





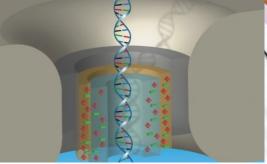
DIRECTOR'S REPORT

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

September 2023











Director's Report-Related Documents

September 2023



No.	Relevant Documents
1	Upcoming Retirement of Director, Division of Extramural Operations
2	New Incoming Director, Division of Extramural Operations
3	New NIH-ACMG Fellows
4	New AAAS Fellow
5	New ASHG-NHGRI Genomics and Public Service Fellowship Program

genome.gov/DirectorsReport

Open Session Agenda

Special Celebration:

100th Meeting of the NACHGR Eric Green

Presentation:

Research Priorities for Office of Data Science Strategy
Susan Gregurick

Concept Clearance:

Machine Learning and Artificial Intelligence Tools to Advance Genomic Translational Research

Sandhya Xirasagar

Open Session Agenda

Concept Clearances:

NHGRI Technology Development Coordinating Center Stephanie Morris

Genomic Medicine eConsult Service Renee Rider

Genomic Research Experiences for Data Scientists
Sandhya Xirasagar

Research Experiences in Genomic Research for Genetic Counselors Heather Colley

Open Session Agenda

Presentations:

Genomics & Society Working Group

Malia Fullerton

NHGRI Office of Communications and History of Genomics Program

Sarah Bates & Christopher Donohue

Using Machine Learning and Artificial Intelligence Methods in History of Genomics Archival Research

Spencer Hong & Thomas Stoeger

Clinical Sequencing Evidence-Generating Research
Gail Jarvik & Kyle Brothers

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- 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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Upcoming Retirement of Director, Division of Extramural Operations



Bettie Graham, Ph.D.



New Incoming Director, Division of Extramural Operations



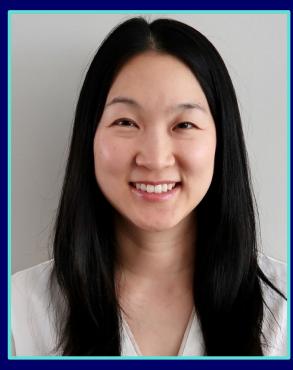
Jennifer Troyer, Ph.D.

New Extramural Program Director



lan Nova, Ph.D.

New NIH-ACMG Fellows



Jessica Chong, P.A.-C.



Karyn Roberts, Ph.D., R.N.

New AAAS Fellow



Molly Bird, Ph.D.

New ASHG-NHGRI Genomics and Public Service Fellowship Program





Genomics and Public Service Fellowship Program

Applications open on October 1, 2023





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New Director, Centers for Disease Control and Prevention



Mandy Cohen, M.D., M.P.H.



Retirement of Director, National Library of Medicine



Patricia Brennan, R.N., Ph.D.



Stephen Sherry, Ph.D.

New Associate Director, NIH Behavioral and Social Sciences Research



Jane Simoni, Ph.D.



New Director, National Institute of Allergy and Infectious Diseases



Jeanne Marrazzo, M.D.



NIH Mission Statement: Request for Information

Request for Information (RFI): Inviting Comments and Suggestions on Updating the NIH Mission Statement

Notice Number:

NOT-OD-23-163

Key Dates

Release Date: August 25, 2023

Response Date: November 24, 2023

Related Announcements

None

Issued by

NATIONAL INSTITUTES OF HEALTH (NIH)

Purpose

This Notice is a Request for Information (RFI) inviting feedback on a proposed update to the NIH mission statement. As the largest public funder of biomedical and behavioral research in the world, NIH works to turn scientific discoveries into better health for all. This RFI will inform NIH's efforts to update its mission statement to ensure that it reflects the NIH mission as accurately as possible.

Review of this entire RFI notice is encouraged to ensure your response is comprehensive and to have a full understanding of how it will be used.





