

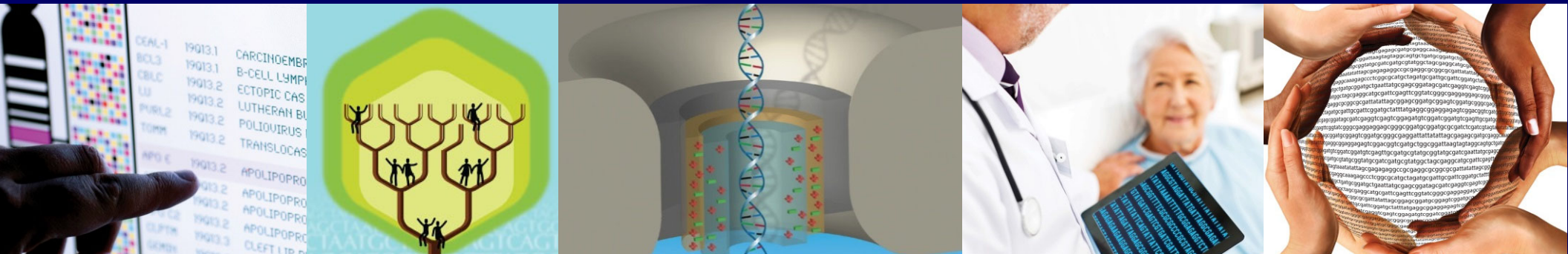


—
The **Forefront**
of **Genomics**
—

DIRECTOR'S REPORT

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

September 2021



Director's Report-Related Documents

September 2021

Director's Report

Director's Report

No.	Relevant Documents
1	New NHGRI-ASHG Fellows
2	New NIH-ACMG Fellows
3	NIH Deputy Director for Intramural Research, Stepping Down
4	Departure of President and Executive Director, Foundation for NIH (FNIH)
5	New NIH Chief Officer, Scientific Workforce Diversity

genome.gov/DirectorsReport

Document #



Open Session Presentations

Presentations:

**The National Cancer Institute: Leading Cancer Research
in 2021 and Beyond**

Ned Sharpless

Genomics, Genetics, and the Eye

Michael Chiang

Open Session Presentations

Concept Clearances:

**Initiative to Maximize Research Education in Genomics:
Courses**

Tina Gatlin

**Enhancing Diversity in Cloud-based Genomic Data Science
Education**

Shurjo Sen

Open Session Presentations

Presentations:

Data Science Working Group

Mark Craven

Community Engagement in Genomics Working Group

Greta Goto

Maya Sabatello

Report on Multi-Omics in Health and Disease Workshop

Joannella Morales

Howard Chang

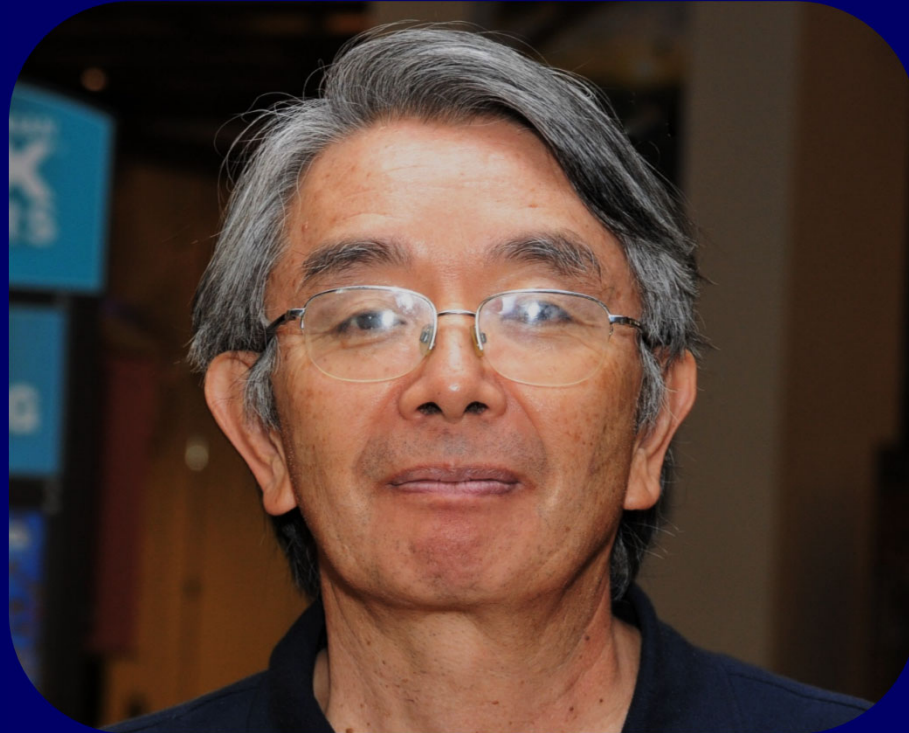
Director's Report Outline

- 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 Scientific Review Officer



Ken Nakamura, Ph.D.

Departure of Extramural Program Director



Luis Cubano, Ph.D.

New NHGRI-ASHG Fellows



Nichole Holm, Ph.D.
Genetics & Public Policy Fellow



Delaney Pagliuso, Ph.D.
**Genetics Education &
Engagement Fellow**

New NIH-ACMG Fellows



Deepika Burkhardt, D.O.

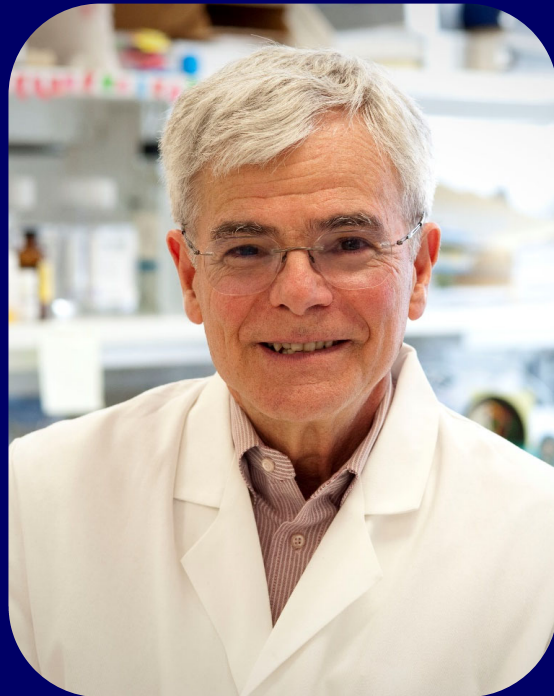


Nguyen Park, M.S., PA-C

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NIH Deputy Director for Intramural Research, Stepping Down



Michael Gottesman, M.D.



Departure of President and Executive Director, Foundation for NIH (FNIH)



Maria Freire, Ph.D.



