# National Human Genome Research Institute

CONGRESSIONAL JUSTIFICATION
FY 2024

Department of Health and Human Services
National Institutes of Health

NIH National Human Genome Research Institute

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DEPARTMENT OF HEALTH AND HUMAN SERVICES

NATIONAL INSTITUTES OF HEALTH

National Human Genome Research Institute (NHGRI)

FY 2024 Budget Table of Contents



**General Notes**

1. FY 2023 Enacted levels cited in this document include the effects of the FY 2023 HIV/AIDS transfer, as shown in the Amounts Available for Obligation table.
2. Detail in this document may not sum to the subtotals and totals due to rounding.

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## Director's Overview

Eric D. Green, M.D., Ph.D.
Director, NHGRI

The Human Genome Project's successful completion nearly 2 decades ago laid the groundwork for the subsequent integration of genomics into virtually all areas of biomedical research and, eventually, healthcare. The field of genomics has been able to build on the style by which the Human Genome Project democratized the pursuit of science, capitalizing on the benefits of large research consortia and the broad sharing of data to drive cutting-edge innovations worldwide in both the research and clinical settings. Attaining a deep foundational understanding about the genomes present in all human populations will be key for ensuring that genomic advances benefit everyone. As the NIH Institute created to lead the United States's efforts in the Human Genome Project, the National Human Genome Research Institute (NHGRI) is uniquely poised to continue providing pivotal leadership at *The Forefront of Genomics*.

NHGRI is committed to developing ever-improving approaches to study the role of genomic variation in health and disease, and such efforts must include the study of diverse populations. This begins with the generation of genome-sequence data and reference genome sequences; however, there has been a historic lack of diversity with respect to such data and sequences – with the correction of this problem now representing a high priority for the field. Knowledge about how genomic variants and their downstream biological effects vary across human populations is key for understanding the genomic contributions to health and disease and how such information can be used in clinical care. To aid these studies, the Human Genome Reference Program (HGRP) is producing a set of high-quality human genome reference sequences that will represent a larger fraction of the world's human population. Together, these reference sequences will contain a more accurate and diverse representation of global genomic variation, improve genomic variant-disease association studies across populations, expand the scope of genomics research to include the most difficult-to-understand regions of the human genome, and serve as the ultimate genome-sequence resource for the genomics community.

The ability to generate high-quality genomic data quickly and cheaply now outpaces the full extraction of meaningful insights from the generated data. For example, it is estimated that

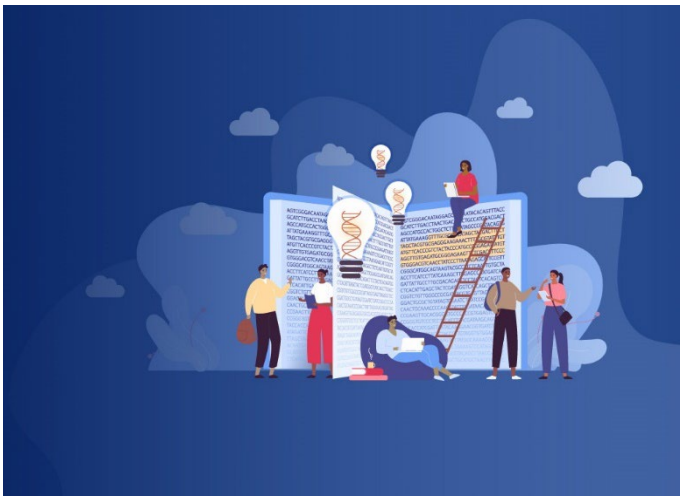
genomics research will generate between 2 and 40 exabytes (an exabyte is 1 billion gigabytes) of data within the next decade, almost certainly at a pace that will outstrip the collective analytical capabilities. To begin closing this gap, NHGRI is spearheading efforts to advance genomic data science, investing in the development and refinement of computational tools and resources that will become foundational for future genomic research studies. For example, NHGRI is leading efforts to create new cutting-edge analysis platforms, data resources, internet browsers, portals, and visualization tools that will allow the research and medical communities to share and analyze the increasingly complex and heterogenous data types (e.g., genome-sequence, environmental, lifestyle, and clinical data) needed to fully realize the potential of genomic medicine.

To enhance NHGRI's leadership in genomic data science, the Institute recently established the Office of Genomic Data Science (OGDS). In addition to serving as the hub for NHGRI's genomic data science efforts, OGDS coordinates NHGRI's involvement in NIH-wide, national, and international genomic data science efforts. Being cognizant of the importance of data sharing, OGDS also provides guidance for NHGRI's implementation of NIH data sharing policies, promoting the proper governance, stewardship, and sustainability of shared data. NHGRI also leads efforts that encourage data scientists to become a part of the genomics research community through the support of training, mentored research experiences, and career development programs.

Among the many high-priority areas that depend on robust data-analysis methods are efforts to connect genomic variants and phenotypes, as exemplified by NHGRI's Impact of Genomic Variation on Function (IGVF) program. Another area relates to continued efforts to understand all functional elements in the human genome, including the function of all genes; for the latter, the recently launched Molecular Phenotypes of Null Alleles in Cells (MorPhiC) program seeks to determine the molecular and cellular phenotypes associated with the knockout of specific genes.

Beyond genome-sequence data, NHGRI also aims to develop technology for generating other data types (e.g., transcriptomic data and epigenetic data) at orders-of-magnitude lower costs, at single-cell resolution, at distinct spatial locations within tissues, and at different time points. Continued advances in myriad high-throughput technologies are increasing access to a wider array of distinct molecular data types. In addition, the newly established Multi-Omics for Health and Disease Consortium seeks to advance the application of multi-omic technologies to study health and disease in ancestrally diverse populations, and the Genomics Research Elucidates Genetics of Rare Disease (GREGoR) Consortium aims to leverage multi-omics data to find the genomic bases for remaining rare diseases for which the mutated genes have yet to be discovered.

For genomic discoveries to be appropriately harnessed for advancing human health at all stages of life and for all people, health equity must be foundational in genomics research. Many of the guiding principles and values for human genomics articulated in the 2020 NHGRI Strategic Vision demonstrate NHGRI's commitment to health equity and the prevention of disparities in



genomics research. Health equity in genomics requires an understanding of how biology influences disease and how disease is influenced by biological and non-biological (e.g., social) determinants of health. The Polygenic Risk Methods in Diverse Populations (PRIMED) Consortium is working to improve genomic risk prediction for common diseases by studying larger numbers of non-European individuals. NHGRI is committed to the systematic inclusion of ancestrally diverse and underrepresented groups in genomic studies so as to close existing evidence gaps among diverse and underserved populations. To aid these efforts, NHGRI recently established the Training, Diversity, and Health Equity (TiDHE) Office, which will develop, support, and lead the Institute's initiatives aimed at addressing health disparities and inequities in genomics as well as its programs focused on attracting, developing, and retaining a diverse genomics workforce.

NHGRI strives to realize one of its featured Bold Predictions for Human Genomics by 2030 described in the 2020 NHGRI Strategic Vision: "Research in human genomics will have moved beyond population descriptors based on historic social constructs such as race." To do so, NHGRI supports examining the use of race, ethnicity, and ancestry as population descriptors in genomics research. The institute is also working to ensure that genomic medicine applications are unbiased and equitably accessible. NHGRI is also committed to addressing and combatting the legacies of eugenics and scientific racism that are linked to the field of genomics, so as to develop an inclusive and welcoming genomics community.


Representing the Telomere-to-Telomere Consortium, Adam Phillippy, Karen Miga, Evan Eichler, and Michael Schatz were named to *Time* magazine's 100 Most Influential People of 2022.

Genomics is still a relatively nascent field, especially compared to other areas of biomedical research. That youthful spirit still brings captivating and inspirational successes, such as the recent generation of the first truly complete sequence of a human genome. This accomplishment garnered significant recognition by the scientific community, the popular press, and the red carpet. There is something universally human about being comprehensive about exploring the very blueprint that makes us *us*. But with that exploration comes great responsibility and potential pitfalls or misuses of genomics. NHGRI embraces its leadership role in navigating these complex issues, so as to ensure that genomics is a science for everyone by everyone.

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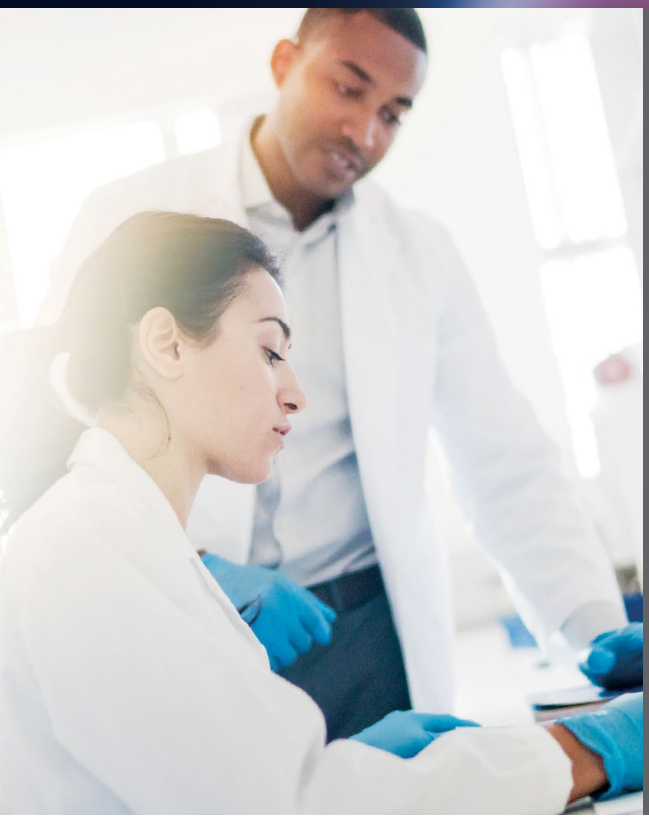
The Forefront of Genomics®

genome.gov

# National Human Genome Research Institute

NHGRI conducts and funds world-class genomics research, trains the next generation of genomics experts, and collaborates with diverse communities to improve human health.



