Region 1
New England Regional Genetics and Newborn Screening Collaborative

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New England Regional Genetics Group

Regional Mapping: Applications and Data

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Key Collaborators

- Title V / MCH Directors New England states
- NBS /Genetics Coordinators New England states
- NBS Program Directors: CT, NENSP, NY

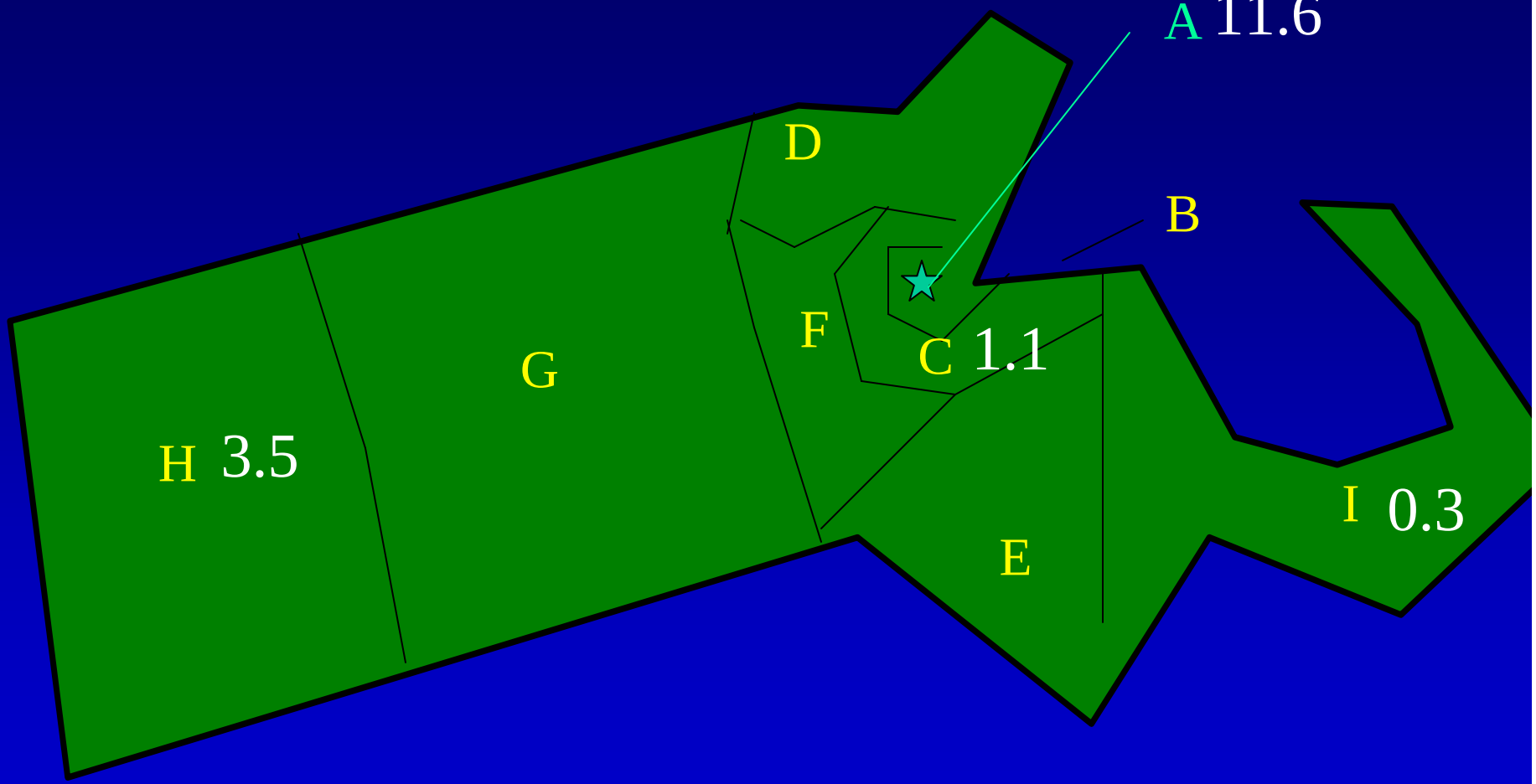
Objectives

- 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…



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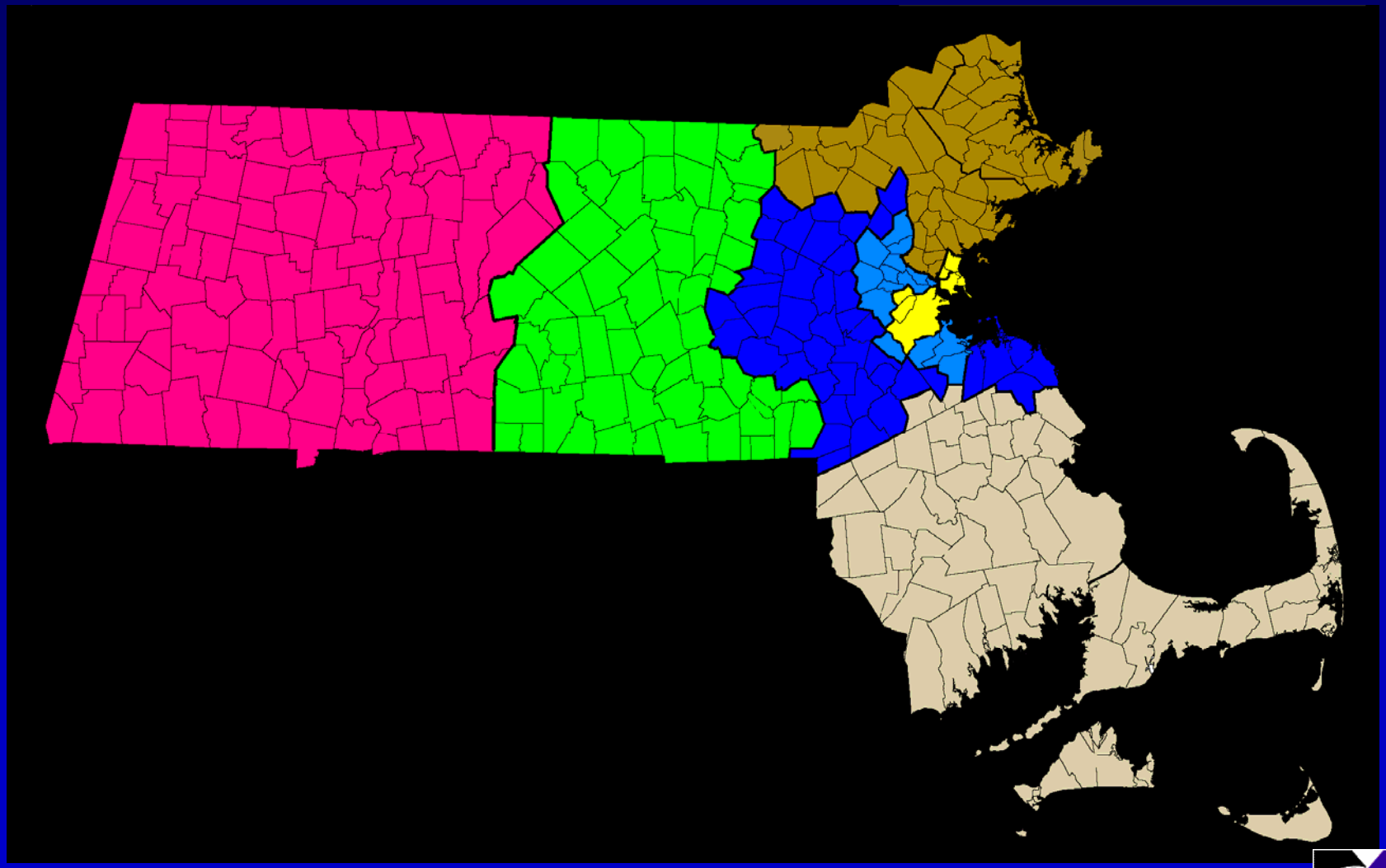
Genetic Epidemiology

- Where are the cases?…
- Where are the services?…



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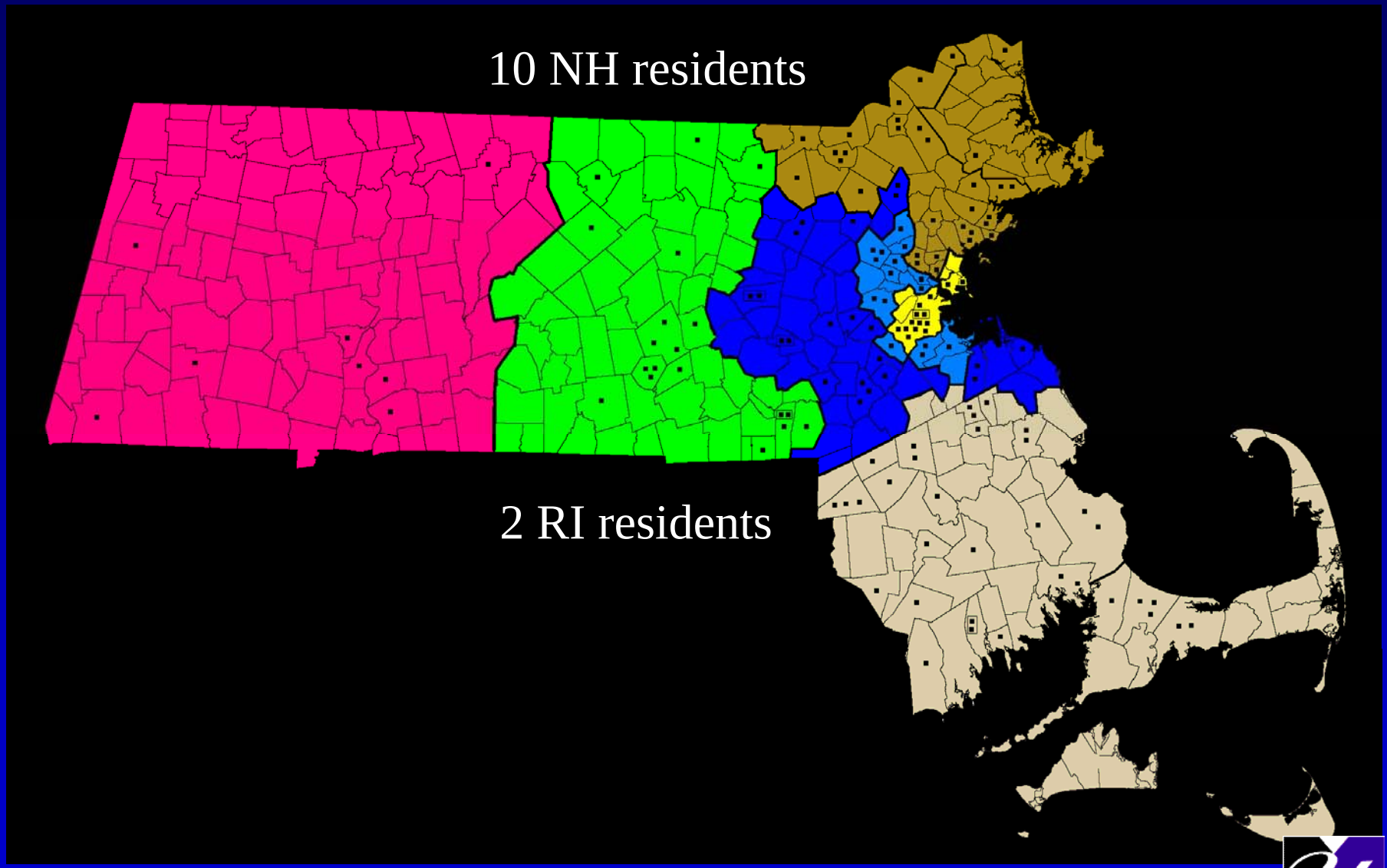
6 years Massachusetts CF Newborn Screening



