




Human The [^]Genomics Landscape Circa 2016


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Current Topics in Genome Analysis 2016

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*No Relevant Financial Relationships with
Commercial Interests*



NATIONAL HUMAN GENOME RESEARCH INSTITUTE
Division of Intramural Research

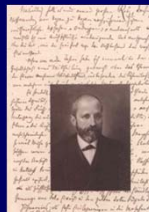
I. Historical Context for Genomics
II. Major Achievements since the Human Genome Project
III. The Human Genomics Landscape: 2016 and Beyond
>> Goal: Place Other Speakers into a Broader Context <<

Foundational Milestones in Genetics & Genomics



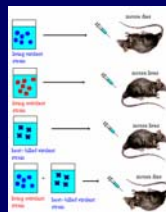
Mendel

1865



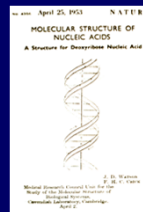
Miescher

1871



Avery

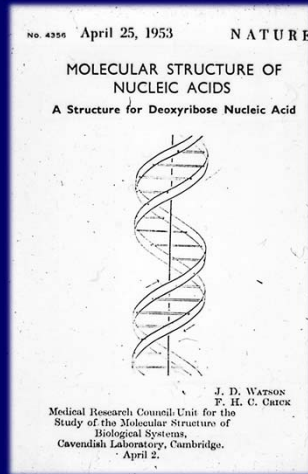
1944



**Watson
& Crick**

1953

April, 1953



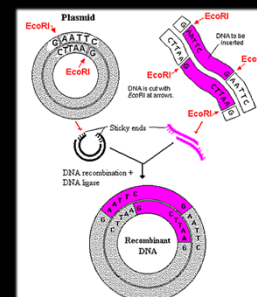
Discovery of Double-Helical Structure of DNA

1960's

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

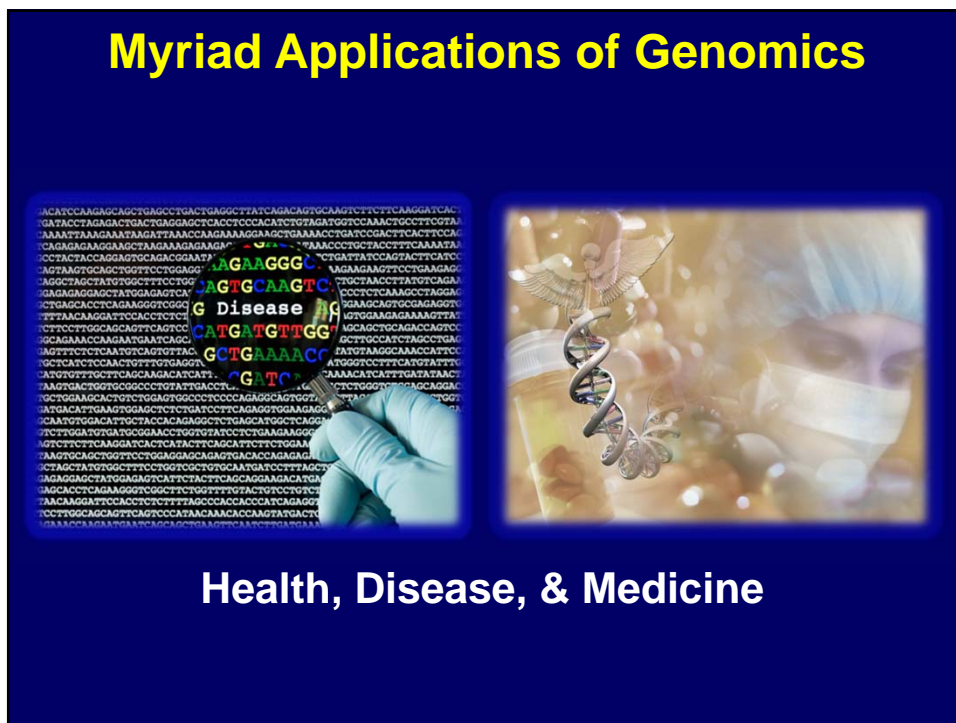
Human Genome Project: 1990-2003



Twenty-five years of big biology

The Human Genome Project, which launched a quarter of a century ago this week, still holds lessons for the consortium-based science it ushered in, say
Eric D. Green, James D. Watson and Francis S. Collins.

Nature (2015)



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



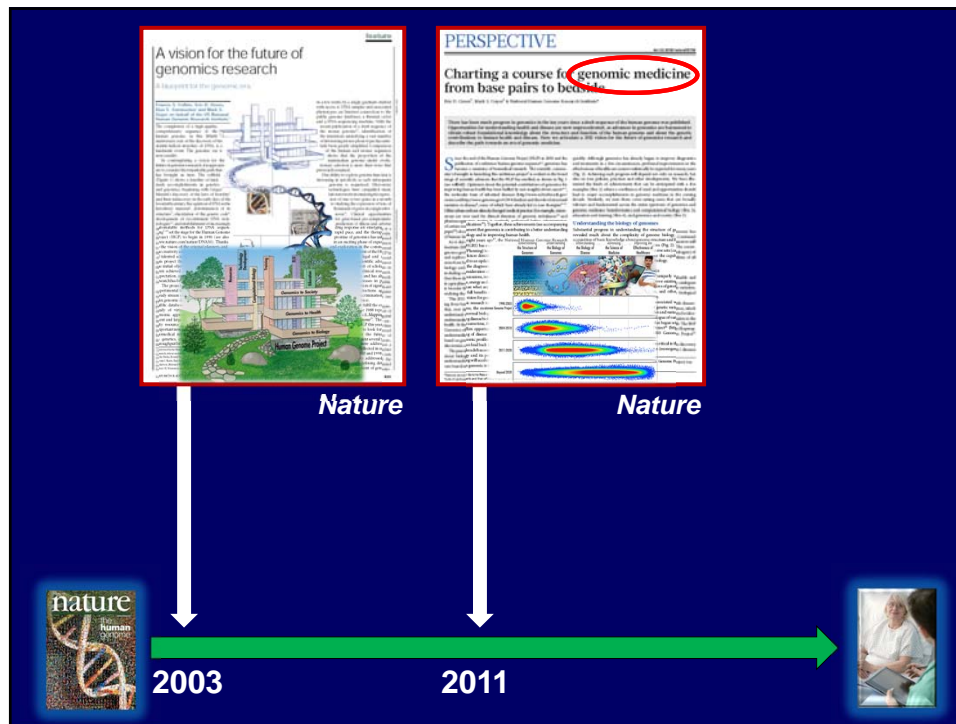
The Path to Genomic Medicine



Human Genome Project



Realization of Genomic Medicine



February, 2011

nature
 THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE

THE FUTURE IS BRIGHT
 Reflections on the first ten years of the human genomics age

THE END OF THE BEGINNING
 The impact of the human genome

MORE BASES PER DOLLAR
 How the cost of sequencing is dropping

FROM LAB TO CLINIC
 How genomics is changing medicine

PERSPECTIVE

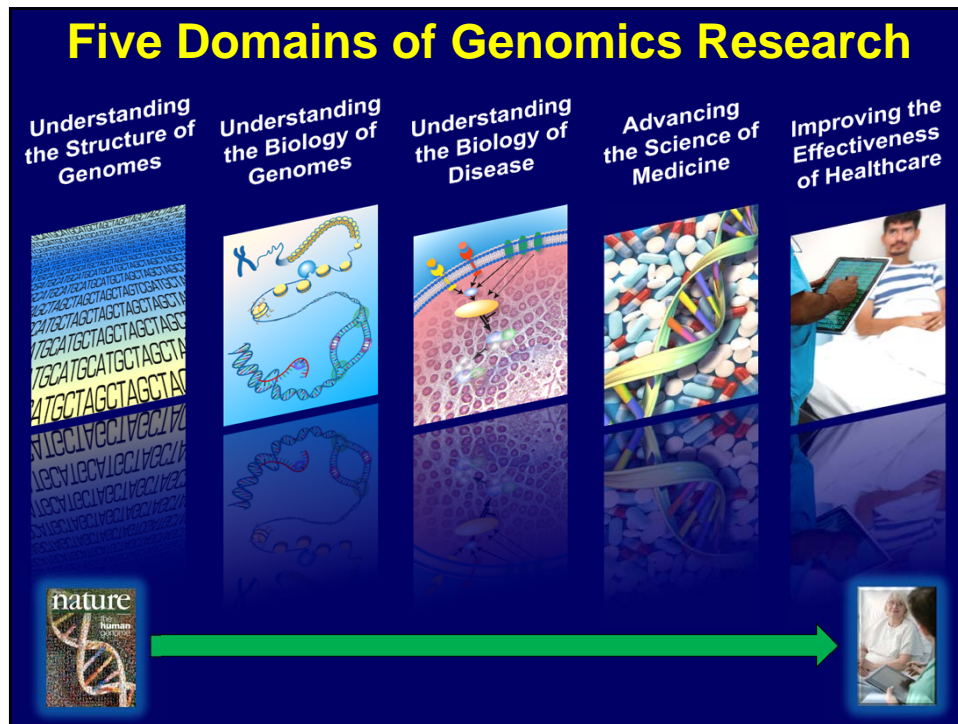
Charting a course for genomic medicine from base pairs to bedside

Eric Green, Mark A. Grady, A 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 expanding, as advances in genomics are harnessed to obtain relevant 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 of the future of genomics research and about the path towards an era of genomic medicine.

