



NYMAC



NEW YORK-MID-ATLANTIC CONSORTIUM FOR GENETIC AND NEWBORN SCREENING SERVICES

Delaware, District of Columbia, Maryland, New Jersey, New York
Pennsylvania, Virginia and West Virginia

NYMAC REGIONAL NEEDS ASSESSMENT 2012

Regional Needs for Genetic Services

The Mid-Atlantic Region is extremely diverse. Within this region is found every possible barrier to health care and every possible solution. Congested cities, rugged mountains; the poorest people, the wealthiest people; frigid, icy, snowy winters; hot, humid, storm-ridden summers; every race, ethnicity and language; outstanding educational systems and those that fail their children; scores of public and private health insurance options and thousands of people who have no insurance; networks of specialty care for those with medical need and people who need that care but cannot seem to access services.

State Genetic Needs Assessments and Plans

- None of the jurisdictions has a Genetics Plan filed with the National Newborn Screening and Genetics Resource Center.
- Delaware has an undated plan that includes raising awareness of Genetic and Newborn Screening Services.
- District of Columbia has an undated draft plan that raises twelve issues, discusses interventions and proposes strategies.
- New Jersey has an undated review of genetic and newborn screening resources in the state. There is no identification of gaps and no plan.
- New York has an undated outline of a plan that identifies needs but proposes no solutions.
- Pennsylvania has a March 2007 draft Genetic Services Overview that proposes to develop a state-wide Genetic Services Plan.
- Virginia has an April 2007 Needs Assessment that includes recommended 2008 scenarios concerning genetic service provider contracts and allocations of funds to local health departments.
- West Virginia has not provided a copy of its plan to NYMAC
- There is no assessment or plan for Maryland.
- In general, while the states are supportive of the NBS Program and recognize the work of the Genetics and Specialty Care Centers, development of a plan is not a DOH priority, given the lack of resources to implement any proposals.

Newborn Screening Programs of Region 2

Each jurisdiction in Region 2 has been screening newborns for congenital conditions for decades – many began in the 1960s. Most jurisdictions have a Newborn Screening Advisory Committee charged with evaluating the composition of the state newborn screening panels in light of population needs, medical and public health standard of care and fiscal constraints. Each jurisdiction has developed a system of specialty care centers and early intervention for the special needs of the children identified by the panels.

With the recent advances in computer technology, MS/MS and DNA mutation analysis, plus increasing public concern about nationwide variations in newborn screening panels, including the March of Dimes and American College of Medical Genetics recommendations and DHHS Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children, all programs have reexamined their test panels and operations.

- All eight NBS Programs screen all infants for all the recommended analytes, except for the newly recommended analytes for SCID and CCHD. As of September 30, 2009 New York screens all infants for SCID. New Jersey added CCHD in 2011.
- There is not a consensus as to whether the newborn screening programs cover the secondary target conditions. Except for defects of biotin cofactor biosynthesis, defects of biotin cofactor regeneration, galactose epimerase, and galactokinase, which require analysis difference from those in the recommended panel, so may not be flagged by newborn screening, the secondary target conditions are likely to be diagnosed in the course of the clinical evaluation conducted by the metabolic centers.

National Newborn Screening Status Report

Updated 12/19/11

STATE	Core ¹ Conditions											Additional Conditions Included in Screening Panel (universally required unless otherwise indicated)	
	Hearin	Endocrine			Hemoglobin			Other					
	HEAR	CH	CAH	S/S	S/A	S/C	BIO	GALT	CF	CCHD	SCID		
D.C.	●	●	●	●	●	●	●	●	●	●			G6PD
Delaware	●	●	●	●	●	●	●	●	●	●			C
Maryland	●	●	●	●	●	●	●	●	●	●			EMA
New Jersey	●	●	●	●	●	●	●	●	●	●	●		
New York	●	●	●	●	●	●	●	●	●	●		●	HIV; HHH; Krabbe Disease
Pennsylvania	●	●	●	●	●	●	●	●	●	●		●	5-OXO; CPS; G6PD; HHH; NKH (B)
Virginia	●	●	●	●	●	●	●	●	●	●			
West Virginia	●	●	●	●	●	●	●	●	●	●		●	C

STATE	Core ¹ Conditions - Metabolic																			
	Fatty Acid Disorders					Organic Acid Disorders								Amino Acid Disorders						
	CUD	LCHAD	MCAD	TFP	VLCAD	GA-I	HMG	IVA	3-MCC	Cbl-A,B	BKT	MUT	PROP	MCD	ASA	CIT	HCY	MSUD	PKU	TYR-I
D.C.	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Delaware	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Maryland	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
New Jersey	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
New York	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Pennsylvania	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Virginia	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
West Virginia	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●

STATE	Secondary Target ¹ Conditions							
	Fatty Acid Disorders		Organic Acid Disorders		Amino Acid Disorders	Other	Hbg	

																Metabolic			Variant Hbg's						
	CACT	CPT-Ia	CPT-II	DE-RED.	GA-II	MCKAT	M/SCHAD	SC AD	2M3HBA	2MBG	3MGA	Chl-C.D	IBG	MAL	ARG	BIOPT-BS	BIOPT-RG	CIT-II		H-PHE	MET	TYR-II	TYR-III	GALE	GALK
D.C.	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	A	A	●	●	●	●	●	●	●	●
Delaware	●	●	●	●	●	D	●	●	D	●	D	●	●	●	●	D	D	●	●	●	●	●	●	●	●
Maryland	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	B	B	●	●	●	●	●	●	●	●
New Jersey	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
New York	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Pennsylvania	B	B	B	B	B		B	B		B	B	B	B	B	B	B	B	●	B	B	B	B	●	●	●
Virginia	D	D	D	D	D	D	D		D	D	D	D		D	D	D	D	D	D	D	D	D	D	D	●
West Virginia	D	D	D	D	D	D	D		D	D	D	D	D	D	D	D	D	D	●	D	D	D	D	●	●

Newborn Screening Follow-up – need to update

All the newborn blood-spot screening programs have developed effective procedures for ensuring confirmation of disease in screen-positive infants. In general, preliminary test results for all specimens are available within two days of receipt at the testing laboratory. Those highly indicative of a disorder are telephoned to the physician of record, the hospital of birth and the recognized specialty care center closest to the address of the mother. Reports of specimens with borderline screen positive results are mailed to the physician of record and/or the hospital of birth. Confirmation of diagnosis for bloodspot screening – positive or negative - is required by all jurisdictions. Two states, District of Columbia and New York do not monitor follow-up of hearing-screening results. As shown in the table below, some programs have long-term follow-up programs. Others are examining cost-effective ways to do so using the evolving system of electronic medical records and electronic transfer of data between systems.