## Empowering the Biomedical Research Community

Building on our leadership role in sequencing the human genome, NHGRI uses a "team science" approach to tackle increasingly complex biological problems. As one of the smaller institutes at the National Institutes of Health (NIH), we make every dollar we spend impactful. We assemble interdisciplinary research teams consisting of scientists, clinicians, ethicists, and other experts, and we ensure they have the funding and resources to advance genomics. This approach has stimulated a cultural change across biomedical research toward enhanced data sharing, increased collaboration in the scientific and medical communities, and a heightened appreciation for the impact of genomics on society.

### Research Funding Areas:

- Structure and Biology of the Genome
- Biology of Disease
- Genomic Medicine
- Data Science
- Early Career Training
- Ethical, Legal, and Social Implications of Genomics
- Small Business Innovation

## Our Institute

**385** FTEs
**FY 2023**

**$660,510,000** FY 2023
Enacted Budget
(1.3% of the total NIH budget)

**$660,510,000** FY 2024
President's Budget Request

**5%** of NHGRI's research budget is dedicated to studying the ethical, legal, and social implications of genomics

## From Bench to Bedside: Transforming Patient Care

By catalyzing new technologies and building translational programs, our experts help understand, diagnose, and treat both rare and common diseases in sophisticated new ways.

### Genomic Technologies

The research we fund paves the way for precision medicine and therapeutic interventions. NHGRI-funded investigators and consortia are developing new genomic and data analysis tools to transform the study of human biology and disease. One such center, the Center for Genome Editing and Recording led by 2020 Nobel Prize winner Dr. Jennifer Doudna, employs CRISPR/Cas9 genome-editing technology to understand how genomic variation contributes to disease and uncover new therapeutic targets.

### Genomic Medicine

We are enabling healthcare professionals to use their patients' genomic information for personalized care. NHGRI is implementing genomic medicine across different clinical settings and in diverse populations as part of the Implementing Genomics in Practice (IGNITE) Pragmatic Trials Network (PTN). By integrating genomic data into electronic medical records and clinical decision making, NHGRI is piloting new approaches for personalized treatments that will be available to everyone in the future.

## Driving Responsible Use of Genomics

NHGRI is committed to advancing genomic knowledge and ensuring that genomics benefits the health of all humans. We consider the ethical, legal, and social implications (ELSI) of genomics in all aspects of our work. The ELSI Research Program supports transdisciplinary Centers of Excellence in ELSI Research (CEER) that examine cutting-edge questions in ELSI research and translate this scholarship into practice and policy.

Our teams:

- Explore privacy concerns and how to communicate study results to research participants and their providers.
- Strive to increase participation of underrepresented populations in research.
- Engage with diverse communities to increase genomic literacy.
- Work with policymakers to inform policy decisions for a future in which genomics is part of daily life.

## A 2020 Vision for Genomics

In October 2020, NHGRI published a strategic vision for the next decade that aims to expand genomics into new frontiers and enable novel applications to human health and disease. Through a multi-year process of strategic engagement, NHGRI collected input from diverse stakeholders at over 50 events. The anticipated advances in technology development, biological insights, and clinical applications will integrate genomics into virtually all areas of biomedical research, mainstream medical and public-health practices, and everyday life. NHGRI's strategic vision highlights research opportunities and priorities that are at *The Forefront of Genomics*:

I. Providing a socially responsible and highly ethical framework for conducting human genomics research by establishing and adhering to guiding principles and values.

II. Facilitating genomic advances by sustaining and improving a robust foundation for genomics research.

III. Breaking down barriers in genomics through advances that create new research opportunities and improve clinical care.

IV. Defining and leading the pursuit of bold and compelling genomics research projects focused on elucidating genome function, understanding human disease, studying the societal implications of genomics, and improving human health.

## Major Changes in the Budget Request

Major changes by budget mechanism and/or budget activity detail are briefly described below. Note that there may be overlap between budget mechanism and activity detail, and these highlights will not sum to the total change for the FY 2024 President's Budget request for NHGRI, which is the same as the FY 2023 Enacted level, $660.5 million. Within this request level, NHGRI will pursue its highest research priorities through strategic investments and careful stewardship of appropriated funds.

Research Project Grants (RPGs) (-$9.0 million, total $313.7 million):
NHGRI will support a total of 445 Research Project Grant (RPG) awards in FY 2024, including SBIR/STTR awards. Costs for non-competing RPGs will decrease by $2.6 million and will support an additional 20 awards compared to the FY 2023 Enacted level. NHGRI will reduce non-competing RPGs by 2.5 percent of the committed levels. Funding for administrative supplements will decrease by $6.7 million compared to the FY 2023 Enacted level. Funding for competing RPGs will increase by $0.4 million relative to the FY 2023 Enacted level, with three additional awards.

Research Centers (+$3.1 million, total $12.3 million):
This increase is to support the Diversity Centers for Genome Research initiative. This program aims to establish Genomic Research Centers at Minority Serving Institutions (MSIs). These centers will support the development of innovative genomic research projects through infrastructure building and the formation of interdisciplinary research teams at MSIs.

Other Research Grants (+$2.9 million, total $117.8 million):
NHGRI will support additional training activities using the Other Research grant mechanism including the Genome Research Experiences to Attract Talented Undergraduates into Genomic Fields to Enhance Diversity program to support educational activities that encourage undergraduates from diverse backgrounds, such as those from groups underrepresented in the biomedical workforce, to pursue further training and careers in the scientific, medical, ethical, social and/or legal areas of genomics research. NHGRI will also continue to increase support to grants in efforts to improve the usability and interoperability of Model Organism Databases.

## NATIONAL INSTITUTES OF HEALTH
## National Human Genome Research Institute

### Budget Mechanism*
(Dollars in Thousands)

| Mechanism | FY 2022 Final | | FY 2023 Enacted | | FY 2024 President's Budget | | FY 2024 +/- FY 2023 | |
|---|---|---|---|---|---|---|---|---|
| | Number | Amount | Number | Amount | Number | Amount | Number | Amount |
| Research Projects: | | | | | | | | |
| Noncompeting | 278 | $226,325 | 296 | $226,547 | 316 | $223,935 | 20 | -$2,613 |
| Administrative Supplements | *(28)* | $6,848 | *(48)* | $13,379 | *(24)* | $6,700 | *-(24)* | -$6,679 |
| Competing: | | | | | | | | |
| Renewal | 11 | $9,023 | 13 | $10,363 | 14 | $10,730 | 1 | $367 |
| New | 86 | $53,572 | 87 | $54,980 | 89 | $55,029 | 2 | $48 |
| Supplements | 0 | $0 | 0 | $0 | 0 | $0 | 0 | $0 |
| **Subtotal, Competing** | **97** | **$62,595** | **100** | **$65,343** | **103** | **$65,759** | **3** | **$415** |
| Subtotal, RPGs | 375 | $295,769 | 396 | $305,270 | 419 | $296,393 | 23 | -$8,877 |
| SBIR/STTR | 30 | $16,682 | 30 | $17,453 | 26 | $17,349 | -4 | -$104 |
| Research Project Grants | 405 | $312,451 | 426 | $322,724 | 445 | $313,743 | 19 | -$8,981 |
| Research Centers | | | | | | | | |
| Specialized/Comprehensive | 2 | $736 | 2 | $3,434 | 4 | $7,815 | 2 | $4,381 |
| Clinical Research | 0 | $0 | 0 | $0 | 0 | $0 | 0 | $0 |
| Biotechnology | 2 | $6,055 | 3 | $5,812 | 3 | $4,495 | 0 | -$1,318 |
| Comparative Medicine | 0 | $0 | 0 | $0 | 0 | $0 | 0 | $0 |
| Research Centers in Minority Institutions | 0 | $0 | 0 | $0 | 0 | $0 | 0 | $0 |
| **Research Centers** | **4** | **$6,792** | **5** | **$9,246** | **7** | **$12,310** | **2** | **$3,063** |
| Other Research: | | | | | | | | |
| Research Careers | 37 | $5,257 | 39 | $5,687 | 38 | $5,589 | -1 | -$98 |
| Cancer Education | 0 | $0 | 0 | $0 | 0 | $0 | 0 | $0 |
| Cooperative Clinical Research | 0 | $0 | 0 | $0 | 0 | $0 | 0 | $0 |
| Biomedical Research Support | 0 | $0 | 0 | $0 | 0 | $0 | 0 | $0 |
| Minority Biomedical Research Support | 0 | $509 | 0 | $494 | 0 | $489 | 0 | -$5 |
| Other | 93 | $104,351 | 103 | $108,707 | 120 | $111,706 | 17 | $2,999 |
| **Other Research** | **130** | **$110,117** | **142** | **$114,888** | **158** | **$117,784** | **16** | **$2,896** |
| Total Research Grants | 539 | $429,359 | 573 | $446,858 | 610 | $443,836 | 37 | -$3,021 |
| Ruth L Kirschstein Training Awards: | FTTPs | | FTTPs | | FTTPs | | FTTPs | |
| Individual Awards | 25 | $1,129 | 25 | $1,176 | 25 | $1,193 | 0 | $16 |
| Institutional Awards | 172 | $10,683 | 174 | $11,517 | 174 | $11,678 | 0 | $161 |
| **Total Research Training** | **197** | **$11,812** | **199** | **$12,693** | **199** | **$12,871** | **0** | **$178** |
| Research & Develop. Contracts | 7 | $24,357 | 7 | $23,230 | 7 | $23,474 | 0 | $244 |
| *SBIR/STTR (non-add)* | *(0)* | *($175)* | *(0)* | *($175)* | *(0)* | *($175)* | *(0)* | *($0)* |
| Intramural Research | 237 | $130,504 | 258 | $135,044 | 258 | $137,044 | 0 | $2,000 |
| Res. Management & Support | 115 | $40,447 | 127 | $42,685 | 127 | $43,285 | 0 | $600 |
| *SBIR Admin. (non-add)* | | *($133)* | | *($0)* | | *($0)* | | *($0)* |
| Construction | | $0 | | $0 | | $0 | | $0 |
| Buildings and Facilities | | $0 | | $0 | | $0 | | $0 |
| **Total, NHGRI** | **352** | **$636,479** | **385** | **$660,510** | **385** | **$660,510** | **0** | **$0** |

* All items in italics and brackets are non-add entries.