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Central Maine & MA Composite Population Comparison

	Central Maine	**MA Towns % White ≥ 97.7**
TOTAL POPULATION	385,176	455, 610
% White	97.65	98.02
% Black	0.36	0.35
% American Indian	0.32	0.14
% Asian	0.47	0.51
% Other	1.19	0.98
% Hispanic	0.71	0.80
% not Hispanic	99.29	99.20

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CF NBS Projections & Locations of CF Centers for State of Maine

Central Maine

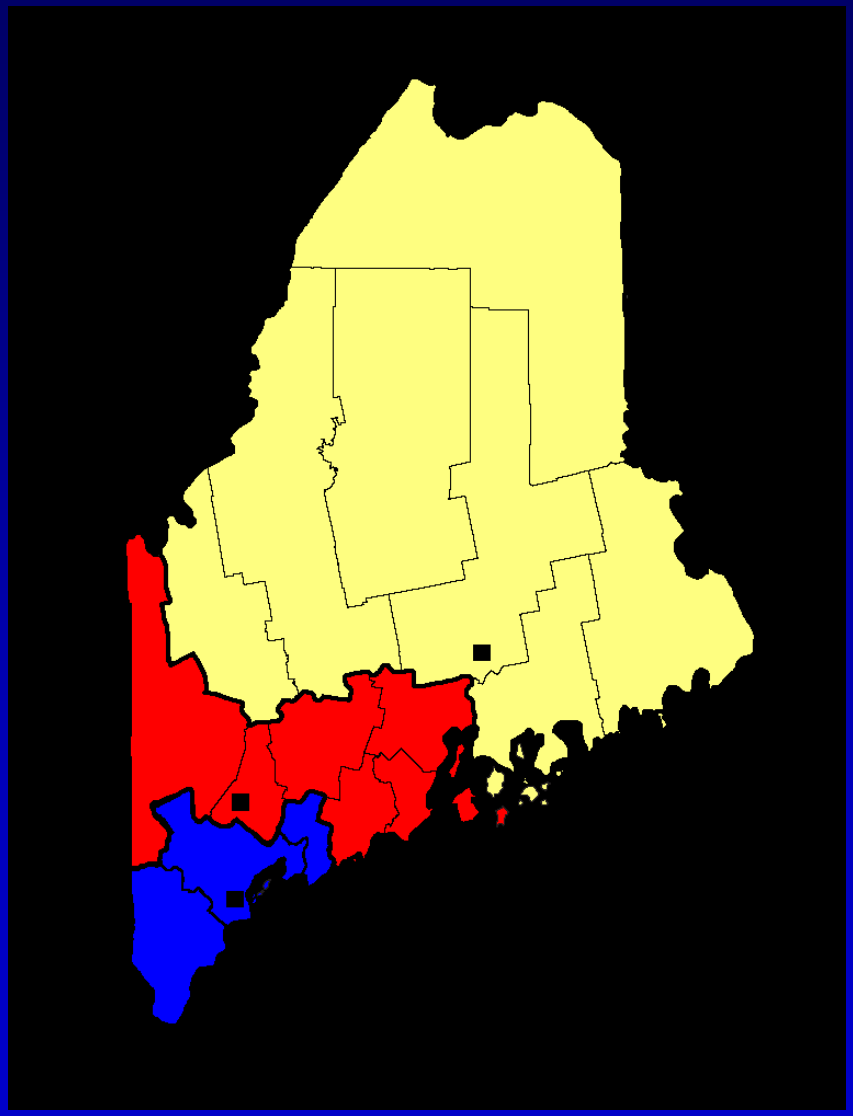
2 CF Births per year

15 Category B per year (sweat negative)