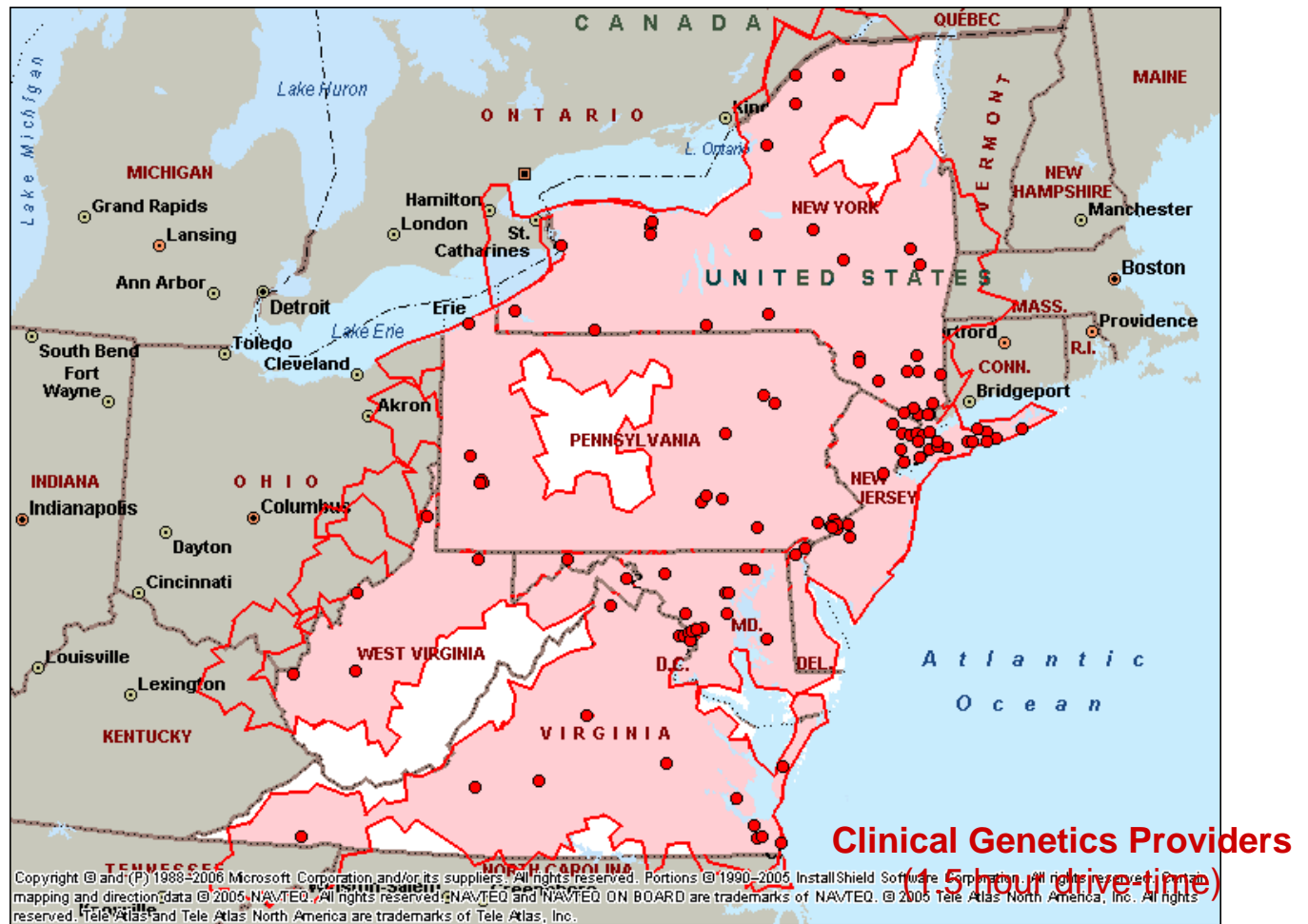
State	Metabolic		Endocrine		Hemoglobin		Cystic Fibrosis		Hearing Screening	
Region 2 (NYMAC)										
DE	1. Yes		1. Yes		1. Yes		1. Yes		1. No	
	2a. Y	2b. 18+	2a. Y.	2b. 3	2a. Y	2b. 20	2a. Y	2b. 2	2a.	2b.
DC	1. No		1. No		1. No		1. No		1. No	
	2a.	2b.	2a.	2b.	2a.	2b.	2a.	2b.	2a.	2b.
MD	1. Yes		1. No		1. Yes		1. No		1. No	
	2a. N	2b.	2a.	2b.	2a. Y	2b. 5	2a.	2b.	2a.	2b.
NJ	1. No		1. No		1. No		1. No		1. Yes (ok LBS)	
	2a.	2b.	2a.	2b.	2a.	2b.	2a.	2b.	2a. Y	2b. 22
NY	1. No		1. No		1. No		1. No		1. No	
	2a.	2b.	2a.	2b.	2a.	2b.	2a.	2b.	2a.	2b.
PA	1. No		1. No		1. No		1. No		1. Yes	
	2a.	2b.	2a.	2b.	2a.	2b.	2a.	2b.	2a. Yes	2b. 3
VA	1. No		1. No		1. No		1. No		1. No	
	2a.	2b.	2a.	2b.	2a.	2b.	2a.	2b.	2a.	2b.
WV	1. No		1. No		1. No		1. No		1. Yes	
	2a.	2b.	2a.	2b.	2a.	2b.	2a.	2b.	2a. Y	2b. 3

See a summary of NYMAC state follow-up protocols at <http://www.wadsworth.org/newborn/nymac/docs/NYMACNewbornScreeningFollow-upProtocols.pdf>

Specialty Care Centers – Blood-spot Programs and Point-of Care Testing

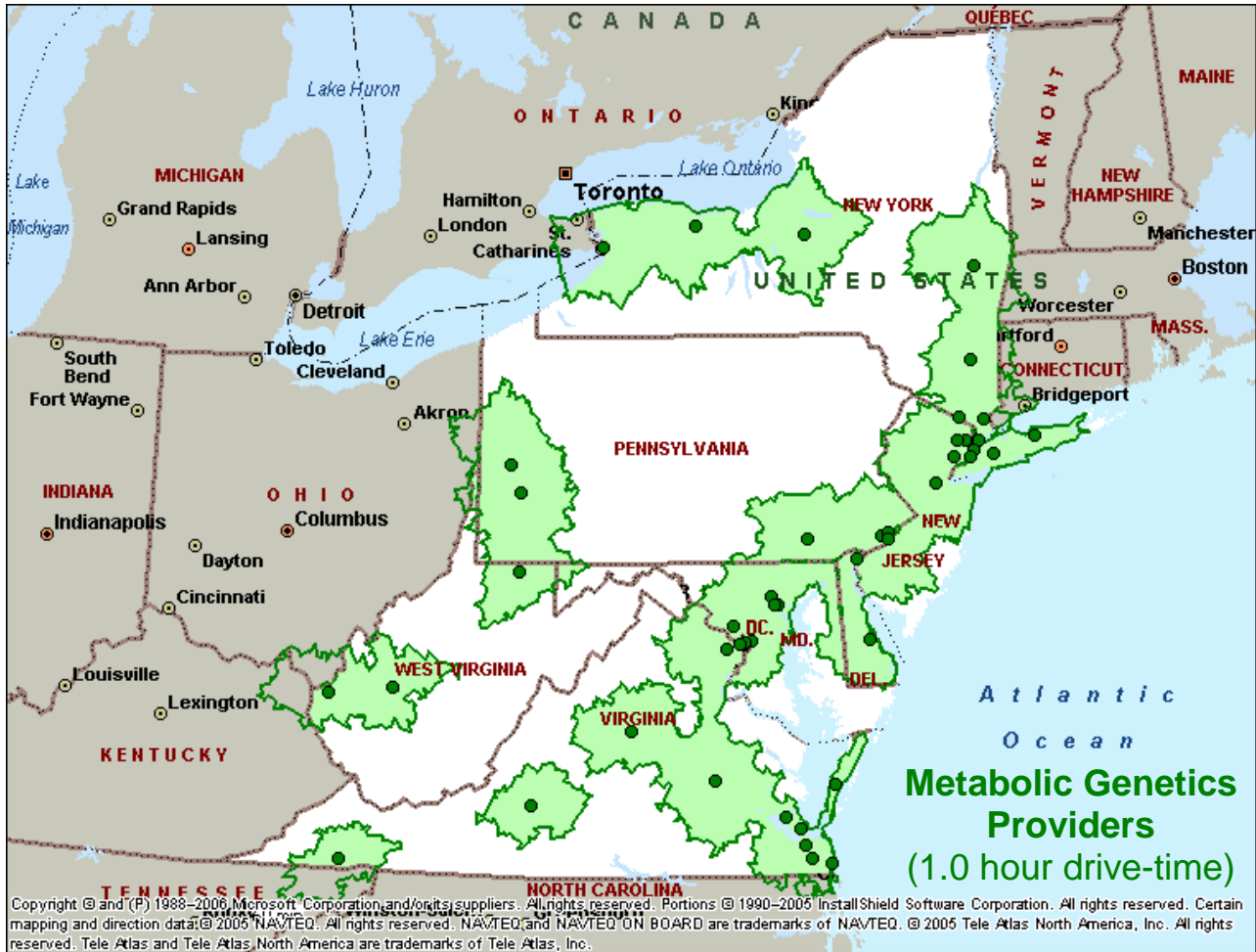
Infants at risk for any of the conditions in the recommended blood-spot panel are generally referred for diagnosis and management to up to eight groups of specialty care centers – Metabolic (metabolic diseases), Hematology (sickle cell disease, other hemoglobinopathies and thalassemias), Endocrinology (congenital hypothyroidism and congenital adrenal hyperplasia), Pulmonary/Respiratory (cystic fibrosis), Neurology (Krabbe disease/lysosomal storage diseases), Immunology (Severe Combined Immunodeficiency, HIV), Cardiology (critical congenital heart disease) and Audiology (hearing deficits). In addition, children with genetic diseases and their parents need access, both prenatally and postnatally, to comprehensive, noncategorical clinical genetics services.