## NATIONAL INSTITUTES OF HEALTH

## NATIONAL HUMAN GENOME RESEARCH INSTITUTE

For carrying out section 301 and title IV of the PHS Act with respect to human genome research, **[**$663,200,000**]** *$660,510,000*.

**NATIONAL INSTITUTES OF HEALTH**
**National Human Genome Research Institute**

**Summary of Changes**
(Dollars in Thousands)

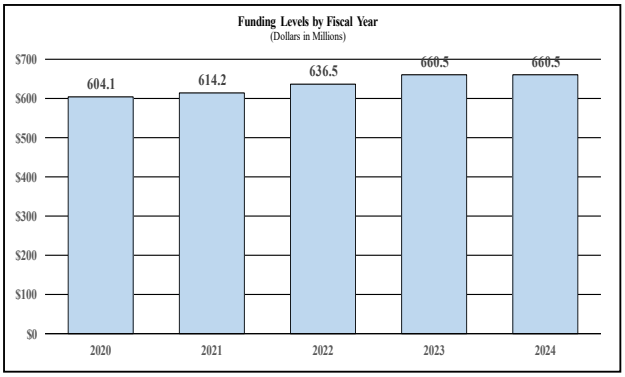
| | |
|---|---|
| **FY 2023 Enacted** | $660,510 |
| **FY 2024 President's Budget** | $660,510 |
| **Net change** | $0 |

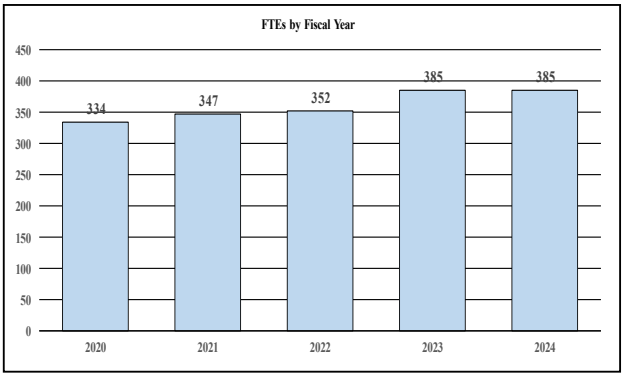
| CHANGES | FY 2023 Enacted | | FY 2024 President's Budget | | Built-In Change from FY 2023 Enacted | |
|---|---|---|---|---|---|---|
| | FTEs | Budget Authority | FTEs | Budget Authority | FTEs | Budget Authority |
| A. Built-in: | | | | | | |
| 1. Intramural Research: | | | | | | |
| a. Annualization of FY 2023 pay and benefits increase | | $51,616 | | $54,376 | | $573 |
| b. FY 2024 pay and benefits increase | | $51,616 | | $54,376 | | $1,978 |
| c. Paid days adjustment | | $51,616 | | $54,376 | | $199 |
| d. Differences attributable to change in FTE | | $51,616 | | $54,376 | | $0 |
| e. Payment for centrally furnished services | | $20,645 | | $20,976 | | $330 |
| f. Cost of laboratory supplies, materials, other expenses, and non-recurring costs | | $62,782 | | $61,692 | | $1,431 |
| Subtotal | | | | | | $4,511 |
| 2. Research Management and Support: | | | | | | |
| a. Annualization of FY 2023 pay and benefits increase | | $20,647 | | $21,750 | | $229 |
| b. FY 2024 pay and benefits increase | | $20,647 | | $21,750 | | $791 |
| c. Paid days adjustment | | $20,647 | | $21,750 | | $79 |
| d. Differences attributable to change in FTE | | $20,647 | | $21,750 | | $0 |
| e. Payment for centrally furnished services | | $1,002 | | $1,018 | | $16 |
| f. Cost of laboratory supplies, materials, other expenses, and non-recurring costs | | $21,036 | | $20,517 | | $491 |
| Subtotal | | | | | | $1,606 |
| Subtotal, Built-in | | | | | | $6,117 |

| CHANGES | FY 2023 Enacted | | FY 2024 President's Budget | | Program Change from FY 2023 Enacted | |
|---|---|---|---|---|---|---|
| | No. | Amount | No. | Amount | No. | Amount |
| B. Program: | | | | | | |
| 1. Research Project Grants: | | | | | | |
| a. Noncompeting | 296 | $239,927 | 316 | $230,635 | 20 | -$9,292 |
| b. Competing | 100 | $65,343 | 103 | $65,759 | 3 | $415 |
| c. SBIR/STTR | 30 | $17,453 | 26 | $17,349 | -4 | -$104 |
| Subtotal, RPGs | 426 | $322,724 | 445 | $313,743 | 19 | -$8,981 |
| 2. Research Centers | 5 | $9,246 | 7 | $12,310 | 2 | $3,063 |
| 3. Other Research | 142 | $114,888 | 158 | $117,784 | 16 | $2,896 |
| 4. Research Training | 199 | $12,693 | 199 | $12,871 | 0 | $178 |
| 5. Research and development contracts | 7 | $23,230 | 7 | $23,474 | 0 | $244 |
| Subtotal, Extramural | | $482,781 | | $480,181 | | -$2,600 |
| 6. Intramural Research | 258 | $135,044 | 258 | $137,044 | 0 | -$2,511 |
| 7. Research Management and Support | 127 | $42,685 | 127 | $43,285 | 0 | -$1,006 |
| 8. Construction | | $0 | | $0 | | $0 |
| 9. Buildings and Facilities | | $0 | | $0 | | $0 |
| Subtotal, Program | 385 | $660,510 | 385 | $660,510 | 0 | -$6,117 |
| Total built-in and program changes | | | | | | $0 |

