

# Health Disparities and the Genetic/Genomic Revolution

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## Our Focus Today

- Current genetic/genomic technology used to improve health
- Not cloning, forensics, newborn screening or paternity testing

# Health Disparities

## Definitions

- Health disparities refer to gaps in the quality of health and health care across racial, ethnic, and socioeconomic groups.

U.S. Department of Health and Human Services (HHS), Healthy People 2010

- Population-specific differences in the presence of disease, health outcomes, or access to health care.

Health Resources and Services Administration

## Who are the Underserved?

- The Uninsured
- The Homeless
- Seasonal Workers
- Minorities
- Recent Immigrants
- Racial & Ethnic Groups
- Forgotten Members of Our Community
- Possibly Your Neighbors

## Uninsured in Michigan

20000 – 11.3% uninsured  
statewide = >1 Million people

Source: U.S. Census Bureau, Small Area Health Insurance Estimates Program - 2005

## Issues

- Access to care
  - Do they have transportation?
  - Rural areas of the state have fewer genetic services
- Reimbursement
  - If they lack reimbursement for basic health how will they find reimbursement for genetic services?
- Can't private pay
  - If they are having trouble meeting basic needs they will lack the ability to private pay for genetic services?
- Literacy issues
  - What is their literacy level? If they can't read they will probably need assistance in the informed consent process.
- Language barriers
  - If they do not speak English is there an interpreter available with knowledge in translating genetics terminology?
- Do they want to know about their risk to develop disease?

## Genetics and Genomics

- Genetics
  - Single gene disorders
    - Eg: Huntington's Disease, Familial Polyposis
- Genomics
  - Multiple genes and their interactions
    - Eg: Asthma, Type II Diabetes

## Why Talk About Genetics/Genomics?

- Human Genome project completed April 2003
- All diseases except some accidents have a genetic component
- We all have some inherited predisposition to disease

## Why Talk About Genetics/Genomics?

- Technological advances require health professionals to have at least a basic amount of genetics/genomics knowledge
  - Eg: ISONG Scope of Practice for Nurses
- Informed decision making as a healthcare consumer
  - Knowing what your family history means for you

## A few words about race, ethnicity & genetics

## Race, Ethnicity & Genes

- Genetically we are all about 99.9% the same
- The 0.1% difference is where the genetic component of the differences in health, and other human traits are.  
Race & Ethnicity in the Genome Era, Bonham, Amer. Psych., Jan 2009
- Race is an external, superficial label
  - Race is a factor in issues such as health disparities, but is not a concept that is substantiated by genetics
- Genes do not determine race
  - Eg: AI Tribe wanted develop "genetic test" to prove tribal membership...found there is no "NA gene"

## Race, Ethnicity & Genes

- To understand and prevent health disparities, we should seek more proximate causes, not depend on the muddy proxies of race and ethnicity

Francis S. Collins, M.D., Ph.D.  
National Leadership Summit to Eliminate  
Racial and Ethnic Disparities in Health  
January 10, 2006

## The Process

## Genetic Risk Assessment

- Risk assessment
  - Is the first step in a process
  - Takes into account known risk factors for a disease or condition
  - Helps determine general level of risk for a disease or condition
  - Identifies moderate and high-risk individuals that might benefit from genetic counseling

## Genetic Counseling

- Performed by healthcare professional with specific training in genetics
  - Genetic Counselor
  - Geneticist
  - MD
  - RN
- A nondirective educational process focusing on persons/families at risk for genetic conditions
- Focuses on medical and emotional aspects
- Results in identification of individuals who meet eligibility criteria for genetic testing
- Goal is informed consent not genetic testing

## Genetic Testing

### Genetic testing:

Analyzing an individual's genetic material to determine predisposition to a particular health condition or to confirm a diagnosis of genetic disease

<http://www.cdc.gov/genomics/gtesting/ACCE/FBR/CF/CFGlossary2.htm#Glossary>

## Genetic Testing

### Context:

- Predisposition genetic testing
  - Utilizing a molecular test to identify a change in a gene that may indicate a risk of developing a certain disease
- Genetic testing in treatment of disease
  - Utilizing a molecular test to identify how a person will respond to treatment or to determine recurrence of disease

## Informed Consent

- A process required prior to any genetic test
- Assists individual/family in understanding
  - The testing process
  - The possible test results & their meaning
  - The risks, benefits and ethical, legal and social implications of having the test
- Includes a signed consent outlining the above information
- Must be free from bias or coercion