David Wholley, M. Phil.

New NIH Chief Officer, Scientific Workforce Diversity

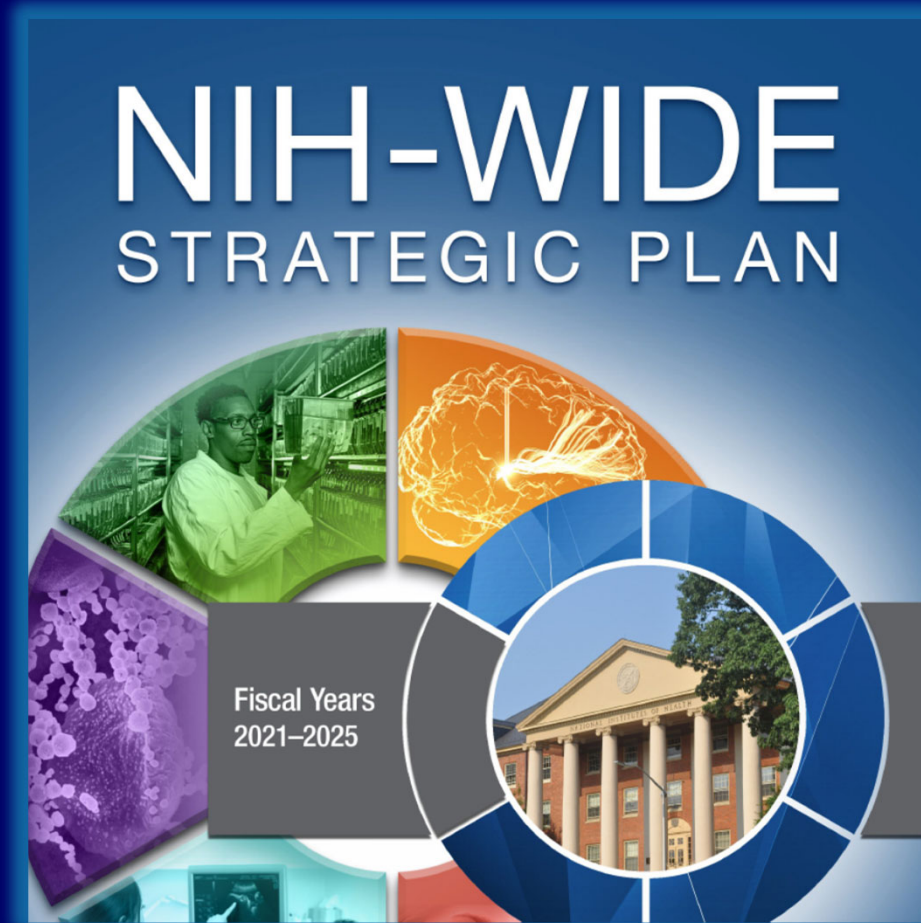


Marie Bernard, M.D.



National Institutes of Health
Office of the Director
Scientific Workforce Diversity

2021-2025 NIH-Wide Strategic Plan



NIH-FDA Next-Generation Sequencing and Radiomics Workshop

Resource Requirements for Acceleration of Clinical Applications

September 29-30 Virtual Workshop

Related Requests for Information (RFIs):

- Open until November 1
- Critical Resource Gaps and Opportunities to Support:
 - NOT-OD-21-162: Next generation sequencing
 - NOT-OD-21-163: Radiological tool development and clinical data interpretation



Fiscal Year 2022 Appropriations

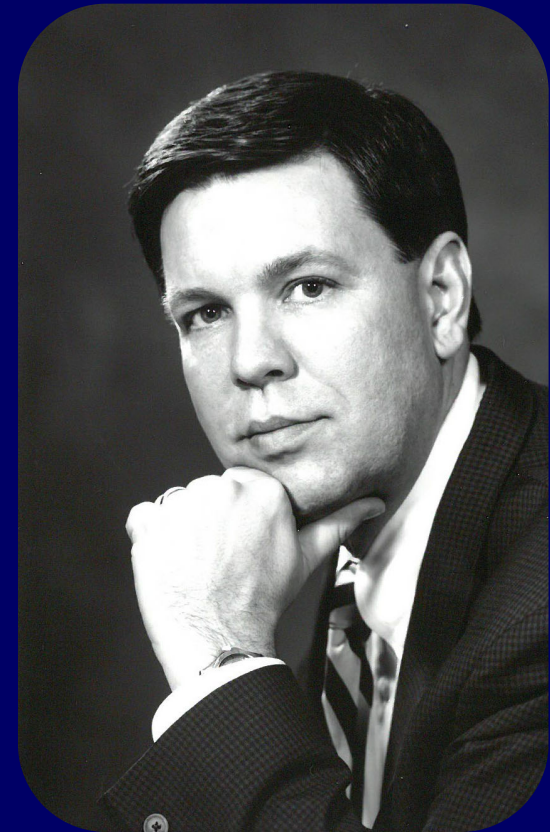
	Fiscal Year 2021 Enacted Labor-HHS Appropriation	Fiscal Year 2022 House Labor-HHS Appropriation	\$ Increase	% Increase
NIH	\$42.9 B	\$49.4 B	\$6.5 B*	15%
NHGRI	\$615.8 M	\$646.3 M	\$30.5 M	5%

*House language designates that ARPA-H would receive \$3 billion of the increase for NIH

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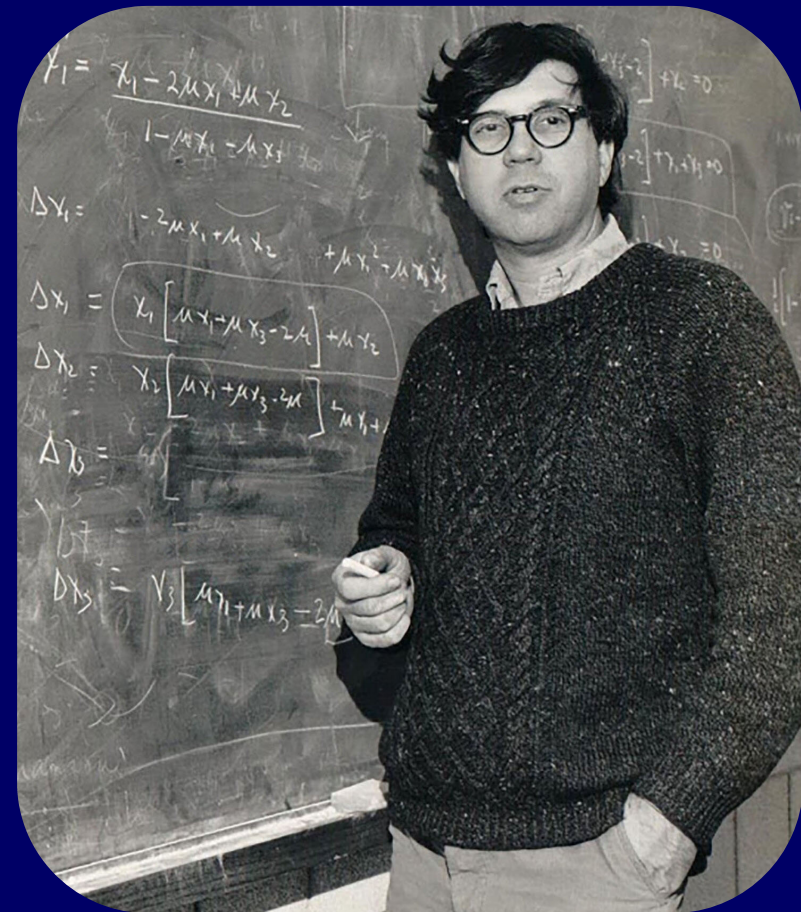
Mourning the Loss of Steve Warren



