

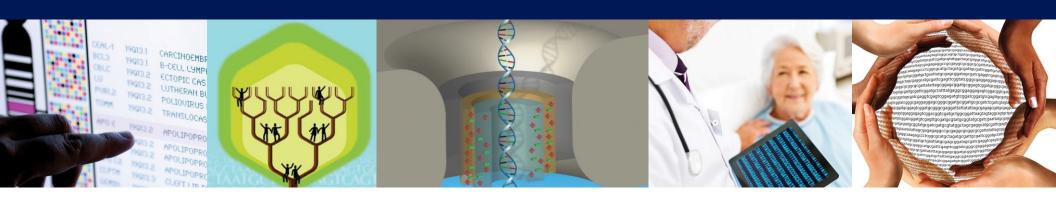


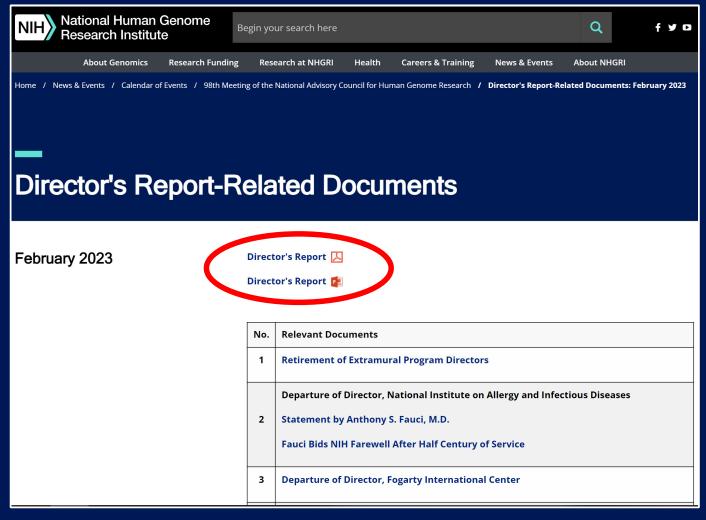


DIRECTOR'S REPORT

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

February 2023





genome.gov/DirectorsReport



Open Session Agenda

Presentations:

Building a Diverse Genomics Workforce Action Agenda: Implementation Plan and Progress Report Vence Bonham

Outstanding Awards for Diversity, Equity, Inclusion, and Accessibility (DEIA) in the Genomics Workforce Lorjetta Schools

Open Session Agenda

Concept Clearances:

ELSI Program Announcements for R01, R03, and R21 Applications

Dave Kaufman

Center for ELSI Resources and Analysis
Nicole Lockhart

Diversity Action Plan
Lucia Hindorff

Advancing Genomic Medicine Research Christine Chang

Human Genome Reference Program
Adam Felsenfeld

Open Session Agenda

Reports:

Genomic Medicine Working Group of Council: Update of Meetings, Activities, and Accomplishments

Teri Manolio

Genomic Data Science Working Group of Council Update Mark Craven

Community Engagement in Genomics Working Group of Council Update

Maya Sabatello & Greta Goto

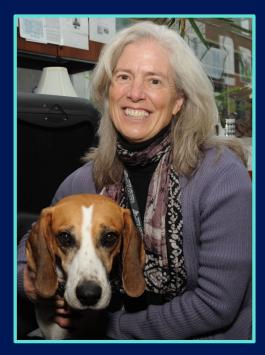
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- VII. NHGRI Intramural Research Program

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Retirement of Extramural Program Directors



Joy Boyer



Elise Feingold, Ph.D.



Mike Smith, Ph.D.

Departure of Extramural Program Director



Ken Wiley, Ph.D.

New Extramural Program Director

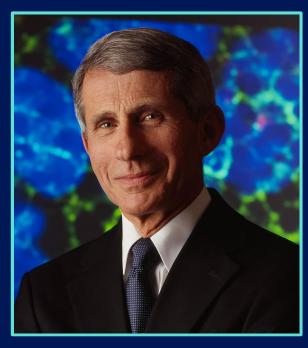


Temesgen Fufa, Ph.D.

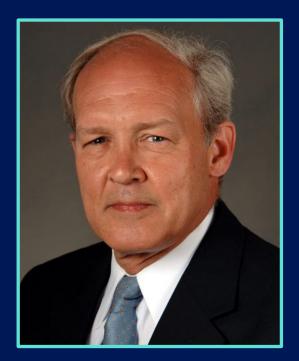
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Departure of Director, National Institute on Allergy and Infectious Diseases

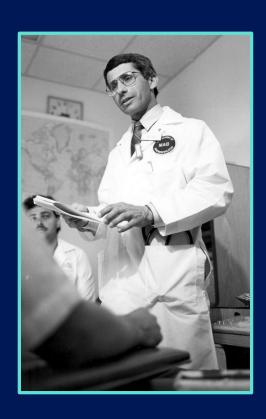


Anthony Fauci, M.D.

