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Current Status of Genetic Service Delivery

Innovations in Service Delivery in the Age of Genomics

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Goal today: Set the stage for a bigger and broader discussion re: genetic services in the U.S.

- What is the scope of current service delivery?
- Who provides what services, where, how & to whom?
- Where are the gaps in our understanding of the delivery system?

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Genetic Services Policy Project

- DHHS/MCHB/Genetic Services Branch Cooperative agreement –
  - How are genetic services delivered?
  - Are there alternative models that better promote cost effective, accessible, and equitable delivery of services?
  - What's on the horizon?
  - What public policies do we need to get from here to there?

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State Genetics Profiles Data

- Socioeconomic/Political Variables
  - Population size, % Pop. Under 200% of FPL, Medicaid expenditures, urban rural status, Party of sitting Governor
- Genetic Services Capacity Variables
  - Varied genetics providers per capita, medical schools, genetics training programs, NBS programs, state genetics plan/implementation grant, FTE State Genetics Coordinator

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State Genetics Profiles Data Continued…

- Legal/Regulatory Variables
  - Privacy statues, insurance or employment discrimination statutes, GC licensure, other

- Data compiled for all 50 states (completed Summer 2005)

What is the scope of current service delivery?

Definitions:

- Genetic Services = genetic testing, diagnosis of genetic conditions, genetic counseling and treatments for individuals with, or at risk of, genetic disorders.

- Genetic testing = lab analysis for DNA, RNA, chromosomes, or gene products.

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What is the scope of current service delivery?

- Includes clinical services, cognitive services and all types of genetic tests *except*
  - Genetic analysis of pathogens
  - "Recreational" genetics (e.g., ancestry, dating services, personal investigations)
  - Paternity testing
  - Forensics

What is the scope of current service delivery?

Preconception: *What is "our" risk of having an affected child?*

Genetic counseling

Carrier test

Predisposition / susceptibility test

Diagnostic test

Prenatal: *How will I manage my pregnancy?*

Genetic counseling

Carrier test

Predisposition / susceptibility test

Diagnostic test

ART

Pre-implantation genetic diagnosis

Prenatal testing

Newborn, Pediatric, and Adult: *How might my genetics affect my health?*

Clinical evaluation

Genetic counseling

Carrier test

Predisposition / susceptibility test

Diagnostic test

Pharmacogenetics

Gene therapy

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Who provides services?

- Potentially all health care providers!
- Two categories -
  - Those formally trained & certified in genetics
    - Clinical geneticists (physicians, genetic technologists, state public health labs, nurses & genetic counselors)
  - All others
  - Service utilization = DATA Gap!

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Concerns about Genetic Services Provided by Non-Geneticists

- 71% of physicians rated their knowledge of genetics and genetic testing as poor and almost all would refer to a genetic counselor.
  Mt. Sinai J of Med 2001. 67(2): 144-51.

- Physicians misinterpreted the results of genetic testing in 31% of cases.
  NEJM 1997. 336(12):823-7

- Surveyed physicians rated genetic counseling information as important on genetic test reports. *Genetics in Medicine 2003. 5(3): 166-71.*

- *42% of MDs surveyed in Massachusetts indicated feeling ill prepared to talk about expanded NBS test results with patients. J. Inherit Metab Dis 2005. 28 : 819-24*

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Who receives services?

- Historic focus on single gene disorder and/or birth defects
- Increasingly utilized by individuals/families receiving information from other specialists (e.g., oncologists, neurologists, cardiologists, ENTs/audiologists, etc.)
- Service utilization = DATA Gap!

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What can we infer about who receives services?

- Nearly all ~4.1 million infants receive NBS (1)
- 14% women 35 y.o./older offered amnio/CVS (2)
- Estimate 600,000 younger women also undergo prenatal screening (3)
- Pediatric and adult utilization in 9 WA RGC's grew an average of 8%/yr between 1995-2004
  - Primarily adults: 29% in 1995 vs 40% 2004 (4)
- DTC uptake "low" (5)

(1) National Newborn Screening & Genetics Resource Center 2006

(2) Centers for Disease Control and Prevention 2006

(3) Rabin et al., 2007

(4) Wang and Watts, 2007

(5) personal communication

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Where are services provided?

- Academic Medical Centers
- Private and public hospitals/medical facilities including HMOs
- Commercial labs, Diagnostic labs, State labs, Biotech
- Private practice
- Insurance industries
  - Note DATA limitation: listed in order of magnitude although professional organizations categorize differently

