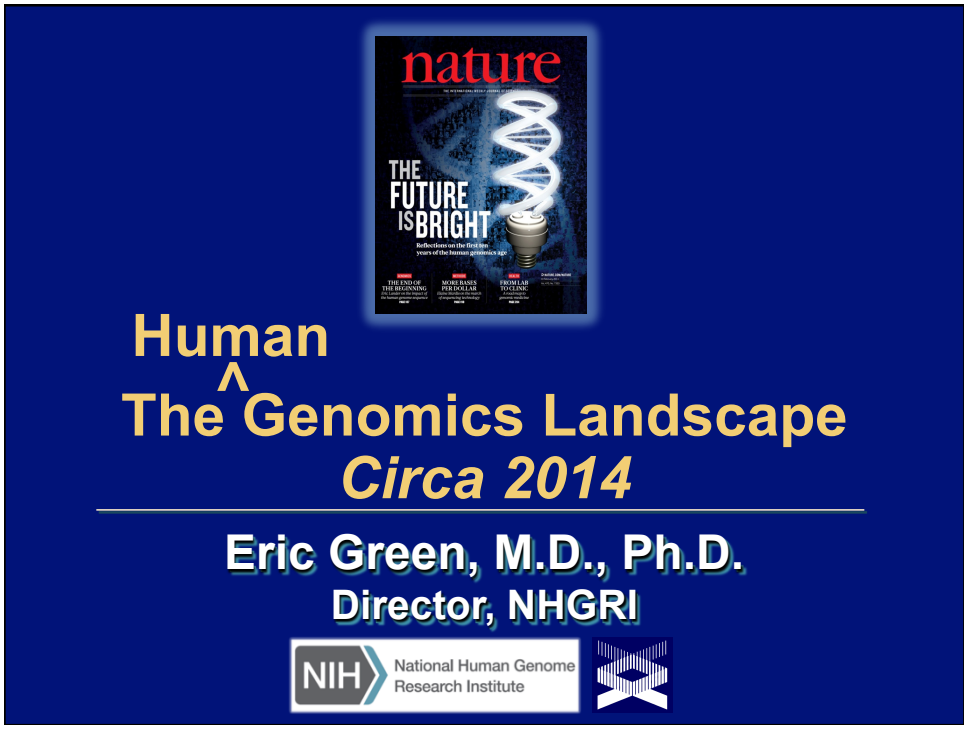
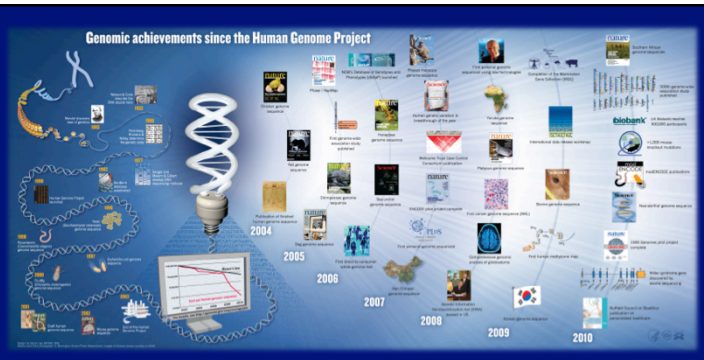

nature
THE FUTURE IS BRIGHT
Reflections on the first ten years of the human genome era
Human
The Genomics Landscape
Circa 2014
Eric Green, M.D., Ph.D.
Director, NHGRI
NIH
National Human Genome Research Institute


JOHNS HOPKINS
MEDICINE
CONTINUING MEDICAL EDUCATION
Current Topics in Genome Analysis 2014
Eric Green
No Relevant Financial Relationships with Commercial Interests
NATIONAL HUMAN GENOME RESEARCH INSTITUTE
Division of Intramural Research



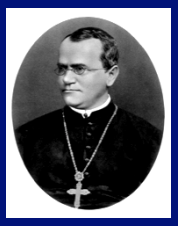
I. Historical Context for Genomics

II. Major Achievements since the Human Genome Project

III. The Human Genomics Landscape: 2014 and Beyond

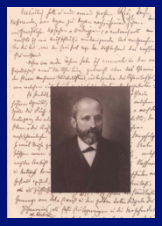
>> Goal: Place Future Speakers into a Broader Context <<

Foundational Milestones in Genetics & Genomics



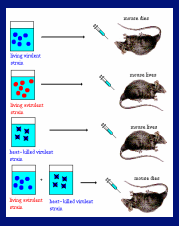
Mendel

1865



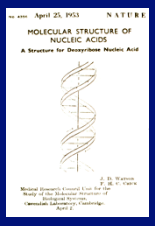
Miescher

1871



Avery

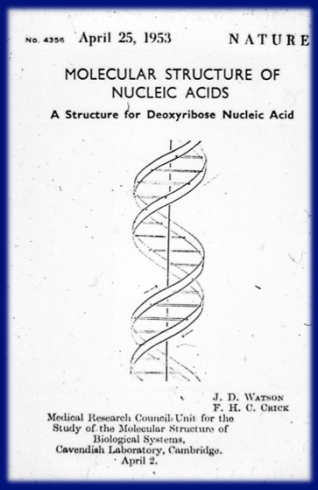
1944



Watson & Crick

1953

April, 1953



No. 4356 April 25, 1953 NATURE

MOLECULAR STRUCTURE OF NUCLEIC ACIDS

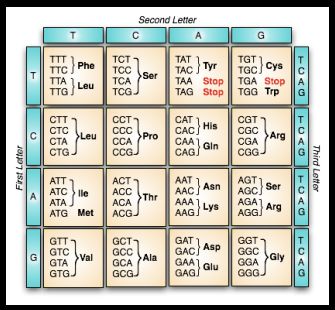
A Structure for Deoxyribose Nucleic Acid

J. D. Watson
F. H. C. Crick

Medical Research Council Unit for the Study of the Molecular Structure of Biological Systems, Cavendish Laboratory, Cambridge. April 2.

Discovery of Double-Helical Structure of DNA

1960's

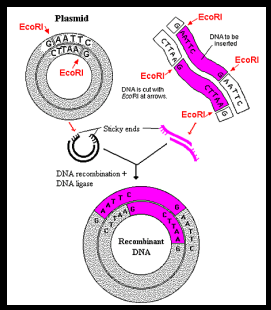


Second Letter

First Letter	T	C	A	G	Third Letter
T	TTT Phe, TTC Phe, TTA Leu, TTG Leu	TCT Ser, TCC Ser, TCA Ser, TCG Ser	TAT Tyr, TAC Tyr, TAA Stop, TAG Stop	TGT Cys, TGC Cys, TGA Stop, TGG Trp	T C A G
C	CTT Leu, CTC Leu, CTA Leu, CTG Leu	CCT Pro, CCC Pro, CCA Pro, CCG Pro	CAT His, CAC His, CAA Gln, CAG Gln	CGT Arg, CGC Arg, CGA Arg, CGG Arg	T C A G
A	ATT Ile, ATC Ile, ATA Ile, ATG Met	ACT Thr, ACC Thr, ACA Thr, ACG Thr	AAT Asn, AAC Asn, AAA Lys, AAG Lys	AGT Ser, AGC Ser, AGA Arg, AGG Arg	T C A G
G	GTT Val, GTC Val, GTA Val, GTG Val	GCT Ala, GCC Ala, GCA Ala, GCG Ala	GAT Asp, GAC Asp, GAA Glu, GAG Glu	GGT Gly, GGC Gly, GGA Gly, GGG Gly	T C A G

The Genetic Code

1980's



DNA Cloning

The Origin of "Genomics": 1987

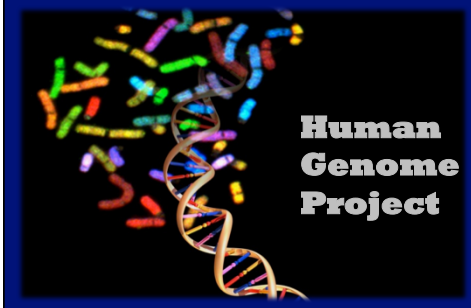
EDITORIAL

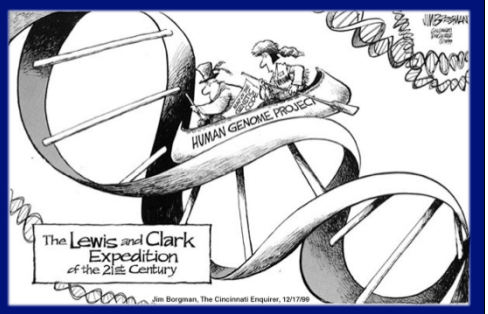
A New Discipline, A New Name, A New Journal

***Genomics* (1987)**

For the newly developing discipline of [genome] mapping/sequencing (including the analysis of the information), we have adopted the term GENOMICS… The new discipline is born from a marriage of molecular and cell biology with classical genetics and is fostered by computational science.

October, 1990





Human Genome Project Begins

April, 2003

Human Genome Project Ends

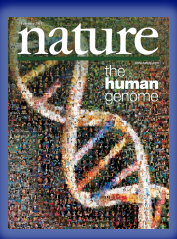
Myriad Applications of Genomics

Health, Disease, & Medicine

Genomic Medicine

An emerging medical discipline that involves using genomic information about an individual as part of their clinical care (e.g., for diagnostic or therapeutic decision-making) and the other implications of that clinical use

The Path to Genomic Medicine



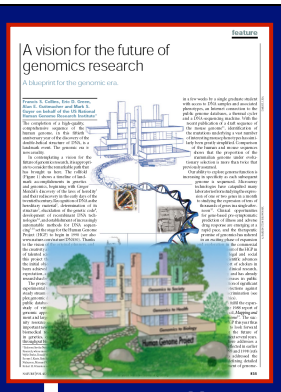
→ → ? → →

Human Genome Project

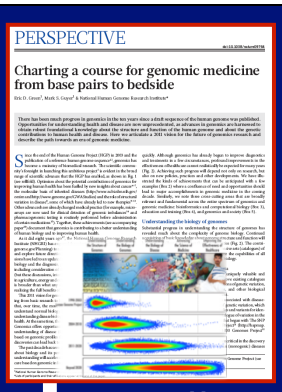
Realization of Genomic Medicine

✓ *"Fulfilling the Promise"*

?



Nature

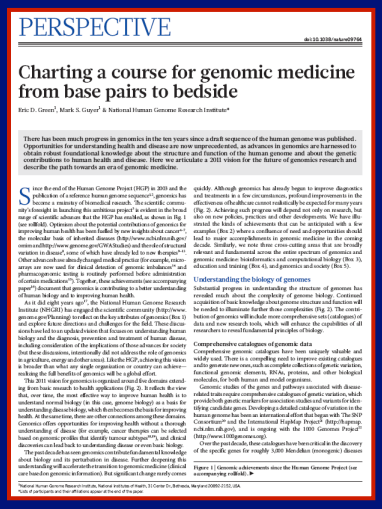


Nature

2003

2011

February, 2011



PERSPECTIVE

doi:10.1038/nature09764

Charting a course for genomic medicine from base pairs to bedside

Eric D. Green[1], Mark S. Guyer[1] & National Human Genome Research Institute*

There has been much progress in genomics in the ten years since a draft sequence of the human genome was published. Opportunities for understanding health and disease are now unprecedented, as advances in genomics are harnessed to obtain robust foundational knowledge about the structure and function of the human genome and about the genetic contributions to human health and disease. Here we articulate a 2011 vision for the future of genomics research and describe the path towards an era of genomic medicine.

Since the end of the Human Genome Project (HGP) in 2003 and the publication of a reference human genome sequence[1], genomics has become a mainstay of biomedical research. The scientific community's foresight in launching this ambitious project[2] is evident in the broad range of scientific advances that the HGP has enabled, as shown in Fig. 1 (see refBAD). Optimism about the potential contributions of genomics for improving human health has been fuelled by new insights about cancer[3–5], the molecular basis of inherited diseases (http://www.ncbi.nlm.nih.gov/omim and http://www.genome.gov/GWAStudies) and thousands of structural variants in diseases[6], some of which have already led to new therapies[7–11]. Other advances have already changed medical practice (for example, microarrays are now used for clinical detection of genomic imbalances[12] and pharmacogenomic testing is routinely performed before administration of certain medications[13]). Together, these achievements (see accompanying paper[14]) document that genomics is contributing to a better understanding of human biology and to improving human health.

As it did eight years ago[15], the National Human Genome Research Institute (NHGRI) has engaged the scientific community (http://www.genome.gov/Planning) to reflect on the key attributes of genomics (Box 1) and explore future directions and challenges for the field. These discussions have led to an updated vision that focuses on understanding human biology and the diagnosis, prevention and treatment of human disease, including consideration of the implications of those advances for society (but these discussions, intentionally did not address the role of genomics in agriculture, energy and other areas). Like the HGP, achieving this vision is broader than what any single organization or country can achieve—realizing the full benefits of genomics will be a global effort.

This 2011 vision for genomics is organized around five domains extending from basic research to health applications (Fig. 2). It reflects the view that, over time, the most effective way to improve human health is to understand normal biology (in this case, genome biology) as a basis for understanding disease biology, which then becomes the basis for improving health. At the same time, there are other connections among these domains. Genomics offers opportunities for improving health without a thorough understanding of disease (for example, cancer therapies can be selected based on genomic profiles that identify tumour subtypes[16–18]), and clinical discoveries can feed back to understanding disease or even basic biology.

The past decade has seen genomics contribute fundamental knowledge about biology and its perturbation in disease. Further deepening this understanding will accelerate the transition to genomic medicine (clinical care based on genomic information). But significant change rarely comes quickly. Although genomics has already begun to improve diagnostics and treatments in a few circumstances, profound improvements in the effectiveness of healthcare cannot realistically be expected for many years (Fig. 2). Achieving such progress will depend not only on research, but also on new policies, practices and other developments. We have illustrated the kinds of achievements that can be anticipated with a few examples (Box 2) where a confluence of need and opportunities should lead to major accomplishments in genomic medicine in the coming decade. Similarly, we note three cross-cutting areas that are broadly relevant and fundamental across the entire spectrum of genomics and genomic medicine: bioinformatics and computational biology (Box 3), education and training (Box 4), and genomics and society (Box 5).

Understanding the biology of genomes

Substantial progress in understanding the structure of genomes has revealed much about the complexity of genome biology. Continued acquisition of basic knowledge about genome structure and function will be needed to illuminate further those complexities (Fig. 2). The contribution of genomics will include more comprehensive sets (catalogues) of data and new research tools, which will enhance the capabilities of all researchers to reveal fundamental principles of biology.