Fiscal Year 2024 Appropriations

Chamber	Agency	Fiscal Year 2024 Draft L-HHS Appropriation	Fiscal Year 2023 Enacted L-HHS Appropriation	2023-2024 Difference \$	2023-2024 Difference %
Цацаа	NIH	\$44 B	\$46.8 B	-\$2.8 B	-6%
House	NHGRI	\$663.2 M	\$660.51 M	\$2.69 M	+0.4%
Senate	NIH	\$47.8 B	\$46.8 B	\$943 M	+2%
Seriale	NHGRI	\$663.2 M	\$660.51 M	\$2.69 M	+0.4%

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Mourning the Loss of Michael Ashburner





Michael Ashburner, Ph.D., Sc.D., F.R.S.

Mourning the Loss of Ian Wilmut





Ian Wilmut, Ph.D., O.B.E., F.R.S., F.Med.Sci., F.R.S.E.

Departure of Chief Executive Officer, American Society of Human Genetics



Mona Miller, M.P.P.



Double Helix: The Musical





Genomes in the "News..."



THE NEW YORKER

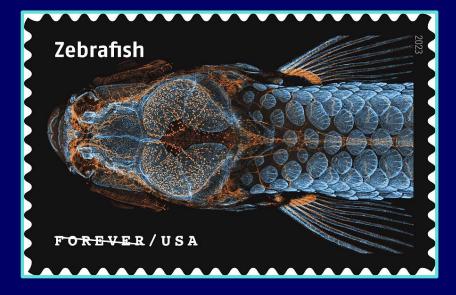
THE RACE TO SAVE THE WORLD'S DNA

A scientific rescue mission aims to analyze every plant, animal, and fungus before it's too late.

By Matthew Hutson

August 9, 2023





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My Week in St. Louis









Human Genome Reference Program



- Program renewal
- Applications received for two components
- Third component:

Open competition for Informatics Tools for the Pangenome

Applications due on November 1, 2023

Human Developmental Genotype-Tissue Expression (dGTEx)

Organ Procurement Organizations (OPOs)

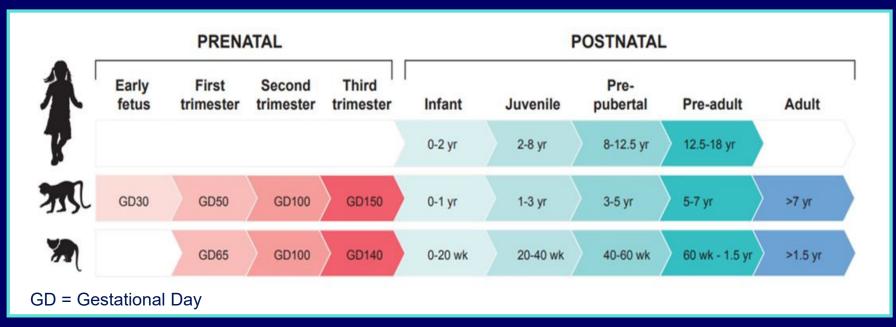




Donor Screening:

- 120 donors
- ~30 donors per age group
- 4 developmental age groups

Non-Human Primate Developmental Genotype-Tissue Expression (dGTEx)



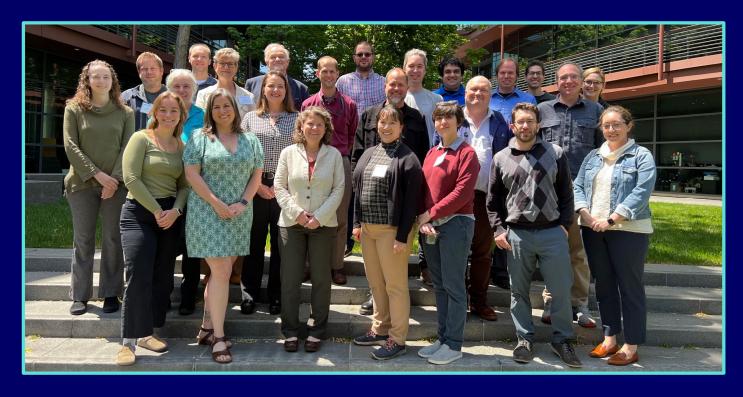
Rhesus macaque sampling to date:

