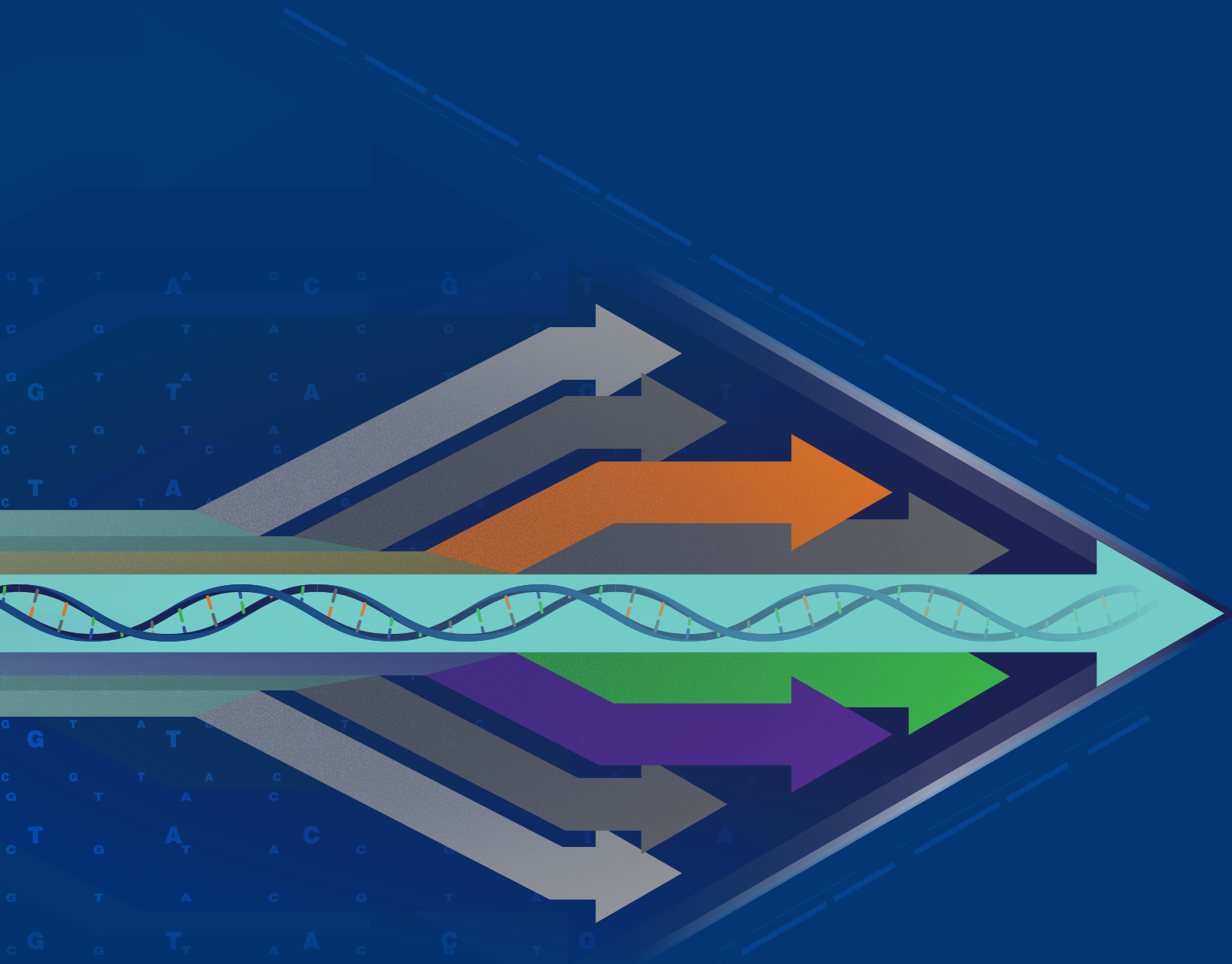


# National Human Genome Research Institute



—  
The **Forefront**  
of **Genomics**<sup>®</sup>  
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The National Human Genome Research Institute (NHGRI) is one of the 27 Institutes and Centers at the U.S. National Institutes of Health (NIH). In addition to being an international leader in funding and conducting genomics research, NHGRI develops resources, technologies, and policies for advancing genomics and its application to improving human health. The Institute also supports the training of researchers and the dissemination of genomic knowledge to the public and to health professionals. Additional information about NHGRI can be found at [genome.gov](http://genome.gov).

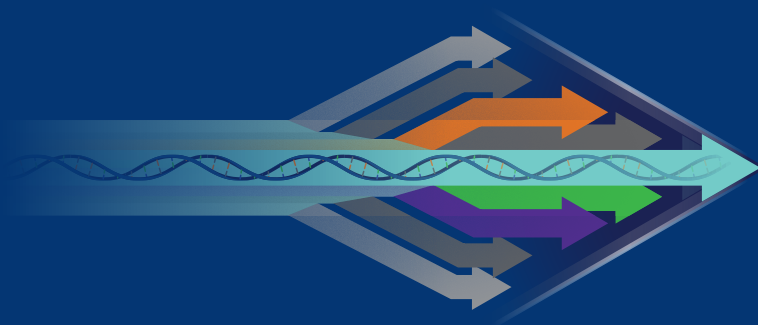
NIH, the nation's medical research agency, is a component of the U.S. Department of Health and Human Services. NIH is the primary federal agency that conducts and supports basic, translational, and clinical research, and investigates the causes, treatments, and cures for both common and rare diseases. For more information about NIH and its programs, visit [nih.gov](http://nih.gov).

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of **Genomics**<sup>®</sup>  
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Starting with the launch of the Human Genome Project in 1990, genomics has become progressively entrenched within the bedrock of the biomedical research enterprise. Capitalizing on the momentum of the Project's successful completion in 2003, genomics now regularly plays a central and catalytic role in basic and translational research, and studies increasingly demonstrate how genomic information can be effectively used in clinical care. I am proud to lead NHGRI, which has been a pioneer in genomics for more than three decades.



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# Director's Message

NHGRI was established in the late 1980s to lead NIH's efforts in the Human Genome Project, the audacious international endeavor that deciphered the order of the approximately 3 billion "letters" that make up the human "blueprint" (the human genome sequence). Completed in 2003, the Human Genome Project laid the foundation for the burgeoning field of genomics. Fast forward to today, and NHGRI is the largest organization in the world dedicated to genomics research.

Genomics is central to understanding human biology and human disease – everything starts with the genome's long strands of deoxyribonucleic acid (DNA). The information encoded in DNA provides the basic instructions for our lives, and subtle variations in our genomes greatly influence our health, our risk for disease, and many of our features. While NHGRI receives less than 2% of NIH's total annual budget, its genomics research programs are critical for the work supported by other NIH Institutes and Centers whose missions are mostly focused on specific disease areas. In addition, NHGRI leads multiple research programs supported by the NIH Common Fund ([commonfund.nih.gov](http://commonfund.nih.gov)), a trans-NIH pool of funds used for short-term, exceptionally high-impact projects that aim to eliminate scientific roadblocks.

The majority of NHGRI's funds are used to support genomics research at leading academic and commercial institutions across the U.S. and around the world. NHGRI is best known for funding and leading large, consortium-based programs, but it also supports the work of many individual investigators. In addition, roughly 20% of NHGRI's funds are used to conduct genomics research in the Institute's laboratories in and around Bethesda, Maryland. These efforts are led by government-based investigators who work at NHGRI and who capitalize on the unique strengths of NIH's Bethesda campus and its associated research and clinical infrastructure to pursue a wide range of genomic studies.

At NHGRI, we believe that it is critical to study the broader societal implications of genomics and genomic advances. From the beginning, the Institute has dedicated a fixed portion of its budget to study the ethical, legal, and social implications of genomics research. In addition, NHGRI staff regularly engage in active dialogue with many societal

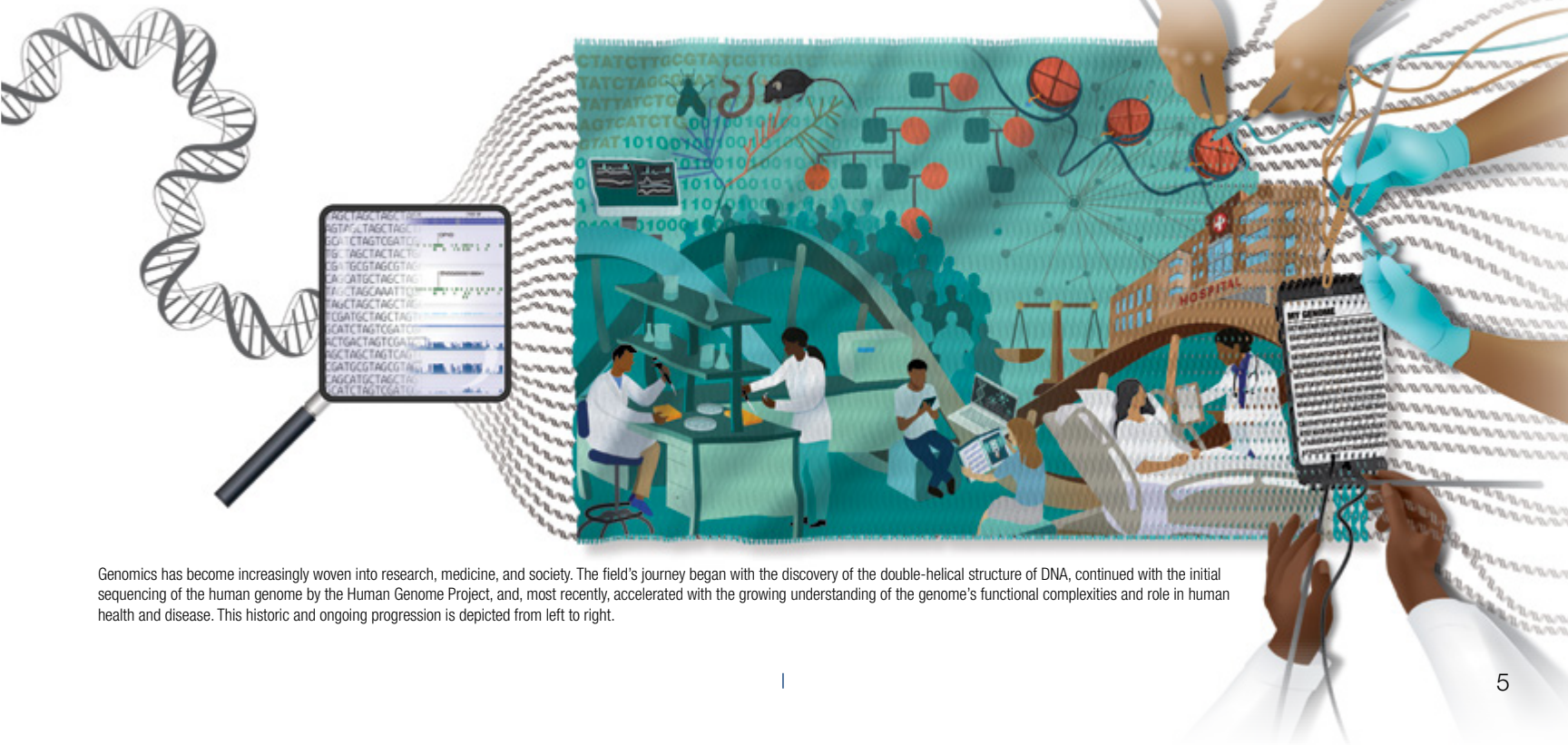
audiences, including educational institutions, community organizations, healthcare professionals, and the public. To facilitate these interactions, we use many different communication tools, including our widely respected website ([genome.gov](http://genome.gov)), our GenomeTV channel on YouTube, and other social media outlets (such as Facebook and Twitter).

The scope, scale, and pace of genomic advances to date were nearly unimaginable when the Human Genome Project began. Even today, such advances continue to yield scientific and clinical opportunities beyond our initial expectations, with many more anticipated in the decade ahead. Going forward, NHGRI remains "laser focused" on helping to fulfill the promise of the Human Genome Project, which precisely aligns with our core vision: *To improve the health of all humans through advances in genomics research. We pursue this vision through our fundamental mission: As a leading authority in the field of genomics, our mission is to accelerate scientific and medical breakthroughs that improve human health. We do this by driving cutting-edge research, developing new technologies, and studying the impact of genomics on society.*

Because of the near-ubiquitous nature of genomics in biomedicine and other areas of the life sciences, NHGRI recognizes that it can no longer lead all aspects of genomics research. Rather, the Institute must focus its attention on the most compelling opportunities at the forefront of the field. This realization is embodied by our recently conceived organizational mantra: *The Forefront of Genomics*. The spirit of this mantra increasingly serves to guide the NHGRI's pursuits.

I invite you to learn more about our research portfolio and our many associated programs by reading this brochure and visiting our website at [genome.gov](http://genome.gov).

Eric D. Green, M.D., Ph.D.  
Director, National Human Genome Research Institute



Genomics has become increasingly woven into research, medicine, and society. The field's journey began with the discovery of the double-helical structure of DNA, continued with the initial sequencing of the human genome by the Human Genome Project, and, most recently, accelerated with the growing understanding of the genome's functional complexities and role in human health and disease. This historic and ongoing progression is depicted from left to right.

# Genomics Primer

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Some basics about the human genome:

- To build a house or a car, you need a blueprint, a parts list, and assembly instructions. For all living creatures, the genome functions as all three.
- Our blueprint – the human genome – resides within tiny double-stranded fibers of DNA that are packed into chromosomes in the nucleus of cells.