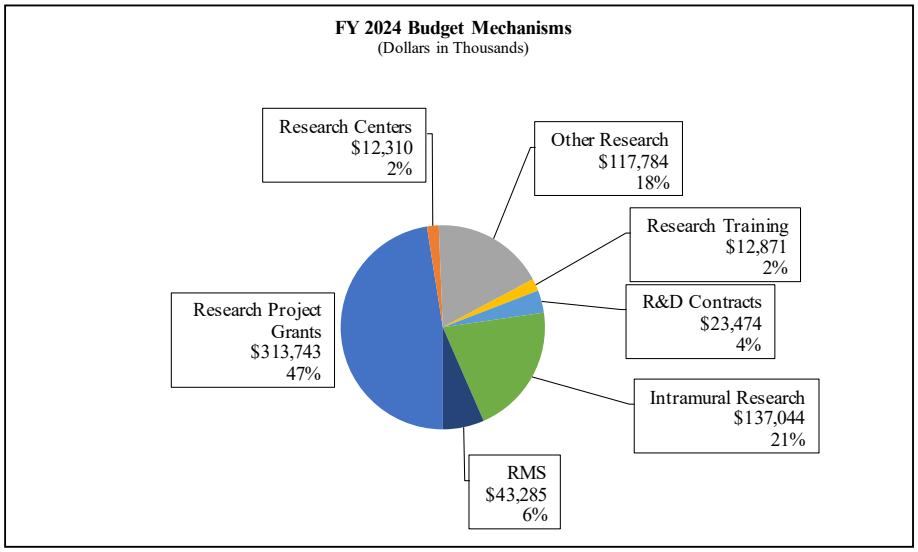
# Fiscal Year 2024 Budget Graphs

## History of Budget Authority and FTEs:

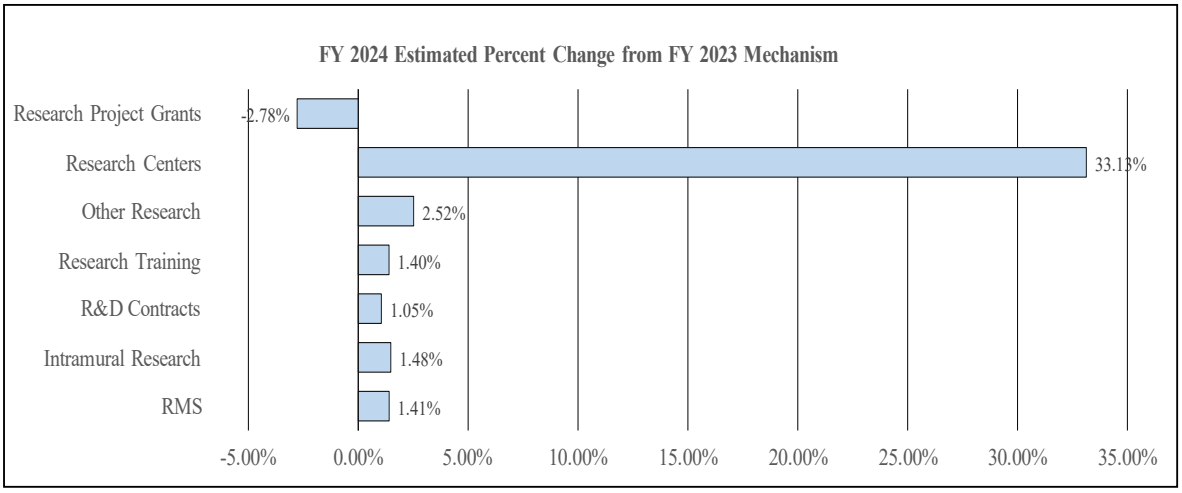


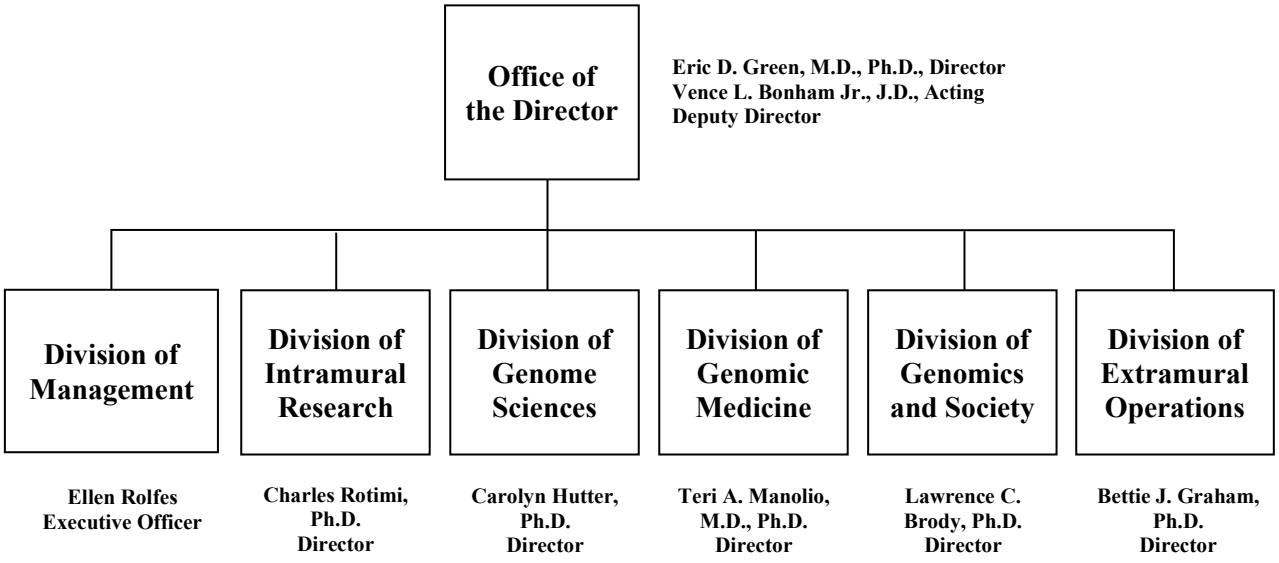

## Distribution by Mechanism:




## Change by Selected Mechanisms:



# NATIONAL HUMAN GENOME RESEARCH INSTITUTE
## Organizational Structure



**NATIONAL INSTITUTES OF HEALTH**
**National Human Genome Research Institute**

**Budget Authority by Activity***
(Dollars in Thousands)

| | FY 2022 Final | | FY 2023 Enacted | | FY 2024 President's Budget | | FY 2024 +/- FY 2023 Enacted | |
|---|---|---|---|---|---|---|---|---|
| **Extramural Research** | **FTE** | **Amount** | **FTE** | **Amount** | **FTE** | **Amount** | **FTE** | **Amount** |
| Detail | | | | | | | | |
| Genome Biology and Technology | | $191,856 | | $198,919 | | $198,282 | | -$637 |
| Genomics and Disease | | $144,707 | | $149,971 | | $150,069 | | $98 |
| Genomics in Medicine | | $105,027 | | $108,858 | | $108,841 | | -$16 |
| Genomic Data Sciences | | $95,780 | | $99,276 | | $99,232 | | -$45 |
| Genomics Workforce | | $27,086 | | $28,071 | | $28,093 | | $23 |
| Genomics and Society | | $31,577 | | $32,730 | | $32,708 | | -$22 |
| **Subtotal, Program Activity[1]** | | **$596,032** | | **$617,825** | | **$617,225** | | **-$600** |
| *(Extramural Research (non-add))* | *(0)* | *($465,529)* | *(0)* | *($482,781)* | *(0)* | *($480,181)* | *(0)* | *-($2,600)* |
| *(Intramural Research (non-add))* | *(237)* | *($130,504)* | *(258)* | *($135,044)* | *(258)* | *($137,044)* | *(0)* | *($2,000)* |
| **Research Management & Support** | **115** | **$40,447** | **127** | **$42,685** | **127** | **$43,285** | **0** | **$600** |
| **TOTAL** | **352** | **$636,479** | **385** | **$660,510** | **385** | **$660,510** | **0** | **$0** |

* Includes FTEs whose payroll obligations are supported by the NIH Common Fund.

[1] The detail programs listed above include both Extramural and Intramural funding.

## National Human Genome Research Institute

Authorizing Legislation: Section 301 and Title IV of the Public Health Service Act, as amended.

Budget Authority (BA):

| | FY 2022 Final | FY 2023 Enacted | FY 2024 President's Budget | FY 2024 +/- FY 2023 |
|---|---|---|---|---|
| BA | $636,479,000 | $660,510,000 | $660,510,000 | $0 |
| FTE | 352 | 385 | 385 | 0 |

Program funds are allocated as follows: Competitive Grants/Cooperative Agreements; Contracts; Direct Federal/Intramural and Other.

Overall Budget Policy: The FY 2024 President's Budget request for NHGRI is $660.5 million, which is the same as the FY 2023 Enacted level.

## Program Descriptions

**Genome Biology and Technology:** The leaders of the Telomere-to-Telomere Consortium (the group responsible for generating the first truly complete sequence of a human genome) were named to *Time* magazine's 100 Most Influential People of 2022. This contribution was made possible through NHGRI's decades of investments in DNA sequencing technology development, which has reduced the cost of genome sequencing by over a million-fold since the end of the Human Genome Project. NHGRI continues to play a leading role in supporting the research that produces important technological advances in DNA sequencing via the Genome Technology Program and Small Business Innovation Research (SBIR) and Small Business Technology Transfer (STTR) grants. The Genome Technology Program has also expanded its focus beyond nucleic acid sequencing to a wider set of efforts, including the development of single-molecule protein-sequencing technologies.

Recognizing the fundamental importance of having representative high-quality genome sequences from many different human populations, the NHGRI-supported Human Genome Reference Program (HGRP) continues to generate reference human genome sequences that represent an increasingly diverse set of human populations. Initial high-quality genome-sequence data from 45 individuals are now available in multiple repositories. The HGRP has also expanded outreach with potential international partners, including the Global Alliance for Genomics and Health (GA4GH), the Human Hereditary and Health in Africa (H3Africa) Consortium, and others; these efforts will continue in fiscal year (FY) 2024.

In order to unlock the potential for scientific discovery that comes with the generation of high-quality genome sequences, it is critically important to invest in efforts to detect and characterize

all functional elements in human DNA. NHGRI continues to be a leader in this core area of genomics. One way to glean insights about the functional landscape of the human genomes is to perform comparative studies of the genomes of various non-human organisms. NHGRI's Comparative Genomics Program is sequencing the genomes of species across the tree of life in order to characterize the genomic similarities and differences that have emerged through millions of years of evolution. Comparative genomics also helps scientists identify similarities among species in order to identify the appropriate models for studying specific human developmental processes and stages. Comparative genomics can also contribute to understanding tissue-specific gene expression at distinct developmental stages, the focus of the Non-Human Primate Developmental Genotype-Tissue Expression (NHP dGTEx) Project that is being conducted in collaboration with the National Institute of Mental Health (NIMH) and the Office of Research Infrastructure Programs (ORIP). The recently funded Multispecies NHP dGTEx Research Center will provide high-quality tissue samples, generate genome-sequence and RNA sequencing data, manage data-quality control, analyze gene-expression patterns in both bulk tissues and single cells, and ensure that the data and samples are accessible to the research community. Through its Comparative Genomics Program and other initiatives, NHGRI will continue to support efforts to compare the human genome with that of model and other non-human organisms in FY 2024 and beyond.

NHGRI's Impact of Genomic Variation on Function (IGVF) Program seeks to build on the successes of the Encyclopedia of DNA Elements (ENCODE) Project by systematically examining the impact of genomic variation on genome function and phenotypes. Through the funding of 26 grants, IGVF researchers are using a variety of high-throughput experimental and computational approaches to collaboratively investigate the links between genomic functional elements, genomic variants, and human and cellular phenotypes. The IGVF Consortium also aims to develop an interactive catalog containing information about the biological impact of genomic variants, which will serve as a resource for the research community. In FY 2024, IGVF will continue developing relationships and collaborations with other consortia.

Gaining a fundamental understanding of genome biology at different life stages will be important for using genomics in medicine. NHGRI is a key contributor to NIH Common Fund efforts in this area, such as the Human BioMolecular Atlas Program (HuBMAP) and the 4D Nucleome (4DN) Program, both of which seek to understand how genomes operate across time and space. To ensure that investigators throughout the research community have access to technologies that allow for robust genomic analyses, NHGRI continues to support the Center for Inherited Disease Research (CIDR). In FY 2024, CIDR will continue its mission to provide cutting-edge genomic services to NIH-funded investigators, including large consortia.

Budget Policy: The FY 2024 President's Budget request for Genome Biology and Technology is $198.3 million, a decrease of $0.6 million or 0.3 percent from the FY 2023 Enacted level. With these funds, NHGRI will continue to support the development of novel nucleic acid sequencing technologies and data-analysis methods. NHGRI will also continue to fund the efforts of the Human Genome Reference Program to produce reference human genome sequences that are more representative of humanity.

**Genomics and Disease:** The path to realizing the promise of genomic medicine requires an ever-growing understanding of both how genomic variation influences biological function and how these effects contribute to human disease. The set of Bold Predictions for Human Genomics by 2030 in the 2020 NHGRI Strategic Vision included one stating that: "The biological function(s) of every human gene will be known; for non-coding elements in the human genome, such knowledge will be the rule, rather than the exception." A powerful way to understand the function of a gene is to study what happens when it is inactivated – or "knocked out." To make progress en route to this prediction, NHGRI recently launched the Molecular Phenotypes of Null Alleles in Cells (MorPhiC) Program, which aims to develop a catalog of molecular and cellular phenotypes that result by inactivating each human gene. In its first five-year phase, which will include important early work in FY 2024, MorPhiC will optimize available methods to knock out 1,000 genes; evaluate the utility of various cell lines and organoids; explore multiple molecular and cellular assays; develop analyses to understand the utility of, and requirements for, the data; and disseminate the results to the research community.

NHGRI's Centers of Excellence in Genomic Science (CEGS) program aims to develop transformative approaches that address important biological and biomedical research problems and advance genomic science. Each CEGS engages a team of interdisciplinary scientists, ranging from basic and clinical researchers to engineers and ethicists, to develop novel approaches and concepts that improve the ability of researchers to produce, analyze, integrate, and use genomic data. In FY 2020, the NHGRI budget included an additional $10 million allocation for emerging Centers of Excellence in Genomic Science (eCEGS), targeting institutions that had not previously received a CEGS award. In FY 2021, this funding increased to $12.5 million; in FY 2022, it increased an additional $2.5 million.

