Region 1
New England Regional Genetics and Newborn Screening Collaborative

PI : Thomas Brewster, MD
New England Regional Genetics Group
Regional Mapping: Applications and Data

Anne Marie Comeau, Ph.D.
Deputy Director
New England Newborn Screening Program
University of Massachusetts Medical School
Key Collaborators

• Title V / MCH Directors New England states

• NBS /Genetics Coordinators New England states

• NBS Program Directors: CT, NENSP, NY
Objectives

• Develop and implement epidemiologic methods for assessment of regional needs for genetic services using three NBS groups (CF, hemoglobinopathies, biochemical genetic disorders) as a model.

• Develop best practice models based upon current need and epidemiologic data.
Newborn Screening Applications

• Early indicators of disorders

• Early indicators of current and future needs of the population accessing specialized health services
Location, location…

New England Newborn Screening Program
Genetic Epidemiology

- Where are the cases?...

- Where are the services?...
6 years Massachusetts CF Newborn Screening

10 NH residents

2 RI residents

New England Newborn Screening Program
## Central Maine & MA Composite Population Comparison

<table>
<thead>
<tr>
<th></th>
<th>Central Maine</th>
<th>MA Towns % White ≥ 97.7</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>TOTAL POPULATION</strong></td>
<td>385,176</td>
<td>455,610</td>
</tr>
<tr>
<td>% White</td>
<td>97.65</td>
<td>98.02</td>
</tr>
<tr>
<td>% Black</td>
<td>0.36</td>
<td>0.35</td>
</tr>
<tr>
<td>% American Indian</td>
<td>0.32</td>
<td>0.14</td>
</tr>
<tr>
<td>% Asian</td>
<td>0.47</td>
<td>0.51</td>
</tr>
<tr>
<td>% Other</td>
<td>1.19</td>
<td>0.98</td>
</tr>
<tr>
<td>% Hispanic</td>
<td>0.71</td>
<td>0.80</td>
</tr>
<tr>
<td>% not Hispanic</td>
<td>99.29</td>
<td>99.20</td>
</tr>
</tbody>
</table>
Northern Maine
2 CF Births per year
13 Category B per year
(sweat negative)

Central Maine
2 CF Births per year
15 Category B per year
(sweat negative)

Southern Maine
2 CF Births per year
16 Category B per year
(sweat negative)
CF NBS Projections & Location of CF Center
for State of New Hampshire

Northern New Hampshire
1 CF birth every other year
5 Category B per year
(sweat negative)

Southern New Hampshire
4 CF Births per year
33 Category B per year
(sweat negative)

New England Newborn Screening Program
CF NBS Projections and Location of CF Center for State of Vermont

State of Vermont
3 CF Births per year
24 Category B per year (sweat negative)

New England Newborn Screening Program
CF NBS Projections and Location of CF Center for State of Rhode Island

State of Rhode Island

3 CF Births per year

31 Category B per year (sweat negative)

New England Newborn Screening Program
Locations of CF Centers in the Northeast

Cystic Fibrosis Centers
Connecticut: Hartford, New Haven
Maine: Bangor, Lewiston, Portland
Massachusetts: Boston (3), Springfield, Worcester
New Hampshire: Lebanon, Manchester
New York: Albany, New Hyde Park, New York City (4), Stony Brook, Syracuse, Valhalla, Watertown, 2 others in far Western New York
Rhode Island: Providence
Vermont: Burlington
New England Newborn Screening Program

1 year Hemoglobinopathies screening results
MCADDs Detected in New England by NBS in MA, ME, RI, & VT Relative to Mother’s Residence

- 985A>G / 985A>G
- 985A>G / other mutation
- Other mutation / other mutation
- Genetics Clinic
Locations of Metabolic, CF, & Hgb Centers in New England

CF Center
Hgb Center
CF & Hgb Center
CF & Metabolic Center
CF, Hgb & Metabolic Center

New England Newborn Screening Program
### Extent of “border babies”

<table>
<thead>
<tr>
<th>State of residence</th>
<th>Requisitioning State</th>
</tr>
</thead>
<tbody>
<tr>
<td>MA</td>
<td>475,876</td>
</tr>
<tr>
<td>ME</td>
<td>79956</td>
</tr>
<tr>
<td>NH</td>
<td>77132</td>
</tr>
<tr>
<td>RI</td>
<td>73664</td>
</tr>
<tr>
<td>VT</td>
<td>32207</td>
</tr>
<tr>
<td>CT</td>
<td>1636</td>
</tr>
<tr>
<td>NY</td>
<td>533</td>
</tr>
<tr>
<td>Other</td>
<td>522</td>
</tr>
</tbody>
</table>

New England Newborn Screening Program
Best practice models responding to current needs

- Border babies
- Laboratory backup
- Clinical backup

New England Newborn Screening Program
Summary

• Newborn Screening is a regional system/itself an early indicator

• Identify components for Continuous Quality Improvement

• Applications for other genetic services