Mourning the Loss of Daniela Gerhard



Mourning the Loss of Richard Lewontin



New President-Elect, American Society of Human Genetics



Brendan Lee, M.D., Ph.D.



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Genomics Research Elucidates Genetics of Rare Disease (GREGoR) Consortium



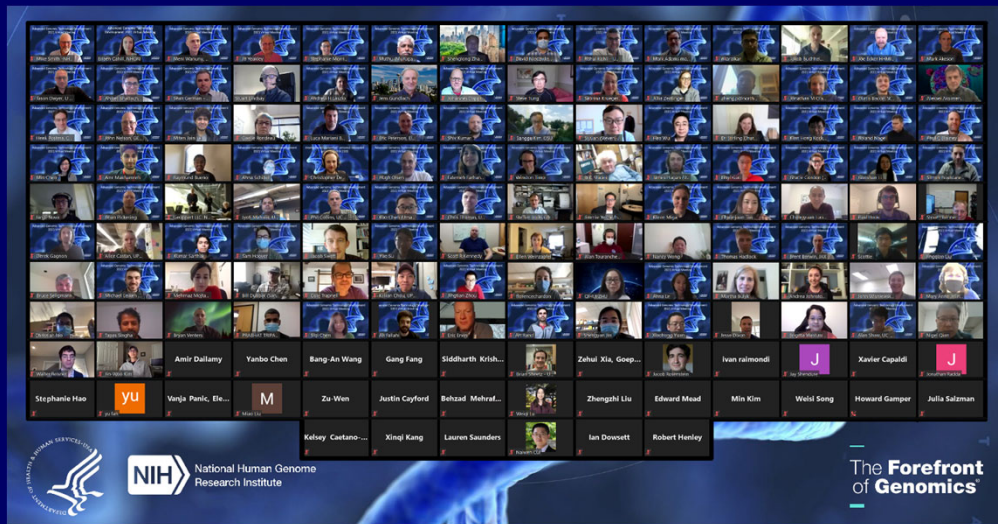
Baylor
College of
Medicine



- Program launched in Summer 2021
- New approaches to find genetic bases of rare diseases

Technology Development Program

Advanced Genomic Technology Development Meeting



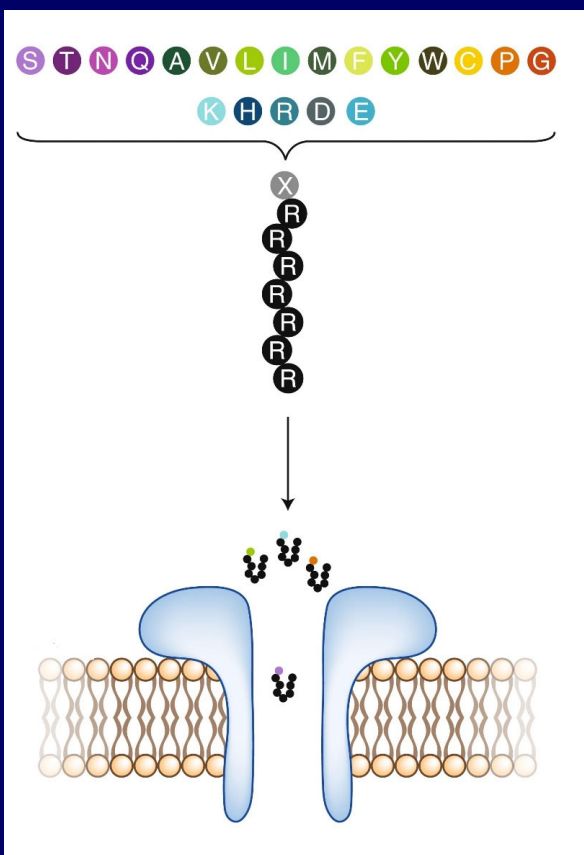
May 2021

Technology Development Coordinating Center



- Facilitating collaborations
- Promoting standards
- Developing resources
- Disseminating information

Technology Development Program



*Courtesy of Stefan Howorka
& Zuzanna Siwy*

Funding Opportunities:

- **Technology Development for Single-Molecule Protein Sequencing**

Collaboration with NCI and NIAID

RFA-HG-21-001 (R01, also linked R21 RFA
and R43/R44 PAR)

First due date: October 1, 2021

Small Business Program



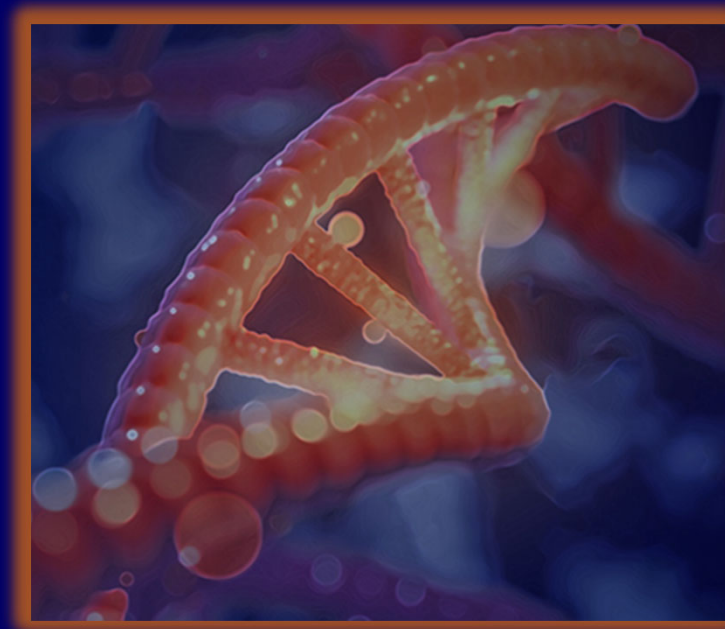
Funding Opportunities

- **Development of Highly Innovative Tools and Technology for Analysis of Single Cells**
 - PA-20-025 (STTR R41/R42 and linked SBIR R43/R44)
 - Standard receipt dates
- **Small Business Initiatives for Innovative Diagnostic Technology for Improving Outcomes for Maternal Health**
 - NOT-EB-21-001, Notice of Special Interest (NOSI)
 - STTR R41/R42 and SBIR R43/R44
 - Standard receipt dates