**Genomics Research to Elucidate the Genetics of Rare Diseases (GREGoR)**

The Human Genome Project's generation of the first human genome sequence in 2003 set the stage for studies aimed at identifying all of the encoded genes and at establishing which ones play a role in health and disease. A high-priority focus was placed on rare genetic diseases. A rare disease is "a disease or condition that affects fewer than 200,000 people in the United States" *(Orphan Drug Act)*. Given their rare prevalence, the study of these diseases can be particularly challenging. However, at a genomic level, these disorders tend to be relatively straightforward to study since each disease is almost always caused by genomic variants (i.e., mutations) that disrupt the function of a single gene. So, characterizing a rare disease most often involves searching for the one 'causative' gene.

NHGRI first launched a major program to systematically identify the causative genes for rare diseases in 2015: the Centers for Mendelian Genomics program. The efforts of these Centers resulted in the successful identification of the causative gene for approximately 6,000 Mendelian phenotypes, a truly monumental advance. However, an estimated one third of rare diseases cases remain unsolved at a genomic level and likely require a more robust programmatic strategy.

Towards that end, the Genomics Research to Elucidate the Genetics of Rare Diseases (GREGoR) Consortium was established in 2021 with the aim of identifying the causal genomic variants and genes for the remaining rare diseases that have to date escaped such identification. Through GREGoR, five research centers and a data coordinating center are working to link diseases to their genomic causes. To accomplish their goals, GREGoR engages in an extensive collaborations among their researchers and brings together experts to apply new genome-sequencing technologies and analytical approaches. Beyond elucidating the genomic underpinning of rare diseases, the methods and approaches developed by GREGoR will advance genomic technologies and aid the diagnosis of other diseases that are challenging to characterize.

Genomic variation is increasingly being associated with human phenotypes (including diseases). The resulting knowledge can often be used in clinical care, both to help diagnose inherited disorders and to inform treatment. The NHGRI-funded Centers for Mendelian Genomics (CMG) program previously led efforts to discover the genomic bases of rare diseases, reporting a total of 3,617 gene-disease pairs since the program's inception in FY 2011. In FY 2021, NHGRI launched the Genomics Research Elucidates Genetics of Rare Disease (GREGoR) Consortium, which aims to develop new approaches to elucidate the bases of those remaining rare diseases which to date have eluded efforts to identify the mutated gene (see Program Portrait).

NHGRI is committed to being a leader in multi-omics research. In FY 2022, NHGRI published three funding opportunities in collaboration with the National Institute of Environmental Health Sciences (NIEHS) and the National Cancer Institute (NCI), to establish the Multi-Omics for Health and Disease Consortium. In FY 2024, this consortium will work to advance the application of multi-omic technologies to study health and disease in ancestrally diverse populations.

Elucidating the genomic bases of some conditions, particularly certain childhood disorders, requires examining patterns of gene expression during human development. Launched in FY 2021, the Developmental Genotype-Tissue Expression (dGTEx) project is co-funded by NHGRI, the *Eunice Kennedy Shriver* National Institute for Child Health and Human Development (NICHD), the National Institute of Neurological Disorders and Stroke (NINDS), and NIMH. The project is studying gene-expression patterns in the first 18 years of life by analyzing tissues from recently deceased pediatric donors. In FY 2024, the dGTEx resource will be a powerful tool that provides a comprehensive dataset of gene expression across a wide range of human tissues throughout development, filling a gap in genomic databases across developmental stages.

Budget Policy: The FY 2024 President's Budget request for Genomics and Disease is $150.1 million, an increase of $0.1 million or 0.1 percent from the FY 2023 Enacted level. These funds will support the CEGS/eCEGS program's efforts in human genomics, as well as the maintenance and expanded capacity of critical genomic databases, catalogs, and knowledge bases that facilitate the use of genomics for studying human disease.

**Genomics and Medicine:** While a foundational understanding of how genomic variation contributes to human disease is essential to realizing the promise of genomic medicine, successful implementation of genomic medicine will only occur following the robust study of how best to integrate genomics into the healthcare ecosystem. NHGRI provides support for essential clinician resources and initiatives designed to bring innovations in genomic medicine implementation among many different real-world settings.

The Clinical Genome Resource (ClinGen) curates information about the relationship between genomic variants and human disease, develops consensus approaches to genomic variant interpretation and classification, and disseminates the resulting information to the genomics and medical communities. In FY 2021, ClinGen was renewed for an additional five years, with co-funding from NCI. In FY 2022, in collaboration with the H3Africa Rare Disease Working

**Intramural Gene Therapy Research**

Sustained progress in genomics since the Human Genome Project has yielded numerous insights into the genomic bases of human disease. With the foundational knowledge provided by direct links between individual genes and specific diseases, researchers can turn their attention to developing strategies that use gene therapy as a therapeutic option. This area of research has many components, including designing and efficiently manufacturing the gene therapy vectors at scale as well as making the treatment protocol affordable. NHGRI's Intramural Research Program has created robust ecosystem for designing and testing gene therapy strategies.

With over 65 years of service, the NIH Clinical Center has served as a central nexus for physician scientists pursuing clinical research, including NHGRI's gene therapy researchers. Importantly, this setting optimally enables gene therapy researchers to concurrently care for their patients, consult with regulatory authorities, and perform well-designed clinical trials. Clinical studies conducted in the NIH Clinical Center are appropriately streamlined to minimize administrative and financial burdens, helping to test new therapies faster than in many other settings. The resulting successes in turn entice other stakeholders—including private companies and nonprofit research institutes—to collaborate with intramural researchers to develop and test therapeutics en route to their broader clinical use.

Several groups in NHGRI's Intramural Research Program are using the NIH Clinical Center in a highly productive way to make major contributions in gene therapy research—from advances in vector design to animal models and clinical trials. For example, the Platform Vector Gene Therapy (PaVe-GT) program, a joint initiative involving NICHD, the National Center for Advancing Translational Sciences (NCATS), the National Institute of Neurological Disorders and Stroke (NINDS), and NHGRI, is examining how the use of a standardized gene therapy delivery approach and manufacturing system can accelerate rare disease clinical trials. By standardizing experimental approaches, PaVe-GT researchers can examine how identical gene therapy vectors differ in efficacy depending on the disease being treated. From this, they can create a plug-and-play roadmap to advance targeted cures for other rare diseases.

Group and the GREGoR Consortium, ClinGen hosted a workshop to provide both didactic and hands-on sessions using real genome-sequence data, which taught users how to perform genomic analyses. The workshop also provided an opportunity to become familiar with other online genomic resources and how to utilize tools on the NHGRI Genomic Analysis, Visualization, and Informatics Lab-space (AnVIL) platform. ClinGen's Ancestry and Diversity Working Group also continued its effort to develop standards and guidelines for the collection, analysis, and reporting of diversity measures in clinical genetics and genomics. In FY 2024, ClinGen will continue to improve upon this essential resource for the research and clinical communities.

Although rare monogenic disorders collectively represent a significant public health burden, the most common diseases afflicting the United States and worldwide population (e.g., heart disease and diabetes) are caused by complex interactions that involve many genomic variants in addition to physical and social environments. Realizing the promise of genomic medicine for these common diseases requires improved methods for genomic risk prediction that can be used for all populations. A polygenic risk score (PRS) is calculated based on the total number of disease-associated genomic variants that a person harbors, and it can be used to assess a person's risk for developing a certain disease. However, at present, these risk scores are less applicable to non-European individuals due to the dearth of genomic data for these populations. The PRIMED Consortium, supported by NHGRI and NCI, is working to improve PRS usage by studying larger numbers of non-European individuals. The PRIMED Consortium will analyze over 120 datasets from 45 countries with the goal of improving the ability of using PRSs to predict disease in diverse populations of individuals.

NHGRI is dedicated to ensuring that genomics improves human health by building on discoveries made through genomic studies and applying them in real-world clinical settings. Building on the successful genomic medicine projects during the initial phase of Implementing Genomics in Practice (IGNITE I), the IGNITE Pragmatic Trials Network (PTN) works to actively implement genomic medicine in clinical settings with diverse populations. IGNITE PTN (or IGNITE II) supports a network of multi-site clinical groups and is conducting two pragmatic clinical trials of genomic medicine interventions: Genetic Testing to Understand and Address Renal Disease Disparities Across the United States **(**GUARDD-US) and A Depression and Opioid Pragmatic Trial in Pharmacogenetics **(**ADOPT-PGx). GUARDD-US has enrolled 6,650 patients as of July 2022. ADOPT-PGx is comparing genotype-guided drug therapy versus conventional approaches to drug therapy selection for acute post-surgical pain, chronic pain, and depression via changes in patient-reported outcomes representing pain and depression control. ADOPT-PGx has accrued over 1,600 patients as of July 2022. The information generated from these trials will contribute to the growing knowledge of using genomic information in patient care.

Investigators in NHGRI's Intramural Research Program are working to develop targeted preventions and cures by spearheading advances in gene therapy (see Program Portrait). NHGRI researchers leverage the unique resources and setting of the NIH Clinical Center to develop new diagnostic tests and treatments for patients with rare genetic disorders, including the design and conduct of gene-therapy trials. In FY 2024, foundational investments in basic science and clinical research remain essential to demonstrating proof-of-concept gene-therapy strategies that facilitate partnerships and enable the scalability necessary to make targeted cures accessible to patients.

Budget Policy: The FY 2024 President's Budget request for Genomics and Medicine is $108.8 million, a decrease of $16,000 or 0.02 percent from the FY 2023 Enacted level. In FY 2024, ClinGen will continue to advance the implementation of genomic medicine by supporting the successful integration of genomics into clinical decision making and care. Using FY 2024 funds, IGNITE will continue to pioneer and study real-world applications of genomics in healthcare to improve patient outcomes, and PRIMED will work to improve the predictive ability and responsible use of PRSs to assess disease risk in diverse populations.