Although genomics has already begun to improve diagnosis and treatment in a few circumstances, potential improvements in the effectiveness of health care cannot be expected for many years. The challenge will be to ensure that the benefits of genomics are realized in a way that is equitable, ethical, and that respects individual privacy and other principles. We have identified a number of key areas that will be critical to the success of genomics in the future. These include: (1) the need to build a strong foundation of genomic medicine in the coming decade; (2) the need to ensure that the benefits of genomics are realized in a way that is equitable, ethical, and that respects individual privacy and other principles; (3) the need to build a strong foundation of genomic medicine in the coming decade; (4) the need to ensure that the benefits of genomics are realized in a way that is equitable, ethical, and that respects individual privacy and other principles.

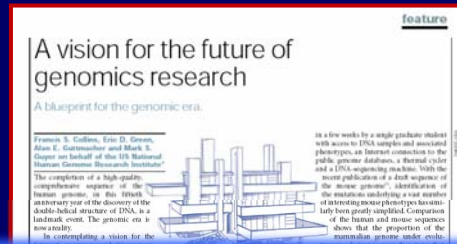
New NHGRI Vision for Genomics Published



A Quarter Century of Genomics

**Human Genome Sequenced for First Time
by the Human Genome Project**

**Cost of Sequencing a Human Genome
Reduced Nearly ~1 Million-Fold**



“...‘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.”

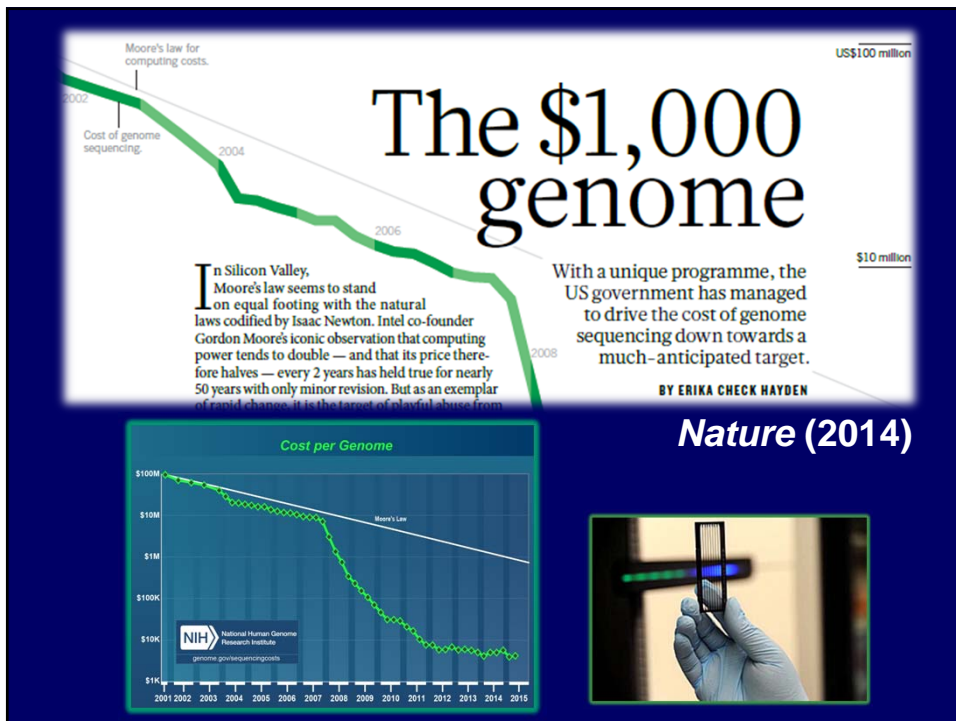
Human Genome Sequence

~\$1,000,000,000



~\$1,000

“The \$1000 Genome”



Sequencing a Human Genome

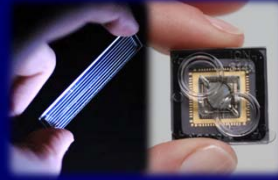
Human Genome Project
(1st Sequence)



~\$1B

~6-8 years

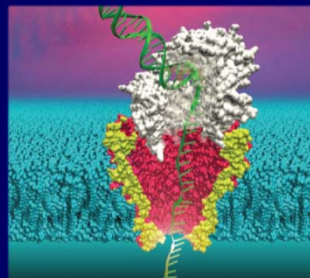
Today



~\$2-3K

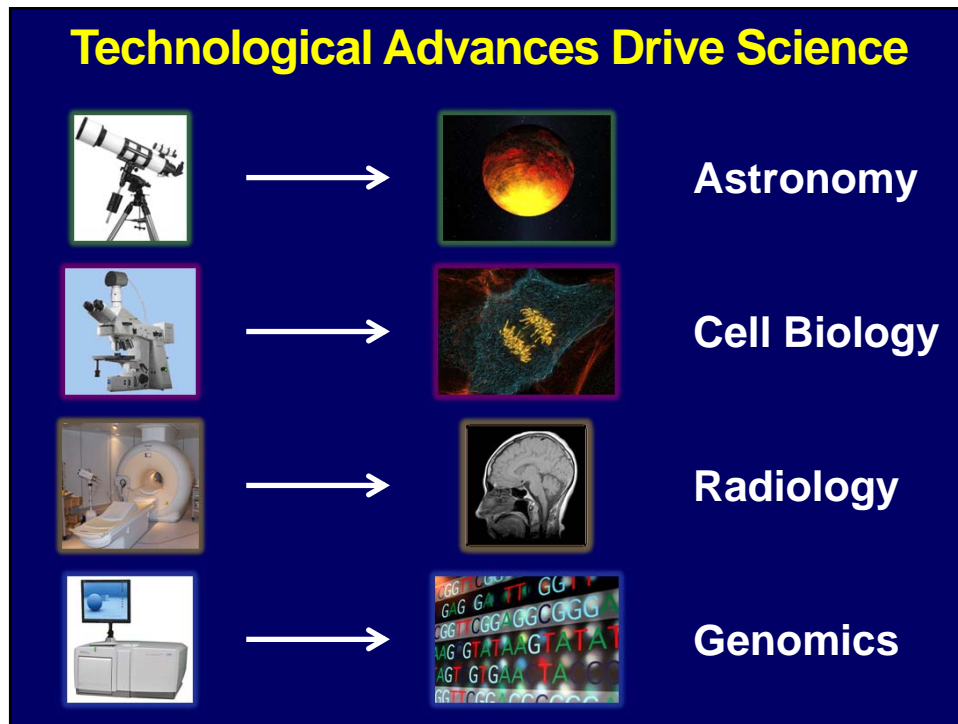
~1-3 days

And Yet Newer Technologies...



