

# The Genomics Landscape a Decade after the Human Genome Project

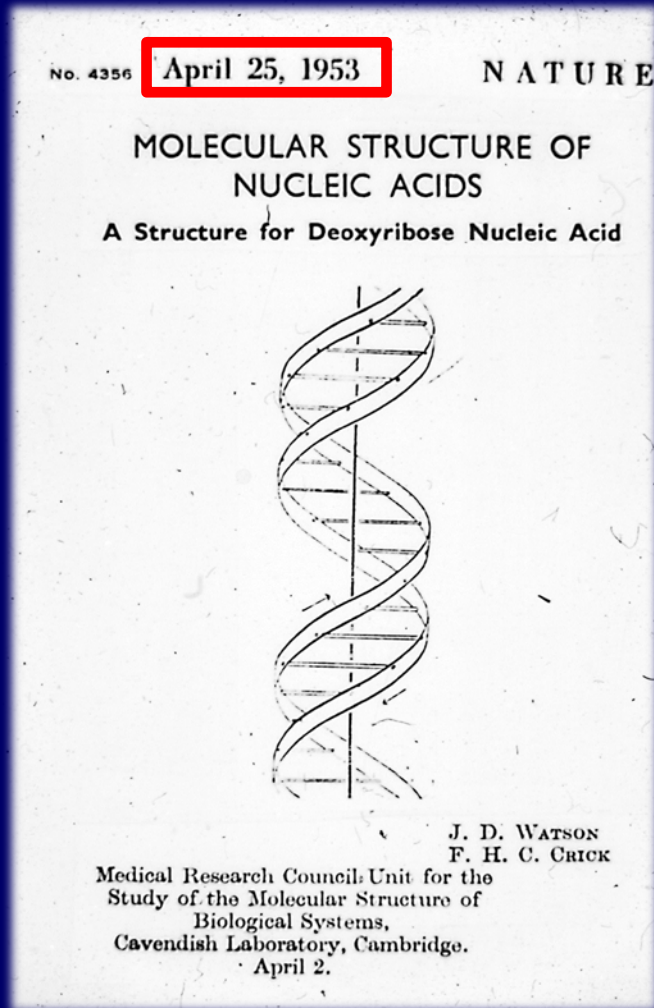
## Special Symposium

Ruth L. Kirschstein Auditorium  
Natcher Conference Center  
National Institutes of Health



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# April 25, 1953



## Discovery of Double-Helical Structure of DNA

# April 14, 2003

National Human Genome Research Institute

*National Institutes of Health  
U.S. Department of Health and Human Services*



National Human Genome Research Institute  
National Institutes of Health  
Department of Health and Human Services  
and  
Office of Science  
U.S. Department of Energy

## **International Consortium Completes Human Genome Project**

*All Goals Achieved; New Vision for Genome Research Unveiled*

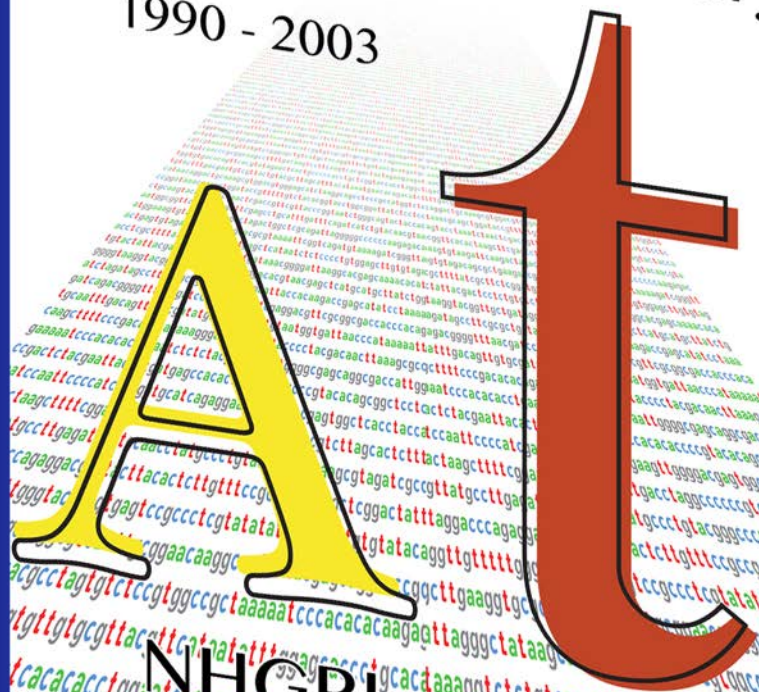
**BETHESDA, Md.**, April 14, 2003 - The International Human Genome Sequencing Consortium, led in the United States by the National Human Genome Research Institute (NHGRI) and the Department of Energy (DOE), today announced the successful completion of the Human Genome Project more than two years ahead of schedule.

