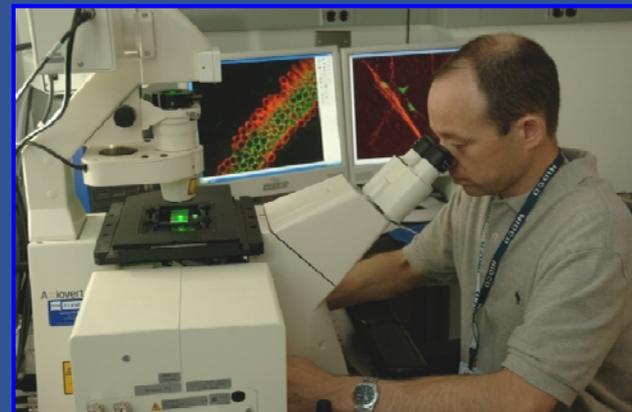


NIH: Steward of Medical and Behavioral Research for the Nation



“Science in pursuit of **fundamental knowledge** about the nature and behavior of living systems ... and the **application of that knowledge** to extend healthy life and reduce the burdens of illness and disability.”





NIH: Support for Bioethics

- Institutes and Centers: ~\$50M/year
 - NHGRI's Ethical, Legal, and Social Implications Research Program: >\$18M/year
- American Recovery and Reinvestment Act funds: \$17.8M (FY 2009 and FY 2010)
 - Supporting 21 grants
 - Select topics: ethical issues posed by emerging technologies; health disparities; research involving data/specimens
- Office of the Director: \$5M (beginning in FY 2010) “to support high priority bioethics research and training projects in coordination with the ICs”

The NIH Bioethics Task Force: Developing a Strategic Plan for Bioethics Research and Training

- Affirms importance of bioethics
 - In part because new scientific and technological challenges – **including genomics and neuroimaging** – pose novel bioethics and policy challenges
- Establishes near-term priorities, including:
 - Encouraging IC support for new mission-related bioethics initiatives
 - Enhancing training initiatives
- Articulates long-term goal: integrate bioethics into full spectrum of biomedical research, starting by:
 - Taking stock of past NIH investments
 - Identifying ethical issues that need to be addressed to advance scientific portfolios and priorities
 - Seeking public input in diverse forums