Comprehensive catalogues of genomic data

Comprehensive genomic catalogues have been uniquely valuable and widely used. There is a compelling need to improve existing catalogues and to generate new ones, such as complete collections of genetic variation, functional genomic elements, RNAs, proteins, and other biological molecules, for both human and model organisms.

Genomic studies of the genes and pathways associated with disease, related traits require comprehensive catalogues of genetic variation, which provide both genetic markers for association studies and variants for identifying candidate genes. Developing a detailed catalogue of variation in the human genome has been an international effort that began with The SNP Consortium[19] and the International HapMap Project[20] (http://hapmap.ncbi.nlm.nih.gov), and is ongoing with the 1000 Genomes Project[21] (http://www.1000genomes.org).

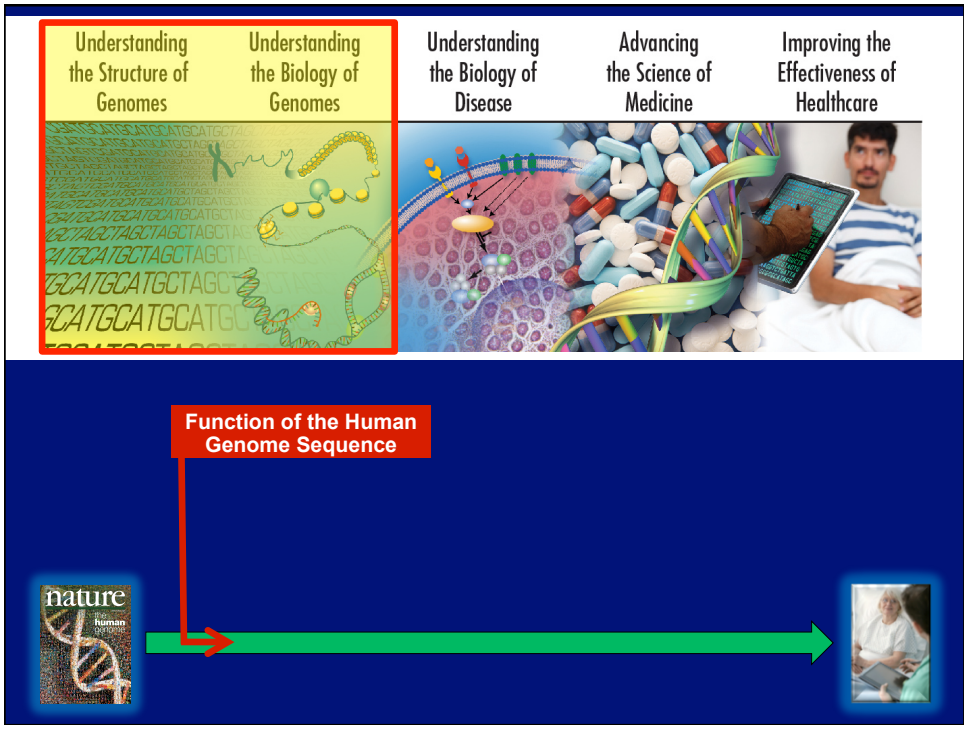
Over the past decade, these catalogues have been critical in the discovery of the specific genes for roughly 3,000 Mendelian (monogenic) diseases

Figure 1 | Genomic achievements since the Human Genome Project (see accompanying editorial). ▶

*National Human Genome Research Institute, National Institutes of Health, 31 Center Dr, Bethesda, Maryland 20892-2152, USA.
†Lists of participants and their affiliations appear at the end of the paper.

New NHGRI Vision for Genomics Published

Five Domains of Genomics Research



Mapping the Human Genome — ~1990 to ~2000

Sequencing the Human Genome — ~1998 to ~2003

The Human Genome Project

Interpreting the Human Genome Sequence — ~2003 to ???

Beyond The Human Genome Project

~3,000 bp (0.0001%) of Human Genome Sequence

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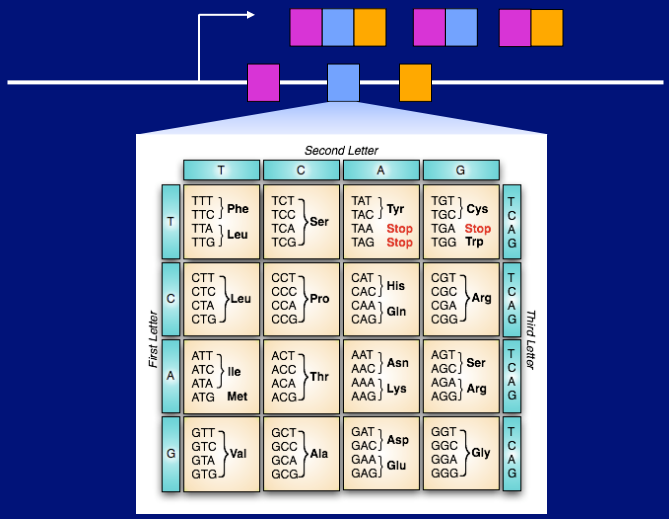
Coding Sequences (i.e., Genes)



The Genetic Code

~3,000 bp (0.0001%) of Human Genome Sequence

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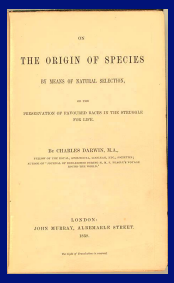
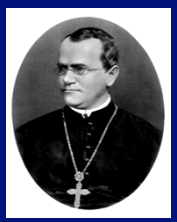
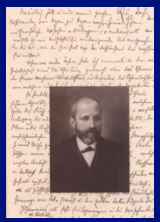
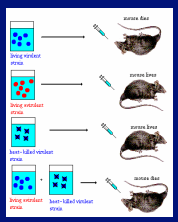
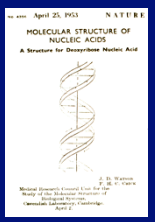
Foundational Milestones in Genetics & Genomics







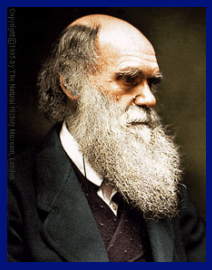




Darwin	Mendel	Miescher	Avery	Watson & Crick
1859	*1865*	*1871*	*1944*	*1953*

"It is not the strongest of the species that survives, nor the most intelligent that survives. It is the one that is the most adaptable to change."

(Attributed to Darwin)

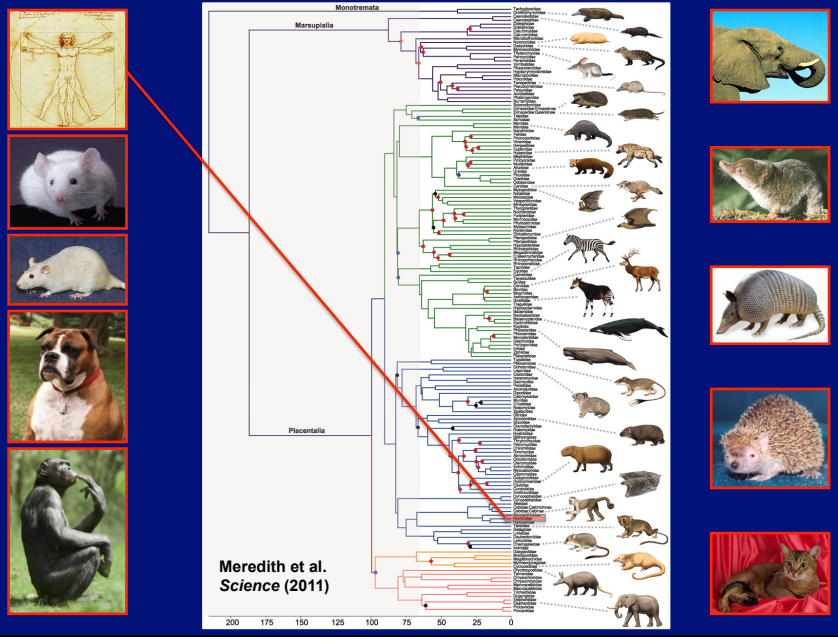


Charles Darwin (1809-1882)

"For the last three and a half billion years, evolution has been taking notes."

— Eric Lander

Comparative Genome Sequencing



The Human Genome: By the Numbers

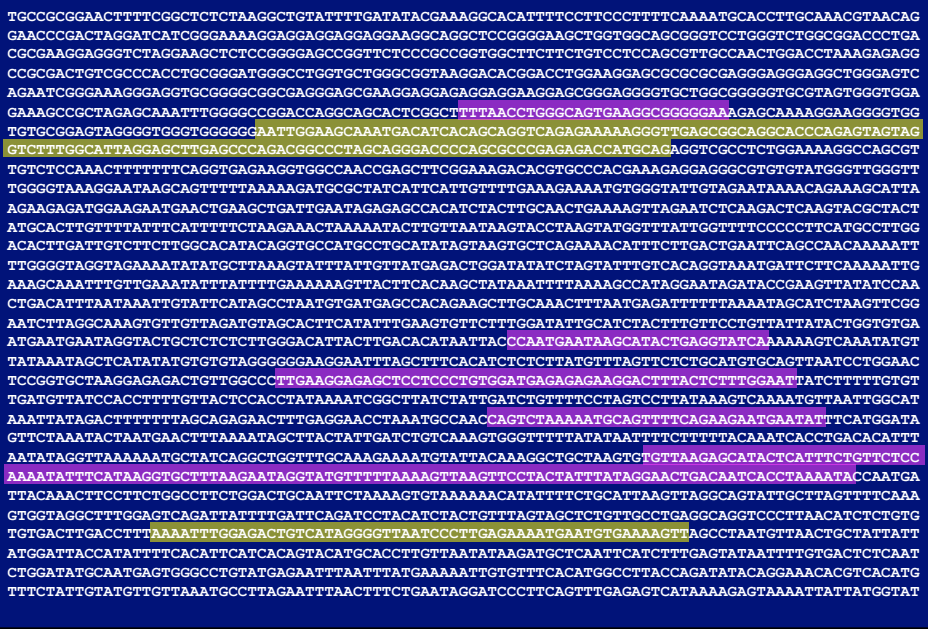
~5% of Human Genome Sequence is Constrained Across Mammals (and Presumed Functional)

- 5% of 3B Bases = ~150M Bases
- Lower Bound for the Amount that is Functional

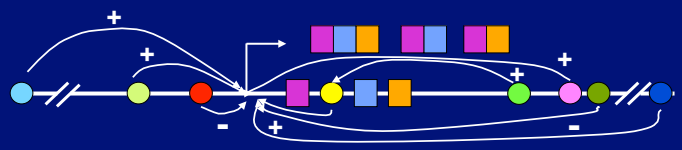
~1.5% Encodes for Protein (Genes)

- Corresponds to ~18-22K Genes
- Many More than ~22K Different Proteins

~3,000 bp (0.0001%) of Human Genome Sequence



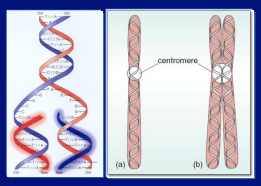
Non-Coding Functional Sequences



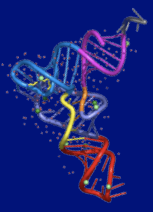
Gene Regulation

Chromosome Packaging

Chromosome Segregation



Chromosome Replication



Non-Coding RNAs

?

The Human Genome: By the Numbers

~5% of Human Genome Sequence is Constrained Across Mammals (and Presumed Functional)

- 5% of 3B Bases = ~150M Bases
- Do NOT Yet Know the Position of these ~150M Functional Bases
- Lower Bound for the Amount that is Functional

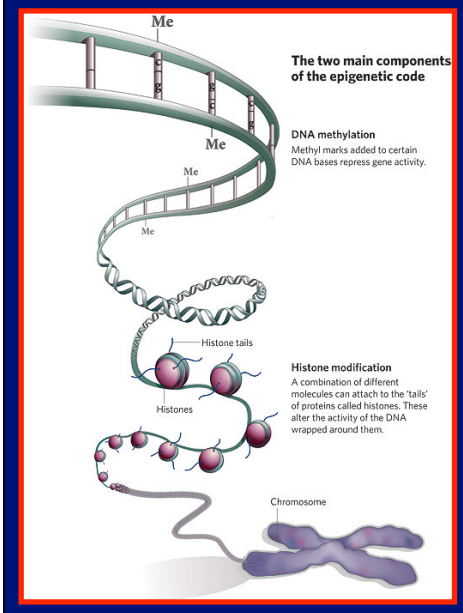
~1.5% Encodes for Protein (Genes)

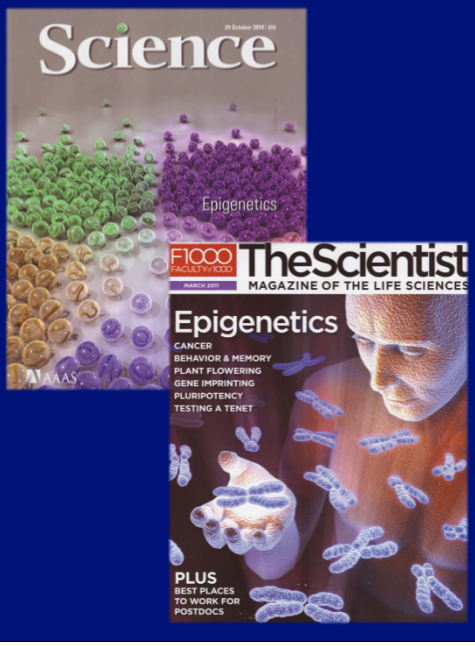
- Corresponds to ~18-22K Genes
- Many More than ~22K Different Proteins

~3.5% Functional But Non-Coding

- Gene Regulatory Elements
- Chromosomal Functional Elements
- Undiscovered Functional Elements (NOT Yet in Textbooks!)