Molecular Phenotypes of Null Alleles in Cells (MorPhiC)

Funding Opportunities:

- **Data Production Research and Development Centers**
RFA-HG-21-029
- **Data Analysis and Validation Centers**
RFA-HG-21-030
- **Data Resource and Administrative Coordinating Center**
RFA-HG-21-031



Application Due Date: November 1, 2021

Centers for Excellence in Genomic Science (CEGS)

- **Chao-Ting Wu (Harvard Medical School)**

- *Center for Genome Imaging*



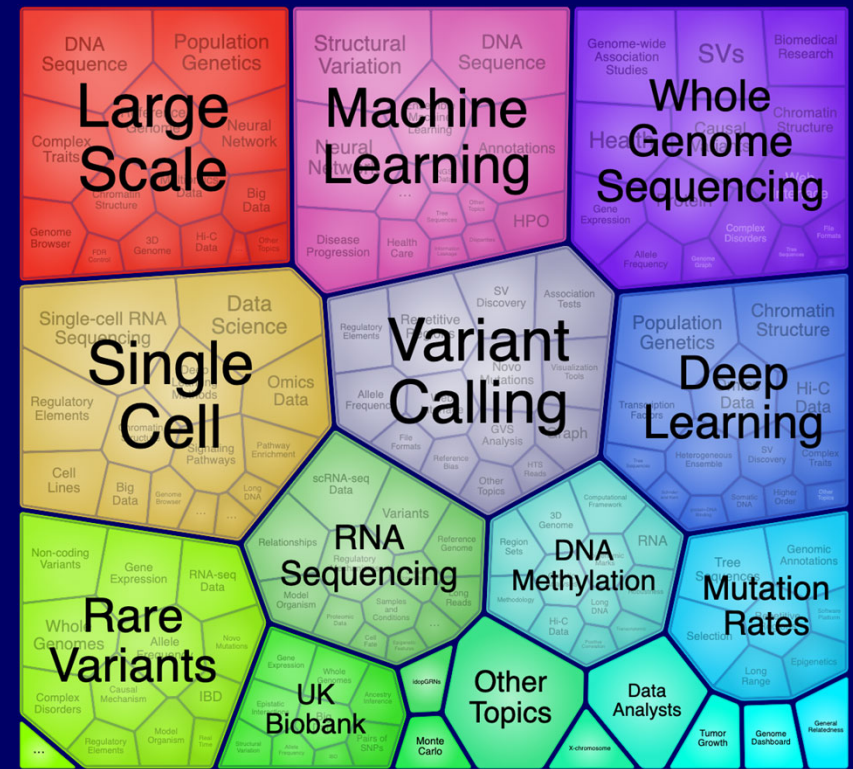
- **Chuan He (University of Chicago)**

- *Center for Dynamic RNA Epitranscriptomes*



Computational Genomics and Data Science Program

- **New funding opportunities released in July**
- **Support innovative research in computational genomics and data science**
- **First due date: October 5, 2021**



GWAS Catalog

nature https://doi.org/10.1038/s41586-021-03767-x

Accelerated Article Preview

Mapping the human genetic architecture of COVID-19

Received: 2 March 2021 COVID-19 Host Genetics Initiative

Accepted: 23 June 2021

Accelerated Article Preview Published online 8 July 2021

Cite this article as: COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19. *Nature* <https://doi.org/10.1038/s41586-021-03767-x> (2021).

This is a PDF file of a peer-reviewed paper that has been accepted for publication. Although unedited, the content has been subjected to preliminary formatting. Nature is providing this early version of the typeset paper as a service to our authors and readers. The text and figures will undergo copyediting and a proof review before the paper is published in its final form. Please note that during the production process errors may be discovered which could affect the content, and all legal disclaimers apply.



GWAS Catalog

The NHGRI-EBI Catalog of human genome-wide association studies

Examples: breast carcinoma, rs7329174, Yao, 2q37.1, HBS1L, 6:16000000-25000000

As of August 16, 2021, the GWAS Catalog contains 5,273 publications and 276,696 associations.

- Repository for summary statistics from COVID-19 Host Genetics Initiative
- Curation team reached milestone of curating >5,000 papers

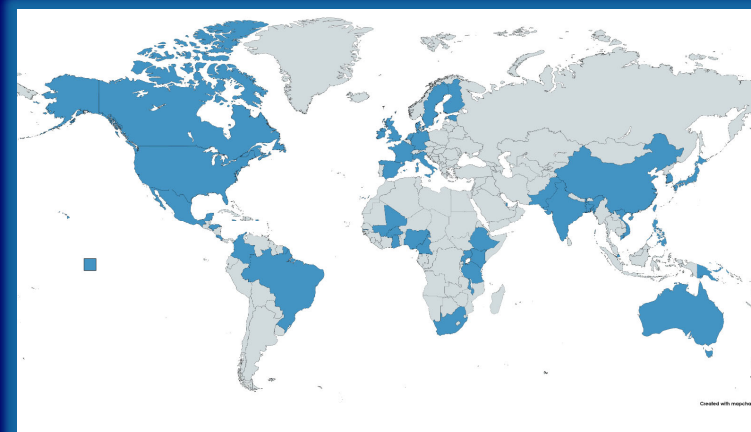
Polygenic Risk Methods in Diverse Populations (PRIMED) Consortium



Improve applicability of PRS in diverse populations



Optimize the integration of large-scale, harmonized genomic and phenotype data



Sally Adebamowo (U. of MD)

David Conti (U. of Southern California)

Amit Khera (Massachusetts General and Broad Institute)

Yun Li (U. of North Carolina)

Josep Mercader (Broad Institute)

Bogdan Pasaniuc (U. of California, Los Angeles)

Kenneth Rice (U. of Washington; Coordinating Center)



Clinical Genome Resource (ClinGen) Virtual Retreat 2021

~ 470 registrants from 38 countries

Topics:

Best practices for gene and variant curation
Curator perspectives
ACMG/AMP updated guidelines
Engaging under-represented Groups
Navigating ClinGen resources





