National Human Genome Research Institute

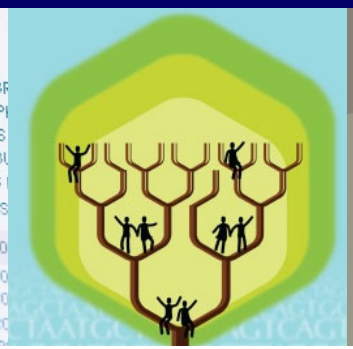
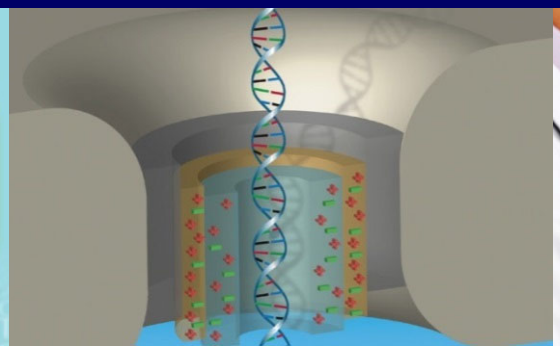
The Forefront of Genomics®

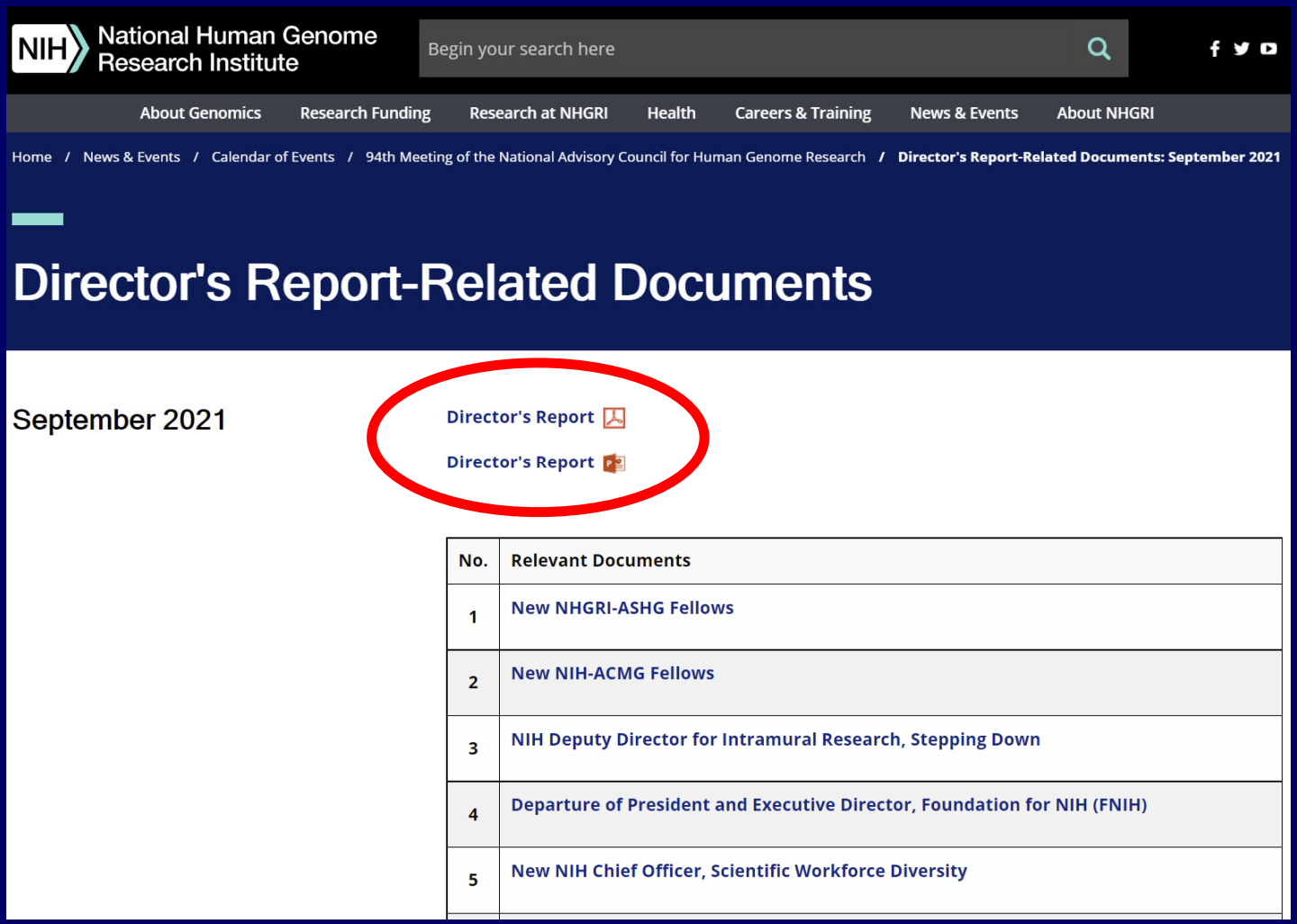
DIRECTOR'S REPORT

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

Director, NHGRI

September 2021



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

Document 1

New NIH-ACMG Fellows

Deepika Burkhardt, D.O.