**Genomic Data Science:** The field of genomic data science enables researchers to use powerful computational and statistical methods to decode the fundamental information within DNA sequences, and then apply that information to help understand genome function and implement genomic medicine. NHGRI is a leader in robust and coordinated genomic data science efforts, including those involved in managing increasing amounts of genomic and clinical data (see Program Portrait). NHGRI-supported research in genomic data science includes developing computational tools and methods for analyzing genomic data; establishing and maintaining genomic data resources accessible to scientists and clinicians worldwide; and developing tools that facilitate clinical decision-making in genomic medicine. NHGRI's new Office of Genomic

Data Science (OGDS) provides leadership, strategic guidance, and coordination for NHGRI activities, programs, and policies in genomic data science.

**Modernization of Genomic Data Science at NHGRI**

Advances in data science and bioinformatics are regularly providing new opportunities to extract insights from genomic data in truly unprecedented ways. These efforts are increasingly being referred to as genomic data science and are now central to how genomics is driving scientific discoveries and being used in medicine.

The 2020 NHGRI Strategic Vision and NIH Strategic Plan for Data Science outline the importance of sustaining and improving a robust foundation for genomics, including the vital role that genomic data science now plays in myriad areas of biomedical research and genomic medicine. Beyond the investment of $150 million per year for genomic data science-related endeavors, NHGRI has been proactive in ensuring the ethical, legal, and societal implications of genomic data sharing are considered. The Institute is also taking steps to ensure it is well-prepared to meet the growing challenges in genomic data science, including implementing internal organizational changes through establishment of a new office.

The NHGRI Office of Genomic Data Science (OGDS) was established in 2022 to coordinate the Institute's genomic data science activities; act as a coordinating hub for NHGRI in trans-NIH, national, and international data science programs; and provide guidance for NHGRI's implementation of NIH data-sharing policies. For example, with the new NIH Data Management and Sharing Policy, researchers must navigate the expectation of sharing data while balancing privacy and confidentiality concerns. OGDS is helping to streamline genomic data sharing and access for genomics researchers and other stakeholders. Another major OGDS responsibility is programmatic leadership of the Genomic Analysis, Visualization and Informatics Lab-space (AnVIL), which is a cloud-based genomic data sharing and analysis platform that facilitates computing on datasets generated by NHGRI and NIH initiatives. OGDS also organizes professional-development initiatives that will help in the building of more diverse genomic data science workforce. Through their leadership, NHGRI and OGDS are prioritizing genomic data science to achieve a healthier society.

NHGRI's Computational Genomics and Data Science (CGDS) Program supports the development of improved computational approaches, innovative data-analysis tools, and data resources that provide scientific utility across all of NHGRI extramural research programs and in alignment with the NIH Strategic Plan for Data Science. Grants and initiatives supported by the CGDS program include AnVIL; the Alliance of Genome Resources (the Alliance); GA4GH; and a set of funding opportunities supporting investigator-initiated research and workforce development efforts. One such effort is the recently launched Educational Hub for Enhancing Diversity in Computational Genomics and Data Science, which is an expansion of the NHGRI-funded Genomic Data Science Community Network (GDSCN); this new program leverages AnVIL and other NIH cloud-based platforms to enhance the diversity of individuals who have access to educational and research opportunities in computational genomics and data science, including those from underrepresented groups. In FY 2024, NHGRI will remain committed to supporting training, research experiences and career development to integrate data scientists into the genomics workforce.

Since its launch in June 2019, AnVIL has onboarded over four and a half petabytes of data, and that number continues to grow. The AnVIL program was renewed in FY 2022 and continues to serve as a scalable and interoperable resource for the basic and clinical genomic research communities by leveraging a cloud-based infrastructure to democratize data access, sharing, and computing across large genomic and genomic-related datasets. As a founding member of the NIH Cloud Platforms Interoperability (NCPI) program, the AnVIL program continues to play a leading role in

trans-NIH activities to facilitate interoperability and maintain communication among NIH cloud-based genomic resources, such as with the NCI Cancer Research Data Commons (CRDC), the NHLBI BioData Catalyst, and the Gabriella Miller Kids First Pediatric Research Program.

In October 2021, NHGRI hosted a virtual workshop entitled "Future Directions of the NHGRI Analysis, Visualization, and Informatics Lab-space (AnVIL)," a gathering that aimed to identify gaps, challenges, and future opportunities related to NHGRI's investments in the AnVIL program. Outcomes from these workshops highlighted support for AnVIL's existing infrastructure and services and identified opportunities to expand and diversify AnVIL activities to support the basic and clinical genomics research communities. The AnVIL program is committed to these activities in FY 2024.

NHGRI also demonstrates its commitment to genomic data science and responsible data sharing via partnerships within the Institute, across NIH, and beyond. The NHGRI Genomic Data Science Working Group — a working group of the National Advisory Council for Human Genome Research — will continue to advise Institute leadership in FY 2024, addressing broad challenges, such as data management, data analysis, biocomputing, and data science policy. NHGRI also plays a leading role in the NIH Data Science Policy Council (DSPC), which aims to address the growing policy challenges and opportunities associated with "big data" and data science in biomedical research and to promote maximum public benefit from data utilization in a responsible manner. In FY 2024, NHGRI will continue to support the strong data-science pursuits of the NIH Intramural Sequencing Center, which serves the entire NIH intramural research community by providing the infrastructure and expertise for bringing genome sequencing to biology and medicine. NHGRI will also continue to support resources, knowledgebases, and repositories that serve the entire genomics community.

Budget Policy: The FY 2024 President's Budget request for Genomic Data Science is $99.2 million, a decrease of $45,000 or 0.04 percent from the FY 2023 Enacted level. With FY 2024 funds, NHGRI will continue to push the frontiers of data-analyses technologies. AnVIL will continue to provide a cutting-edge resource for the storage, analysis, and sharing of genomic data.

**Genomics Workforce:** Attracting, developing, and retaining a diverse and talented genomics workforce is a top priority for NHGRI and will be necessary to ensure that genomics-based breakthroughs improve human health. NHGRI recently established the TiDHE Office, which harmonizes the Institute's training and workforce-development efforts with its research initiatives in health disparities and health equity. The TiDHE Office oversees the implementation of the NHGRI Action Agenda for Genomics Workforce Diversity, which outlines an ambitious set of goals and objectives towards establishing a more inclusive genomics workforce. The TiDHE Office also provides NHGRI leadership within NIH-wide efforts, such as the UNITE Initiative, and partners with professional societies, the National Academies of Sciences, Engineering and Medicine (NASEM), and industry to promote greater diversity in the genomics workforce. Through the efforts of the TiDHE Office, NHGRI will continue to be a leader in fostering a diverse and talented genomics workforce in FY 2024 and beyond.

NHGRI has a series of funding opportunities focused on genomics workforce diversity that will remain active in FY 2024. A new extramurally funded program, the Diversity Centers for Genome Research, aims to increase the diversity of the genomics workforce by funding innovative research projects, genomics education, and training at minority-serving institutions with a mission to serve historically underrepresented populations. The Genome Research Experiences to Attract Talented Undergraduates into the Genomics Field to Enhance Diversity (GREAT) program supports educational activities encouraging undergraduates from diverse backgrounds, including those from groups underrepresented in the biomedical workforce, to pursue training and careers in the scientific, medical, ethical, social, and/or legal areas of genomics. The Faculty Institutional Recruitment for Sustainable Transformation (or FIRST) program, a NIH Common Fund initiative, is designed to facilitate building a self-reinforcing community of scientists at institutions by recruiting early-career faculty who have demonstrated a commitment to inclusive excellence. Finally, the NIH Science Education Partnership Award (SEPA) program supports pre-kindergarten to grade 12 as well as informal science education activities that both enhance the diversity of the biomedical, behavioral, and clinical research workforce and foster a better understanding of NIH-funded biomedical, behavioral, and clinical research and its public health implications. NHGRI has developed a new awards program — the Genomics Workforce DEIA Award — to recognize extramural investigators who have made sustained and substantial contributions to enhancing diversity, equity, inclusion, and accessibility (DEIA) in the genomics workforce.

In FY 2021, NHGRI launched a new collaborative training opportunity with the United States Food and Drug Administration (FDA), and the Center for Disease Control and Preventions's Office of Minority Health and Health Equity (CDC OMHHE): the Postdoctoral Fellowship in Genomic Science and Health Equity. This fellowship program is designed to prepare fellows to use genetic, genomic, and pharmacogenomic approaches to advance minority health and health equity; in addition, it will train individuals in the research methodology and medical product development processes that facilitate the delivery of drugs, biologics and devices from bench to bedside. Fellows will gain unique experiences by working closely with mentors at both the FDA and NIH, including the opportunity to advance their knowledge of health disparity and regulatory sciences.

NHGRI also continues to fund the Genomic Innovator Awards and — in partnership with the American Society of Human Genetics — the Human Genetics Scholars Initiative (HGI); both programs support early-career researchers. In FY 2022, NHGRI launched a new seminar series based on its Genomic Innovator Award Program, pairing an early-career researcher with a more established researcher for each seminar. These seminars will cover specific topics that showcase the creative ways that early-career investigators are advancing genomics research.

In addition to supporting summer interns, post-baccalaureate fellows, graduate students, and postdoctoral fellows of all backgrounds, the NHGRI Intramural Research Program participates in the NIH Office of Intramural Training and Education's High School Scientific Training and Enrichment Program 2.0 (HiSTEP 2.0), which is offered to high school seniors who have little research experience, providing them the opportunity to perform biomedical research at NIH. NHGRI's Education and Community Involvement Branch (ECIB) provides genomics education and training resources for healthcare providers, tribal colleges, high school teachers, and other

professionals. NHGRI remains committed to supporting the training of the next generation of genomics professionals in FY 2024 and beyond.

Budget Policy: The FY 2024 President's Budget request for Genomics Workforce is $28.1 million, an increase of $23,000 or 0.1 percent from the FY 2023 Enacted level. These funds will be used by NHGRI to continue supporting diverse trainees in genomics at all stages of their careers and to aid in the development of early-stage investigators.

**Genomics and Society:** The 2020 NHGRI Strategic Vision outlined a set of principles and values for human genomics, and these have been heavily informed by a lengthy history of ethical, legal, and social implications (ELSI) research. In fact, NHGRI dedicates at least five percent of its research budget to ELSI research. While the origins of ELSI research date back to the beginning of the Human Genome Project, this area of research has grown to include a large portfolio of studies that examine the intersection of genomics, law, ethics, philosophy, and society.