- 18 macaques across 7 developmental stages
- 766 samples from 54 distinct tissues



GREGoR Consortium

First In-Person GREGoR Meeting



Focused discussion of high-priority research themes

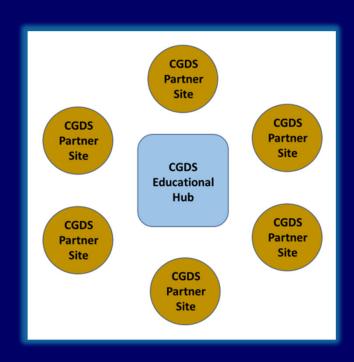
Educational Hub for Enhancing Diversity in Computational Genomics and Data Science

"The Hub" will leverage AnVIL and All of Us platforms to increase access to educational and research opportunities in CGDS

Awarded to North Carolina Agricultural and Technical State University

Funding opportunity:

Broadening Opportunities for Computational Genomics and Data Science Education



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

AnVIL renewed for Phase II



Addition of the AnVIL Clinical Resource (ACR)









Multi-Omics for Health and Disease

Disease Study Sites



Krzysztof Kiryluk



Joseph McCormick



Lida Chatzi



Tanika Kelly



Louise Laurent



Stephanie Christenson

'Omics Production Center





Gary Patti

Data Analysis & Coordinating Center







National Human Genome Research Institute





Document 21

Clinical Genome Resource (ClinGen)

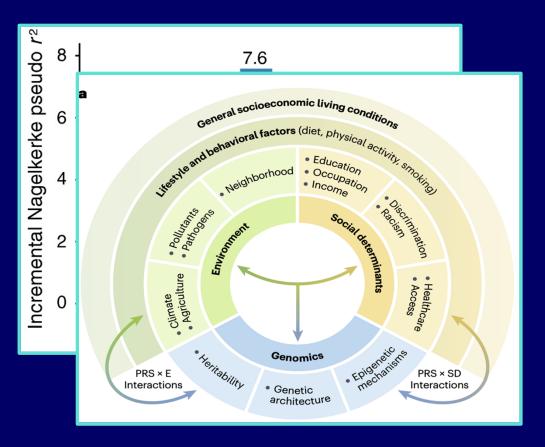
10th Anniversary



- American Society of Human Genetics November 2, 2023
 Lunch reception
- American College of Medical Genetics & Genomics March 12-16, 2024
- Curating the Clinical Genome May 9-10, 2024

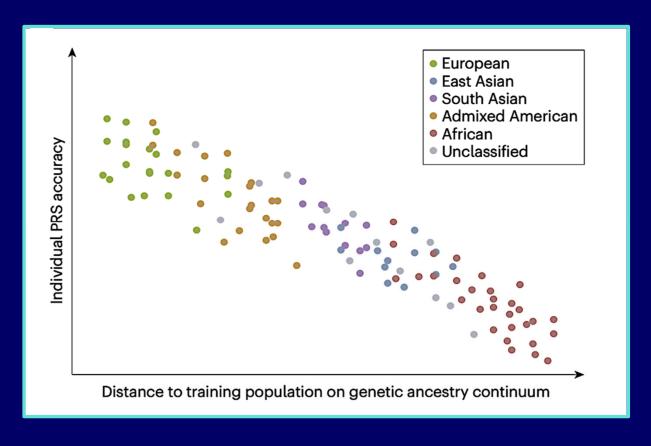
Stay tuned for event updates in the coming year!