Nguyen Park, M.S., PA-C

Document 2

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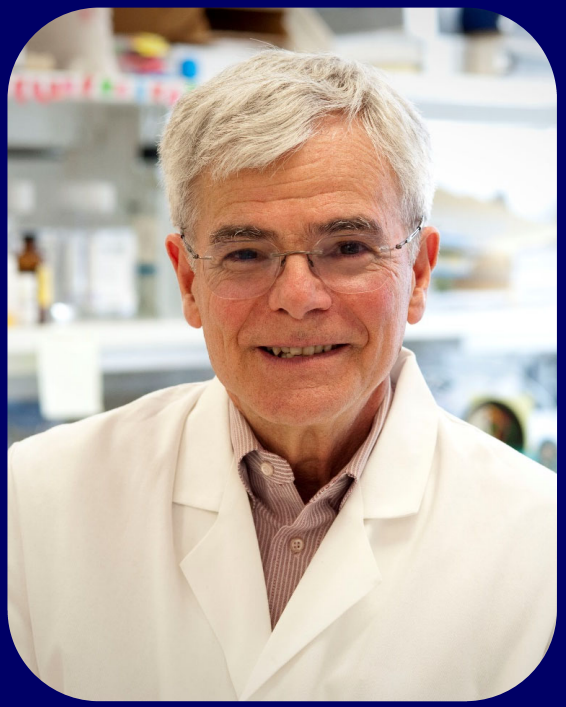
NIH Deputy Director for Intramural Research, Stepping Down



Michael Gottesman, M.D.

NIH Intramural

Document 3

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

Maria Freire, Ph.D.

David Wholley, M. Phil.

Document 4

New NIH Chief Officer, Scientific Workforce Diversity

Marie Bernard, M.D.

National Institutes of Health
Office of the Director
Scientific Workforce Diversity

Document 5

2021-2025 NIH-Wide Strategic Plan

Document 6

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

Document 7

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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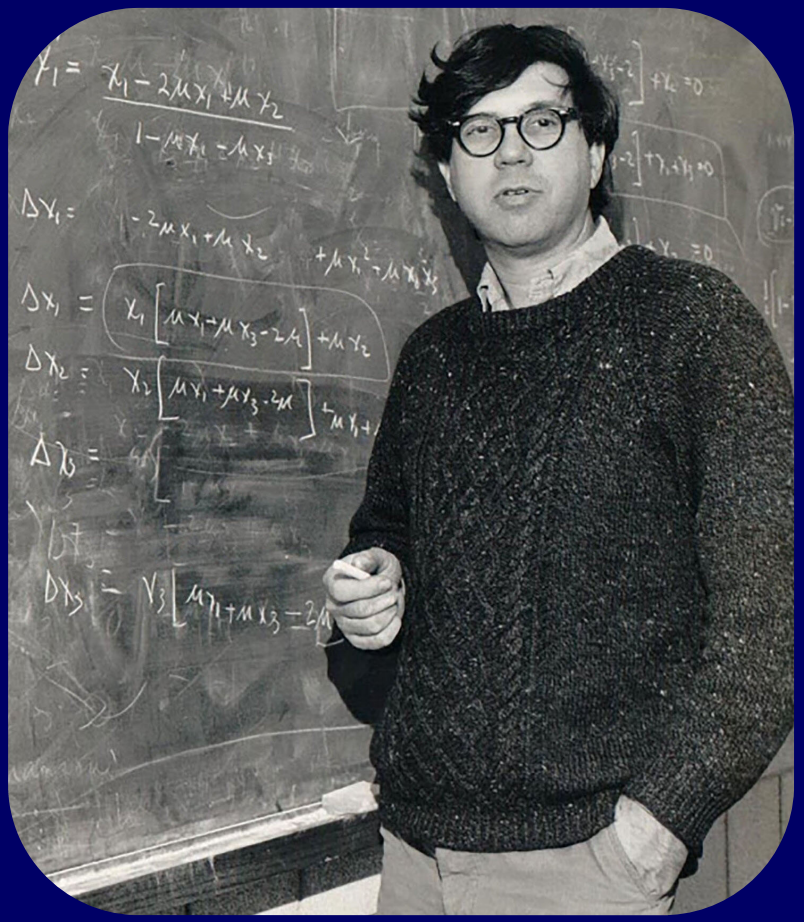
Mourning the Loss of Steve Warren

