National Human Genome Research Institute

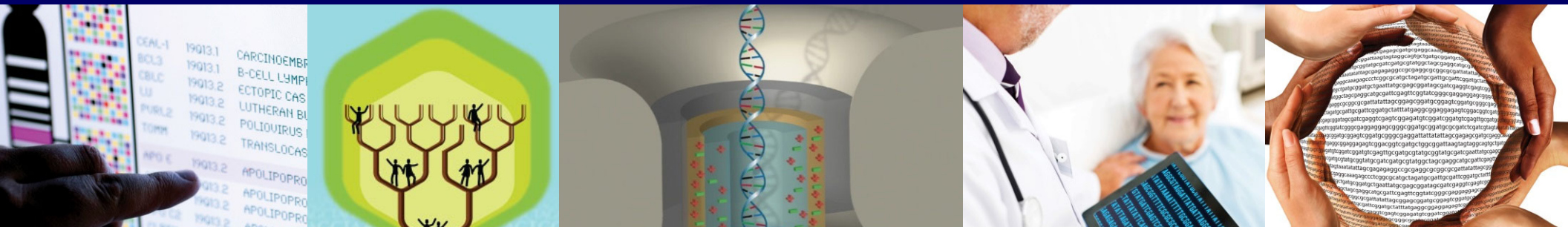
The Forefront of Genomics®

DIRECTOR'S REPORT

Eric Green, M.D., Ph.D.

Director, NHGRI

September 2024



Director's Report-Related Documents

September 2024

Director's Report

Director's Report

No.	Relevant Documents
1	Departure of Director, Division of Genome Sciences
2	New Director, Training, Diversity, and Health Equity Office
3	New NIH-ACMG Genomic Medicine Program Management Fellows
4	Journeys in Human Genetics and Genomics Colloquium
5	Eric Goes 'Down Under' in July 2024
6	Request for Information on Recommendations for Re-envisioning U.S. Postdoctoral Research Training

genome.gov/DirectorsReport

Document #

Open Session Agenda

Presentations:

Looking Back and Looking Forward: NHGRI's Training, Diversity, and Health Equity Program

Vence Bonham and Rob Rivers

A New Vision and Role for the NIH Center for Information Technology (CIT)

Sean Mooney

Administration Priorities and NIH Policy Update

Lyric Jorgenson

Open Session Agenda

Concept Clearances:

Impact of Genomic Variation on Function (IGVF) Renewal
Stephanie Morris

Genomics Research to Elucidate the Genetics of Rare Disease (GREGoR) Renewal
Lisa Chadwick

Electronic Medical Records and Genomics (eMERGE) Renewal
Robb Rowley

Advancing Genomic Medicine Research (AGMR) Renewal
Christine Chang

Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

IV. NHGRI Extramural Research Program

V. NIH Common Fund/Trans-NIH

VI. NHGRI Communications, Policy, and Education

VII. NHGRI Intramural Research Program

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Retirement of Chief, Scientific Review Branch, Division of Extramural Operations

Rudy Pozzatti, Ph.D.

Departure of Director, Division of Genome Sciences

Carolyn Hutter, Ph.D.

NIH National Institutes of Health

Office of Strategic Coordination - The Common Fund

Document 1

Departure of Program Director, Training, Diversity, and Health Equity Office

Lucia Hindorff, Ph.D., M.P.H.

New Director,
Training, Diversity, and Health Equity Office

Robert Rivers, Ph.D.

New Extramural Program Director, Division of Genome Sciences

Idan Gabdank, Ph.D.

New Extramural Program Director, Division of Genomic Medicine

Mollie Minear, Ph.D.

New Genomic Program Administrator, Division of Extramural Operations

Sharna Tingle, M.P.H.

New Lead Education Outreach Specialist, Education and Community Involvement Branch

Christopher Williams, Ph.D.

New NIH-ACMG Genomic Medicine Program Management Fellows

Rachel Nusbaum, M.S., C.G.C.

Nicole Thompson, M.S., L.G.C.


Marie Luise Brennan


Hwaida Hannoush


Renee Rider


Valerie Willis

NIH-ACMG Fellowship in Genomic Medicine Program Management

- **Full-time, two-year, and paid experience**
- **Acquire credentials and experience to lead genomic medicine research and implementation programs**
- **Open to qualified physicians, physician assistants/associates, advanced practice nurses, and genetic counselors**
- **Post-fellowship positions include the NIH, WHO, and major medical institutions**

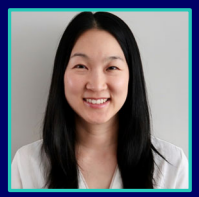
Applications Due Date: December 6, 2024


Deepika Burkhardt


Nguyen Park


Veronica Abraham


Julius Militante


Jessica Chong


Karyn Roberts


Rachel Nusbaum


Nicole Thompson

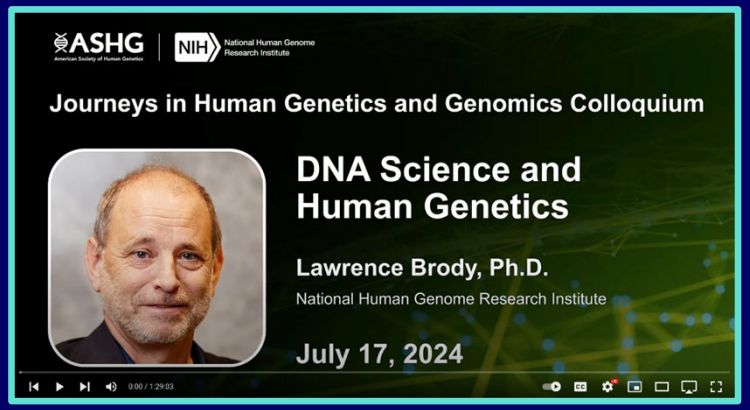
Document 3

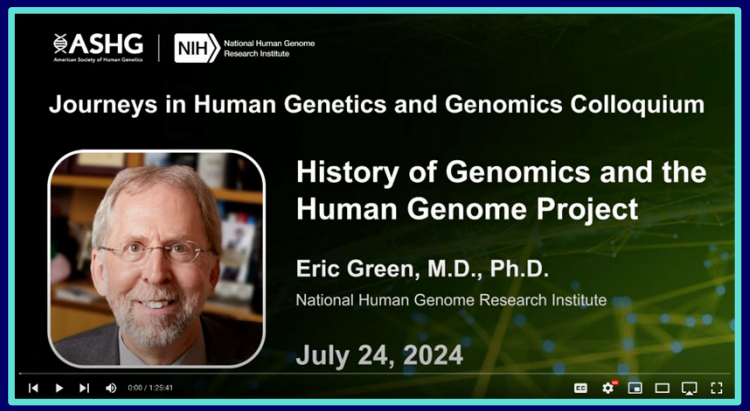
Journeys in Human Genetics and Genomics Colloquium

Journeys in Human Genetics and Genomics Colloquium

ASHG
American Society of Human Genetics

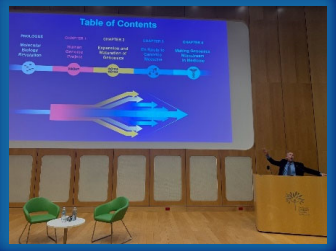
NIH
National Human Genome Research Institute

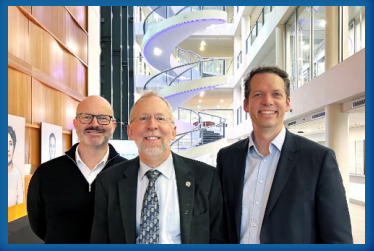




Document 4

Eric Goes ‘Down Under’ in July 2024





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New Director,
NIH Office of AIDS Research

Geri Donenberg, Ph.D.

