ASHG

American Society of Human Genetics

HUMAN GENETIC & GENOMIC RESEARCH

Public Funding | Public Benefit

Sustained investment in the National Institutes of Health → Groundbreaking genetic and genomic discoveries → Revolutionizing medicine and saving lives

RESEARCH ADVANCES INCLUDE

- **Next-generation sequencing techniques for earlier and more accurate detection and diagnosis of genetic conditions.**
- **New gene therapy treatments that are delivering hope to millions.**
- **Improved access of individuals to genomic medicine.**

ASHG acknowledges and values Congress' commitment to biomedical research and urges continued robust support to deliver new knowledge, treatments, answers, and hope for people everywhere.

TIMELINE

1989 U.S. Department of Health and Human services establishes the National Center for Human Genome Research (NCHGR)

1990 The Human Genome Project begins

First gene therapy success story (severe combined immuno-deficiency)

1994 The Human Genome Project develops a comprehensive human genetic linkage map (relative order of and approximate spacing between specific DNA patterns positioned on chromosomes)

1997 NCHGR becomes National Human Genome Research Institute (NHGRI)

1998 First anti-sense oligonucleotide (ASO) approved by the FDA (cytomegalovirus retinitis in immuno-compromised AIDS patients)

2003 The Human Genome Project is completed (two years ahead of schedule; all of the genes in the human genome mapped and sequenced, 90% of the human genome)

2009 Long-read sequencing developed

First successful application of whole-exome sequencing to monogenic disease

2010 Improved viral vectors for gene therapy

2012 CRISPR-Cas9 successfully repurposed as a gene-editing tool

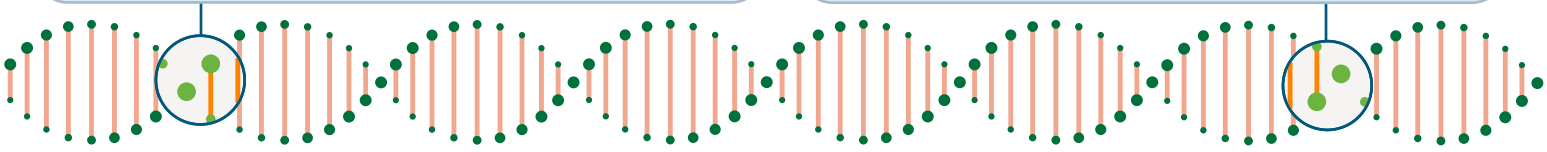
2015 Framework for *All of Us* Research Program developed

2017 First gene therapy is FDA approved (CAR T-cell therapy, Kymriah, acute lymphoblastic leukemia)

2018 First siRNA-based therapy approved by the FDA (patisiran; transthyretin-mediated amyloidosis)

2022 The NIH-funded Telomere to Telomere (T2T) project completes, establishing the first complete, gapless sequence of a human genome

2023 First CRISPR-based treatment approved by FDA (Casgevy, sickle cell disease and beta-thalassemia)



policy@ashg.org · www.ashg.org

NIH FUNDING FOR HUMAN GENETICS & GENOMICS RESEARCH

Institutes and Centers across the NIH fund human genetics and genomics research, which is foundational to modern medicine.

NHGRI is uniquely equipped to ask human genetics and genomics questions, with other institutes and centers applying the techniques and tools funded by grants from NHGRI.

All people benefit from the groundbreaking work initiated and implemented through NHGRI.

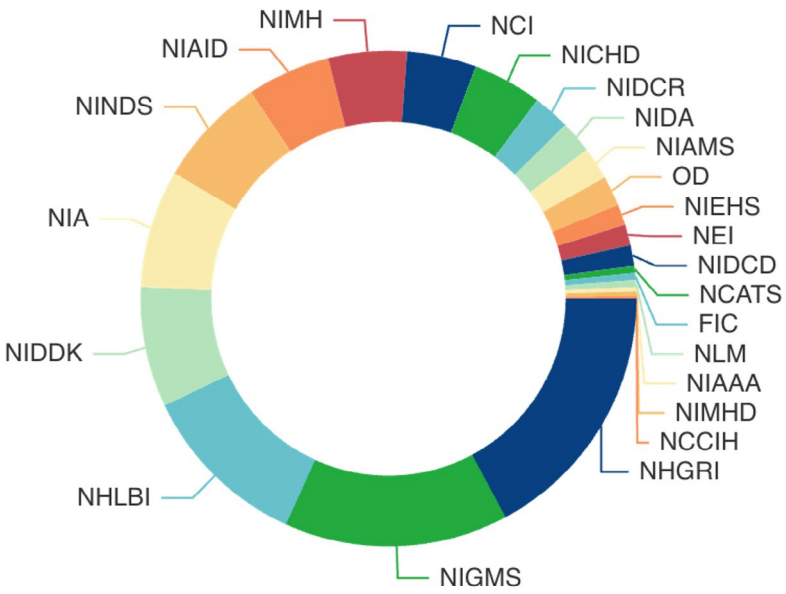


Figure 1. 24 of the 27 Institutes and Centers at NIH fund human genetics and genomics research projects. (NIH RePORTER)

GENE THERAPY/RARE DISEASE TREATMENT

- FDA has approved **36 gene therapies** for treatment of rare diseases, cancers, and neuromuscular disorders.
- An additional **500 therapies** are in the pipeline for FDA approval.
- Based on the current pipeline, the FDA anticipates approving **10 to 20** new gene therapies a year.