Polygenic Risk Methods in Diverse Populations (PRIMED) Consortium



- Developing improved PRS models across multiple disease domains and populations
- Recent review: "Principles and methods for transferring polygenic risk scores across global populations"

Polygenic Risk Methods in Diverse Populations (PRIMED) Consortium



- Advancing PRS
 methods to recognize
 the continuous nature
 of genomic variation
- Pioneering Consortium secondary use of data in AnVIL

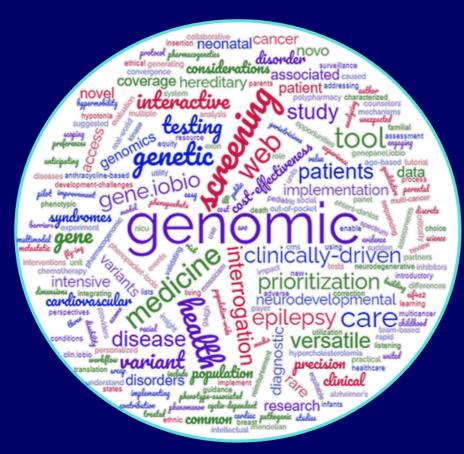


Advancing Genomic Medicine Research (AGMR)

Scientific Area	#
Pharmacogenomics	3
Computational Methods for Clinical Genomics	5
Demonstrating Effectiveness of Genomic Medicine	9
Health Disparities, Health Equity, and Access to Genomic Medicine	1
Return of Results and Outcome Research	8
Electronic Medical/Health Records-related Genomic Studies	5
Total	31

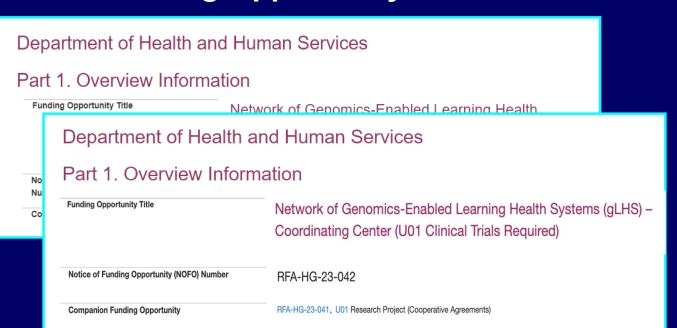
NIH Funding Source	#
Only AGMR	5
Multiple NIH Grants	26
Total	31

Publication Titles



Genomics-Enabled Learning Health Systems (gLHS)

- Establish a Network to identify and create LHS tools and resources to improve adoption of genomics into clinical care
- New Funding Opportunity Announcements:





Applications due on November 07, 2023

Genomics and Health Funding Opportunities



Funding Opportunity Title
Investigator-Initiated Research in Genomics and Health Equity
(R21 Clinical Trial Optional)

Activity Code
R21 Exploratory/Developmental Research Grant

Announcement Type
New

Notice of Funding Opportunity (NOFO) Number
RFA-HG-23-018

- Support investigator-initiated research in genomics and health equity (R01 and R21)
- Research spanning across genomics research areas
- Collaboration with NIA, NCI, All of Us, and ORWH

Applications due on November 8, 2023

Genomic Medicine XV: Genomics and Population Screening

November 8-9, 2023

Identify needs, opportunities, and challenges for using genomics in population screening

- Review current state of population genomic screening in U.S. and the available evidence of the impact of such screening
- Examine obstacles and opportunities for expanded population screening
- Identify research directions to inform expanded screening



NHGRI Funding Opportunity for Early Stage Investigators



Supporting Talented Early Career Researchers in Genomics





Applications due on February 27, 2024

Genome Technology Program

Advances in Genomic Technology Development 2023 Hybrid Meeting



- Commercialization opportunities
- Engagement with NHGRI consortia
- Overviews of NHGRI programs

Small Business Program

Funding Opportunities



- PAR-23-219: SBIR/STTR Commercialization Readiness Pilot Program Technical Assistance and Late-Stage Development
- NOT-HG-22-007: Methods Development for Genomic Studies of Genetic Variation, Function, and Disease
- NOT-HG-21-038: Genomics Education Interactive Digital Media Resources (SBIR/STTR)

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



Reimagining the Benefits of Genomic Science

SAVE THE DATE

June 10-12, 2024

Columbia University | NYC

Abstract submission open from September 5 to December 1, 2023



Add the event to your calendar!