The Epigenomic Landscape

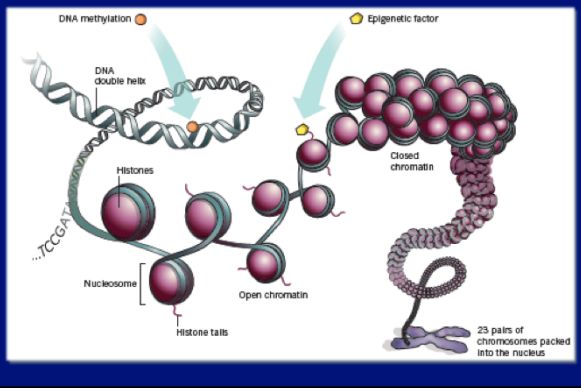




TECHNOLOGY FEATURE

READING THE SECOND GENOMIC CODE

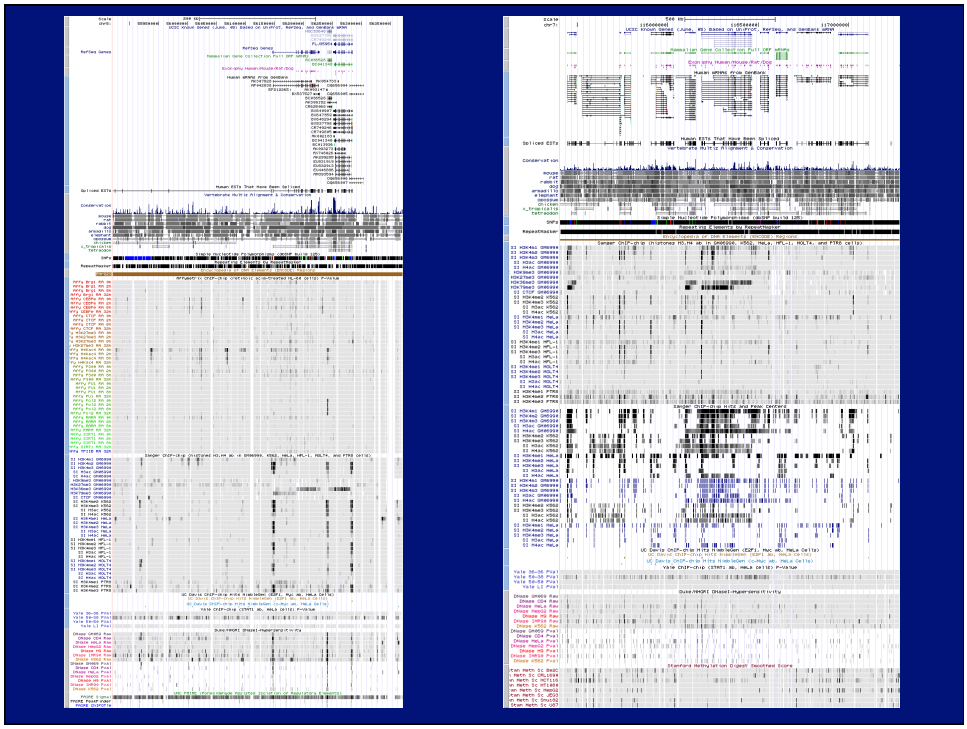
***Nature* (2012)**

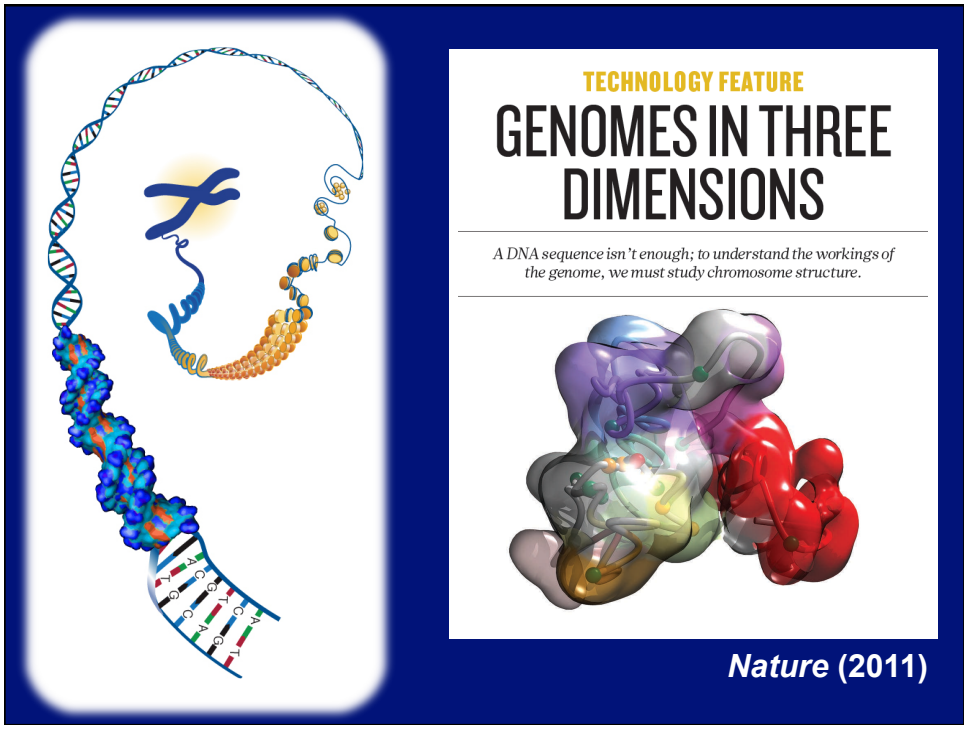


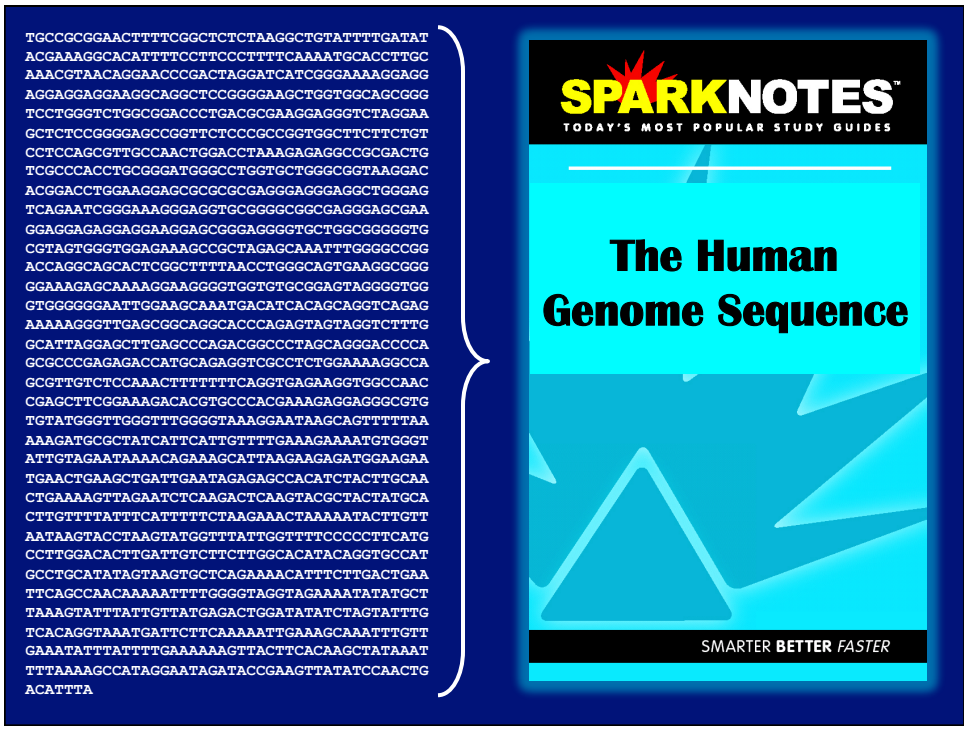
The ENCODE Portfolio

ENCODE

mod ENCODE




TECHNOLOGY FEATURE
GENOMES IN THREE DIMENSIONS
A DNA sequence isn't enough; to understand the workings of the genome, we must study chromosome structure.
Nature (2011)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AAACGTAACAGGAACCCGACTAGGATCATCGGGAAAAGGAGG
AGGAGGAGGAAGGCAGGCTCCGGGGAAGCTGGTGGCAGCGGG
TCCTGGGTCTGGCGGACCCTGACGCGAAGGAGGGTCTAGGAA
GCTCTCCGGGGAGCCGGTTCTCCCGCCGGTGGCTTCTTCTGT
CCTCCAGCGTTGCCAACTGGACCTAAAGAGAGGCCGCGACTG
TCGCCCACCTGCGGGATGGGCCTGGTGCTGGGCGGTAAGGAC
ACGGACCTGGAAGGAGCGCGCGCGAGGGAGGGAGGCTGGGAG
TCAGAATCGGGAAAGGGAGGTGCGGGGCGGCGAGGGAGCGAA
GGAGGAGAGGAGGAAGGAGCGGGAGGGGTGCTGGCGGGGGTG
CGTAGTGGGTGGAGAAAGCCGCTAGAGCAAATTTGGGGCCGG
ACCAGGCAGCACTCGGCTTTTAACCTGGGCAGTGAAGGCGGG
GGAAAGAGCAAAAGGAAGGGGTGGTGTGCGGAGTAGGGGTGG
GTGGGGGGAATTGGAAGCAAATGACATCACAGCAGGTCAGAG
AAAAAGGGTTGAGCGGCAGGCACCCAGAGTAGTAGGTCTTTG
GCATTAGGAGCTTGAGCCCAGACGGCCCTAGCAGGGACCCCA
GCGCCCGAGAGACCATGCAGAGGTCGCCTCTGGAAAAGGCCA
GCGTTGTCTCCAAACTTTTTTTCAGGTGAGAAGGTGGCCAAC
CGAGCTTCGGAAAGACACGTGCCCACGAAAGAGGAGGGCGTG
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AAAGATGCGCTATCATTCATTGTTTTGAAAGAAAATGTGGGT
ATTGTAGAATAAAACAGAAAGCATTAAGAAGAGATGGAAGAA
TGAACTGAAGCTGATTGAATAGAGAGCCACATCTACTTGCAA
CTGAAAAGTTAGAATCTCAAGACTCAAGTACGCTACTATGCA
CTTGTTTTATTTCATTTTTCTAAGAAACTAAAAATACTTGTT
AATAAGTACCTAAGTATGGTTTATTGGTTTTCCCCCTTCATG
CCTTGGACACTTGATTGTCTTCTTGGCACATACAGGTGCCAT
GCCTGCATATAGTAAGTGCTCAGAAAACATTTCTTGACTGAA
TTCAGCCAACAAAAATTTTGGGGTAGGTAGAAAATATATGCT
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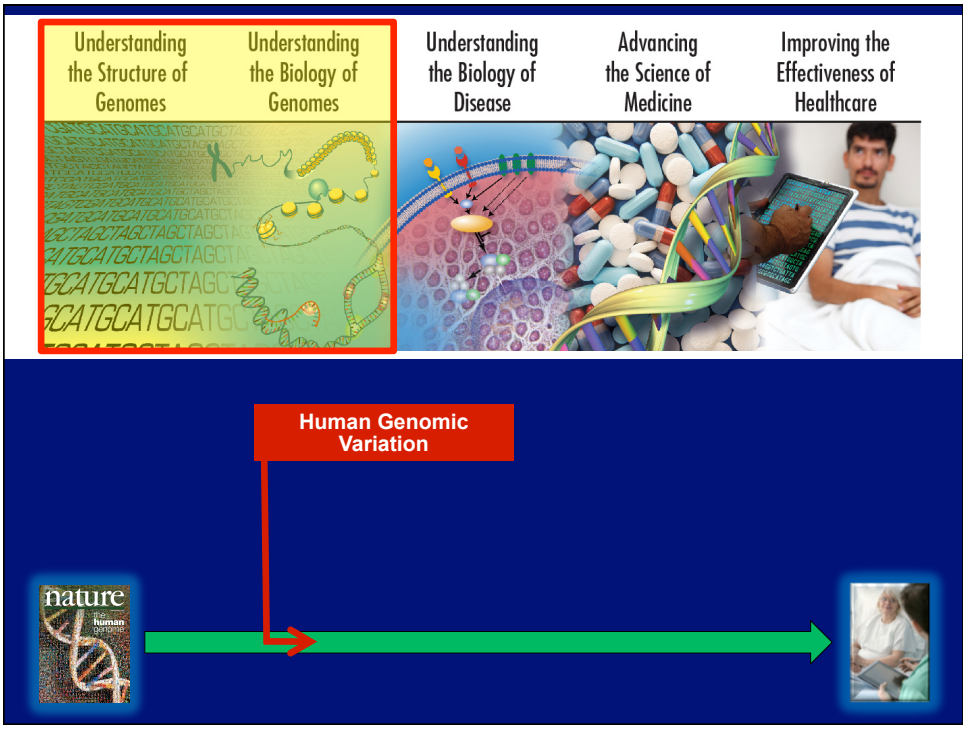
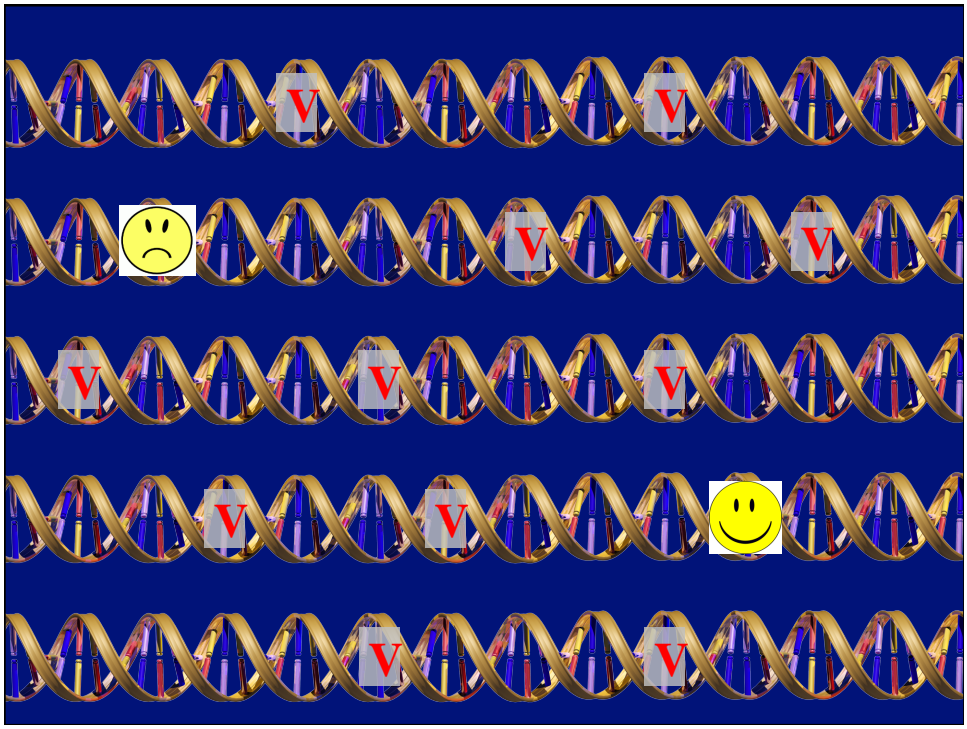
TCACAGGTAAATGATTCTTCAAAAATTGAAAGCAAATTTGTT
GAAATATTTATTTTGAAAAAAGTTACTTCACAAGCTATAAAT
TTTAAAAGCCATAGGAATAGATACCGAAGTTATATCCAACTG
ACATTTA
SPARKNOTES
TODAY'S MOST POPULAR STUDY GUIDES
The Human Genome Sequence
SMARTER BETTER FASTER