10<sup>th</sup>  
Anniversary  
Human  
Genome  
Project  
1990 - 2003



NHGRI

Human  
Genome  
Project  
1990 - 2003  
10<sup>th</sup>  
Anniversary



NHGRI

# Milestones in Genomics

**1990**

**HGP  
Begins**



**2003**

**HGP  
Ends**



**2013**

**10 Years  
after HGP**



# U.S. Presidents

1990



**Bush #1**

2003



**Bush #2**

2013



**Obama**

# Music Devices

1990



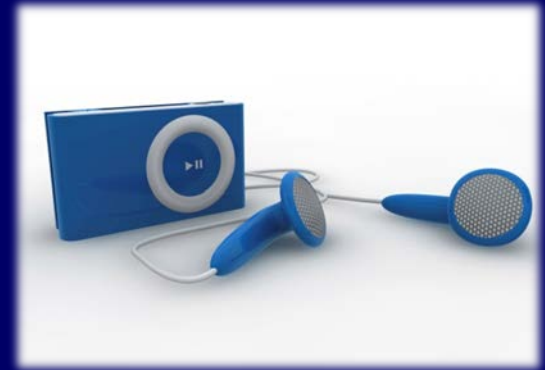
Walkman

2003



Discman

2013



MP3 Player

# Computer Devices

1990



**Desktop**

2003



**Laptop**

2013



**Smartphone**



# Routine Communication

1990



Fax

2003



E-mail

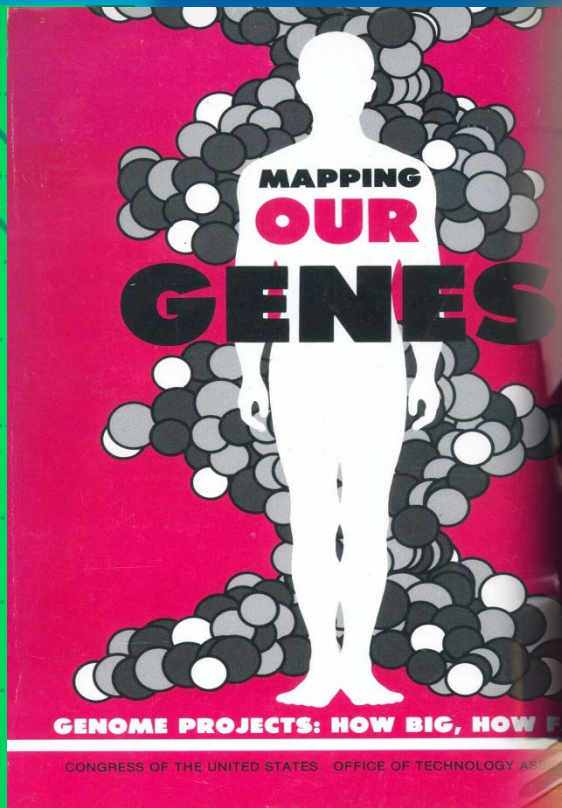
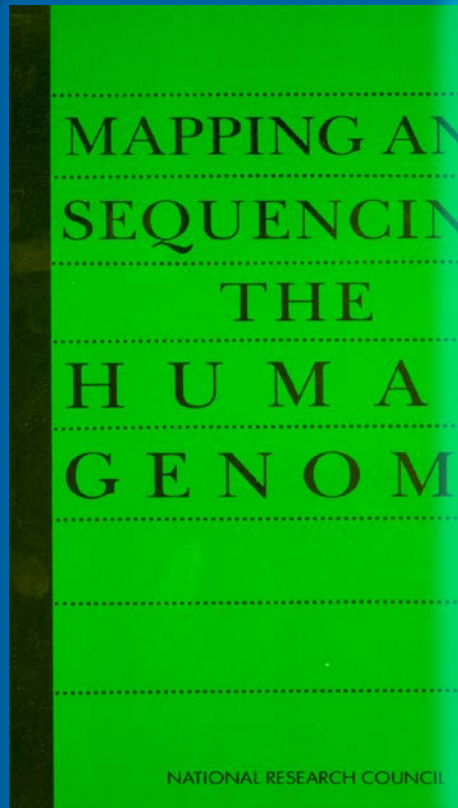
2013



