Discovery

- **Evolution and genetic variation**: Researchers at Vanderbilt in Tennessee are analyzing how Neanderthal-derived DNA in the modern human genome can influence disease risk such as excessive blood clotting and tobacco addiction.
- **Microbiome**: The Human Microbiome Project is using DNA sequencing to characterize the microorganisms that live on the human body to better understand how the microbiome impacts health and disease.

Diagnosis

- **In the NICU**: Researchers at UC San Diego are using new DNA sequencing technologies to speed up the time to diagnosis for newborns suspected to have a genetic disease.
- **Ultra-rare diseases**: At University of North Carolina Chapel Hill, a woman regained the ability to walk after DNA sequencing was used to diagnose her with a rare condition called dopa-responsive dystonia.

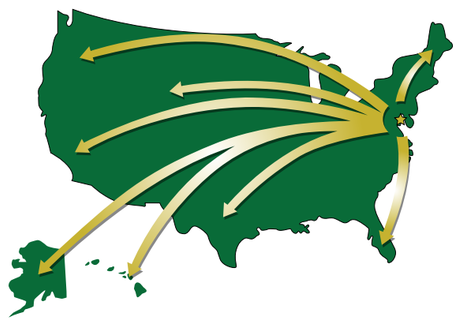
Determinants of disease

- **Cardiovasular diseases**: Researchers in Boston, Massachusetts, have developed a new method of analyzing millions of points in a person's genome to assess their risk of heart disease. This 'polygenic risk score' approach has taken off in many disease areas.
- **Rare diseases**: The Centers for Mendelian Genomics are working to identify the cause of thousands of diseases suspected to be genetic.

Treatments

- **Cancer CAR T-cell immunotherapy**: In research recently approved by the FDA, two new therapies modify patients' own immune cells to target certain types of non-Hodgkin's lymphoma and acute lymphoblastic leukemia in children.
- **Gene therapy**: Researchers at Dana-Farber in Massachusetts are testing a gene therapy for treating sickle cell disease, by removing a patient's own stem cells, and enabling the production of healthy hemoglobin.

The world's largest funder of genetics research, the NIH spent $3.30 billion on research on the human genome in 2017.

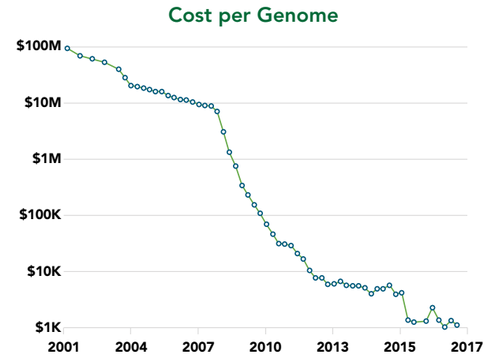


Around 83% of NIH research dollars are distributed to institutes across all 50 states and worldwide.

Our genome has a fundamental influence on human disease. Accordingly, human genome research is a significant component of the research portfolio of many NIH institutes (2017 data).

$764M	*National Cancer Institute*
$485M	*National Human Genome Research Institute*
$235M	*National Heart Lung and Blood Institute*
$159M	*National Institute on Aging*
$118M	*National Institute of Diabetes and Digestive and Kidney Diseases*
$85M	*Eunice Kennedy Shriver National Institute of Child Health and Human Development*

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



As well as advancing science and health, genetics and genomics research is advancing the economy.

Federal Investment in the Human Genome Project and related research (1988-2012)

$12.3B Investment

$293B Personal Income

$965B Economic Impact

4.3M Job Years

ASHG Policy Priorities

Research

Research funding
We need robust, predictable, sustained NIH funding to fuel advances in genetics and genomics.

Public participation in research
The public is a vital partner in genetics research studies, and research participants deserve clear informed consent and strong protections to ensure their rights and welfare.

Genome editing
It is important to realize the potential of this powerful technology in an appropriate and responsible way. Regarding heritable genome editing, ASHG recommends against genome editing that culminates in human pregnancy. It supports publicly funded, *in vitro* research into its potential clinical applications and additional steps before implementation of such clinical applications is considered. The Society actively supports current global dialogue underway to establish rigorous governance models and prevent misuse.

Diversity in research cohorts
Policies supporting improved diversity among research participants strengthen our ability to study the genome and the genetic factors that influence health and disease, ensuring that everyone can benefit from clinical advances.

Healthcare

Coverage of genetic testing
Government health insurance programs and private payers should establish clear policies covering the use of genetic tests that improve healthcare, as well as coverage of the associated patient services.

Medicare coverage for genetic counseling
Genetic counselors play a key role in healthcare, and it is essential that their services are accessible to patients.

Society

Genetic discrimination
There should be strong implementation of the Genetic Information Nondiscrimination Act, which established protections against genetic discrimination in the workplace and in health care, as well as protections for individuals with genetic diseases. This will help ensure that an individual's genetic information can only be used in ways that benefit them, and assure them that they can participate in research without fear of harm.

Genetic privacy
Whether for research, clinical use, or any other purpose, disclosure of individuals' genome information should be entirely voluntary. In establishing broad data privacy protections, it is critical to avoid imposing undue restrictions on the conduct of genetics research, which is already subject to regulations that protect the rights of research participants.

References: https://www.ashg.org/policy/NIH-research.shtml

ASHG
American Society of Human Genetics

ASHG is the primary professional scientific membership organization for human genetics specialists worldwide. The Society's 8,000 members include researchers, clinicians, laboratory practice professionals, genetic counselors, nurses and others who have a special interest in the field of human genetics. Our members work in a wide range of settings, including universities, hospitals, biotech, and medical and research laboratories. The ASHG Annual Meeting is the largest human genetics meeting in the world with over 9,000 attendees and 250 exhibiting companies.

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Human Genetic & Genomic Research:
Driving Discovery and Better Health

"There's no question that from a scientific standpoint, the transformation of biomedical research by genomic technology has been absolutely profound."
–Dr. Francis Collins, NIH Director

Thanks to decades of robust federal funding for National Institutes of Health research, new genetic and genomic knowledge is transforming medicine and saving lives.

Advances include more precise diagnoses, drugs, and therapies; new understanding and earlier screening of rare and common disease; and a better understanding of the rich evolutionary heritage of all peoples. New research frontiers such as gene therapy and gene editing present opportunities to treat—and even reverse—disease more effectively.

ASHG recognizes and appreciates Congress' commitment to medical research. We urge continued strong support to deliver new knowledge, treatments, answers, and hope.

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