All the jurisdictions have specialty care centers for each newborn screening specialty. In addition most of the tertiary care medical centers can meet the specialty care and developmental needs of all children with special health care needs. (See the NYMAC Google map <http://maps.google.com/maps/ms?ie=UTF8&hl=en&msa=0&msid=106181927411964196664.0004764de2695b00efc85&z=6>) In every state it is recognized that some families do or must routinely seek specialized medical care in a neighboring state.



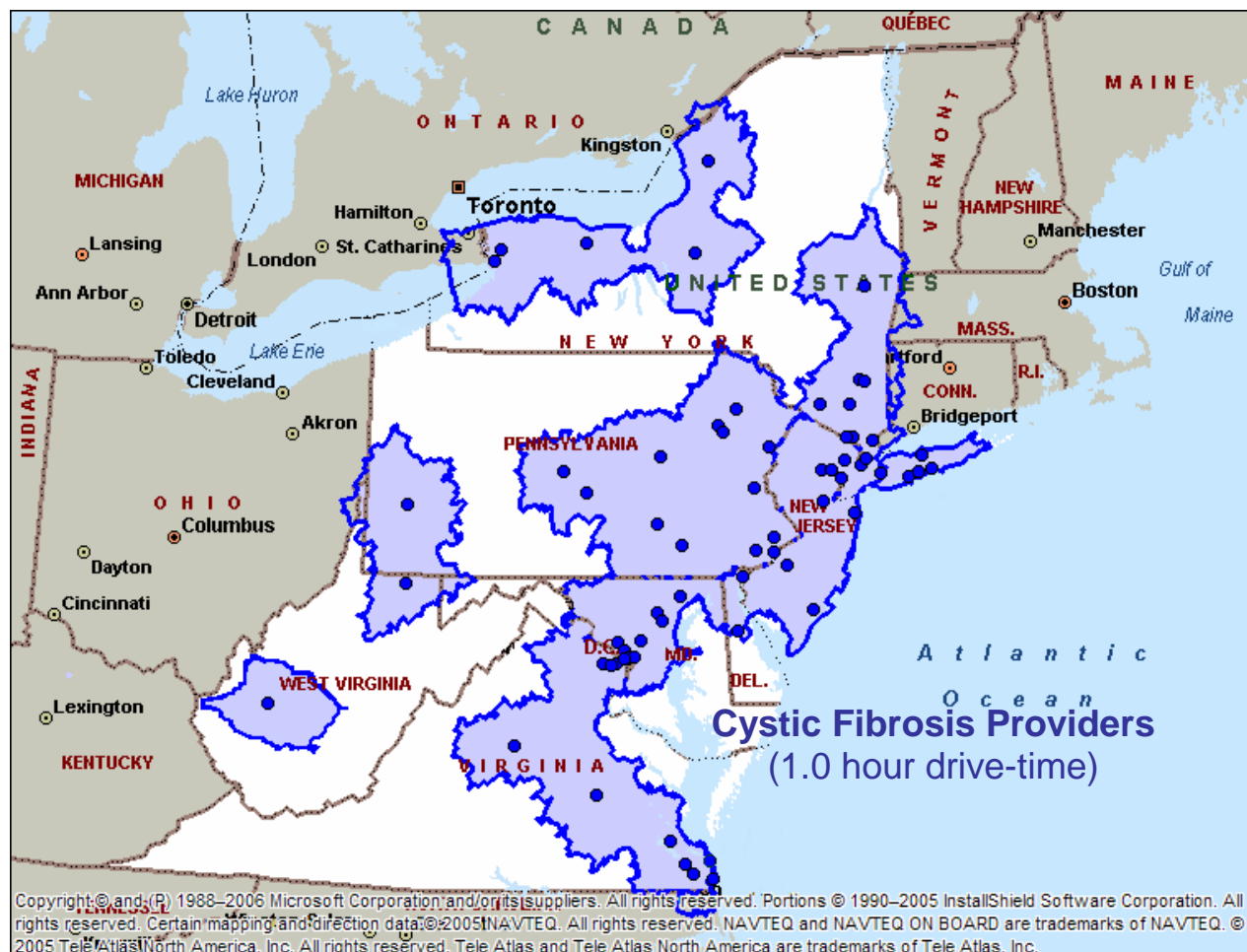
●	NYMAC Clinical Non-categorical Genetics Centers
	1.5 hour Drive time

This NYMAC map of clinical genetic centers shows that most areas of the region are within an hour-and-a-half travel time (clinical genetic services are understood to be sought for a specific situation and not necessarily needed on a regular and on-going basis). That said there are still areas of upstate New York, central western Pennsylvania, southeastern West Virginia and the western half of Virginia where clinical genetic services are not easily accessible.