The Genomics of Human Evolution





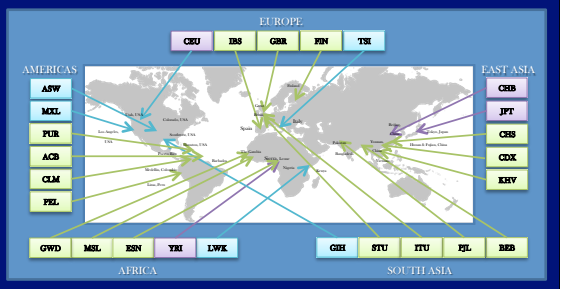

International HapMap Project
27 October 2005 | www.nature.com/nature | $10
THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE
nature
INSIDE
Why do we sleep?
OPTOELECTRONICS
Germanium boost for silicon chips
LAW OF THE JUNGLE
Don't ask a chimpanzee for help
MEN OF LETTERS
If Darwin and Einstein had e-mail...
THE HAPMAP PROJECT
Chapter and verse on human genetic variation
NATUREJOBS
Biodefence boom
A haplotype map of the human genome
The International HapMap Consortium*
Inherited genetic variation has a critical but as yet largely uncharacterized role in human disease. Here we report a public database of common variation in the human genome: more than one million single nucleotide polymorphisms (SNPs) for which accurate and complete genotypes have been obtained in 269 DNA samples from four populations, including ten 500-kilobase regions in which essentially all information about common DNA variation has been extracted. These data document the generality of recombination hotspots, a block-like structure of linkage disequilibrium and low haplotype diversity, leading to substantial correlations of SNPs with many of their neighbours. We show how the HapMap resource can guide the design and analysis of genetic association studies, shed light on structural variation and recombination, and identify loci that may have been subject to natural selection during human evolution.
2005
A second generation human haplotype map of over 3.1 million SNPs
The International HapMap Consortium*
We describe the Phase II HapMap, which characterizes over 3.1 million human single nucleotide polymorphisms (SNPs) genotyped in 270 individuals from four geographically diverse populations and includes 25–35% of common SNP variation in the populations surveyed. The map is estimated to capture untyped common variation with an average maximum r^2 of between 0.9 and 0.96 depending on population. We demonstrate that the current generation of commercial genome-wide genotyping products captures common Phase II SNPs with an average maximum r^2 of up to 0.8 in African and up to 0.95 in non-African populations, and that potential gains in power in association studies can be obtained through imputation. These data also reveal novel aspects of the structure of linkage disequilibrium. We show that 10–30% of pairs of individuals within a population share at least one region of extended genetic identity arising from recent ancestry and that up to 1% of all common variants are untaggable, primarily because they lie within recombination hotspots. We show that recombination rates vary systematically around genes and between genes of different function. Finally, we demonstrate increased differentiation at non-synonymous, compared to synonymous, SNPs, resulting from systematic differences in the strength or efficacy of natural selection between populations.
2007
Integrating common and rare genetic variation in diverse human populations
The International HapMap 3 Consortium*
Despite great progress in identifying genetic variants that influence human disease, most inherited risk remains unexplained. A more complete understanding requires genome-wide studies that fully examine less common alleles in populations with a wide range of ancestry. To inform the design and interpretation of such studies, we genotyped 1.6 million common single nucleotide polymorphisms (SNPs) in 1,184 reference individuals from 11 global populations, and sequenced ten 100-kilobase regions in 692 of these individuals. This integrated data set of common and rare alleles, called 'HapMap 3', includes both SNPs and copy number polymorphisms (CNPs). We characterized population-specific differences among low-frequency variants, measured the improvement in imputation accuracy afforded by the larger reference panel, especially in imputing SNPs with a minor allele frequency of <5%, and demonstrated the feasibility of imputing newly discovered CNPs and SNPs. This expanded public resource of genome variants in global populations supports deeper interrogation of genomic variation and its role in human disease, and serves as a step towards a high-resolution map of the landscape of human genetic variation.
2010

1000 Genomes

A Deep Catalog of Human Genetic Variation

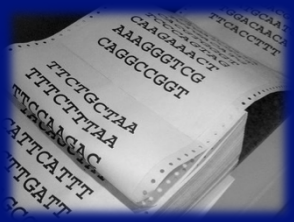
Nature (2010)

Nature (2012)

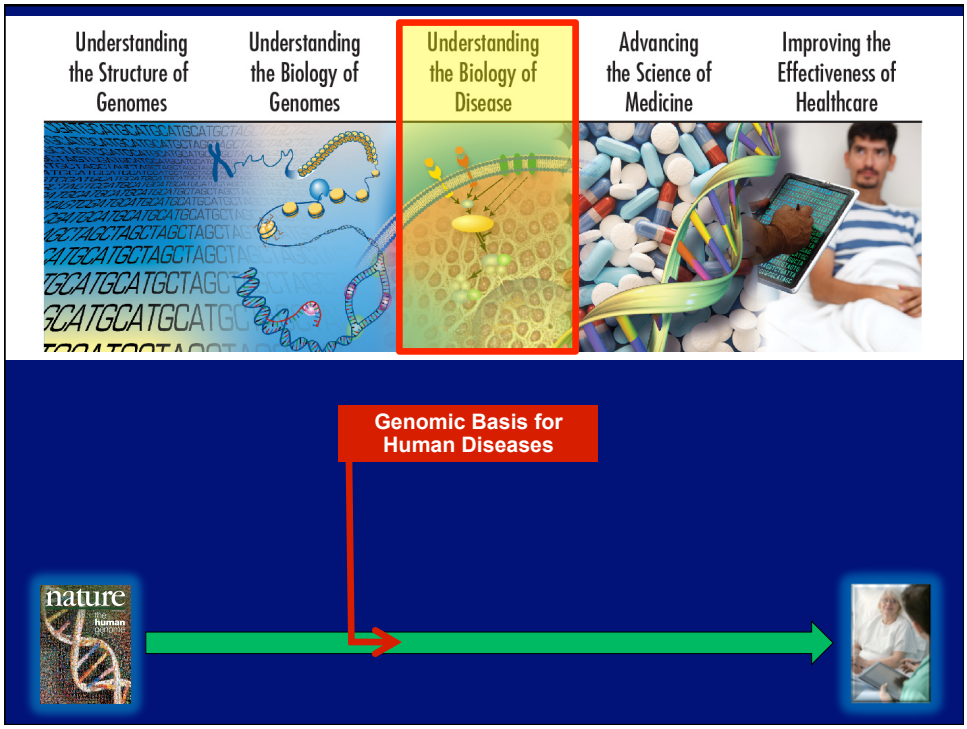


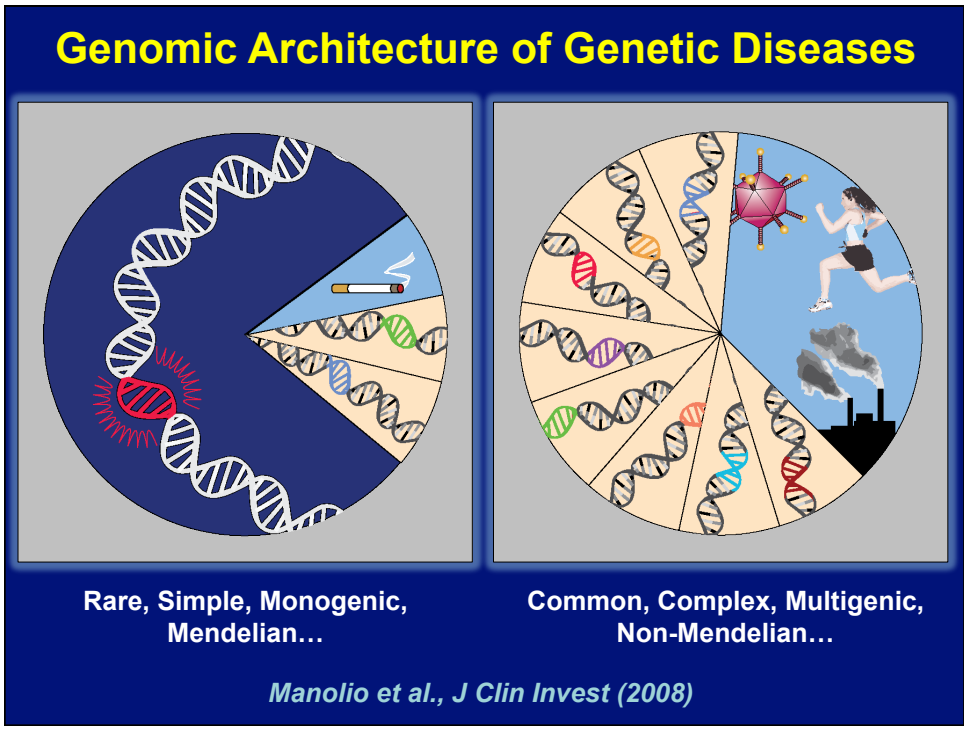
2535 Humans, 26 Populations

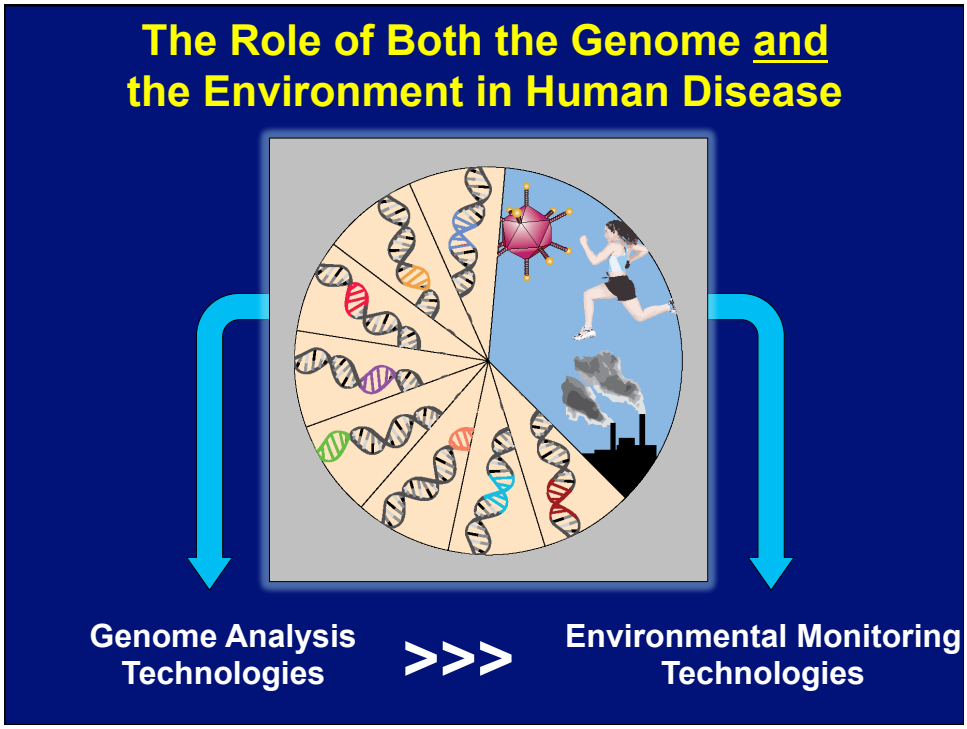
Your Genome: By the Numbers

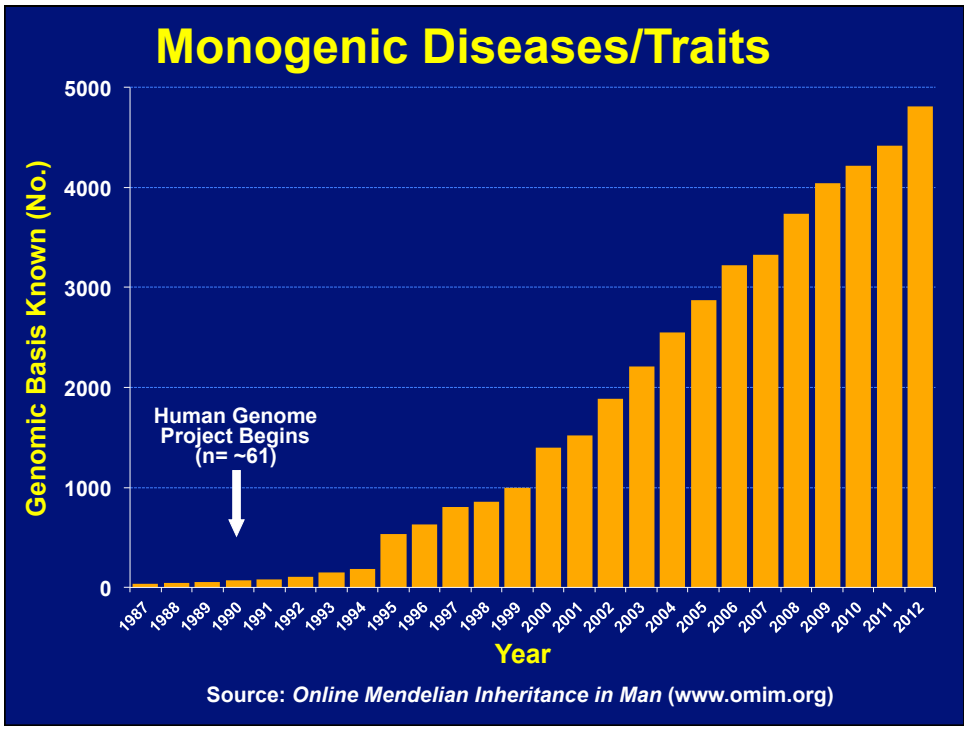


- ~6B nucleotides
- ~3-5M single-nucleotide variants
  - ~150K not in databases
  - ~60 not in either parent
- ~100 'disruptive' variants in genes
- ~20 completely inactivated genes (both copies)

