

Continuum from Service to Research: Massachusetts Research Models

Challenges and Opportunities in Using Newborn Screening Samples for
Translational Research: a Workshop

INSTITUTE OF MEDICINE

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New England Newborn Screening Program

Newborn Screening is

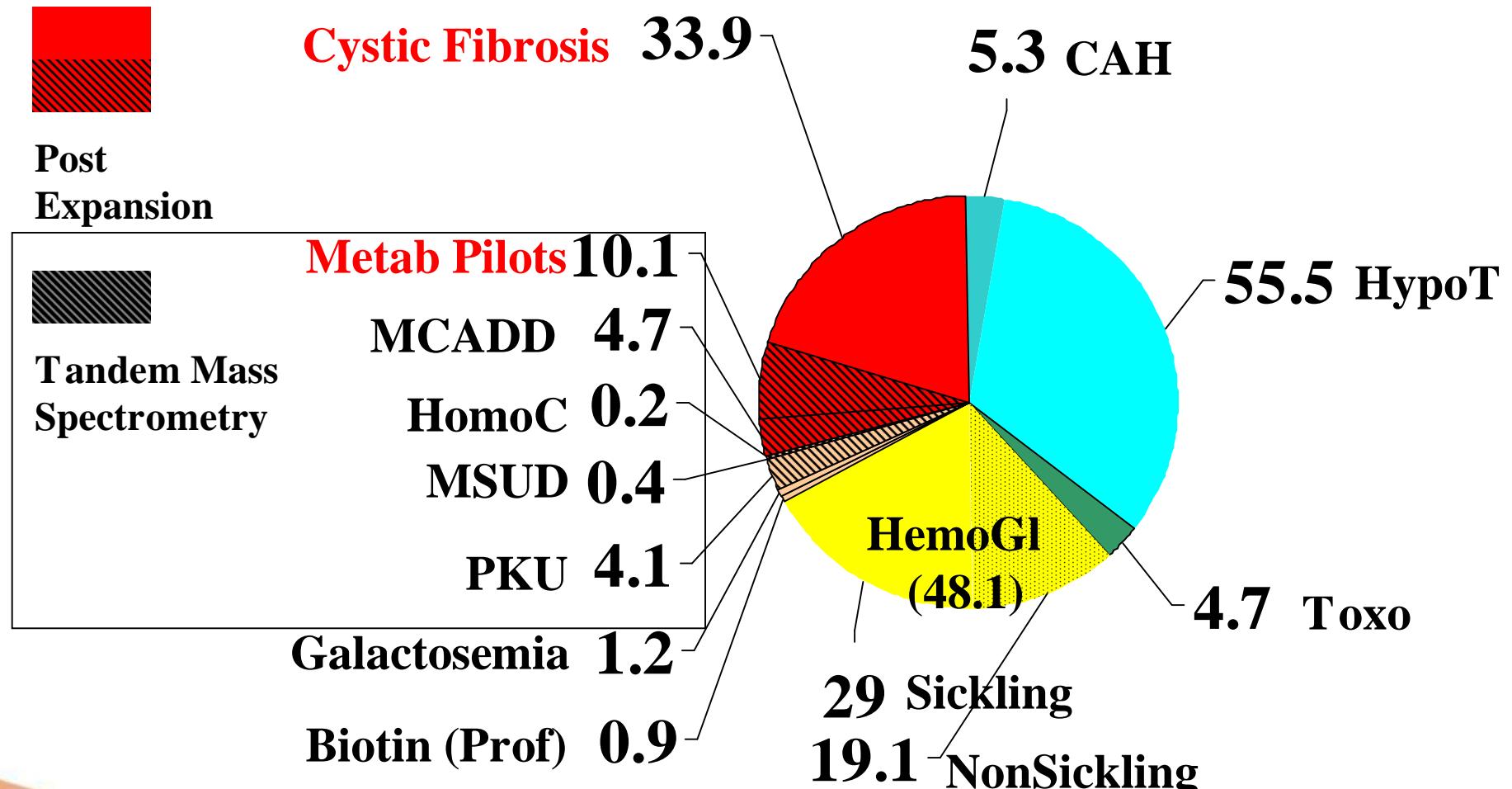
- a public health program that
- provides an opportunity for early identification and early treatment
- of infants with conditions that otherwise would go unrecognized
- prior to irreversible clinical damage.