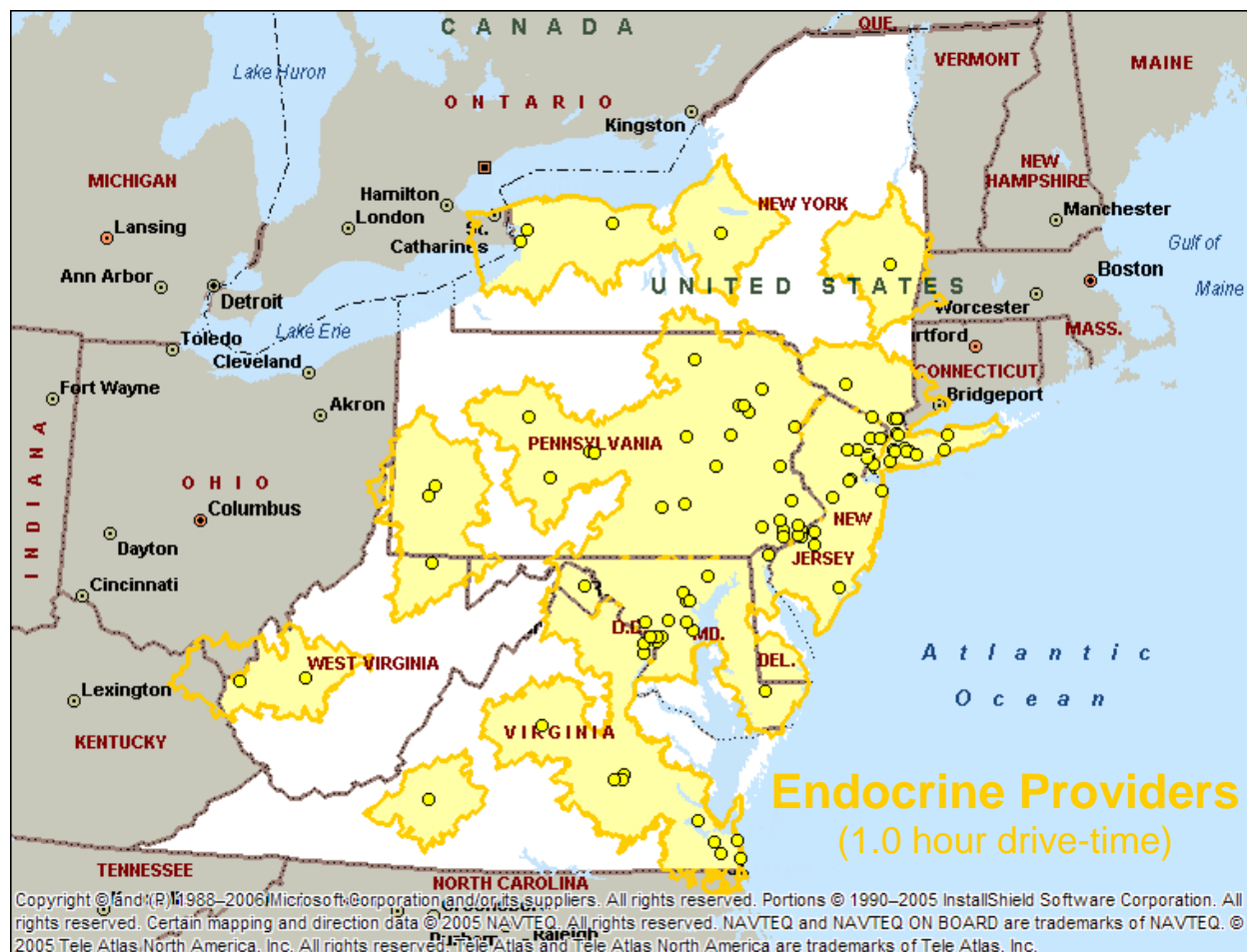
●	NYMAC Metabolic Specialty Care Centers
■	1.0 hour drive time

This map of the metabolic specialty care centers shows significant gaps in geographic accessibility for services in all states but Delaware, New Jersey and the District of Columbia. More than two-thirds of the area is more than an hour travel time from specialty care centers. It is understood that metabolic specialty care is needed by many patients and their families on a regular basis and at times in emergency situations. A one-way hour drive-time seems reasonable for most situations.



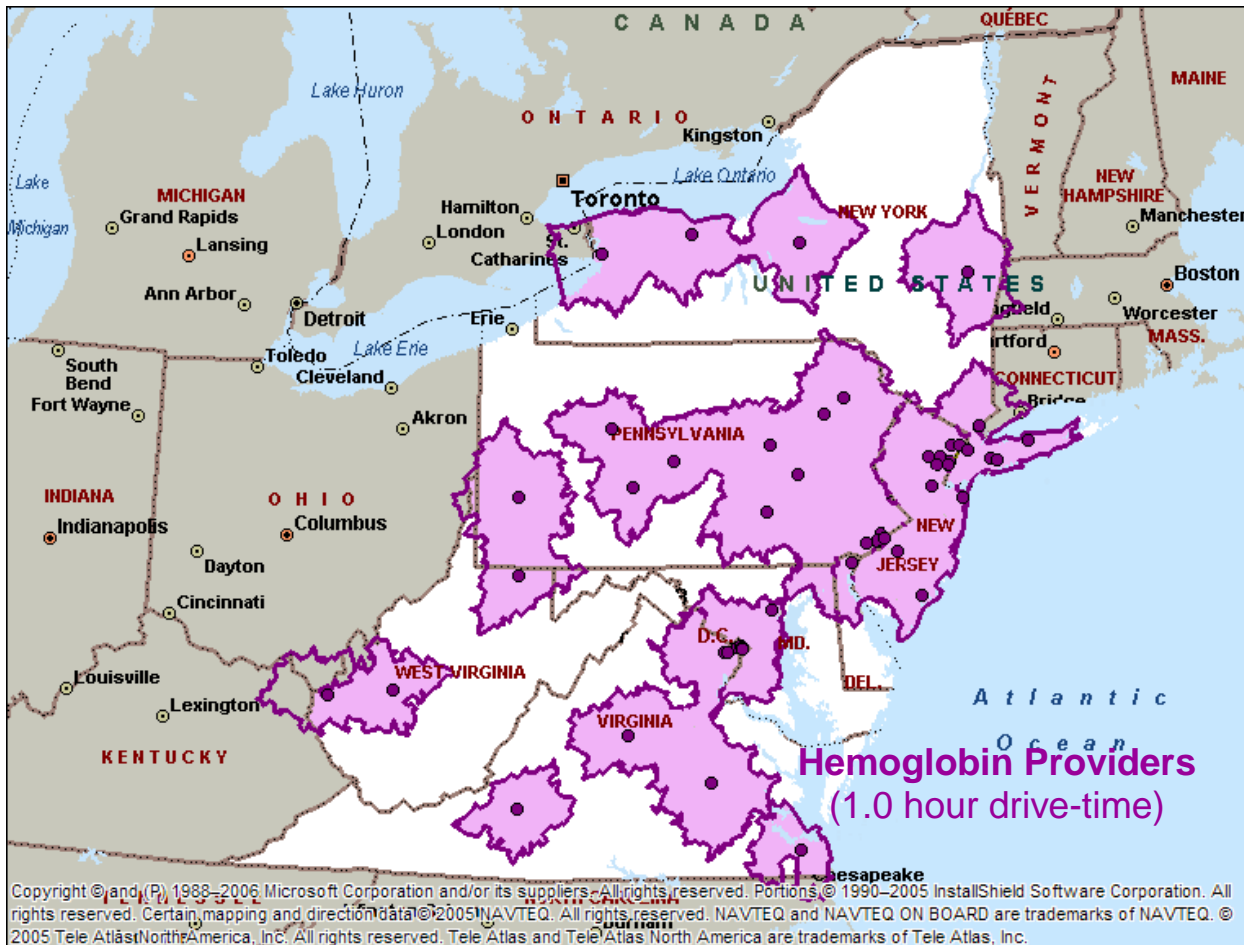
●	NYMAC Cystic Fibrosis Specialty Care Centers
□	1.0 hour drive time

This map of the cystic fibrosis specialty care centers shows significant gaps in geographic accessibility for services in all states but New Jersey and the District of Columbia. More than half of the area is more than an hour travel time from specialty care centers. It is understood that cystic fibrosis specialty care is needed by many patients and their families on a regular basis and at times in emergency situations. A one-way hour drive-time seems reasonable for most situations.



●	NYMAC Endocrine Specialty Care Centers
■	1.0 hour drive time

This map of the endocrine specialty care centers shows significant gaps in geographic accessibility for services in all states but Delaware, New Jersey and the District of Columbia. About a third of the area is more than an hour travel time from specialty care centers. It is understood that endocrine specialty care is needed by many patients and their families on a regular basis and at times in emergency situations. A one-way hour drive-time seems reasonable for most situations.



●	NYMAC Hemoglobinopathy Care Centers
■	1.0 hour drive time

This map of the endocrine specialty care centers shows significant gaps in geographic accessibility for services in all states but Delaware, New Jersey and the District of Columbia. More than half of the area is more than an hour travel time from specialty care centers. It is understood that hemoglobinopathy specialty care is needed by many patients and their families on a regular basis and at times in emergency situations. A one-way hour drive-time seems reasonable for most situations.