15 February 2011

na

nature

THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE

THE FUTURE IS BRIGHT

Reflections on the first ten
years of the human genomics age



Nuclear fission
Five-dimensional
energy landscapes

Seafloor spreading
The view from under
the Arctic icepack

Career prospects
Sequence creates new
opportunities

naturejobs
genomics special

GENOMICS

**THE END OF
THE BEGINNING**
*Eric Lander on the impact of
the human genome sequence*
PAGE 187

METHODS

**MORE BASES
PER DOLLAR**
*Elaine Mardis on the march
of sequencing technology*
PAGE 198

HEALTH

**FROM LAB
TO CLINIC**
*A road map to
genomic medicine*
PAGE 204

NATURE.COM/NATURE
10 February 2011 \$10



16 February 2011

nature

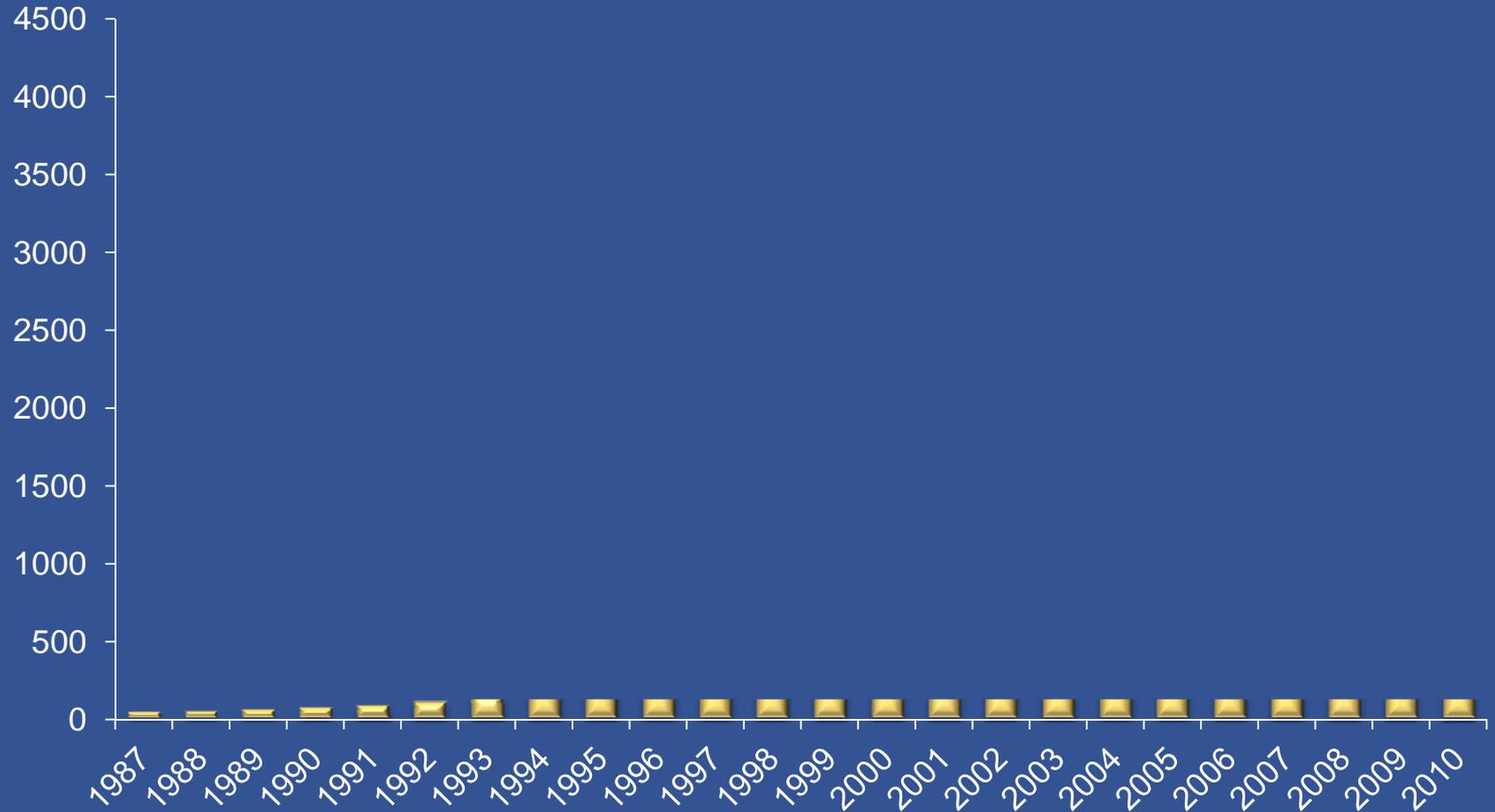
Vol 291 No 5507
Pages 1145-1434 \$9



ADVANCEMENT OF SCIENCE



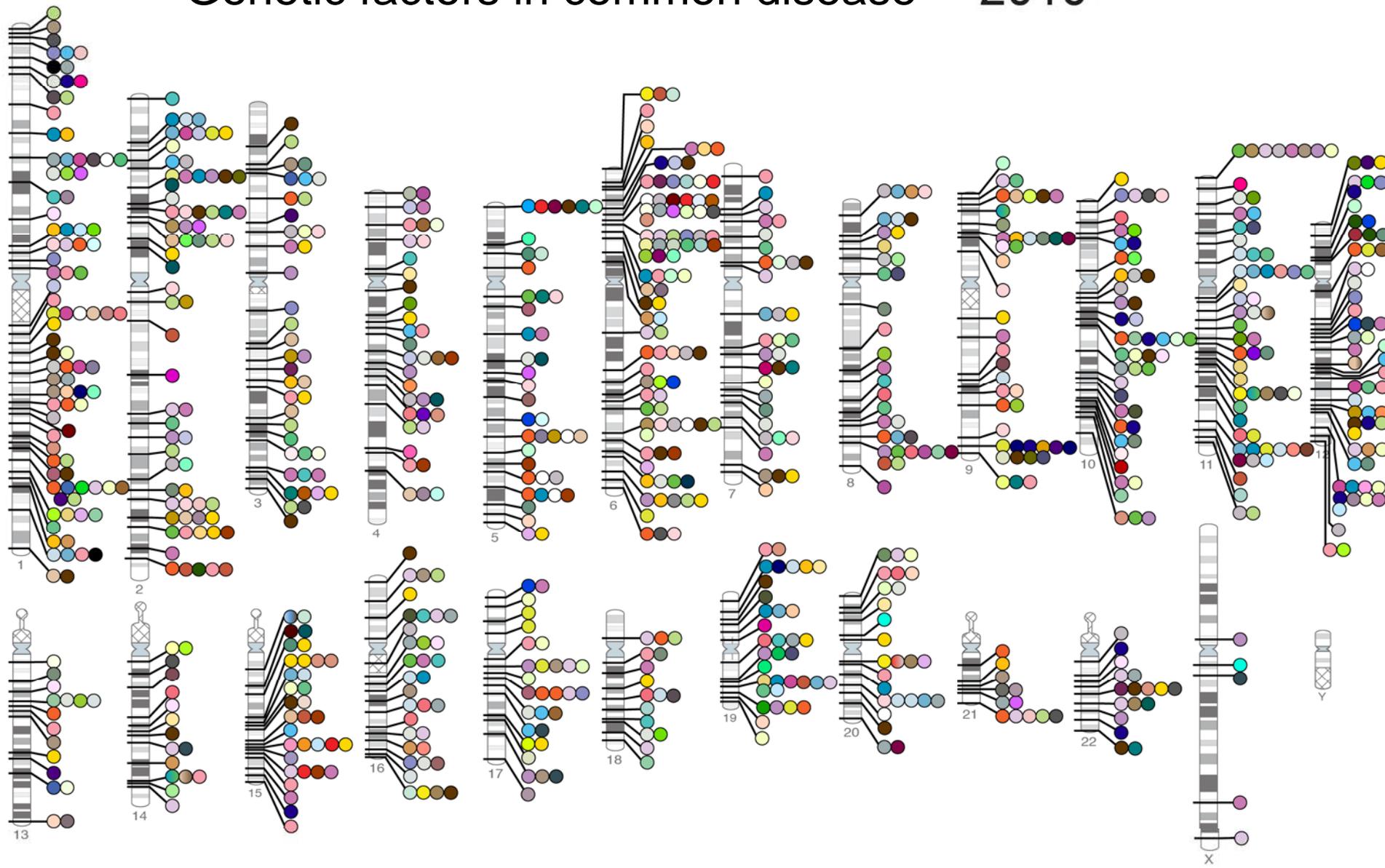
Disorders with Known Molecular Basis



Source: Online *Mendelian Inheritance in Man*, Morbid Anatomy of the Human Genome