Clinical Sequencing Evidence-Generating Research (CSER) Program

Genetics
inMedicine

www.nature.com/gim



BRIEF COMMUNICATION

GenomeDiver: a platform for phenotype-guided medical genomic diagnosis

Nathaniel M. Pearson^{1,13}, Christian Stolte^{2,11,13}, Kevin Shi^{2,13}, Faygel Beren³, Noura S. Abul-Husn^{4,5,6}, Gabrielle Bertier⁷, Kaitlyn Brown⁸, George A. Diaz^{2,7}, Jacqueline A. Odgis⁴, Sabrina A. Suckiel⁴, Carol R. Horowitz⁷, Melissa Wasserstein^{8,9}, Bruce D. Gelb⁷, Eimear E. Kenny^{4,5,6}, Charles Gagnon², Vaidehi Jobanputra², Toby Bloom^{2,12} and John M. Greally^{8,9,10}✉

ANNUAL
REVIEWS

Annual Review of Genomics and Human Genetics

Scaling Genetic Counseling
in the Genomics Era

Laura M. Amendola,¹ Katie Golden-Grant,¹
and Sarah Scollon²

ORIGINAL ARTICLE

GENETICS

Pitfalls and challenges in genetic test interpretation:
An exploration of genetic professionals experience
with interpretation of results

Katherine E. Donohue¹ ✉ | Catherine Gooch^{2,3} | Alexander Katz⁴ |
Jessica Wakelee⁵ | Anne Slavotinek⁶ ✉ | Bruce R. Korf²

Survey sent of genetics professionals asking about experiences with test result misinterpretation

- 83% of respondents described at least one example of misinterpretation
- Incorrect result interpretation is biggest perceived factor leading to misinterpretation
- Results highlight need for better communication among labs, providers, and families

Multi-omics in Health and Disease Workshop

The poster features a central graphic of a DNA double helix in white on a dark blue circular background. This central element is surrounded by several overlapping, semi-transparent circles in various colors (blue, green, yellow, orange, red, purple). Four large, curved bands radiate from the center, each containing a different type of data: a grey band with DNA base pairs (A, G, C, T), a green band with binary code (0s and 1s), a purple band with binary code (0s and 1s), and an orange band with binary code (0s and 1s). The background is white with scattered small colored dots.

**Multi-omics in
Health and Disease**
Current Applications, Challenges
and Future Directions

June 17-18, 2021

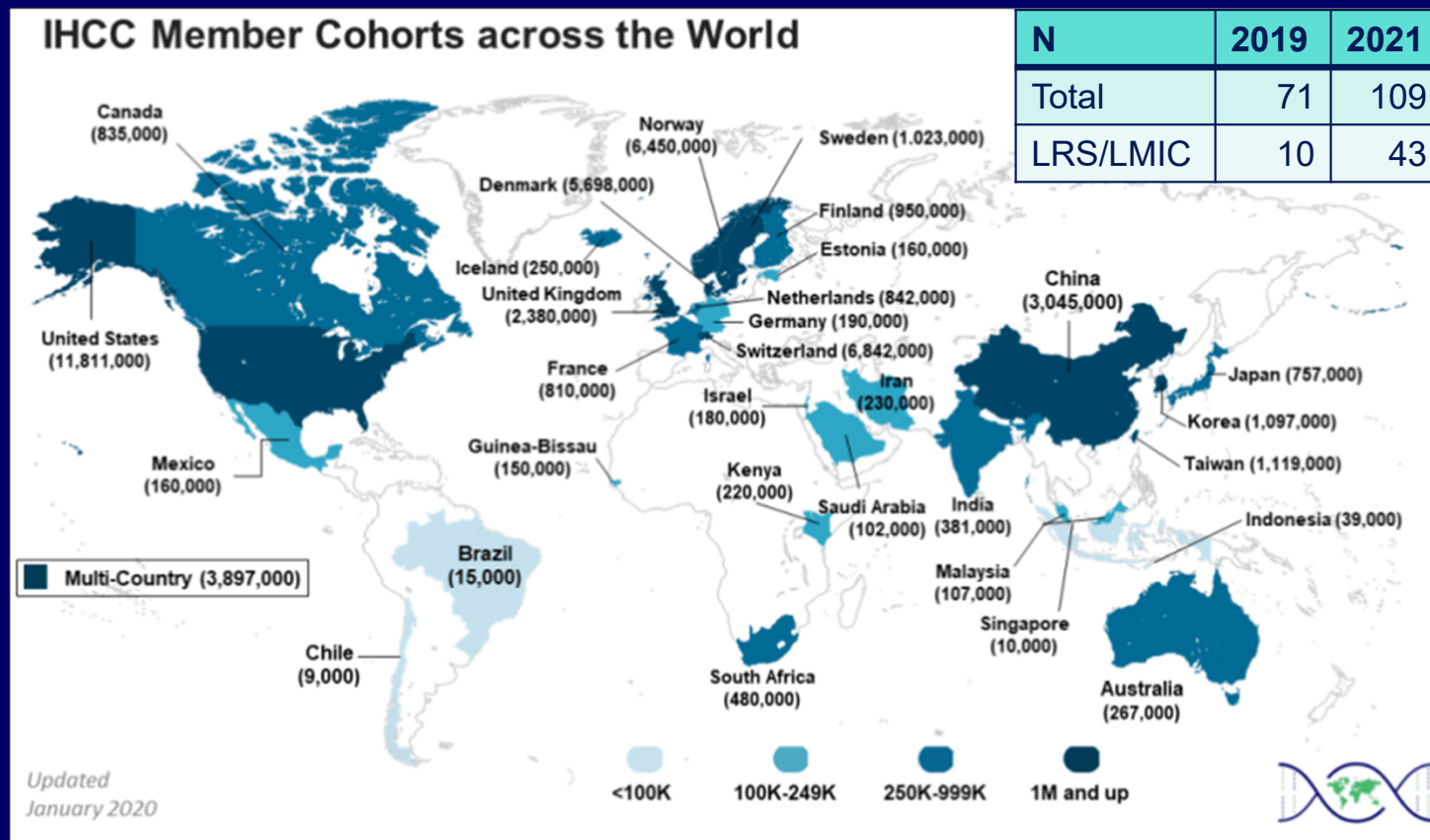
 NIH
NHGRI

 The **Forefront**
of **Genomics**

International 100K+ Cohort Consortium (IHCC)

4th International Cohorts Summit

- Galvanize IHCC around a long-term strategic plan
- Training, Sharing, and Capacity Development Working Group



The 5th ELSI Congress

The **5th**
ELSI
CONGRESS

ELSIcon2022: Innovating for a Just and Equitable Future

Save the Date

June 1-3, 2022

Columbia University

Abstract Submission: Open from September 13 – December 1, 2021

Training and Career Development

New Diversity-Focused FOAs

PAR-21-143

Next due date: December 8, 2021

NHGRI Predoctoral to Postdoctoral Transition Award for Diverse Genomics Workforce (F99/K00)

PAR-21-214

Next due date: February 12, 2022

Short-term Mentored Research Career Enhancement Award to Promote Diversity (K18)