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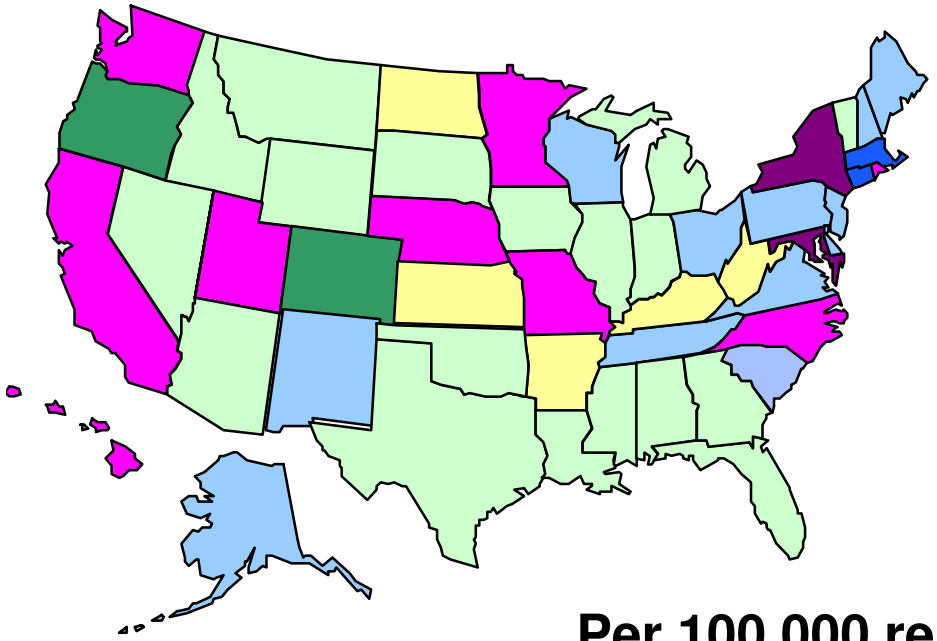
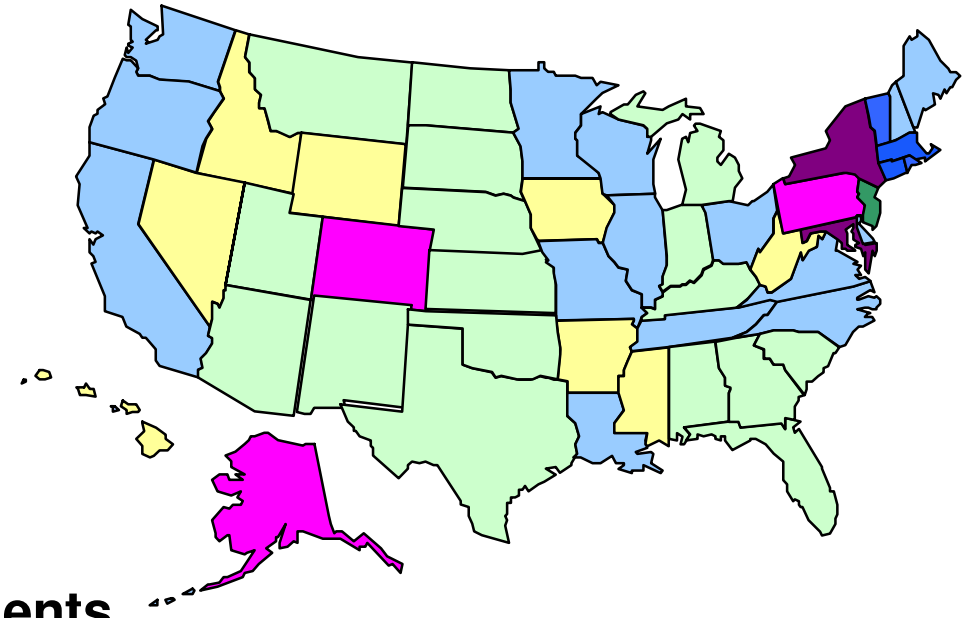
Genetics Providers per capita*



All MDs per capita*



Per 100,000 residents

- 0.00 – 0.50
- 0.51 – 1.00
- 1.01 – 1.50
- 1.51 – 2.00
- 2.01 – 2.50
- 2.51 – 3.00
- 3.01 – 4.00

- 160 - 195
- 196 - 231
- 232 - 267
- 268 - 303
- 304 - 339
- 340 - 375
- 3.01 – 4.00

* 2004 data

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Genetic Capacity Indicators - Correlation Results:

- Total, Biochemical, Molecular and Cytogeneticist highly correlated (r^2.53-.67)

- Combined biochemical/molecular, nurses (APNG & GCN) not correlated

- More likely to see an increase in all provider types given the presence of one, with the exception of nurses or the combined MD certificates.

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Where are the gaps in our understanding of the delivery system?

- **Systematic data collection or published reports on service utilization/expenditures (other than NBS)**
  - Claims data from Payers– including outpatient services - may be useful to reflect services rendered by non-genetics and genetics professionals and possibly self-referrals to retail genetics but pose significant limitations
    - Data are often proprietary hence not available or costly
    - GREAT variability in use of CPT and ICD – 9 codes used hence masking services and making difficult to compare/contrast

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Where are the gaps in our understanding of the delivery system?

- Data analyzed revealed generalized low levels of certified genetic service providers nationally yet NO data to indicate actual or estimated numbers needed for optimal access and/or quality

- Very little data to demonstrate consumer demand/utilization of retail genetics (marketing either DTC or DTP)

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