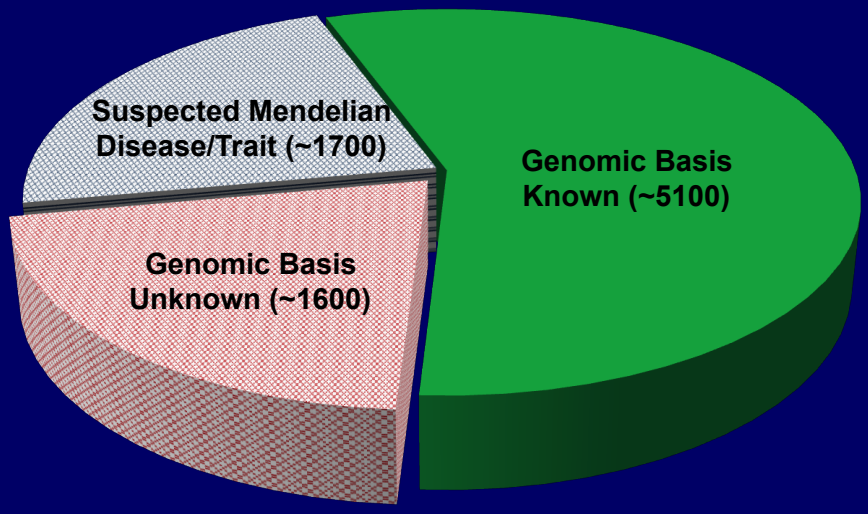

Understanding the Structure of Genomes
Understanding the Biology of Genomes
Understanding the Biology of Disease
Advancing the Science of Medicine
Improving the Effectiveness of Healthcare
Genomic Basis for Human Diseases
nature


Genomic Architecture of Genetic Diseases
Rare, Simple, Monogenic, Mendelian…
Common, Complex, Multigenic, Non-Mendelian…
Manolio et al., J Clin Invest (2008)


The Role of Both the Genome and the Environment in Human Disease
Genome Analysis Technologies
>>>
Environmental Monitoring Technologies

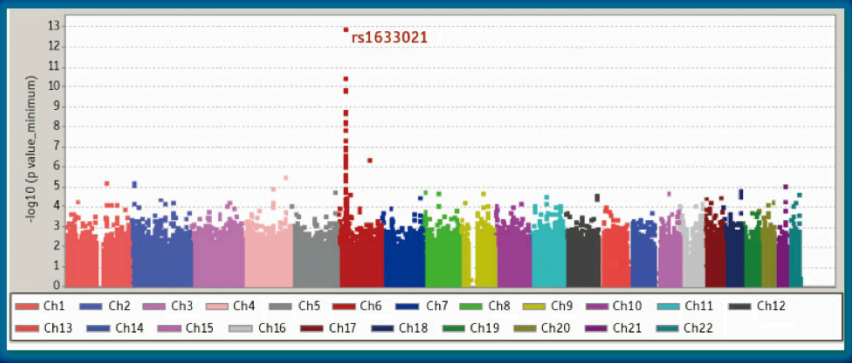

Monogenic Diseases/Traits
Genomic Basis Known (No.)
5000
4000
3000
2000
1000
0
Human Genome Project Begins (n= ~61)
1987
1988
1989
1990
1991
1992
1993
1994
1995
1996
1997
1998
1999
2000
2001
2002
2003
2004
2005
2006
2007
2008
2009
2010
2011
2012
Year
Source: Online Mendelian Inheritance in Man (www.omim.org)

Monogenic Diseases/Traits



Source: ***Online Mendelian Inheritance in Man*** (www.omim.org)

Genome-Wide Association Studies (GWAS)



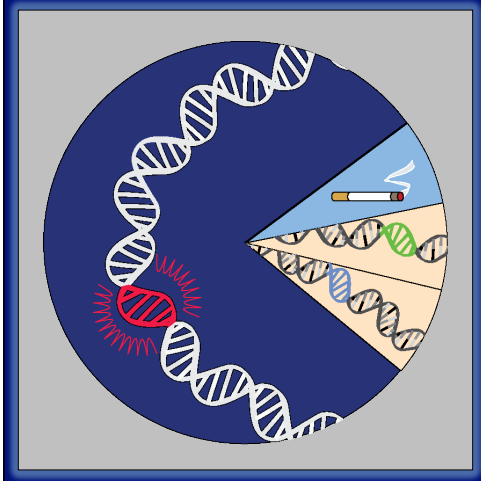
The First GWAS Success Story: Age-Related Macular Degeneration

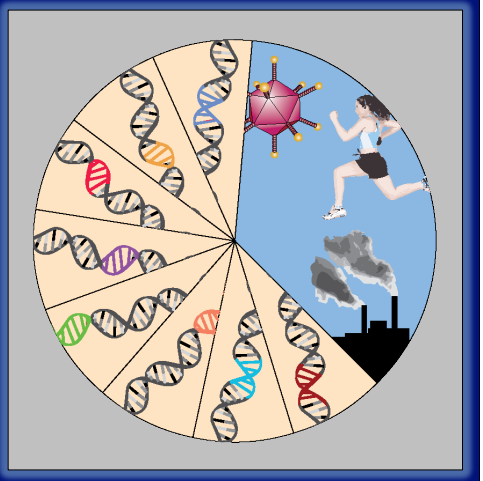
Complement Factor H Polymorphism in Age-Related Macular Degeneration

Robert J. Klein,[1] Caroline Zeiss,[2*] Emily Y. Chew,[3*] Jen-Yue Tsai,[4*] Richard S. Sackler,[1] Chad Haynes,[1] Alice K. Henning,[5] John Paul SanGiovanni,[3] Shrikant M. Mane,[6] Susan T. Mayne,[7] Michael B. Bracken,[7] Frederick L. Ferris,[3] Jurg Ott,[1] Colin Barnstable,[2] Josephine Hoh[7†]

***Science* (2005)**

Genomic Architecture of Genetic Diseases



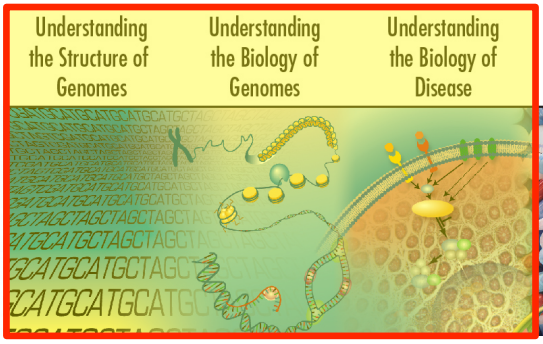


Rare, Simple, Monogenic, Mendelian…

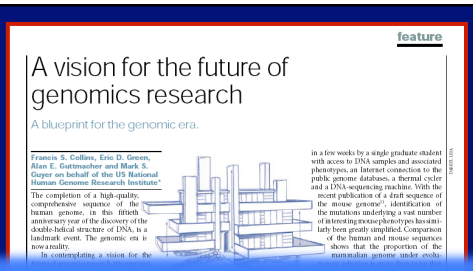
Mostly *Coding* Mutations

Common, Complex, Multigenic, Non-Mendelian…

Mostly *Non-Coding* Mutations



Routine Whole-Genome Sequencing


feature

A vision for the future of genomics research

A blueprint for the genomic era.

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

The completion of a high-quality, comprehensive sequence of the human genome, in this fiftieth anniversary year of the discovery of the double-helical structure of DNA, is a landmark event. The genomic era is now a reality.

In contemplating a vision for the

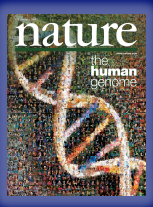
in a few weeks by a single graduate student with access to DNA samples and associated phenotypes, an Internet connection to the public genome databases, a thermal cycler and a DNA-sequencing machine. With the recent publication of a draft sequence of the mouse genome[1], identification of the mutations underlying a vast number of interesting mouse phenotypes has similarly been greatly simplified. Comparison of the human and mouse sequences shows that the proportion of the mammalian genome under evolu-

"…'technological leaps' that seem so far off as to be almost fictional but which, if they could be achieved, would revolutionize biomedical research and clinical practice.

[For example,]…the ability to sequence DNA at costs that are lower by four to five orders of magnitude than the current cost, allowing a human genome to be sequenced for $1,000 or less."

Nature, April 2003

Human Genome Sequence



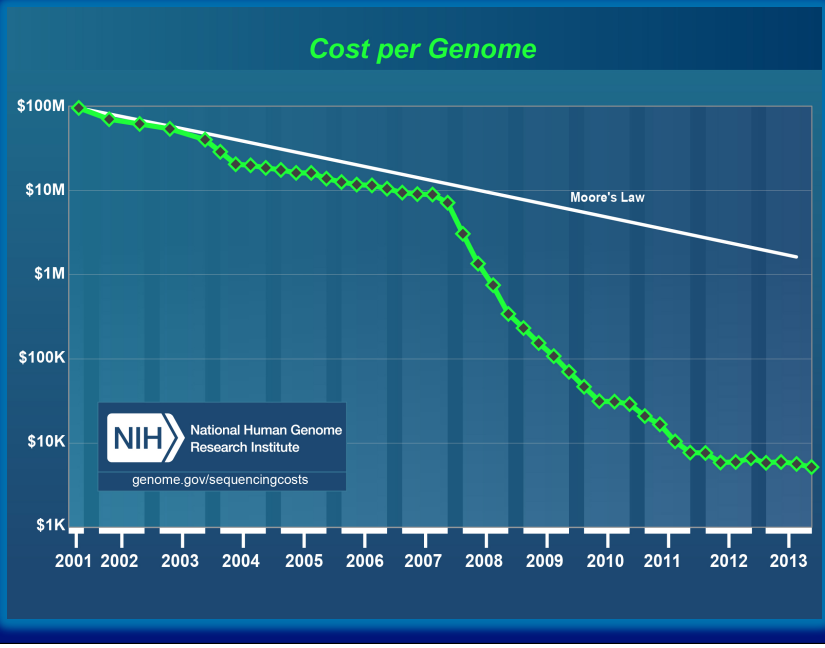
~$1,000,000,000

~$1,000

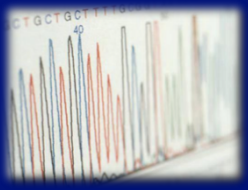
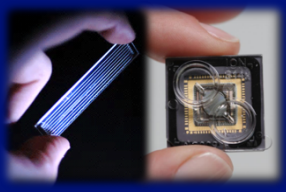
"The $1000 Genome"