Request for Information on Recommendations for Re-envisioning U.S. Postdoctoral Research Training

- RFI on re-envisioning NIH-supported postdoctoral training
- Recommendations:
  - Limit the total number of years a person can be supported by NIH funds in a postdoctoral position to no more than 5 years
  - Revise K99/R00 mechanism to focus on ideas and creativity over productivity
  - Promote training and professional development of postdoctoral scholars and their mentors
- Responses requested by October 23

Document 6

NIH Director's Statement Supporting Our Valued Asian Research Colleagues

- NIH has implemented measures to protect research integrity
- NIH is committed to promoting a diverse and inclusive research community
- NIH is working to restore relationships and ensure a welcoming environment while safeguarding research integrity

Document 7

Fiscal Year 2025 Appropriations

Portion	FY 2024 Enacted ($M)	FY 2025 House ($M)	FY 2025 House - FY 2024 Enacted (%)	FY 2025 Senate ($M)	FY 2025 Senate - FY 2024 Enacted (%)
NIH	$48,174	$48,581	+0.8%	$50,224	+4%
NHGRI	$663	~~	-1.4%	$663	0%

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Mourning the Loss of Maxine Singer

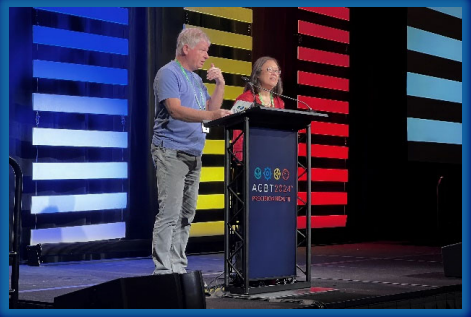
Document 8

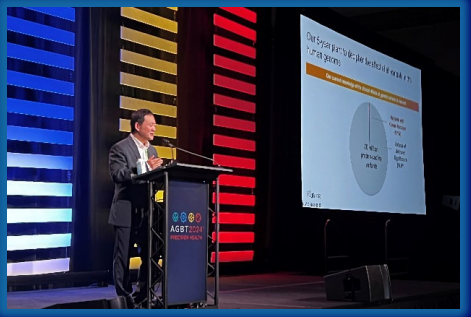
Lurie Prize in Biomedical Sciences

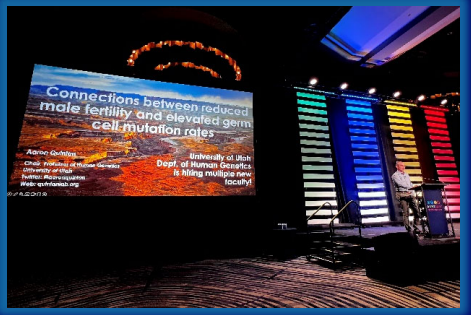
Howard Chang,
M.D., Ph.D.

Document 9

2024 AGBT Precision Health Meeting







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Centers for Excellence in Genomic Science (CEGS)

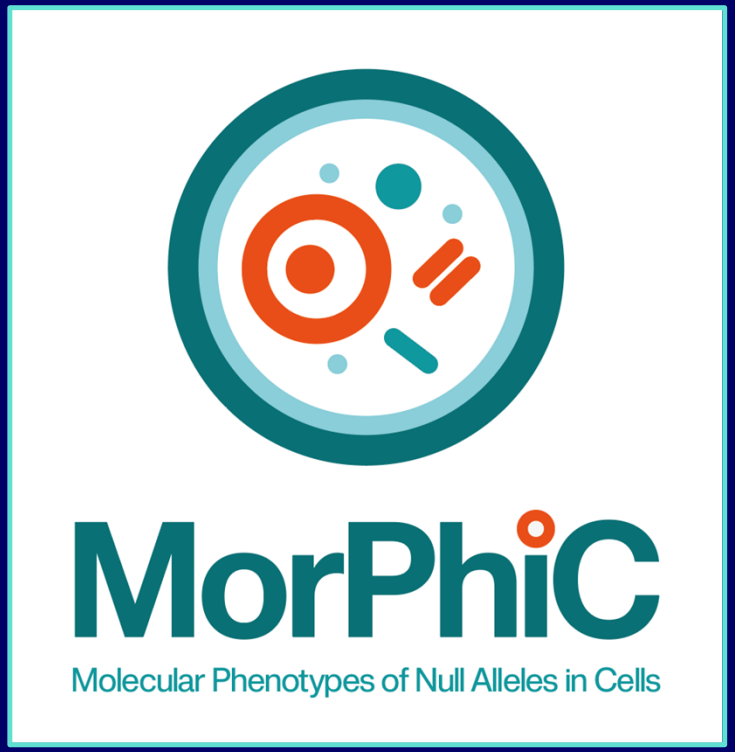
Click to add text

Doug Fowler, Ph.D.

Next Receipt Date: June 23, 2025

Molecular Phenotypes of Null Alleles in Cells (MorPhiC) Phase 1

- Virtual consortium meeting in July 2024
- Highlighted experimental design, data pipeline plans, individual scientific results, and progress updates
- Trainee organizers, presenters, and moderators



Molecular Foundations in Biotechnology (MFB)

Workshop:

- Translating knowledge into biotechnology solutions
- Identifying research directions, translational opportunities, and commercial relevance

Genome Technology Program

- **Notice of Special Interest : Advancing Genomic Technology Development for Research and Clinical Application**