Southern Maine

2 CF Births per year

16 Category B per year (sweat negative)



Northern Maine

2 CF Births per year

13 Category B per year (sweat negative)

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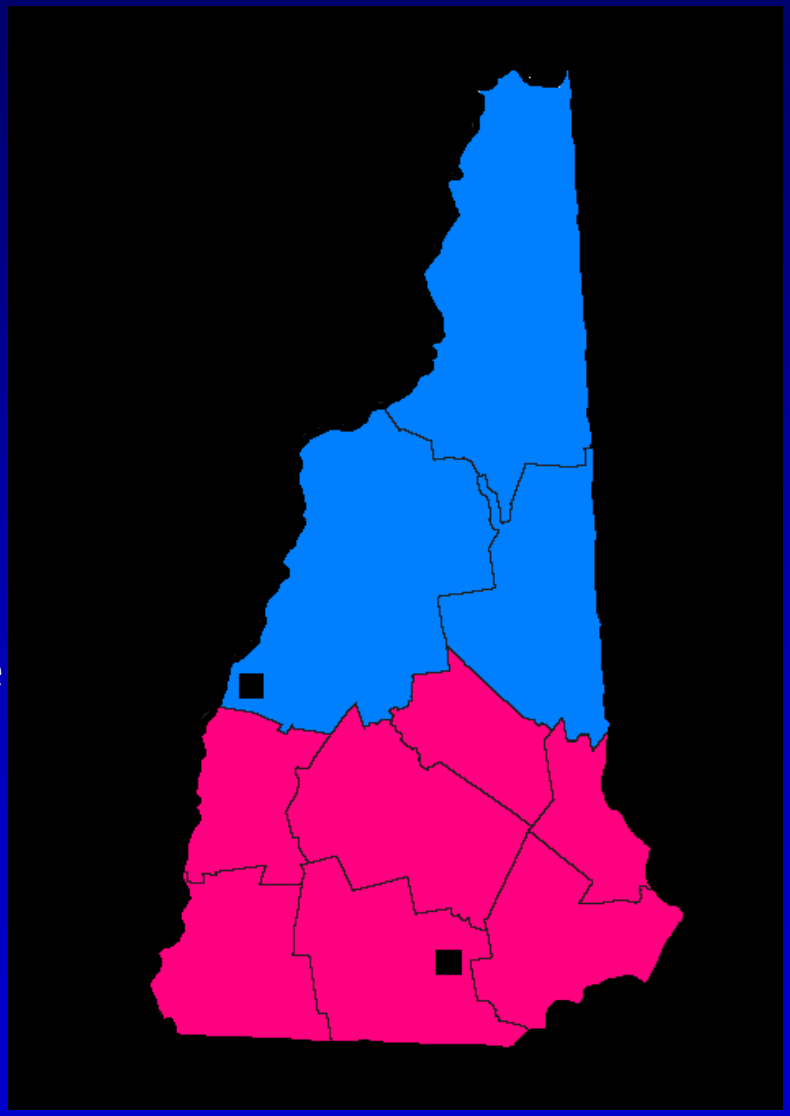
CF NBS Projections & Location of CF Center for State of New Hampshire



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)

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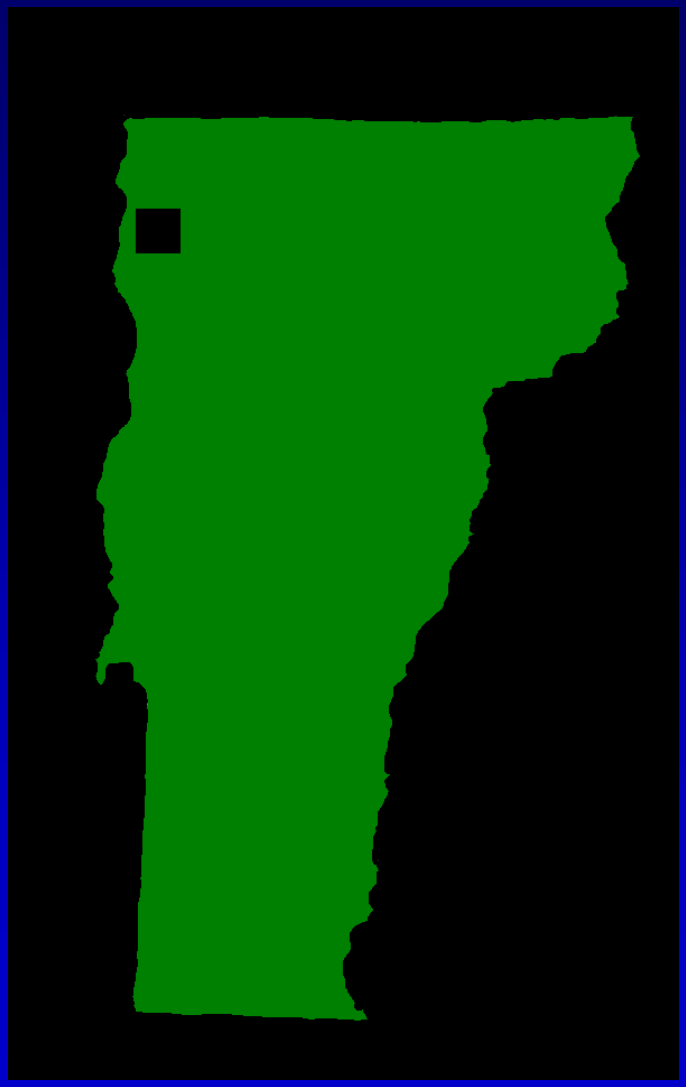
CF NBS Projections and Location of CF Center for State of Vermont



