Genome v. Phenome

Environment-gene interactions and interpretation of personal genome analysis

Case 3: Gender, Race, and the Complexities of Science and Technology

Varki et al. (2008) question: why so little genetic difference between humans and non-human hominids, when they have so many phenotypic differences?

Some definitions

- Gene expression
- Disease risk
- Environment

Multiple analyses, multiple interpretations

Varki et al. reviewed them all…

- ## Phenomic analysis

  - Manifestation of gene-environment interactions

- ## Systems analysis

  - Differences in organ system and performance

- ## Molecular/genetic analysis

  - single nucleotide differences, gene deletions, repeat elements…

Copy Number Variants

QuickTime™ and a
decompressor
are needed to see this picture.

4

Varki et al., 2008

# Environmental impacts on disease risk

- Most personal disease risk is jointly determined by genes and complexities of the individual's environment.

- Gene-environment outcomes are evident only over decades.

- Late-life diseases and conditions are especially subject to interactions between the personal genome and personal sequence of social and physical environments.

Frank, 2005

Gene expression: modifying factors

Environment

Culture

Learning
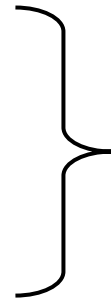
Social & physical behavior

Varki et al., 2008

Darwin, shmarwin

QuickTime™ and a
decompressor
are needed to see this picture.

- Baldwin: learned behavior becomes hard-wired
- Wallace: pre-conditional adaptation

Varki et al., 2008

Assessing disease risk and ancestry: one and the same?

Methods for calculating disease risk rely on comparison within "human populations."

Rose 2001, 2008

Promises…

Knowing your ancestry will empower you.

- Proving your legitimacy as a member of a respected group

- Increasing self-knowledge and therefore self-direction

- Providing a sense of belonging.

AfricanDNA.com

Promises…

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Do you belong to a "high-risk" group?

Promises.

- Information about your disease risk will empower you.
  - You can prevent or lessen disease impact.
  - You can protect your family -- or unborn children

deCODE.com

Promises.

- Information about your disease risk will empower you.
  - You can prevent or lessen disease impact.
  - You can protect your family -- or unborn children

Are you a "risky individual"?

Unanticipated results?

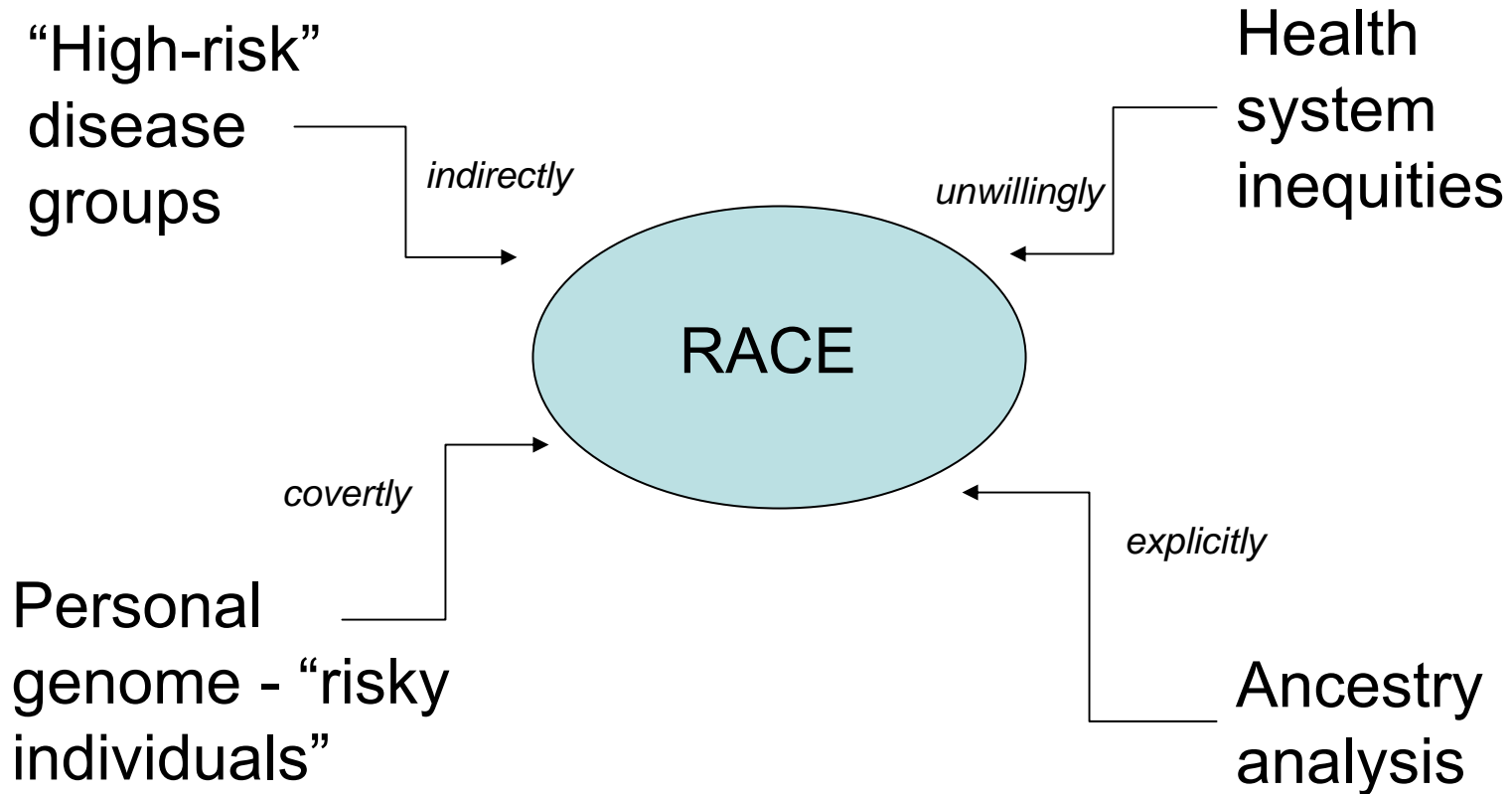High-risk groups, risky individuals both experience undesired outcomes:

- – Invasive monitoring and surveillance
- – Coercion
- – Comparison with an elusive "normal"

Eugenics

Rose 2001, 2008

All roads lead to issues of race

U.S. society links these issues…

"High-risk" disease groups

indirectly

Health system inequities

unwillingly

RACE

covertly

Personal genome - "risky individuals"

explicitly

Ancestry analysis

Uncertainties abound

- Which "population" is your genome compared with? What method was used?

- How will you interpret reports of risk -- or no risk?

- How will you account for environmental factors?

- What will you do with the information? What will your insurance company and your employer do with the information?

- Which risk group do you belong to?

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WGS.693 Gender, Race, and the Complexities of Science and Technology: A Problem-Based
Learning Experiment

Spring 2009