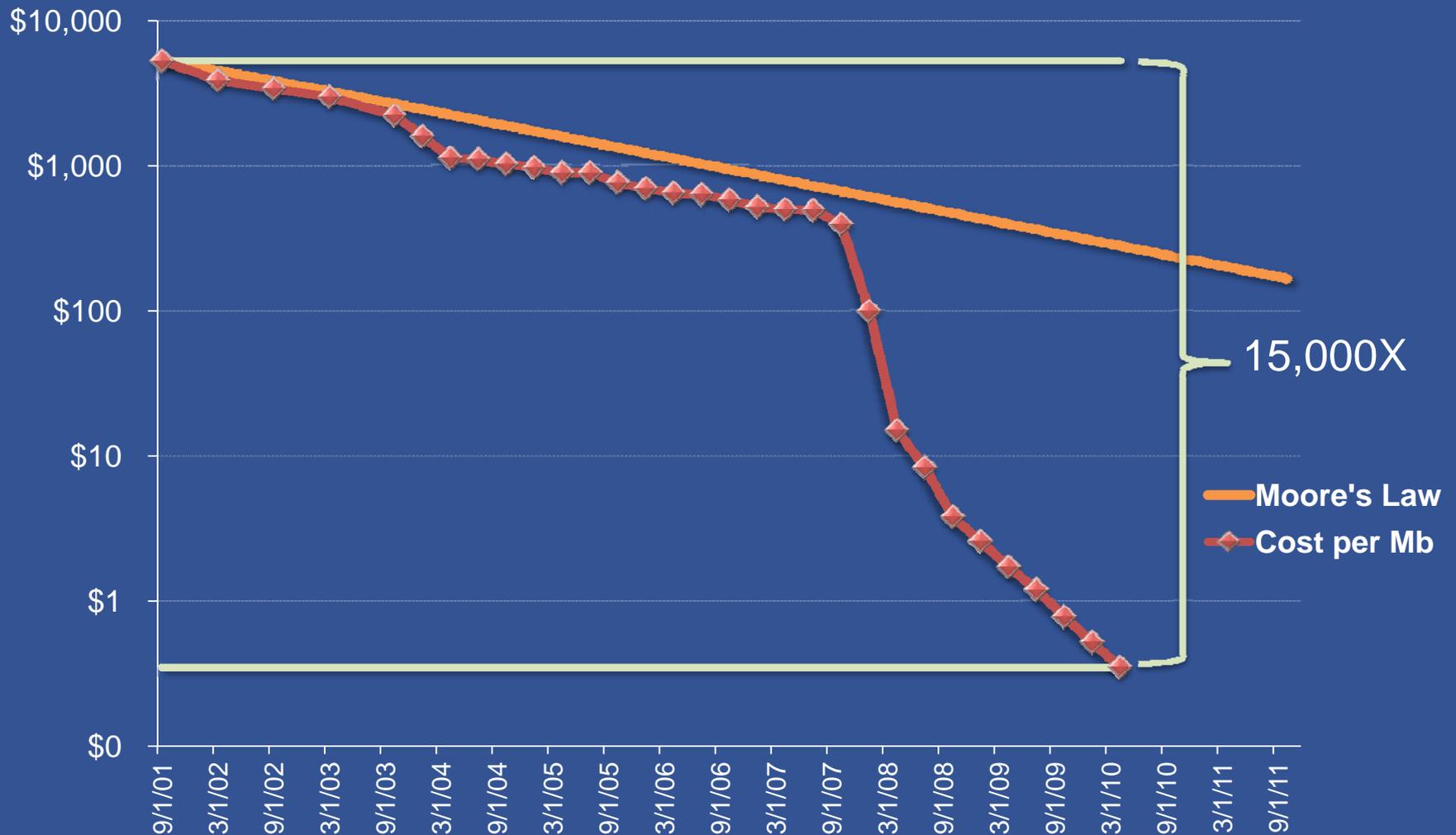
Genetic factors in common disease

2010



Sequencing Costs Decrease Rapidly ...

Cost per Megabase of DNA Sequence



Clinical Applications of Genomic Analysis: Diagnosis and Treatment

- Patient: 6-year-old Nic
 - Severe inflammatory bowel disease from just before 2nd birthday
 - 100+ surgeries – little solid food – **no diagnosis**
- Whole exome sequencing
 - Found mutation in *XIAP* gene
 - Gene previously linked to blood disorder; curable by bone marrow transplantation
- Diagnosis allows treatment
 - July 2010: Nic receives stem cell transplant from healthy donor
 - Today: doing well; recovery continues



Credit: Gary Porter, Milwaukee Journal Sentinel



Charting a course for genomic medicine from base pairs to bedside

Eric D. Green¹, Mark S. Guyer¹ & National Human Genome Research Institute*

Francis S. Collins, Eric D. Green, Alan E. Guttmacher and Mark S. Guyer on behalf of the US National Human Genome Research Institute

Psychosocial and ethical issues in genomics research

Psychosocial and ethical issues in genomic medicine

Legal and public policy issues

Broader societal issues

BOX 5

Genomics and society



Effectively examining the societal implications of genomic advances requires collaborations involving individuals with expertise in genomics and clinical medicine and experts in bioethics, psychology, sociology, anthropology, history, philosophy, law, economics, health services research and related disciplines.

Psychosocial and ethical issues in genomics research. These include ensuring appropriate protection of human research participants and addressing the perceptions of risks and benefits of participating in genomic studies; expanding the diversity of research cohorts; incorporating biological ancestry markers and self-identified race and ethnicity as variables in genomic studies; accomplishing effective community engagement; and including vulnerable populations (for example, children and the disabled) and deceased individuals in genomics research.