  Next Due Date: January 5, 2025

- **2024 Advances in Genomic Technology Development Meeting**

AGTD

Advances in Genomic Technology Development

Document 13

Genome Technology Program

AGTD 2024 Meeting: Open Science Day

Small Business Workshops

AlidaBio

EpiCypher®

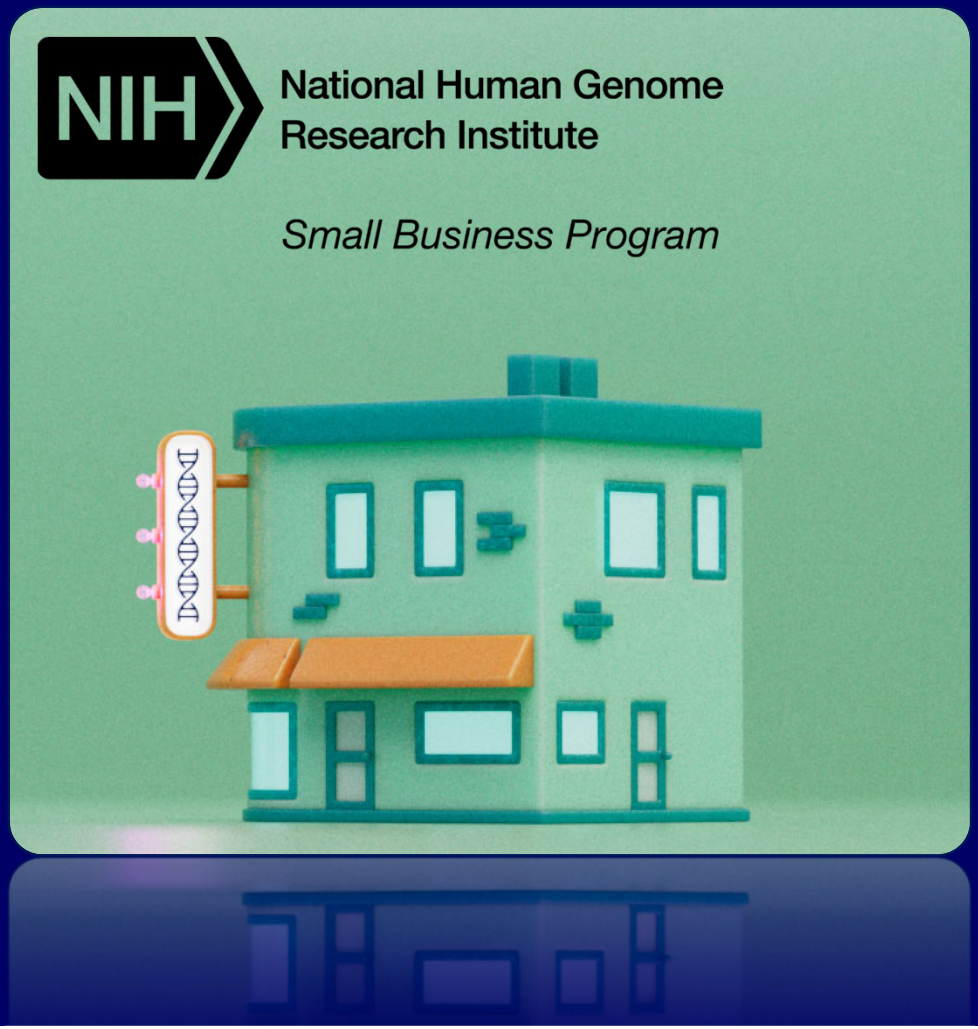
Keynote Speaker

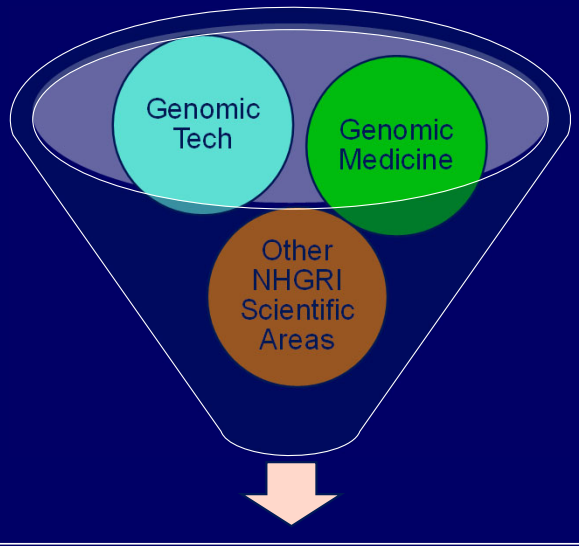
Stacey Gabriel, Ph.D.

@genomeTDCC

Document 13

Small Business Program





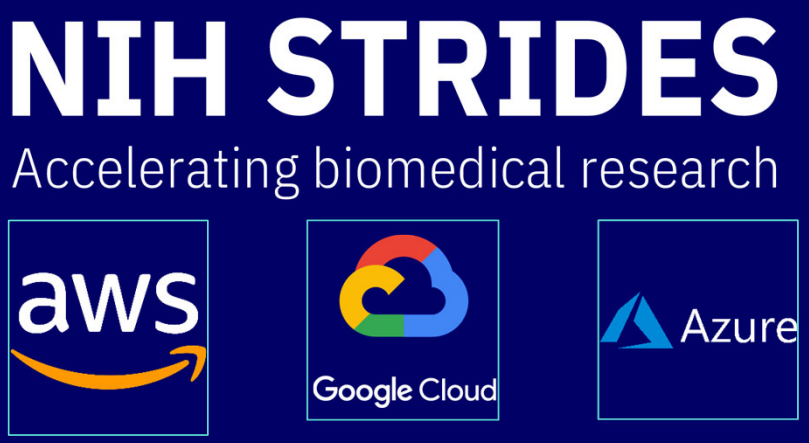
NHGRI Small Business Program

Small Business Listserv:

- Funding opportunities
- General announcements
- Resources to maximize commercialization

Document 14

NIH Cloud Lab



- 90-day, no-cost program providing $500 in cloud credits
- NIH-funded researchers at eligible institutions
- Addresses cloud adoption challenges by offering relevant training

NIH Office of Data Science Strategy Funding Opportunities

Four Data Science Funding Opportunities

Link	NOFO Title	Receipt Dates
NOT-OD-24-078	Notice of Special Interest (NOSI): Supporting the Exploration of Cloud in NIH-supported Research (Competing Revision)	June 18 2024 - 2026
NOT-OD-24-096	Notice of Special Interest (NOSI): Promoting Data Reuse for Health Research	March 4, July 3, November 3 2024 - 2026
RFA-OD-24-010	Building Sustainable Software Tools for Open Science (R03 Clinical Trial Not Allowed)	June 4, December 4 2024 - 2026
RFA-OD-24-011	NIH Research Software Engineer (RSE) Award (R50 Clinical Trials Not Allowed)	June 4, December 4 2024 - 2026

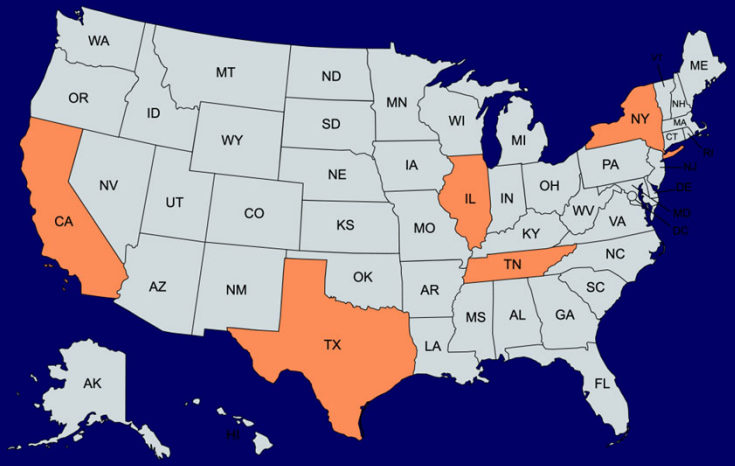
emerge network

ELECTRONIC MEDICAL RECORDS AND GENOMICS

- **Recruitment is complete: 25,375 participants**
- **48% self-reported underrepresented race or ethnicity**
- **17,516 GIRA generated and 16,609 GIRA returned to the electronic health records**
- **24% of GIRAs: high-risk for 10 common, complex diseases**