Newborn Screening is

Highly successful

Newborn Screening in New England

Number of Cases Detected (169)/100,000 Screened



Newborn Screening is

more than just a laboratory test.

Newborn Screening success:

What makes it so?

What keeps it so?

How do we further improve NBS?

Can we – should we - how do we

build upon NBS success to benefit
other areas and systems of health?

Newborn Screening success:

Quality people

Quality systems

Continuum



Based upon public trust

Public Health Program

Authorized by the states

Individual benefit – *Parents Patriae*

Societal benefit

Universal access

MASSACHUSETTS DEPARTMENT OF PUBLIC HEALTH POLICY ON THE PURPOSE, STORAGE AND USE OF SPECIMENS RESIDUAL TO THOSE COLLECTED FOR NEWBORN SCREENING SERVICES

- **Overview**
- **Rationale for Retaining Identified or Anonymized Residual Specimens**
- **Continuous Quality Improvement**
- **Research**
- **Individual Clinical Investigations**
- **Forensics**
- **Notice**

- **II. Rationale for Retaining Identified or Anonymized Residual Specimens**
- **legal accountability** – reconfirmation of newborn screening analytical results, number of punches taken for analysis, the existence of a specimen and its adequate collection
- **laboratory quality control** – specimens are necessary to conduct continuous quality improvement for testing methodology
- **new method evaluations and comparisons** – specimens are necessary to validate new analytical methods and/or to compare methods
- **epidemiologic research to benefit the public health** - research on anonymized and identified specimens can advance newborn screening efforts as well as other areas of public health medicine
- **basic research to benefit general medical knowledge**- research on select identified specimens can advance understanding of newborn biochemistry and genetics in the early postnatal development

Research for Public Health Benefit

- Continuous Quality Improvement
- Epidemiological research
- Basic research to enhance general medical knowledge

Research Applications

De-identified

Linked

Research for Public Health Benefit

- Continuous Quality Improvement
- Epidemiological research
- Basic research to enhance general medical knowledge