Document 9

Mourning the Loss of Daniela Gerhard

Document 10

Mourning the Loss of Richard Lewontin



Document 11

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

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

Technology Development Program

Advanced Genomic Technology Development Meeting

May 2021

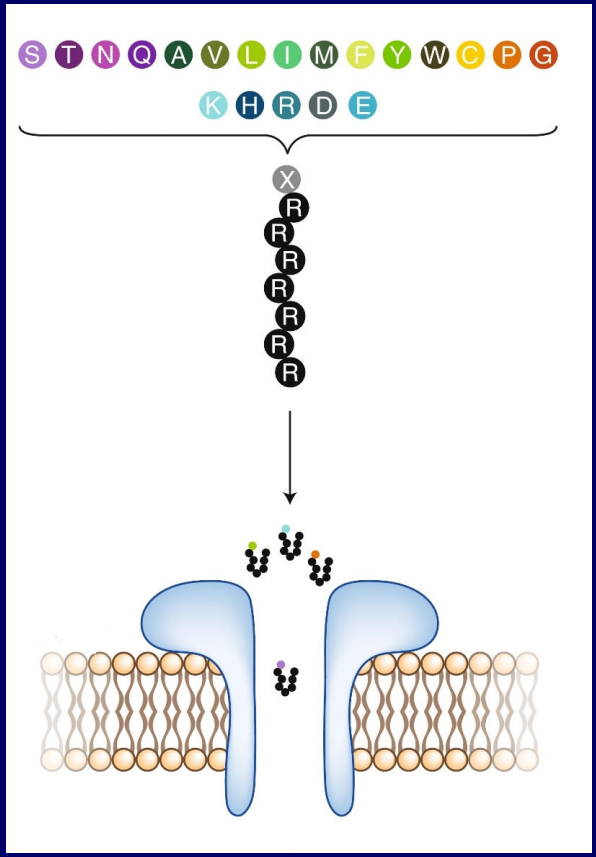
Technology Development Coordinating Center

The Jackson Laboratory

Leading the search for tomorrow's cures

- 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**

Technology Development Program

Funding Opportunities:

- **Novel Synthetic Nucleic Acid Technology Development**
  - RFA-HG-20-014 (R01, also linked R21 and R43/R44)
  - Next due date: February 4, 2022
- **Transformative Nucleic Acid Sequencing Technology Innovation and Early Development**
  - RFA-HG-21-006 (R01, also linked R21 and R43/R44)
  - Next due date: March 1, 2022
- **Advancing Genomic Technology Development for Research and Clinical Application**
  - NOT-HG-21-018, Notice of Special Interest (NOSI)
  - Standard receipt dates

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

Impact of Genomic Variation on Function (IGVF)

