

# **Applying and Integrating Genomics into Public Health Practice: An Overview**

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# Outline

- Terms and Definitions
- Human Genome Project
- HapMap Project
  - What can we take from these projects to help us with our public health activities?
- Challenges
  - Examples from the field
- Northwest Center for Genomics and Public Health
  - Who we are, what we do and how we can help you
- Conclusions

# Terms and Definitions

- **Genome:**

- All the DNA in an organism, including it's genes
- 23 pairs of human chromosomes, including 1 pair of sex chromosomes (XX=female, XY=male)
- Chromosome 1 is the biggest, 22 is the smallest

- **Genomics:**

- Study of the functions and interactions of all the genes in the genome, including their interactions with environmental factors

- **Public Health Genomics:**

- Assess the impact of *genes and their interaction* with behavior, diet and the environment on the populations health

# Human Genome Project

- A 15 year international project, started in 1990 by the Dept. of Energy and the National Institutes of Health
- **Goals**
  - Determine the complete sequence of DNA subunits
    - create a reference sequence
  - Identify all genes in human DNA
    - Approximately 20-50,000 genes
    - Some estimates as high as 80-140,000 genes in the human
    - Address the Ethical, Legal, Social Implications (ELSI) of this information
- **Private sector companies were also sequencing the human genome**

# Whose genome are we looking at in the HGP?

- Subjects are anonymous
- DNA was obtained from “a large number of donors”
  - white cells from females
  - sperm from males
  - sequence does not represent any single individual
- In the private sector (Celera Genomics)
  - samples from anonymous donors of European, African, American (North, Central and South), and Asian ancestry were included.

# Completing the HGP

- Completed in 2003, 2 years ahead of schedule
  - Number of genes is still unknown, but *estimates* are much lower, perhaps as few as 20-25,000
  - Genes makeup approximately 2% of the genome
  - 99.9% of nucleotide sequences are the same for all people
  - The functions are known for half of the discovered genes
    - Humans share most of the same protein families with worms, flies and plants, but some protein families have expanded (immunity and development)
- Provides a “road map” of the human genome

# Haplotype Map of the Human Genome



## Goals:


- Define *patterns of common genetic variation* across the human genome
- Guide selection of SNPs (single nucleotide polymorphisms) efficiently to “tag” variants
- Public release of all data (assays, genotypes)

# The HapMap Populations

- Yoruba in Ibadan, Nigeria
  - 30 trios (90 individuals, 60 unrelated)
- Centre d'Etude du Polymorphisme Humain collection (CEPH) in Utah, USA
  - 30 trios (90 individuals, 60 unrelated)
- Han Chinese in Beijing, China
  - 45 unrelated individuals
- Japanese in Tokyo, Japan
  - 45 unrelated individuals

# Findings and Implications from the HapMap project

- PHASE II results released on web October 2005
  - >3,500,000 SNPs typed in all 270 HapMap samples
- Variation across groups
- Frequency of some polymorphisms differs between groups
- Implications for application and health disparities
  - Predictive value positive and negative can differ between groups if the prevalence of the polymorphism differs