State of Vermont

3 CF Births per year

24 Category B per year (sweat negative)

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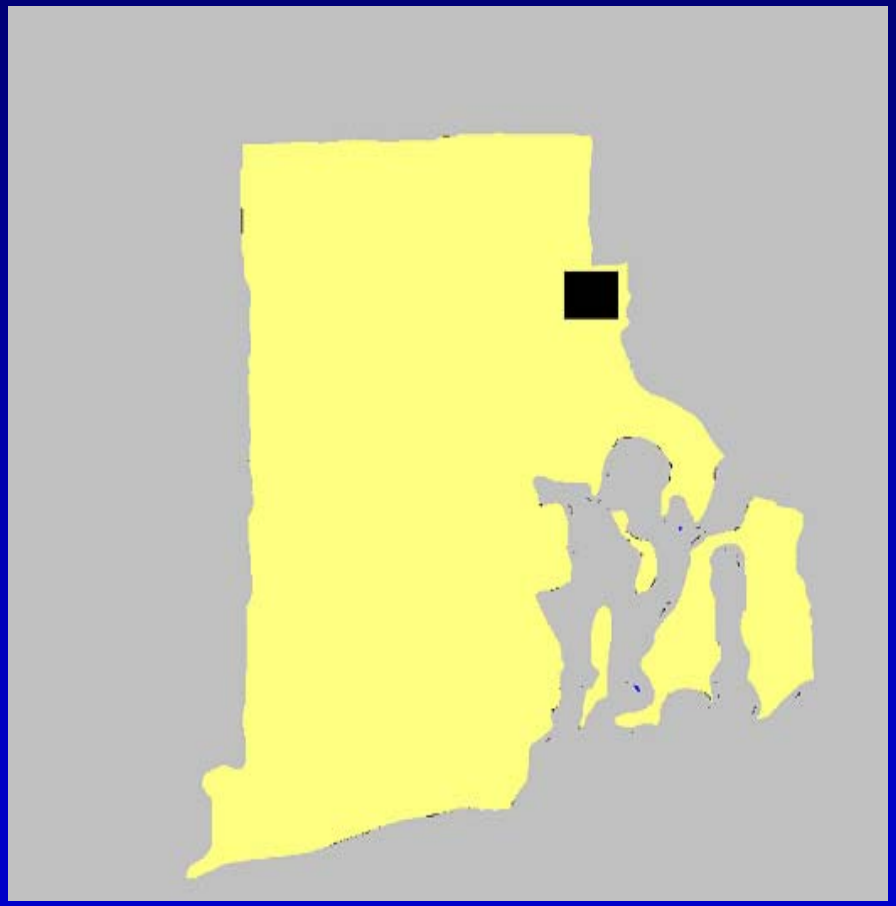
CF NBS Projections and Location of CF Center for State of Rhode Island



State of Rhode Island

3 CF Births per year

31 Category B per year (sweat negative)

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Locations of CF Centers in the Northeast