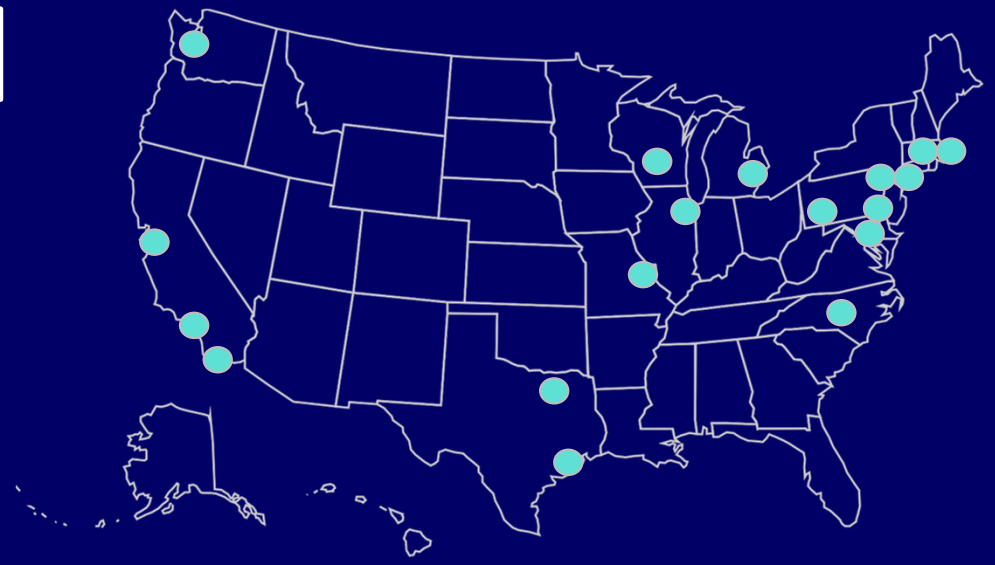
Effects of Genomic Variation

↓

Genome Function

↓

Health & Disease

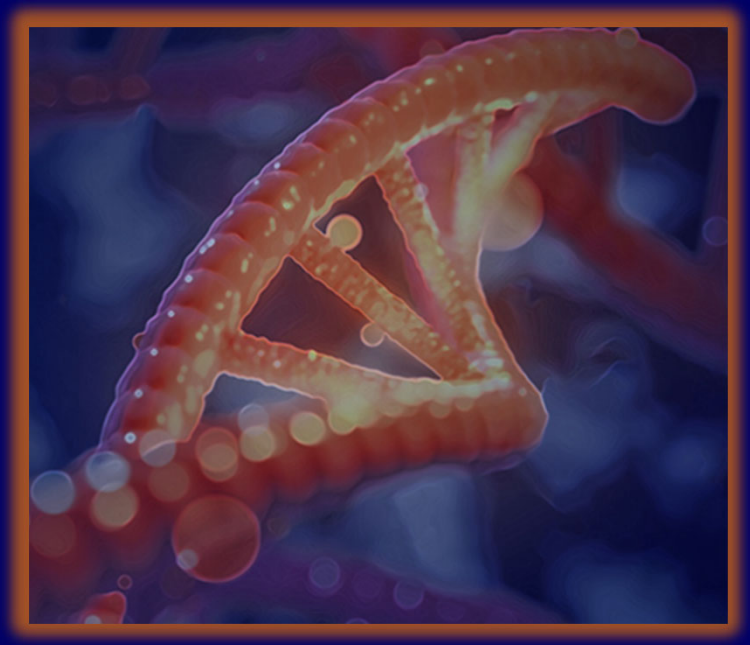


● **Award Locations in U.S.**

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

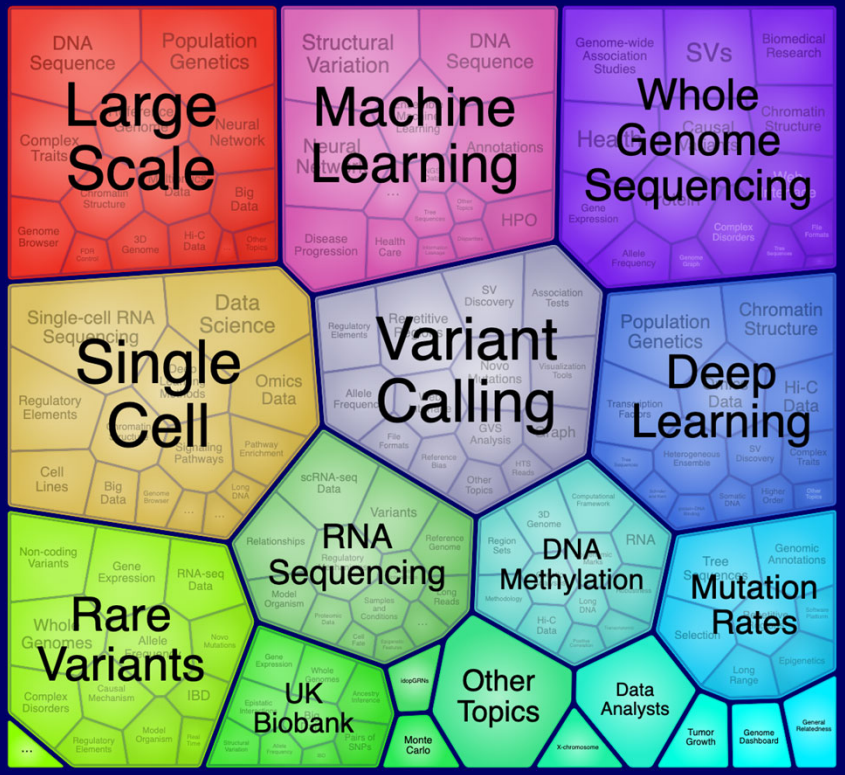
Centers for Excellence in Genomic Science (CEGS)

- **Chao-Ting Wu (Harvard Medical School)**
  - ***Center for Genome Imaging***

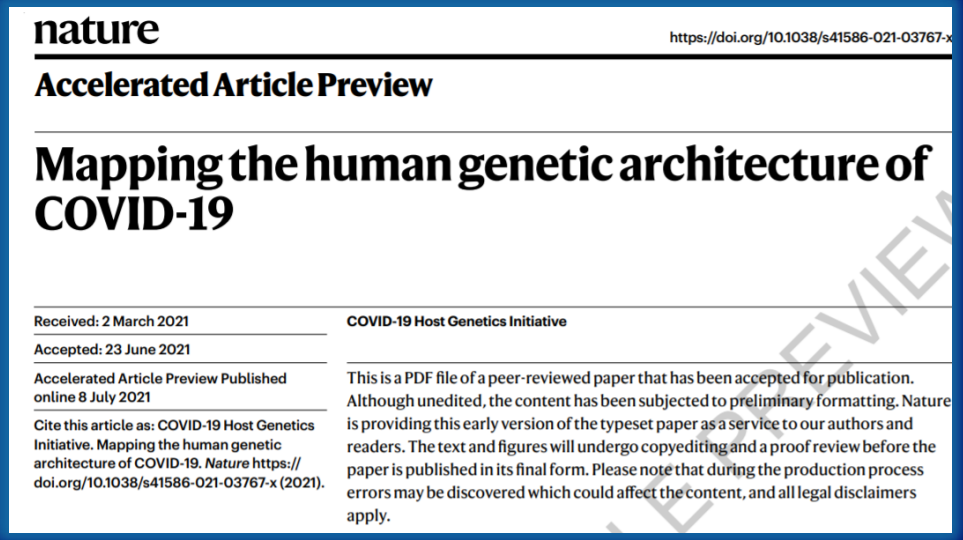
- **Chuan He (University of Chicago)**
  - ***Center for Dynamic RNA Epitranscriptomes***