Training and Career Development

New Diversity-Focused FOAs

RFA-HG-21-033

Due date: December 1, 2021

Genome Research Experiences to Attract Talented Undergraduates into the Genomics Field to Enhance Diversity (GREAT, R25)

RFA-HG-21-041

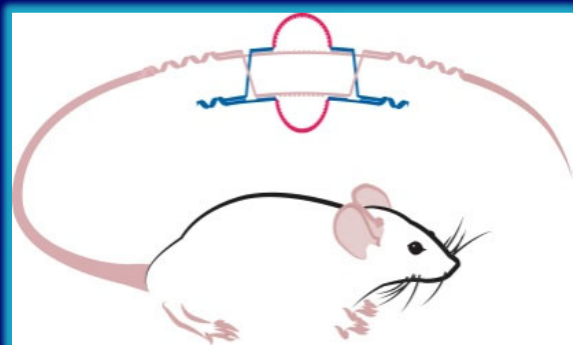
Due date: February 22, 2022

New Investigators to Promote Workforce Diversity in Genomics, Bioinformatics, or Bioengineering and Biomedical Imaging Research (R01 Clinical Trial Optional)

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Knockout Mouse Phenotyping Project (KOMP2)



Limited Competition Funding Opportunities:

- RFA-HG-21-036

Knockout Mouse Production and Phenotyping Project (UM1)

- RFA-HG-21-037

Knockout Mouse Phenotyping Project Data Coordination Center
and Database (UM1)

Application due date: November 1, 2021

Human Biomolecular Atlas Program (HuBMAP)

Two funding opportunities:



- **HuBMAP Integration, Visualization, & Engagement (HIVE) Collaboratory**

OTA-21-012

Application due date: December 3, 2021

- **Demonstration Projects**

RFA-RM-21-027

Application due date: November 19, 2021

All of Us

RESEARCH PROGRAM

Researcher Workbench

Year One: By The Numbers

Participant Data Available in the Researcher Workbench



263,000+

Physical Measurements



203,000+

Electronic Health Records (EHR)



8,000+

Fitbit Records



7 Surveys

Capturing participant data such as:
 • Lifestyle
 • Health care utilization and access
 • COVID-19 participant experience

Participants at a Glance



Participants
316,000+



80%

are from underrepresented communities, including

50% racial and ethnic minorities.

Research Currently Underway



575
Active projects



13
Publications in peer-reviewed journals



Top conditions being studied

in the Researcher Workbench include:
 • Cardiovascular disease
 • Hypertension • Cancer
 • Mental health • Diabetes

Our Researchers

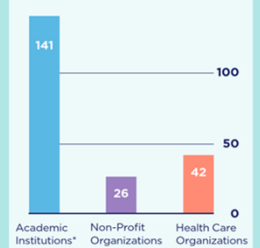


826
Registered Researchers

across a range of institutional roles and career stages.



209
Institutions



*Includes 23 Historically Black Colleges and Universities and Hispanic-Serving Institutions.

Data and Usage



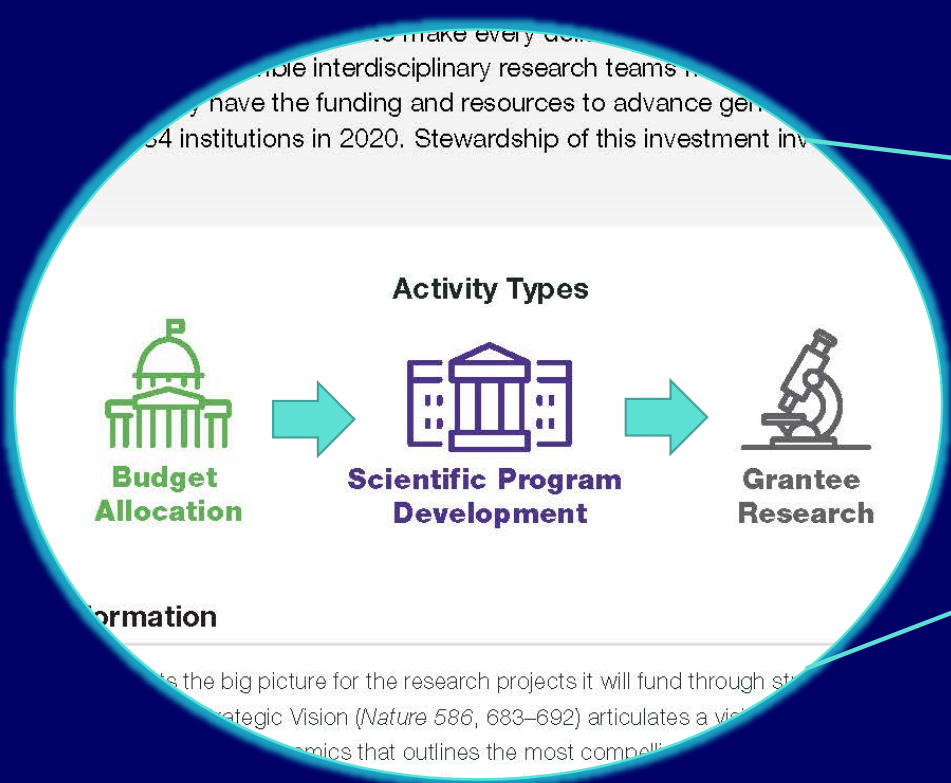
More than **28,000** estimated compute hours spent.

Data as of May 24, 2021

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New Resource on NHGRI Grant Funding Process



NHGRI Grant Funding, Review and Award

The Path from NHGRI Appropriations to Scientific Discovery

The National Institutes of Health (NIH) is the largest public funder of biomedical research in the world. NIH provides funding through its 27 institutes and centers, each with unique research missions, priorities, budgets, and funding strategies. The National Human Genome Research Institute (NHGRI) builds on its leadership role in sequencing and understanding the human genome to advance genomics research at NIH and the scientific and medical community at-large.

As one of the smaller institutes at NIH, NHGRI works to make every dollar we spend as impactful as possible by using innovative, team-based methods. We assemble interdisciplinary research teams made up of scientists, clinicians, ethicists and other experts, and we ensure they have the funding and resources to advance genomic research. NHGRI funded \$430 million in extramural research at 184 institutions in 2020. Stewardship of this investment involves a rigorous application and review process.