Diversity Action Plan

 PAR-22-268, Initiative to Maximize Research Education in Genomics: Diversity Action Plan

 Support undergraduates and postbaccalaureates from diverse backgrounds

Next application due date is November 15, 2023

Diversity Research Supplements

 PA-23-189: Research Supplements to Promote Diversity in Health-Related Research

 Enhance the diversity of the research workforce by recruiting, mentoring and supporting individuals from diverse backgrounds

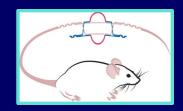
Applications accepted throughout the fiscal year until May 15



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Knockout Mouse Phenotyping Project (KOMP2)





 International Mouse Phenotyping Consortium Annual Conference, July 2023



- Online Workshop: "Home Cage Monitoring in Behavioral Phenotyping"
- NIH Office for Data Science Strategy supplement to KOMP2

Human Biomolecular Atlas Program (HuBMAP)



Nature cross-journal collection released July 20, 2023





Somatic Mosaicism Across Human Tissues (SMaHT)



- Network kickoff meeting in May 2023
- Hackathon with GREGoR & HGSVC



NIH Cloud Platform Interoperability Effort (NCPI)



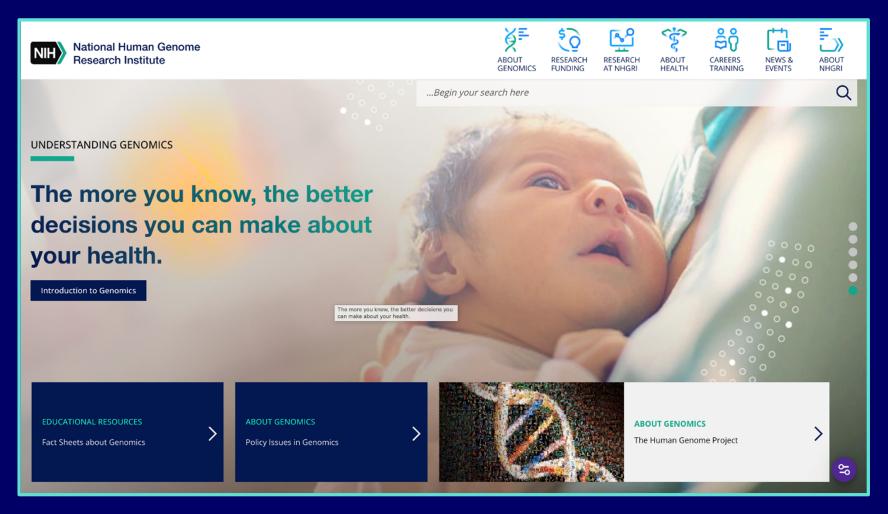


NCPI accepted as a 2024 GA4GH Driver Project

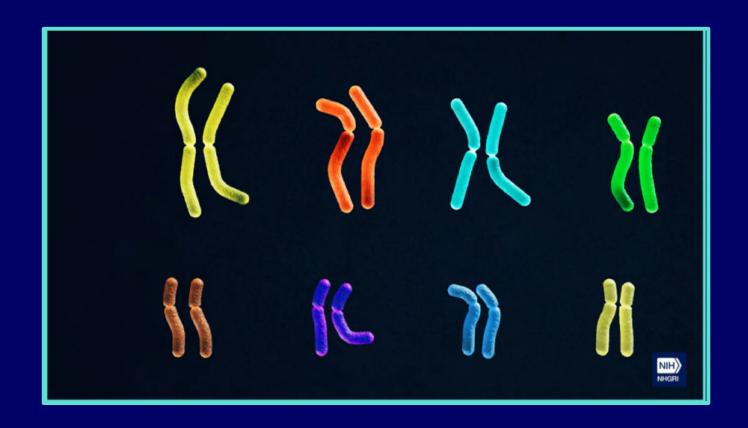
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Refresh of NHGRI Homepage for genome.gov



A Complete Human Y Chromosome Sequence



Virtual Reunion of the Human Genome Project 'G5'



NHGRI History of Genomics Program ArchivesSpace



NHGRI History of Genomics Program

Archival Spotlight: 1st Meeting of NHGRI Advisory Council

NATIONAL ADVISORY COUNCIL FOR HUMAN GENOME RESEARCH SUMMARY OF MEETING!