## **ge·nome**

noun [JEE-nohm]: all of the DNA in a cell

## **ge·no·mics**

noun [je-NO-miks]: the study of genomes

- The typical human cell contains 23 pairs of chromosomes; one set of 23 chromosomes comes from each parent.
- In humans, males and females have two copies of 22 of these chromosomes (called autosomes). For the 23rd pair, known as the sex chromosomes, males and females differ – typically, females have two X chromosomes, while males have one X chromosome and one Y chromosome.
- DNA is made of four different building blocks, and each is a different chemical (called a nucleotide or base): adenine (**A**), thymine (**T**), cytosine (**C**), and guanine (**G**).
- The order of the A, T, C, and G building blocks (“letters”) in the human genome encodes the biological instructions that tell each cell what to do and when to do it.
- Each set of 23 chromosomes (essentially one copy of the human genome) contains about 3 billion A, T, C, and G letters – so the typical human cell contains roughly 6 billion of these genomic letters in total.
- The genome of each person is slightly different from the genome of every other person. The genome sequences (the order of the genomic letters) of any two people typically contain a different letter roughly once in every 1,000 positions.
- Most genomic differences (or variants) are subtle in their effects or even inconsequential, but some are known to influence our physical traits and health.
- The advanced technologies used in genomics research routinely generate massive amounts of big data that are challenging to collect, store, organize, analyze, and share. For instance, a single research project can generate terabytes or even petabytes of genomic data each month.



# Benefits of Genomics Research

## *Genomics is an engine for both scientific and economic growth*

The scientific leaders who proposed the Human Genome Project in the 1980s envisioned numerous benefits from the multibillion-dollar investment, among them that: (1) Mapping and sequencing the human genome would increase the basic understanding about how the genome works and, therefore, how cells work; (2) Genomic knowledge would accelerate medical research, yielding fundamental insights about inherited diseases and disorders like cancer; and (3) Technological advances coupled with new genomics-enabled scientific opportunities would stimulate the biotechnology industry (then less than a decade old), helping to propel the world economy into the next century.

### **Increasing basic understanding of the genome**

Genomics research has expanded our grasp of the interplay between genome structure and function and has clarified many misconceptions. As the Human Genome Project got under way, many scientists estimated that humans – being the complex creatures that we are – contain 100,000 or more genes in our genomes. Today, we know that number is much lower, more like about 20,000 genes. We have also learned that those vast stretches of DNA that do not code for proteins – which some used to refer to as “junk DNA” – are far from useless. In fact, studies over the last two decades have revealed the presence of hundreds of thousands of functional elements within the large portions of the human genome that do not directly code for proteins. Many of these elements play crucial roles in controlling the activity of our genes.

### **Accelerating medical research**

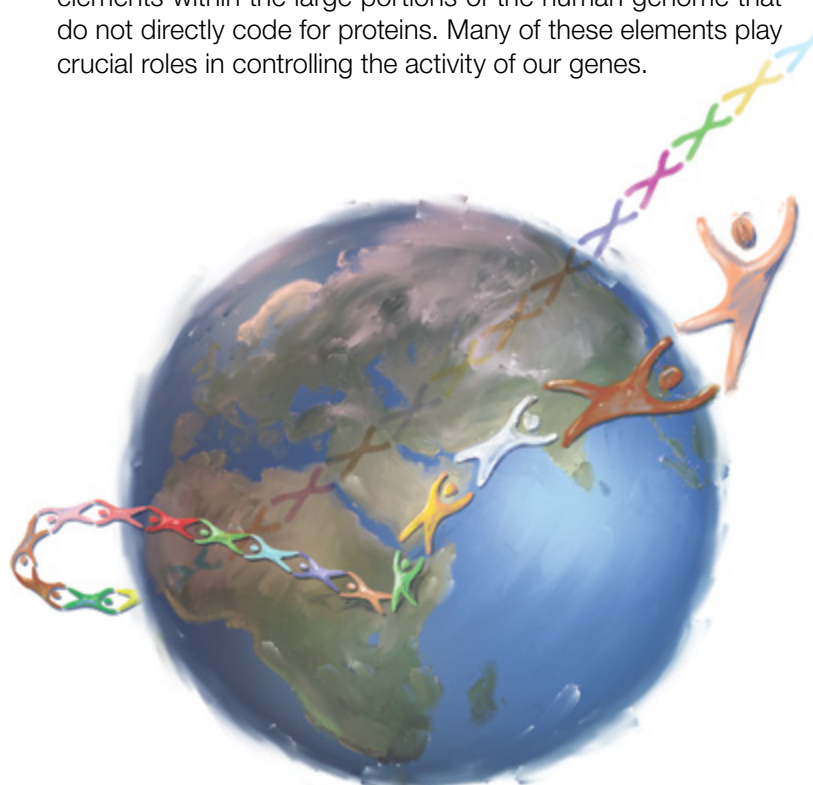
Genomics has become a central discipline of biomedical research, quickly spreading across the entire research landscape. Nearly all other NIH Institutes and Centers, as well as many other private and public institutions in the U.S. and around the world, now have made major investments in genomics.

NHGRI and other components of NIH have launched important partnerships using genomics to study areas of mutual interest. Past examples of such partnerships include: (1) The Cancer Genome Atlas (TCGA), a joint endeavor between NHGRI and the National Cancer Institute, which investigated the genomics of cancer; and (2) The Newborn Sequencing in Genomic Medicine and Public Health (NSIGHT) program, a collaboration between NHGRI and the Eunice Kennedy Shriver National Institute of Child Health and Human Development, which explored the implications, opportunities, and challenges of using genome sequence information in the newborn period.

### **Stimulating the economy**

Since the completion of the Human Genome Project, the largest driver of genomic advances has been the stunning progress in developing more powerful technologies for sequencing DNA. Catalyzed by an NHGRI program in technology development and significant investments by the private sector, the costs of sequencing DNA have plummeted at a pace exceeding Moore’s Law (the observation that computing power doubles roughly every 24 months). Along with cost reductions, the speed of sequencing genomes has increased substantially (see graphic on page 13).

These technological developments, in combination with other genomic advances, have been a boon for the economy. A 2013 analysis by the Battelle Technology Partnership Practice determined that from 1988 to 2012, the U.S. government invested \$11.3 billion (\$14.5 billion in 2012 dollars) in the Human Genome Project and related areas of genomics research. This investment in turn generated nearly \$1 trillion in economic activity, more than 4.3 million job-years of supported employment, and \$54.8 billion in tax revenues from genomics research, development, and commercial activities. The 2013 analysis also noted that for every \$1 invested in genomics by the U.S. government, there was a return on investment of roughly \$65 for the U.S. economy. These and other measures of economic output repeatedly demonstrate that the overall effects of the federal investment in genomics have been positive and substantial.



# History of Genomics

*Genomics is a young discipline built on the work of early geneticists and, later, molecular biologists.*

In the 1860s, an Austrian monk named Gregor Mendel studied pea plants to understand patterns of inheritance, but he had no idea what carried traits from one generation to the next. The Swiss physician Friedrich Miescher discovered DNA in the 1870s; he named it “nuclein” because it existed in the cell’s nucleus. Decades later, in the 1940s, the Canadian American physician Oswald Avery and colleagues proved that DNA is the molecule containing life’s inherited information.

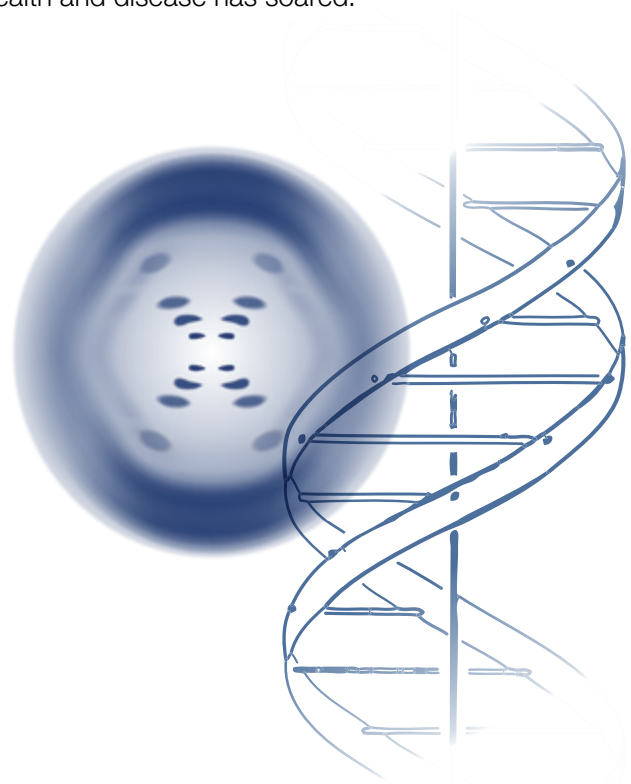
In 1953, American biologist James Watson and British physicist Francis Crick described the double-helical structure of DNA, a finding deduced from British chemist and crystallographer Rosalind Franklin’s X-ray diffraction images of DNA. This pivotal insight provided the definitive piece of the puzzle about how DNA serves as the molecule of heredity, carrying genetic information from one cell to the next and from one generation to the next.

The 1960s brought critical insights about DNA structure and function. This included deciphering the genetic code – the fundamental rules about how DNA’s nucleotides (represented by As, Ts, Cs, and Gs) encode the instructions for making proteins – and developing a more refined view of the units of DNA responsible for encoding proteins (genes).

A detailed understanding of how genes work remained beyond reach, however, until the molecular biology revolution of the 1970s and 1980s, an era that brought powerful new tools to study and manipulate DNA. Among other major advances of that time, British biochemist Frederick Sanger and American biologist Walter Gilbert independently developed the first techniques for determining the sequence of nucleotides in DNA.

Improvements to the early methods for isolating, analyzing, and sequencing DNA led to the notion of being comprehensive in studying an organism’s DNA (its genome). This, in turn, led to the Human Genome Project, which launched in 1990 with the signature aim of generating the first sequence of the human genome. The ambitious and compelling nature of the Project stimulated interest in genomics among scientists and the public alike.

Within 10 years, researchers generated an initial draft of the human genome sequence. In April 2003, leaders of the Human Genome Project revealed a finished sequence of the 3-billion-nucleotide human genome, marking the end of the 13-year odyssey. In many ways, that historic milestone marked the starting line for what has since transpired, as knowledge about genome function and the genome’s role in health and disease has soared.





# Overview of NHGRI's Organization

*The Office of the Director and six divisions together serve to align the Institute's structure with its mission.*

NHGRI leads the field of genomics by strategically investing in highly innovative research studies, technologies, and data resources that are needed to determine how the human genome works and how to use that knowledge for advancing human health. The Institute was initially established as an NIH Center in 1989 to lead NIH's effort in the Human Genome Project; later, it was elevated to an Institute and renamed the National Human Genome Research Institute in 1997. Today, the Institute's organizational structure allows for the productive pursuit of all aspects of its mission, including those related to research, training, and outreach. These efforts are executed through NHGRI's two overarching programmatic components:

- The **Extramural Research Program**, which funds genomics research and training opportunities at universities, medical centers, and other institutions throughout the U.S. and abroad.
- The **Intramural Research Program**, which supports government-based genomics research and training at NIH.

## National Human Genome Research Institute

### Office of the Director

Provides overall leadership for the entire Institute; oversees general operations, fiscal stewardship, scientific management, and communications; and coordinates genomics research within NIH and with other federal, private, and international programs.

### Division of Management

Leads a wide range of activities on behalf of the entire Institute, including financial management, administrative services, information technology, and employee ethics oversight.

### Division of Intramural Research

Constitutes the Institute's Intramural Research Program, which conducts genomics research in NHGRI's laboratories on and around the main NIH campus in Bethesda, Maryland.

### Division of Genome Sciences

Leads the Institute's extramural efforts in basic genomics research, technology development, and genomic data science.

### Division of Genomic Medicine

Leads the Institute's extramural efforts in the application of genomics to medical science and clinical care.

### Division of Genomics and Society

Leads the Institute's extramural efforts in studying the societal issues relevant to genomics research through the Ethical, Legal, and Social Implications (ELSI) Research Program; also oversees policy development, legislative affairs, and educational outreach programs.

### Division of Extramural Operations

Manages operational aspects of the Institute's Extramural Research Program, such as grants management and scientific review.

