

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 2023 Funding for the National Institutes of Health

May 11, 2022

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 \$2.03 billion increase provided for Fiscal Year (FY) 2022 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 \$49 billion for NIH's base budget in FY 2023, with any additional funding for the newly established Advanced Research Projects Agency for Health (ARPA-H) to supplement, not supplant, the core investments in the NIH base budget.**

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.¹ Through the development of powerful DNA sequencing and computational tools, in the past year, an NIH-supported consortium generated the first complete assembly of a human genome, greatly expand our understanding of genomic variation and providing an essential tool for exploring the genetic underpinnings of disease.² Researchers are also developing new 'polygenic score' tools to assess one's risk for many of the leading causes of death in the United States, including cardiovascular diseases, immune disorders, and cancers.³ Such tools hold promise for enabling a personalized approach to preempting and preventing disease.

Genetic science is delivering major advances in the detection and treatment of chronic diseases, notably cancer. Through 'liquid biopsy' blood tests, clinicians can identify genetic changes in cancerous tumors in a non-invasive way to guide targeted treatments; this testing method is being further developed for the early screening and detection of multiple cancers. Because of federally funded research findings, we now have novel cancer treatment options such as CAR-T gene therapies. As of this year, the FDA currently lists 23 approved cellular and gene therapy products to treat cancers and other diseases.⁴

Human genetic and genomic research is also delivering hope for the millions of people in the United States living with rare diseases. For example, results from NIH-funded trials investigating gene therapies and gene editing technologies indicate that genetic

approaches will allow patients afflicted with sickle cell disease to live pain-free and no longer in need of frequent blood transfusions. Effective gene therapy is now available for spinal muscular atrophy, a rare childhood disease characterized by progressive muscle weakness, and is being tested to treat other devastating diseases like Huntington's disease and familial amyotrophic lateral sclerosis (Lou Gehrig's disease).

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. The inclusion of populations representing diverse ancestries helps us gain a fuller understanding of the genetics of health and disease, knowledge which can be used to develop more accurate diagnostic tests and more effective treatments that benefit all Americans.⁵ Diverse participation in research is essential if we are to realize the full promise of human genetics and genomics research and the equitable application of genetic discoveries in healthcare and society.

Genetics research studies illustrate the importance of research cohorts reflecting humanity's diversity.⁶ For example, because most individuals participating in genetics research are of European ancestry, polygenic risk score tests are more effective for assessing disease risk in people of European ancestry than for individuals with Hispanic, South Asian, East Asian or African ancestries.⁷ The Society commends NIH's efforts to advance diverse participation in research, particularly the All of Us Research Program.⁸ Significantly, in 2022, this program released a database of health information and whole-genome sequences from almost 100,000 individuals,⁹ half of whom are from historically underrepresented racial or ethnic backgrounds.¹⁰

Return on Investment: Genetics Research Benefits the Economy

As the United States moves towards recovery from the COVID-19 pandemic, economic activity across all sectors remains key for our return to normalcy. In addition to its importance for addressing health care needs in the United States, federal investments in research and development have been shown to drive economic activity. A 2021 study commissioned by ASHG and conducted by TEconomy Partners highlights the growth of a dynamic ecosystem derived from human genetics and genomics research, and that the development and manufacturing of genomic technologies, diagnostics and therapeutics, and the associated healthcare services, "generate substantial U.S. economic activity and support a large volume of jobs across the nation."¹¹ The report estimates that the human genetics and genomics sector supports 850,000 jobs and generates \$265 billion in total economic activity annually,¹² demonstrating that this sector has grown around five-fold in the last decade.

Broad Data Sharing: Essential for Human Genetics and Genomics Research

Broad sharing of human genome data from NIH-funded research is essential for advancing science and maximizing the public's return on investment in biomedical research. Since the human genome houses sensitive information, the genetics and

genomics research community is a leader in developing best practices for sharing data while protecting individuals' privacy. We strongly support policies including the Common Rule, the Genetic Information Nondiscrimination Act (GINA), the 21st Century Cures Act, the NIH Genomic Data Sharing Policy, and HIPAA, which together act to protect individuals from the inappropriate disclosure of data for non-research purposes.¹³ As Congress encourages NIH to explore the national security risks associated with the sharing of individuals' health information, 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.

Summary

ASHG joins the Ad Hoc Group for Medical Research in recommending at least a \$49 billion base budget for NIH for FY 2023. 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. ASHG also recognizes the important and catalytic role of ARPA-H for advancing science and biomedicine, building on the foundation of basic research supported by NIH. Funding to establish ARPA-H should complement NIH's current investments in basic research.

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.

¹ American Society of Human Genetics. (2021). Success Stories in Human Genetics and Genomics Research (Chronic Diseases) [Fact sheet]. <https://www.ashg.org/wp-content/uploads/2022/01/The-Benefits-of-human-genetics-Noncommunicable-Diseases-factsheets-v3.pdf>

² Nurk, S. et al. The Complete Sequence of a Human Genome. *Science* 376(6588): 44-53 (2022). <https://doi.org/10.1126/science.abj6987>

³ <https://www.cdc.gov/nchs/fastats/leading-causes-of-death.htm>

⁴ <https://www.fda.gov/vaccines-blood-biologics/cellular-gene-therapy-products/approved-cellular-and-gene-therapy-products>

⁵ Collins, F., Doudna, J.A., Lander, E., and Rotimi, C.N. Human Molecular Genetics and Genomics — Important Advances and Exciting Possibilities. *N.Engl.J.Med* 384: 1-4 (2021)

⁶ Wojcik G., et al. Genetic Analysis of diverse populations improves discovery for complex traits. *Nature* 570(7762): 514-518 (2019). <https://doi.org/10.1038/s41586-019-1310-4>.

⁷ Ibid.

⁸ <https://allofus.nih.gov/>

⁹ <https://www.researchallofus.org/>

¹⁰ <https://www.nih.gov/news-events/news-releases/nih-s-all-us-research-program-releases-first-genomic-dataset-nearly-100000-whole-genome-sequences>

¹¹ Tripp, S., and Grueber, M. 2021. The Economic Impact and Functional Applications of Human Genetics and Genomics.

¹² Ibid.

¹³ American Society for Human Genetics. (2021). Perspectives: Research and Privacy [Fact sheet]. <https://www.ashg.org/wp-content/uploads/2021/08/Factsheet-DataPrivacy.pdf>