Multi-Omics for Health and Disease (MOHD) Consortium

Aim: 1,800 participants
Diverse populations
6 Disease Study Sites

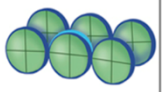
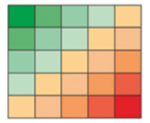


Clinical visits

Surveys

Biospecimens

Multi-Omics Profiles

Genomics	Epigenomics	Transcriptomics	Proteomics	Metabolomics
				
DNA (e.g. SNP or WGS	Chromatin accessibility Chromatin structure DMA methylation	mRNA Non-coding RNA (e.g. miRNA, piRNA, lncRNA)	Secreted and intracellular proteins	Endogenous circulating metabolites Xenobiotics

Adapted from Rhee *et al.* 2020

Measurements

Document 18

Clinical Genome Resource (ClinGen)

Implementation of ClinGen's Gene Curation Results

Selection of Germline Genetic Testing Panels in Patients With Cancer: ASCO Guideline

Nadine Tung, MD[1]; Charité Ricker, MS, CGC[2]; Hans Messersmith, MPH[3]; Judith Balmaña, MD, PhD[4]; Susan Domchek, MD, FASCO[5]; Elena Martinez Stoffel, MD, MPH[6]; Khaldoun Almhanna, MD, MPH[7]; Banu Arun, MD, FASCO[8]; Yanin Chavarri-Guerra, MD, MSc[9]; Stephanie A. Cohen, MS, LCGC[10]; Deborah Cragun, PhD, CGC[11]; Katherine D. Crew, MD, MS[12]; Michael J. Hall, MD, MS[13]; Gregory Idos, MD, MS[14]; Ghecemy Lopez, DSW(C), MAED[15]; Tuya Pal, MD[16]; Sara Pirzadeh-Miller, MS, CGC[17]; Colin Pritchard, MD, PhD[18]; Huma Q. Rana, MD, MPH[19]; Umang Swami, MD[20]; and Gregory A. Vidal, MD, PhD[21]

DOI https://doi.org/10.1200/JCO.24.00662

ABSTRACT

ASCO Guidelines provide recommendations with comprehensive review and analyses of the relevant literature for each recommendation, following the guideline development process as outlined in the ASCO Guidelines Methodology Manual. ASCO Guidelines follow the ASCO Conflict of Interest Policy fo Clinical hensive should b applicab and anal does not for any Appendi

ACCOMPANYING CONTENT

Appendix

Data Supplement

Table 3: Justification for Inclusion of Genes in Guideline Table 2

Gene	Justification
APC	Colorectal Cancer: recommended in several documents[2,11-17,20], supported by ClinGen[71] validity classification. Gastric Cancer: recommended in two documents[17,27], supported by ClinGen[71] validity classification. Adrenocortical Tumors: recommended in one document[33], based on link to FAP supported by ClinGen[71] validity classification Pancreatic Adenocarcinoma: Recommended in one document[23]

"The evidence assessment of the ClinGen expert panels informed the final selection of genes and the strength of the recommendation for each."

Document 19

Clinical Genome Resource (ClinGen)

ClinGen Awards of Excellence

Joannella Morales, PhD
Ancestry and Diversity Working Group

Justice, Equity, Diversity, and Inclusion (JEDI) Award

Document 19

Polygenic Risk Methods in Diverse Populations (PRIMED) Consortium

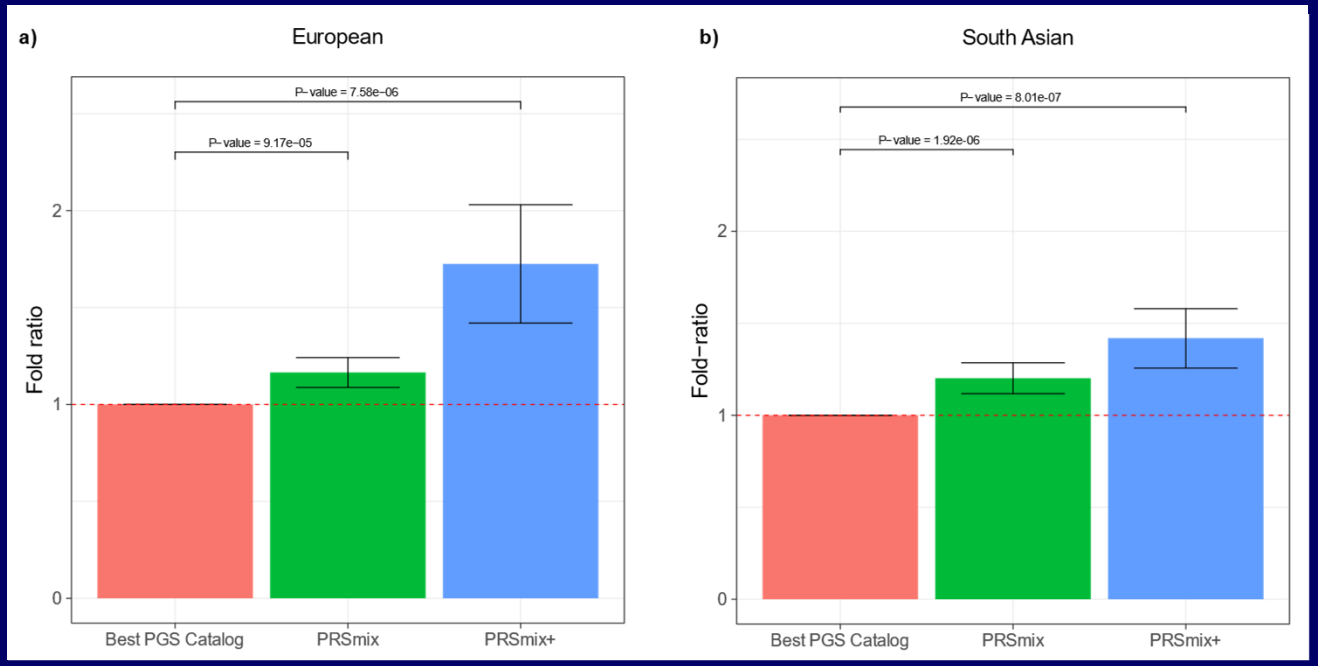
Cell Genomics

Volume 4, Issue 4, 10 April 2024, 100523

Article

Integrative polygenic risk score improves the prediction accuracy of complex traits and diseases

Buu Truong [1 2], Leland E. Hull [3 4], Yunfeng Ruan [1 2], Qin Qin Huang [5], Whitney Hornsby [1 2], Hilary Martin [5], David A. van Heel [6], Ying Wang [1 7 8], Alicia R. Martin [7 8], S. Hong Lee [9], Pradeep Natarajan [1 2 4 10]