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

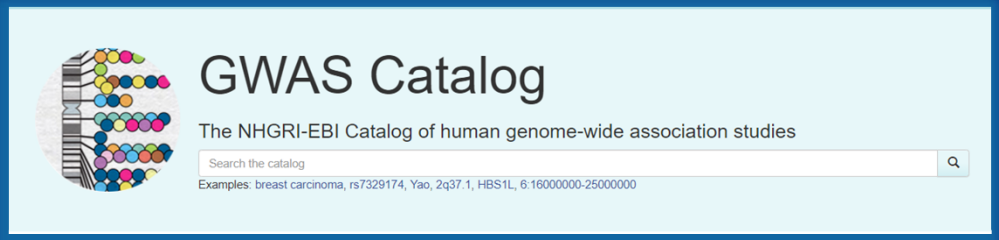
Accepted: 23 June 2021

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).

COVID-19 Host Genetics Initiative

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.



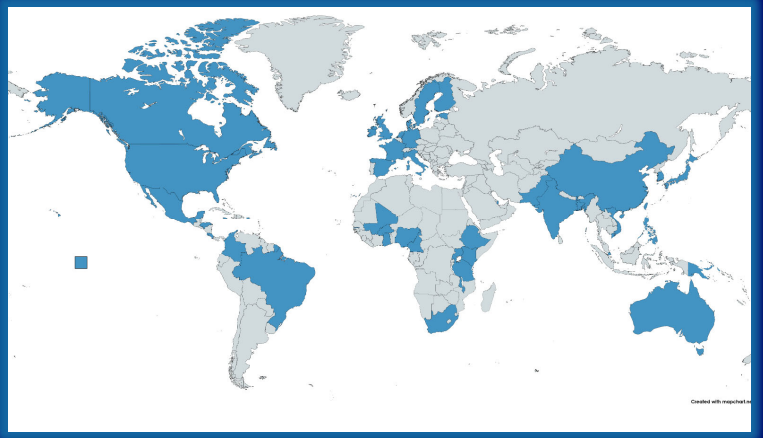
As of August 16, 2021, the GWAS Catalog contains 5,273 publications and 276,696 associations.

- **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)

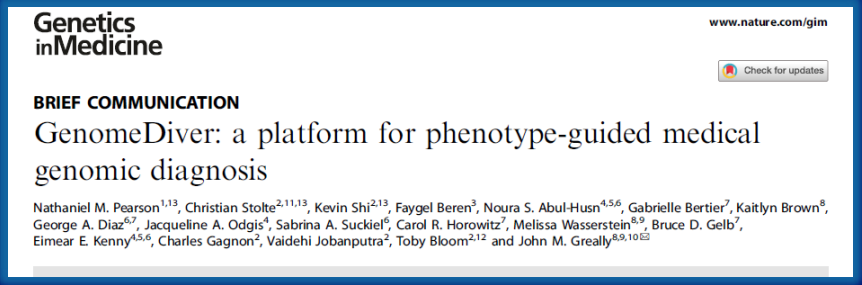
Virtual Retreat 2021

~ 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

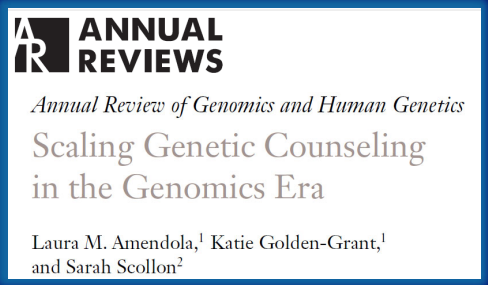
www.nature.com/gim

Check for updates

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 Beren[3], Noura S. Abul-Husn[4,5,6], Gabrielle Bertier[7], Kaitlyn Brown[8], George A. Diaz[6,7], Jacqueline A. Odgis[4], Sabrina A. Suckiel[6], Carol R. Horowitz[7], Melissa Wasserstein[8,9], Bruce D. Gelb[7], Eimear E. Kenny[4,5,6], Charles Gagnon[2], Vaidehi Jobanputra[2], 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 Scollon[2]


ORIGINAL ARTICLE

CLINICAL GENETICS

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

Katherine E. Donohue[1] | Catherine Gooch[2,3] | Alexander Katz[4] | Jessica Wakelee[5] | Anne Slavotinek[6] | Bruce R. Korf[2]

