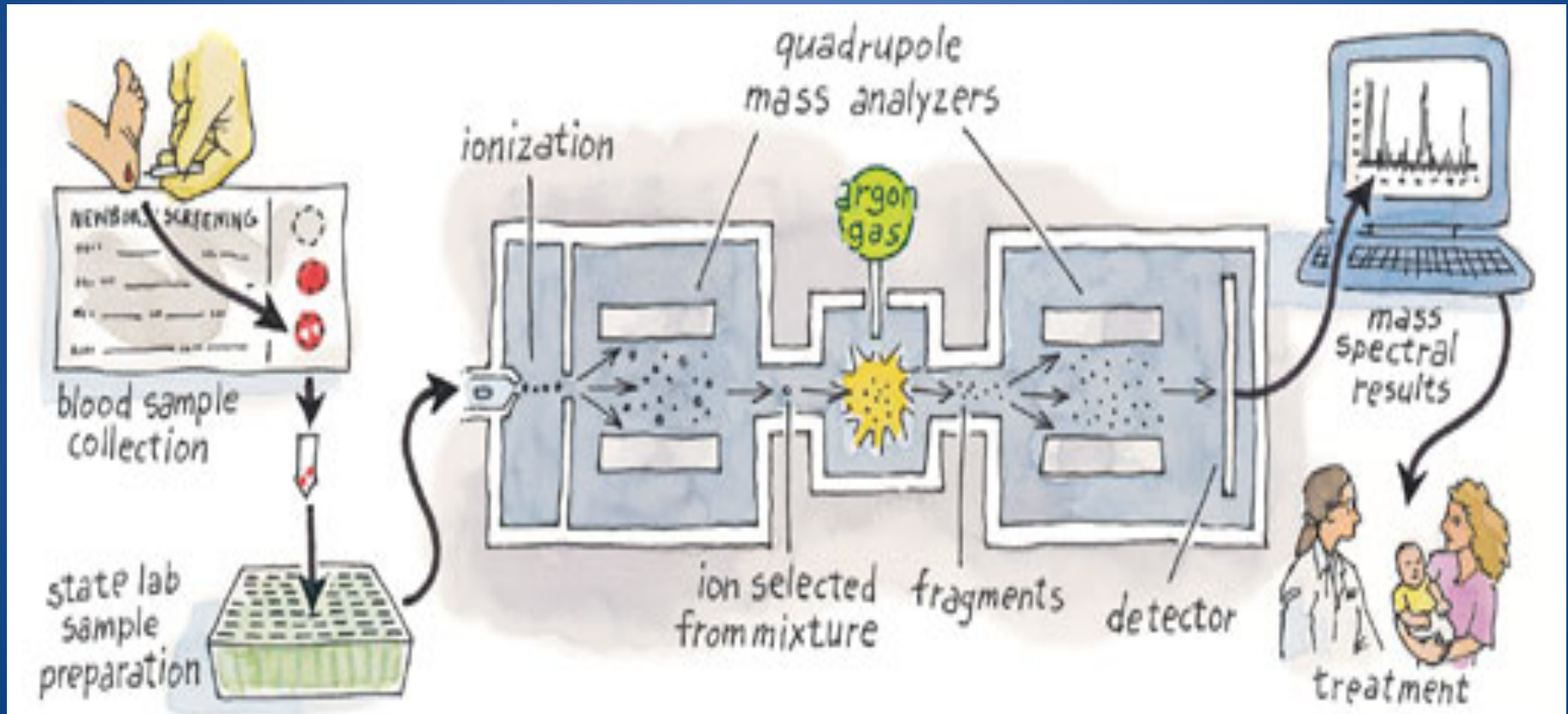
- 2005 – SACHDNC endorses uniform screening panel (29 conditions)
- 2010 – SACHDNC endorses addition of SCID to universal newborn screening panel
- 2012 – Critical congenital heart disease - addition to uniform screening panel

Source: “The Newborn Screening Story: How one simple test changed lives, science and health in America”, APHL, 2013