The National Advisory Council for Human Genome Research was convened for its first meeting at 10:00 A.M. on Tuesday, January 22, 1991, at the Hyatt Regency Hotel in Bethesda, Maryland. Dr. Elke Jordan, Deputy Director of the National Tenter for Human Genome Research (NCHGR) presided as chairman in the absence of Dr. James Watson, Director of the NCHGR. The members of the Council had met with staff of the Center from 8:00 to 10:00 in an orientation session to acquaint them with the Center and its programs and with the role of Advisory Councils in the peer review of research grants.

In accordance with the provisions of Public Law 92-463, the meeting was open to the public from 10:00 to 11:00 A.M. The meeting was closed to the public from 11:00 A.H. to 5:00 P.M. for the review, discussion, and evaluation of grant applications.

Dr. Francisco Ayala

Dr. David Botstein Dr. K. Danner Clouser Dr. Francis Collins

Dr. P. Michael Conneally

Dr. Jerome R. Cox

Dr. Norman Davidson Dr. Joe W. Gray

Dr. Joe W. Gray Dr. Kay Jamison

Dr. Shirley Tilghman Dr. Donald A. B. Lindberg**

* Ad Hoc Consultant

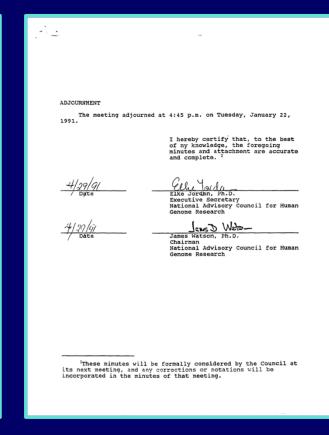
** Ex Officio

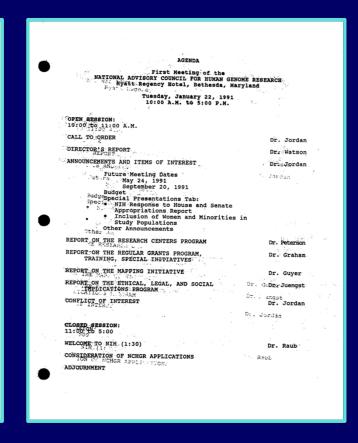
Staff of the National Center for Human Genome Research attending, in addition to Dr. Jordan, were:

Midge Bajefsky, Grants Technical Assistant Dr. David Benton, Assistant to the Director for Scientific Dr. David Benton, Assistant t Data Management Erin Burgess, Budget Officer

Michelle Coleman, Committee Management Officer

'For the record, it is noted that to avoid a conflict of interest, Council members absent themselves from the meeting when the Council discusses applications from their respective institutions or in which a conflict of interest may occur. Members are asked to sign a statement to this effect. This does not apply to "en bloc" actions.





2023 Virtual Short Course in Genomics



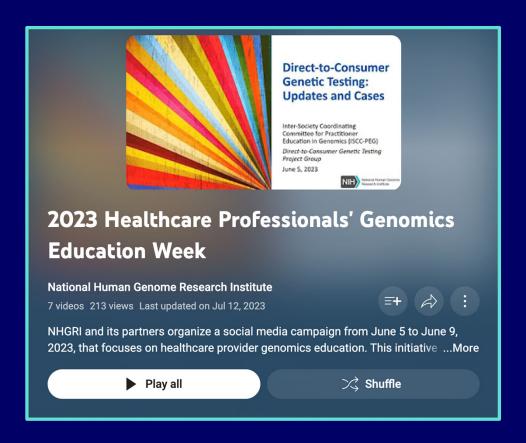


New Gene-Environment Interactions Lesson Plans



- NHGRI collaboration with National Science Teaching Association
- Teacher training webinars 68 teacher participants
- 2022 Lesson Plans statistics:
 10K clicks
 1.6M impressions
 940,000 students served

Healthcare Professionals' Genomics Education Week



New educational resources:

- Nursing Genomics Frequently Asked Questions
- Pharmacogenomics Learning Series
- Patient Module: Autism Spectrum Disorder and Vaccines

YouTube Playlist

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Retirement of Intramural Investigator



William Pavan, Ph.D.

