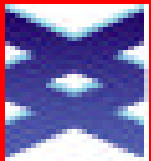


THE HUMAN GENOME PROJECT: IMPLICATIONS FOR HEALTH CARE, RESEARCH AND SOCIETY

Washington, DC
March 25, 2002



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What We Will Consider

- “Traditional genetics”
- The Human Genome Project
- The “new genetics” - genomics
- Genomic health care
- Genomics and society
- Genomics and research



“Traditional Genetics”

- Involves conditions wholly caused by:
 - An extra or missing complete chromosome or part of a chromosome
 - e.g., Down syndrome
 - A mutation in a single gene
 - e.g., cystic fibrosis, fragile-x syndrome, sickle cell disease



“Traditional Genetics”

- These conditions
 - Are of great importance to individuals and families with them
 - But, even when added together, are relatively rare
 - Most people not directly affected
 - Thus, genetics played small role in health care (and in society)



≥ 9 of the 10 Leading Causes of Mortality Have Genetic Components

- 1. Heart disease (29.5% of deaths in '00)
- 2. Cancer (22.9%)
- 3. Cerebrovascular diseases (6.9%)
- 4. Chronic lower respiratory dis. (5.1%)
- ? • 5. Injury (3.9%)
- 6. Diabetes (2.9%)
- 7. Pneumonia/Influenza (2.8%)
- 8. Alzheimer disease (2.0%)
- 9. Kidney disease (1.6%)
- 10. Septicemia (1.3%)



Genomic Medicine

- Will come largely from knowledge emanating from the Human Genome Project



The Human Genome Project

- An international government project that is ahead of schedule!
- And under budget!!
- And from its start has earmarked funds for consideration of its ethical, legal, and social implications (ELSI) - the largest funding ever devoted to bioethics!!!



The Human Genome Project

- Human genome consists of three billion base pairs – Adenine, Cytosine, Guanine, Thymine
- Printing out the A,C,G,T would fill over 150,000 telephone book pages
- Disease is often caused by a single variation in the three billion bases - one letter in 150,000 pages different