Tweet

# Different Eras for Genomics & NHGRI

1990



# Different Eras for Genomics & NHGRI

2003

From Double Helix

to Human Sequencing  
- and Beyond



Scientific Symposium  
Natcher Auditorium  
National Institutes of Health  
April 14 - 15, 2003

The Human Genome Project

and Beyond



Mark Walport

Aravind Parthasarathy

# Different Eras for Genomics & NHGRI

2013

nature

THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE



THE FUTURE IS BRIGHT

Reflections on the first ten years of the human genomics age

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

**METHODS**  
MORE BASES PER DOLLAR  
Elaine Mardis on the march of sequencing technology  
PAGE 188

**HEALTH**  
FROM LAB TO CLINIC  
A road map to genomic medicine  
PAGE 194

NATURE ASIA.COM  
10 February 2011  
Vol. 470, No. 7323

## PERSPECTIVE

### Charting a course for genomic medicine from base pairs to bedside

Eric D. Green<sup>1</sup>, Mark S. Geyer<sup>1</sup> & National Human Genome Research Institute\*

There has been much progress in genomics in the ten years since a draft sequence of the human genome. Opportunities for understanding health and disease are now unprecedented, as advances in genomics obtain robust foundational knowledge about the structure and function of the human genome and also contributions to human health and disease. Here we articulate a 2011 vision for the future of genomics: 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<sup>1</sup>, genomics has become a mainstay of biomedical research. The scientific community's foresight in launching this ambitious project is evident in the broad range of scientific advances that the HGP has enabled, as shown in Fig. 1 (see ref60). Optimism about the potential contributions of genomics for improving human health has been fuelled by new insights about cancer<sup>2</sup>, the molecular basis of inherited diseases (http://www.ncbi.nlm.nih.gov/omim and http://www.genome.gov/GWASdb) and the role of structural variation in disease<sup>3</sup>, some of which have already led to new therapies<sup>4,5</sup>. Other advances have already changed medical practice (for example, microarrays are now used for clinical detection of genomic imbalances<sup>6</sup> and pharmacogenomic testing is routinely performed before administration of certain medications<sup>7</sup>). Together, these achievements (see accompanying paper<sup>8</sup>) document that genomics is contributing to a better understanding of human biology and to improving human health.

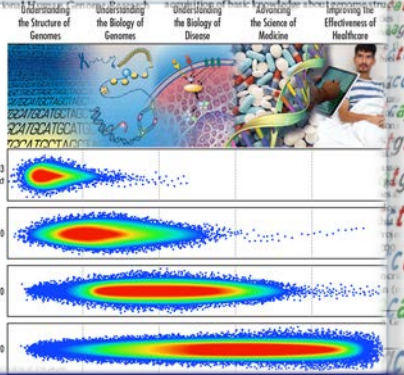
As it did eight years ago<sup>9</sup>, the National Human Genome Research Institute (NHGRI) has organized a series of workshops to plan and explore future directions have led to an updated vision for genomics in biology and the diagnosis and treatment of disease, including consideration of the use of genomics in agriculture, energy and other areas, and is broader than what is realizing the full benefits of genomics.

This 2011 vision for genomics is based on research that, over time, the most fundamental to understanding disease and health. At the same time, genomics offers opportunities for understanding disease based on genomic predisposition, and also to understand normal biology and the path to disease. The path to disease has been about biology and its contribution to understanding will accelerate based on genomic medicine.