New Deputy Scientific Directors, NHGRI Intramural Research Program



Paul Liu, Ph.D.



Sara Chandros Hull, Ph.D.



Shawn Burgess, Ph.D.

30th Anniversary of the NHGRI Intramural Research Program



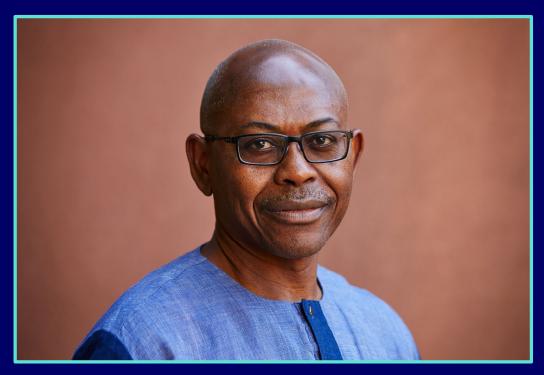


Todd Golub, M.D.

Jeffrey M. Trent Lectureship in Cancer Research Series

November 13, 2023 2-3 p.m. ET

New York Academy of Medicine Medal for Distinguished Contributions in Biomedical Science





Charles Rotimi, Ph.D.



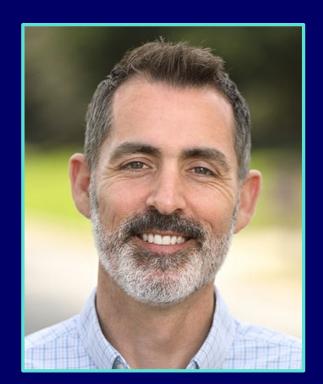
2024 Breakthrough Prize in Life Sciences



Ellen Sidransky, M.D.



Thomas Gasser, M.D., Ph.D.



Andrew Singleton, Ph.D.

Document 46

Genomic Science and Health Equity Postdoctoral Fellowship

Training to use genetic, genomic, and pharmacogenomic approaches to advance minority health and health equity





Next year's fellowship application available soon



Lauren Edgar, DNP, MSN-Ed., RN, FNP

NHGRI Intramural Research Highlights



Leukemia

Characterization of leukemia progression in the Cbfb-MYH11 knockin mice by single cell RNA sequencing



PRENATAL **DIAGNOSIS**

Patients' perspectives on prenatal screening results that suggest maternal cancer: A qualitative analysis



AJHG

Haplotype-based analysis resolves missing heritability in oculocutaneous albinism type 1B

'One-Stop-Shop' to Stay Connected genome.gov/stayconnected





2020 NHGRI Strategic Vision>

Building a Diverse Genomics

in the genomics workforce.

Read about NHGRI's plans to increase the

number of individuals from diverse backgrounds

Read NHGRI's strategic vision for improving



NHGRI Brochure>

Watch GenomeTV, a vast collection of NHGRI

videos, including live event coverage, scientific

seminars, and educational films.



Subscribe to Dr. Eric Green's monthly newsletter Follow Dr. Eric Green on Twitter summarizing NHGRI and genomics highlights.



Learn more about NHGRI and the institute's history, core principles and values, research portfolio, and various other programs.



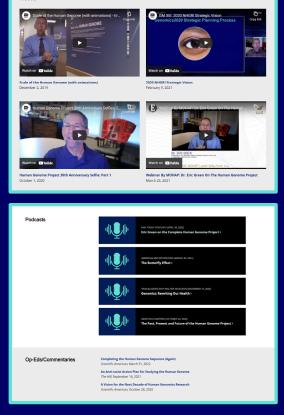
Gain a better understanding of hundreds of genetics and genomics terms and concepts using this educational NHGRI resource.



NHGRI's Oral History Collection>



and experiences in genomics programs and other NHGRI endeavors



Among the Best of Eric Green



Thanks!



Special Thanks!