Budget Allocation



Scientific Program Development



Grantee Research

Activity	Information	Timeframe
Strategic Planning of NHGRI Research Priorities	NIH sets the big picture for the research projects it will fund through strategic planning. The NHGRI Strategic Vision (NIH, 2018) articulates a vision for genomics research at The Potomac Center that guides the most compelling opportunities for human genomics research, informed by a multi-year planning process that involved over 50 events nationally and internationally with a broad and diverse range of stakeholders.	Ongoing process
Congressional Justification	The annual NHGRI Congressional Justification provides the Senate and House Appropriations Committees details on needs and justifications for research and research support activities (e.g., infrastructure and administrative).	-
Appropriation of Funds	The federal appropriations process provides NHGRI with budget authority each fiscal year. Budget authority is given to carry out Section 301 and Title IV of the Public Health Service Act with respect to human genome research. Authority provided by appropriations is not immediately available for obligation or expenditure.	Before Oct. 1 or Variable
Apportionment by the Office of Management and Budget	The Office of Management and Budget (OMB) distributes funding available for obligation to NIH. NIH must receive the funds from OMB no later than 30 days after the start of the new fiscal year or 30 days after enactment of the appropriation act.	Before Nov. 1 or < 30 days after appropriation
Allocation by NIH Office of Budget	Allocation by the NIH Office of Budget (OB) transfers authority over the funds as allowances from the director of NIH to directors of institutes and centers, such as the NHGRI director, to incur obligations within a specified amount, within a specified period.	-
Sub-Allotment by NHGRI Budget Office	The NHGRI Budget Office further divides these funds within the spending budget for the Institute, following NHGRI's policies and priorities. First, NHGRI funds projects and centers that have commitments from previous years. NHGRI funds each grant for about 3 years, on average. The Institute uses the remainder of the funds on operations, new strategic scientific funding, small business grants, and ethical, legal and social implications (ELSI) research.	-



Genomics and the Media

Previous events



Apoorva Mandavilli
Reporter (Science and
Global Health),
The New York Times

May 20, 2021



John Inglis
Executive Director, Cold
Spring Harbor Laboratory
Press; Co-founder,
bioRxiv, medRxiv

July 28, 2021



Amy Harmon
National Correspondent
(Science and Society),
The New York Times

September 20, 2021



Dorothy Roberts
Professor of Law and
Sociology, Civil Rights,
University of Pennsylvania;
Author, *Fatal Invention*

November 4, 2021



Elizabeth Wayne
Assistant Professor of
Chemistry, Carnegie Mellon
University; Co-host,
PhDivas Podcast

January 20, 2022



Joe Palca
Science Correspondent,
NPR

March 2022



Magdalena Skipper
Editor in Chief, Nature;
Chief Editorial Advisor,
Nature Research

May 25, 2022

**Conversations with
trailblazing science
communicators**

Infographic on Completing the Human Genome Sequence

Why was it so difficult to fully complete the human genome sequence?

The Human Genome Project ended in 2003, but genomic researchers had not yet determined every last base (or letter) of the human genome sequence. Instead, they had only completed about 92% of the sequence at that time. Why did they stop there?

Wow, this doesn't look simple to me!

Reason 1

The human genome contains a massive amount of DNA.

The human genome consists of about 3 billion bases in a precise order, each of which can be represented by a letter (G, A, T or C). A genome's sequence cannot be read out end-to-end. Rather, researchers must first determine the sequence of random pieces of DNA and then use those smaller sequences to put the whole genome sequence back together like a massive puzzle.

Good thing I'm smart!

Reason 2

Some parts of our DNA are painfully repetitive.

Some sections of the human genome consist of long, repetitive stretches of letters that are difficult to put in the right place. Over the past few decades, researchers developed new technologies to read longer stretches of DNA — from only about 500 to now over 100,000 letters at a time — which allowed them to assemble the full length of the most difficult repeats.

Where do these go??

After meeting with my...

Reason 3

The first 92% was hard. The last 8% was excruciating.

Those DNA repeats and other obstacles stood between the genomic researchers and the final 8% of the human genome sequence until new laboratory and computational technologies were developed. It took almost twice as long to finish the last 8% of the human genome as it did the first 92%.

Wow, we made it!

Reason 4

The last 8% needed a generation of dedicated genomic researchers with a vision.

Even with new technologies, genome sequencing is still tough, time-consuming work that requires a lot of skill and dedication. The current generation of genomic researchers are true perfectionists and brought everything together to finally complete the human genome sequence.

This is a failure!

It's a much bigger sequence than I thought!

These new methods are so powerful!

It's almost twice as long to finish the last 8% of the human genome as it did the first 92%!

genome.gov

NIH NHGRI

Reason 1

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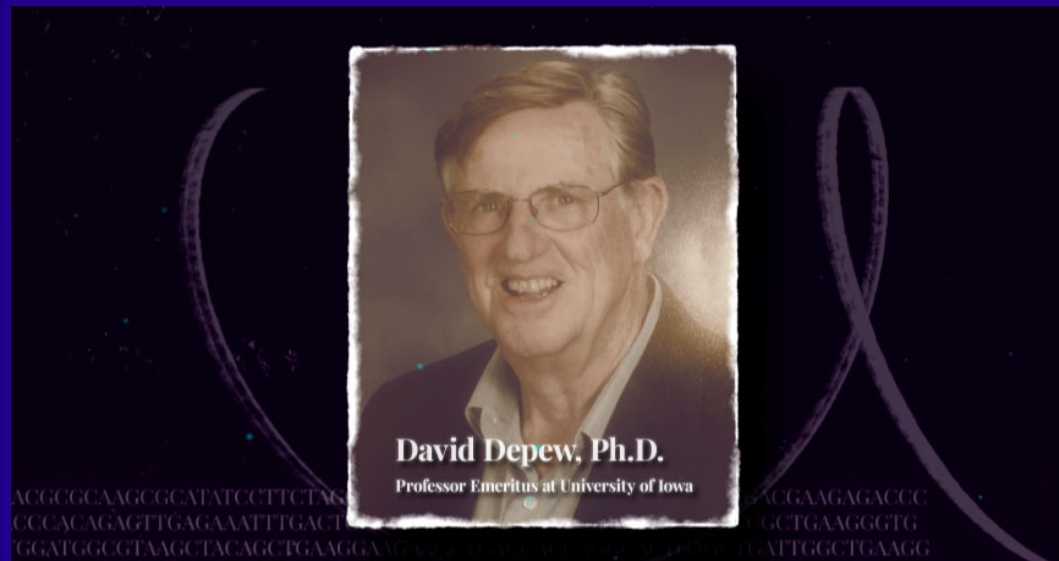
Road trip anyone?

If you printed out the ~3 billion letters of the human genome in size 12 font, it would stretch from Houston to Boston!

1,804 miles

Note: Only 20 letters of DNA sequence shown. To imagine what ~3 billion letters of the human genome sequence would look like, multiple this by 153 million!