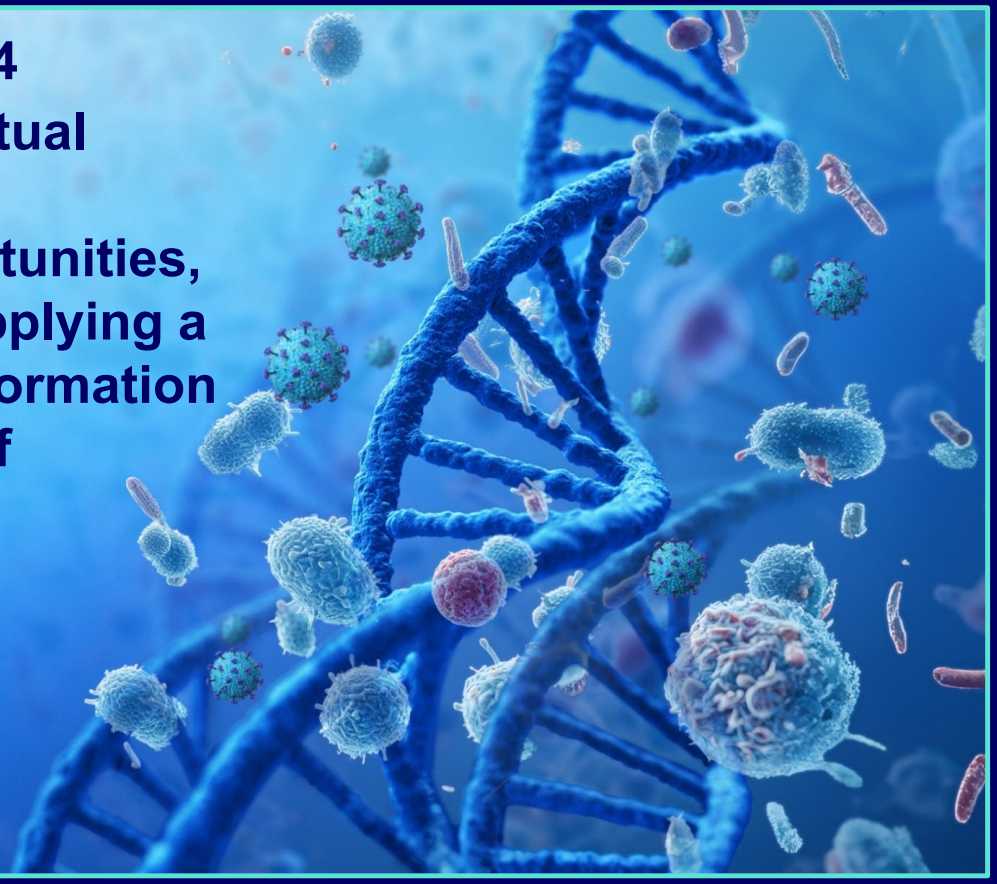


PRIMED

Genomic Medicine XVI: Host Genomics and Infectious Diseases

December 12-13, 2024
Bethesda MD and Virtual

Identify needs, opportunities, and challenges for applying a patient's genomic information in the management of infectious diseases.



NIH National Human Genome Research Institute

NIH National Institute of Allergy and Infectious Diseases

Document 21

Precision Health Research and International Health Cohorts Consortium Conference

- **Aims:**
  - Bring diverse perspectives on global health challenges
  - Identify issues related to integrating cohort studies and biobanks for translational research
  - Champion early-career scientists in genomic and precision medicine
- **Important Topics:**
  - Biobanks for precision medicine
  - Data validation and discovery
  - Data sharing, ethics, and policy
  - Genetic counseling and community engagement

Ethical, Legal, and Social Implications (ELSI) Research Program

ELSIcon2024: 6th ELSI Congress

Reimagining the Benefits of Genomic Science

Presentations now available on the ELSIhub resource

New Funding Opportunity: RFA-HG-24-028

Seeking applicants to organize ELSI Congress meetings in 2026, 2028, and 2030

Application Deadline: November 18, 2024

Ethical, Legal, and Social Implications (ELSI) Research Program

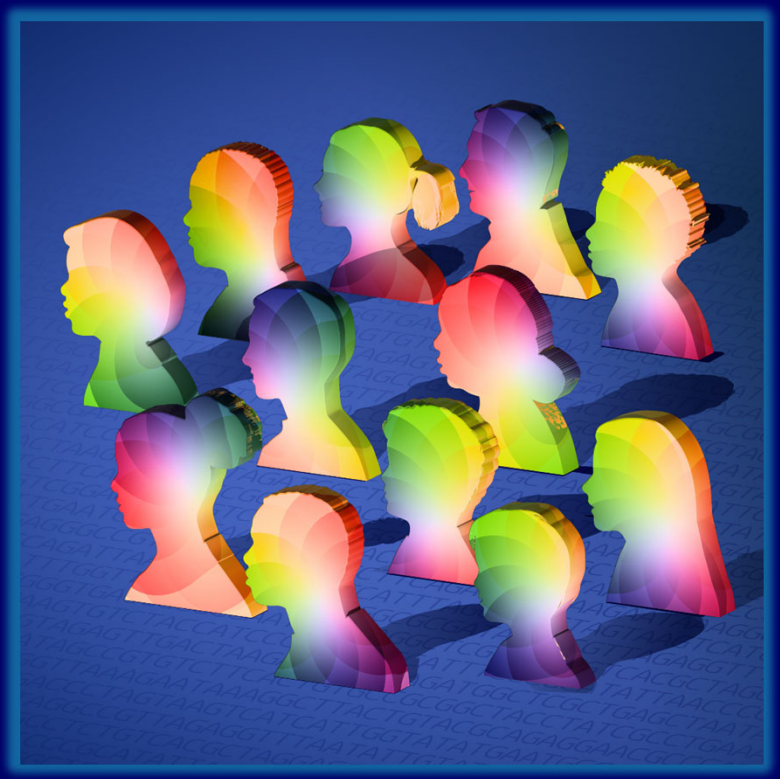
Building Partnerships and Broadening Perspectives to Advance ELSI Research (BBAER) Program

- **Eligible Applicants: U.S. domestic organizations receiving <$30M/year in NIH funding for last 3 fiscal years**

Application Due Date: November 15, 2024

Population Descriptors in Genomic Legacy Data

May 2024 Workshop



- Thoughtful study design and transparent justification of descriptors
- Expanded engagement with participant communities
- Increased education to avoid misuse of labels
- Promoting responsible use of descriptors
- Development of guardrails and guidelines

Extramural Investigator-Initiated Highlights

DeepGSEA: explainable deep gene set enrichment analysis for single-cell transcriptomic data

Guangzhi Xiong, Nathan J LeRoy, Stefan Bekiranov, Nathan C Sheffield, Aidong Zhang

Bioinformatics, Volume 40, Issue 7, July 2024, btae434, https://doi.org/10.1093/bioinformatics/btae434