Search for Pore-fection



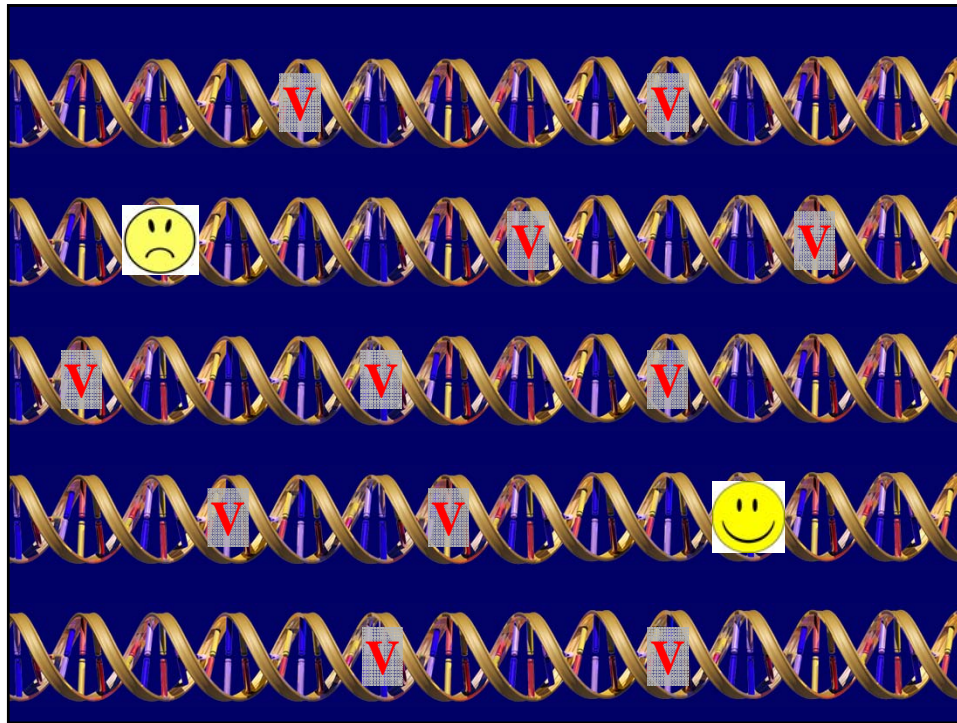


A Quarter Century of Genomics

Human Genome Sequenced for First Time
by the Human Genome Project

Cost of Sequencing a Human Genome
Reduced Nearly ~1 Million-Fold

Tens of Thousands of Human
Genomes Sequenced



International HapMap Project

27 October 2005 | www.nature.com/nature | \$10

THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE

nature

INSIDE
Why do we sleep?
CULTURE
HapMap

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 the 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 25–30% of common SNP variation in the populations surveyed. The map is extended to capture rare genetic 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 0.76 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 10–20% 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

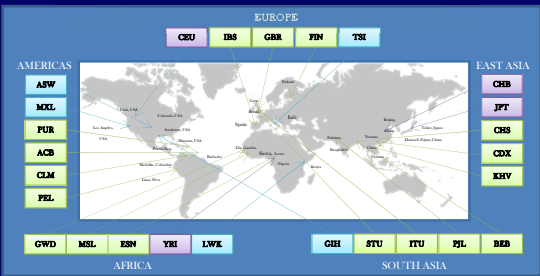
Integrating common and rare genetic variation in diverse human populations

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 1,068 reference individuals from 11 global populations, and sequenced two 500-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 $\le 1\%$, and demonstrated the feasibility of imputing newly discovered CNPs and SNPs. This expanded public resource of genome variants in global populations requires deeper interpretation of genomic variation and its role in human disease, and serves as a step toward a high-resolution map of the landscape of human genetic variation.

2010

1000 Genomes
A Deep Catalog of Human Genetic Variation






nature
THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE

A THOUSAND GENOMES
Pilot studies prepare the way for population-scale gene sequencing

2535 Humans, 26 Populations


AMERICAS: ASW, MXL, PUR, ACB, CLM, PEL
EUROPE: CEU, IBS, GRB, FIN, TSI
AFRICA: GWD, MSL, ESN, YRI, LWK
SOUTH ASIA: GIH, STU, ITU, PZL, BEB
EAST ASIA: CHB, JPT, CHS, CDX, KHV

Your Genome: By the Numbers



- ~6B nucleotides
- ~3-5M single-nucleotide variants
 - ~150K not in databases
 - ~60 not in either parent

A Quarter Century of Genomics

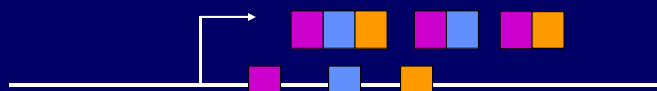


- Human Genome Sequenced for First Time
by the Human Genome Project**
- Cost of Sequencing a Human Genome
Reduced Nearly ~1 Million-Fold**
- Tens of Thousands of Human
Genomes Sequenced**
- Profound Advances in Understanding
How the Human Genome Functions**

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

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Coding Sequences (i.e., Genes)



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

The Genetic Code

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

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Foundational Milestones in Genetics & Genomics



Darwin

1859



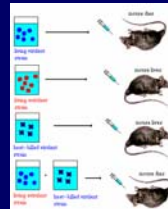
Mendel

1865



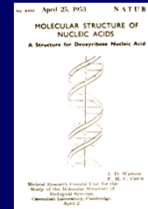
Miescher

1871



Avery

1944

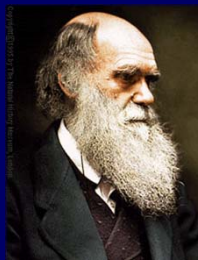


Watson
& Crick

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."

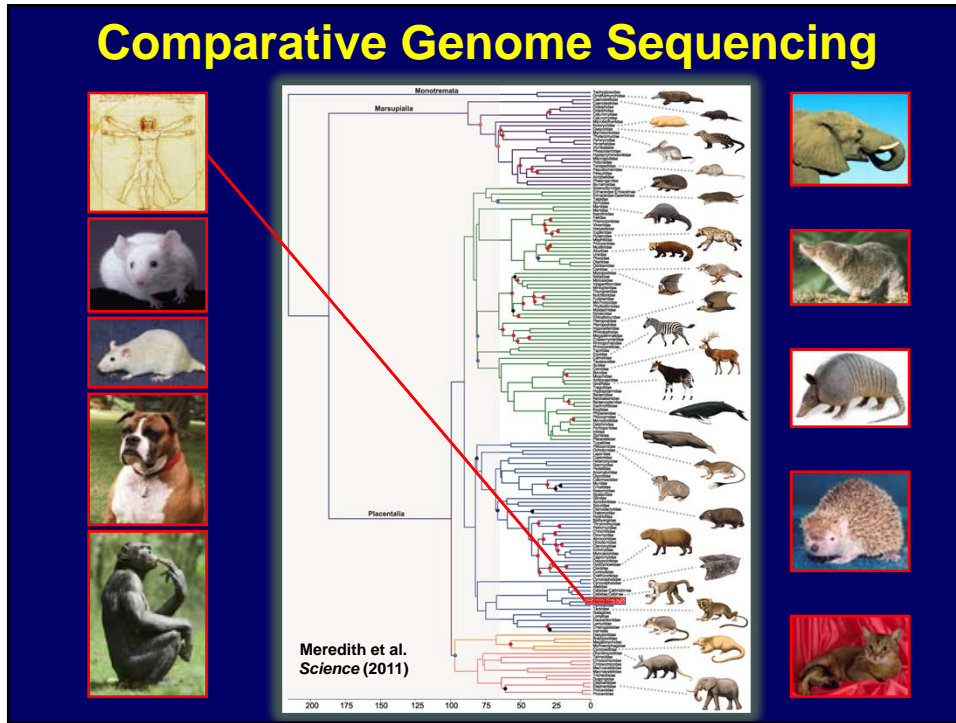
(Attributed to Darwin)



Charles Darwin (1809-1882)

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

— Eric Lander

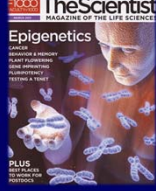



~3,000 bp (0.0001%) of Human Genome Sequence

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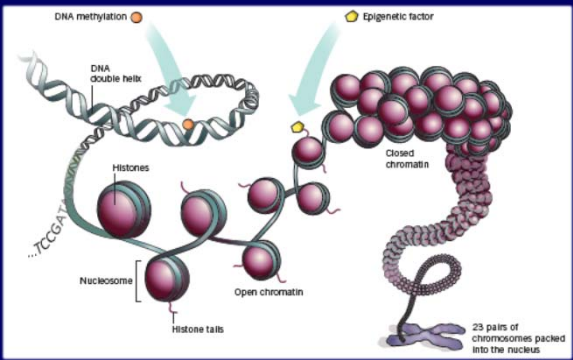

The Epigenomics Landscape



TECHNOLOGY FEATURE

READING THE SECOND GENOMIC CODE

Nature (2012)

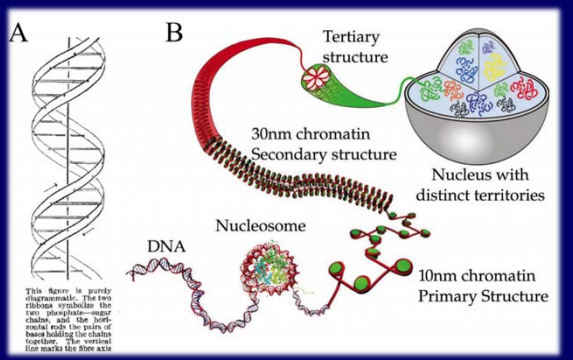


TECHNOLOGY FEATURE

GENOMES IN THREE DIMENSIONS

A DNA sequence isn't enough; to understand the workings of the genome, we must study chromosome structure.