### Extramural Research Program

Strategic Plan:  
*New Goals for the U.S.  
Human Genome Project*

1998

Strategic Plan:  
*A Vision for the Future of  
Genomics Research*

2003

Eric Green appointed  
NHGRI Director

2009

NHGRI reorganizes  
to accommodate  
expanding research mission

2012

President Obama announces the  
U.S. Precision Medicine Initiative  
NIH's *All of Us* Research Program

2015

2003

Human Genome Project  
completed

2008

Genetic Information  
Nondiscrimination Act  
(GINA) becomes U.S. law

2011

Strategic Plan:  
*Charting a Course for  
Genomic Medicine from  
Base Pairs to Bedside*

2013

NHGRI-Smithsonian exhibition  
*Genome: Unlocking Life's Code*  
opens

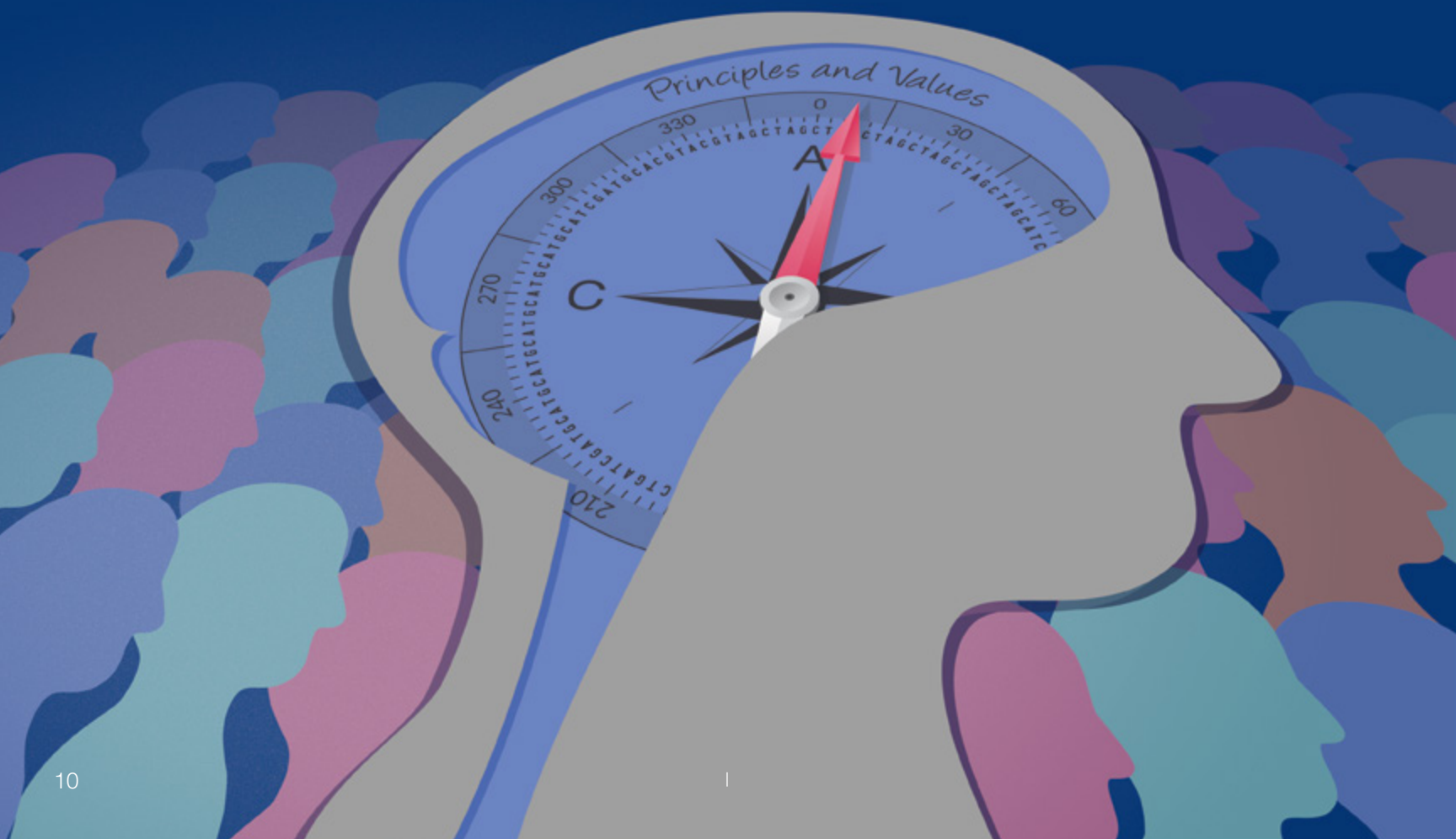
2020

NHGRI Strategic Vision:  
*Strategic Vision for Improving  
Human Health at The Forefront  
of Genomics*

# NHGRI Core Principles and Values

Successful organizations are guided by core principles and values that shape how they pursue their missions and influence their people and cultures. NHGRI embraces a growing set of fundamental principles and values that together serve as a guiding compass for genomics – some of these emerged organically, whereas others have been adopted from the broader scientific community.

- Maintain an overarching focus on using genomics to understand biology, to enhance knowledge about disease, and to improve human health.
- Strive for global diversity in all aspects of genomics research, committing to the systematic inclusion of ancestrally diverse and underrepresented individuals in major genomic studies.
- Maximize the utility of genomics for all members of the public, including the ability to access genomics in healthcare.
- Champion a diverse genomics workforce.
- Provide a conceptual research framing that consistently examines the role of both genomic and non-genomic contributors to health and disease.
- Promote robust and consistently applied standards in genomics research.
- Embrace the interdisciplinary and team-oriented nature of genomics research.
- Adhere to the highest expectations and requirements related to open science, responsible data sharing, and rigor and reproducibility in genomics research.
- Pursue advances in genomics as part of a vibrant global community of genomics researchers and funders.



# Priority Research Areas for NHGRI

Realizing the health benefits of genomics will take decades of dedicated work and a global effort that builds on past findings and expands as new productive avenues emerge. NHGRI has a long-term commitment to this promising enterprise through the following five priority research areas.

## **Understanding how the human genome works**

The Human Genome Project delineated the order of the nucleotides in the human genome sequence, but it will take decades of research to reveal all the complexities of how the genome functions. These efforts involve using laboratory and computational approaches to assemble inventories of functional elements in the human genome, establish the choreography by which these elements confer biological function, and catalog the differences among people's genomes. Key to these advances is the ongoing development of new technologies and strategies for studying genome structure and deciphering genome function.

## **Establishing the role of genomic variants in health and disease**

Diseases are a consequence of the complex interplay of influences from our genomes, our environment, and our social context. Rare diseases typically result from the presence of genomic variants (mutations) in a single gene, with environmental and social influences playing a lesser role. Common diseases typically result from the presence of multiple risk-conferring genomic variants in conjunction with important environmental and social influences. Large-scale genomic studies involving diverse participants can establish the role that genomic variants play in rare and common diseases, the response to medications, and the preservation of health.

## **Using genomic information to advance medical care and human health**

Genomic medicine is an emerging medical discipline that involves using genomic information about an individual as part of their clinical care (such as for diagnostic or therapeutic decision-making) and the health outcomes and policy implications of that clinical use. The systematic implementation of genomic medicine relies on research programs that establish tools, resources, and knowledgebases that empower healthcare professionals to use genomic information effectively when delivering clinical care. Another important requirement is the appropriate training of clinicians in genomics so that they can help patients and their families understand the role that genomics plays in making healthcare decisions.

## **Providing infrastructure for secure data analysis and access**

As genomics researchers worldwide generate increasingly large, complex, and diverse datasets, the challenges of managing and empowering others to productively use those data also increase. Addressing those challenges requires the collective expertise of both quantitative scientists and genomics researchers for developing innovative approaches, methods, and technologies. Ultimately, such efforts provide greater access to secure data and computational tools that facilitate genomic studies as well as the implementation of genomic medicine.

## **Addressing the societal impact of genomic advances**

Studying the ethical, legal, and social implications of genomics research has been a cornerstone of the field since its inception. As new technologies increase our capability to generate genomic information and research expands our understanding of what that information might mean, society needs to determine how best to use the technologies and information responsibly. The successive integration of genomics in society depends on ensuring that the research moves forward in ways that thoughtfully consider the implications of the generated knowledge.



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# Genome Structure and Function

# Understanding How the Human Genome Works

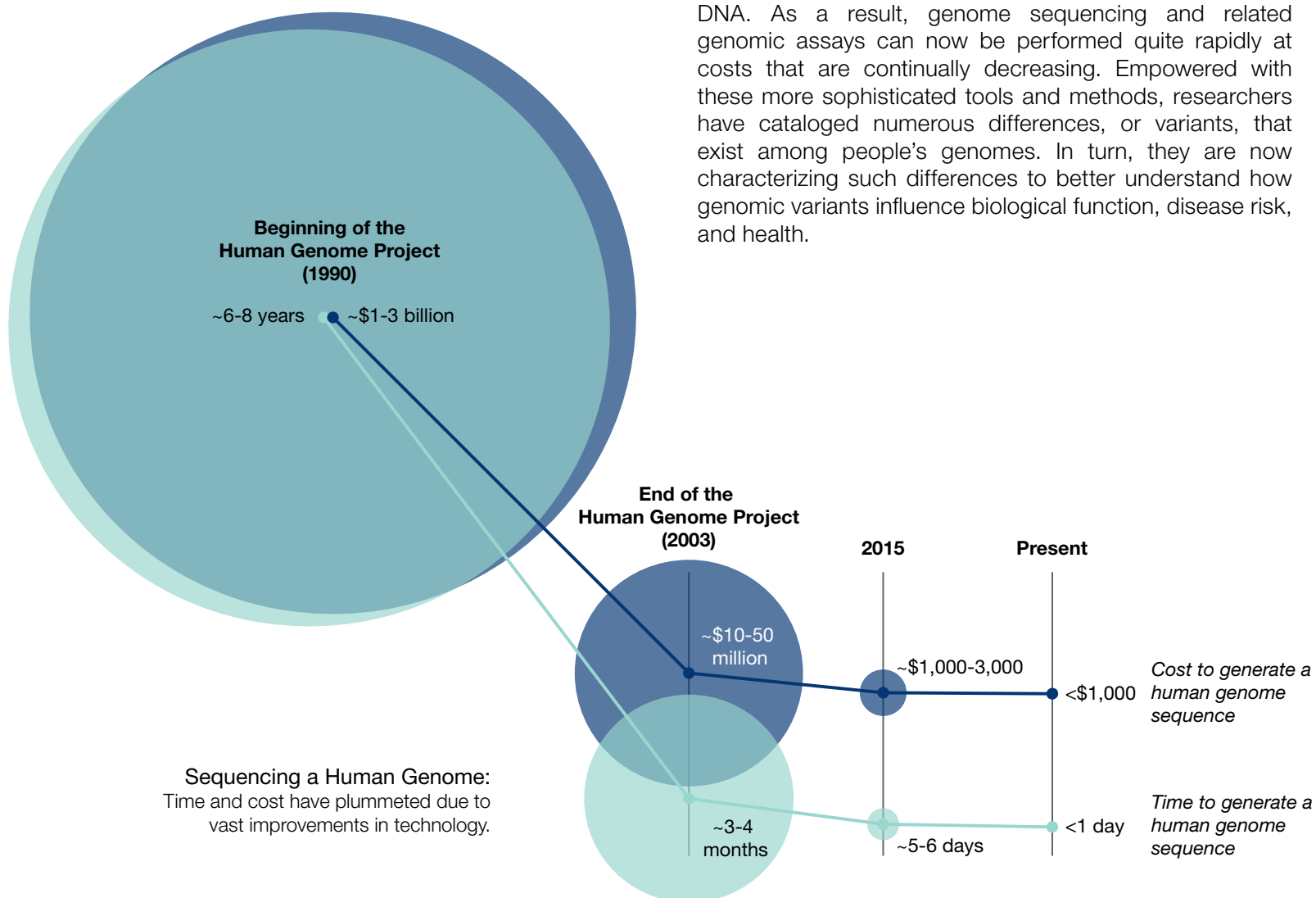
## *Establishing how genomes confer biological information*

When the Human Genome Project ended in 2003, genomics researchers had generated a human genome sequence available for further study but only a limited comprehension of what that sequence meant. NHGRI immediately pivoted to acquire a deeper understanding of genome function and to uncover the genomic underpinnings of human health and disease.

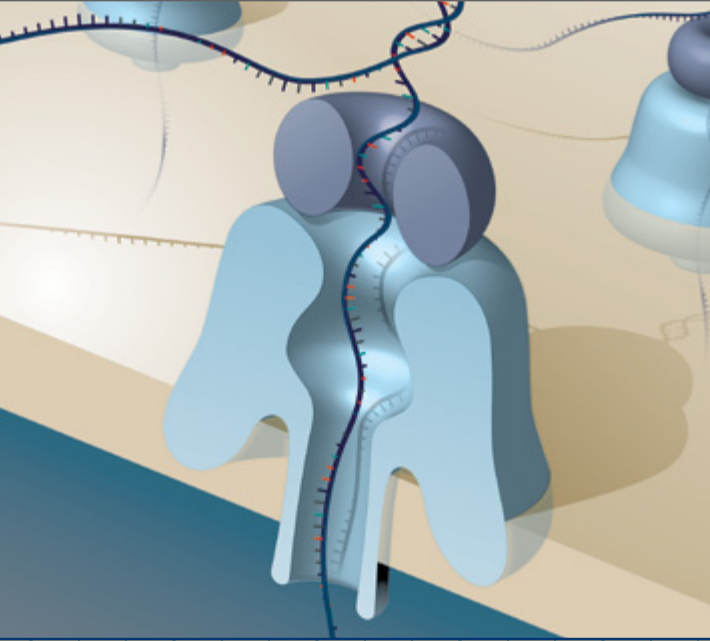
Embedded in the billions of A, T, C, and G nucleotides that make up the human genome is a fundamental yet complicated code for human biology. Interpreting that code requires the application of novel laboratory and computational approaches. Part of that work involves understanding the genomic differences and similarities among an array of organisms – an area known as

comparative genomics. Overall, researchers have made substantial progress in identifying and characterizing the thousands of genes and other functional elements encoded in the human genome. While a complete understanding of human genome function will take decades to achieve, the information collected to date already provides critical insights for scientists and clinicians to use in studying genomic contributions to human health and disease.