\*National Human Genome Research Institute, Bethesda, MD, USA

quickly. Although genomics has already begun to lead to new treatments in a few circumstances, profound effectiveness of the therapies is currently realized by only a few (Fig. 2). Achieving such progress will depend not only on new policies, practices and other developments, but also on the kinds of achievements that can be realized in the next decade (Box 2) where a confluence of need and opportunity will lead to major accomplishments in genomic medicine. Similarly, we note three cross-cutting areas that are relevant and fundamental across the entire spectrum of genomic medicine: bioinformatics and computational genomics, education and training (Box 4), and genomics and

Understanding the biology of genomes. Substantial progress in understanding the structure and function of the genome revealed much about the complexity of genomes and the role of structural variation in disease.

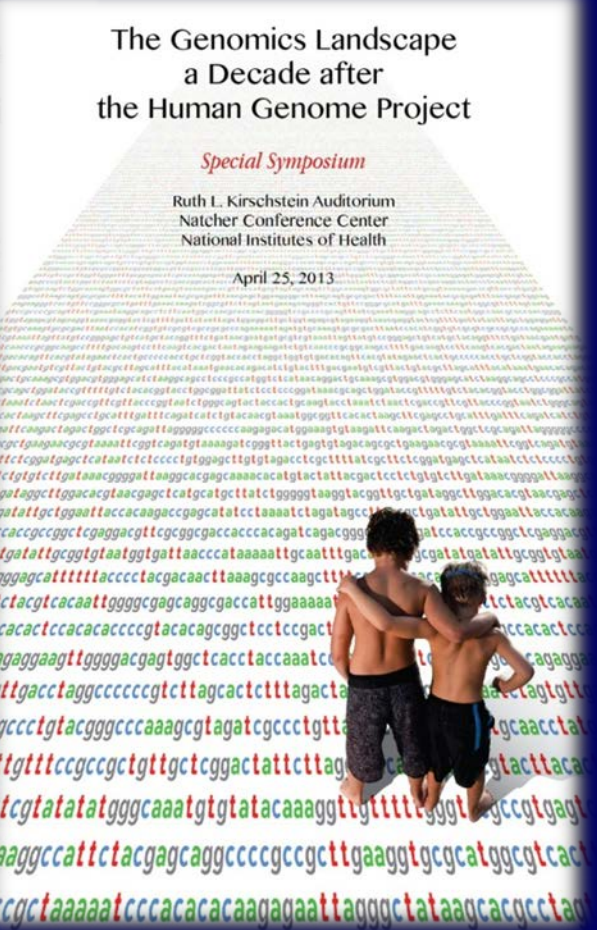


### The Genomics Landscape a Decade after the Human Genome Project

Special Symposium

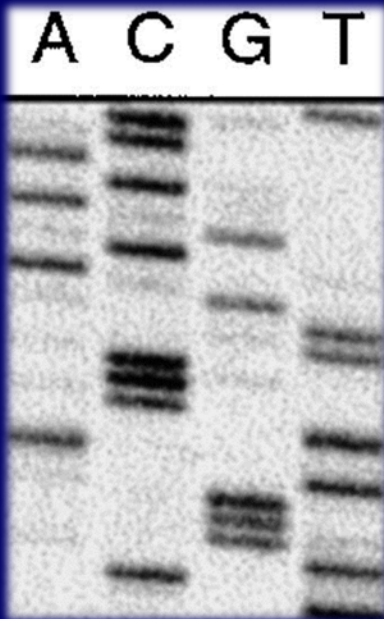
Ruth L. Kirschstein Auditorium  
Natcher Conference Center  
National Institutes of Health