POLONATOR G.007

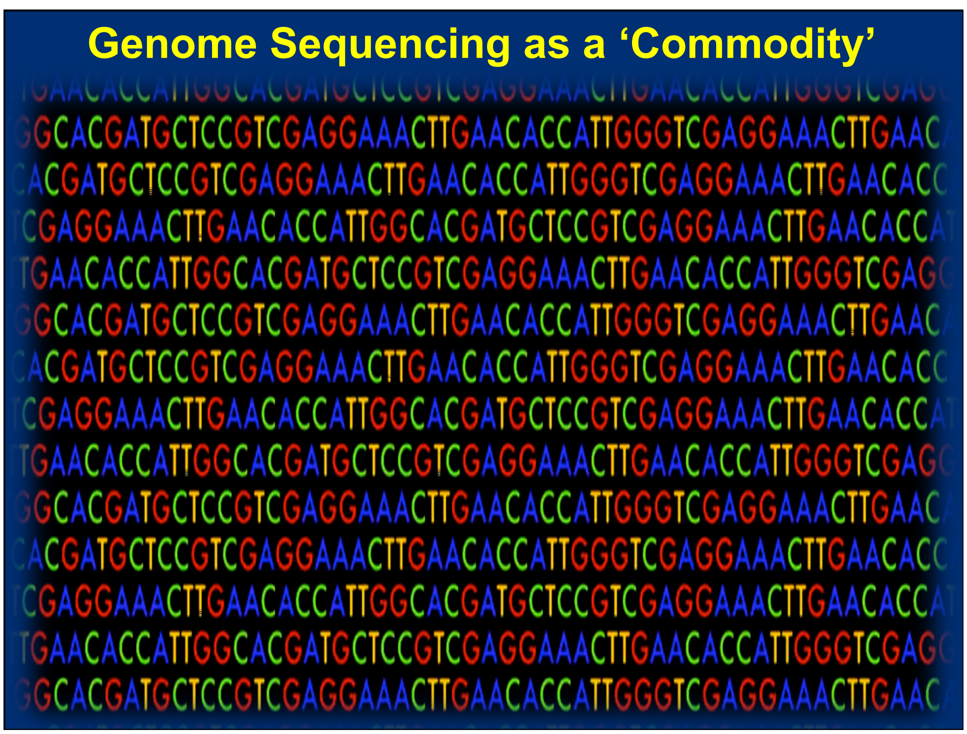
Cost per Sequenced Human Genome

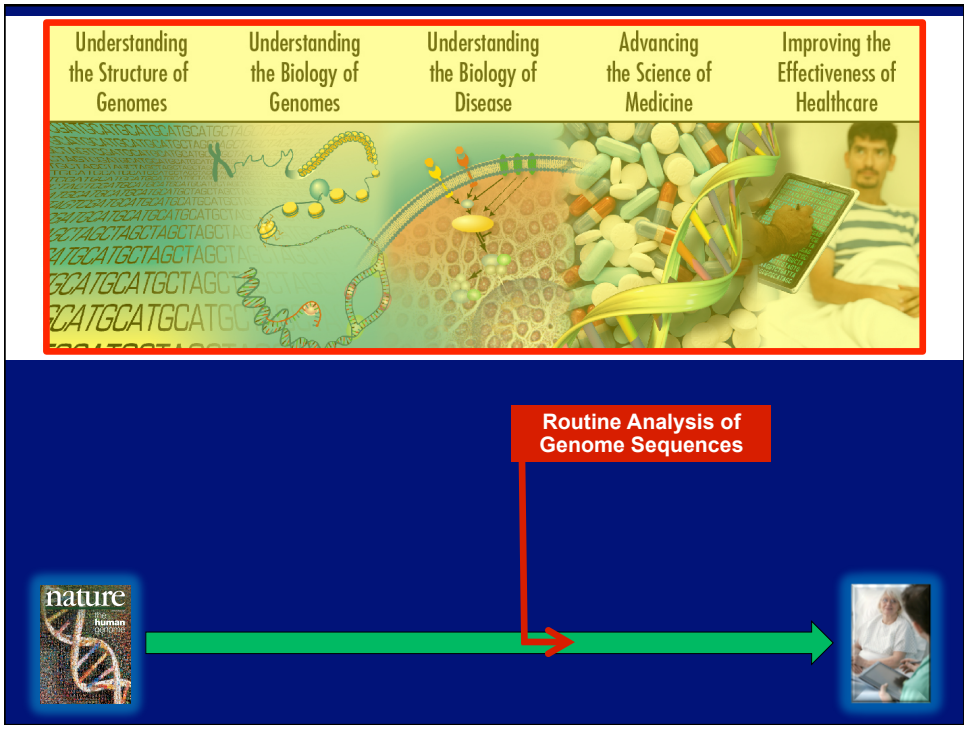


Sequencing a Human Genome

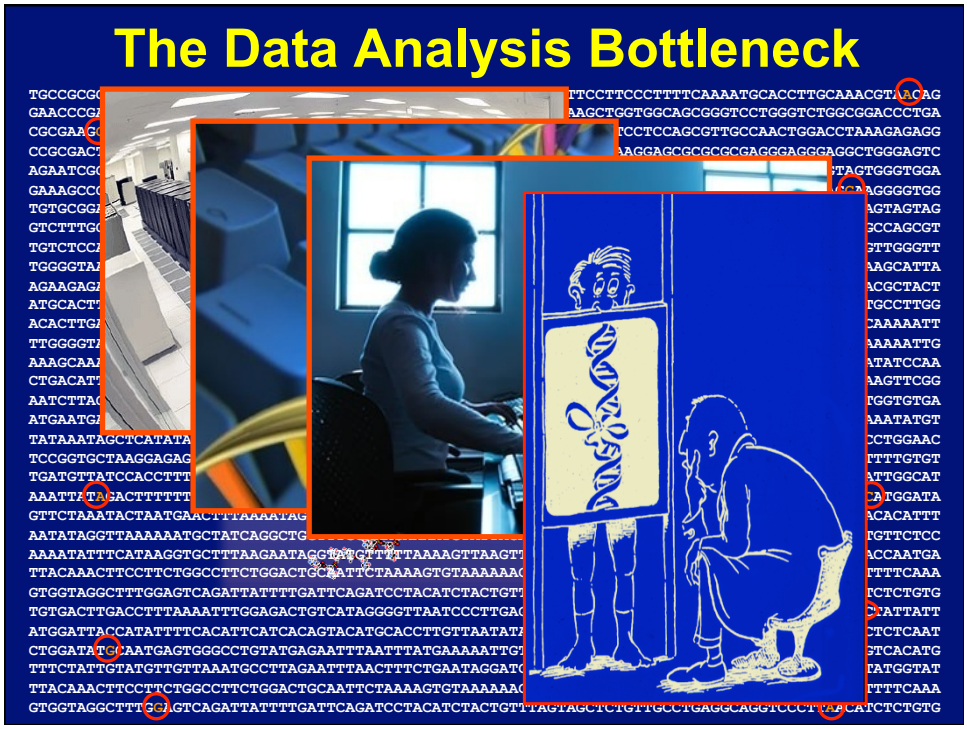
HGP (1st Sequence)	*Immediate Post-HGP*	*Today*
		
~6-8 years	~3-4 months	~2-3 days
~$1B	~$10-50M	~$4-6K

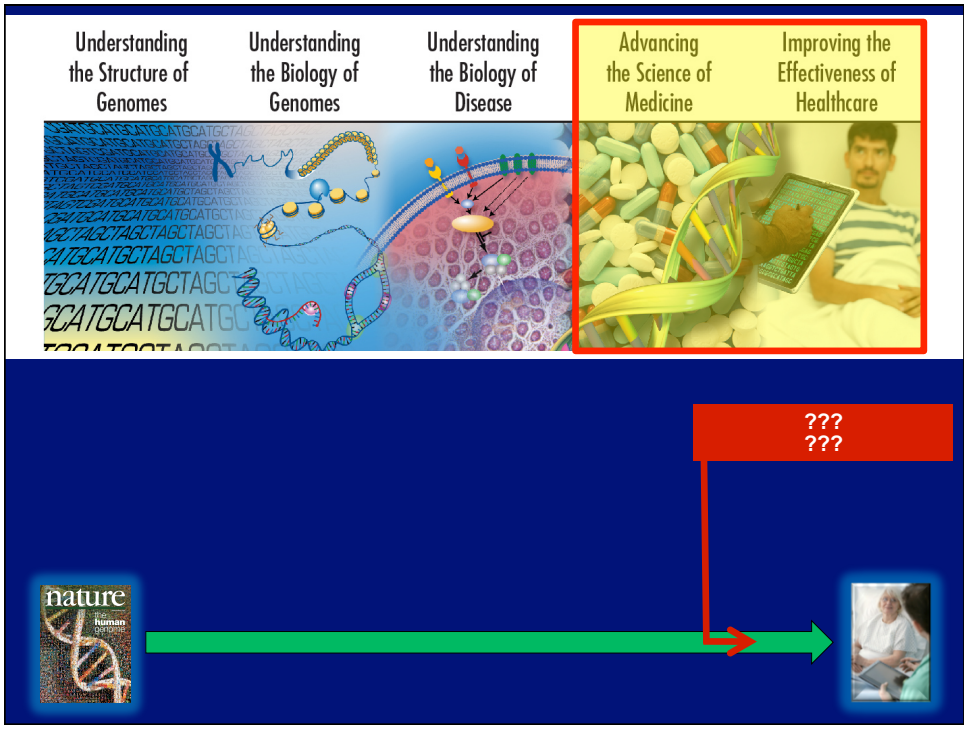

And Yet Newer Technologies…
Search for Pore-fection
MinION


Genome Sequencing as a 'Commodity'

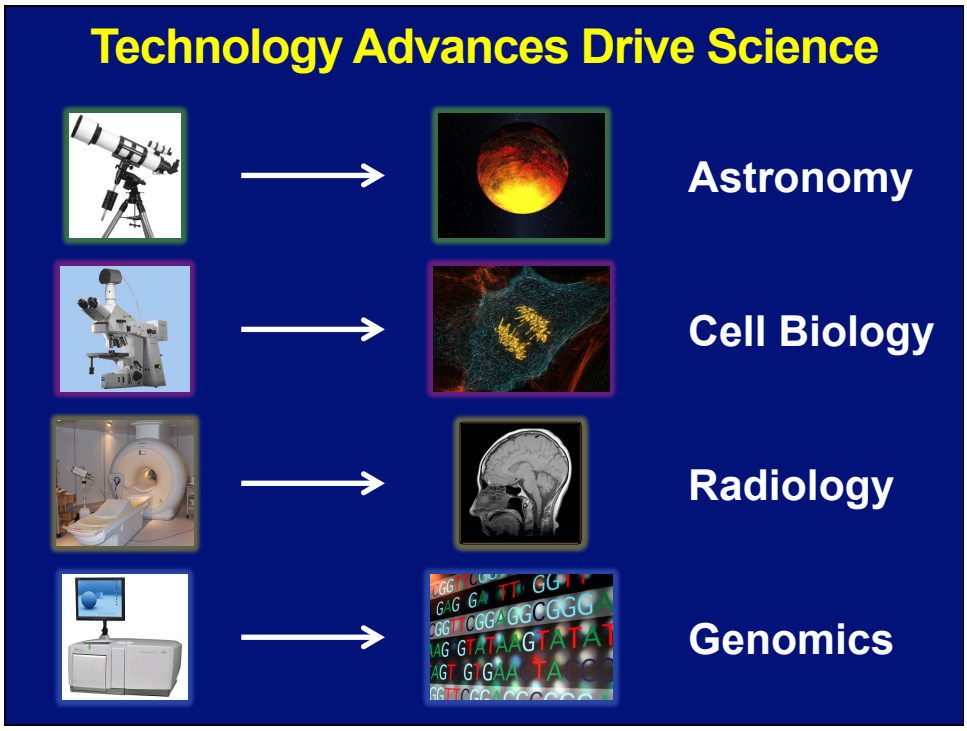

Understanding the Structure of Genomes
Understanding the Biology of Genomes
Understanding the Biology of Disease
Advancing the Science of Medicine
Improving the Effectiveness of Healthcare
Routine Analysis of Genome Sequences
nature

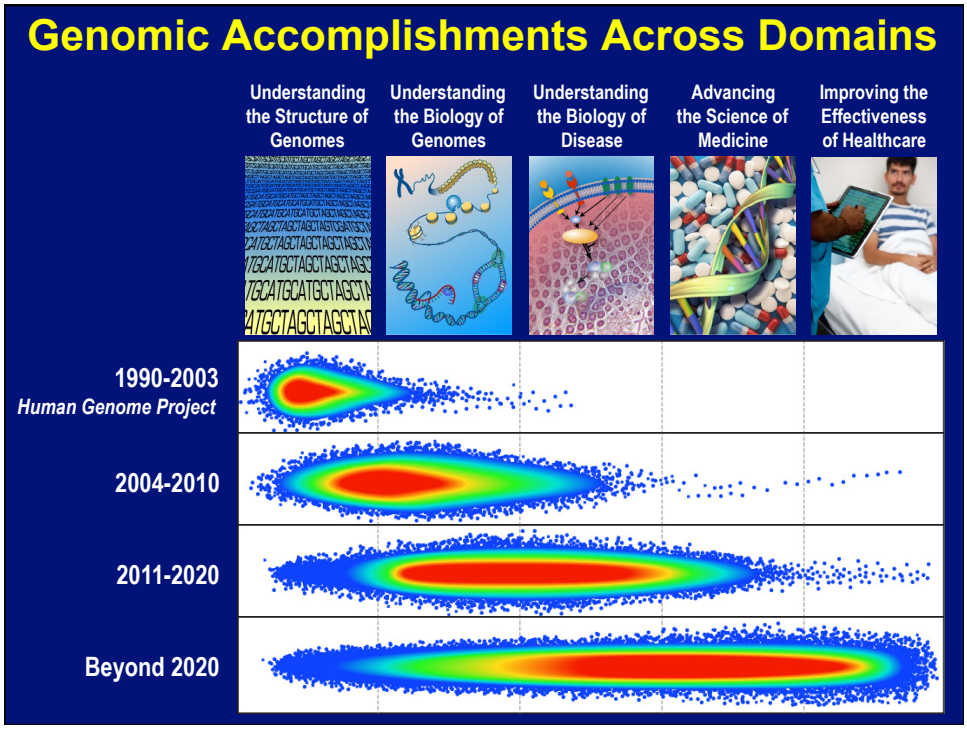

The Largest Current Bottleneck in Genomics…

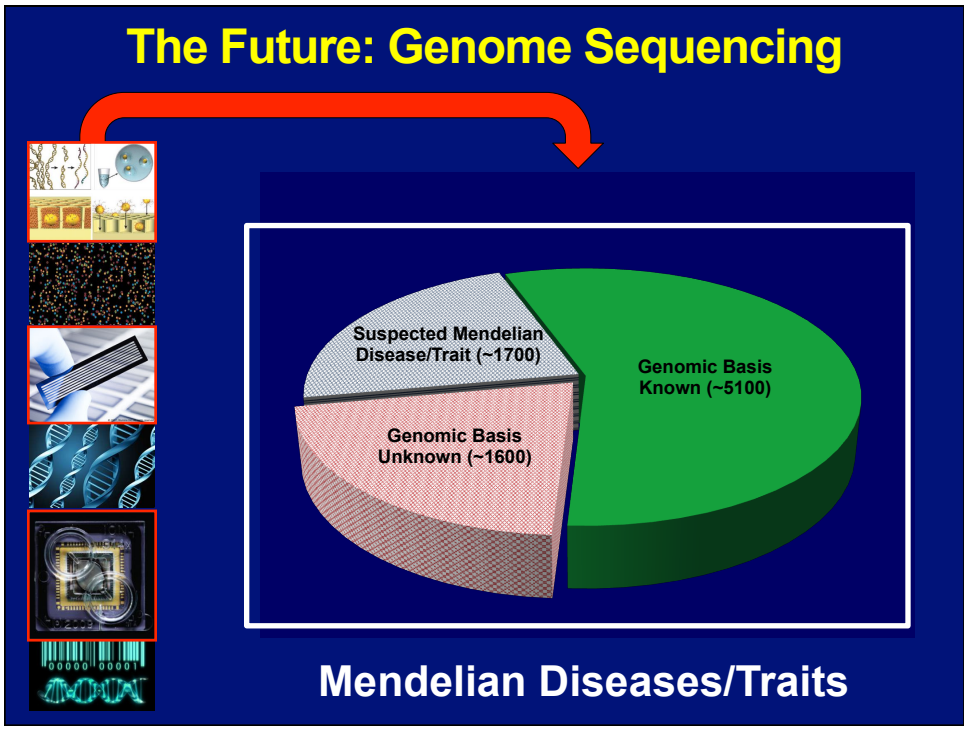

The Data Analysis Bottleneck


Understanding the Structure of Genomes
Understanding the Biology of Genomes
Understanding the Biology of Disease
Advancing the Science of Medicine
Improving the Effectiveness of Healthcare
???
???
nature

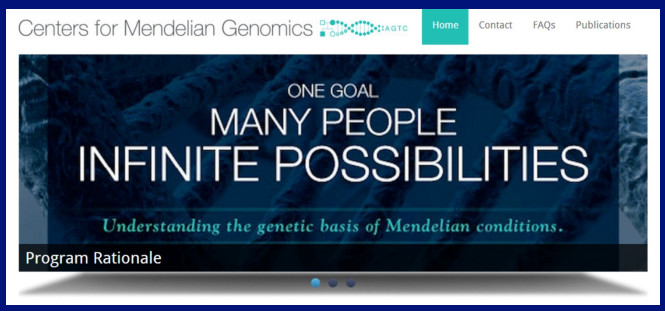

The Future
NEXT EXIT


Technology Advances Drive Science
Astronomy
Cell Biology
Radiology
Genomics


Genomic Accomplishments Across Domains
Understanding the Structure of Genomes
Understanding the Biology of Genomes
Understanding the Biology of Disease
Advancing the Science of Medicine
Improving the Effectiveness of Healthcare
1990-2003
Human Genome Project
2004-2010
2011-2020
Beyond 2020


The Future: Genome Sequencing
Suspected Mendelian Disease/Trait (~1700)
Genomic Basis Known (~5100)
Genomic Basis Unknown (~1600)
Mendelian Diseases/Traits

Centers for Mendelian Genomics



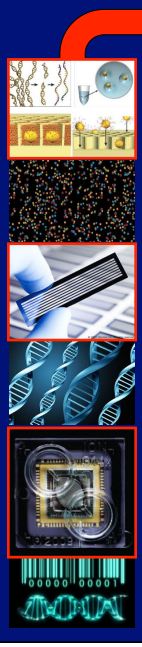
www.mendelian.org

The Centers for Mendelian Genomics: A New Large-Scale Initiative to Identify the Genes Underlying Rare Mendelian Conditions