# Integrating Genomics into Public Health Practice

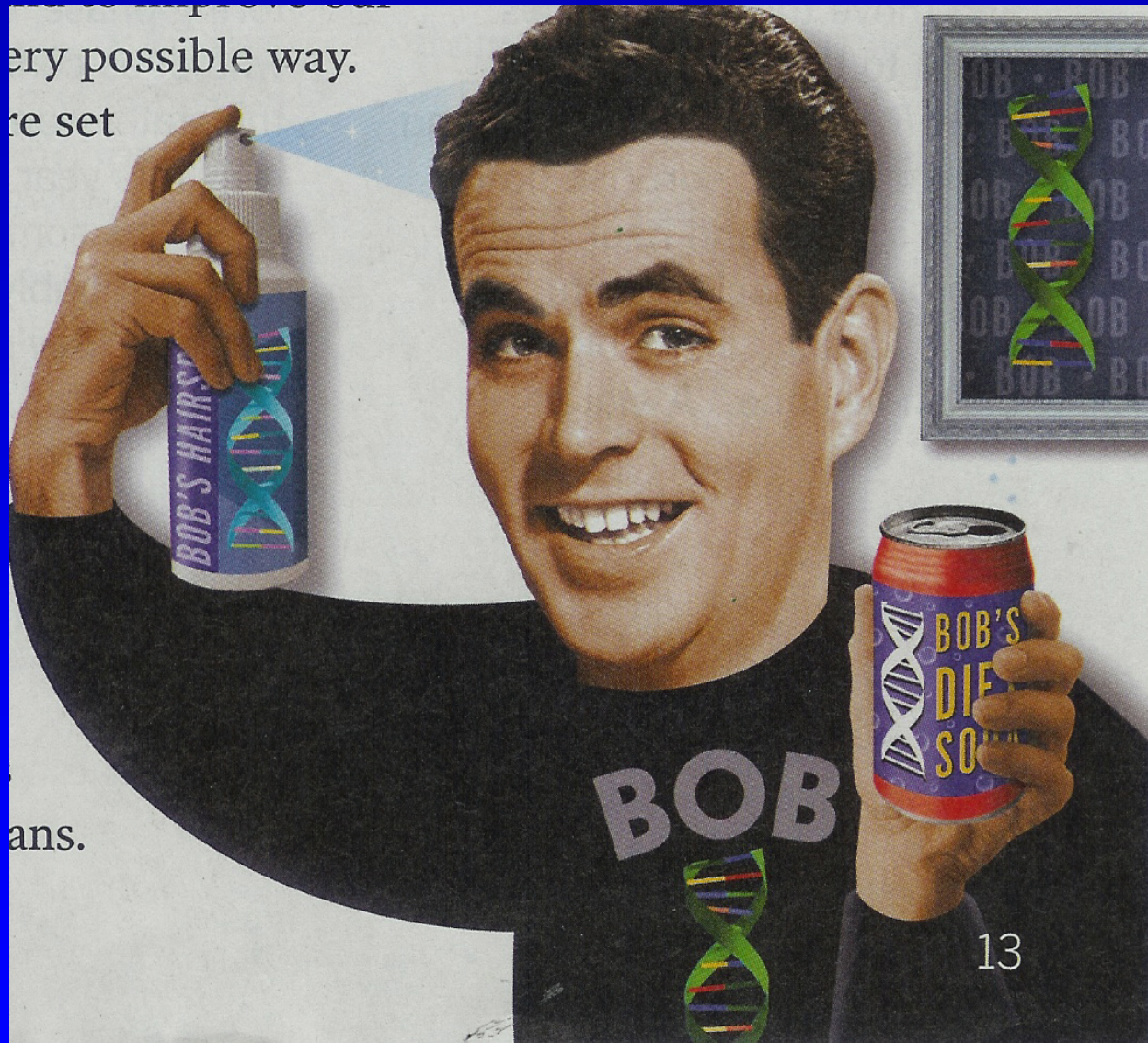
"Now this is not the end. It is not even the beginning of the end. But it is, perhaps, the end of the beginning."

Winston Churchill, 1942

# Integrating Genomics into Public Health Practice: Challenges

- The avalanche of genome data grows daily. The new challenge will be to use this vast reservoir of data to explore how DNA and proteins work with each other and the environment to create complex, dynamic living systems
  - Identify what is ready for prime time application and what isn't
  - Identify approaches to keep up to date
  - How to integrate new tools into existing programs
- Biologic explorations in this century will encompass studies in transcriptomics, proteomics, structural genomics, new experimental methodologies, and comparative genomics (functional genomics).
  - Improve health of (all) populations
  - Reduce health disparities
  - Ethical, legal and social implications (ELSI)

# Direct To Consumer Marketing



# Nutritional Genomics

The study of how different foods can interact with particular genes to increase the risk of diseases such as type 2 diabetes, obesity, heart disease and some cancers

**Goal:** Use of personalized diets to prevent or delay the onset of disease and optimize and maintain human health

<http://nutrigenomics.ucdavis.edu/pressarticles.htm>

# Weight Loss

***“DNA Diet Builds Customized Weight-Loss Plan”***

***“One-size-fits-all diets could be a thing of the past”***

The newest weight-loss plan is a customized diet based on your DNA. The DNA diet is a personalized meal plan that claims to be based on your unique genetic blueprint (NBC station KNSD in San Diego ).