Psychosocial and ethical issues in genomic medicine. These include communicating with patients about the uncertainty and evolving nature of predictions based on genomic information; interpreting information from direct-to-consumer genetic tests; ensuring fair access to genomic medicine; assessing the effectiveness of genomically informed diagnostics and therapeutics; using genomic information to improve behaviour change interventions; addressing issues associated with pre-implantation, prenatal and postnatal genetic diagnoses; and determining how constructs of race and ethnicity relate to the biology of disease and the potential to advance genomic medicine.

Legal and public policy issues. These include intellectual property in genomics; insurance reimbursement for genomic services; regulation of genetic testing; regulatory and non-regulatory approaches for dealing with direct-to-consumer genetic testing; the regulation of pharmacogenomics and genomics-based therapeutics; protection against genetic discrimination and stigmatization; and uses of genomics in non-medical settings.

Broader societal issues. These include the implications of increasing genomic knowledge for conceptualizing health and disease; for understanding identity at the individual and group levels, including race and ethnicity; for gaining insights about human origins; and for considering genetic determinism, free will and individual responsibility.

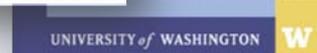
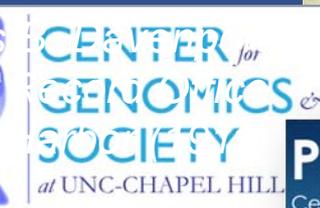
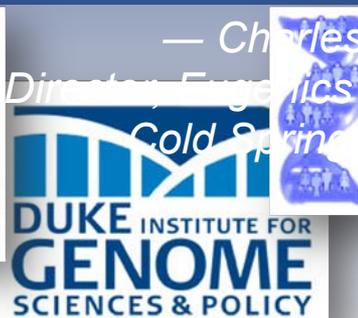
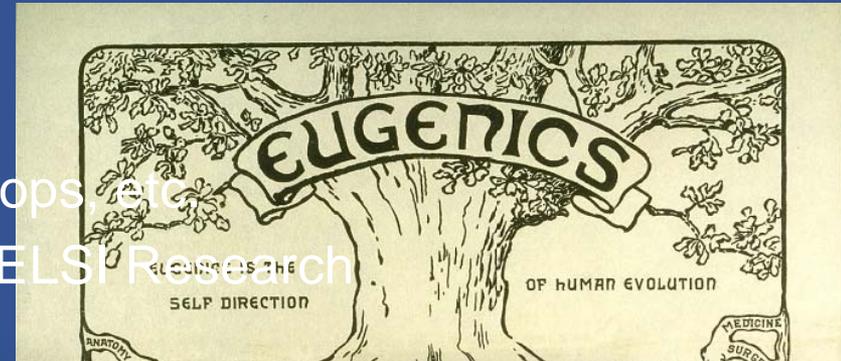
ELSI: Response to the Past; Vision for the Future

Ethical, Legal, and Social Implications Research Program

- Historical concern: eugenics
- ELSI established in 1990 as an integral part of the Human Genome Project
 - 3–5% of HGP annual budget
 - Remains active today

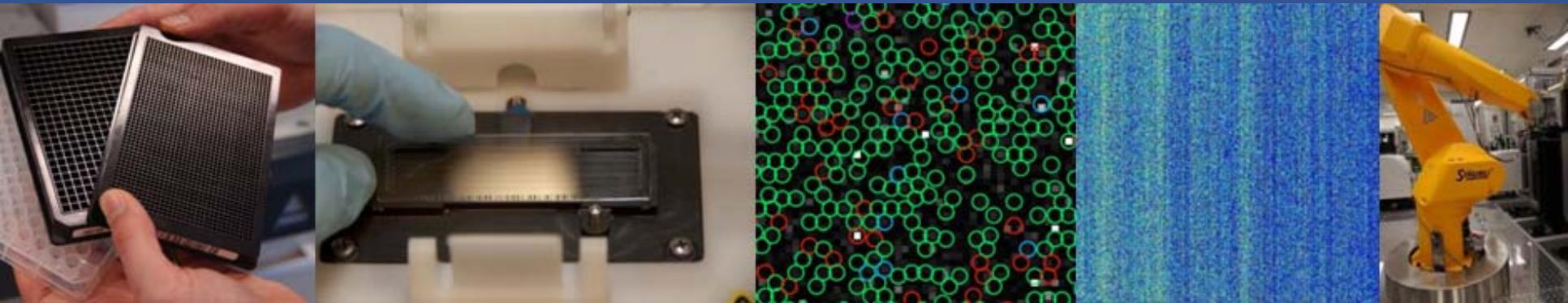
- ELSI at NHGRI

Eugenics is “the science of the improvement of the human race by better breeding”