The technological innovations that occurred along with the generation of genomic data were integral to the success of the Human Genome Project. The same is true today – genomic advances continue to depend heavily on the development of new technologies. NHGRI programs have fostered the emergence of powerful genomic technologies, including better methods for sequencing and analyzing DNA. As a result, genome sequencing and related genomic assays can now be performed quite rapidly at costs that are continually decreasing. Empowered with these more sophisticated tools and methods, researchers have cataloged numerous differences, or variants, that exist among people's genomes. In turn, they are now characterizing such differences to better understand how genomic variants influence biological function, disease risk, and health.



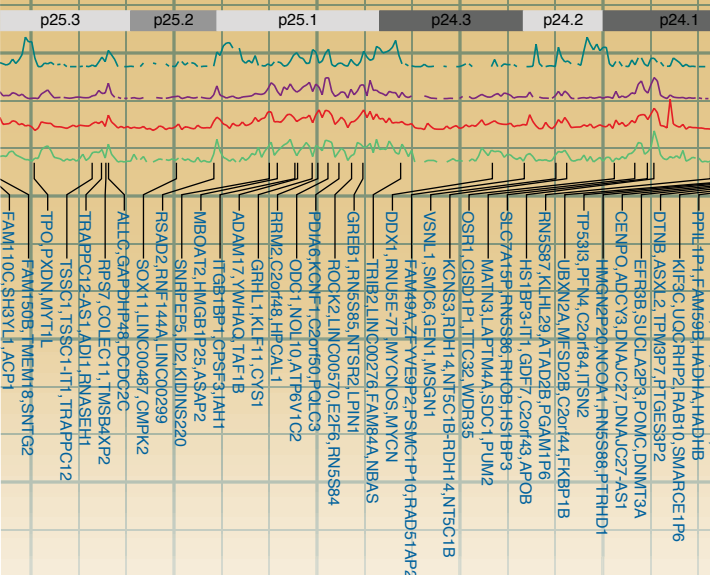
# Genome Structure and Function



## Developing Technologies for DNA Sequencing and Synthesis

Dedicated technology development has always driven genomics forward. After the Human Genome Project was completed in 2003, NHGRI launched a major technology development effort to greatly reduce DNA sequencing costs. Working with and alongside the private sector, NHGRI's "\$1,000 Genome" program was a resounding success. NHGRI continues to foster improvements to DNA sequencing and, in addition, now aims to make similar inroads with synthetic nucleic acid technologies. To that end, the Institute's Genome Technology Program supports the development of cost-effective technologies that will markedly improve the quality and efficiency of nucleic acid synthesis. Progress in this area – for example, the ability to generate large amounts of DNA molecules of any sequence or size – holds transformative potential for biomedical research.

### Chromosome 2



## New Approaches to Study Increasingly Complex Genomic Data

Technological advances are now allowing researchers to generate genomic data in complex ways, including from single cells, at distinct spatial locations within the same tissue, and longitudinally over time. In addition, interdisciplinary and team-science efforts are yielding new ways to collect multiple data types from a single sample. Investigators are finding ways to integrate such data with other information in sophisticated ways to provide insights about genome variation and function in different biological and clinical contexts. The growing scale and multi-dimensional nature of these genomic datasets provide researchers new opportunities to design transformative analytic methods and tools to assimilate, share, visualize, and analyze complex and heterogeneous data types, which is greatly advancing the opportunities for gaining new insights into genome structure and function.

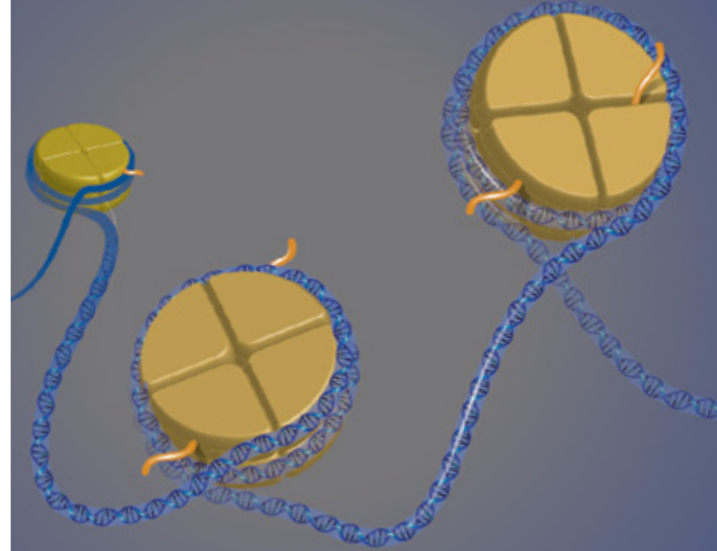
## Establishing a Human Pangenome Reference Sequence

Virtually all researchers who generate human genome sequences use a human genome reference as a comparator to properly align and assemble the primary sequence data. Scientists also rely on this universally accepted reference sequence as a consensus coordinate system for reporting genomic results in a consistent and organized fashion. In partnership with the Wellcome Sanger Institute, NHGRI is supporting the Human Genome Reference Program (HGRP) to generate a high-quality human genome reference sequence that better represents humanity's diversity and to make that "pangenome" reference freely available to the global scientific community. The program is also developing novel computational approaches to visualize the pangenome reference sequence more effectively. A related NHGRI-supported program, the Telomere-to-Telomere (T2T) Consortium, is an open, community-based effort to generate the first truly complete – literally end-to-end (with no missing pieces) – human genome sequence.



## Delineating Gene and Genome Function

Establishing a detailed accounting of all functional sequences in the human genome is essential for understanding the interplay of genomics and human biology, health, and disease. Defining the function of each individual element in the human genome and determining how these elements interact to perform biological processes is a high-priority area of NHGRI research. Many of these studies use cutting-edge technologies, such as CRISPR and other genome-editing tools, to characterize specific genes and gene-regulatory elements, while others use multiplexed methods to examine multiple variants or genes at one time. In some cases, characterizing gene-encoded proteins provides information that helps assign function to specific genes. Ultimately, this accumulating knowledge about genome function will contribute to a broader understanding of fundamental pathways and networks that are central to human biology.



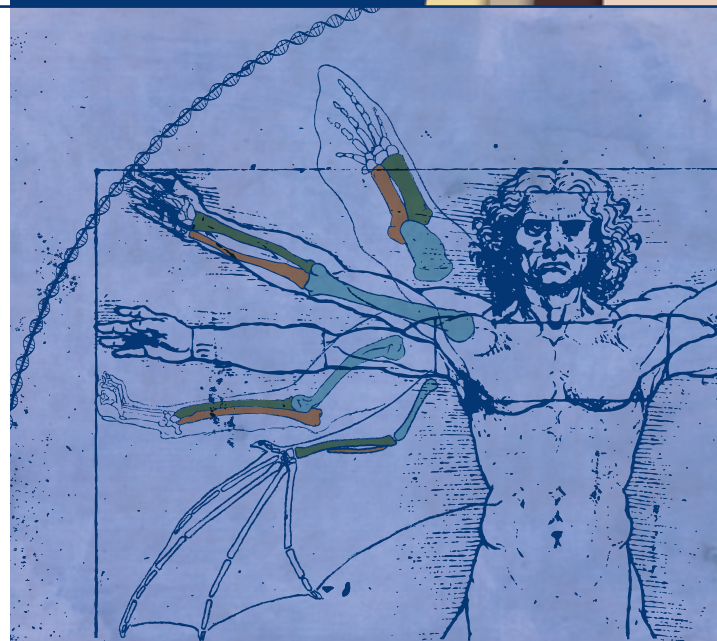
## Understanding How Genomic Variation Affects Genome Function

Scientists have now cataloged hundreds of thousands of variants in the genomes of people from different populations, but they have not yet established which of those genomic variants affect biological function. NHGRI's Impact of Genomic Variation on Function (IGVF) Consortium is applying experimental and computational approaches to examine how genomes function, how genome function shapes phenotypes, and how these processes are influenced by genomic variation. A complementary effort co-led by NHGRI and the Eunice Kennedy Shriver National Institute of Child Health and Human Development, the Developmental Genotype-Tissue Expression (dGTEx) project is studying how genomic variants affect biological function during the neonatal and pediatric periods. This work aims to fill gaps in our understanding about the role of gene expression in human development and provide insights into how processes that occur before maturation influence health later in life.

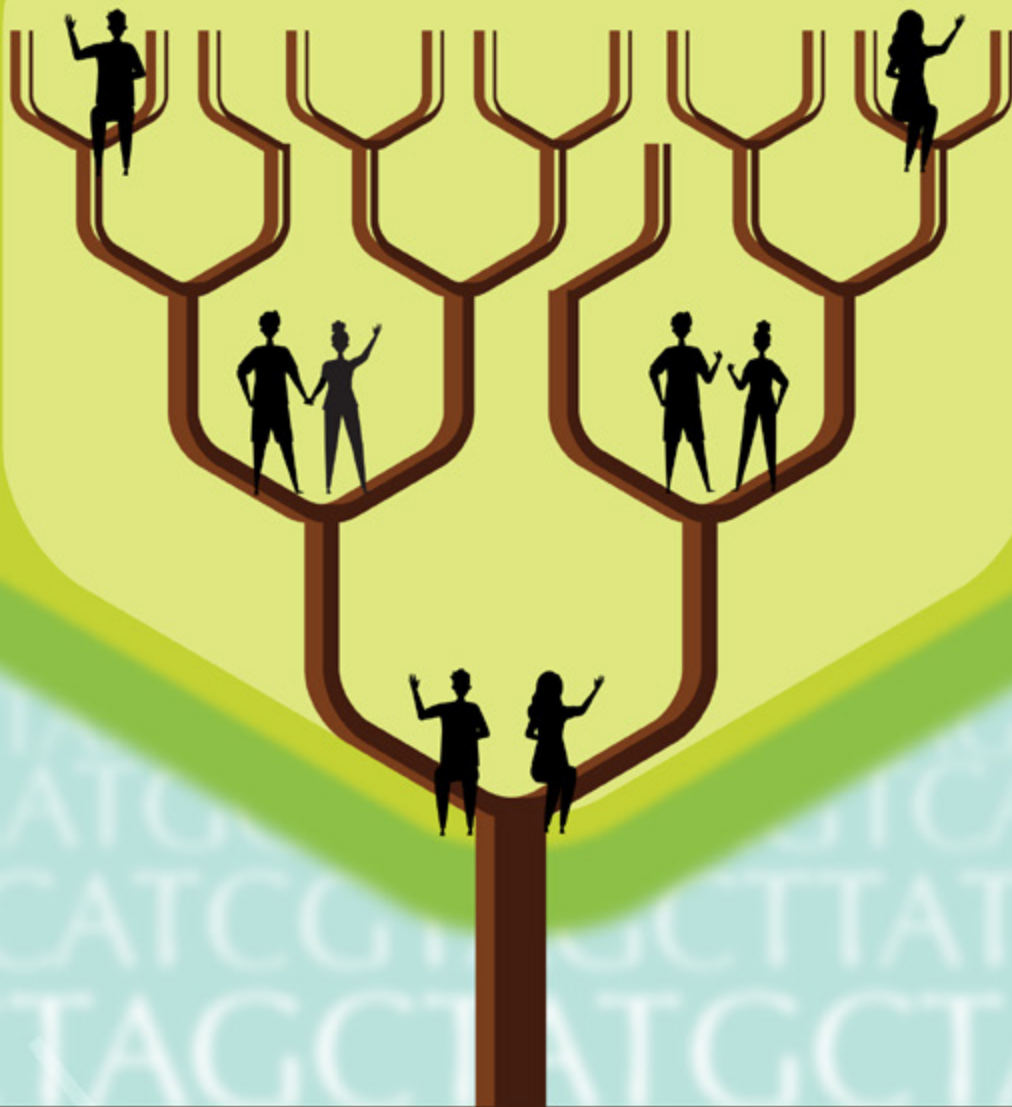


## Leveraging Comparative, Evolutionary, and Population Genomics

Predicting which genomic sequences are relevant to biological function requires researchers to understand genomic variation within and between species, the conservation of genomic regions across species, and evolutionary changes that correlate with specific traits. NHGRI supports research approaches that use model organisms to study genome structure and function and how genomic changes influence biology. This work includes using the tools of comparative genomics and clues provided by evolutionary processes to deduce genome function. NHGRI also supports research efforts that explore the rich genomic diversity of the world's human populations. In addition to providing the benefits of increased representation and equity, including more ancestrally diverse populations in genomics research facilitates the ability to search for disease-associated genes, perform forensic DNA analyses, clarify relationships with our near and distant ancestors, and reveal aspects of human population history.