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**

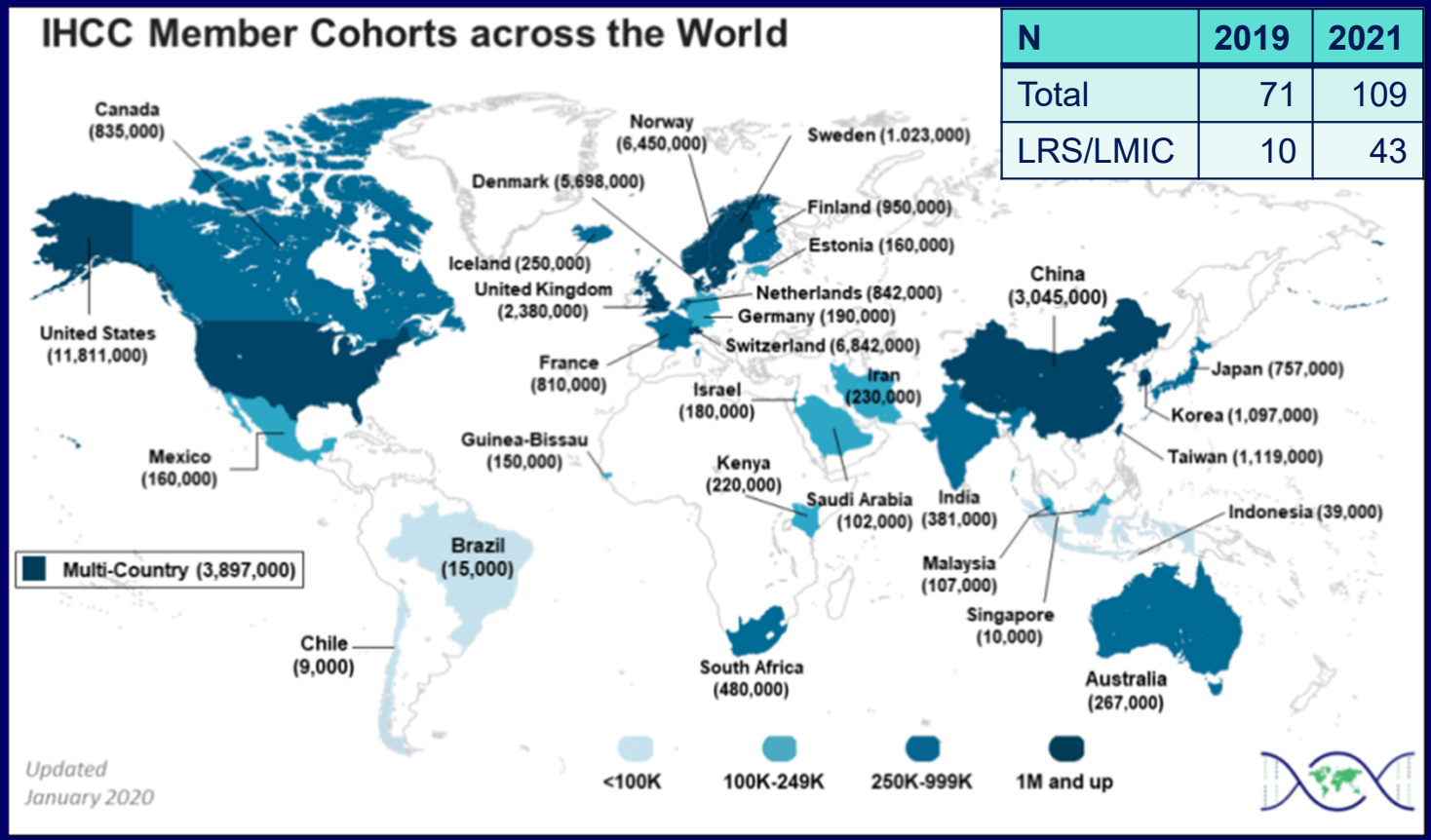
Multi-omics in Health and Disease Workshop

Document 23

International 100K+ Cohort Consortium (IHCC)

4th International Cohorts Summit

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



N	2019	2021
Total	71	109
LRS/LMIC	10	43

Document 24

The 5th ELSI Congress

Save the Date

June 1-3, 2022

Columbia University

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

Document 25

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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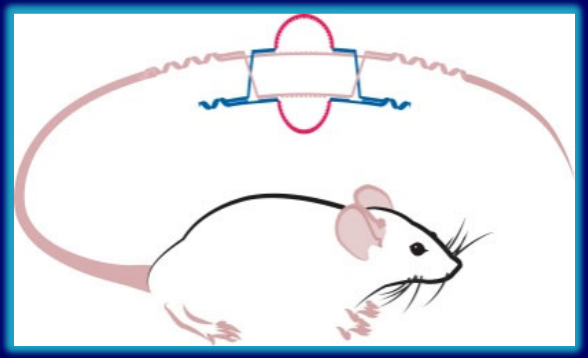
Knockout Mouse Phenotyping Project (KOMP2)



IMPC
International Mouse Phenotyping Consortium

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

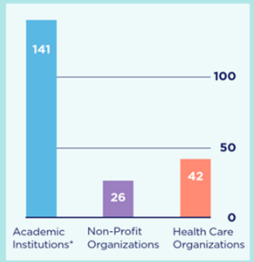
Top conditions being studied
in the Researcher Workbench include:
- Cardiovascular disease
- Hypertension
- Cancer
- Mental health
- Diabetes

