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Testimony on behalf of the American Society of Human Genetics,
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Submitted to the Senate Appropriations Subcommittee on
Labor, Health and Human Services, Education, and Related Agencies,
Fiscal Year 2024 Funding for the National Institutes of Health

May 18, 2023

The American Society of Human Genetics (ASHG) thanks the Subcommittee for its continued strong support and leadership in funding the National Institutes of Health (NIH). The $2.5 billion increase provided for NIH in Fiscal Year (FY) 2023 reinforces our nation's commitment to the health and well-being of all Americans at a time when investing in biomedical research and scientific innovation is more important than ever. **ASHG urges the Subcommittee to appropriate at least $50.924 billion for NIH's base budget in FY 2024. This amount would allow NIH's base budget to keep pace with the biomedical research and development price index (BRDPI) and allow meaningful growth of five percent.**

Extraordinary Progress in Human Genetics & Genomics Research

Federal funding for human genetics and genomics research is enabling new insights into the structure of, and variation in, the human genome, and leading to new discoveries in preventing, diagnosing, and treating disease. Advances in genomic sequencing, both in sequencing speed and cost reduction, have been leveraged to enhance the ability to detect and diagnose rare neurogenetic disorders, genetic predisposition to early-onset heart conditions, and rare conditions that predispose individuals to colorectal, endometrial, and other cancers.[1] Understanding the genomic contribution to disease enables improved diagnosis and treatment, subsequently reducing healthcare costs and improving well-being.[2] Continued federal funding to support research exploring the genetic underpinning of cancer has led to better techniques for early detection and prevention of cancerous tumors, leading to increased survival,[3] and has created a national ecosystem for sharing and analyzing cancer data so that researchers can more efficiently collaborate, increasing the potential for new discoveries.[4]

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[1] https://pubmed.ncbi.nlm.nih.gov/36459977/
[2] Health NRC (US) and I of M (US) R on TGBR for. *Impact on Health Care and Public Health*. National Academies Press (US); 2010. Accessed March 28, 2023. https://www.ncbi.nlm.nih.gov/books/NBK209648/
[3] https://acsjournals.onlinelibrary.wiley.com/doi/full/10.1002/cncr.34479; https://doi.org/10.1002/cncr.34479
[4] https://www.cancer.gov/research/key-initiatives/moonshot-cancer-initiative

Additionally, NIH funding for human genetics and genomics has directly unleashed the power of precision medicine to improve health and well-being. Due to critical investments like NIH's *All of Us* Research Program, a groundbreaking effort to collect and study data from over one million people, researchers and clinicians can now compare an individual's personal and family history to more accurately identify predisposition to disease. For example, heart disease is responsible for 1 in 5 deaths[5] in the United States, and it disproportionately affects individuals from racial and ethnic communities that have been historically underrepresented in medical research. There are now more than 200 research projects on heart disease and more than 15 peer-reviewed studies about heart disease that used the *All of Us* dataset. *All of Us* is building one of the largest, most diverse, and broadly accessible datasets for researchers, enabling studies addressing health disparities and investigating novel treatments for a range of conditions, including heart disease.[6]

Human genetics and genomics research is also delivering hope for the millions of people in the United States living with rare diseases. For example, decades of work funded by the NIH have led to the first FDA approved drug for treating progeria, a rare pediatric genetic disorder that causes accelerated aging and premature death in children.[7] This advancement was made possible by our understanding of human genetics and subsequent knowledge of exactly how a single gene mutation leads to disease. In addition, the NIH's Undiagnosed Diseases Network (UDN) was created to establish a nationwide network of researchers and clinicians who leverage advances in genomics tools and research to uncover the mechanisms underlying rare diseases and conditions, which collectively impact approximately 25 million Americans. Funding for the UDN has improved the ability for patients suffering from undiagnosed conditions to receive the clarity of a diagnosis and has contributed to major advances in therapies (e.g., gene and cell therapy) that can be used to treat both rare and common genetic diseases. Knowledge gained from the study of rare genetic diseases will also impact our approach to treating common diseases. Critically, these advances are driven by NIH's robust support for fundamental research to better understand disease mechanisms. The *Consolidated Appropriations Act 2023* reaffirmed the importance of the UDN and ASHG supports its continuation as a priority program at the NIH.