Some Ethical Issues in High Throughput Technologies

- Genetic discrimination



Genetic Information Nondiscrimination Act (GINA) Becomes Law



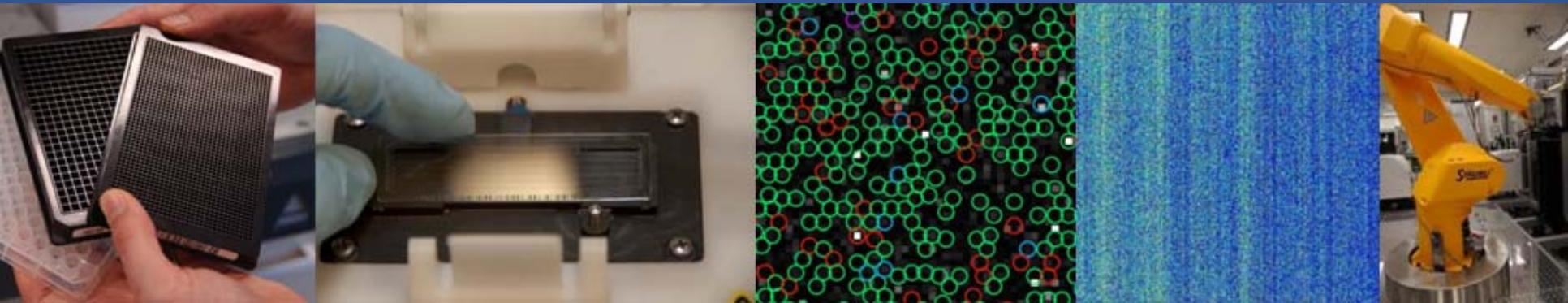
May 21, 2008

Unresolved potential risks of genetic discrimination

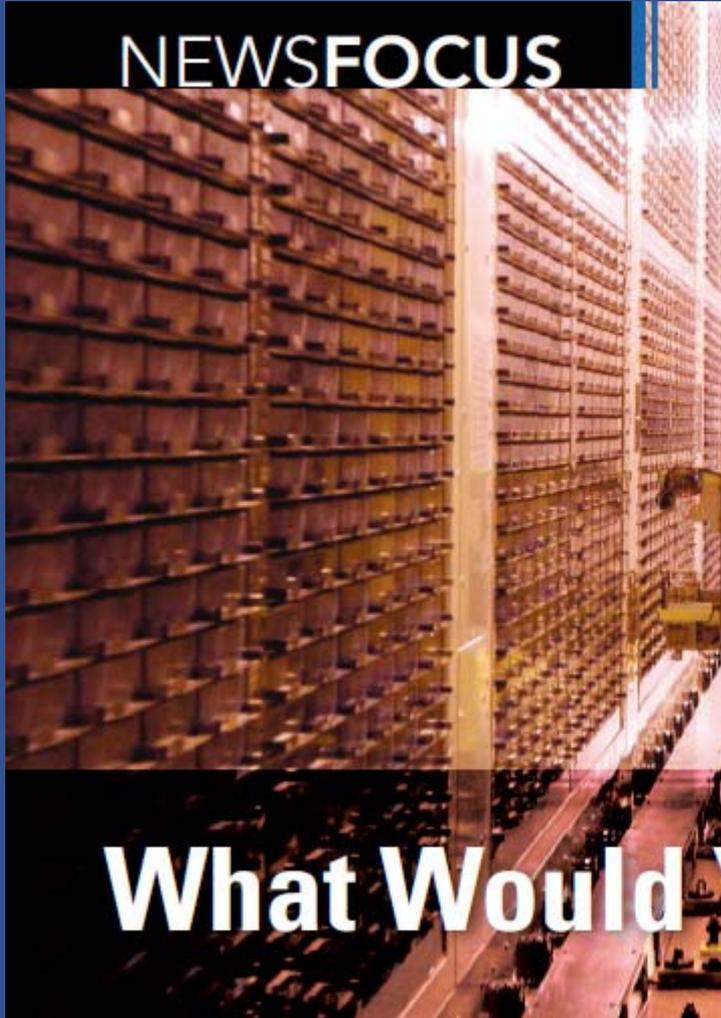
- Life insurance
- Long term care insurance
- Disability insurance
- Educational opportunities
- Military service
- Court decisions about child custody