Published: 01 July 2024 **Article history**

Young adults' reasoning for involving a parent in a genomic decision-making research study

Julia M. Pascal, Michelle L. McGowan, Amy A. Blumling, Cynthia A. Prows, Ellen A. Lipstein, Melanie F. Myers

First published: 26 August 2023 | https://doi.org/10.1002/jgc4.1768

Article | Open access | Published: 13 December 2023

GDF15 linked to maternal risk of nausea and vomiting during pregnancy

M. Fejzo, N. Rocha, I. Cimino, S. M. Lockhart, C. J. Petry, R. G. Kay, K. Burling, P. Barker, A. L. George, N. Yasara, A. Premawardhena, S. Gong, E. Cook, D. Rimmington, K. Rainbow, D. J. Withers, V. Cortessis, P. M. Mullin, K. W. MacGibbon, E. Jin, A. Kam, A. Campbell, O. Polasek, G. Tzoneva, ... S. O'Rahilly

+ Show authors

Nature **625**, 760–767 (2024) | Cite this article

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All of Us RESEARCH PROGRAM

- Notice (NOT-PM-24-004) describing how the program's data provides opportunities for research, training, and career development
- Recently started enrolling children from birth though age 4
- Now accepting applications for access to the Researcher Workbench from commercial organizations

Document 26

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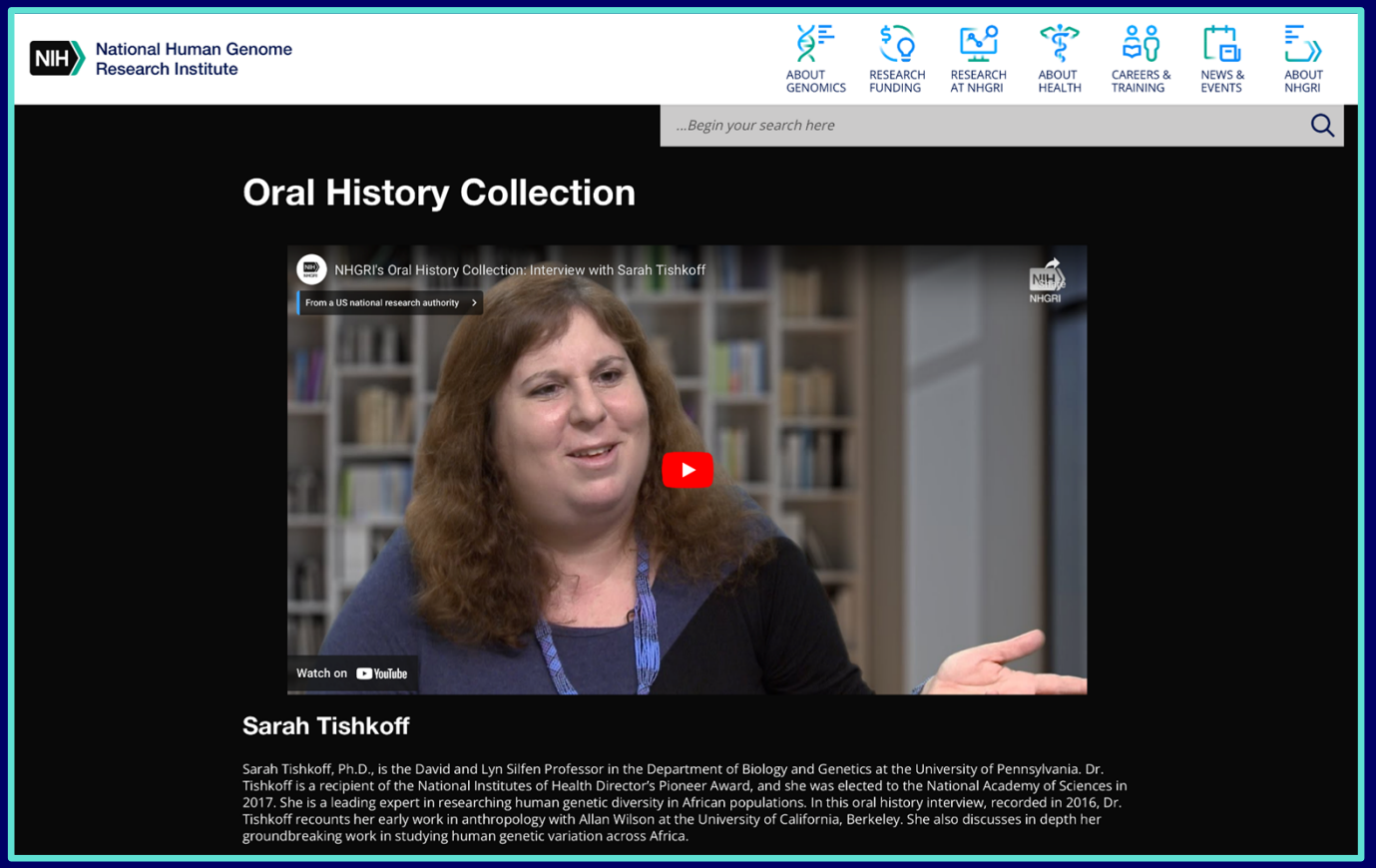
NHGRI History of Genomics Program

