

# The Genomic Era: A Public Health Perspective

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# Objectives

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- 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 (eg., pharmacogenetics)
- Metabolic/proteomic screening for disorders

# IOM: Core public health services

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- 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)

# Policy development and genomics

- 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



# Policy development and genomics

- 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

# Genomic applications and 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 (eg., pharmacogenetics)
  - » 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 (eg, 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