From the Human Genome Project to Genomic Medicine

A Journey to Advance Human Health

Eric Green, M.D., Ph.D.
Director, NHGRI
"For the newly developing discipline of [genome] mapping/sequencing (including the analysis of the information), we have adopted the term GENOMICS..."
‘The Genome Institute’

Office for Human Genome Research
1988-1989

National Center for Human Genome Research
1989-1997

National Human Genome Research Institute
1997-present
NHGRI: Circa 1990-2003

Human Genome Project
NHGRI Today: Characteristic Features

- Relatively young (~28 years)
- Relatively small (~1.7% of NIH)
- Unusual historical origins (think ‘Human Genome Project’)
- Emphasis on ‘Team Science’ (think managed ‘consortia’)
- Rapidly disseminating footprint (think ‘genomics’)
- Novel societal/bioethics research component (think ‘ELSI’)
- Over-achievers for trans-NIH initiatives (think ‘Common Fund’)
- Vibrant (and large) Intramural Research Program
A Quarter Century of Genomics

Human Genome Sequenced for First Time by the Human Genome Project
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
A vision for the future of genomics research

Nature

Charting a course for genomic medicine from base pairs to bedside

Nature

Base Pairs to Bedside

2003

2011

Heli to Health
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
In Silicon Valley, Moore’s law seems to stand on equal footing with the natural laws codified by Isaac Newton. Intel co-founder Gordon Moore’s iconic observation that computing power tends to double — and that its price therefore halves — every 2 years has held true for nearly 50 years with only minor revision. But as an exemplar of rapid change, it is the target of playful abuse from...
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
A global reference for human genetic variation

The 1000 Genomes Project Consortium*

A Quarter Century of Genomics

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- 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
ENCODE: Giving ‘GPS’ Views of Genomes
A Quarter Century of Genomics

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- Significant Advances in Unraveling the Genomic Bases of Human Disease
Genomic Architecture of Genetic Diseases

NHGRI Centers for Common Disease Genomics

Cardiovascular Disease
Hemorrhagic Stroke
Autism
Epilepsy
Asthma

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

Manolio et al., J Clin Invest (2008)
A Quarter Century of Genomics

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- Vivid Examples of Genomic Medicine in Action Now Emerging
Bringing Genomic Medicine Into Focus
‘Hot Areas’ in Genomic Medicine

Cancer Genomics

Pharmacogenomics
Electronic Medical Records and Genomics (eMERGE) Network

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

emerge.mc.vanderbilt.edu
‘Hot Areas’ in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Rare Genetic Disease Diagnostics
“...disorders not readily explained by standard tests can sometimes be diagnosed through genome sequencing and analysis.”

‘Hot Areas’ in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Rare Genetic Disease Diagnostics
- Newborn Genome Sequencing
In 2025, Everyone Will Get DNA Mapped At Birth

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.

Time (2014)
Newborn Sequencing In Genomic medicine and public Health (NSIGHT)

UCSF
San Francisco, CA

Harvard Hospitals
Boston, MA

Children’s Mercy
Kansas City, MO

UNC – Chapel Hill
Chapel Hill, NC
‘Hot Areas’ in Genomic Medicine

- Cancer Genomics
- Pharmacogenomics
- Rare Genetic Disease Diagnostics
- Newborn Genome Sequencing
- Clinical Genomics Information Systems
Clinical Genome Resource (ClinGen)


Technological advances are quickly allowing genome-wide analysis to become commonplace in the care of patients. However, the ability to detect DNA variants has greatly surpassed the ability to interpret their clinical impact, limiting patient benefit. Improving genomic interpretation will require a coordinated effort from both the clinical and research communities. Learn more »

clinicalgenome.org

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)
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.
Advancing human health through genomics research