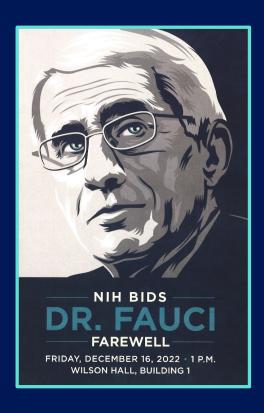


Hugh Auchincloss, M.D.

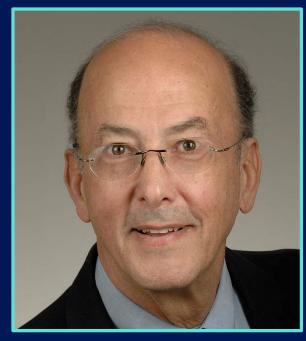
Departure of Director, National Institute on Allergy and Infectious Diseases



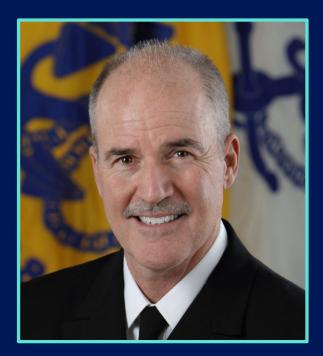




Departure of Director, Fogarty International Center



Roger Glass, M.D., Ph.D.



Peter Kilmarx, M.D.

Departure of Director, Center for Information Technology



Andrea Norris, M.B.A.



Ivor D'Souza, M.S.



Dennis Papula, M.S.

Departure of Associate Director for Clinical Research



John Gallin, M.D.



Departure of Associate Director for Legislative Policy and Analysis



Adrienne Hallett, M.A.



Lauren Higgins

New Deputy Director for Intramural Research



Nina Schor, M.D., Ph.D.



New Director, National Center for Advancing Translational Sciences



Joni Rutter, Ph.D.



NIH Data Management and Sharing Policy



- Effective January 25, 2023
- NHGRI's preparation for implementation of the policy:
 Guidance on where to share scientific data
 NHGRI sample DMS Plans

NIH Institute/Center Research Collaborations

Report for Fiscal Years 2019-2021

IC	FY2019	FY2020	FY2021
NHGRI	36%	33%	33%
All NIH	17%	18%	18%

NIH Inter-Institute/Center Research Collaborations



Office of Science and Technology Policy Guidance

Ensuring Free, Immediate, and Equitable Access to Federally Funded Research



Memorandum to federal departments and agencies:

- Agencies must make publications that describe taxpayer-funded work freely available to the public by the end of 2025
- Agencies required to develop plans to improve transparency

Science Policy Forum on NIH-led COVID-19 Response

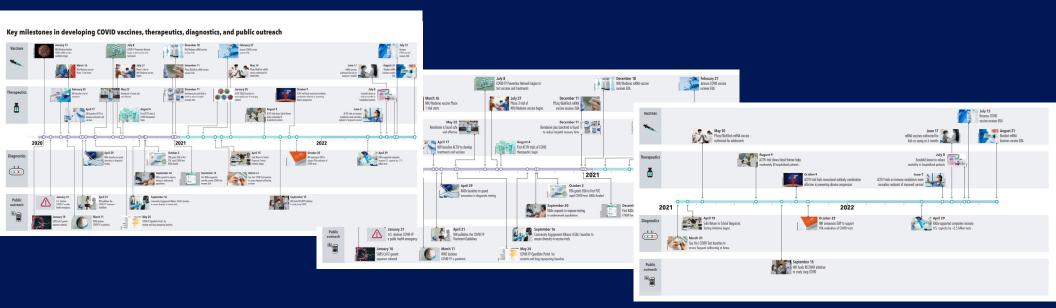


BIOMEDICINE

The NIH-led research response to COVID-19

Investment, collaboration, and coordination have been key

By Francis Collins¹, Stacey Adam², Christine Colvis³, Elizabeth Desrosiers⁴, Ruxandra Draghia-Akli⁵, Anthony Fauci⁶, Maria Freire², Gary Gibbons⁷, Matthew Hall³, Eric Hughes⁸, Kathrin Jansen⁹, Michael Kurilla³, H. Clifford Lane⁶, Douglas Lowy¹⁰, Peter Marks¹¹, Joseph Menetski², William Pao⁹, Eliseo Pérez-Stable¹², Lisa Purcell¹³, Sarah Read⁶, Joni Rutter³, Michael Santos², Tara Schwetz¹⁴, Jeffrey Shuren¹¹, Timothy Stenzel¹¹, Paul Stoffels¹⁵, Lawrence Tabak¹⁴, Karen Tountas², Bruce Tromberg¹⁶, David Wholley², Janet Woodcock¹¹, John Young¹⁷