Nature (2011)



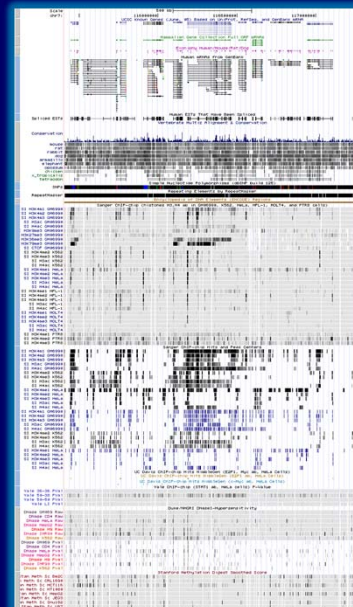
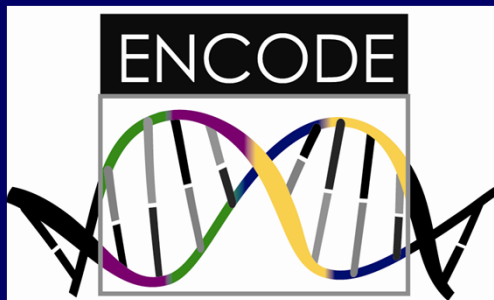
This figure is partly schematic. The two ribbons illustrate the two phosphate-sugar chains and the horizontal and vertical lines mark the bases holding the chains together. The vertical line marks the three axes.

Elucidating Genome Function



'Team Science'

ENCODE: Giving 'GPS' Views of Genomes



Elucidating Genome Function



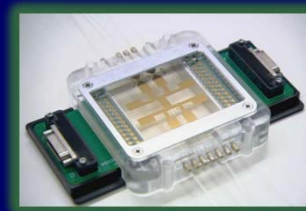
'Team Science'