Researchers in Boston, Massachusetts and Philadelphia, Pennsylvania have shown that anti-sense oligonucleotide therapies and CRISPR-based therapies can be customized to treat individual patients with ultra-rare diseases.

CLINICAL GENOMIC SEQUENCING

The availability of next-generation sequencing technologies such as exome sequencing (ES) and genome sequencing (GS) has transformed rare genetic disease diagnosis. Multiple studies have demonstrated the effectiveness of ES and GS in diagnosing individuals with rare diseases, often after diagnoses had previously been missed with other testing approaches. This provides an opportunity to avoid what is commonly referred to as 'the diagnostic odyssey' by offering ES or GS earlier in the life course.

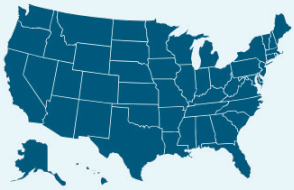
Newborn screening currently targets genetic conditions where early intervention – often within the first days of life – can either prevent serious health complications or significantly improve long-term outcomes. Genome-based newborn screening (gNBS) has the potential to expand traditional newborn screening methods to help address hundreds of severe childhood disorders more effectively.

BeginNGS, a health care delivery system at Rady Children's Hospital in San Diego, California designed to screen newborns for genetic disease and connect their doctors with effective interventions, screens for 412 genetic disorders with 1,603 therapies. In a pilot NIH-funded clinical trial, BeginNGS had sufficient efficacy and effectiveness to proceed to a multicenter, international adaptive clinical trial.

ACCESS TO GENOMIC MEDICINE

Rare disease and other genetic conditions affect at least **200 million** individuals worldwide.

Rare disease and other genetic conditions affect at least **30 million** individuals in the United States.



Rare disease diagnosis takes an average of **5 to 7 years** and up to **12 specialists.**

The expansion of the Undiagnosed Disease Network (UDN) to over a dozen sites throughout the United States enables more individuals to access experts to help solve the most challenging medical mysteries using advanced technologies.

Thanks to NIH-funded research, biotechnology companies and NIH-funded researchers have developed cheaper and more accurate ways of sequencing the human genome, enabling clinical genomic testing to be provided to hundreds of thousands of individuals in the United States.

Cost per Genome

NIH funding has supported the development of innovative DNA sequencing technologies, and genome sequencing can now be done quickly and cheaply, enabling new research and clinical applications.

In 2001, the cost to sequence a human genome was ~$100,000,000. By 2024, the cost dropped to ~$500 for a complete human genome.

Research from the NIH *All of Us* Research Program has found more than

275 million

previously unreported genetic variants identified in data shared by nearly

250,000 participants

+

New data on participant environmental exposures and lifestyles

This ongoing effort gives researchers new pathways to better understand the genetic and environmental influences on health and disease.

policy@ashg.org · www.ashg.org

ECONOMIC IMPACT OF HUMAN GENETICS AND GENOMICS RESEARCH

$52 BILLION

As an agency, the NIH supports 76,362 active projects at universities, hospitals, clinics, medical schools, research centers and more totaling over $52 billion in all 50 US states (NIH RePORTER).

$1 ➠ $2.56

United for Medical Research (UMR) highlights that every $1 invested in NIH research generates $2.56 in new economic activity, including creating jobs across the nation.

$4.9 BILLION

In 2024, NIH allocated an estimated $4.9 billion toward human genome research.

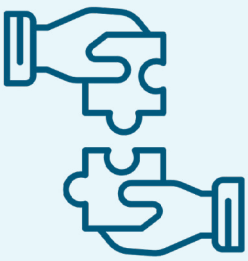
4.75:1.00 ROI

The 2021 economic impact report produced in partnership with ASHG demonstrated how human genetics and genomics is fundamental for biomedical research and healthcare, and boosts the U.S. economy with a 4.75:1.00 federal return on investment.

850,000 JOBS

The human genetics and genomics research field supports at least 850,000 jobs across the United States, including 152,000 industry positions.

NIH's Institutes also support research funding for small businesses through the Small Business Innovation Research (SBIR) and Small Business Technology Transfer (STTR) programs, also known as America's Seed Fund. These grants support public-private partnerships to advance scientific discovery.



- In Fiscal Year 2023, **275 projects** received funding through the SBIR/STTR programs, totaling **over $168 million.**
- Currently, there are **81 active projects across 27 states** awarded to small businesses, with a combined funding amount **exceeding $44 million.**
- The SBIR/STTR funding mechanisms underscore that **collaborations** between small businesses and academic institutions can drive **meaningful change** at the state and local level.

LOOKING AHEAD

Human genetics and genomics research has achieved extraordinary progress, laying the foundation for innovations that improve health and well-being worldwide. Looking ahead, the next generation of discoveries — driven by emerging scientists and transformative technologies — holds even greater promise. Through continued investment, we can ensure these advances benefit all people, reflecting our broad community and global impact: One Humanity, Many Genomes™.

WHAT'S NEXT?

Making an effort to capture the entirety of the human genome across individuals (NIH-funded Human Pangenome Reference Sequencing Project)

Expanding access across the U.S. to advanced clinical genomic testing (NIH-funded Undiagnosed Diseases Network)

Understanding how mutations impact human health (NIH-funded Somatic Mutation across Human Tissues Network)

Staying updated on statements in the ASHG Advocacy Center

Published July 2025

policy@ashg.org · www.ashg.org