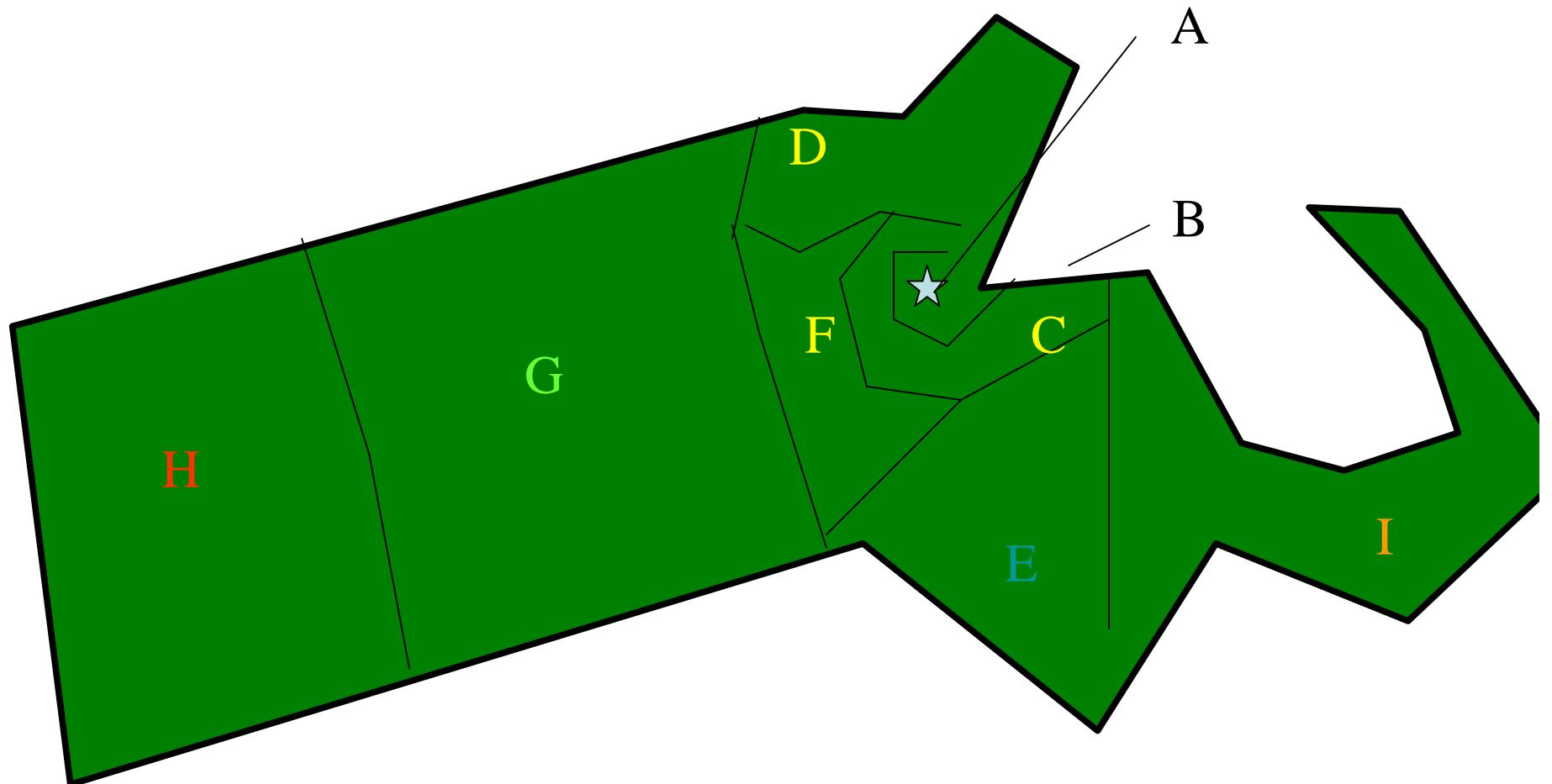
Research Applications

(Specimens Stripped of Identifiers)

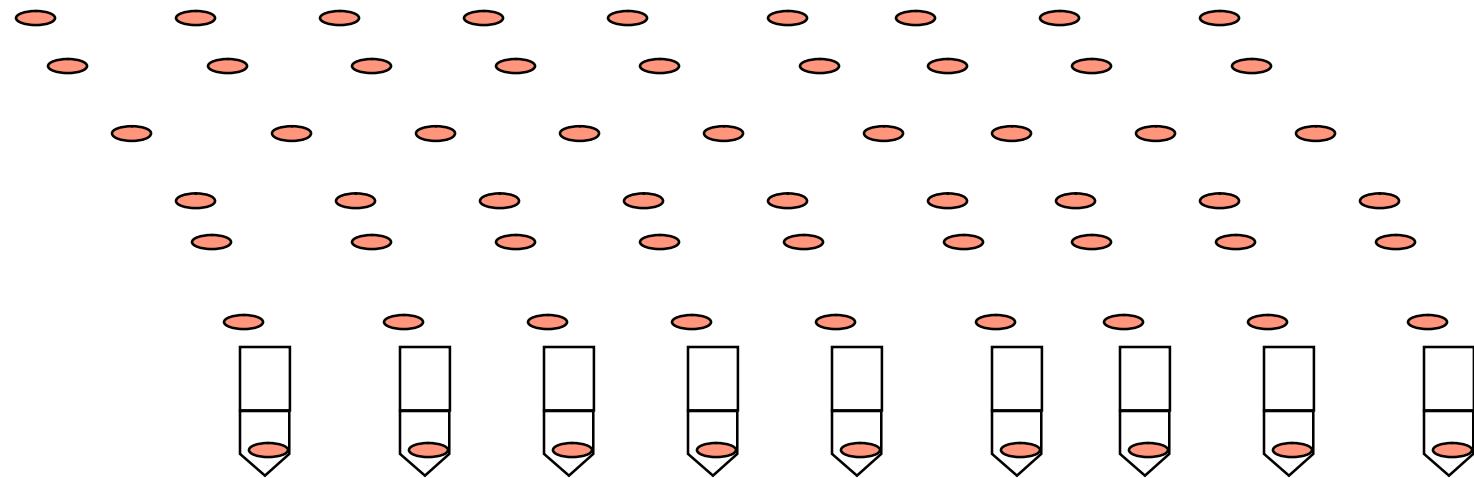
Seroprevalence of Human Immunodeficiency Virus Among Childbearing Women