Model Organisms

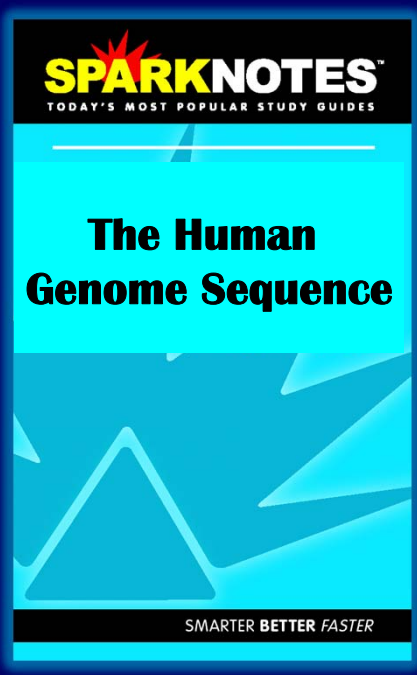


Computational Modeling



Technology Development

```
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CCATAGGAATAGATACCGAAGTTATATCCAACTGACATTA
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A Quarter Century of Genomics

Human Genome Sequenced for First Time
by the Human Genome Project

Cost of Sequencing a Human Genome
Reduced Nearly ~1 Million-Fold

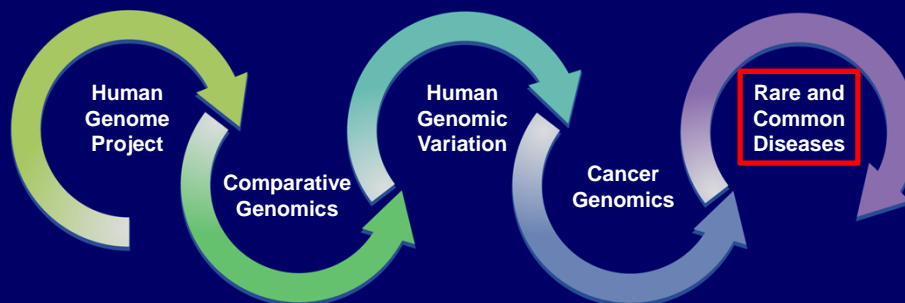
Tens of Thousands of Human
Genomes Sequenced

Profound Advances in Understanding
How the Human Genome Functions

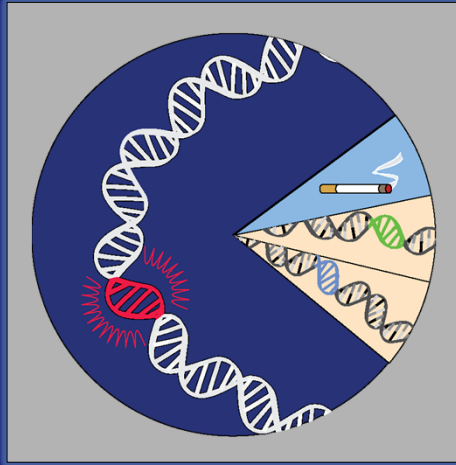
Significant Advances in Unraveling the
Genomic Bases of Human Disease

NHGRI Genome Sequencing Program

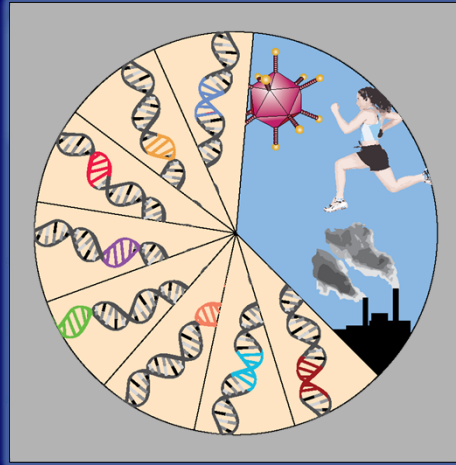
At the cutting edge of genome analysis



Genomic Architecture of Genetic Diseases



Rare, Simple, Monogenic,
Mendelian...



Common, Complex, Multigenic,
Non-Mendelian...

Manolio et al., J Clin Invest (2008)

The Largest Current Bottleneck in Genomics...

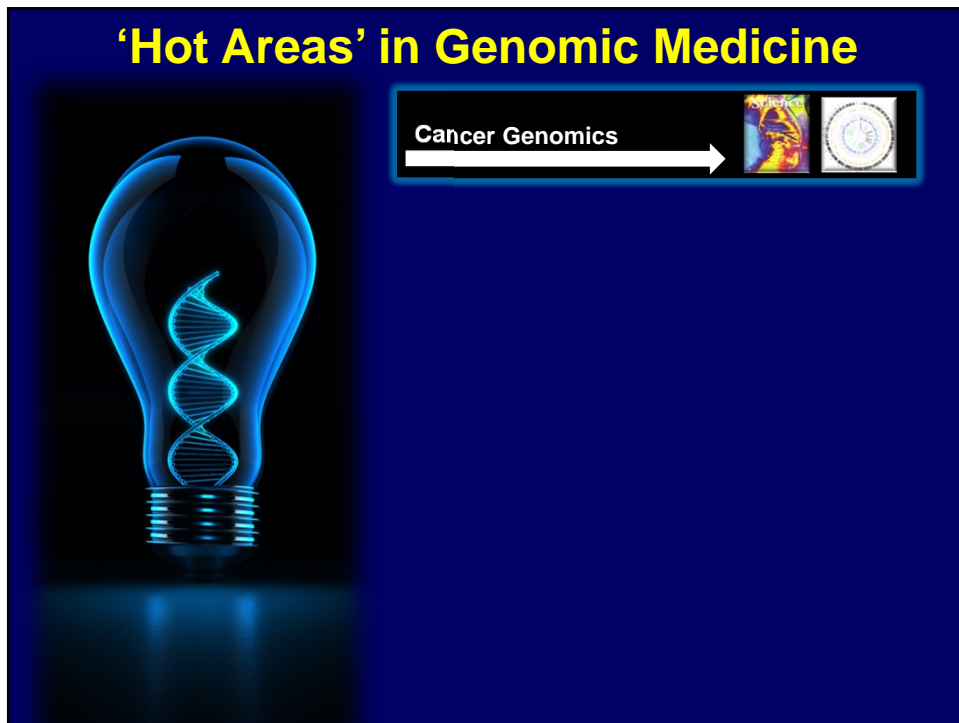
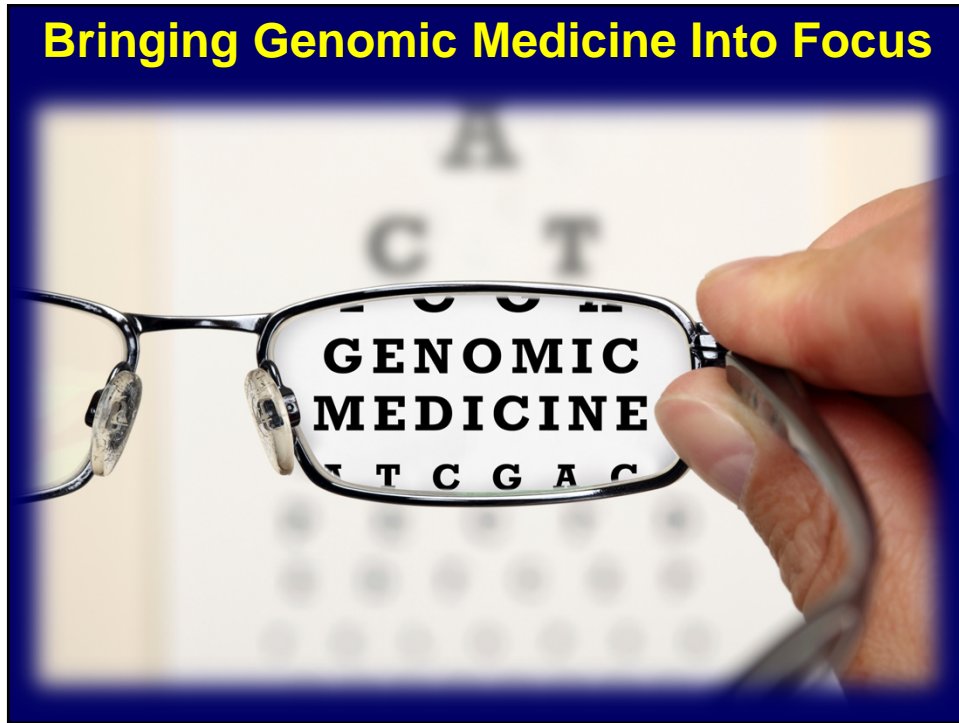


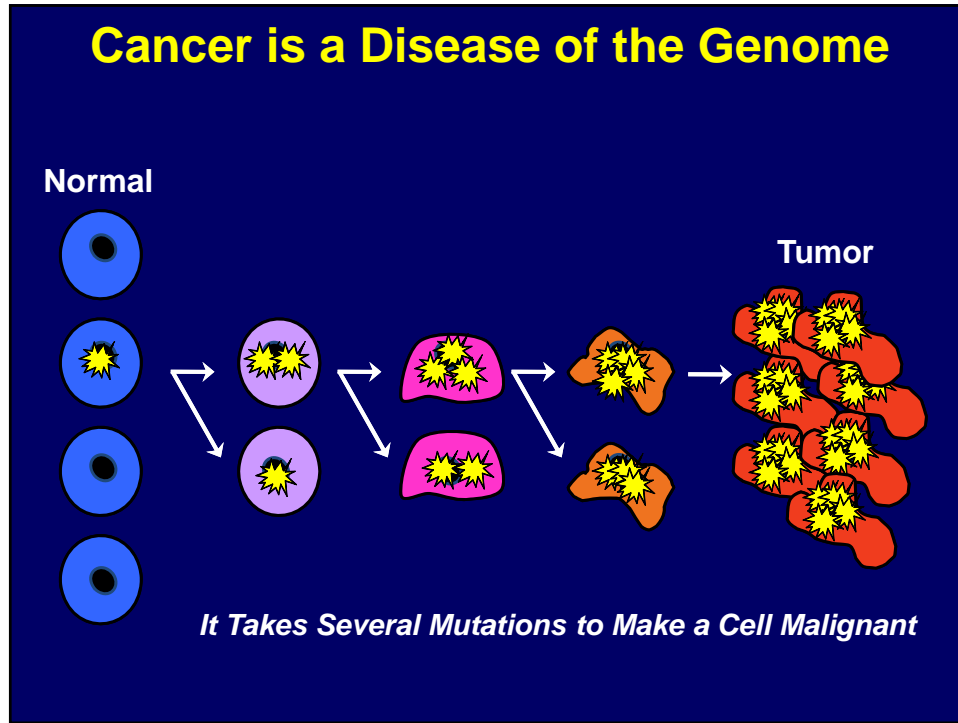
The Data Analysis Bottleneck

The image is a composite illustrating the 'Data Analysis Bottleneck'. It features a woman working at a computer workstation in a server room, a 3D model of a DNA double helix, and a cartoon illustration of a man looking at a DNA helix on a screen while another man sits on a toilet. The background is a blue gradient with white DNA sequence text.


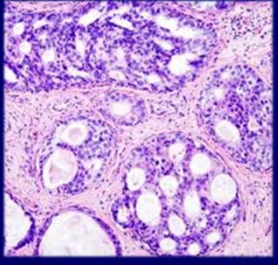

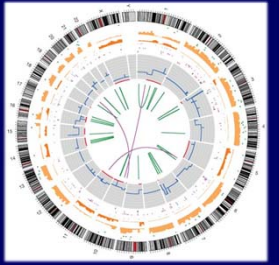
A Quarter Century of Genomics

- Human Genome Sequenced for First Time by the Human Genome Project
- Cost of Sequencing a Human Genome Reduced Nearly ~1 Million-Fold
- Tens of Thousands of Human Genomes Sequenced
- Profound Advances in Understanding How the Human Genome Functions
- Significant Advances in Unraveling the Genomic Bases of Human Disease
- Vivid Examples of Genomic Medicine in Action Now Emerging





Routine Cancer Diagnostic Tools

<p>Cancer Histopathology</p>  	<p>Cancer Genome Sequencing</p>  
--	---

Genomics and Cancer: Here and Now

Cancer Treatment Centers of America

We're available 24/7 to discuss treatment options. Call anytime (800) 931-9299 Chat online now

ABOUT YOUR CANCER | HOW WE TREAT CANCER | OUR HOSPITALS | COMMUNITY & SUPPORT

HOW CAN GENOMIC TESTING HELP PATIENTS NOW?

Every cancer is different. Genomic testing helps our doctors understand a patient's cancer at the molecular level and may reveal more personalized treatment options.

LEARN MORE »

"Genomic testing is the future of cancer treatment."

Dr. Shayma Kazmi, Medical Oncologist
Cancer Treatment Centers of America

HUNTSMAN
CANCER INSTITUTE
UNIVERSITY OF UTAH

CHANGING THE DNA OF CANCER CARE

huntsmancancer.org

'Hot Areas' in Genomic Medicine

Cancer Genomics →

Pharmacogenomics →



All of these work.

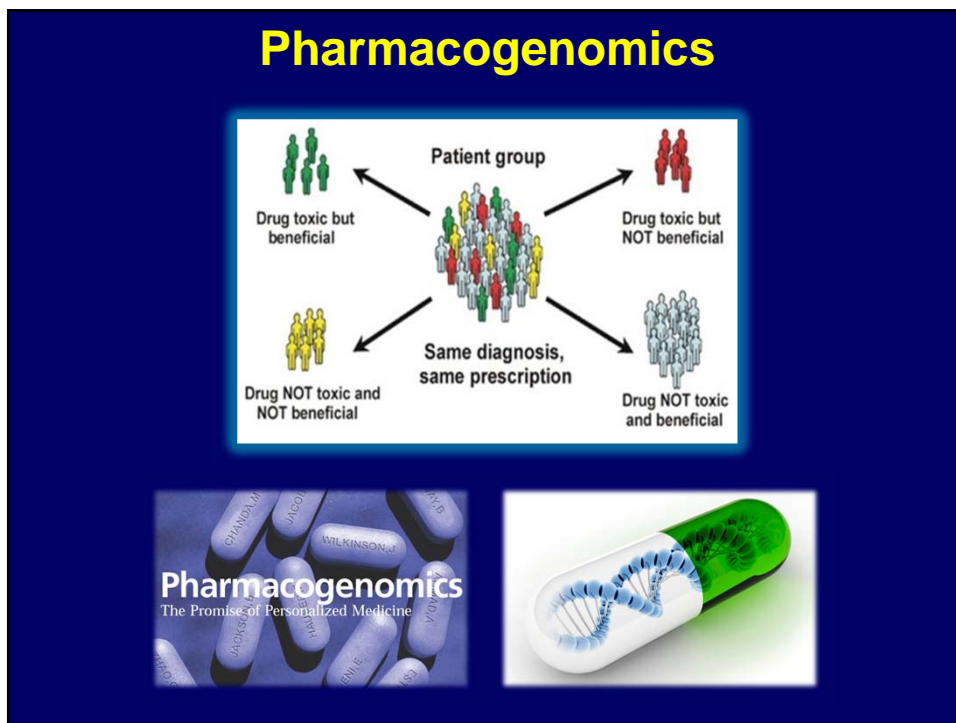
Just not for everyone.

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.

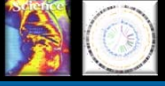


Perlegen's exceptional coverage of the genome and experienced team of analysts could help you get clinically relevant answers, not just data, in a matter of months.

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** → 
- Rare Genetic Disease Diagnostics** → 

TECHNOLOGY FEATURE

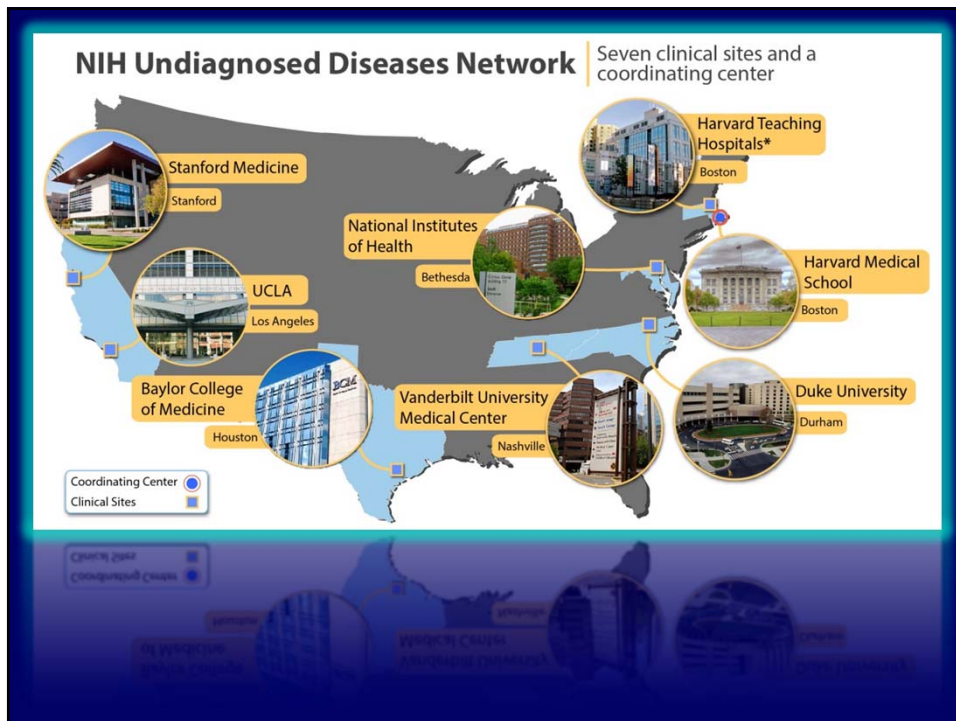
WHEN DISEASE STRIKES FROM NOWHERE

When healthy parents have a child with a genetic disorder, the cause is sometimes a new mutation. Tools are emerging to meet the challenge of finding such changes.




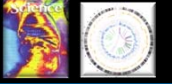



“ ...disorders not readily explained by standard tests can sometimes be diagnosed through genome sequencing and analysis.”

Nature (2014)




'Hot Areas' in Genomic Medicine

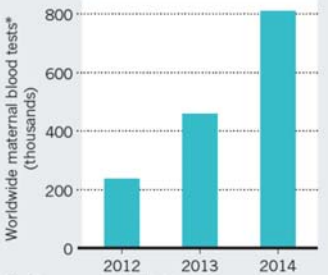


- Cancer Genomics** → 
- Pharmacogenomics** → 
- Rare Genetic Disease Diagnostics** → 