Our Researchers

826
Registered Researchers
across a range of institutional roles and career stages.

209
Institutions

Researchers **nationwide** are using the **Researcher Workbench**



*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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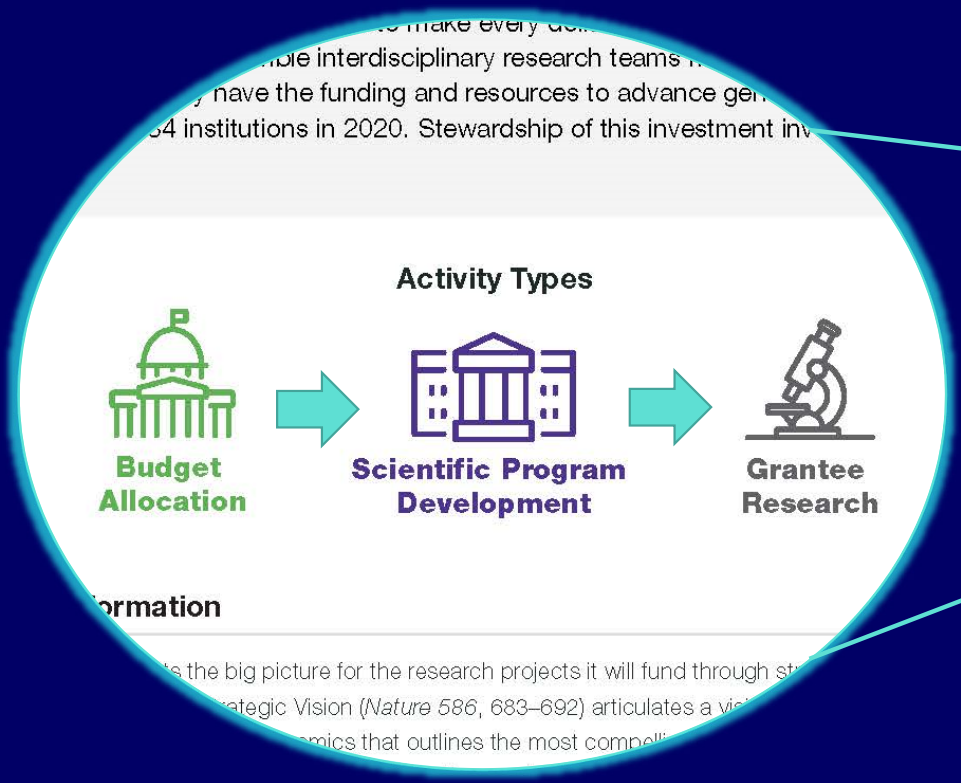
New Resource on NHGRI Grant Funding Process



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 Correspondant (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

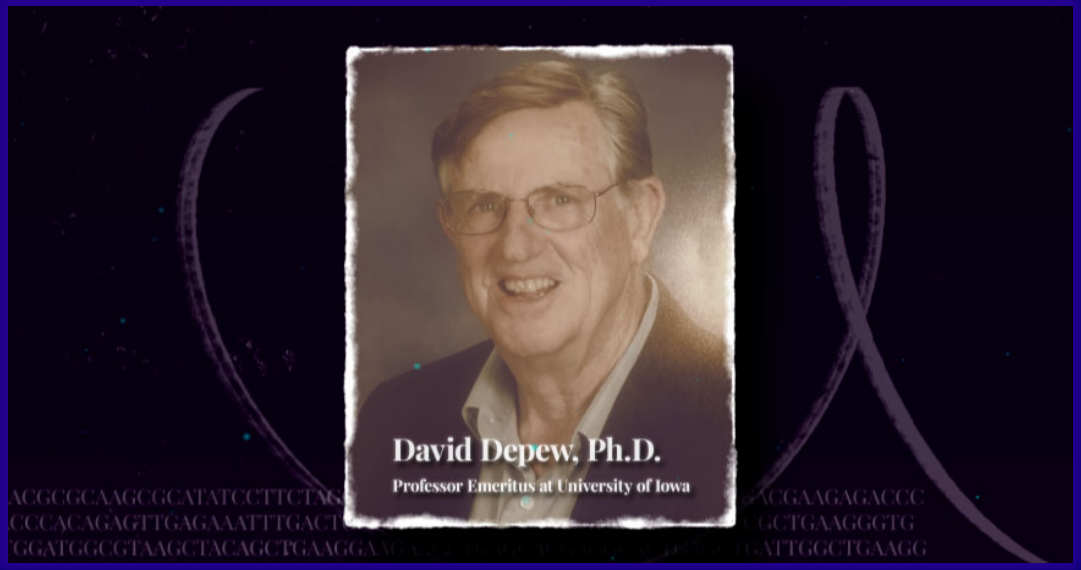
Conversations with trailblazing science communicators