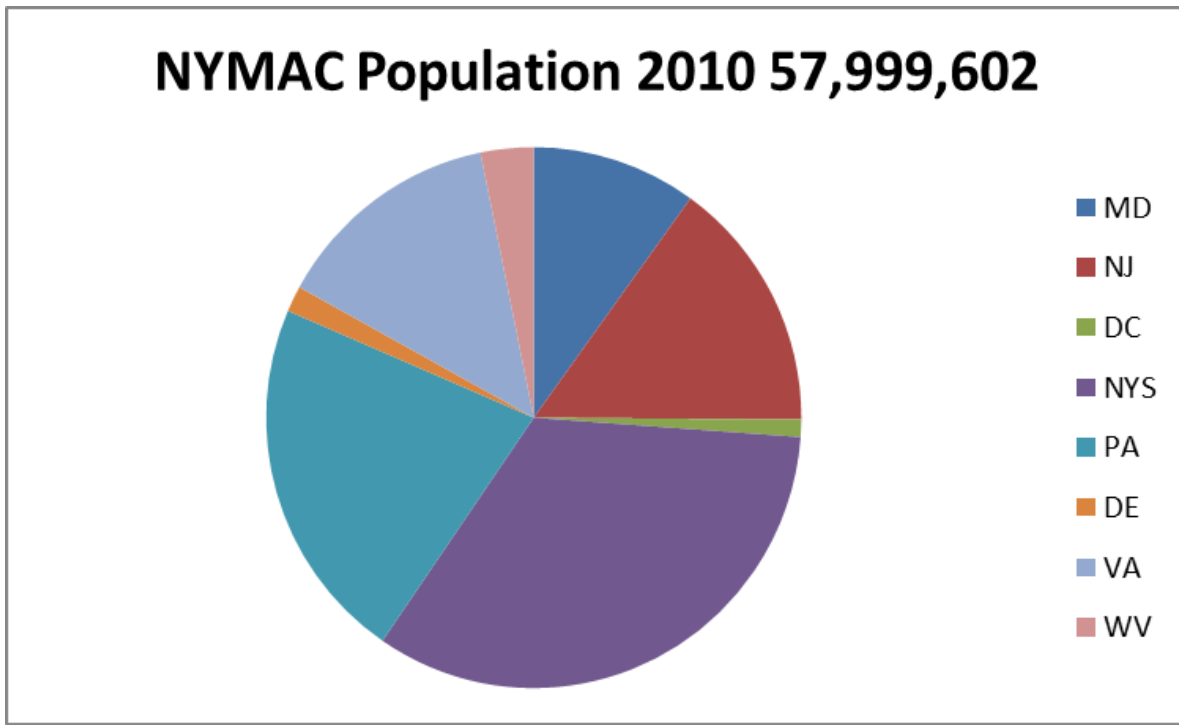
Demographics of the New York – Mid-Atlantic Region

There are eight states in the New York – Mid-Atlantic Region (Region 2). See the pie charts for the populations of the eight jurisdictions and the number of live births. See the table below for number and percentage of infants born by race and Hispanic ethnicity in 2009.

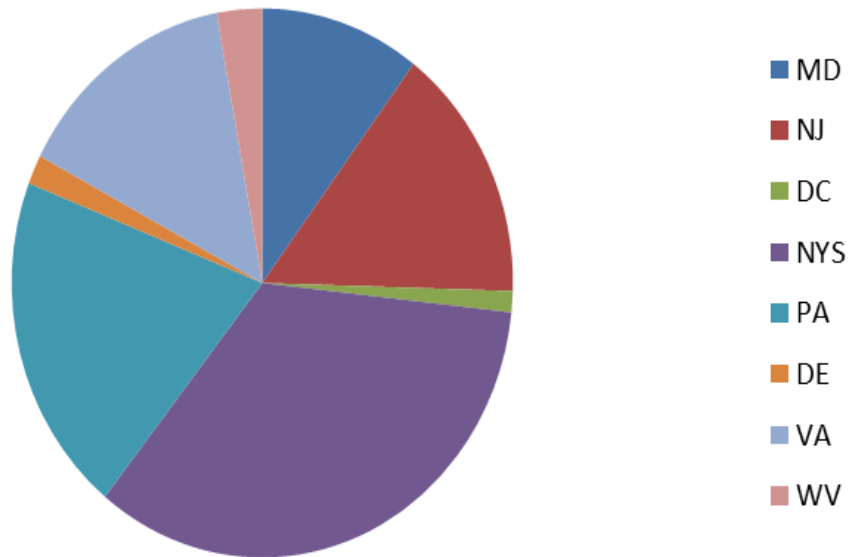
NYMAC Region Live Births 2009							
	Non-Hispanic White	Non-Hispanic Black	Non-Hispanic American Indian	Non-Hispanic Asian or Pacific Islander	Hispanic	Not Stated	Total

Delaware	6,183	3,178	22	517	1,648	14	11,562
District of Columbia	2,344	4,720	28	406	1,510	36	9,044
Maryland	34,014	24,992	201	5,388	10,612		75,061
New Jersey	52,161	17,131	179	11,668	29,003	182	110,324
New York	119,530	40,982	748	23,274	59,791	3785	248,110
Pennsylvania	103,302	21,482	413	6,416	14,115	704	146,432
Virginia	60,404	23,021	179	7,780	13,688		105,056
West Virginia	19,962	831	20	146	231	80	21,270
Total	397,900	136,337	1,790	55,595	130,598	4,801	726,859

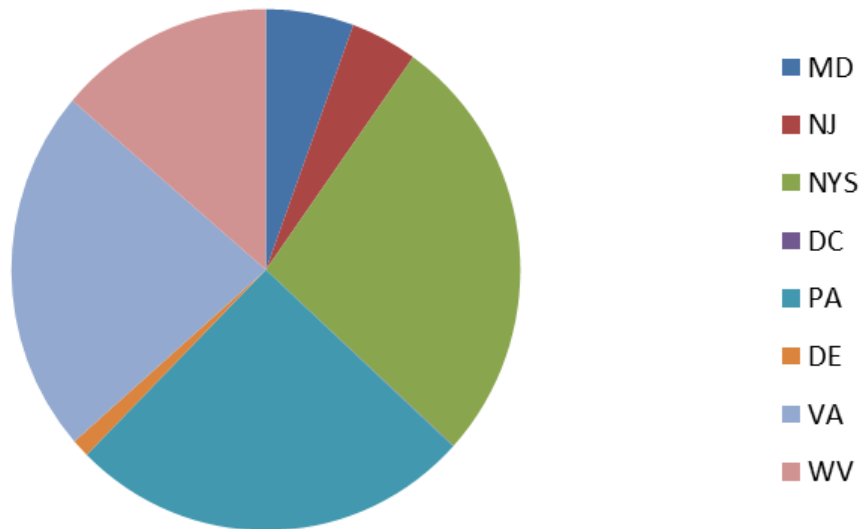
	Non-Hispanic White	Non-Hispanic Black	Non-Hispanic American Indian	Non-Hispanic Asian or Pacific Islander	Hispanic	Not Stated	Total
Delaware	53.5%	27.5%	0.2%	4.5%	14.3%	0.1%	100.0%
District of Columbia	25.9%	52.2%	0.3%	4.5%	16.7%	0.4%	100.0%
Maryland	45.3%	33.3%	0.3%	7.2%	14.1%	0.0%	100.0%
New Jersey	47.3%	15.5%	0.2%	10.6%	26.3%	0.2%	100.0%
New York	48.2%	16.5%	0.3%	9.4%	24.1%	1.5%	100.0%
Pennsylvania	70.5%	14.7%	0.3%	4.4%	9.6%	0.5%	100.0%
Virginia	57.5%	21.9%	0.2%	7.4%	13.0%	0.0%	100.0%
West Virginia	93.9%	3.9%	0.1%	0.7%	1.1%	0.4%	100.0%
NYMAC	54.7%	18.8%	0.2%	7.6%	18.0%	0.7%	100.0%