## Possible Benefits of Genetic Testing

- Possible disease prevention for those at high risk
  - Lifestyle choices – diet, smoking, alcohol, environmental exposures
  - Medical choices – eg: removal of colon polyps or entire colon in hereditary CRC syndromes
- Early detection of disease for those at high risk
  - Use of alternative screening frequency/methods for women with BRCA mutations

## Possible Benefits of Genetic Testing

- To allow for decision making
  - Identifying a mutation within a family to allow others in family to undergo testing
  - Preimplantation genetic testing to avoid having a child with a known inherited mutation
  - Advanced decision making in life altering diseases such as Huntington's Disease

## Possible Benefits of Genetic Testing

- Targeted therapy
  - Cancer treatment based on each specific tumor and whether it will be responsive to a treatment

## Possible Risks Of Genetic Testing

- Guilt
  - Parents discover they passed a mutation on to children
  - Survivor guilt in persons who test negative for a families mutation
- Anxiety
  - Once test result is known it can't be "undone"

## Possible Risks Of Genetic Testing

- Genetic test available but no treatment
  - Huntington's Disease
- Negative result may provide false sense of security
  - Still at general population risk for diseases like cancer, heart disease, diabetes, etc.
- Possible Discrimination
  - Possible loss of insurance
  - Possible loss of employment

## Ethical, Legal & Social Implications of Genetic Testing

- Appropriate informed consent process
- Access to genetic information & confidentiality
- Intellectual property rights
  - Who “owns” genetic information
    - Companies sell a “gene” they have identified in a group of people. The company makes the profit, not the people who donated their DNA

## Ethical Issues

- For profit lab paid NA women \$100 to submit blood for genetic testing
- Purpose was to add NA data to their database
- Inadequate informed consent
- No ethical oversight to the project

## Targeted Treatment

- Iressa (gefitinib)
  - FDA approved for treatment of advanced non-small-cell lung cancer Expensive
  - Only 13.6% patients responded very well
  - Further research found tumors with a specific genetic mutation were susceptible to the drug

## Genetic Discrimination

“Unequal treatment of persons with either known genetic abnormalities or the inherited propensity for disease. Genetic discrimination may have a negative effect on employability, insurability, and other socioeconomic variables.”

Taber's Cyclopedic Medical Dictionary – 20<sup>th</sup> Edition, 2001

## Why Talk About Genetic Discrimination?

- Additional Barrier for those who are disadvantaged
  - Lack of funds to access genetic services
  - Added “risk” for those trying to get back into workforce/reinsured
  - Risk of social stigma

## Genetic Discrimination

- American's with Disabilities Act - 1990
  - provides protection from discrimination for "genetically disabled" people<sup>1</sup>
- Executive Order 13145 in 2000
  - prohibits genetic discrimination in the workplace for federal employees
- Genetic Information Nondiscrimination Act of 2007
  - First introduced in 2003

<sup>1</sup><http://www.genome.gov/12513983>

## Genetic Discrimination

- HIPAA Provides some protection but **does not**:
  - Prohibit the use of genetic information as a basis for charging a group more for health insurance.
  - Limit the collection of genetic information by insurers and prohibit insurers from requiring an individual to take a genetic test.
  - Limit the disclosure of genetic information by insurers.
  - Apply to individual health insurers except if covered by the portability provision.

## Solutions

## Initiatives to Eliminate Health Disparities

### Federal Initiatives:

- National Center on Minority Health and Health Disparities (NCMHD) - NIH
  - Healthy People 2010 – Goal #2 Eliminate Health Disparities - HHS, CDC
  - Health Disparities Collaboratives - HRSA
  - Others:
    - American Medical Association
    - American Public Health Association
    - Prevention Institute (California)
    - Healthy Americans.org
- And the list goes on... 1.4 million hits

## Technological Solutions

- Lower cost genetic tests
- Individualized health care
  - Increase knowledge about targeted therapies = each tumor treated based on specific molecular signature

## Health System Solutions

- Health professionals with increased genetics knowledge
- Cancer genetics moving toward primary care (NIH prediction)
- Better methods of risk assessment to identify individuals in need of genetic services

## Community Based Solutions

Community Based  
Participatory Research  
(CBPR)

“Research that is conducted as  
an equal partnership between  
traditionally trained "experts"  
and members of a  
community”

[Wikipedia.org](https://en.wikipedia.org/wiki/Community_based_participatory_research)