Hoff et al 1988 NEJM 318(9): 525-530

Massachusetts Regions



Rate per thousand



Group	A	B	C	D	E	F	G	H	I
HIV Ab	11.6	2.2	1.1	0.7	2.2	2.9	2.1	3.5	0.3

Research for Public Health Benefit

- Continuous Quality Improvement
- Epidemiological research
- Basic research to enhance general medical knowledge

Research Priorities

- Further newborn screening activities
- Further diagnostic/treatment developments for newborns and young infants
- Further understanding of disease in newborns and young infants
- Further understanding of general medical knowledge

Criteria for adding to Newborn Screening List (by Regulation)

- Infant should benefit
- Condition serious and frequent enough to justify screening entire population
- Treatment is effective and available to all screened
- Test exists that is simple, safe, precise, validated

American Academy of Pediatrics, August, 2000

Human Subjects Review

2 Independent Review Boards

- Presumed benefit to infant
- No additional blood
- Protocols to assure clinical follow up for infants
- Practical implications for 80,000 annual births
 - Alternate form of informed consent

METHOD OVERVIEW

- Verbal Informed Consent
- Mechanism:
 - distribute expanded brochure
 - ask parent

CONSENT PROTOCOL IMPLEMENTATION

- Descriptive Brochure (pre- and neonatal)
- NINE languages
- Consent form (collection card)
- In-service trainings and grand rounds
- Apprise pediatric providers
- QA/QC monitoring