April 25, 2013



# DNA Sequencing Technology

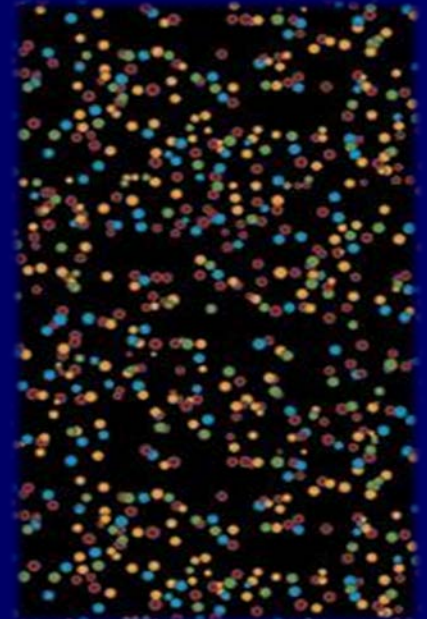
1990



2003



2013



# Human Genome Sequences

1990

2003

2013

Time to Generate a Human Genome Sequence

~6-8 years

~3-4 months

~2-3 days

Cost to Generate a Human Genome Sequence

~\$1B

~\$10-50M

~\$4-6K

Generated Human Genome Sequences

0

1

Thousands

# Genome Sequences

1990

2003

2013

## Vertebrate Genome Sequences

0

3

112

## Non-Vertebrate, Eukaryotic Genome Sequences

0

14

455

## Prokaryotic Genome Sequences

0

167

8760

# Genome Sequence Data

1990

2003

2013

## Total DNA Bases in GenBank

~49M

~31T

~150T

## Whole-Genome Shotgun Bases in GenBank

0

~9.6T

~391T



# Human Single-Nucleotide Polymorphisms in Public Databases

1990

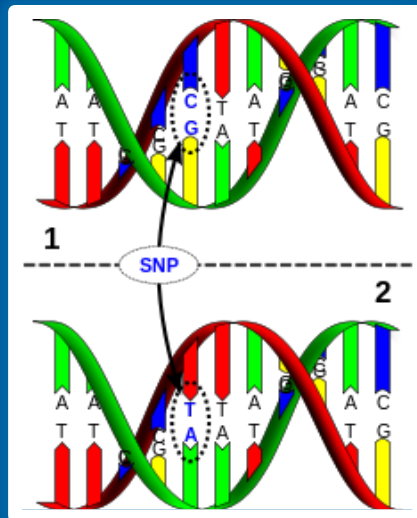
2003

2013

~4.4K

~3.4M

~54M



```
AGAGTTCCTGTCGAGA  
AGGGTTATGGCGAGA  
CGTTACGGGAATCC  
CCTTACGGGAATCT  
TCTTTGACGACTCCT  
CCTTAGAGGACTCCT  
AGAGCTGGTCTAGAT  
AGAACTGGTATAGGT  
CCTAGGGCGTTACAA  
CCTTGGCGTGAACA  
AAGCTTGGCCGAACC  
AGGCTTAGCCGAACC  
CCAGTACATGAACGA  
CCGGTACATGTACGA
```

# Rare Genetic Diseases

1990

2003

2013

**No. Diseases with Known Molecular Basis**

61

2264

4847

**No. Genes with Known Disease-Causing Mutations**

53

1474

2972

# Common Genetic Diseases

1990

2003

2013

**No. Published Genome-Wide Association Studies (GWAS)**

0

0

~1550

**No. Replicated Disease-Associated Variants**

0

6

~2900

# Drugs with Pharmacogenomics Information on Label

1990

2003

2013

4

46

106



# The Changing Genomics Landscape

1990

2003

2013



The word "Thanks!" is rendered in a bold, 3D, red font. The letters are thick and have a slight shadow underneath, giving them a three-dimensional appearance. The text is centered at the top of the slide.

**Rudy Pozzatti & Brad Ozenberger (Co-Chairs)**  
**Annette Sante (All Things Logistic)**  
**Darryl Leja (Graphics Extraordinaire)**  
**Larry Thompson & Team (Videography)**

**Jessica Barry**  
**Vence Bonham**  
**Larry Brody**  
**Adam Felsenfeld**  
**Allison Mandich**  
**Derek Scholes**

**Barbara Biesecker**  
**Joy Boyer**  
**Priscilla Crockett**  
**Lucia Hindorff**  
**Bill Pavan**  
**Kris Wetterstrand**

# NHGRI-Smithsonian Partnership



National Human Genome  
Research Institute



Smithsonian  
*National Museum of Natural History*

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