# Genomics and Human Disease





# Establishing the Role of Genomic Variants in Health and Disease

## *Determining the genomic bases of rare and common diseases*

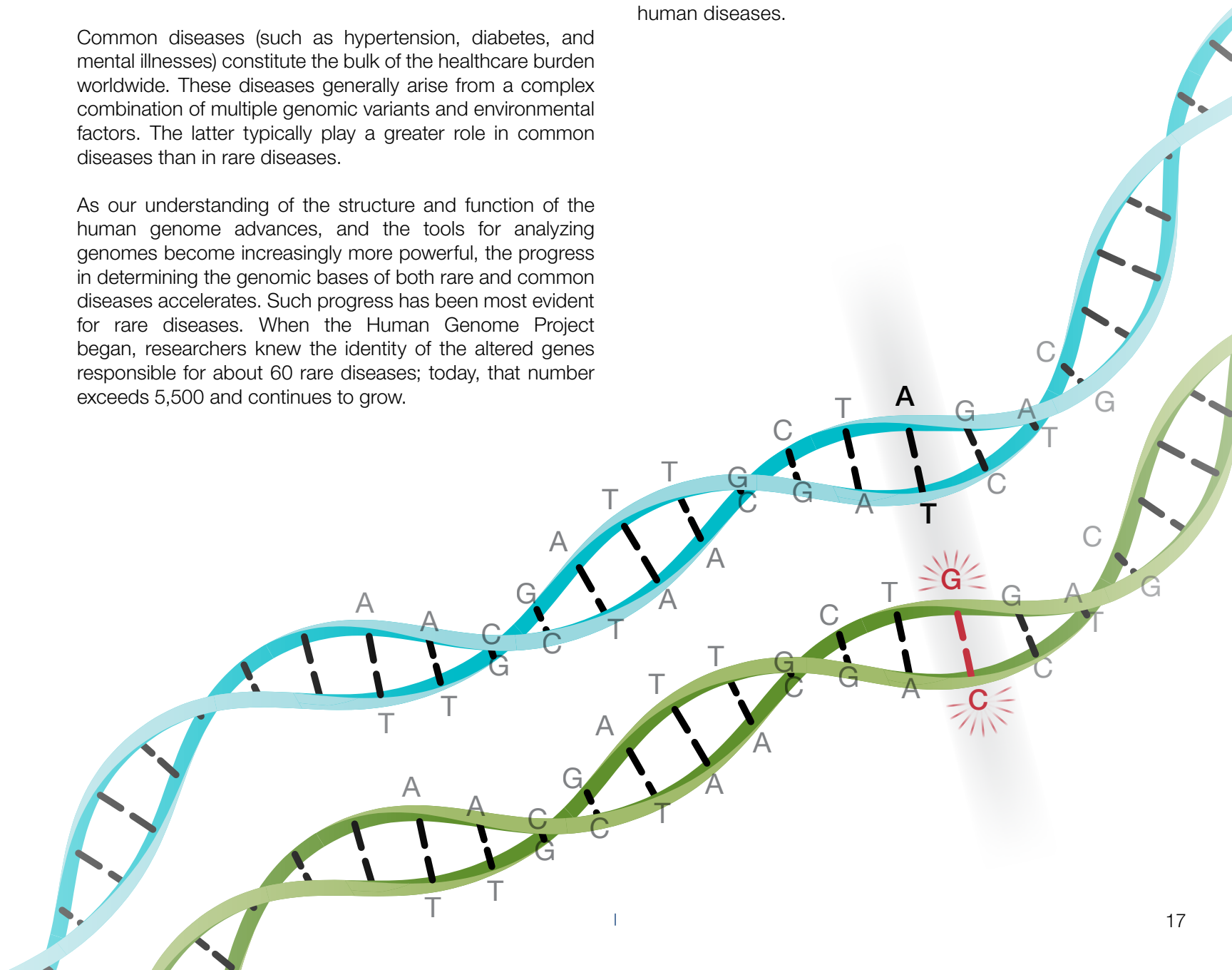
Genomic variants play a central role in essentially all human diseases, but the nature of that role generally differs for rare diseases versus common diseases.

So-called rare diseases include those such as sickle cell anemia, cystic fibrosis, and Huntington's disease, which occur relatively infrequently in the population. However, while individually rare, these disorders in aggregate likely affect more than one in 30 people. Such diseases are most often caused by genomic mutations that disrupt a single gene. While other genomic variants and environmental factors may influence the severity of the disease, the fundamental cause is the altered form of one gene.

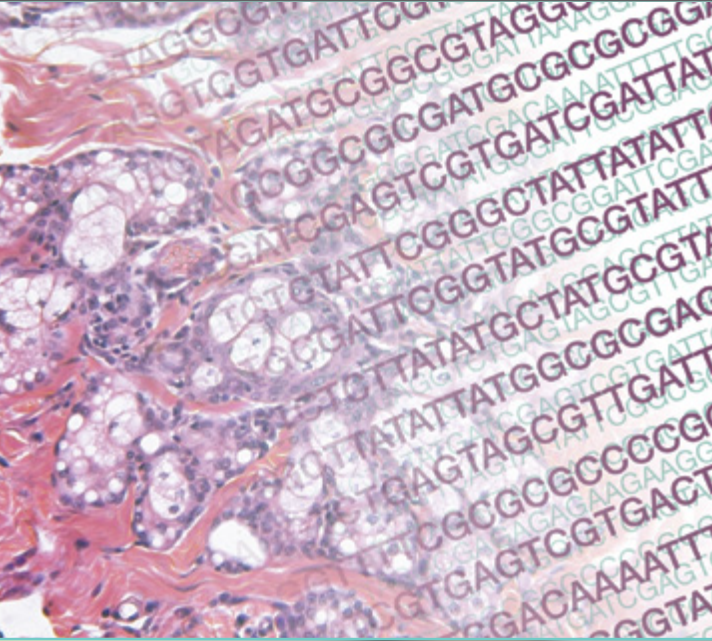
Common diseases (such as hypertension, diabetes, and mental illnesses) constitute the bulk of the healthcare burden worldwide. These diseases generally arise from a complex combination of multiple genomic variants and environmental factors. The latter typically play a greater role in common diseases than in rare diseases.

As our understanding of the structure and function of the human genome advances, and the tools for analyzing genomes become increasingly more powerful, the progress in determining the genomic bases of both rare and common diseases accelerates. Such progress has been most evident for rare diseases. When the Human Genome Project began, researchers knew the identity of the altered genes responsible for about 60 rare diseases; today, that number exceeds 5,500 and continues to grow.

The multifactorial nature of common diseases makes their study more complicated but advances in genomic knowledge and technologies help drive efforts forward. One approach for studying the genomic contributions to common diseases uses a detective-like strategy called a genome-wide association study (GWAS), which involves scanning the genomes of thousands of individuals in search of variants that are associated with an increased risk for disease. Researchers have published roughly 5,000 such studies that together reported over 200,000 associations between a genomic variant and a human disease (or trait), collectively giving critical clues about the location and identity of genomic variants that play a role in common human diseases.



# Genomics and Human Disease



## Interplay of Genomic Variation and Function with Human Disease

Variants in the human genome sequence, along with environmental and lifestyle factors, determine a person's risk for developing certain diseases. By studying genomic variation between individuals and populations, researchers have identified tens of thousands of disease-associated genomic variants. NHGRI's IGVF Consortium focuses on the challenge of establishing which of these variants directly influence human health. This group of researchers applies experimental and computational approaches to examine where and when genes function, how genome function orchestrates human physiology, and how genomic variation influences these processes in a fashion that confers risk for human disease. The IGVF Consortium also ensures that data resources which catalog the association of genomic variants with human diseases are fully operational and readily available to clinicians for use in patient care.



## Rare Diseases

An estimated 25-30 million Americans have a disease that is categorized as rare (most of these disorders affect fewer than 200,000 people, although some affect many more). Of the roughly 7,000-8,000 rare diseases (often referred to as Mendelian disorders) described to date, researchers have determined the underlying genomic cause for more than 5,500 of them. Attention is now being focused on those rare diseases for which the genomic causes have proven to be more difficult to establish. NHGRI's Mendelian Genomics Research Consortium is developing new approaches to interrogate the human genome, looking for novel ways that genomic mutations may be responsible for these remaining rare diseases. This group of researchers also aims to empower their ongoing studies and accelerate the pace of discovery by others through the development of secure data-sharing resources and routines for sharing all generated data.



## Common Diseases

The monumental challenges of defining the genomic contributions to common diseases obstruct progress in the diagnosis and treatment of many human ailments. Researchers can reliably find genomic variants that are statistically associated with a common disease (for example, by GWAS); however, identifying the specific variant(s) conferring disease risk can be a herculean task. Tackling these challenges requires developing new strategic ideas, cutting-edge approaches, and democratized resources that are broadly shared by the international human genomics community. NHGRI is coordinating with global organizations focused on accelerating such efforts, such as the International Common Disease Alliance (ICDA). In this and other work, NHGRI is committed to increasing the diversity of the participants in human genomics research, so that the generated data is more representative of regional and global populations.

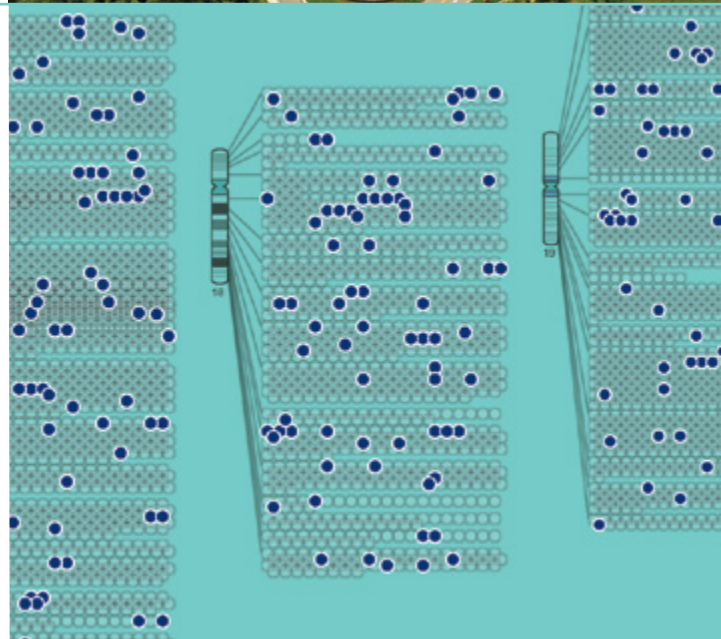
## Genome Sequencing at the NIH Clinical Center

The NIH Clinical Center is America's research hospital – the largest hospital in the world dedicated to clinical research. This facility has become a valuable venue for conducting a number of major clinical research programs that involve human genome sequencing. For example, the NIH Clinical Center pioneered early clinical genomics research studies through the ClinSeq® and Clinical Center Genomics Opportunity programs, which paved the way for other NIH Institutes to launch their own clinical genomics research projects. That momentum continues with the NHGRI-hosted Center for Precision Health Research, which aims to develop next-generation paradigms for conducting precision medicine research, including the integration of large-scale genome-sequence data with electronic medical record information and detailed clinical phenotype data generated at the NIH Clinical Center.



## Genomic Resources for Human Disease Studies

Progress in understanding the genomic influences on human disease requires that research findings are readily shared in a systematic, reliable, and timely manner. NHGRI seeks to amplify the collective power of individual genomic studies through its support of key data and information resources that facilitate such sharing. For example, studies that use PhenX (consensus measures for Phenotypes and eXposures) benefit from the ability to perform cross-study analyses to increase statistical power and to replicate results. The GWAS Catalog is a searchable database used by scientists and clinicians worldwide to identify disease-related genomic variants and understand disease mechanisms. The Genome Aggregation Database (gnomAD) contains aggregated and harmonized genome-sequencing data that are used to determine which genomic variants confer risk for a particular disease. These resources are freely available to the biomedical research and clinical communities.



## Global Genomics

Understanding that the benefits of genomic advances must be realized throughout the world, NHGRI aims to stimulate genomics research on a global level by participating in and supporting the work of international research organizations focused on genomics and human disease. One example is the International Human Epigenome Consortium (IHEC), which provides the research community with access to data from comprehensive epigenomic analyses of healthy and abnormal cells. Another international organization, the International HundredK+ Cohorts Consortium (IHCC), aims to connect large human cohort studies that researchers perform in different countries to encourage data sharing, improve efficiencies, and address scientific questions that none could answer alone. Such global coordination and cooperation help both to increase the diversity of the genomic data available for study and to make those data more accessible to investigators worldwide.