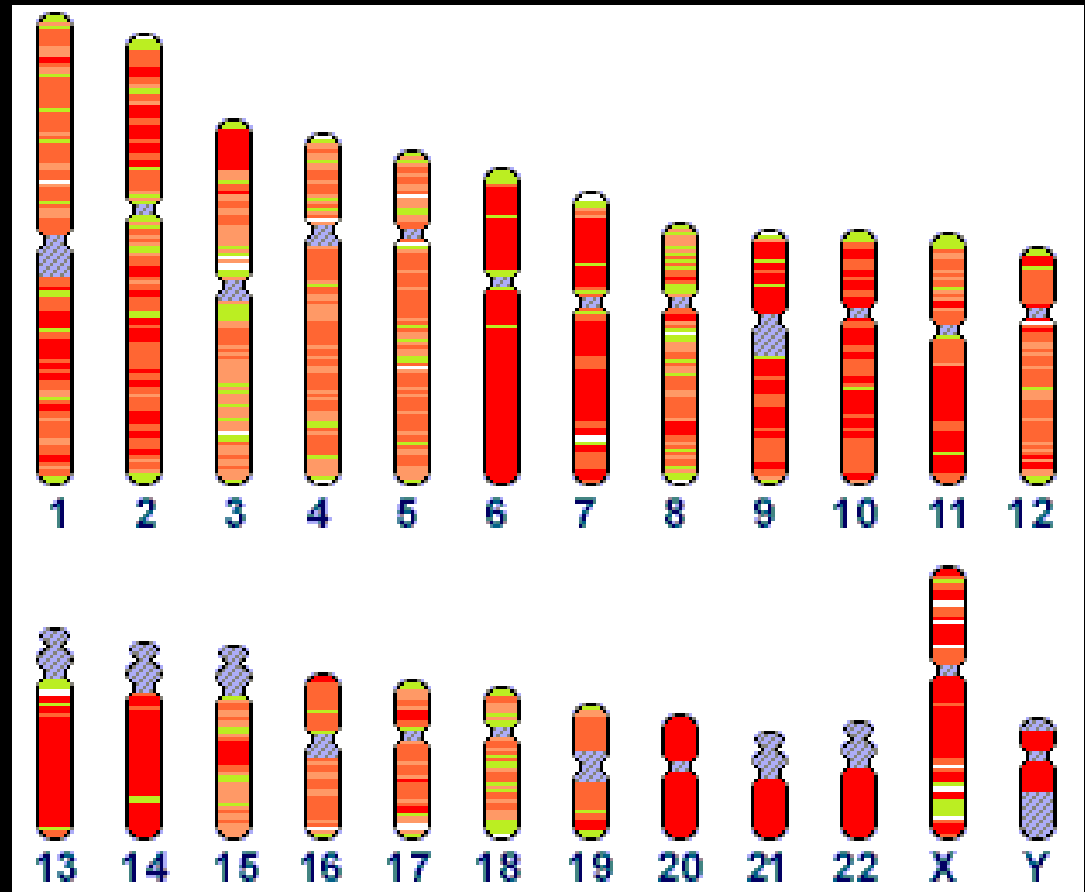


Human Genome Project - Sequence Progress (12/31/01)

Draft =
34.8%

Finished =
63.0%

Total =
97.8%



■ > 1000 kb ■ 250 - 1000 kb ■ < 250 kb
■ draft sequence ■ heterochromatin

Genomic Medicine

- About conditions partly:
 - Caused by mutation(s) in gene(s)
 - e.g., breast cancer, colon cancer, atherosclerosis, inflammatory bowel disease, diabetes, Alzheimer disease, mood disorders, many others
 - *Prevented* by mutation(s) in gene(s)
 - e.g., HIV (CCR5), ?atherosclerosis, ?cancers, ?diabetes, many others



Genomic Medicine

- These conditions
 - Are also of great importance to individuals and families with them
 - But are quite common
 - Directly affect virtually everyone
 - Will make genetics play large role in health care and in society



Genomic Medicine

- These conditions are common enough that:
 - Genetics care will be supplied with occasional involvement of medical geneticists and genetic counselors, but primarily by primary care providers and other non-genetics specialists



Genomic Medicine

- Will change health care by...
 - Creating a fundamental understanding of the biology of many diseases, even many “non-genetic” ones
 - Leading to defining disorders by mechanism of causation, rather than by symptoms



Genomic Medicine

- Will change health care by...
 - providing knowledge of individual genetic predispositions
 - creating pharmacogenomics



Genomic Medicine

- Knowledge of individual genetic predispositions will allow:
 - Individualized screening
 - Individualized behavior changes
 - Presymptomatic medical therapies, e.g., antihypertensive agents before hypertension develops, anti-colon cancer agents before cancer occurs



Genomic Medicine

- Pharmacogenomics will allow:
 - new approaches to drug design
 - individualized medication use based on genetically determined variation in effects and side effects
 - new medications for specific genotypic disease subtypes



Genomic Medicine

- Will change health care by...
 - providing better understanding of non-genetic (environmental) factors in health and disease
 - allowing genetic engineering
 - emphasizing health maintenance rather than disease treatment



The “New Genetics”

- Will include knowledge about traits that most of us see as human characteristics, rather than as “diseases”
 - e.g., height, intelligence, alcoholism, violence, etc.



The “New Genetics”

- Will also change our lives...
 - Knowing our own (and others’) disease predispositions
 - Knowing our (and others’) “characteristics” predispositions
 - Showing that we are all mutants



The “New Genetics”

- May also change society...
 - Genetic stratification, e.g., in employment or marriage
 - Genetic engineering against (and for) diseases and characteristics
 - Cloning
 - Increased opportunity for “private eugenics”



The “New Genetics”

- Raises new concerns, such as:
 - Discrimination against individuals
 - Discrimination against groups
 - Nature over nurture? (genetic determinism)
 - Genes run in families
 - Confidentiality/privacy



The “New Genetics”

- And more concerns, such as:
 - Fairness in access
 - Right not to know and not to act
 - What is the appropriate informed consent process for genetic testing
 - Patenting and licensing



Areas of Genomics Related Research

- Model organisms
- Annotation of the genome
- Proteomics
- Biological pathways, networks, cellular mechanisms, etc.
- New technologies
- Pathophysiology of disease



Areas of Genomics Related Research

- Gene-gene-environment interactions
- Pharmaceutical design and use
- Health outcomes
- Health care delivery
- Ethical, legal and social issues
- Etc., etc.