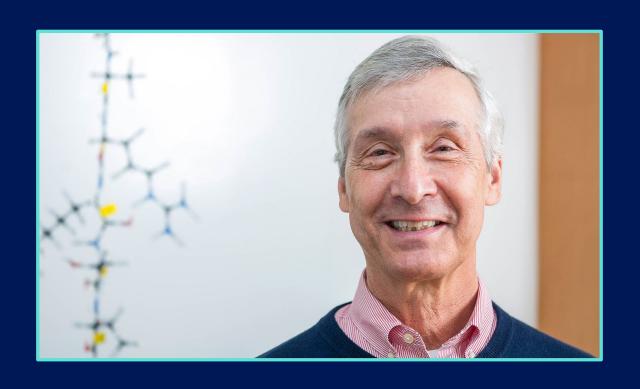
Fiscal Year 2023 Appropriations

	FY 2022 (\$)	FY 2023 (\$)	Increase (\$)	Increase (%)
NIH	\$45.0 billion	\$47.5 billion	\$2.5 billion	5.6
NHGRI	\$639 million	\$663 million	\$24 million	3.8

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Mourning the Loss of David Allis



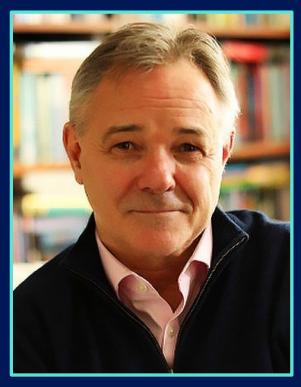


Mourning the Loss of Barbara Migeon





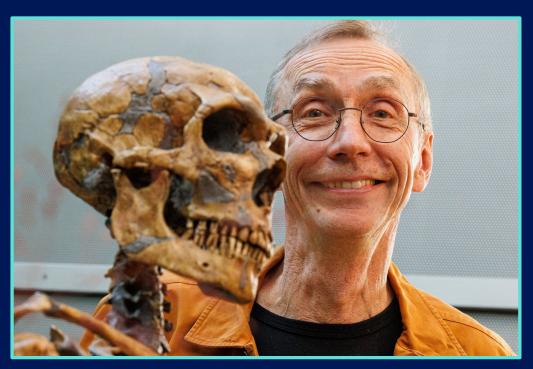
New Chief Scientist, World Health Organization



Jeremy Farrar, OBE, FMedSci, FRS



2022 Nobel Prize in Physiology or Medicine





Svante Pääbo, Ph.D.

2022 Lasker~DeBakey Clinical Medical Research Award

Noninvasive Prenatal Testing Using Fetal DNA

2022 Lasker~DeBakey Clinical Medical Research Award



Yuk Ming Dennis Lo
The Chinese University of Hong Kong



Elected to National Academy of Medicine

Steven Joffe Norman Sharpless

Eugene Koonin Bruce Tromberg

Katherine Pollard Marylyn Ritchie

John Quackenbush Sarah Tishkoff



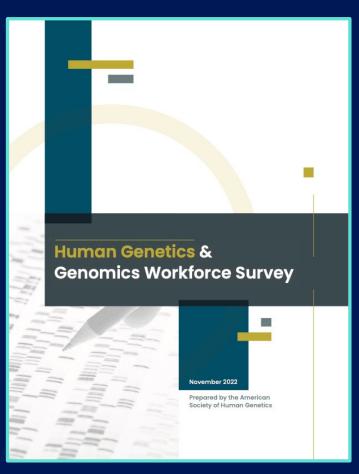
Elected to AAAS

Linda Birnbaum
Atul Butte
Cynthia Dunbar
Paul Liu

Alan Love Robert Tjian Elias Zerhouni



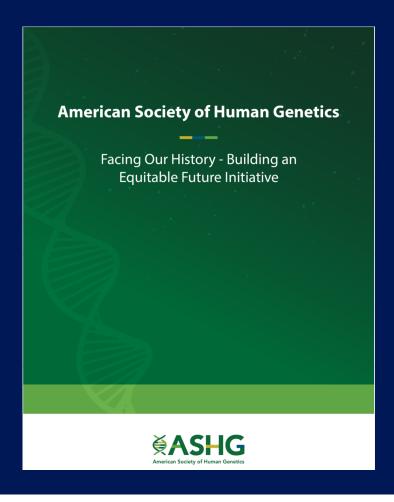
Human Genetics & Genomics Workforce Survey Report