NYMAC 2009 Live Births 727,842



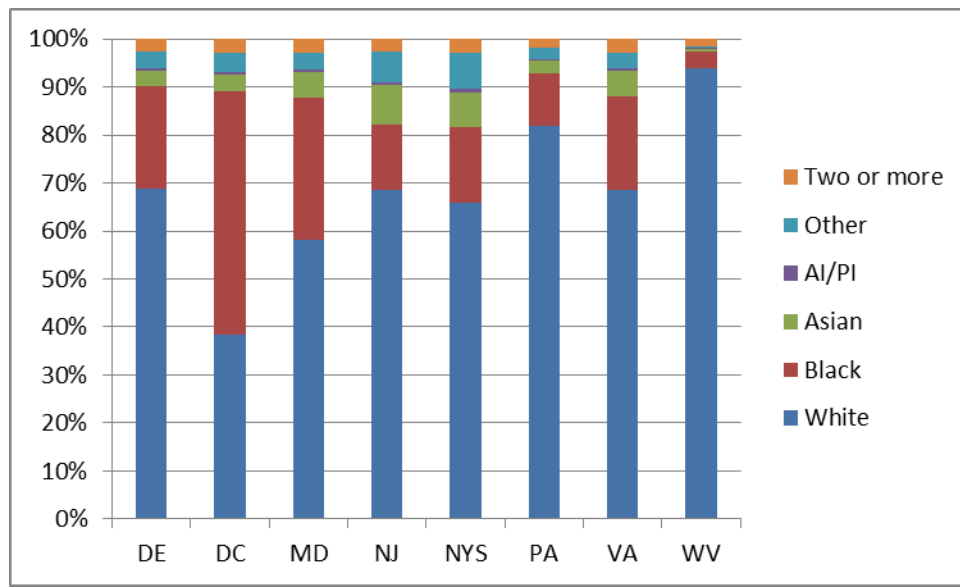
NYMAC Land Area 175,011 sq mi



The population of the region varies widely among the states. Over 50% of the residents live in New York and Pennsylvania. Nearly 40% more live in Maryland, New Jersey and Virginia. The remaining 6% reside in Delaware, the District of Columbia and West Virginia. The number of live births reflects this same dichotomy. As for land area, New York, Pennsylvania and Virginia are of comparable size, with the other four states comprising only 25% of the area. The District

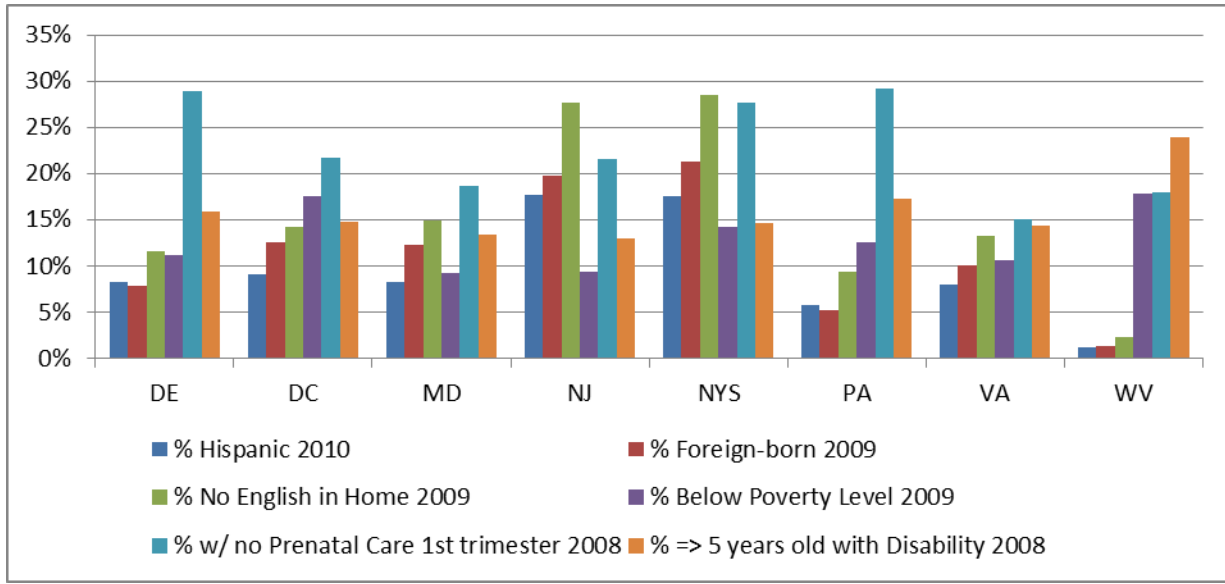
of Columbia has negligible land area. Population densities range from 27,402 persons per square mile in New York City and 9,864 people per square mile in the District of Columbia, to 1,185 in New Jersey, 460 in Delaware, 410 in Maryland, 283 in Pennsylvania, 236 in upstate New York, 202 in Virginia and 86 in West Virginia. In the larger states, there are vast regions that are rural and less-populated. Three of the states, New York, West Virginia and Virginia include mountain ranges that greatly impede access to services, especially during inclement weather events. These two issues suggest barriers to care for those people living in the rural areas, far from urban areas where specialty services are available.

NYMAC Racial Mix



This table shows the differences within the region in terms of racial mix. West Virginia is quite homogeneous with a 94% White population. District of Columbia, on the other hand, is only 39% White, with Blacks making up 51%. The other states have comparable white population percentages with Blacks, Asians and Others comprising varying proportions within their populations.