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## Genomic Medicine

# Using Genomic Information to Advance Medical Care and Human Health

## *Applying genomics to clinical care and improving the effectiveness of healthcare*

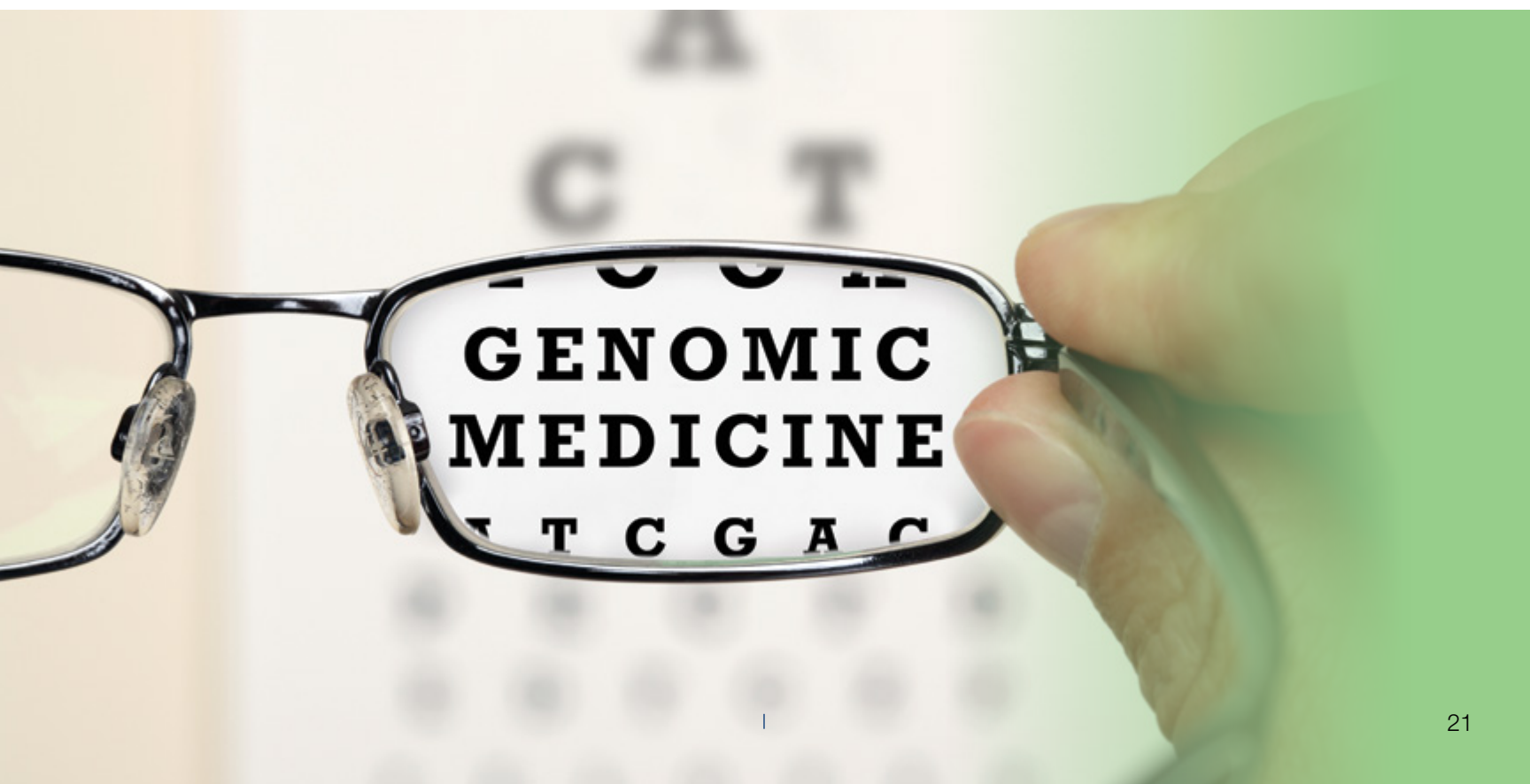
Early examples of the exciting potential of genomic medicine are emerging. By using genomic technologies and investigative findings, clinical care can be tailored to an individual's genomics-related predispositions to disease and disability. Genomic applications already deployed into routine clinical practice include using specific tumor mutations to guide cancer treatment and genomics-based drug hypersensitivity testing associated with treating HIV/AIDS. These examples provide added enthusiasm for additional research to capitalize on genomic discoveries and to help realize genomic medicine's full potential.

To establish a strong foundation for implementing genomic medicine, NHGRI invests in multiple research efforts. Current studies evaluate the impact of using genomic information in clinical care, operationalize genome analyses as a diagnostic tool, and assess the ability to generalize genomic findings across populations and clinical settings.

Some of these NHGRI genomic medicine programs are prototypes for broader initiatives which aim to individualize medical care that advances human health, such as the NIH *All of Us* Research Program, a long-term research effort

launched in 2016. In this way, NHGRI is not alone; other NIH Institutes and Centers, government agencies, funding organizations, not-for-profit groups, and the private sector are actively working to advance genomic medicine.

The journey to improved medical care and human health through effective application of genomics also requires other important advances. Healthcare professionals need effective, accessible tools and systems so that they can use genomic information for clinical decision-making; they also require training and education to use these resources effectively. Patients need a basic understanding of genomics to communicate with their healthcare providers and their own families so that they can make informed decisions about their medical care and other important life choices. Finally, the broader medical system needs to adapt to the ongoing genomic advances so that clinical benefits are rapidly and fairly made available to all people.



# Genomic Medicine



## Rare and Undiagnosed Diseases

Undiagnosed diseases are conditions that even skilled physicians struggle to diagnose. The disease may be one that is rarely seen or that has never been described. Arriving at a diagnosis for a rare or unknown condition can be a long, frustrating process for patients and their families, often involving years of testing and multiple doctors. NHGRI supports collaborative research projects that aim to systematically improve the evaluation of patients with rare and undiagnosed diseases. With an emphasis on cutting-edge genomic studies combined with rigorous clinical evaluations, these efforts aim to define the biological mechanisms that underlie these diseases and to identify best practices to diagnose and manage them. A long-term objective is to have the clinical evaluation of undiagnosed diseases become routine in medical practice.



## Implementing Genomic Medicine in Clinical Care

Recognizing the potential for using genomics as part of clinical care, NHGRI supports studies to determine optimal processes for integrating genomics into clinical workflows. The Clinical Sequencing Evidence-generating Research (CSER) Consortium aims to assess the challenges and opportunities of using genome sequencing in the medical care of patients. The desire to better understand the interactions among patients, families, providers, and laboratories with respect to genomic information, particularly among medically underserved populations, drives CSER to examine a range of scientific questions in diverse and underrepresented populations. The consortium is evaluating the use of genomic information for clinical management and follow-up care; it is also tailoring processes for interpreting genomic variants and disclosing genomic test results. Some of these areas are particularly well-suited for investigation in robustly designed clinical trials.



## Generating Evidence for Implementing Genomic Medicine

NHGRI's Implementing Genomics in Practice (IGNITE) Pragmatic Clinical Trials Network, which consists of multi-site clinical groups that involve diverse settings and populations, are conducting clinical trials to study the utility and cost-effectiveness of implementing genomic medicine in different situations. These efforts include assessing approaches for the real-world application of genomic medicine, with the results yielding generalizable knowledge and methods for conducting clinical trials that examine the use of genomics in clinical care. Ultimately, IGNITE aims to advance the implementation of genomic medicine in routine healthcare by providing evidence that demonstrates improved patient outcomes from the use of genomic information; in turn, these efforts also involve disseminating the resulting methods and best practices beyond the network to facilitate a broader uptake of genomics in clinical care.

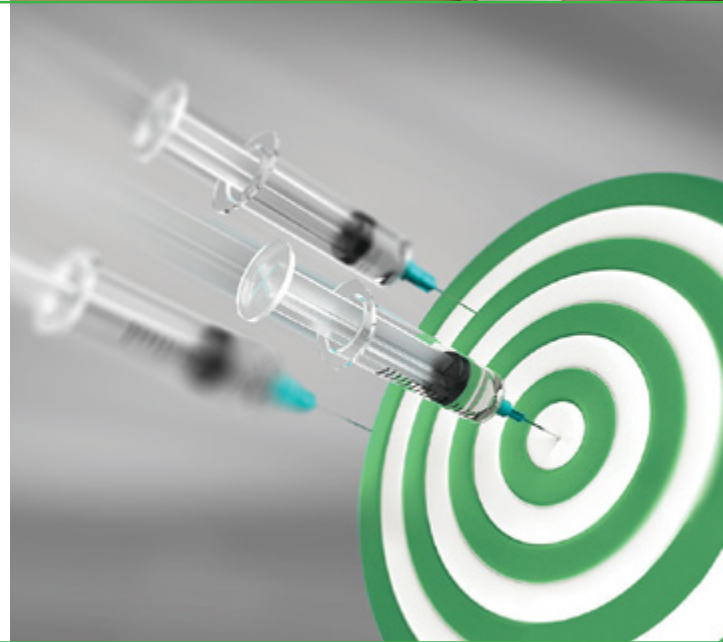
## Readying Healthcare Professionals for Genomics

For genomics to be adopted into routine medical practice, clinicians must be able to interpret their patients' genomic information as part of medical decision-making. NHGRI supports several endeavors aimed at preparing frontline healthcare professionals for this process, which is key for implementing genomic medicine. The Clinical Genome Resource (ClinGen) is developing a consensus approach for identifying clinically relevant genomic variants, with the resulting information being made available to clinicians. NHGRI also promotes genomic literacy among healthcare professionals through a vetted repository of genomics education resources and a series of simulated clinical case studies in genomic medicine (both are available on genome.gov). Finally, NHGRI co-leads the Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG), which facilitates interactions between NIH and professional medical societies and creates genomics educational resources for use by non-genetics medical professionals.



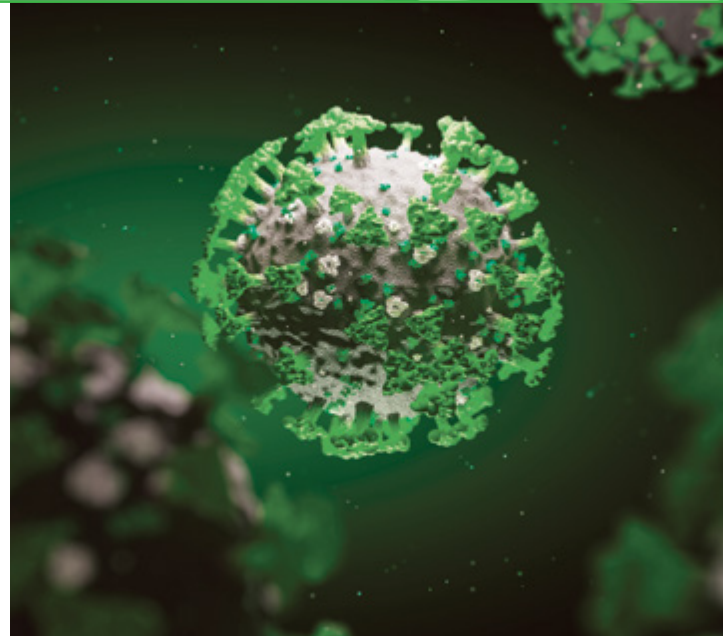
## Using Genomic Information to Estimate Disease Risk

Studies that examine large cohorts of people along with their electronic medical records can be used to refine approaches for genomic medicine and precision health. The NHGRI-funded Electronic Medical Records and Genomics (eMERGE) network has been advancing work in this area since the program's inception in 2007. eMERGE investigators have made major contributions in developing, testing, and implementing methods that integrate genomic information into electronic medical records for use in clinical care. These researchers are now focused on combining clinical data, family health history, and genomic information in an effort to improve estimates of disease risk and, in turn, to facilitate the development of more precise prevention strategies. Importantly, the work of the eMERGE network involves both estimating disease risk for individuals from diverse populations and then measuring the impact of such knowledge on patient health over time.



## Changing Face of Infectious Disease Genomics

Advanced genome-sequencing and sequence-analysis methods have created exciting opportunities in clinical microbiology, especially related to public health and hospital epidemiology. The ability to rapidly sequence microbial DNA from clinical samples has transformed infectious disease diagnostics and mainstreamed systematic genomic surveillance of infectious agents. Researchers can now determine how infections spread among patients and healthcare workers, identify new infection-causing microbes, characterize the genetics underlying antibiotic resistance, and support vaccine development. Epidemiologists can rapidly trace the origins of food-borne illnesses and compare the genomes of infectious microbes from different patients. Scientists also use genome sequencing to identify features of bacteria and viruses that influence the nature of the infections they cause. The adoption of genomic approaches in response to global pandemics illustrates the integral and vital nature of genomics in modern research and medicine.



The background features a complex network of interconnected nodes and lines, overlaid with various DNA sequences (e.g., GATCG, TAGCT, GAGAG, TATAT) in shades of blue and green. The overall color palette is warm, with orange and yellow tones. A solid orange rectangular box is positioned in the upper right quadrant, containing the main title.