- Report from American Society of Human Genetics (ASHG) – with support from NHGRI
- Report lays important groundwork for future assessment and action
- Key respondent findings: 67.7% identified as White; 73.3% as U.S. Citizens; women made up majority of respondents
- NHGRI hosted webinar with report partners to explore key findings and define next steps

American Society of Human Genetics

Facing Our History – Building an Equitable Future Initiative



2022 American Society of Human Genetics Awards









Heidi Rehm, PhD, FACMG 2022 Curt Stern Award



David L. Nelson, PhD 2022 Victor A. McKusick Leadership Award



Eimear Kenny, PhD 2022 Early Career Award

NASEM Study

Toward Sequencing and Mapping of RNA Modifications

- Consensus study on direct sequencing of RNA modifications
- Sponsored by: Warren Alpert Foundation, NIEHS, & NHGRI



■ Upcoming Workshop: March 14-15, 2023

Global Alliance for Genomics and Health (GA4GH)



- 10th plenary meeting in September 2022
- 11 NIH-supported GA4GH Driver Projects (out of 24 projects)
- Call for new GA4GH Driver Projects and Strategic Partnerships
- GA4GH Assigned Expert program extends global reach

Global Biodata Coalition



- First set of Global Core Biodata Resources (GCBRs) selected
- GCBRs of fundamental importance to life sciences around the world, and the long-term preservation of biological data
- Of the 37 resources selected, 16 are funded by NHGRI

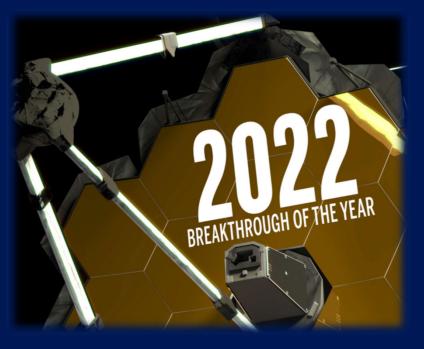


International Common Disease Alliance

- ICDA Scientific Plenary Meeting in December 2022
- Meeting focused on proposals for moving from Maps to Mechanism to Medicine



Science's 2022 Breakthrough of the Year & Runners Up





Black Death's legacy detected in the genes of Europeans



Ancient ecosystem reconstructed from 2-million-year-old DNA

Nature's 10 People Who Helped Shape Science in 2022





The Scientist's Top Innovations of 2022



Machine to sequence the genome for \$100
Instruments for single-cell analysis
Non-invasive prenatal testing
3D view of gene expression

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Human Genome Reference Program (HGRP)



Annual HPRC meeting (October 2022)

Sample selection and collection Embedded ELSI research Changes in DNA sequence-production costs

HGRP planning meeting (October 2022)

Genomics Research Enhances Genetics of Rare Disease (GREGoR) Consortium



- First in-person GREGoR Consortium Meeting November 2022
 Over 140 participants
- Key themes of networking and collaboration
 Identifying challenges to solving Mendelian disorders
 Internal and external collaboration

Impact of Genomic Variation on Function (IGVF)

First Annual Consortium Meeting



September 2022

Meeting focus:

- Coordinating activities
- Engaging trainees
- Fostering interactions
- Defining future goals

Impact of Genomic Variation on Function (IGVF)

IGVF Affiliate Membership

- Affiliate role
 Contribute to data
 sharing and analysis
 Engage in Consortium
 Broaden participation
- Current affiliate members17 membersU.S. and Europe



Affiliate Sites in Continental U.S.

Molecular Foundations for Biotechnology (MFB) Partnerships to Transform Emerging Industries

RNA Tools/Biotechnology

- NSF23-554: Partnership between NSF & NHGRI
- Soliciting synergistic proposals:
 Study RNA function in complex biological systems
 Harness RNA research to advance biotechnology
 Integrate across disciplines
- Letters of intent due: March 16, 2023
- Full proposal due: May 11, 2023

Analysis, Visualization, and Informatics Lab-space (AnVIL)