NHGRI History of Genomics Program Conference Celebrating the Work of David Depew



- Eight internationally recognized scholars
- Presentations drawing from themes in Depew's work
- 108 total attendees

NHGRI History of Genomics Program

Symposium on the History of Eugenics and Scientific Racism



- December 2-3, 2021
- Examine history of eugenics, scientific racism, and complex legacies in the modern health sciences

UNITED STATES
HOLOCAUST
MEMORIAL
MUSEUM

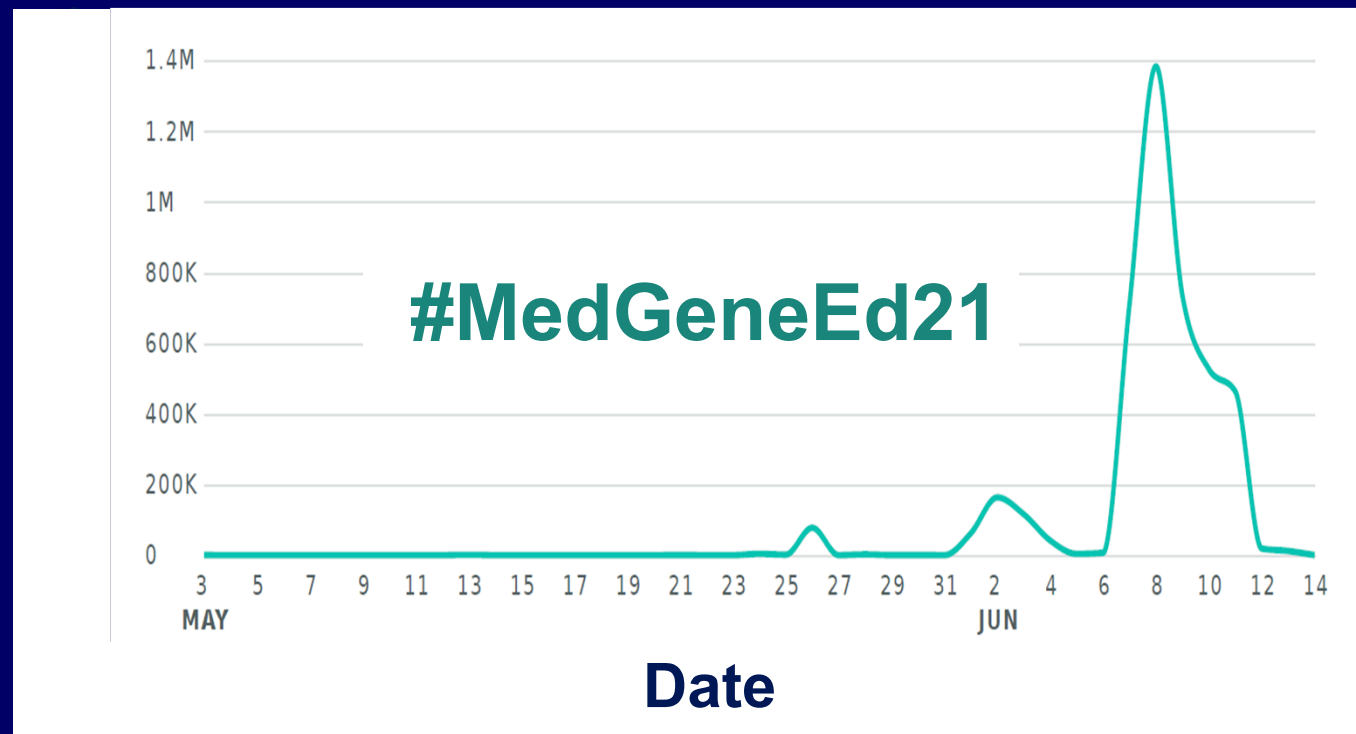


Smithsonian Institution

NATIONAL
MUSEUM *of*
**NATURAL
HISTORY**

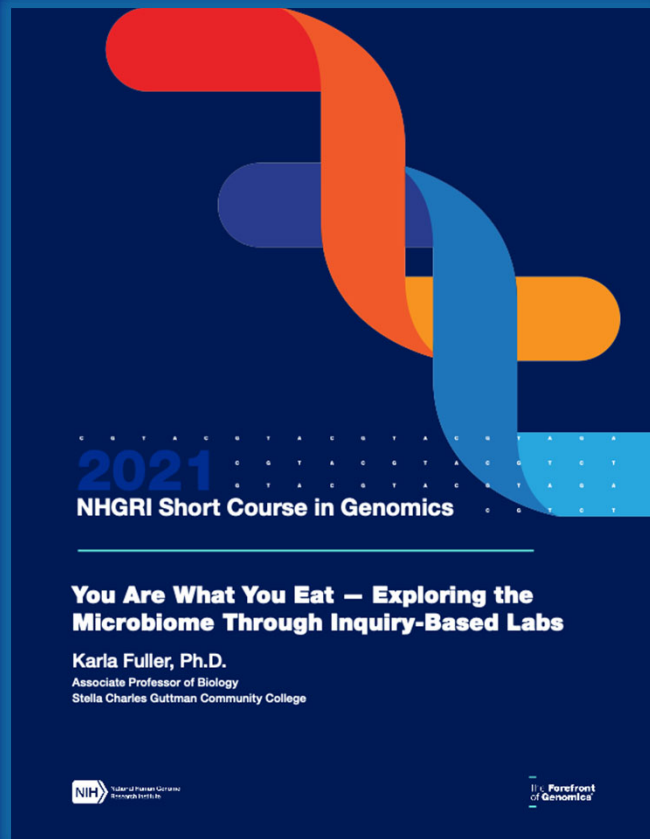
Healthcare Provider Genomics Education Week 2021

Social Media Campaign



2021 Short Course in Genomics

Microbiome Lesson Plans



- **A Glimpse Into the Microbiome: An Introductory Lecture**
- **You Are What You Eat – Exploring the Microbiome Through Inquiry-Based Labs**
- **Microbiome Virtual Lab Exploration!**
- **Exploring the Microbiome and Its Connection to Metabolic Syndrome**

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NHGRI-FDA Genomic Science and Health Equity Postdoctoral Fellowship



Raven Hardy, M.S., Ph.D.

NHGRI Director on Twitter



A screenshot of a Twitter profile card for Eric Green. The background features a glowing blue DNA double helix and a hexagonal grid pattern with faint DNA sequence letters. The profile picture shows a man wearing glasses and a white face mask. The name 'Eric Green' is displayed in bold black text with a blue verified checkmark icon. Below the name is the handle '@NHGRI_Director'. The bio reads '@Genome_gov Director. Genomicist. St. Louis Cardinals fan.' To the right of the profile picture are three circular icons: a three-dot menu, a notification bell with a plus sign, and a blue 'Following' button.

Eric Green ✓
@NHGRI_Director
[@Genome_gov](#) Director. Genomicist. St. Louis Cardinals fan.

⋮ 🔔 Following



The Genomics Landscape



National Human Genome
Research Institute

Thanks!



Special Thanks!



The **Forefront**
of **Genomics**[®]