Infographic on Completing the Human Genome Sequence

Document 32

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

UNITED STATES HOLOCAUST MEMORIAL MUSEUM

Smithsonian Institution

NATIONAL MUSEUM of NATURAL HISTORY

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

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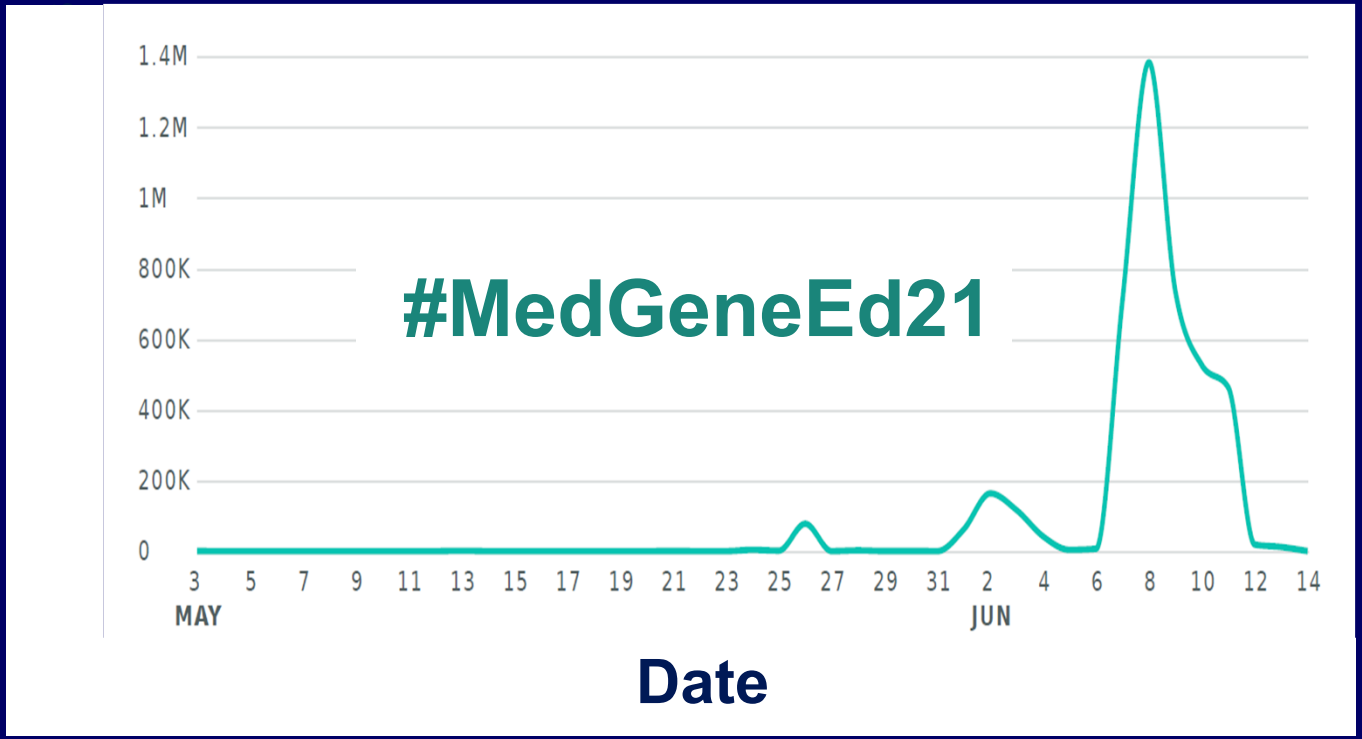
Healthcare Provider Genomics Education Week 2021

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.

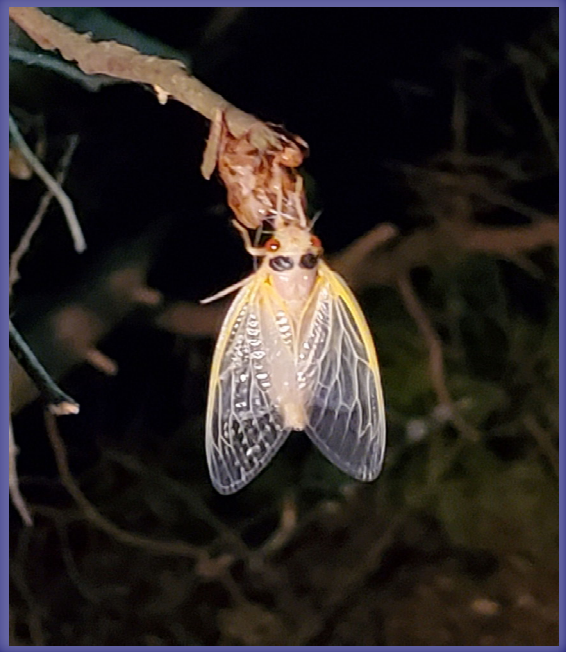
NHGRI Director on Twitter

Document 37

The Genomics Landscape

National Human Genome Research Institute

Thanks!



Special Thanks!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