Data Use Oversight System (DUOS) Pilot



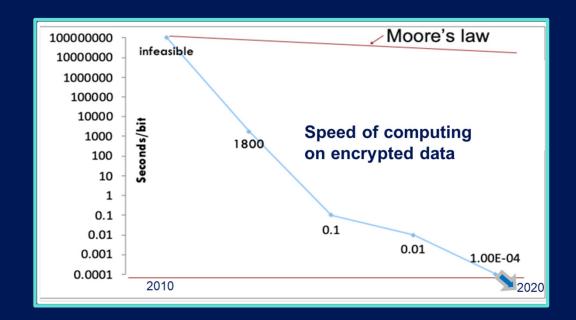
Data Use Oversight System

Expediting data access for researchers, by facilitating and enhancing data access committee's workflows

- DUOS expedites controlled data access
- By mid-2023, DUOS will be used by the NHGRI Data Access Committee to process all data access requests

iDASH Genomic Privacy Challenge 2022

- Privacy algorithms rapidly becoming practical in real-world use
- Compute time decreasing faster than Moore's Law



■ iDASH challenge winners building useful genomics applications

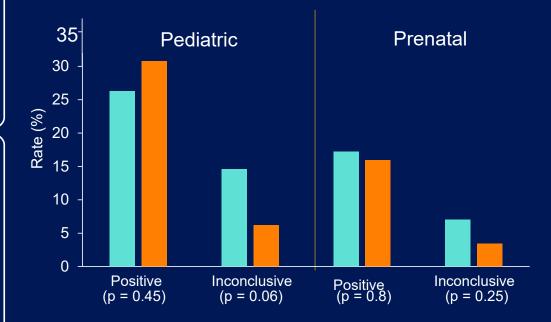


Clinical Sequencing Evidence-generating Research Program (CSER)



- 10 presentations
- P3EGS: Rates of positive diagnoses and inconclusive findings were similar between URM and non-URM children
- KidsCanSeq: Combined exome sequencing and panel testing may improve utility in pediatric cancer

Similar Rates of Positive Diagnoses and Inconclusive Findings in URM and non-URM Pediatric and Prenatal Exome Sequencing



Courtesy of Anne Slavotinek (unpublished)

Document 37

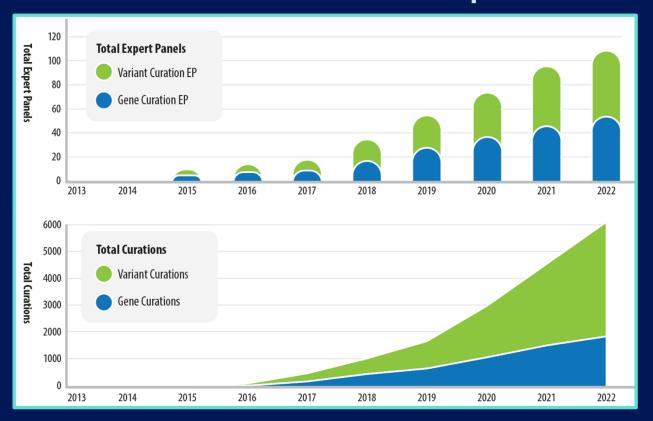


Clinical Genome Resource (ClinGen)

Efforts Toward Sustainability



Increased Curation Output



Other NIH Funding

NICHD NCI **NINDS** NEI

Non-NIH Funding



Phenotypes and Exposures (PhenX) Toolkit



PhenX Toolkit renewed (Fiscal Years 2022-2026)

- Carol Hamilton, RTI International
- Funded as Trans-NIHBiomedical Knowledgebase
- Co-funding from NHLBI, NIMHD, NINDS, and OBSSR

Phenotypes and Exposures (PhenX) Toolkit

Expanded Social Determinants of Health Collection

PhenX Social Determinants of Health Assessments Collection

An open-access resource for standard social determinants of health measures.







- 15 new protocols
- Protocols include:

 Internet access
 Minimum wage
 Healthcare discrimination





Polygenic Risk MEthods in Diverse populations Consortium (PRIMED)



Working Groups:

- Data Sharing
- Genotype Harmonization
- Methods Review
- Phenotype Harmonization
- Social and Ethical Implications of Polygenic Risk

Establishment of PRIMED External Scientific Panel

Small Business Program

- ~\$16.5 million in Fiscal Year 2022
- 19 Phase I Proof of Principle awards
- 11 Phase II Pre-Commercialization awards













Extramural Training Program

- Outreach at the 2022 Annual Biomedical Research Conference for Minority Students (ABRCMS)
- Publication of the conference report on the 2022 NHGRI Research Training and Career Development Annual Meeting

Conference Report

For reprint orders, please contact: reprints@futuremedicine.com

Personalized Medicine



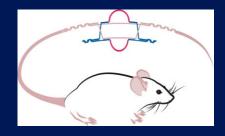
Susanne B Haga*,10

¹Duke University School of Medicine, Department of Medicine, Division of General Internal Medicine, Durham, NC 27708, USA *Author for correspondence: Tel.: 919 684 0325; susanne.haga@duke.edu

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Knockout Mouse Phenotyping Project (KOMP2)





Awards funded for a final five-year project period









Annual meeting held December 2022

Human Heredity & Health in Africa (H3Africa)



Setting the agenda in research

Comment



Scientists at the African Centre of Excellence for Genomics of Infectious Diseases in Ede, Nigeria, prepare samples for DNA sequencing

Could Africa be the future for genomics research?