# Genomic Data Science



# Facilitating the Management and Analysis of Large Amounts of Genomic Data

## *Providing a robust infrastructure for secure genomic data analysis and access*

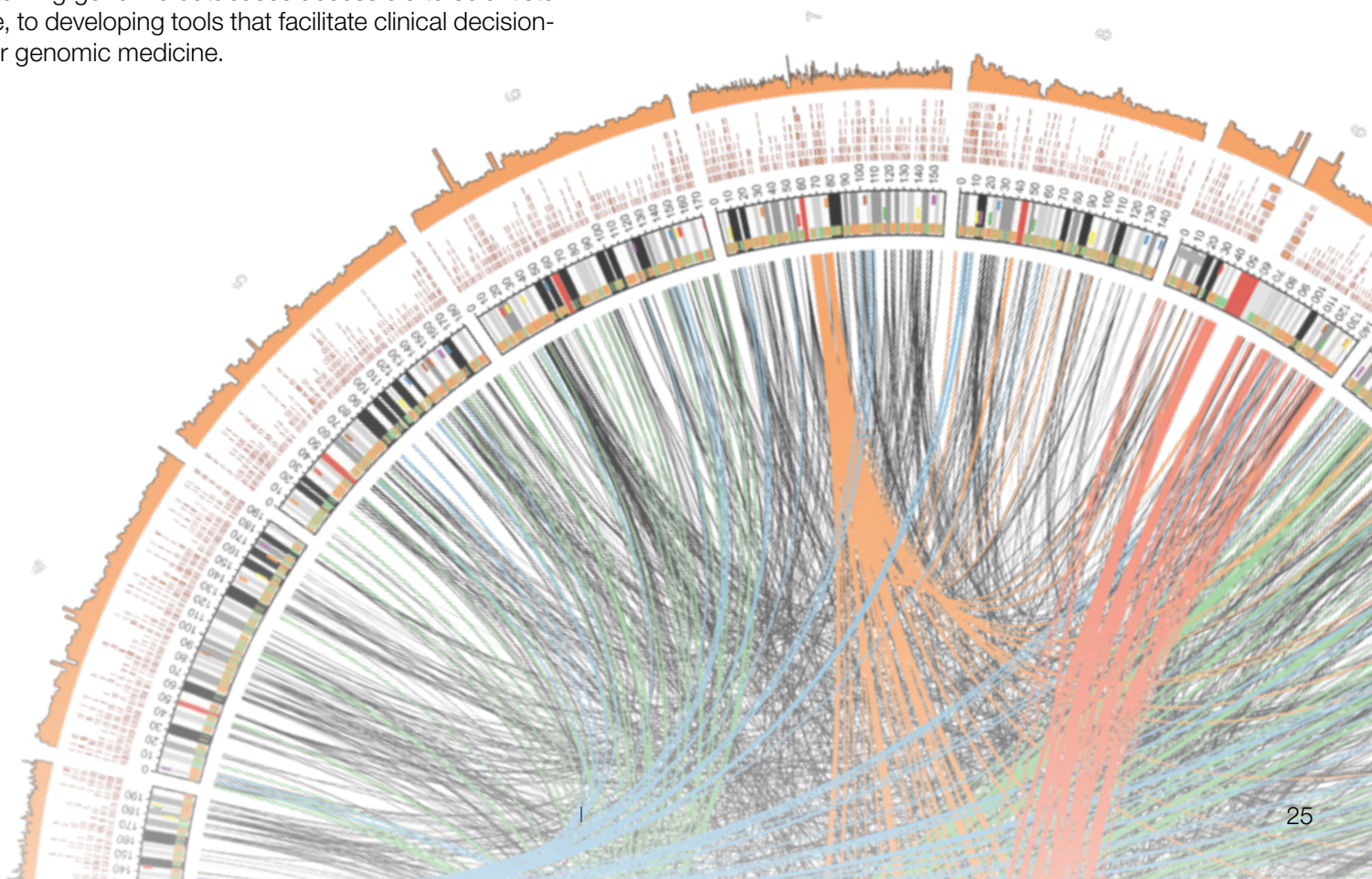
Genomic technologies now readily generate massive amounts of data – often terabytes to petabytes from a single study. In fact, genomics researchers routinely produce far more data than they can analyze in real time. While a human genome can now be sequenced in a day, analyzing the resulting data to obtain a comprehensive and meaningful interpretation remains a lengthy and laborious task. Outputs from other genomic technologies present similar challenges in terms of data processing and analysis.

Data science as it relates to genomics (that is, genomic data science) involves the study of increasingly large, complex, and diverse datasets. These data arise from various types of research projects – for example, population studies examining the role of genomic variants in common diseases or studies characterizing the functional elements in the human genome. As such genomic data science requires expertise in quantitative scientific disciplines, such as bioinformatics, computational biology, biostatistics, and clinical informatics.

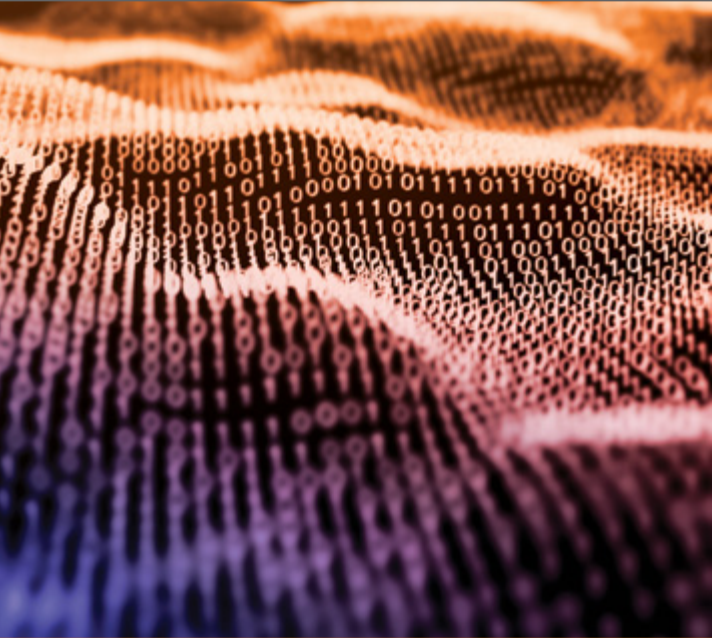
The portfolio of NHGRI-supported research includes many projects and programs that focus on genomic data science. These efforts range from developing computational tools and methods for analyzing genomic data, to establishing and maintaining genomic databases accessible to scientists worldwide, to developing tools that facilitate clinical decision-making for genomic medicine.

Issues related to data security, access, and privacy are central to many aspects of genomic data science. The growing focus on understanding the role of genomic variation in human disease has prompted major research initiatives aimed at cataloging human genomic variants and determining their biological and clinical consequences. Such work requires maintaining data security and privacy while also providing data access to appropriate researchers worldwide, a delicate balancing act that requires ongoing attention as well as both technical and policy expertise.

Through multiple initiatives, NHGRI works to build a workforce of data scientists trained in genomics as well as a cadre of genomics scientists and clinicians empowered to use cutting-edge data science resources and tools for genomic research and genomic medicine.



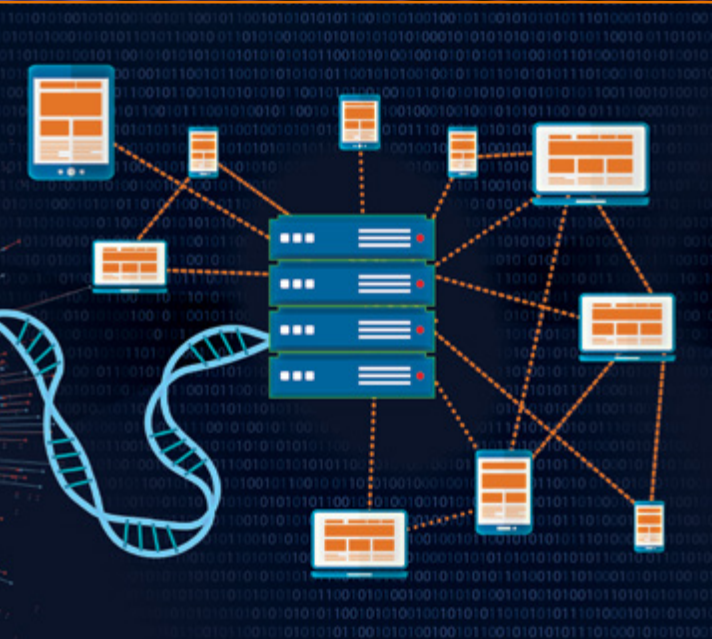
# Genomic Data Science



## Tools and Methods for Genomic Data Science

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The volume and complexity of genomic data that researchers now generate are continually increasing at a breathtaking pace, which presents a prototypic big data challenge that often impedes progress in genomics research. This situation requires the ongoing development of new tools and methods to assimilate, analyze, visualize, and share terabytes of data in a form and format that allow new knowledge to be extracted by researchers. NHGRI supports many projects aimed at developing innovative data science tools and methods, including those that use artificial intelligence and machine learning approaches, for a wide array of genomic applications. Such efforts require enlisting investigators with expertise in quantitative scientific disciplines, including biostatistics and computer science, to help develop appropriate solutions for the data-related obstacles encountered in genomics.



## Sharing Genomic Data

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Genomic data have the greatest impact when made broadly available to the scientific community, well beyond the researchers who originally generated them. Often, such data are then used in productive ways beyond that originally envisioned at the time of generation, thereby amplifying their value. Successful data sharing requires both practical policies that account for the needs of data generators and users alike as well as effective computational infrastructures to facilitate data access in an appropriately secure environment. These efforts must offer appropriate privacy protections to the individuals who provide samples for the research studies. NHGRI is a leader in genomic data sharing across the full range of activities, from policy development and implementation to the design of computational systems that operationalize broad data sharing.



## Facilitating International Access to Genomic Data

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Genomic data are continually generated by scientists and clinicians around the world. The research enterprise benefits enormously by the global sharing of these data, but this requires accounting for privacy laws of different countries and myriad technical hurdles. NHGRI is extensively involved in establishing policies and technical capabilities that allow international researchers to access shared genomic data, regardless of the data's origin. NHGRI supports a number of international groups who are enabling international access to genomic data, including the Global Alliance for Genomics and Health (GA4GH), which creates standards and frameworks to enable genomic and health-related data sharing; Global Biodata Coalition (GBC), which works to sustain the key data resources used by biomedical researchers worldwide; and H3ABioNet, which focuses on developing a Pan-African informatics infrastructure to support genomics research on the African continent.

## Community Data Resources

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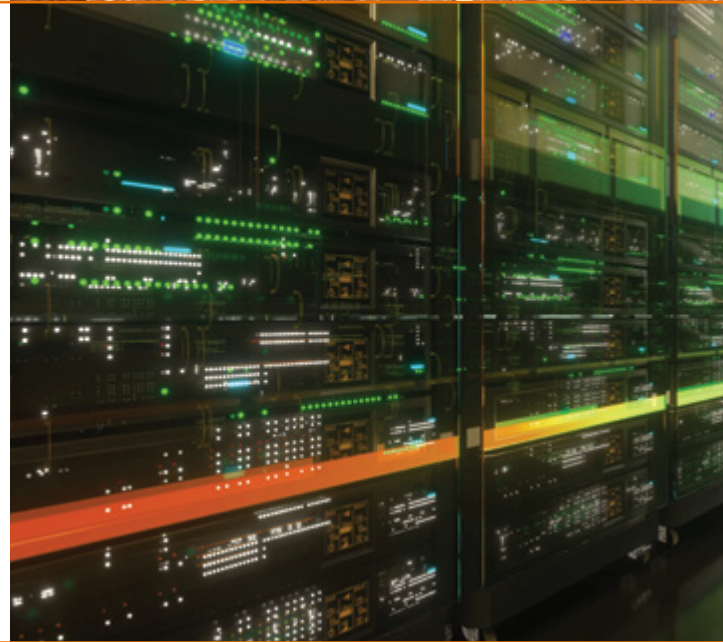
Large-scale analyses of genomic data rely extensively on freely accessible data resources that are well-organized, secure, and user-friendly. NHGRI is at the forefront of developing and sustaining genomic data resources for use by the broader scientific community. Prominent examples include data repositories, such as the Model Organism Databases and UniProt, and informatics platforms, such as the Genomic Data Science Analysis, Visualization, and Informatics Lab-space (AnVIL). The design of these data resources emphasizes interoperability and open, web-based access, including through an NIH-wide cloud-based effort, to facilitate their broad use by researchers with diverse data science skills. NHGRI is committed to the long-term sustainability of these resources and will continue to support their development and maturation, thereby benefiting the broader research community.



## Coordinating Genomic Data at Scale

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NHGRI-supported research projects have ever-growing capacities for generating and analyzing genomic data. Such efforts continually make influential contributions to understanding genome structure and function, the genomics of human disease, and how to implement genomic medicine. In many cases, NHGRI organizes these studies as part of large overarching programs designed to operate at considerable scale, with each subproject capable of generating prodigious amounts of genomic data in a relatively complete, rapid, and cost-effective fashion. An inherent (but exciting) consequence of such scale is the challenge of then handling, organizing, analyzing, and sharing the resulting massive genomic datasets. Increasingly, researchers require robust approaches for coordinating such large-scale data generation and analysis, including the development and dissemination of data and meta-data standards, to maximize the broader value of the genomic data.



## Data Science for Genomic Medicine

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Healthcare professionals are increasingly able to access genomic information about their patients that can be used to make treatment and management decisions. The effective use of that information requires a number of data science elements, which are largely still being developed and refined. Protecting patient privacy is paramount, requiring a secure data infrastructure that allows clinicians to access and analyze a patient's genomic data. Similarly, clinicians rely on the availability of cutting-edge and user-friendly decision-support tools that provide guidance about the clinical relevance of genomic variants in conjunction with other clinical information. These tools are essential for simplifying how genomic information is incorporated into the clinical workflow. NHGRI continues to provide pivotal leadership in this area by supporting the development of effective solutions, thereby facilitating the implementation of genomic medicine.





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# Genomics and Society

# Addressing the Societal Impact of Genomic Advances

## *Promoting the ethical and equitable integration of genomics into society*

The founders of the Human Genome Project recognized that the information gained from mapping and sequencing the human genome would have profound implications for society. How would the new genomic knowledge be interpreted and used? Who should have access to an individual's genomic information, and how can that person be protected from its improper disclosure or use? Such questions gave rise to NHGRI's ELSI Research Program.

For over three decades, the ELSI Research Program has worked to understand, anticipate, and address the impact of genomic advances for individuals and society; stimulate public discussion of relevant genomic issues; and help develop policy options that ensure the information is used for societal benefits.

NHGRI is committed to examining the societal issues associated with large biomedical research projects. ELSI research components are therefore embedded within many major NHGRI-led projects to explore important issues, such as how to recruit and include diverse populations in biomedical research and how to work with communities to disseminate information about research in a culturally responsive manner.

Such efforts involve ensuring transparent communication between researchers and study participants to promote a clear understanding of two important issues: the participants' role in research and the protections in place to safeguard their interests. NHGRI staff have emerged as leading consultants about genomics and society because of their experience in ELSI research.

To enhance broader societal knowledge about genomics and its applications for improving personal and family health, NHGRI also leads several high-profile genomic educational activities aimed at students, educators, healthcare professionals, and the public. These efforts seek to improve genomic literacy in many different communities.