Some Ethical Issues in High Throughput Technologies

- Genetic discrimination
- Incidental findings and return of research results



NEWSFOCUS

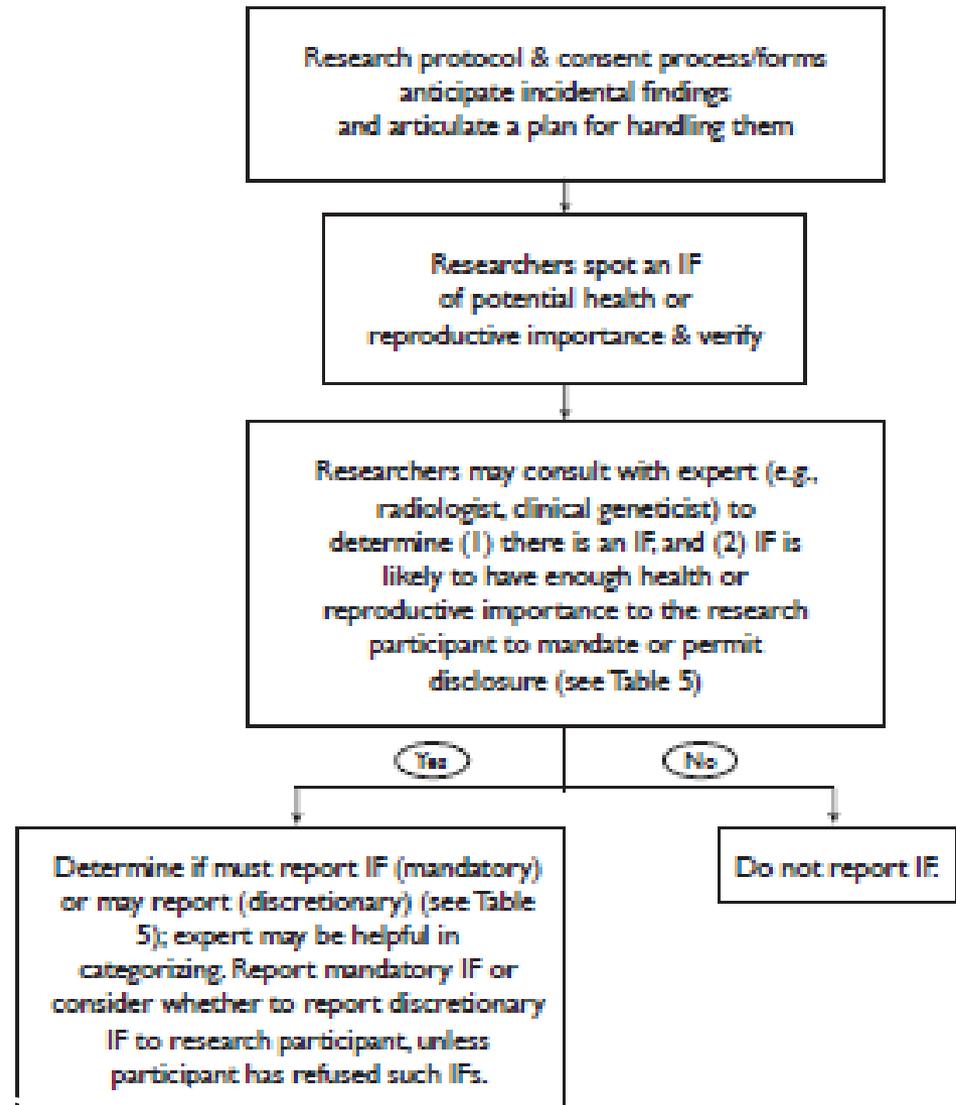


What Would

Science, 11 February 2008

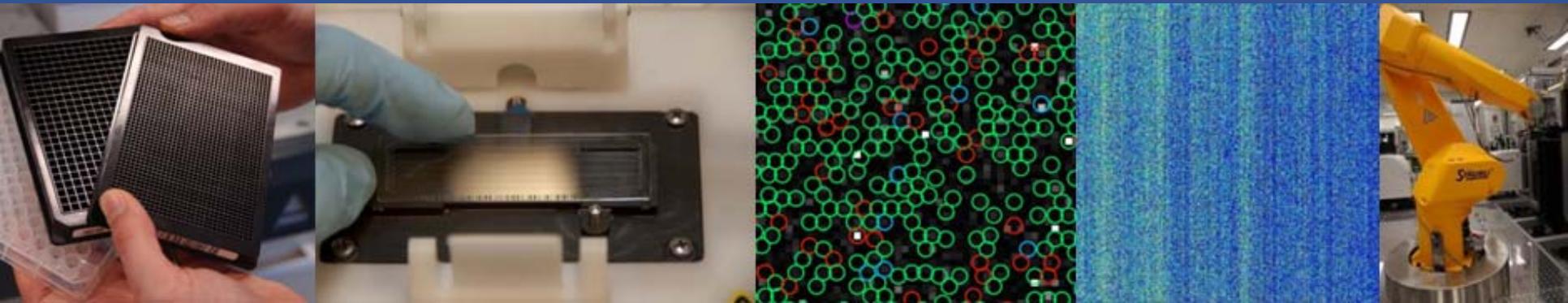
Table 4

Recommended Pathway for Handling IFs in Research



Some Ethical Issues in High Throughput Technologies

- Genetic discrimination
- Incidental findings and return of research results
- Forensic applications of DNA analysis



DNA and Forensics/Courts

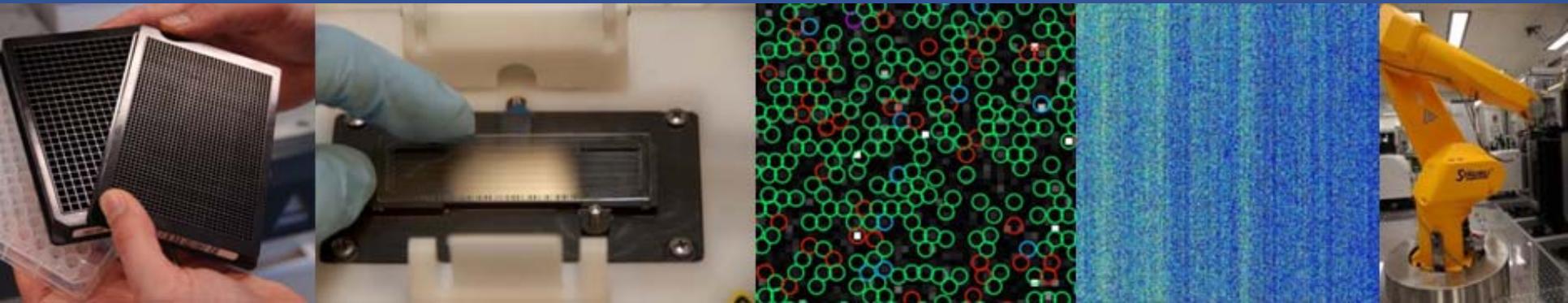
- Surreptitious collection
- Tracking through relatives
- Predicting age
- Predicting ancestry
- Predicting physical appearance
- Predicting recent travel (microbiome)
- Assessing presence of disease
- As a defense in a criminal case?



Science, 18 February 2011

Some Ethical Issues in High Throughput Technologies

- Genetic discrimination
- Incidental findings and return of research results
- Forensic applications of DNA analysis
- Neuroimaging

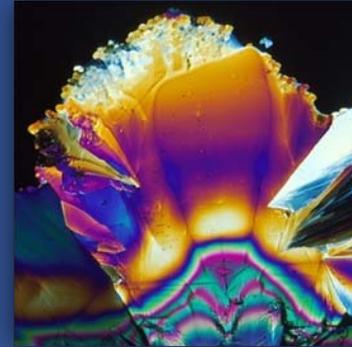


Neuroimaging – some ethical issues

- Incidental findings
- Lie detection
- Personality prediction/profiling
- Neuromarketing
- Exploration of spirituality
- Limited consciousness states
- Prediction of future disease risk

“We must not allow our technology to exceed our humanity.”

~ Albert Schweitzer



NIH *Turning discovery into health*