Zané Lombard & Guida Landouré for the H3Africa Steering Committee

Human Biomolecular Atlas Program (HuBMAP)

Now in Production Phase (Fiscal Years 2022-2026)

6 HuBMAP Integration, Visualization, and Engagement Groups

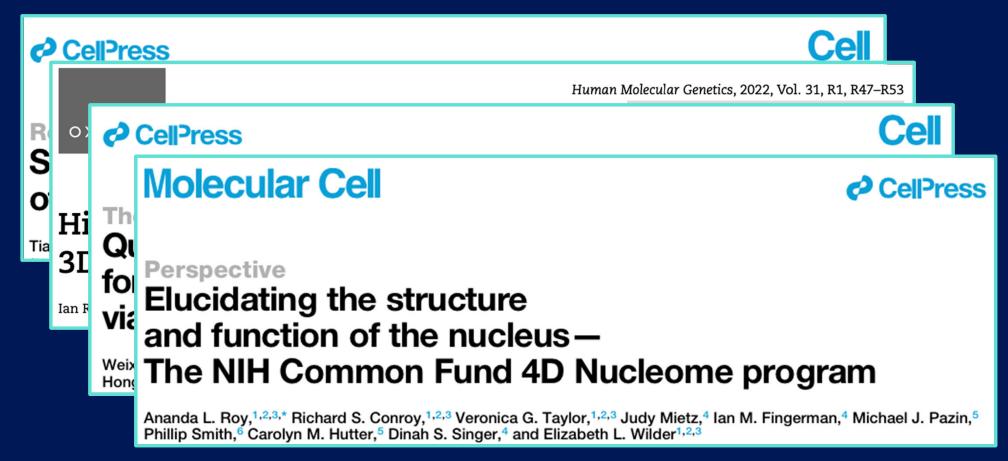
10 Tissue Mapping Centers

4 Demonstration Projects





4D Nucleome (4DN)





Faculty Institutional Recruitment for Sustainable Transformation (FIRST)

2022 Awardees

Institution	Principal Investigators
Northwestern University in Chicago	Melissa Simon, Eric Perreault, Clyde Yancy
University of California, San Diego	JoAnn Trejo, María Elena Martinez
University of Maryland Baltimore	James Kaper, William LaCourse
The University of New Mexico	Jane Ellen Smith, Irene Salinas
University of South Carolina in Columbia	Angela D. Liese, Coretta Jenerette



- Return of health-related genetic results to participants
- New Funding Opportunity Announcements

RFA-PM-23-001

RFA-PM-23-002

Due date: March 1, 2023



Bespoke Gene Therapy Consortium (BGTC)



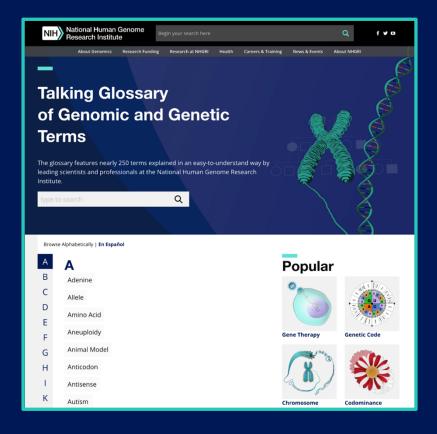
Candidate Diseases:

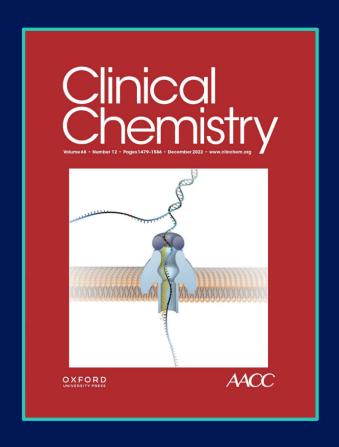
- Congenital Hereditary Endothelial Dystrophy
- MPS VI Corneal Disease
- Leber Congenital Amaurosis 16
- Retinitis Pigmentosa CNGB1
- Retinal Degeneration NPHP5
- Charcot Marie Tooth Disease, Type4J
- Multiple Sulfatase Deficiency
- Spastic Paraplegia, Type 47
- Spastic Paraplegia, Type 50
- Barth Syndrome
- PGM1-Congenital Disorder of Glycosylation
- Propionic Acidemia
- Fibrodysplasia Ossificans Progressiva
- Mucopolysaccharidosis IVA (Morquio A Syndrome)