Other Characteristics of NYMAC's Population

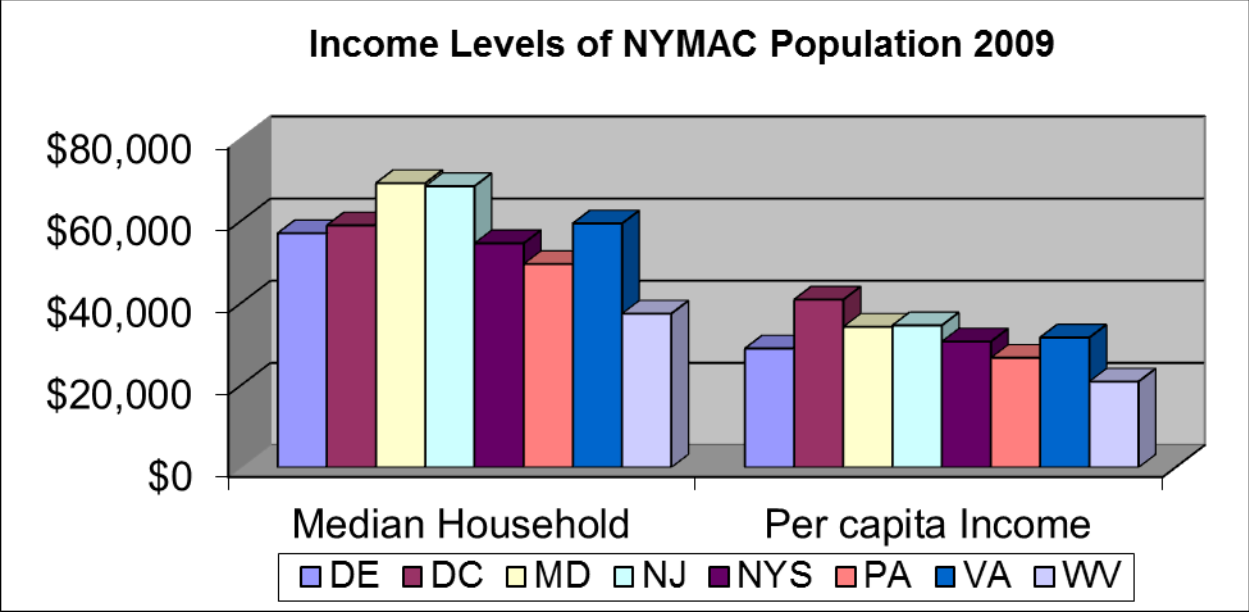


Other Characteristics of NYMAC's Population

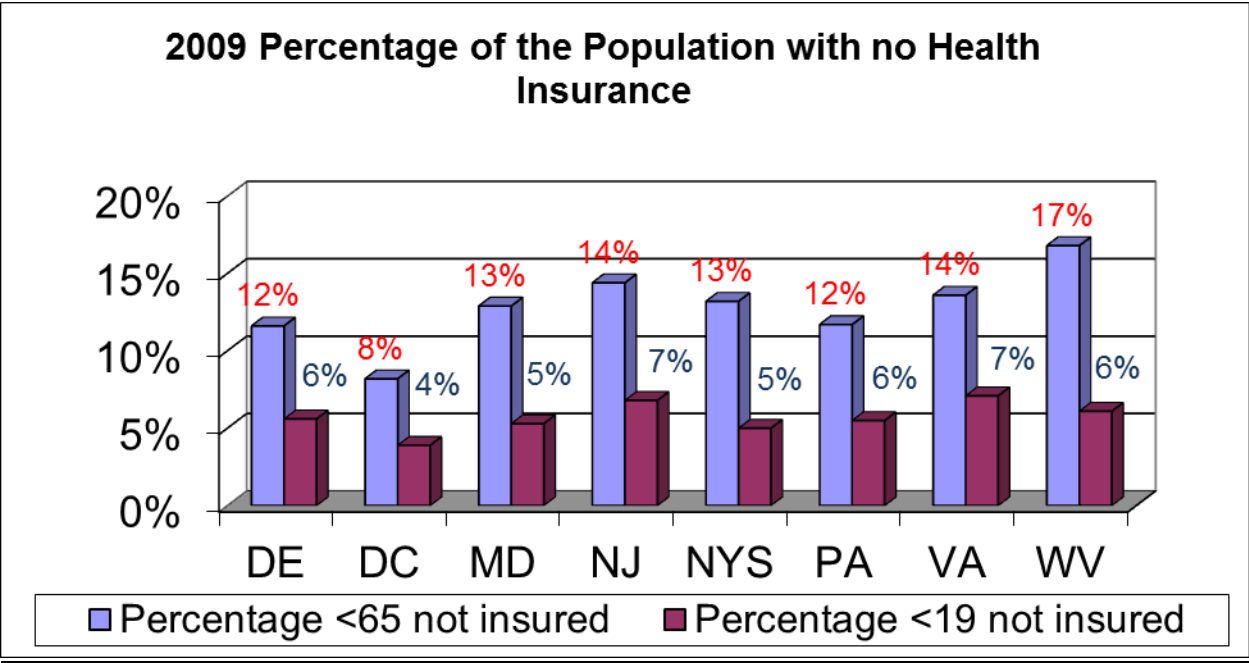
	DE	DC	MD	NJ	NYS	PA	VA	WV
% Hispanic 2010	8%	9%	8%	18%	18%	6%	8%	1%
% Foreign-born 2009	8%	13%	12%	20%	21%	5%	10%	1%
% No English in Home 2009	12%	14%	15%	28%	29%	9%	13%	2%
% Below Poverty Level 2009	11%	18%	9%	9%	14%	13%	11%	18%
% w/ no Prenatal Care 1st trimester 2008	29%	22%	19%	22%	28%	29%	15%	18%
% => 5 years old with Disability 2008	16%	15%	13%	13%	15%	17%	14%	24%

The percentage of Hispanics is quite variable, being highest in New Jersey and New York at 18% each. The number of foreign-born residents and the number of households where English is not spoken, likewise, is highest in New Jersey at 20% and 28% and New York at 28% and 29%. These numbers suggest that any outreach and education about genetics, newborn screening and health care in general must reach peoples with widely different experiences.

Delaware and Pennsylvania has the highest percentage of women who receive no prenatal care in the first trimester at 29%, with New York at 28% each. All the states have disability rates between 13% and 24%. DC and West Virginia have the highest percentage of people below the poverty level at 18% each.



Statewide median household and per capita income are relatively consistent within the region, except, perhaps, for West Virginia which is significantly lower in both categories. That said, there are wide variations within each state with very wealthy neighborhoods and desperately poor ones. Access to health care will vary widely across the income spectrum.



The states in the region have similar percentages of uninsured, between 8% and 17% for all people under 65 years of age and between 4% and 7% for children less than 19 years of age. While most specialty care centers are in medical centers that serve their communities regardless of patients' abilities to pay, access to prescriptions, medical foods, supplies, ancillary care and

specialists is affected by insurance status. Other barriers to care – educational level, language, transportation, for example – are more likely issues for those without medical insurance.

Available Resources in Region

There are tremendous resources within the Mid-Atlantic Region. All jurisdictions participate in the federal Medicaid and Child Health Plus programs, receive Title V (Maternal and Child Health Block Grant) funds and have well-established newborn screening programs, follow-up protocols, and a system of specialty care centers for care of those children identified by the program with a genetic or other congenital disorder. There is public and private health insurance available in each jurisdiction; primary medical care providers, focusing on pediatrics, obstetrics and gynecology and family medicine can be found throughout the region. Medical and public health professionals dedicated to the needs of their populations are found in every jurisdiction.

NYMAC’S REGIONAL GENETIC PLAN

NYMAC’s Role in Meeting Needs

The vision of NYMAC is that individuals with heritable disorders and their families have access to quality care and appropriate genetic expertise and information. At a strategic Planning Meeting in October 2009 attended by Advisory Council members, Work Group chairs and other involved NYMAC members, NYMAC dissolved its work groups and is presently working with designated champions to address problems and issues identified at the Meeting. In the planning process for the 2012 non-competitive continuation application, many NYMAC constituents participated in conference calls, provided e-mailed ideas and made separate phone calls. NYMAC will continue to operate with champions assisted by NYMAC staff. See the table below for current projects, champions and NYMAC staff facilitators:

See the attached table of Strategic Goals and Objectives with current and proposed projects, including champions and NYMAC staff facilitators

NYMAC CENTRAL ADMINISTRATION EFFORTS

Vision: Individuals with heritable disorders and their families have access to quality care and appropriate genetic expertise and information. This should be provided in the context of a medical home that provides accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally-effective care.

Central administrative efforts are designed to support the overall efforts of the regional collaborative and its work groups and to maintain a regional perspective for implementing and institutionalizing its efforts. Central administration efforts also focus on inter-regional and national collaborations, while enabling communication among participating entities. Many NYMAC work groups have their counterparts at the National Coordinating Center and within other regions, so communication is especially crucial to enable each group to learn and benefit from the efforts of the comparable group. Administration strives to identify similar and complementary efforts.