Cystic Fibrosis Centers

Connecticut: Hartford, New Haven

Maine: Bangor, Lewiston, Portland

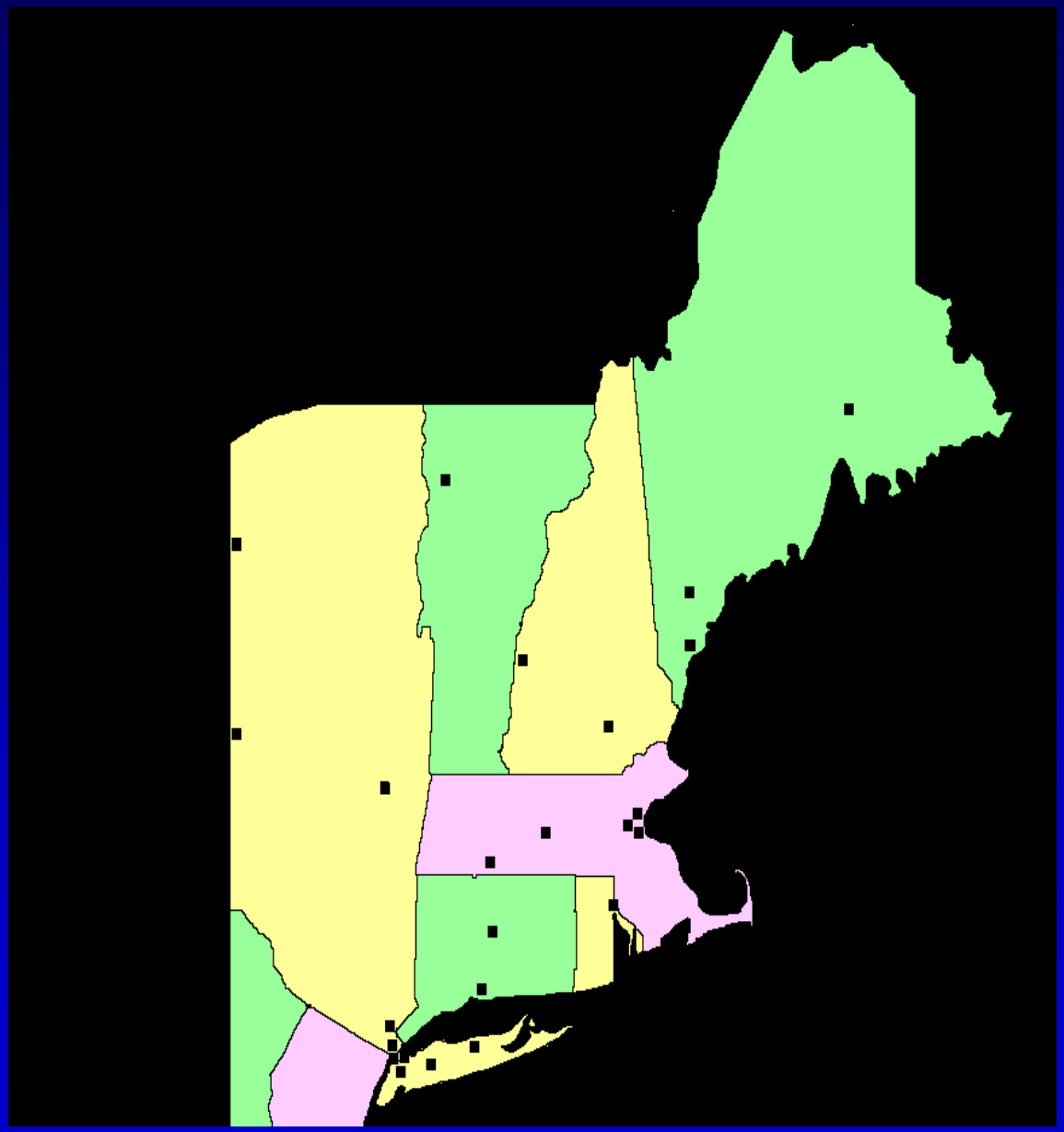
Massachusetts: Boston (3), Springfield, Worcester

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



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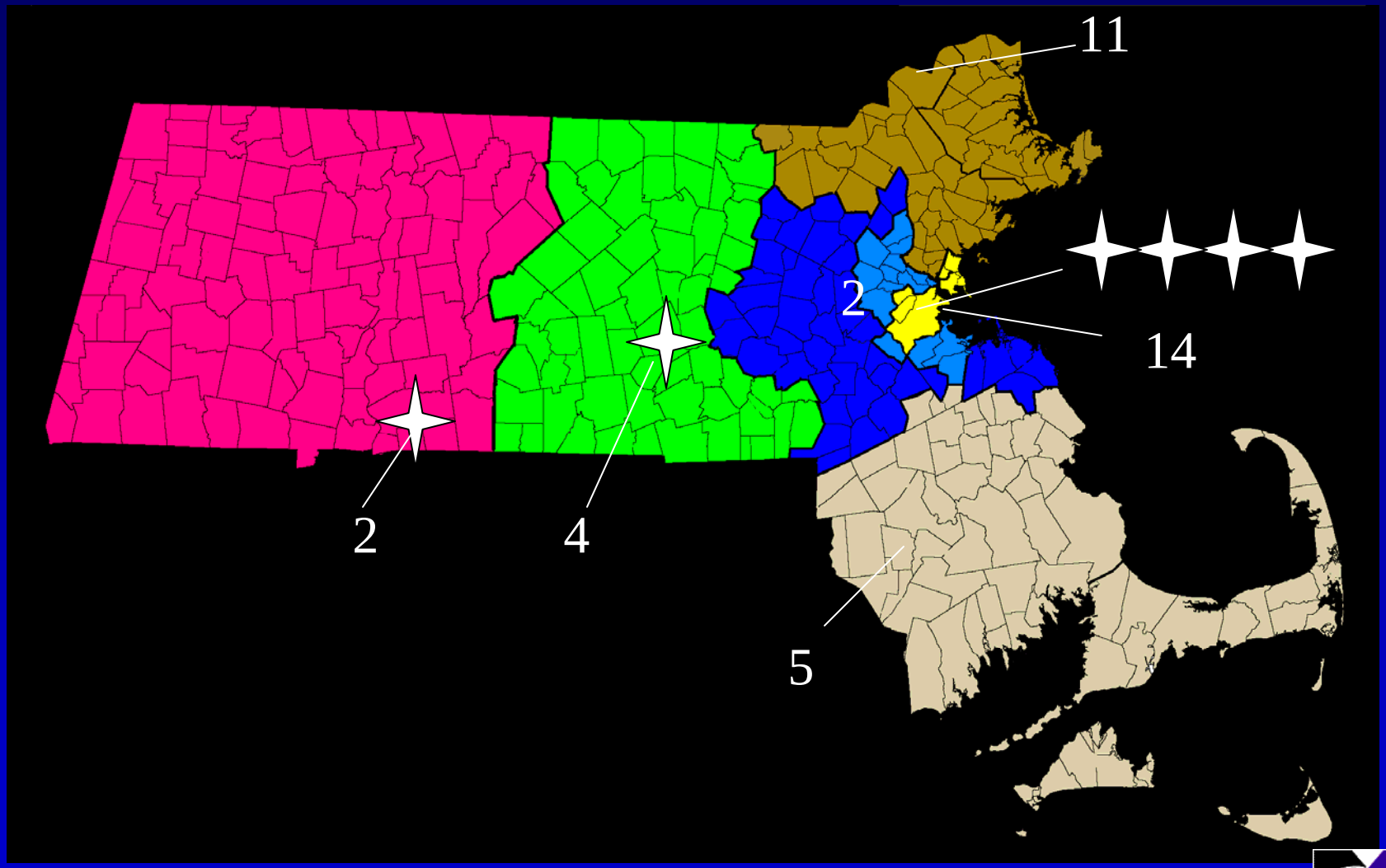
1 year Hemoglobinopathies screening results