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Talking Glossary of Genomic and Genetic Terms

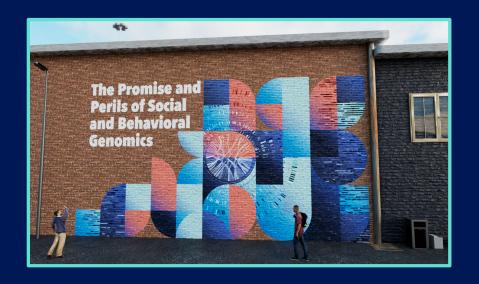


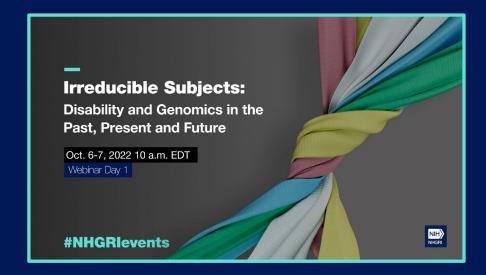


~18.4 million page views

NHGRI History of Genomics Program

Recent Events





NHGRI History of Genomics Program

Archive Highlight



THE WHITE HOUSE

April 11, 2003

I send greetings to the International Human Genome Sequencing Consortium as you mark the successful completion of the Human Genome Project. This historic milestone is also the 50th anniversary of the description of the double-helix structure of DNA

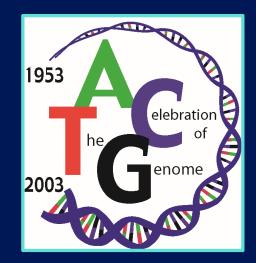
The American system of medicine is a model of skill and innovation and is adding good years to countless lives. The completion of the sequencing of the human genome, begins another era of medical progress. New gene-based screening tools are alerting patients when they have an elevated risk of diseases so they can take an active role in preventing them. Scientists believe many new therapies will be tailor-made to an individual's genetic makeup, resulting in fewer adverse effects. The scientific accomplishments and international collaboration of the Consortium bring hope and promise to countless individuals who suffer from disease and others at 100 per some constant of the constraints of th

My Administration has demonstrated our strong commitment to medical research by completing a five-year doubling of the National Institutes of Health (NIH) budget to more than \$27 billion. As a result of this increase, the NIH now trains 1,500 more scientists per year and issues 10,000 more research grants than it did in 1998. This investment will help turn today's research opportunities into more of formorrow's medical success stories.

I commend the scientists, researchers, and all others involved in the project for your tireless work to attain new scientific breakthroughs that enhance lives. Your efforts contribute to an improved system of medicine and will benefit the health and well-bening of all mankind.

Laura joins me in sending our best wishes.

Jusac.



108TH CONGRESS 1ST SESSION S. CON. RES. 10

Designating April 2003 as "Human Genome Month" and April 25 as "DNA Day".

IN THE SENATE OF THE UNITED STATES

February 27, 2003

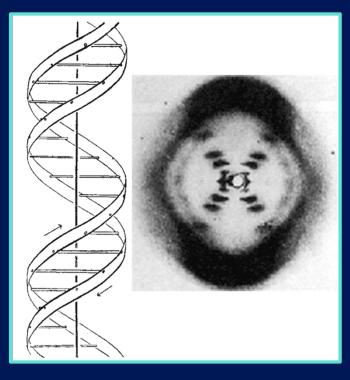
Mr. Gregg (for himself, Mr. Kennedy, Ms. Snowe, and Mr. Daschle) submitted the following concurrent resolution; which was considered and agreed to

CONCURRENT RESOLUTION

Designating April 2003 as "Human Genome Month" and April 25 as "DNA Day".