Oral History Collection



Document 27

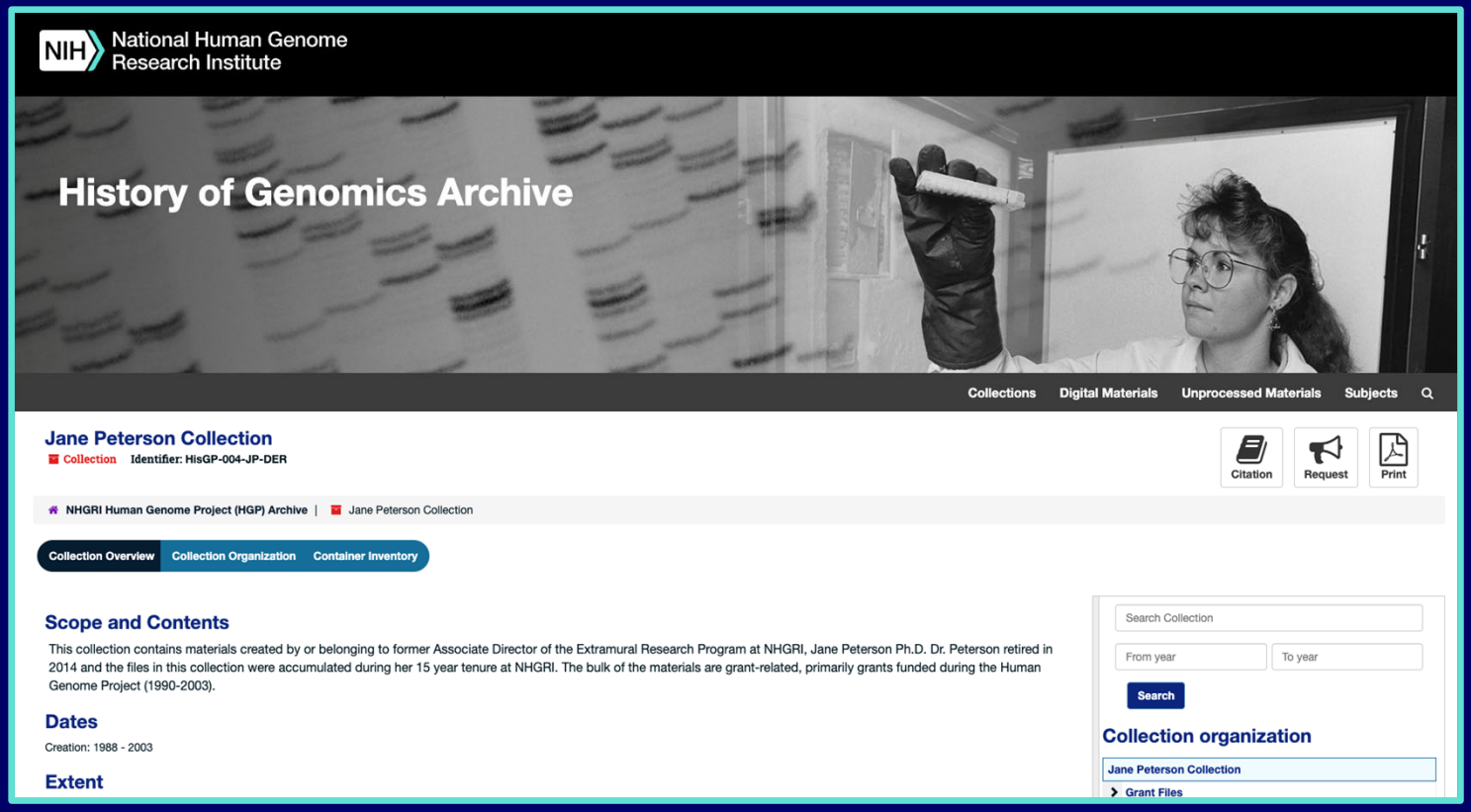
NHGRI History of Genomics Program

Virtual Symposium

Document 27

NHGRI History of Genomics Program

Jane Peterson Collection



Document 27

NHGRI History of Genomics Program

Archival Spotlight

February 20, 1989

Senator Mitch McConnell
US Senate
Washington D.C. 20510-6225

Dear Senator McConnell:

I was just reading about how ambitiously the government is funding the Human Genome Project. Undoubtedly, developing a road map of our genes is very important, but the voting public probably isn't terribly concerned with the benefits derived from a project like this. I would think the voting public might be more interested in having our tax dollars go to a project with more immediate and tangible benefits. Now I am not against the Human Genome project but I believe we should shift just a little of our ambition to other more consumer oriented projects. For example, I submit the following:

I have conducted a small survey of twenty-five of my friends and twenty-three of them agreed that the federal government would be held in higher esteem if it funded a project that would put the flavor of a home grown summer tomato into a winter, store-bought, hot-house tomato. The two who disagreed didn't like tomatos but I think its because they've never tried a summer tomato. I wouldn't like them either if hot-house tomatos were my only experience.

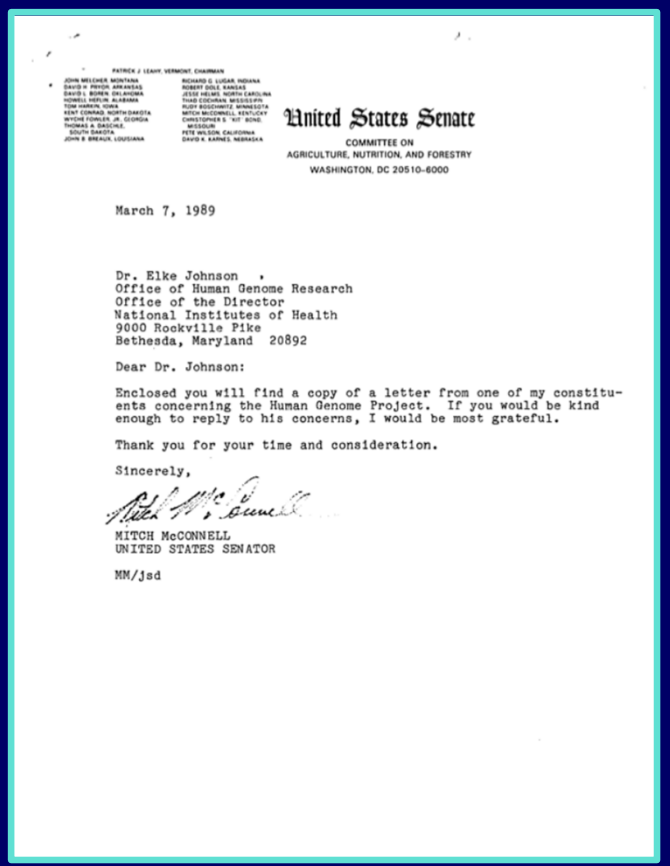
I am sure that if the Honorable Senator is like an ordinary voting consumer, you are always displeased with the quality of a winter tomato. Let's put some of those Human Genome Genetic Engineers onto the tomato flavor project. The increased sales of winter tomatos could possibly generate enough revenue to fund our genome road atlas.

Yours for Better Government,

[redacted]

University City, MO 63130

P.S. My normal residence is in Wilmore KY; I am presently studying at Washington University in St. Louis, that's why my address is out of your district.



PATRICK J. LEAHY, VERMONT, CHAIRMAN

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ROBERT DOLE, KANSAS
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RUDY BOSCHWITZ, MINNESOTA
MITCH McCONNELL, KENTUCKY
CHRISTOPHER S. "KIT" BOND, MISSOURI
PETE WILSON, CALIFORNIA
DAVID K. KARNES, NEBRASKA

United States Senate

COMMITTEE ON
AGRICULTURE, NUTRITION, AND FORESTRY
WASHINGTON, DC 20510-6000

March 7, 1989

Dr. Elke Johnson
Office of Human Genome Research
Office of the Director
National Institutes of Health
9000 Rockville Pike
Bethesda, Maryland 20892

Dear Dr. Johnson:

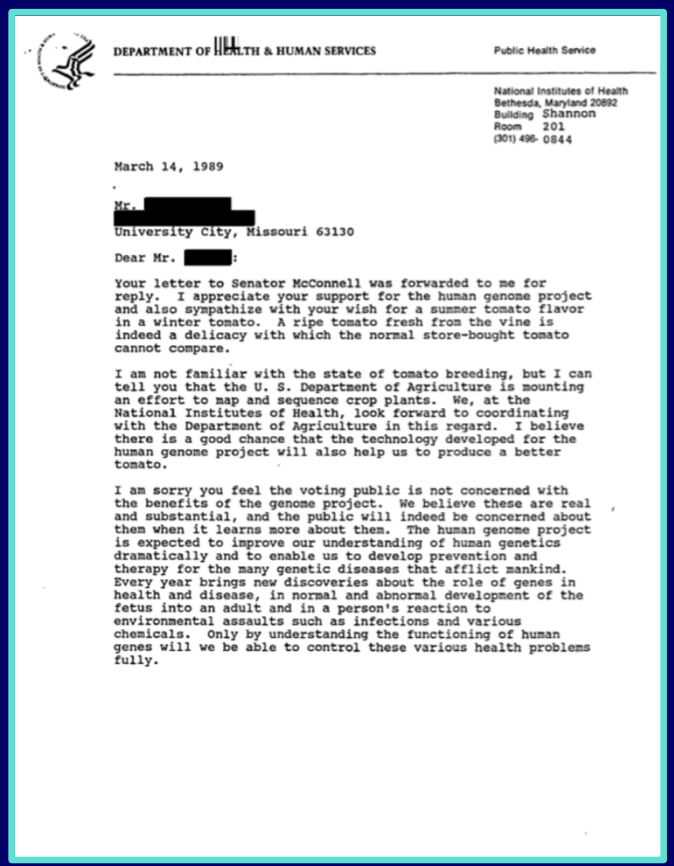
Enclosed you will find a copy of a letter from one of my constituents concerning the Human Genome Project. If you would be kind enough to reply to his concerns, I would be most grateful.

Thank you for your time and consideration.

Sincerely,

MITCH McCONNELL
UNITED STATES SENATOR

MM/jsd



DEPARTMENT OF HEALTH & HUMAN SERVICES — Public Health Service

National Institutes of Health
Bethesda, Maryland 20892
Building Shannon
Room 201
(301) 496- 0844

March 14, 1989

Mr. [redacted]
University City, Missouri 63130

Dear Mr. [redacted]:

Your letter to Senator McConnell was forwarded to me for reply. I appreciate your support for the human genome project and also sympathize with your wish for a summer tomato flavor in a winter tomato. A ripe tomato fresh from the vine is indeed a delicacy with which the normal store-bought tomato cannot compare.

I am not familiar with the state of tomato breeding, but I can tell you that the U. S. Department of Agriculture is mounting an effort to map and sequence crop plants. We, at the National Institutes of Health, look forward to coordinating with the Department of Agriculture in this regard. I believe there is a good chance that the technology developed for the human genome project will also help us to produce a better tomato.

I am sorry you feel the voting public is not concerned with the benefits of the genome project. We believe these are real and substantial, and the public will indeed be concerned about them when it learns more about them. The human genome project is expected to improve our understanding of human genetics dramatically and to enable us to develop prevention and therapy for the many genetic diseases that afflict mankind. Every year brings new discoveries about the role of genes in health and disease, in normal and abnormal development of the fetus into an adult and in a person's reaction to environmental assaults such as infections and various chemicals. Only by understanding the functioning of human genes will we be able to control these various health problems fully.

Document 27

NHGRI Office of Communications

Primate XY Video

NHGRI Office of Communications

Science Communications Talks

Kira Dineen

Emily Graslie

NHGRI Office of Communications

RNA Media Roundtable

Document 28

2024 NHGRI Short Course in Genomics

Document 29

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

New Co-Chair

Rebecca Kronk, Ph.D.

Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

IV. NHGRI Extramural Research Program

V. NIH Common Fund/Trans-NIH

VI. NHGRI Communications, Policy, and Education

VII. NHGRI Intramural Research Program

New Director, Intramural Training Office

Marcus G. Hodges, Ph.D.

The New York Times: A Disease That Makes Children Age Rapidly Gets Closer to a Cure

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NHGRI Intramural Research Highlights

AJHG

Evaluating large language models on medical, lay language, and self-reported descriptions of genetic conditions

scientific data

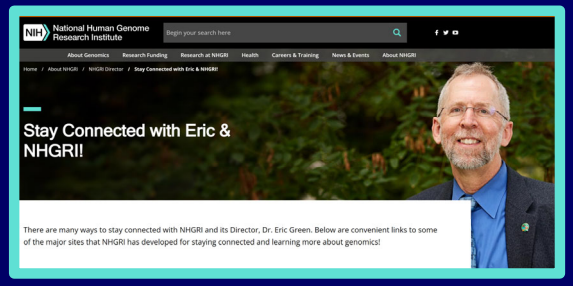
The reference genome of *Macropodus opercularis* (the paradise fish)

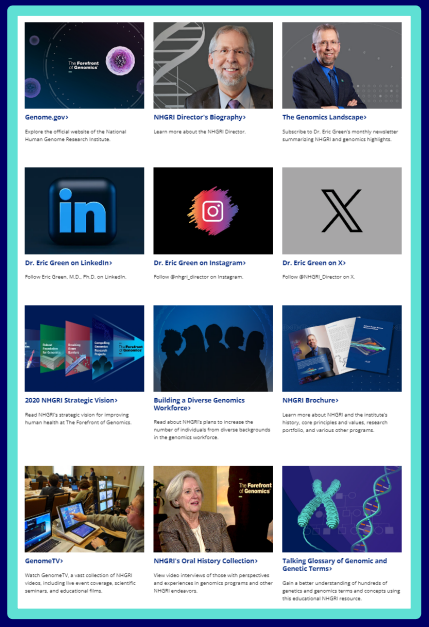
Diabetologia

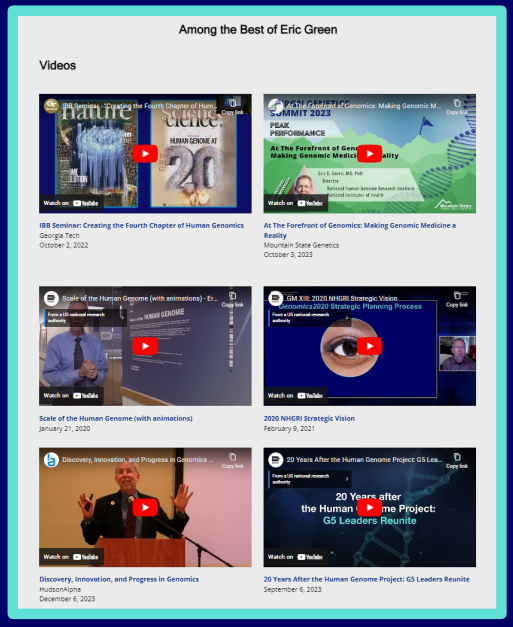
Single-cell transcriptomic profiling of human pancreatic islets reveals genes responsive to glucose exposure over 24h

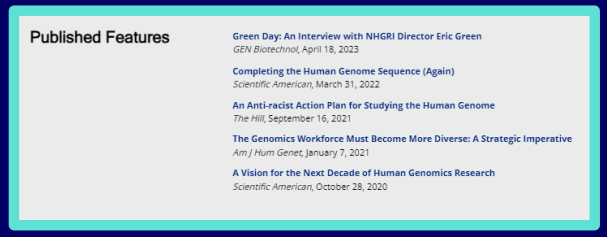
‘One-Stop-Shop’ to Stay Connected

genome.gov/stayconnected









NIH National Human Genome Research Institute

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National Human Genome Research Institute

Thanks!

Special Thanks!


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of Genomics®