GOAL 1: Ensure communication among project principles and staff

Current projects

- Hold biweekly staff conference calls

GOAL 2: Elicit input from advisors (Advisory Council)

Current projects

- Hold annual Advisory Council meetings
- Hold Ac conference calls every two months
- Identify new Advisory Council members as needed

GOAL 3: Facilitate Project Activities

Current projects

- NYMAC Staff assigned to all projects to work with the Champion to ensure that the projects proceed at an acceptable rate
- Hold Project conference calls bimonthly or as needed
- Establish and review Project progress

GOAL 4: Monitor and participate in activities of the Advisory Council on Hereditary Diseases in Newborns and Children

Current projects

- Attend ACHDNC meetings and conference calls
- Participate in ACHDNC activities
- Participate in ACHDNC Subcommittees on Long-term Follow-up and Treatment, Laboratory Standards and Procedures and Education and Training

GOAL 5: Coordinate with HRSA/MCHB

Current projects

- Work collaboratively to insure that NYMAC meets expectations
- Respond to requests for assistance, collaboration and information

GOAL 6: Work with the National Coordinating Center and other Regional Collaboratives, the National Newborn Screening and Genetics Resource Center and the Newborn Screening Translational Research Network

Current projects

- Participate in all NCC and NCC WG meetings, activities and conference calls
 - Work Group members in: Long-term Follow-up and Data Collection (Louis Bartoshesky, Katharine Harris); Emergency Preparedness (Kenneth Pass – co-chair, Barbara Eckerd, Katharine Harris); Evaluation (Katharine Harris, Susanna Ginsburg); Medical Home (Robert Ostrander); NBSTRN (Steering Committee: Jane Getchell, Kenneth Pass); State Policy: Natasha Bonhomme (Genetic Alliance), Alan Fleishman (MOD), Jane Getchell, Christopher Kus (ASTHO and AMCHP), Cindy Pellegrini (AAP), Sharon Terry (Genetic Alliance), Ann Willey; Planning Meeting (Natasha Bonhomme (Genetic Alliance), Michele Caggana, Jerry Vockley), Regional Managers (Katharine Harris), Telegenetics (Luba Djurdjinovic) and Transition (Louis Bartoshesky – co-chair)
- Identify inter-regional collaborations, sharing of resources, and building on accomplishments
- Submit articles to the NCC Collaborator and sharing quarterly issues with the members of the region
- Submit Evaluation forms annually
- Participate in NCC/MCHB quarterly conference calls

GOAL 7: Manage subcontracts

Current and planned subcontracts

- AI duPont Hospital/Nemours – Patient and Family Coordinator
- SG Associates – Evaluation Consultant
- Genetic Alliance subcontract – Update and modify Understanding Genetics with NYMAC region information
- Asian Outreach and Education
 - Cornell/Weill Medical Center
 - New York Methodist Hospital
 - University of Medicine and Dentistry of New Jersey
- Electronic Personal Health Record
 - Albany Medical College
 - Research Foundation of SUNY at Stony Brook
 - Children’s Foundation at Westchester Medical Center
- Medical Home Collaboration among Specialists, Primary Care Providers and Parents
 - Albany Medical College
 - Brookdale University Hospital and Medical Center

- University of Maryland School of Medicine
- Participation in the Region 4 IBEM-IS Project
 - Hackensack University Medical Center
 - University of Buffalo
 - University of Pittsburgh
 - University of Rochester
- Transition Navigators
 - AI DuPont Hospital for Children
 - Children's Research Institute
 - University of Maryland
- Consumer Outreach and Education
 - CARES Foundation
 - Genetic Alliance
 - Hunter's Hope Foundation
 - Immune Deficiency Foundation
 - Sickle Cell Association of New Jersey
 - Sickle Cell Thalassemia Patient Network
- Other subcontracts as NYMAC and Work Groups develop projects

GOAL 8: NYMAC Core Activities

Current projects

- Maintain appropriate staff
 - Project Coordinator
 - Patient and Family Coordinator
 - Evaluation Consultant
- Maintain NYMAC Website
 - Include NYMAC products
 - Add resource links
 - Promote website
- Develop and maintain NYMAC Resource Directory
 - Regional Specialty Care Centers for genetics, metabolic diseases, endocrine disorders, cystic fibrosis and hemoglobinopathies
 - Using Google Maps for Directory:
<http://maps.google.com/maps/ms?ie=UTF8&hl=en&msa=0&msid=106181927411964196664.0004764de2695b00efc85&z=6>
- Assist in Region 4 MS/MS Project
 - Encouraging participation of all states in data submission 7 states currently or soon-to-be participating
 - Facilitating participation in Region 4 MS/MS Project meetings
 - Facilitating training of MS/MS technologists at Mayo Clinic
- Emergency response plans in the 8 states of the region and in the other regions
 - Monitor the development and maintenance of plans by NBS programs
 - Monitor NBS program responses to real emergency situations
 - Assist NBS programs to evaluate and amend responses

- William Perry, under contract with the ACMG, and Hans Andersson, co-PI for Region 3, will present COOP for clinical service programs at the May 2009 Advisory Council meeting
- Hold a seminar for specialty care center directors about clinical emergency preparedness in August 2010. Information will be presented by William Perry and Hans Andersson.
- Hold a Table-top Specimen Exchange Exercise 12/11
- Paired states to do actual specimen exchange 2012

GOAL 9: Evaluate NYMAC Activities within Region and within NCC/HRSA

Current projects

- Develop NYMAC's vision, goals and activities within a logic model that clearly indicates progress in achieving defined goals
- Develop NYMAC WGs' visions, goals and activities within a logic model that clearly indicates progress in achieving defined goals
- Conduct periodic evaluation of NYMAC activities and overall NYMAC progress in achieving objectives
- Develop and reviewing NYMAC Regional Genetic Plan
- Assist in development of NCC priorities
- Submit NCC/MCHB evaluation forms annually

Other Projects

- Use the *Understanding Genetics* Manual as an educational tool
- Develop and update lists of consumer groups
- Update consumer resource directory for genetic services
- Identify appropriate web sites to be posted on the NYMAC website
- Develop and implement a market strategy for consumers' use of the website
- Develop, pilot test and disseminating *Genetics and Your Health* brochures
 - a. Three NYMAC genetic counseling master's level programs identified students who worked collaboratively to design and implement a study plan and survey to evaluate the use and effectiveness of these brochures
 - b. Howard University, Virginia Commonwealth University, Sarah Lawrence College
 - c. A recently-graduated student from Arcadia University translated the brochures into Spanish for use by one of the students
 - d. Publish the results
- Participate in meetings and conferences to increase awareness about NYMAC in the region and to facilitate NYMAC's mission
- Develop contacts with Parent/Family/Advocacy groups
- Develop a telemedicine model for areas in need of genetics services including potential piloting in federally qualified health centers
- Disseminate maps of underserved areas to support identification of needed resources

- Develop an approach and pilot to address formalizing medical homes for children diagnosed-positive through newborn screening
- Develop directories of resources for providers/consumers/community organizations (see consumer workgroup)
- Fund 3 projects at Albany Medical Center, Brookdale University Hospital and Medical Center and University of Maryland School of Medicine to use care plans, emergency cards, fax-back forms and other tools to improve communication among the primary health care provider (PCP), the specialty care provider (SCP) and the family. This includes aggressive monitoring of billing and reimbursement activities to evaluate the viability of these activities.
- Encourage, and fund if appropriate, implementation of medical home systems and tools between SCPs and the PCPs for infants who are newly diagnosed by the newborn screening programs.
- Improve the diagnostic process including confirmation of diagnosis, specificity of the screen and reduction of false-positives by developing and disseminating a diagnostic evaluation tool
- Support improvements in reimbursement for testing and evaluation by identifying reimbursement issues and strategies to address them
- Evaluate NBS lab cut-off values
- Standardize of NBS metabolic lab screening
- Examine NBS follow-up protocols
- Develop and disseminate laminated cards to be carried by patients and their families for use during a medical emergency
- Conduct a pilot to examine the use of personal health records in clinical settings
- Pilot test an educational program for young adults transitioning into adult care at Children's Hospital of Pittsburgh
- Develop a pilot on initiating transition project

SUMMARY

Projects and other activities in the NYMAC region both of the central administration and of the work groups have been and will continue to be developed to address issues identified in the Needs Assessment. Examination of needs will continue. Evaluation of completed projects will lead to new activities. Input from NYMAC staff, members of the Advisory Council, NYMAC Work Groups members and other participants will be sought to further define needs and possible solutions.