The NHGRI ELSI Research Program funds research studies, training opportunities, and workshops. It also develops and supports research consortia and conferences in four broad areas: (1) Genomics and Sociocultural Structures and Values; (2) Genomics at the Institutional and System Level; (3) Genomic Research Design and Implementation; and (4) Genomic Healthcare. The ELSI Research Program has a particular focus on studies that explore these issues with and within communities that have been underrepresented, underserved, and/or mistreated in biomedical research and healthcare.

In addition to its three active program announcements, the ELSI Research Program supports the Centers of Excellence in ELSI Research (CEER) program, which funds four research centers at universities across the country, and the Center for ELSI Resources and Analysis (CERA), which assists in establishing a more integrated ELSI research community. In July 2019, the ELSI Research Program funded a conference grant to support the biennial ELSI Research Congress. The fifth ELSI Congress, *ELSIcon2022: Innovating for a Just and Equitable Future*, was held virtually on May 31 – June 3, 2022; the next ELSI Congress will be held in 2024. The ELSI Research Program also helps support ELSI research that is embedded within other NHGRI programs and projects, such as dGTEx, the PRIMED consortium, and the Human Pangenome Reference Consortium.

Budget Policy: The FY 2024 President's Budget request for Genomics and Society is $32.7 million, a decrease of $22,000 or 0.1 percent from the FY 2023 Enacted level. In FY 2024, NHGRI will continue to fund ELSI research that is critical for ensuring ethical and successful advances in genomics.

**Research Management and Support:** Within NHGRI's Division of Genomics and Society and alongside the ELSI Research Program, the Policy and Program Analysis Branch (PPAB) and ECIB use Research Management and Support funds to support NHGRI's mission of furthering genetics and genomics research by engaging with policymakers, stakeholders, educators, researchers, clinicians, and the public.

PPAB supports NHGRI in a broad range of activities, including Congressional relations; program and portfolio analysis; policy tracking, dissemination, analysis, and development; stakeholder engagement; and resource development for both scientific and lay audiences. In FY 2022, PPAB created an educational resource to help understand the FDA Investigational Device Exemption (IDE) regulation in the context of genomics. PPAB also recently spearheaded an update to NHGRI's informed consent webpages, which provide vital information about considerations specific to genomics research and contributing genomic data. In FY 2024, PPAB will continue to focus its portfolio to address emerging policy issues in genomics, such as data sharing, human subjects research, legal and regulatory landscapes, the implementation of genomic medicine, and other salient issues.

ECIB works with a range of audiences to provide educational resources about the ever-changing landscape of genetics and genomics. Led by ECIB, National DNA Day is an annual community-driven initiative that provides resources to the public, students at all levels, and educators to inspire everyone to learn about genomics. In FY 2022, ECIB hosted the annual Short Course in Genomics, which enables high school and community college science educators to hear lectures and receive teaching resources from leading researchers, clinicians, and NIH staff. To enrich the experience of NHGRI trainees, ECIB partners with the Intramural Training Office to provide opportunities in program management, training, and community outreach. ECIB also hosts the Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG) to create resources and promote collaborations among healthcare professionals. Among ISCC-PEG initiatives is the Scholars Program, which pairs trainees with mentors to research and create products in genomics education. ISCC-PEG also supports GenomeEd, an updated repository of genomics education resources built on the foundation of the Genetics/Genomics Competency Center (G2C2). ECIB will continue to lead programs and initiatives to promote the engagement of diverse communities in understanding genomics and its translation to health and society.

NHGRI's Office of Communications (OC) creates media in the form of written articles, videos, graphics, and social media posts to provide high-quality and timely genomics information to the public. OC also serves as the point of contact for all news media requests, inquiries from the scientific community, and questions from the public. In FY 2022, OC continued its virtual Genomics and the Media Seminar Series, featuring leaders in science communication in all forms of media. On National DNA Day in FY 2022, OC released a fully revamped version of its popular Talking Glossary under a new name: the "Talking Glossary of Genomic and Genetic Terms." The revamped Talking Glossary has 222 terms, 142 of which have new accompanying illustrations and 15 of which have new associated animations. This educational resource features audio recordings from 37 NHGRI experts. OC was also recently nominated for a Golden Post Award for its Twitter presence; this award recognizes outstanding use of social media by government agencies in the United States. Out of more than 400 entries, NHGRI was named 1 of the top 5 finalists.

In FY 2022, the NHGRI History of Genomics Program, a component of OC, hosted a symposium that examined the history of eugenics and scientific racism and their complex legacies in the modern health sciences. The symposium illustrated the need for more informed public discussions about the connection between existing and emerging genetic and genomic technologies and the historical and present-day uses of genetics and genomics in social and

behavioral research. The History of Genomics Program followed that symposium with two roundtable discussions: one that explored historical and present-day eugenics and scientific racism in the context of existing and developing genetic and genomic screening technologies and one that addressed the promise and perils of social and behavioral genomics research.

In FY 2024, PPAB, ECIB, and OC will continue to work in concert to promote sound policies, perform outreach and stakeholder engagement, and keep the public informed about the latest developments in genomics and genomic medicine.

Budget Policy: The FY 2024 President's Budget request for Research Management and Support is $43.3 million, an increase of $0.6 million or 1.1 percent from the FY 2023 CR level. These funds will be used to support a number of outreach, education, and stakeholder-engagement initiatives in FY 2024.

# NATIONAL INSTITUTES OF HEALTH
## National Human Genome Research Institute

## Appropriations History

| Fiscal Year | Budget Estimate to Congress | House Allowance | Senate Allowance | Appropriation |
|---|---|---|---|---|
| 2015 | $498,451,000 | | | $499,356,000 |
| Rescission | | | | $0 |
| 2016 | $515,491,000 | $505,551,000 | $526,166,000 | $518,956,000 |
| Rescission | | | | $0 |
| 2017 [1] | $513,227,000 | $531,438,000 | $534,516,000 | $528,566,000 |
| Rescission | | | | $0 |
| 2018 | $399,622,000 | $536,774,000 | $546,934,000 | $556,881,000 |
| Rescission | | | | $0 |
| 2019 | $512,979,000 | $563,531,000 | $575,882,000 | $575,579,000 |
| Rescission | | | | $0 |
| 2020 | $495,448,000 | $603,710,000 | $607,999,000 | $606,349,000 |
| Rescission | | | | $0 |
| 2021 | $550,116,000 | $611,564,000 | $623,862,000 | $615,780,000 |
| Rescission | | | | $0 |
| 2022 | $632,973,000 | $646,295,000 | $634,598,000 | $639,062,000 |
| Rescission | | | | $0 |
| 2023 | $629,154,000 | $659,233,000 | $658,873,000 | $663,200,000 |
| Rescission | | | | $0 |
| 2024 | $660,510,000 | | | |

[1] Budget Estimate to Congress includes mandatory financing.

**NATIONAL INSTITUTES OF HEALTH**
**National Human Genome Research Institute**

**Authorizing Legislation**

| | PHS Act/ Other Citation | U.S. Code Citation | 2023 Amount Authorized | FY 2023 Enacted | 2024 Amount Authorized | FY 2024 President's Budget |
|---|---|---|---|---|---|---|
| Research and Investigation | Section 301 | 42§241 | Indefinite | $660,510,000 | Indefinite | $660,510,000 |
| National Human Genome Research Institute | Section 401(a) | 42§281 | Indefinite | | Indefinite | |
| Total, Budget Authority | | | | $660,510,000 | | $660,510,000 |

# NATIONAL INSTITUTES OF HEALTH

## National Human Genome Research Institute

### Amounts Available for Obligation [1]

(Dollars in Thousands)

| Source of Funding | FY 2022 Final | FY 2023 Enacted | FY 2024 President's Budget |
|---|---|---|---|
| Appropriation | $639,062 | $663,200 | $660,510 |
| Secretary's Transfer | $0 | $0 | $0 |
| OAR HIV/AIDS Transfers | -$2,583 | -$2,690 | $0 |
| Subtotal, adjusted budget authority | $636,479 | $660,510 | $660,510 |
| Unobligated balance, start of year | $0 | $0 | $0 |
| Unobligated balance, end of year (carryover) | $0 | $0 | $0 |
| **Subtotal, adjusted budget authority** | **$636,479** | **$660,510** | **$660,510** |
| Unobligated balance lapsing | -$45 | $0 | $0 |
| Total obligations | $636,434 | $660,510 | $660,510 |

[1] Excludes the following amounts (in thousands) for reimbursable activities carried out by this account:
FY 2022 - $28,801 FY 2023 - $30,978 FY 2024 - $31,938