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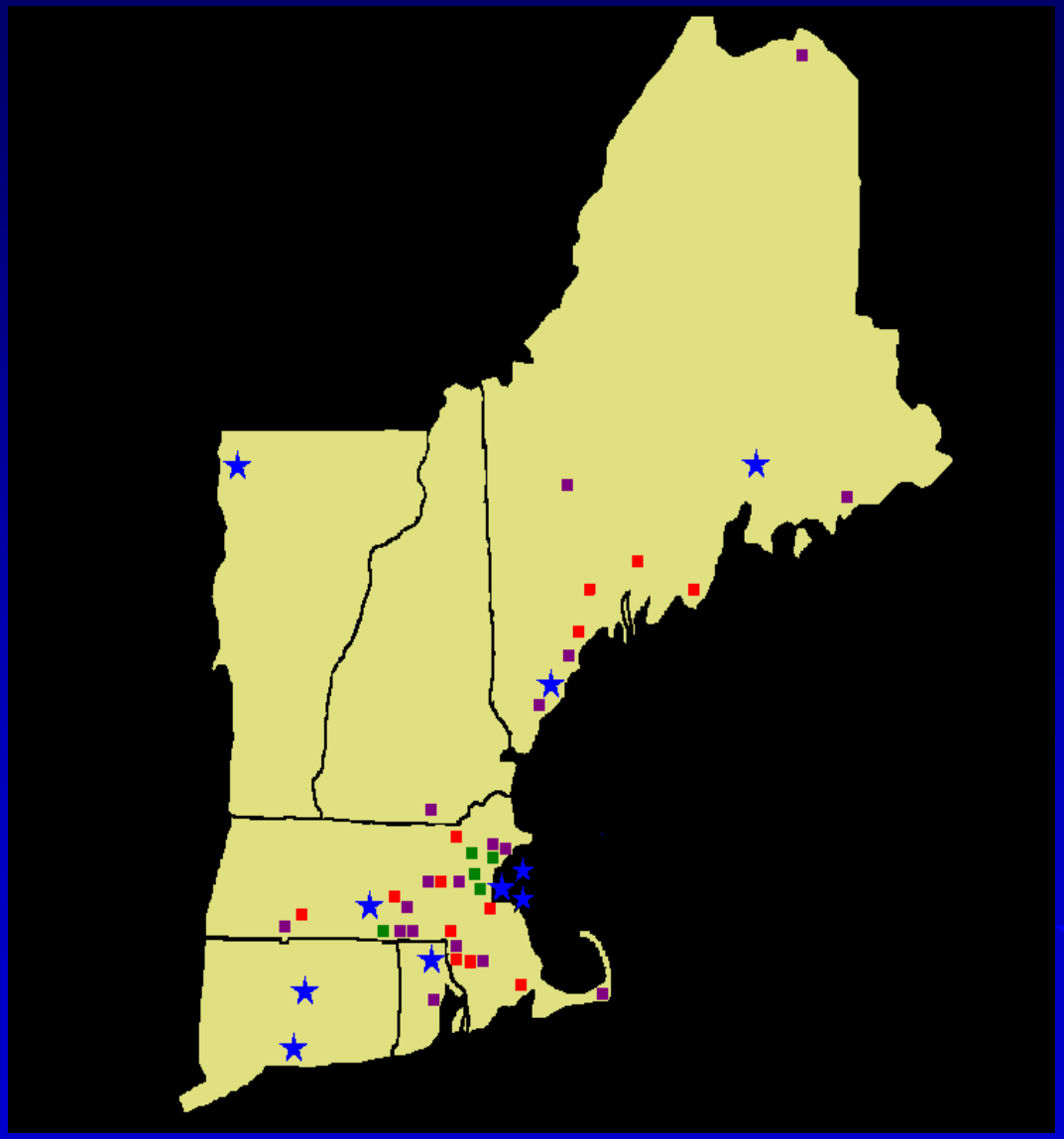
MCADDs Detected in New England by NBS in MA, ME, RI, & VT Relative to Mother's Residence