- Genomics of Pregnancy** → 

Noninvasive Prenatal Genome Sequencing



Since late 2011, clinicians have been able to screen mothers' blood for fetal chromosome problems using circulating DNA.



Year	Worldwide maternal blood tests* (thousands)
2012	~250
2013	~450
2014	~800

*Numbers as reported by Illumina, Sequenom, Ariosa Diagnostics, Berry Genomics and BGI in GenomeWeb articles.

DW Bianchi, *Nature* (2015)

Newborn Genome Sequencing

HEALTH RESEARCH

In 2025, Everyone Will Get DNA Mapped At Birth

Alice Park @alleeparkny June 30, 2014

Scientists have scoured trends in research grants, patents and more to come up with these 10 innovations that will be reality in 10 years (or so they think)

Everybody likes to blue-sky it when it comes to technology. Driverless cars! Fat-burning pills! Telepathic butlers! But the folks at Thomson Reuters Intellectual Property & Science do it for a living—and they do it with data.

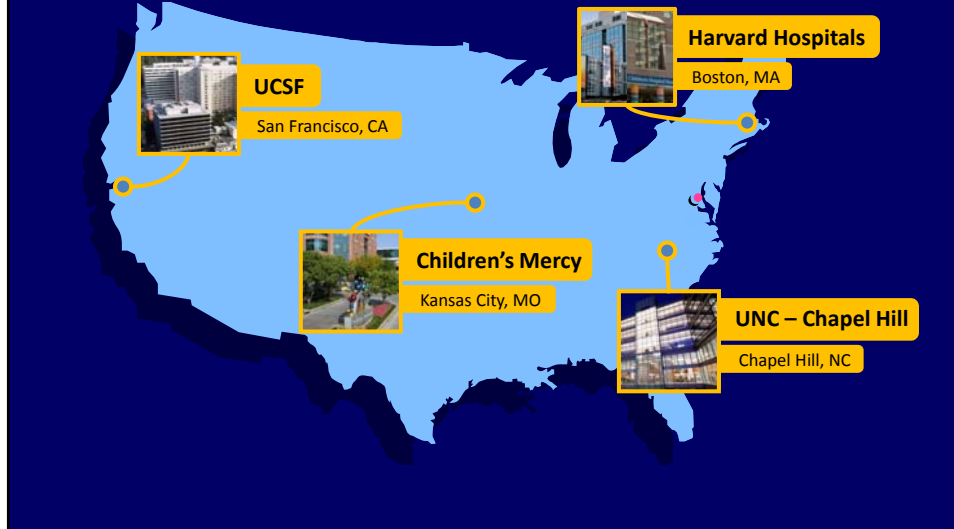


What will the future hold?
© Thomson Reuters Intellectual Property & Science

Time (2014)



Newborn Sequencing In Genomic medicine and public Health (NSIGHT)



Genome Sequencing of Acutely Sick Newborns




The genomes of ill newborns can be sequenced in less than 24 hours to give clinicians a rapid diagnosis.

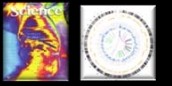




Fast sequencing saves newborns

Rapid analysis of infant genomes is aiding diagnosis and treatment of inexplicably ill babies.

Nature (2014)

'Hot Areas' in Genomic Medicine

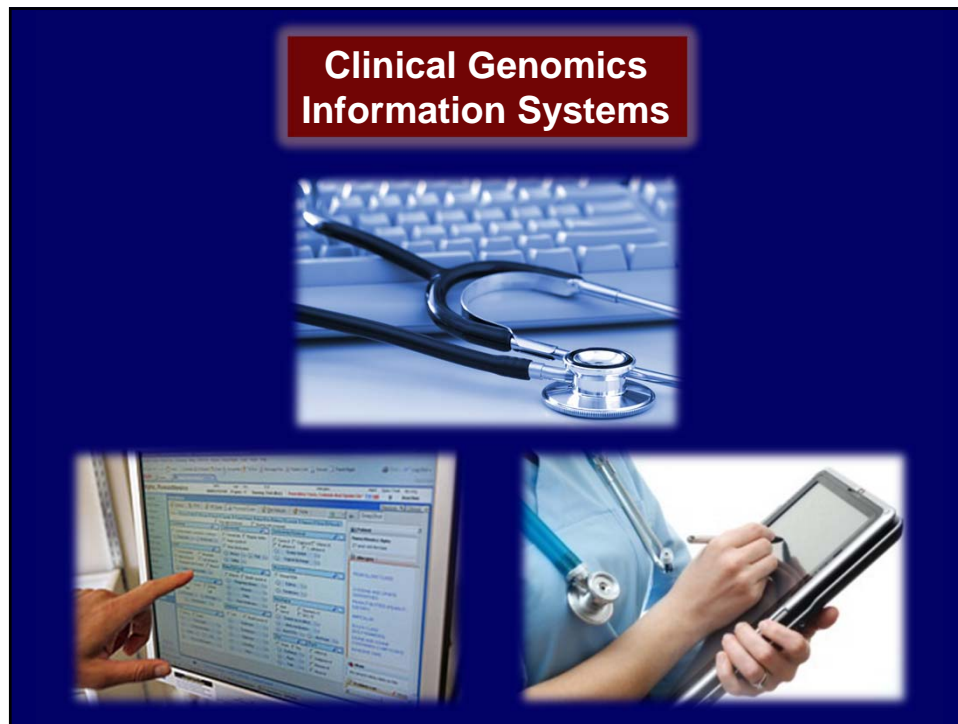


- Cancer Genomics** → 
- Pharmacogenomics** → 
- Rare Genetic Disease Diagnostics** → 
- Genomics of Pregnancy** → 
- Clinical Genomics Information Systems** → 

Generating a Human Genome Sequence is (Almost) Trivial

TGAACACCAATTGGCACGATGCTCCGTCGAGGAAACTTGAACACCAATTGGGTCGAGGAAACTTGAACACCA
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Clinical Genome Resource (ClinGen)

The screenshot shows the ClinGen website interface. It includes a navigation menu with links for "About", "Data Sharing", "Knowledge Curation", "Machine Learning", "GenomeConnect", and "Events & News". A search bar and "Contact" link are also visible. The main content area features a blue banner with the text "ClinGen: Sharing Data. Building Knowledge. Improving Care." and a paragraph of introductory text. Below the banner, the URL "clinicalgenome.org" is displayed in large white letters.

ClinGen — The Clinical Genome Resource

Heidi L. Rehm, Ph.D., Jonathan S. Berg, M.D., Ph.D., Lisa D. Brooks, Ph.D., Carlos D. Bustamante, Ph.D., James P. Evans, M.D., Ph.D., Melissa J. Landrum, Ph.D., David H. Ledbetter, Ph.D., Donna R. Maglott, Ph.D., Christa Lese Martin, Ph.D., Robert L. Nussbaum, M.D., Sharon E. Plon, M.D., Ph.D., Erin M. Ramos, Ph.D., Stephen T. Sherry, Ph.D., and Michael S. Watson, Ph.D., for ClinGen

NEJM (2015)

The Genomic Medicine Ecosystem

Healthcare Delivery



The Genomic Medicine Ecosystem

Education & Genomic Literacy



The Genomic Medicine Ecosystem

Regulatory Oversight



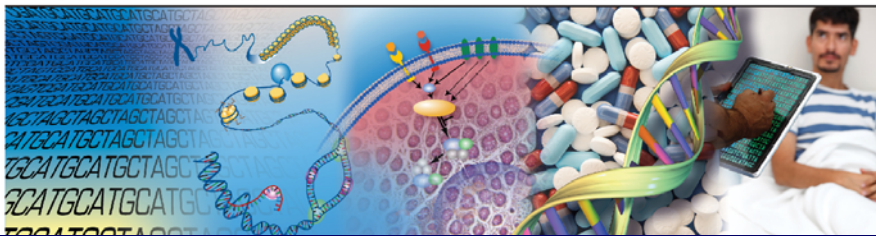
Understanding
the Structure of
Genomes

Understanding
the Biology of
Genomes

Understanding
the Biology of
Disease

Advancing
the Science of
Medicine

Improving the
Effectiveness of
Healthcare



**A pessimist sees the difficulty in every opportunity.
An optimist sees the opportunity in every difficulty.**

--Winston Churchill

The Relevance of Genomics



Biomedical Researchers



Healthcare Professionals



Patients (and Friends & Relatives of Patients)

Genomics, Society, and Public Health



Precision Medicine

- Today: most medical care based on expected response of the average patient
- Tomorrow: medical care based on individual genomic, environmental, and lifestyle differences that enable more precise ways to prevent and treat disease



How do we get from today to tomorrow?





