Current Status of Genetic Service Delivery

Innovations in Service Delivery in the Age of Genomics

Debra Lochner Doyle, MS, CGC

July 28, 2008

Debra.lochnerdoyle@doh.wa.gov
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?
• 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?
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
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.
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?

<table>
<thead>
<tr>
<th>Preconception: What is “our” risk of having an affected child?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic counseling</td>
</tr>
<tr>
<td>Carrier test</td>
</tr>
<tr>
<td>Predisposition / susceptibility test</td>
</tr>
<tr>
<td>Diagnostic test</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Prenatal: How will I manage my pregnancy?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Genetic counseling</td>
</tr>
<tr>
<td>Carrier test</td>
</tr>
<tr>
<td>Predisposition / susceptibility test</td>
</tr>
<tr>
<td>Diagnostic test</td>
</tr>
<tr>
<td>ART</td>
</tr>
<tr>
<td>Pre-implantation genetic diagnosis</td>
</tr>
<tr>
<td>Prenatal testing</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Newborn, Pediatric, and Adult: How might my genetics affect my health?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical evaluation</td>
</tr>
<tr>
<td>Genetic counseling</td>
</tr>
<tr>
<td>Carrier test</td>
</tr>
<tr>
<td>Predisposition / susceptibility test</td>
</tr>
<tr>
<td>Diagnostic test</td>
</tr>
<tr>
<td>Pharmacogenetics</td>
</tr>
<tr>
<td>Gene therapy</td>
</tr>
</tbody>
</table>
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!
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


- 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
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!
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
(5) personal communication
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
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.
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
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)
Acknowledgements

- Final report posted: http://depts.washington.edu/genpol/

University of Washington
- Carolyn (Cindy) Watts, PhD
- Grace Wang, PhD candidate
- Rick Carlson, JD
- Anne Doherty, MPH candidate
- Wylie Burke, PhD, MD
- Scott Ramsey, MD, PhD
- Dave Veenstra Pharm D., PhD

WA DOH
- Deb Lochner Doyle, MS, CGC
- Amber Roche, MPH

HRSA
- Michele Lloyd-Puryear, MD, PhD
- Penny Kyler, MPH

And special thanks to the many Advisors and Consultants who shared their expertise and time!

- This work was supported in part by Projects #U35MCO2601 and U35MCO2602 from the Maternal and Child Health Bureau (Title V, Social Security Act), #1126, Health Resources and Services Administration, Department of Health and Human Services