Basic scheme of an MS/MS analysis



NEWBORN SCREENING



NBS

Tennessee NBS

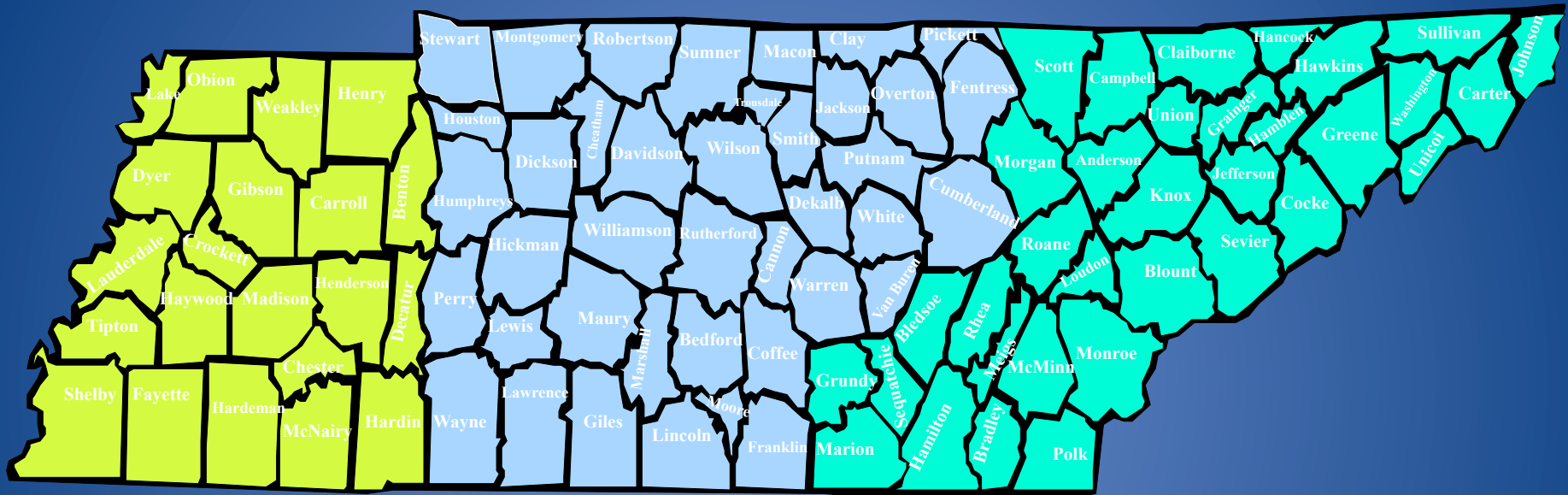
- 1968 - PKU
- 1980 - Hypothyroidism
- 1988 - Hemoglobinopathies
- 1992 – Galactosemia
- 2001 - Hearing (voluntary, 2008 universal)
- 2003 - Biotinidase deficiency
- 2004 - TMS based disorders (amino acids, organic acids, fatty acid oxidation
(all primary targets, >95% secondary targets)
- 2008 - Cystic 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%)

Referral Pattern for Birth Population



-  U T Memphis
-  Vanderbilt
-  U T Knoxville

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

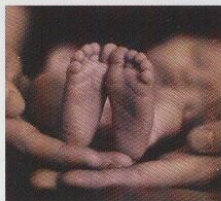
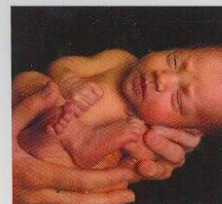
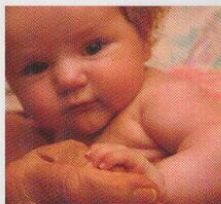
- Genetics Advisory Committee (GAC)
 - Advisory to Genetics and NBS section of TN Maternal and Child Health, and State Newborn Screening Lab (2 sessions/year)
 - Established in 1970's
 - Representation from
 - Comprehensive Genetic Centers/Satellite Genetic Centers
 - Hematology/Endocrine/Pulmonary/Pediatric 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



TENNESSEE
DEPARTMENT OF
HEALTH

It's About Time!

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>
- 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 Genetics Resource Center, MOD, ACMG ACT Sheets, AAP, etc)

NBS

TN NBS – Unsatisfactory Specimens

- Unsatisfactory rate issues
 - Adopted goal of $< \text{ or } = 2\%$ 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