**NATIONAL INSTITUTES OF HEALTH**
**National Human Genome Research Institute**

**Budget Authority by Object Class**[1]
(Dollars in Thousands)

| | | FY 2023 Enacted | FY 2024 President's Budget | FY 2024 +/- FY 2023 |
|---|---|---|---|---|
| **Total compensable workyears:** | | | | |
| | Full-time equivalent | 385 | 385 | 0 |
| | Full-time equivalent of overtime and holiday hours | 0 | 0 | 0 |
| | Average ES salary | $212 | $223 | $11 |
| | Average GM/GS grade | 12.8 | 12.8 | 0.0 |
| | Average GM/GS salary | $131 | $137 | $7 |
| | Average salary, Commissioned Corps (42 U.S.C. 207) | $129 | $136 | $7 |
| | Average salary of ungraded positions | $174 | $183 | $9 |
| | **OBJECT CLASSES** | **FY 2023 Enacted** | **FY 2024 President's Budget** | **FY 2024 +/- FY 2023** |
| | Personnel Compensation | | | |
| 11.1 | Full-Time Permanent | $25,452 | $26,841 | $1,388 |
| 11.3 | Other Than Full-Time Permanent | $21,126 | $22,279 | $1,152 |
| 11.5 | Other Personnel Compensation | $1,369 | $1,444 | $75 |
| 11.7 | Military Personnel | $232 | $245 | $13 |
| 11.8 | Special Personnel Services Payments | $6,291 | $6,634 | $343 |
| **11.9** | **Subtotal Personnel Compensation** | **$54,471** | **$57,442** | **$2,971** |
| 12.1 | Civilian Personnel Benefits | $17,686 | $18,572 | $885 |
| 12.2 | Military Personnel Benefits | $107 | $113 | $6 |
| 13.0 | Benefits to Former Personnel | $0 | $0 | $0 |
| | **Subtotal Pay Costs** | **$72,264** | **$76,126** | **$3,862** |
| 21.0 | Travel & Transportation of Persons | $568 | $582 | $14 |
| 22.0 | Transportation of Things | $234 | $240 | $6 |
| 23.1 | Rental Payments to GSA | $0 | $0 | $0 |
| 23.2 | Rental Payments to Others | $0 | $0 | $0 |
| 23.3 | Communications, Utilities & Misc. Charges | $92 | $94 | $2 |
| 24.0 | Printing & Reproduction | $12 | $12 | $0 |
| 25.1 | Consulting Services | $23,557 | $23,949 | $392 |
| 25.2 | Other Services | $19,453 | $18,799 | -$654 |
| 25.3 | Purchase of Goods and Services from Government Accounts | $62,149 | $61,871 | -$278 |
| 25.4 | Operation & Maintenance of Facilities | $314 | $314 | $1 |
| 25.5 | R&D Contracts | $1,032 | $1,056 | $25 |
| 25.6 | Medical Care | $428 | $445 | $18 |
| 25.7 | Operation & Maintenance of Equipment | $10,425 | $10,415 | -$10 |
| 25.8 | Subsistence & Support of Persons | $0 | $0 | $0 |
| **25.0** | **Subtotal Other Contractual Services** | **$117,356** | **$116,849** | **-$507** |
| 26.0 | Supplies & Materials | $6,628 | $6,786 | $158 |
| 31.0 | Equipment | $3,153 | $2,946 | -$207 |
| 32.0 | Land and Structures | $651 | $167 | -$484 |
| 33.0 | Investments & Loans | $0 | $0 | $0 |
| 41.0 | Grants, Subsidies & Contributions | $459,551 | $456,707 | -$2,844 |
| 42.0 | Insurance Claims & Indemnities | $0 | $0 | $0 |
| 43.0 | Interest & Dividends | $1 | $1 | $0 |
| 44.0 | Refunds | $0 | $0 | $0 |
| | **Subtotal Non-Pay Costs** | **$588,246** | **$584,384** | **-$3,862** |
| | **Total Budget Authority by Object Class** | **$660,510** | **$660,510** | **$0** |

[1] Includes FTEs whose payroll obligations are supported by the NIH Common Fund.

# NATIONAL INSTITUTES OF HEALTH

## National Human Genome Research Institute

### Salaries and Expenses

(Dollars in Thousands)

| Object Classes | FY 2023 Enacted | FY 2024 President's Budget | FY 2024 +/- FY 2023 |
|---|---|---|---|
| Personnel Compensation | | | |
| Full-Time Permanent (11.1) | $25,452 | $26,841 | $1,388 |
| Other Than Full-Time Permanent (11.3) | $21,126 | $22,279 | $1,152 |
| Other Personnel Compensation (11.5) | $1,369 | $1,444 | $75 |
| Military Personnel (11.7) | $232 | $245 | $13 |
| Special Personnel Services Payments (11.8) | $6,291 | $6,634 | $343 |
| **Subtotal, Personnel Compensation (11.9)** | **$54,471** | **$57,442** | **$2,971** |
| Civilian Personnel Benefits (12.1) | $17,686 | $18,572 | $885 |
| Military Personnel Benefits (12.2) | $107 | $113 | $6 |
| Benefits to Former Personnel (13.0) | $0 | $0 | $0 |
| **Subtotal Pay Costs** | **$72,264** | **$76,126** | **$3,862** |
| Travel & Transportation of Persons (21.0) | $568 | $582 | $14 |
| Transportation of Things (22.0) | $234 | $240 | $6 |
| Rental Payments to Others (23.2) | $0 | $0 | $0 |
| Communications, Utilities & Misc. Charges (23.3) | $92 | $94 | $2 |
| Printing & Reproduction (24.0) | $12 | $12 | $0 |
| Other Contractual Services | | | |
| Consultant Services (25.1) | $23,557 | $23,949 | $392 |
| Other Services (25.2) | $19,453 | $18,799 | -$654 |
| Purchase of Goods and Services from Government Accounts (25.3) | $43,759 | $43,442 | -$317 |
| Operation & Maintenance of Facilities (25.4) | $314 | $314 | $1 |
| Operation & Maintenance of Equipment (25.7) | $10,425 | $10,415 | -$10 |
| Subsistence & Support of Persons (25.8) | $0 | $0 | $0 |
| **Subtotal Other Contractual Services** | **$97,508** | **$96,919** | **-$589** |
| Supplies & Materials (26.0) | $6,628 | $6,786 | $158 |
| **Subtotal Non-Pay Costs** | **$105,042** | **$104,633** | **-$409** |
| **Total Administrative Costs** | **$177,305** | **$180,759** | **$3,454** |

## NATIONAL INSTITUTES OF HEALTH
## National Human Genome Research Institute

### Detail of Full-Time Equivalent Employment (FTE)

| Office | FY 2022 Final | | | FY 2023 Enacted | | | FY 2024 President's Budget | | |
|---|---|---|---|---|---|---|---|---|---|
| | Civilian | Military | Total | Civilian | Military | Total | Civilian | Military | Total |
| Division of Intramural Research | | | | | | | | | |
| Direct: | 200 | 1 | 201 | 218 | 1 | 219 | 218 | 1 | 219 |
| Reimbursable: | 34 | 2 | 36 | 37 | 2 | 39 | 37 | 2 | 39 |
| Total: | 234 | 3 | 237 | 255 | 3 | 258 | 255 | 3 | 258 |
| Office of the Director | | | | | | | | | |
| Direct: | 17 | - | 17 | 17 | - | 17 | 17 | - | 17 |
| Total: | 17 | - | 17 | 17 | - | 17 | 17 | - | 17 |
| Division of Management | | | | | | | | | |
| Direct: | 47 | - | 47 | 48 | - | 48 | 48 | - | 48 |
| Total: | 47 | - | 47 | 48 | - | 48 | 48 | - | 48 |
| Division of Genome Sciences | | | | | | | | | |
| Direct: | 10 | - | 10 | 13 | - | 13 | 13 | - | 13 |
| Reimbursable: | 3 | - | 3 | 3 | - | 3 | 3 | - | 3 |
| Total: | 13 | - | 13 | 16 | - | 16 | 16 | - | 16 |
| Division of Genomic Medicine | | | | | | | | | |
| Direct: | 11 | - | 11 | 14 | - | 14 | 14 | - | 14 |
| Reimbursable: | 1 | - | 1 | 2 | - | 2 | 2 | - | 2 |
| Total: | 12 | - | 12 | 16 | - | 16 | 16 | - | 16 |
| Division of Genomics and Society | | | | | | | | | |
| Direct: | 9 | - | 9 | 12 | - | 12 | 12 | - | 12 |
| Total: | 9 | - | 9 | 12 | - | 12 | 12 | - | 12 |
| Division of Extramural Operations | | | | | | | | | |
| Direct: | 16 | - | 16 | 17 | - | 17 | 17 | - | 17 |
| Reimbursable: | 1 | - | 1 | 1 | - | 1 | 1 | - | 1 |
| Total: | 17 | - | 17 | 18 | - | 18 | 18 | - | 18 |
| Total | 349 | 3 | 352 | 382 | 3 | 385 | 382 | 3 | 385 |
| Includes FTEs whose payroll obligations are supported by the NIH Common Fund. | | | | | | | | | |
| FTEs supported by funds from Cooperative Research and Development Agreements. | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |

| FISCAL YEAR | Average GS Grade |
|---|---|
| 2020 | 12.6 |
| 2021 | 12.7 |
| 2022 | 12.8 |
| 2023 | 12.8 |
| 2024 | 12.8 |

# NATIONAL INSTITUTES OF HEALTH
## National Human Genome Research Institute

### Detail of Positions[1]

| GRADE | FY 2022 Final | FY 2023 Enacted | FY 2024 President's Budget |
|---|---|---|---|
| Total, ES Positions | 1 | 1 | 1 |
| Total, ES Salary | $203,700 | $212,103 | $222,815 |
| General Schedule | | | |
| GM/GS-15 | 41 | 45 | 45 |
| GM/GS-14 | 35 | 40 | 40 |
| GM/GS-13 | 79 | 88 | 88 |
| GS-12 | 43 | 48 | 48 |
| GS-11 | 10 | 15 | 15 |
| GS-10 | 0 | 0 | 0 |
| GS-9 | 7 | 9 | 9 |
| GS-8 | 10 | 10 | 10 |
| GS-7 | 1 | 1 | 1 |
| GS-6 | 0 | 0 | 0 |
| GS-5 | 0 | 0 | 0 |
| GS-4 | 0 | 0 | 0 |
| GS-3 | 2 | 2 | 2 |
| GS-2 | 0 | 0 | 0 |
| GS-1 | 0 | 0 | 0 |
| Subtotal | 228 | 258 | 258 |
| Commissioned Corps (42 U.S.C. 207) | | | |
| Assistant Surgeon General | 0 | 0 | 0 |
| Director Grade | 2 | 2 | 2 |
| Senior Grade | 0 | 0 | 0 |
| Full Grade | 1 | 1 | 1 |
| Senior Assistant Grade | 0 | 0 | 0 |
| Assistant Grade | 0 | 0 | 0 |
| Subtotal | 3 | 3 | 3 |
| Ungraded | 140 | 145 | 145 |
| Total permanent positions | 232 | 262 | 262 |
| Total positions, end of year | 372 | 407 | 407 |
| Total full-time equivalent (FTE) employment, end of year | 352 | 385 | 385 |
| Average ES salary | $203,700 | $212,103 | $222,815 |
| Average GM/GS grade | 12.8 | 12.8 | 12.8 |
| Average GM/GS salary | $125,812 | $130,873 | $137,482 |

[1] Includes FTEs whose payroll obligations are supported by the NIH Common Fund.