Genetics & Genomics: Striving for Equity and Research Cohort Diversity

Genetic science can advance health equity through the deliberate, meaningful inclusion and participation of individuals from diverse groups in human genetics and genomics research. While human beings are 99.9 percent identical in our genetic makeup, the variation in the remaining 0.1% can influence a person's risk of disease or how the body responds to medications, stress, or environmental factors. Polygenic-risk scores (PRS) are one way to

[5] https://www.cdc.gov/heartdisease/facts.htm

[6] Lunt C, Mayo K, Master H, Growing a research ecosystem in the cloud: Early insights from the *All of Us* Research Program; (Abstract #PB3215). Presented at the annual meeting of the American Society of Human Genetics, October 27, 2022, in Los Angeles, California.

[7] https://www.fda.gov/news-events/press-announcements/fda-approves-first-treatment-hutchinsongilford-progeria-syndrome-and-some-progeroid-laminopathies

provide insight into a person's risk for disease based on their genes. It is imperative for participation in genetics and genomics research to reflect humanity's diversity so that we can gain a fuller understanding of the genetics of health and disease, so all people can enjoy its benefits, and genetic discoveries can be equitably applied in healthcare.

While human genetics and genomics research has advanced biomedical research and improved the lives of patients, the field has historically failed to account for the rich diversity of patients that the research should be designed to serve. For example, because most individuals participating in genetics research are of European ancestry, PRS tools are more effective for assessing disease risk in people of European ancestry than for individuals with Hispanic, South Asian, East Asian or African ancestries.[8] Recently, scientists have taken a step to improve the accuracy of these genetic scores for a broader population by creating a new method called PRS-CSx[9]. This new approach incorporates population level differences in DNA patterns and further pushes this approach closer to being useful in clinical contexts. However, the lack of participation of underrepresented communities in human genetics and genomics research can largely be attributed to issues around scientific mistrust, miscommunication, and barriers that make it challenging for individuals to participate in this much needed research.

During the COVID-19 pandemic, NIH established the Community Engagement Alliance (CEAL) Initiative consisting of researchers, policy-makers, community-based organizations, faith-based organizations, and other relevant stakeholders to tackle scientific misinformation and build public trust in biomedical research and its advances among communities across our nation.[10] Community engagement to foster sustainable partnerships and trust is also key for science when there is public hesitancy to participation in research, as is the case with human genetics and genomics research

ASHG was pleased to see that the *Consolidated Appropriations Act 2023* included language urging the National Human Genome Research Institute (NHGRI) to build on these successes and establish a community engagement program to support efforts to increase the participation of individuals historically underrepresented and hesitant to participate in human genetics and genomics research.

Broad Data Sharing: Essential for Human Genetics and Genomics Research

The funding provided by the U.S. government to the NIH is instrumental in promoting the health and stability of the United States, and its benefits extend globally. Broad sharing of human genome data from NIH-funded research drives cutting-edge innovations worldwide and is essential for advancing science and maximizing the public's return on investment in biomedical research. The genetics and genomics research community is a leader in developing best practices for sharing data while protecting individuals' privacy. We strongly support

[8] https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9072305/
[9] Ruan, Y. *et al.* Improving polygenic prediction in ancestrally diverse populations. *Nat Genet* **54**, 573–580 (2022).
[10] https://covid19community.nih.gov/about/who-we-are

policies including the Common Rule, the *Genetic Information Nondiscrimination Act* (GINA), the *21st Century Cures Act*, the NIH Genomic Data Sharing Policy, and the *Health Insurance Portability and Accountability Act* (HIPAA), which together act to protect individuals from the inappropriate disclosure of data for non-research purposes.[11] As Congress continues its oversight of the nation's research enterprise, we urge the Committee to recognize the privacy protections already established by Congress and NIH for genetic research data, and to ensure that broad data-sharing can continue to fuel scientific progress and maintain the United States' leadership and global competitiveness.

Summary

ASHG joins the Ad Hoc Group for Medical Research in recommending at least a $50.924 billion base budget for NIH for FY 2024. This funding level would allow NIH's base budget to keep pace with inflation, specifically the biomedical research and development price index, and support crucial research on human genetics and genomics across all of the NIH's 27 Institutes and Centers.

The American Society of Human Genetics (ASHG), founded in 1948, is the primary professional membership organization for human genetics specialists worldwide. The Society's nearly 8,000 members include researchers, clinicians, genetic counselors, nurses, and others who have a special interest in the field of human genetics.

[11] American Society for Human Genetics. (2021). Perspectives: Research and Privacy [Fact sheet]. https://www.ashg.org/wp-content/uploads/2021/08/Factsheet-DataPrivacy.pdf