Katzin claims that based on your DNA profile she can “determine whether someone should increase the amount of folic acid, B-6 or B-12, for example. So, we would choose foods that are rich in those supplements.”

... “interprets the data and makes a customized meal plan. Her suggestions range from “ taking more vitamins to eating more meat.”

# Sciona: Heart Health Panel


- “Analyzes thirteen of your genes that may play an important role in determining how your body manages overall heart health”
- “...assesses nine key diet and lifestyle action areas”

<http://sciona.com/products.html>

Gene Analyzed	Role of the Gene in Heart Health	Genetic Variation Screened For Variations Found in Your Gene	Percentage of Population with this Gene Variation*
MTHFR	Use of Folic Acid for DNA Synthesis or DNA Repair	C677T	28.7
		A1298C	30.0
MS_MTRR	Metabolism of Vitamin B12	A66G	47.3
MTR	Removal of Homocysteine	A2756G	17.4
CBS	Metabolism of Vitamin B6 and Removal of Homocysteine	C699T	28.0
MnSOD	Antioxidant Defense	C(-28)T	54.2
		T175C	
SOD3		C760G	
IL-6	Inflammatory Response	G(-174)C	36.3
TNF- $\alpha$		G(-308)A	16.5
APOC3	Triglyceride Metabolism	C3175G	12.6
CETP	Cholesterol	G279A	37.0
LPL	Metabolism	C1595G	9.9
eNOS	Blood Flow	G894T	35.6
ACE		DEL	61.0

\*The population frequencies given are normalized for the U.S. population data from the U.S. 2002 Census Report. Population frequencies can vary for different ethnic groups, so for more detailed information, please turn to the Population Frequency Data Table in the Reference Section of your report.

# In Store Sales




Sciona™ Optimal health through genetics™

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## Retail Partners

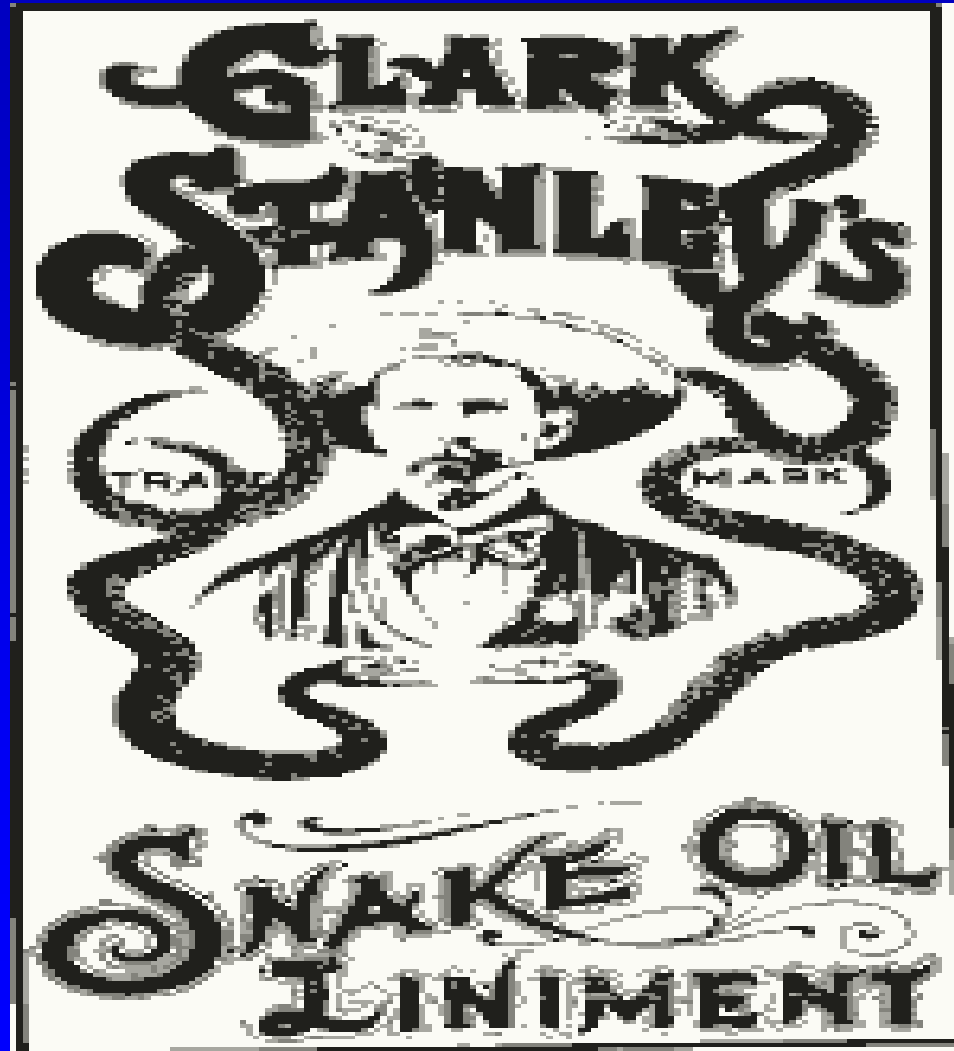
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<a href="http://www.hy-vee.com">www.hy-vee.com</a>		
<a href="http://www.ukrops.com">www.ukrops.com</a>		