985A>G / 985A>G

985A>G / other mutation

Other mutation / other mutation

Genetics Clinic

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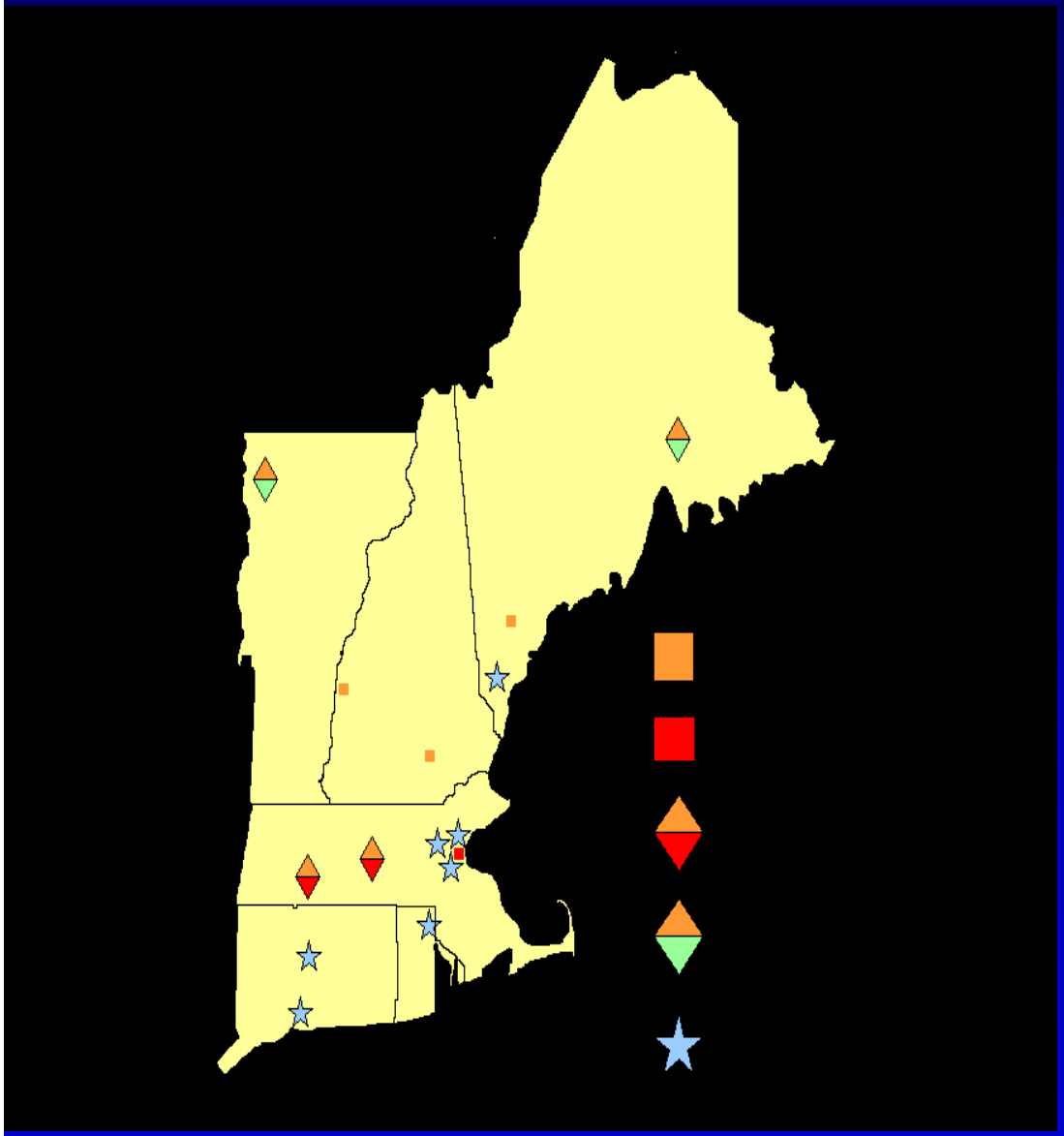
Locations of Metabolic, CF, & Hgb Centers in New England



CF Center

Hgb Center

CF & Hgb Center

CF & Metabolic Center

CF, Hgb & Metabolic Center

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Extent of “border babies”

	Requisitioning State				
State of residence	MA	ME	NH	RI	VT
MA	**475,876**	39	949	5349	77
ME	188	**79956**	1352	5	0
NH	8356	470	**77132**	11	454
RI	2127	1	5	**73664**	0
VT	294	4	3868	4	**32207**
CT	1636	7	19	1020	4
NY	533	13	25	17	2071
Other	522	360	117	329	1817

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Best practice models responding to current needs

- Border babies
- Laboratory backup
- Clinical backup

Summary

- Newborn Screening is a regional system/ itself an early indicator
- Identify components for Continuous Quality Improvement
- Applications for other genetic services