The Genomic Era: A Public Health Perspective

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Objectives

- Summarize the current scope of genomic applications
- Discuss the relationship between public health and genomic applications
- Discuss carrier screening and public health
Genomic applications

- Screening in asymptomatic individuals for genetic pre-disposition for disease
- Screening for reproductive risks
- Screening for acquired disease
- Diagnostic testing for symptomatic disease
- Testing to inform/alter therapeutic approaches (e.g., pharmacogenetics)
- Metabolic/proteomic screening for disorders
IOM: Core public health services

- Assessment (of the health of the community)
- Policy development (to support the health of the community)
- Assurance (that public health goals are met)
Assessment and genomics

Opportunities

» Population-based genetic epidemiology
  – Surveillance
  – Research

» Population-based screening
  – Newborn screening
  – Impact on “medical standard of care”
  – Expansion of public health role in screening?

» Interface with “personalized health care”
HHS Personalized Health Care Initiative

- Provide federal leadership supporting research addressing individual aspects of disease and disease prevention with the ultimate goal of shaping preventive and diagnostic care to match each person’s unique genetic characteristics.

- Create a “network of networks” to aggregate anonymous health care data to help researchers establish patterns and identify genetic “definitions” to existing diseases.
Assessment and genomics

- Challenges
  - Funding for data collection, analysis and maintenance
  - Funding for population-based services
  - Privacy attitudes and laws; personal rights
  - Disconnect between public health and direct medical care services (especially beyond childhood)
Opportunities

» Public policy (and associated resources) can provide population access to important genomics services

» Public policy can impact the medical standard of care (mandated coverage and/or testing)

» Public policy can impact the balance of public and private interests
Challenges

- Impact of advocacy on public policy
- Impact of ideology on public policy
- Impact of market and other economic forces on public policy
- Translation of genomics knowledge into policy
- Gap between genomics science and technology (what we can do) and ethics and wisdom (what we should do)
Assurance and genomics

Opportunities

- Assurance of availability of effective genomics services for individuals
- Assurance of follow-up and treatment for abnormal tests and detected diseases
- Protection from unwarranted use of tests and test results: genomic application regulation and oversight
Assurance and genomics

**Challenges**

- Appropriate availability and use of genomic tests
- Education of providers and the public
- Assurance of accuracy of genomic tests
- Resources to support appropriate delivery of genomics services (pre and post test counseling, testing, interpretation, diagnosis, treatment and follow-up)
- Significant potential for harm (ethical, social, legal; physical health, mental health)
Public health and specific genomic applications

- There is national public health interest and involvement in genomic applications
  - CDC office
  - Evaluation Genomic Applications in Practice and Prevention (EGAPP) Workgroup
- There is great variation in state public health involvement in genomic applications
Genomic applications and public health

- Screening in asymptomatic individuals for genetic pre-disposition:
  - Early in the development stage in public health involvement
  - Few examples of publicly-funded programs
  - Regulatory issues include informed consent, approval of tests, laboratory quality, clinical validity and utility
  - EGAPP and USPSTF involvement in evidence-based recommendation development
Genomics applications and public health

- Screening for reproductive risks
  » Multiple areas of testing:
    – Carrier screening (pre-conception, other)
    – Pre-natal testing for disease or disease pre-disposition
    – Pre-implantation testing for disease or disease pre-disposition
  » Early in the development stage in public health involvement
Genomics applications and public health

- Genetic screening for acquired disease
  - Example: fecal DNA screening for colorectal cancer
  - USPSTF involvement in evidence-based recommendation development
  - Little other public health involvement; little anticipation of a role for public health
Diagnostic testing for symptomatic disease
  » Almost entirely in the realm of health care delivery
  » Little public health involvement; little anticipation of a role for public health
Genomic applications and public health

- Genetic testing to alter therapeutic approaches (e.g., pharmacogenomics)
  - EGAPP involvement in evidence-based recommendation development
  - Little other involvement of public health, little anticipation of a role for public health
Genomic applications and public health

- Metabolic/proteomic screening for disorders
  - Public health is integrally involved in population-based newborn screening
  - Generates interesting policy issues:
    - Mandatory testing vs. voluntary, opt-in vs. opt-out
    - Is information itself an important health outcome?
    - Uniformity vs. local (state, institution) variation
  - HHS/ MCHB/ ACHDGDNC involvement in evidence-based recommendation development for core tests
  - EGAPP involvement in evidence-based recommendation development (for applications other than newborn screening)
Carrier screening and public health

- Currently there is a variable interface between public health and carrier screening
- Most carrier screening is done within the context of direct reproductive health care delivery
- Carrier screening is primarily implemented through the “medical standard of care”, impacted by professional groups (ACOG, AAP, AAFP), insurers, and advocates
Carrier screening and public health

The primary public health objectives for carrier screening would be to reduce the prevalence of the disease and to inform reproductive choices.

Current areas of interface:
- Laboratory oversight
- Informed consent regulations
- Public and provider education

Potential areas of interface:
- Population-based screening
- Policies based on analytic and clinical validity
- Policies based on clinical utility
Future of public health and genomics

- Addition of carrier screening to population-based newborn screening? (already available for cystic fibrosis)
- Public health programs for pre-conception carrier screening?
- “Complete” genetic screening/assessment at birth?
Future of public health and genomics

- Complex policies are needed to provide individual protection and access to services
- Complex policies are needed to assure appropriate use of information to promote health and wellness
- There are significant educational challenges (e.g., risk does not equal disease, all screening has potential for harm, etc.)
- Evaluating the health value of genomic applications and prioritizing these within the larger context of health care services is essential, and will be challenging