## New DNA kits to help you manage your health?

A \$99 test, available in some supermarkets, may help consumers manage risks associated with heart health, bone health, insulin resistance and more

- **Lund Foods CEO:** “...plan is to create a link between the evaluations performed by Sciona and his stores’ food experts, which have long provided consumers with diet and nutritional advice and information.”
- **Today Food Editor:** “The idea, which is a good one, is to help shoppers understand what they can do in their daily food choices to either maintain their good health or help correct certain genetic defects that the test may have identified.”

# Genetic Profiteering



# GAO Report: July 27, 2006

- 10 month investigation of internet based companies selling nutrigenetic test kits
  - Genelex, Seattle, WA
  - Sciona, Boulder CO
  - Suracell, Montclair, N.J.
  - Market America, Greensboro, N.C.
- Test kits range in price from \$100-\$400
- DNA from 2 subjects were collected
  - 48 yr male, 9 month old female
  - Created 14 “fictitious consumers”
- Lifestyle and health information were created and submitted with DNA to 4 companies

# GAO Report: July 27, 2006

- All 4 companies mislead consumers
  - Health related predictions that are medically unproven
  - Claims about disease risk that may be interpreted as diagnoses
  - Below average risk
- Results are not based on unique genetic profile
- Expensive supplements were promoted by 2 companies
  - \$1,200 year for a personalized customized nutritional formula  
*“for those areas of your body that need special support”*
  - \$1,880 for personalized supplements to *“compensate for genetic deficiencies and repair DNA”*
- Lack of QC in laboratory performing analyses

# Northwest Center for Genomics and Public Health

**Mission:** To integrate advances in genomics into public health practice

- Contribute to the knowledge base on genomics and public health, focusing on chronic diseases with modifiable environmental risk factors
- Provide technical assistance to local, state, & regional public health organizations
- Serve as a reliable and credible source of information
- Serve as a regional resource for public health
- Develop and provide training for the public health work force

# Major Areas of Emphasis

- Family history as a public health tool
- Translation and practical application
- EGAPP project
- Education
- Technical Assistance
  - Disease specific emphases: Diabetes, Obesity, CVD, Asthma, Colon cancer
  - Talks
  - Assistance in developing survey questions
  - Provide support for data analyses
  - Website: <http://depts.washington.edu/cgph>

# Rationale for using Family History as a Public Health Tool

1. Screening for single major gene(s) is unlikely
2. Reflects unique *Genomic* information
  - genomic, environmental, and interactions
3. Effective interventions for high-risk individuals
4. Identify individuals for targeted intervention
5. Family-Centered approaches

# Conclusions: Integrating genomics into public health practice

- Fast moving field – keep up to date
  - National Office of Public Health Genomics: <http://www.cdc.gov/genomics/default.htm>
  - State Depts of Health: MI, MN, ORE, Utah
  - Centers for Genomics and Public Health
    - Ann Arbor, Michigan
    - Seattle, Washington
- Need for partnerships
  - Academic Centers with State and Local Public Health organizations
- Networking with others is important
  - Don't reinvent the wheel



# Northwest Center for Genomics and Public Health

<http://depts.washington.edu/cgph>

Funded by the National Office of Public Health Genomics  
Centers for Disease Control and Prevention