Michael J. Bamshad,[1,2,3]* Jay A. Shendure,[2] David Valle,[4] Ada Hamosh,[4] James R. Lupski,[5,6,7,8] Richard A. Gibbs,[5,8] Eric Boerwinkle,[8,9] Richard P. Lifton,[10] Mark Gerstein,[11] Murat Gunel,[10,12] Shrikant Mane,[10] and Deborah A. Nickerson[2] on behalf of the Centers for Mendelian Genomics

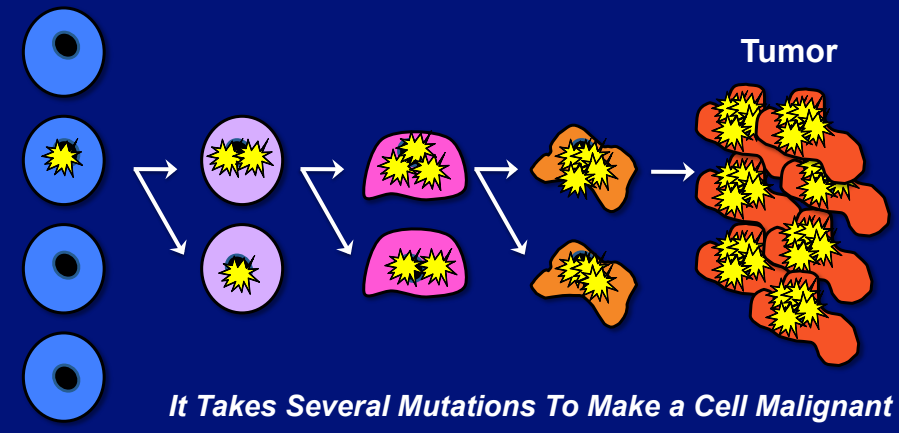
***Am J Med Genet* (2012)**

The Future: Genome Sequencing



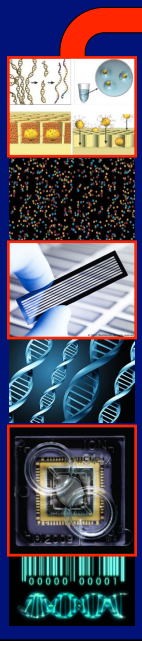
Complex Diseases/Traits

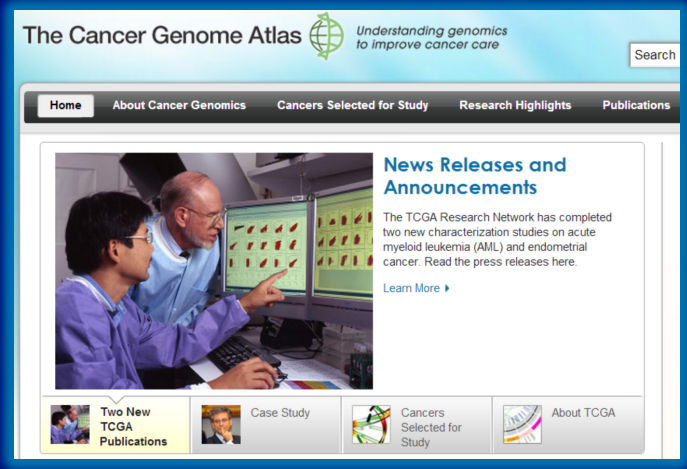
Cancer is a Disease of the Genome



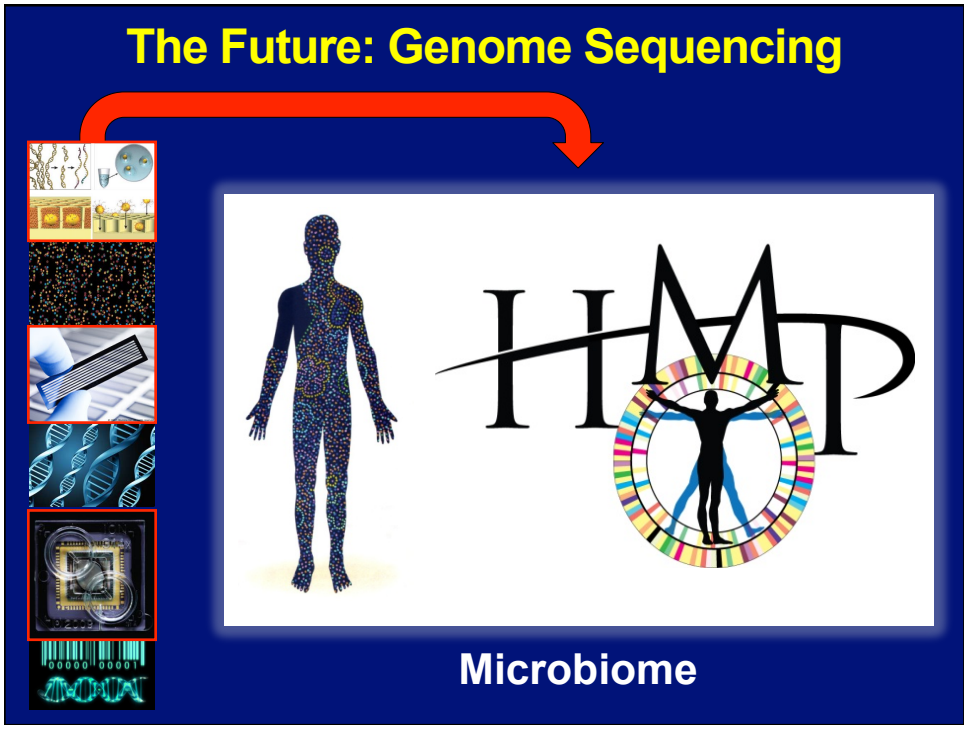
It Takes Several Mutations To Make a Cell Malignant

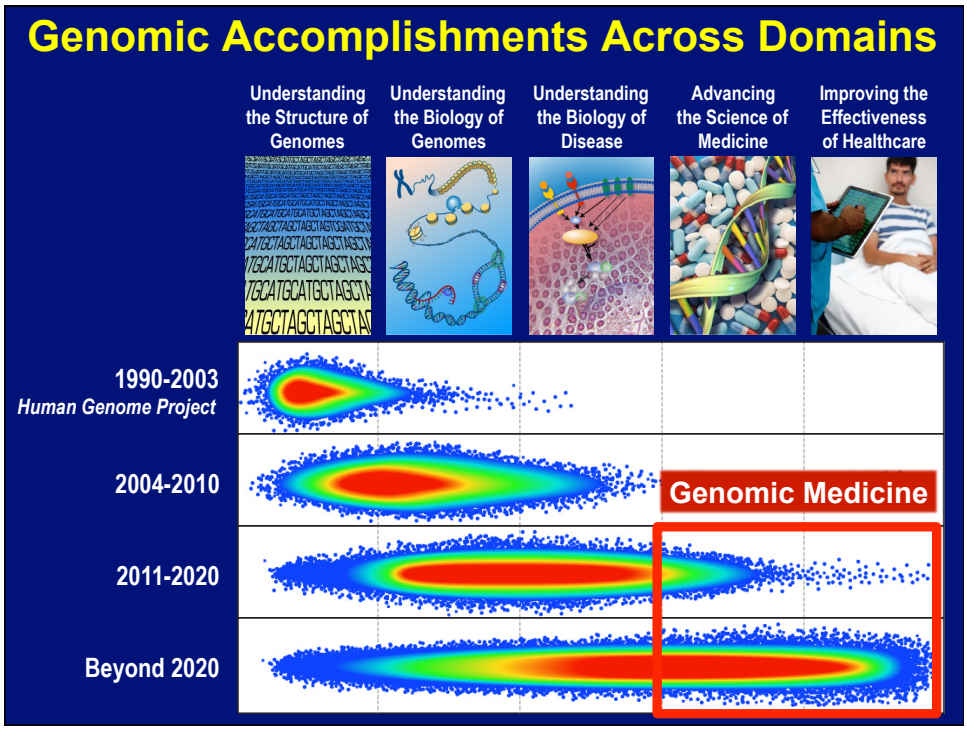
The Future: Genome Sequencing





Cancer Genomics


The Future: Genome Sequencing
HMP
Microbiome


Genomic Accomplishments Across Domains
Understanding the Structure of Genomes
Understanding the Biology of Genomes
Understanding the Biology of Disease
Advancing the Science of Medicine
Improving the Effectiveness of Healthcare
1990-2003
Human Genome Project
2004-2010
2011-2020
Beyond 2020
Genomic Medicine

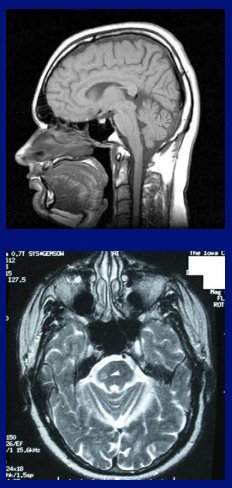
Genomic Medicine Comes Into Focus

'Hot Areas' in Genomic Medicine

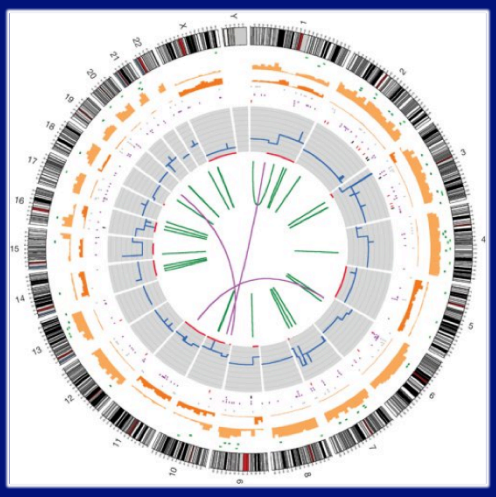
Cancer Genomics

Routine Clinical Diagnostic Tools

Radiographic Imaging

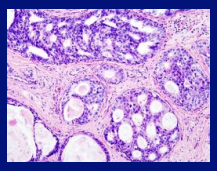


Cancer Genome Sequencing

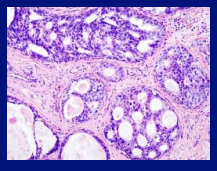


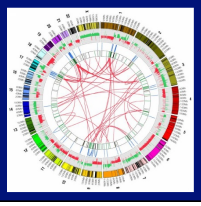
Genomic Medicine: Cancer Diagnostics

Now

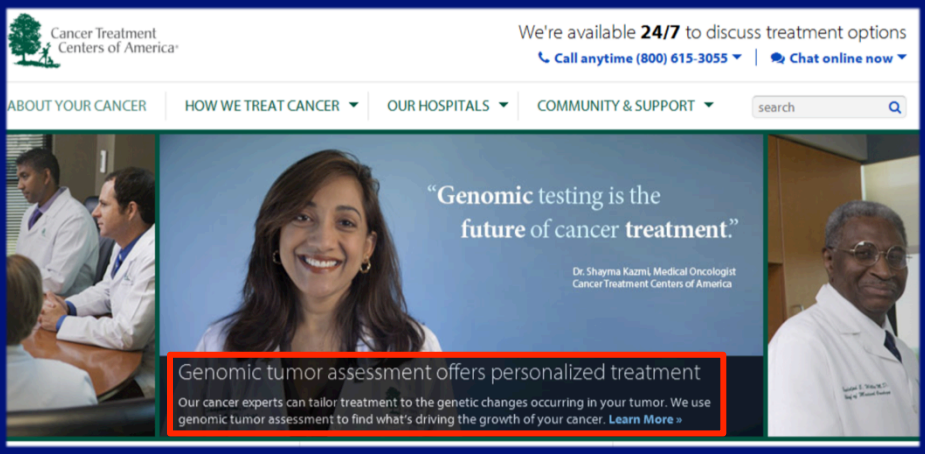


Future





Cancer Genomics: Here and Now



www.cancercenter.com

'Hot Areas' in Genomic Medicine

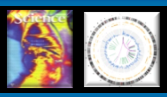
Cancer Genomics →

Pharmacogenomics →


Because Everyone Responds Differently.


All of these work.
Just not for everyone.
Perlegen may be able to help you sort out which medicine helps which patient.
Working with you, we can comprehensively analyze the DNA from thousands of patients taking your drug. Out of the millions of genetic variations between patients, we may be able to help you identify the ones that are associated with strong efficacy, poor efficacy, or side effects.
Perlegen's exceptional coverage of the genome and experienced team of analysts could help you get clinically relevant answers, not just data, in a matter of months.
We partner with the top pharmaceutical companies around the world. We also license late-stage drugs. If you have a drug that can benefit from our approach, please contact us.

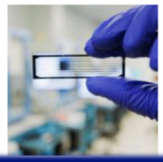
'Hot Areas' in Genomic Medicine

- Cancer Genomics 
- Pharmacogenomics 
- Genomic Medicine 'Test Drive' Programs 

Clinical Sequencing Exploratory Research (CSER)

cser
Clinical Sequencing Exploratory Research

Moving the genome into the clinic



In the past, standard medical practice for genetic testing involved looking at one gene at a time. With new advances in our understanding of the genomic basis of health and disease and in technology, it is now possible to test all of our genes at once using tests called whole exome or whole genome sequencing. Medical uses of genome sequencing are being applied and adapted on a case-by-case basis, but research to study the optimal uses and implementation of these tests is needed.

cser-consortium.org

Implementing Genomics into Clinical Practice Network (IGNITE)

Implementing Genomics in Practice (IGNITE)

Share Print

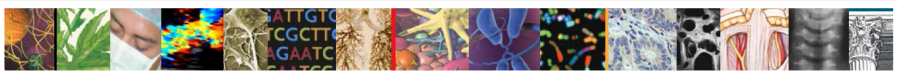
Overview

Findings from the genomics field have slowly started to find applications in clinical care. The field of "genomic medicine" could potentially improve patient health and treatment strategies or better predict the likelihood of disease.

The Implementing Genomics in Practice (IGNITE) consortium (RFA-HG-12-006, RFA-HG-12-007 and RFA-HG-13-004) was created to enhance the use of genomic medicine by supporting the development of methods for incorporating genomic information into clinical care and exploration of the methods for effective implementation, diffusion and sustainability in diverse clinical settings.