Things we DO know from Massachusetts

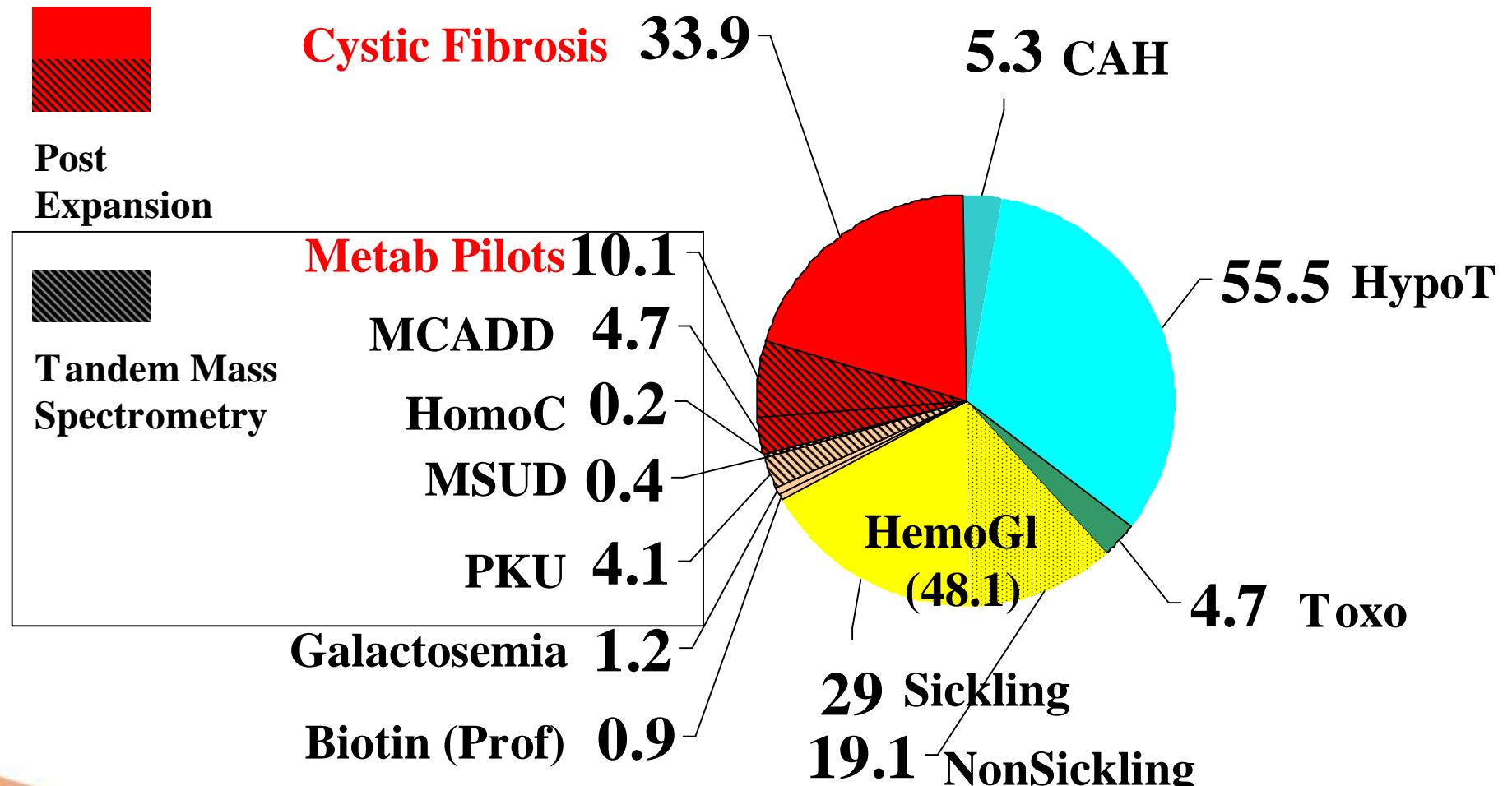
Initial Specimens...CF and metabolics

DOB 2/1/99 –2/1/09

- Total Number of Babies > **784,000**
- Total number declined **8,000 1%**
- **Use of Brochures**
- Consistent with pre and postnatal distribution

Newborn Screening in New England

Number of Cases Detected (169)/100,000 Screened



Massachusetts' Implementation of SCID NBS

- Panel of conditions determined by Regulation
- New Regulations:
 - Many MS/MS conditions moved
 - CF mandatory
 - SCID and other MS/MS to pilot program
 - required outcomes reporting to NENSP
- IRB complete
- Screening begun February 2009

Initial Specimens....SCID DOB 2/1/09 –2/1/10

- Total Number of Babies > 75,000
- Total number declined < 1%
- **Use of Brochures**
- Consistent with pre and postnatal distribution

The next study-

- Justification ?
- Stewardship of specimens, of data?
- Study begins :
 - At enrollment for newborn screen?
 - At enrollment for clinical trial?

Newborn Screening success:

Quality people

Quality systems

Continuum



Based upon public trust

Considerations

- Repository entails significant costs
- Physical/Environment
- Quality Control
- Data management: ingoing outgoing
- Study management/ethics assurance
- Data management: study design

Virtual Repository Model -

PHYSICAL LOCATION	VIRTUAL LOCATION	STEWARDSHIP /CONTROL	SHARE LEVEL
Destroyed			
Individual		Individual	
State	State	State	
STATE	State: details Central: general	Committee of Contributors	Project-Specific
Central National		Federal	