“...[the] new Precision Medicine Initiative [will bring] America closer to curing diseases like cancer and diabetes, and gives all of us access, potentially, to the personalized information that we need to keep ourselves and our families healthier.”

President Barack Obama
January 30, 2015



The NEW ENGLAND JOURNAL of MEDICINE

January 30, 2015

Perspective

A New Initiative on Precision Medicine

Francis S. Collins, M.D., Ph.D., and Harold Varmus, M.D.

“Tonight, I’m launching a new Precision Medicine

Initiative to bring us closer to curing diseases like cancer and diabetes — and to give all of us access to the personalized information we need to keep ourselves and our families healthier.”

— President Barack Obama, State of the Union Address, January 20, 2015

The proposed initiative has two main components: a near-term focus on cancers and a longer-term aim to generate knowledge applicable to the whole range of health and disease. Both components are now within our reach because of advances in basic research, including molecular biology, genomics, and bioinformatics. Furthermore, the initiative

U.S. National Research Cohort



- >1 million U.S. volunteers
- Participants to share genomic data, lifestyle information, biological samples – all linked to their EHRs
- Forge new model for ‘doing science’ that emphasizes:
 - Engaged participants
 - Open, responsible data sharing
 - Strong privacy protections

Everything Old is New Again

insight commentary


The case for a US prospective cohort study of genes and environment

Francis S. Collins

National Human Genome Research Institute, National Institutes of Health, Building 31, Room 4B09, MSC 2152, 31 Center Drive, Bethesda, Maryland 20892-2152, USA (e-mail: fc23a@nih.gov)

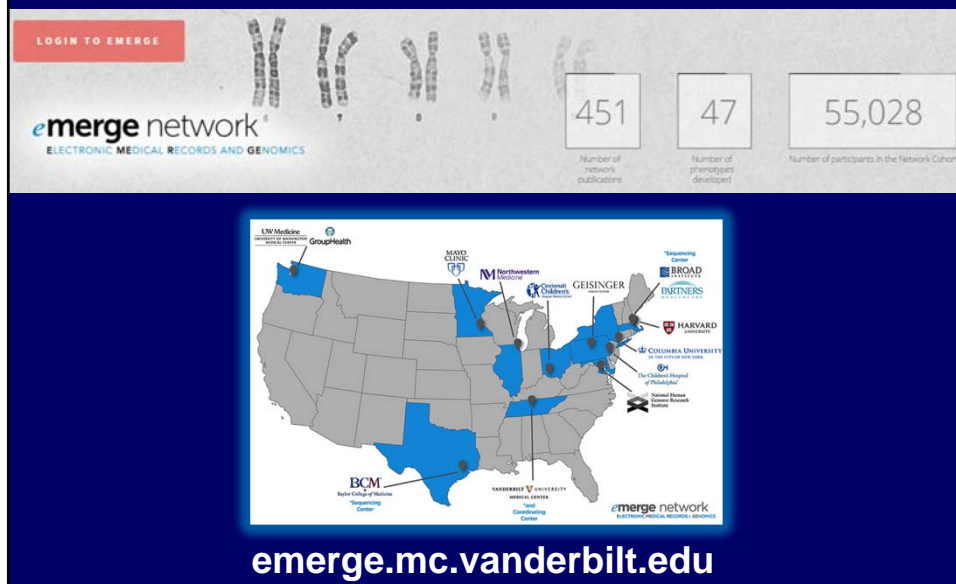
Information from the Human Genome Project will be vital for defining the genetic and environmental factors that contribute to health and disease. Well-designed case-control studies of people with and without a particular disease are essential for this, but rigorous and unbiased conclusions about the causes of diseases and their population-wide impact will require a representative population to be monitored over time (a prospective cohort study). The time is right for the United States to consider such a project.

Nature (2004)



Genomics **EHRs**

Electronic Medical Records and Genomics (eMERGE) Network




LOGIN TO EMERGE

emerge network
ELECTRONIC MEDICAL RECORDS AND GENOMICS

451	47	55,028
Number of network publications	Number of phenotypes developed	Number of participants in the Network Cohort

emerge.mc.vanderbilt.edu



Genomics

EHRs

Technologies

THE BODY ELECTRIC

RESEARCHERS WANT TO WIRE THE HUMAN BODY WITH SENSORS THAT COULD HARVEST REAMS OF DATA — AND TRANSFORM HEALTH CARE.

BY ELIZABETH GIBNEY

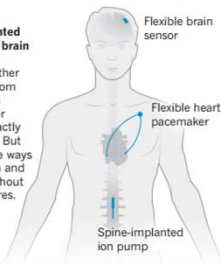
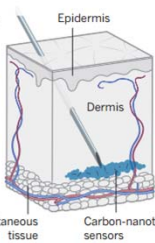
Nature (2015)

WIRED FOR LIFE Sensors woven into the body could alert people to medical problems before they become seriously ill — if the devices can overcome some daunting challenges.

Sensors mounted on the skin are easy to apply and remove, and can obtain high-quality data on breathing, heart rate, blood pressure and other vital signs. But they must be flexible and stretchy enough to follow the natural movement of the body.

Sensors injected under the skin can access the trove of information carried in the blood by chemical signals called biomarkers. The devices must be long-lived and biocompatible, so that they don't trigger an immune response.

Devices implanted into the heart, brain or other deep tissues can gather data directly from the source and deliver drugs or stimulation exactly where needed. But they must have ways to get power in and data out — without resorting to wires.







Genomics


EHRs

Technologies

Data Science

Participant Partnerships

Report on Precision Medicine Initiative Cohort Program



The Precision Medicine Initiative Cohort Program – Building a Research Foundation for 21st Century Medicine