Whereas April 25, 2003, will mark the 50th anniversary of the description of the double-helix structure of DNA by James D. Watson and Francis H.C. Crick, considered by many to be one of the most significant scientific discoveries of the 20th Century;

Genomics Anniversaries in 2023

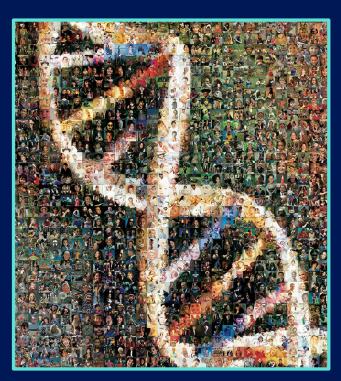




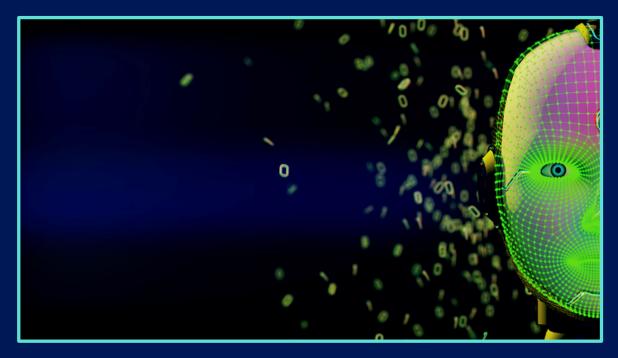






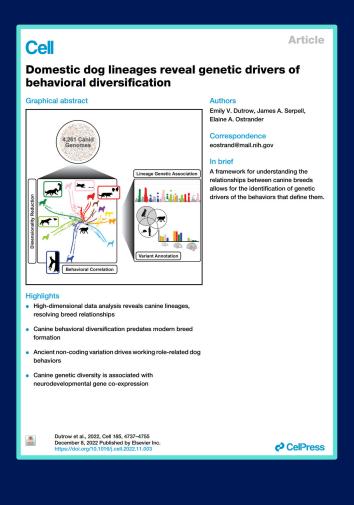


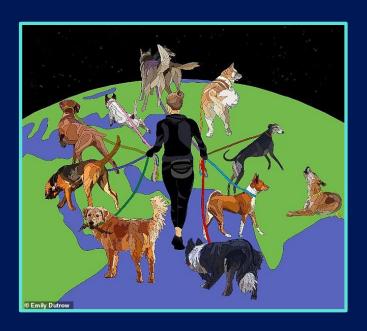
Communications on Artificial Intelligence in Genomics



- Feature story on Al and ML research at NHGRI
- Video with Benjamin Solomon, Susan Persky, and Oleg Shchelochkov
- Twitter Q&A with Spencer Hong (Ph.D. Candidate)

Communications on the NHGRI Dog Genome Project





The New York Times









Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

2022-2024 Scholars



Ava Willoughby



Molly Felix

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Retirement of Intramural Investigators



Joan Bailey-Wilson, Ph.D.



David Bodine, Ph.D. Jim Mullikin, Ph.D.



New Chief, NHGRI Metabolic Medicine Branch



Charles Venditti, M.D., Ph.D.



NHGRI Intramural Research Highlights



Genetics Medicine Educational considerations based on medical student use of polygenic risk information and apparent race in a simulated consultation



AJHG

Genotype first: Clinical genomics research through a reverse phenotyping approach



PNAS

A family of unusual immunoglobulin superfamily genes in an invertebrate histocompatibility complex









Where's Eric?































Where's Eric?

Where's Laura?

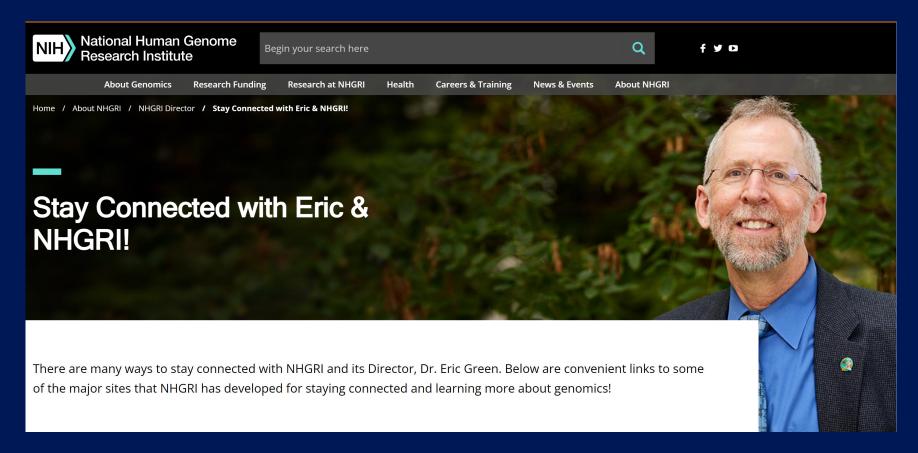
Where's Stephen?

Where's Gail?

Where's Hal?

'One-Stop-Shop' to Stay Connected

genome.gov/stayconnected





Thanks!



Special Thanks!