# Genomics and Society



## Transdisciplinary ELSI Studies

Many of the studies funded by the NHGRI ELSI Research Program cross disciplines to focus on problems of broad relevance to genomics researchers and the public alike, including the conduct of genomics research and the translation of research findings into healthcare. The NHGRI Centers of Excellence in ELSI Research Program supports centers of transdisciplinary research and training that promote the translation of ELSI scholarship into practices and policies that guide genomics research and genomic medicine. To facilitate ELSI research more broadly, the NHGRI-funded Center for ELSI Resources and Analysis (CERA) provides an established platform for sharing ELSI research tools and methods; curating, synthesizing, and distributing important research findings; and convening ELSI investigators to encourage transdisciplinary interactions and build a more synergistic research community.



## Informing Public Policy

NHGRI helps inform policies surrounding human subjects research, the incorporation of genomics into healthcare, and the sharing of genomic data in a manner that enables scientific discovery while protecting research participant privacy. NHGRI played a significant role in the development and eventual passage of the Genetic Information Nondiscrimination Act (GINA) of 2008. The Institute also helped shape policies on the patenting and licensing of intellectual property based on genomic discoveries. NHGRI collaborates with other federal partners, including the Federal Trade Commission (to examine advertising for products claiming a genomic basis for their use) as well as the Food and Drug Administration and the Centers for Medicare and Medicaid Services (to inform the oversight of genomic tests and targeted genomic-based therapies and how they are reimbursed by health insurers).



## Engaging Students and Educators in Genomics

National DNA Day, held each April, commemorates the reporting of DNA's double-helical structure in 1953 and the completion of the Human Genome Project in 2003. This annual celebration offers students, teachers, and the public exciting opportunities to learn about the latest advances in genomics and explore what these advances may mean for their lives. Each year, NHGRI partners with professional societies and advocacy groups to promote DNA Day, and it supports DNA Day activities at schools, museums, libraries, and community centers around the world. The annual NHGRI Short Course in Genomics offers science, technology, engineering, and mathematics (STEM) educators the opportunity to hear lectures and receive teaching resources from leading NIH researchers, clinicians, and staff. The course is designed to provide participants appropriate background information and materials to bring genomics-oriented content into their classrooms across a wide range of topics.

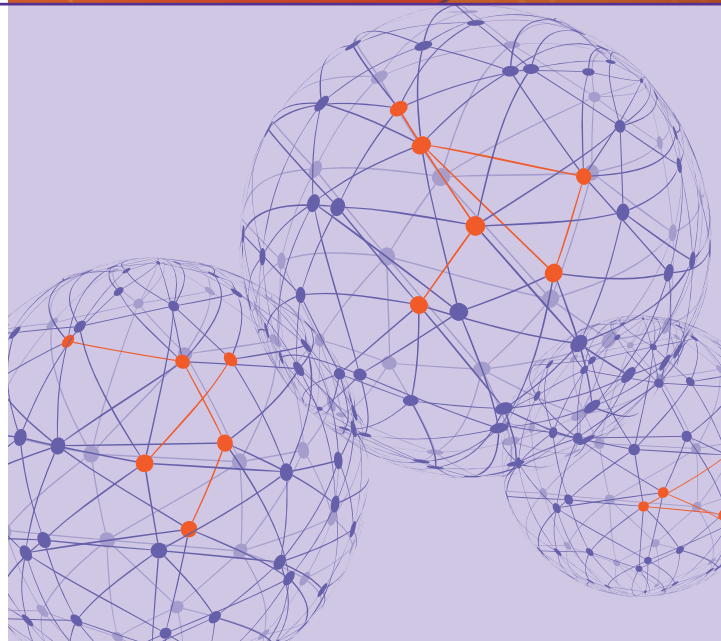
## Improving Public Genomic Literacy

NHGRI is helping to prepare the public for the widespread use of genomic information as a routine part of medical care and other aspects of life. Through community engagement activities, workshops, and forums, Institute staff work with diverse communities to enhance genomic literacy. Since 2013, NHGRI has partnered with the Smithsonian's National Museum of Natural History on the exhibition "Genome: Unlocking Life's Code." More than 3.6 million people visited the exhibition at the museum in 2013-2014, and many more experienced it during a subsequent tour across North America. The exhibition shows the revolutionary nature of genomics, its impact on fields as varied as biodiversity and medicine, and the challenges it presents to modern society. NHGRI also partnered with the Smithsonian to develop a series of free educational programs, including a Genome "Do It Yourself" (DIY) toolkit and app.



## Embedding ELSI Research

NHGRI supports an increasing number of ELSI research projects that are strategically embedded within larger programs. For these studies, behavioral and social scientists, legal scholars, and bioethicists work closely with other genomics researchers to identify and address ethical, legal, and societal issues as they arise and to anticipate potential future challenges. These collaborative ELSI research projects represent vital components of several large NHGRI initiatives, including efforts to catalog human genomic variation, establish a pangenome human reference sequence, incorporate genomic information into electronic medical records, and implement genomic medicine. Directly embedding ELSI research studies within larger genomics projects provides a proactive and mutually beneficial means to ensure that the overall work products address important genomics and societal issues of the day.



## Genomics and Health Disparities

NHGRI is committed to supporting research that increases our understanding of health disparities as they relate to advances in genomics. Ongoing efforts include studying the genomic basis of diseases and conditions that contribute to health disparities and examining the barriers to ensuring that genomic medicine is available to all patient groups. The Institute seeks to advance research to define relevant gene-environment interactions, including identifying inherited susceptibility factors for disease and developing technologies to reliably measure relevant environmental contributions. NHGRI is also committed to expanding the inclusion of research participants from diverse ancestral backgrounds in large genomic studies, aiming to broaden and deepen knowledge about the role of genomic variation in human health and to provide genomics-based benefits to all human populations.



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# Training and Workforce Development in Genomics

## *Developing a strong and diverse workforce for genomics research and genomic medicine*

NHGRI's mission includes cultivating a strong and diverse pool of future researchers, healthcare providers, and educators with appropriate expertise in genomics. The Institute's support of training and professional development resulted in a workforce that was prepared for completing the Human Genome Project and for the genomics programs that have followed. To meet the needs of the continually growing field of genomics, NHGRI regularly adapts its training and workforce development programs. These efforts recently broadened to incorporate an emphasis on genomic medicine and genomic data science; the latter includes the training of data scientists in genomics and, reciprocally, genomics researchers in data science.

NHGRI supports genomics training in multiple ways: grants to teaching institutions, fellowships to individual trainees, loan repayment, internships, courses, and online resources, among others. These training opportunities span the Institute's Extramural Research Program and Intramural Research Program.

The NHGRI Extramural Research Program offers support to academic institutions for pre- and postdoctoral trainees and for researchers who are already established in their careers. These training programs are designed to develop a diverse pool of trainees with expertise in broad areas (such as genome structure and function, genomic data science, genomic medicine, and the societal implications of genomics). NHGRI also supports early career scientists in genomics through programs such as the Genomic Innovator Awards, thereby helping to ensure that the growing workforce is poised to build upon new advances in the future.

The NHGRI Intramural Research Program offers a diverse array of training programs. Pre- and postdoctoral trainees can conduct research in the laboratories of NHGRI investigators. In addition, the Institute offers specific fellowship opportunities in areas such as medical genetics, medical biochemical genetics, genetic counseling, health disparities, education, and policy. NHGRI also conducts short-term summer programs in genomics for students, teachers, and faculty from around the world.

All NHGRI training programs include a core commitment to maximize the diversity of the genomics workforce. The Institute supports training and career development programs to help recruit underrepresented investigators into genomics research and ensure their paths to success. These efforts include creating networks that provide resources and mentors to help diverse students and faculty advance through their careers and become independent genomics researchers. Toward that end, NHGRI is developing research partnerships with minority-serving institutions to enhance their technical capabilities for conducting genomics research, building relationships with individuals from underserved communities and minority organizations to increase awareness of their genomics research-related concerns, and providing opportunities for underserved students and researchers to pursue careers in genomics.



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# Future Horizons

## *NHGRI at The Forefront of Genomics*

Over the last three-plus decades, genomics has progressed from a just-named, emerging discipline led by a small community of trailblazing researchers to a vital component of biomedical research. Genomics has rapidly become profoundly important for most areas of biological inquiry and is now poised to become a fundamental part of medical care.

With this transition, the field has gained relevance to a much broader audience. Once only relevant to biomedical researchers and, later, healthcare professionals, genomics is increasingly becoming important to patients (and friends and relatives of patients) – which means all of us. Indeed, the words “genome” and “genomics” are now regularly seen in the popular press, heard on the radio, mentioned on television, and featured on social media. In short, genomics is now highly relevant to society more broadly.

Since its inception, NHGRI has been a world leader in genomics research, training, education, and outreach. While the Institute’s origins were deeply rooted in the highly focused goals of the Human Genome

Project, its mission and corresponding programs have grown in parallel with the expanding relevance of genomics; these now touch the entire landscape of biomedical research – from basic science to clinical implementation.

Looking toward the future horizons, the scope and relevance of genomics will continue to grow. Genomics will catalyze important scientific and medical advances on a regular basis, bringing new opportunities along with new challenges. NHGRI stands at The Forefront of Genomics, ready to pursue those opportunities and to address the associated challenges through its research programs, engagement efforts, and outreach initiatives.

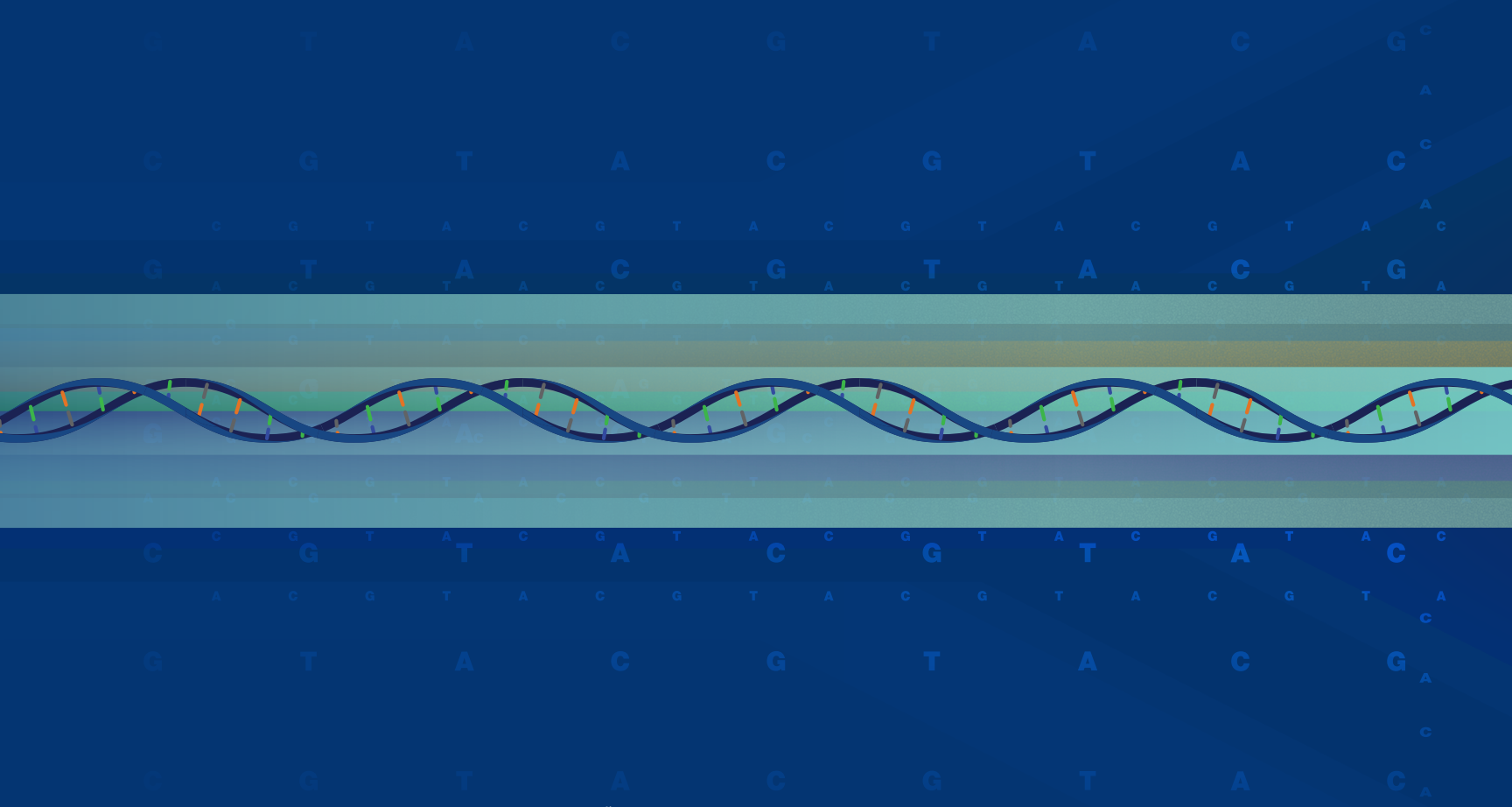
At the launch of the Human Genome Project, the full potential of genomics was hard to fathom. Three decades later, progress in genomics – the research achievements, technological developments, and societal integration – has been nothing short of spectacular, bolstering NHGRI’s fundamental and core belief that genomics offers great promise and hope for improving the human condition.

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**The Forefront  
of Genomics®**

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2021

[genome.gov](http://genome.gov)