Precision Medicine Initiative (PMI) Working Group Report to the Advisory Committee to the Director, NIH

September 17, 2015

For Immediate Release: Thursday, September 17, 2015

NIH framework points the way forward for building national, large-scale research cohort, a key component of the President's Precision Medicine Initiative

The National Institutes of Health Advisory Committee to the Director (ACD) today presented to NIH Director Francis S. Collins, M.D., Ph.D., a detailed design framework for building a national research participant group, called a cohort, of 1 million or more Americans to expand our knowledge and practice of precision medicine. Dr. Collins embraced the design recommendations made by the ACD, noting the need to remain nimble and adaptable as the Initiative progresses. He also thanked the Committee for their recommendations on policy issues and welcomed the opportunity to review them. NIH plans to move quickly to build the infrastructure so that participants can begin enrolling in the cohort in 2016, with a goal of enrolling at least 1 million participants in three to four years.

Precision Medicine Initiative

Health Information Grants & Funding News & Events Research & Training Institutes at NIH About NIH

NIH Home > Research & Training

PRECISION MEDICINE INITIATIVE

Precision Medicine Initiative

- Near-term Goals
- Longer-term Goals
- Scale and Scope
- Participation
- PM Working Group
- Events
- Announcements
- PMJ in the News
- Multimedia

Faces of the Precision Medicine Initiative – Dr. Russ Altman

NIH Director's blog: Read precision medicine-related blogs by the NIH Director.

ABOUT THE PRECISION MEDICINE INITIATIVE

Far too many diseases do not have a proven means of prevention or effective treatments. We must gain better insights into the biology of these diseases to make a difference for the millions of Americans who suffer from them. Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person. While significant advances in precision medicine have been made for select cancers, the practice is not currently in use for most diseases. Many efforts are underway to help make precision medicine the norm rather than the exception. To accelerate the pace, President Obama unveiled the Precision Medicine Initiative (PMI) – a bold new enterprise to revolutionize medicine and generate the scientific evidence needed to move the concept of precision medicine into every day clinical practice.

Email Updates

To sign up for updates please enter your e-mail address.

Related Links

- NEJM Perspective: A New Initiative on Precision Medicine
- White House Precision Medicine Web Page
- White House Fact Sheet: President Obama's Precision Medicine Initiative
- Precision Medicine Initiative and Cancer Research
- Storify: #PMINetwork Twitter Chat
- Storify: The Precision Medicine Initiative Announcement
- Precision Medicine Initiative YouTube Channel

www.nih.gov/precisionmedicine

Déjà Vu, All Over Again?




Human Genome Project

Circa Winter 1990



Precision Medicine Initiative

Circa Winter 2015

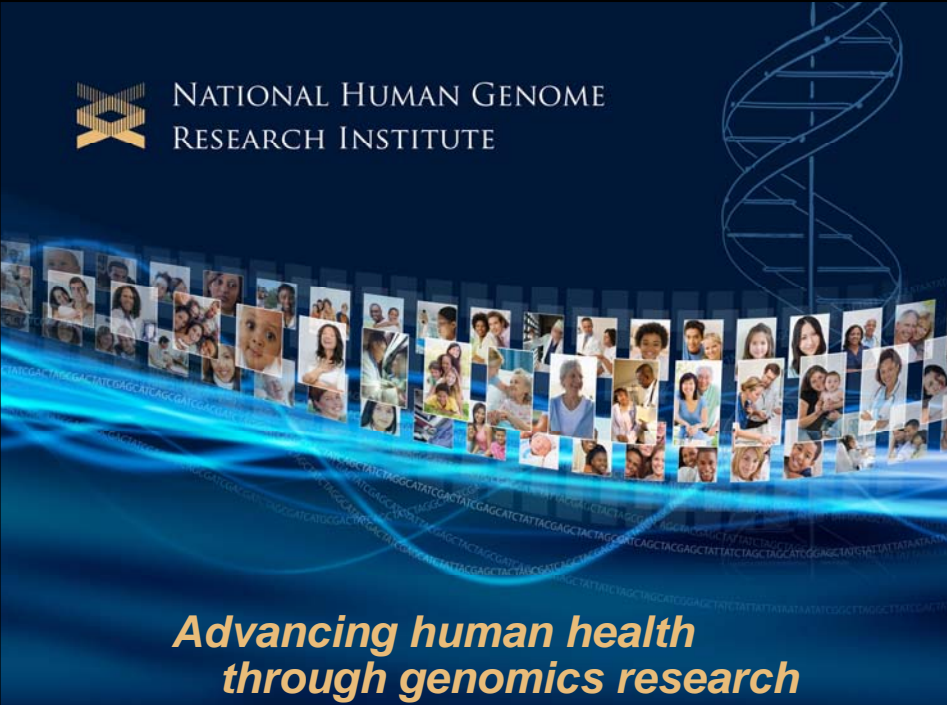


The Genomics Landscape
A monthly newsletter from the NHGRI Director

October 6, 2015

This month brought a historic 'odometer moment' for the field of genomics – October 1, 2015, marked the 25th anniversary of the launch of the Human Genome Project. I, for one, cannot believe a quarter-century has now passed since many of us started working on the Project. At the same time, it is truly incredible to think about how far genomics has progressed since that time. I thought the significance of this anniversary warranted making this topic the lead story in this month's *The Genomics Landscape*; in addition, I reflect on this important anniversary in a recent video interview now available on the NHGRI web site.

To subscribe, follow link from:
genome.gov/Director



**NATIONAL HUMAN GENOME
RESEARCH INSTITUTE**

**Advancing human health
through genomics research**