These demonstration projects will incorporate genomic information into the electronic medical record (EMR) and provide clinical decision support (CDS) for implementation of appropriate interventions or clinical advice. The sites will work together to develop new methods and projects and disseminate their findings to the public. Dissemination of these methods and developing best practices for implementation is a key goal so that the information generated from the program will contribute to the growing knowledge base of using genomic information in patient care.

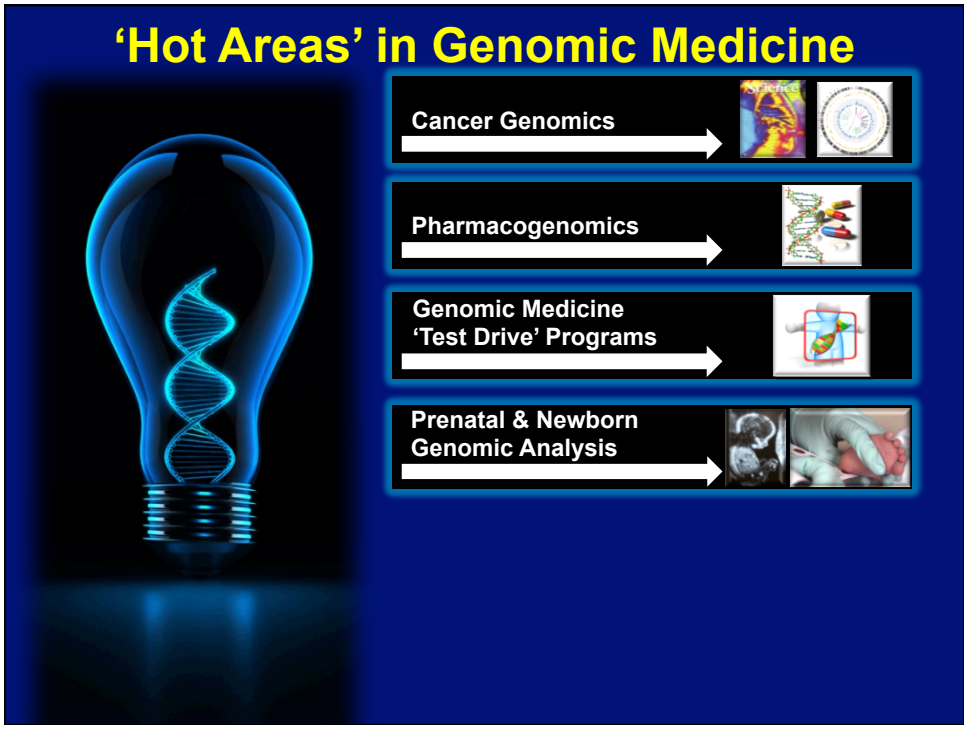
genome.gov/27554264



The NEW ENGLAND JOURNAL of MEDICINE

First FDA Authorization for Next-Generation Sequencer

Francis S. Collins, M.D., Ph.D., and Margaret A. Hamburg, M.D.


'Hot Areas' in Genomic Medicine
Cancer Genomics
Pharmacogenomics
Genomic Medicine
'Test Drive' Programs
Prenatal & Newborn
Genomic Analysis


Noninvasive Prenatal Genome Sequencing
CNNMoney
A Service of CNN, Fortune & Money
The next big thing in pregnancy: Sequencing your baby's genome
August 12, 2013: 7:35 AM ET
MIT Technology Review
10 BREAKTHROUGH TECHNOLOGIES 2013
Prenatal DNA Sequencing

Genomic Sequencing in Newborns (NSIGHT)

NIH program explores the use of genomic sequencing in newborn healthcare

Bethesda, Md., Wed., Sept. 4, 2013 - Can sequencing of newborns' genomes provide useful medical information beyond what current newborn screening already provides? Pilot projects to examine this important question are being funded by the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) and the National Human Genome Research Institute (NHGRI), both parts of the National Institutes of Health. Awards of $5 million to four grantees have been made in fiscal year 2013 under the Genomic Sequencing and Newborn Screening Disorders research program. The program will be funded at $25 million over five years, as funds are made available.

"Genomic sequencing has potential to diagnose a vast array of disorders and conditions at the very start of life," said Alan E. Guttmacher, M.D., director of NICHD. "But the ability to decipher an individual's genetic code rapidly also brings with it a host of clinical and ethical issues, which is why it is important that this program explores the trio of technical, clinical, and ethical aspects of genomics research in the newborn period."

The awards will fund studies on the potential for genome and exome sequencing to expand and improve newborn health care. Genomic sequencing examines the complete DNA blueprint of the cells, and exome sequencing is a strategy to selectively sequence exons, the short stretches of DNA within our genomes that code for proteins.

genome.gov

Sequenced from the start

Four US studies are set to explore how genomic data can best help healthy and ill newborns. They must also settle some questions of ethics.

Genetic sequencing has established itself as a powerful tool for diagnosis, but it is not yet clear how useful it will be for disease prevention or health management. A US$25-million project announced last week aims to explore that issue in perhaps the most high-stakes patient group: newborn babies.

In the Genomic Sequencing and Newborn Screening Disorders (GSNSD) programme, four teams will sequence the exomes — the protein-coding portions of the genome — or the whole genomes of more than 1,500 babies, including not only infants who are ill, whether or not the disease has been diagnosed, but also healthy babies. The programme is funded by the US National Human Genome Research Institute and the Eunice Shriver Kennedy National Institute of Child Health and Human Development (NICHD). The studies will examine how useful sequencing information is for families and doctors, and whether it is superior to data gathered through conventional newborn-screening methods, which check for about 60 genetic disorders.

"The day when all children will be sequenced at birth — if not before — draws ever nearer."

plans to give the raw genetic data to the children's families, even though that could allow the children to benefit from it throughout their lives.

Finally, should the data be shared with other researchers? This would be the best way for scientists to help tackle the tough question of how genes contribute to disease. But it is increasingly difficult to guarantee the privacy of genetic data (see *Nature* **493**, 451; 2013), and this is an important issue for babies, whose information will be known for their entire lives even though they themselves have not consented to the disclosure. One of the GSNSD projects will share data with the NICHD's Newborn Screening Translational Research Network, and another with the National Center for Biotechnology Information's Database of Genotypes and Phenotypes. The other two are still deciding.

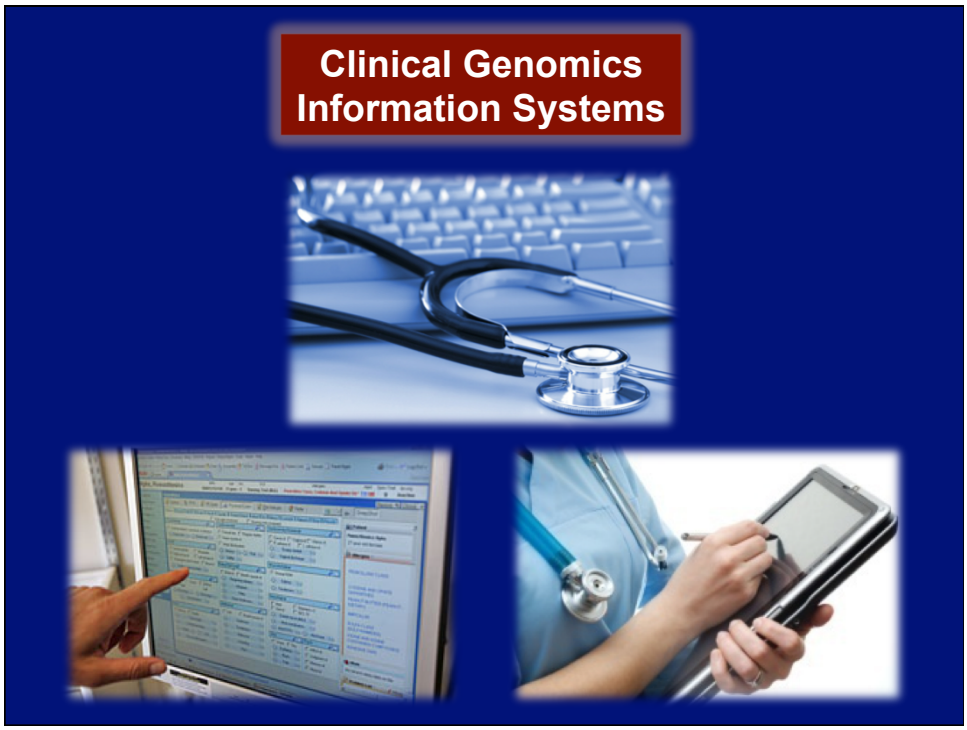
As researchers explore these questions, sequencing costs continue

***Nature* (2013)**

'Hot Areas' in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Genomic Medicine 'Test Drive' Programs
- Prenatal & Newborn Genomic Analysis
- Clinical Genomics Information Systems


GTACT
CAAGT
CTA
ATCC
TAC
CCGA
TAGG
ATCC
TGG
ATCC


Clinical Genomics
Information Systems

Clinical Genome Resource (ClinGen)

New NIH-funded resource focuses on use of genomic variants in medical care

Bethesda, Md., Wed., Sept. 25, 2013 - Three grants totaling more than $25 million over four years will help three research groups to develop authoritative information on the millions of genomic variants relevant to human disease and the hundreds that are expected to be useful for clinical practice. The awards are from the National Institutes of Health.

More and more medical and research centers are sequencing the DNA of whole genomes (the body's entire genetic blueprint) or exomes (the genome's protein-coding region) of patients. Each time, millions of DNA differences in genes and the regions between the genes are detected. But doctors struggle to know which of those differences, called variants, are relevant to disease and for a patient's medical care. As a result, information on few genomic variants is used in clinical practice.

The grants will support a consortium of research groups to develop the Clinical Genome Resource (ClinGen). The investigators will design and implement a framework for evaluating which variants play a role in disease and those that are relevant to patient care, and will work closely with the National Center for Biotechnology Information (NCBI) of the National Library of Medicine (NLM), which will distribute this information through its ClinVar database. The grants are funded by the National Human Genome Research Institute (NHGRI) and the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), which, along with NCBI and NLM, are part of NIH. ClinGen was developed from NHGRI's Clinically Relevant Variants Resource program.

genome.gov

'Hot Areas' in Genomic Medicine

- **Cancer Genomics**
- **Pharmacogenomics**
- **Genomic Medicine 'Test Drive' Programs**
- **Prenatal & Newborn Genomic Analysis**
- **Clinical Genomics Information Systems**
- **Ultra-Rare Genetic Disease Diagnostics**

Ultra-Rare Genetic Disease Diagnostics

Exome Sequencing: Dual Role as a Discovery and Diagnostic Tool

Chee-S Clinical application of exome sequencing in undiagnosed genetic conditions

Anna C
Kevin V

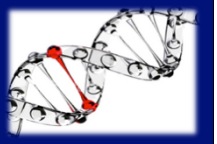
Next-Generation Sequencing for Clinical Diagnostics

Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders

Ya
Matt
Alici
Matth
Magalie

Genomics in Clinical Practice: Lessons from the Front Lines

Howard J. Jacob,[1,5,6*] Kelly Abrams,[12] David P. Bick,[1,5,10] Kent Brodie,[1] David P. Dimmock,[1,5,10] Michael Farrell,[3] Jennifer Geurts,[1,7] Jeremy Harris,[1,5] Daniel Helbling,[1,5] Barbara J. Joers,[12] Robert Kliegman,[5] George Kowalski,[1] Jozef Lazar,[1,2] David A. Margolis,[5] Paula North,[4,9,11] Jill Northup,[1] Altheia Roquemore-Goins,[11] Gunter Scharer,[1,5,10] Mary Shimoyama,[1,7] Kimberly Strong,[1,8] Bradley Taylor,[1] Shirng-Wern Tsaih,[1] Michael R. Tschannen,[1] Regan L. Veith,[1,10] Jaime Wendt-Andrae,[1] Brandon Wilk,[1,5] Elizabeth A. Worthey[1,5,9]



***Sci Transl Med* (2013)**

Undiagnosed Diseases Network (UDN)

- **Build upon the successful experience with the NIH Undiagnosed Diseases Program to improve the diagnosis and care of patients with undiagnosed diseases**
- **Facilitate research into the etiology of undiagnosed diseases**
- **Create a highly collaborative research community to identify best practices for the diagnosis and management of undiagnosed diseases**

The Relevance of Genomics

Biomedical Researchers

Healthcare Professionals

Patients (and Friends & Relatives of Patients)

Genomics and Society

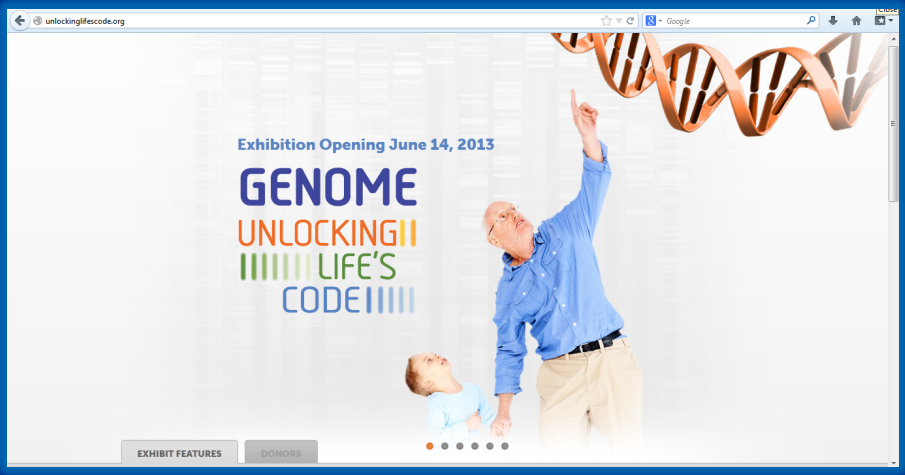
NHGRI-Smithsonian Genome Exhibition

GENOME UNLOCKING LIFE'S CODE

NIH National Human Genome Research Institute

Smithsonian *National Museum of Natural History*

Smithsonian Exhibition: Website



unlockinglifescode.org


The Genomics Landscape
A monthly update from
the NHGRI Director
February 4, 2014
For this second month of 2014, I hope you enjoy reading about the new trans-NIH Big Data to Knowledge (BD2K) Initiative, the centerpiece of NIH's efforts to address the 'Big Data' problem facing biomedical research. And while parts of the country continue to suffer the chilling effects of a polar vortex, I am relieved to report that Washington, D.C. shows some signs of a thaw with regard to the budget battles. The politicians in our nation's
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genome.gov/Director


National Human Genome
Research Institute
Advancing human health
through genomics research