

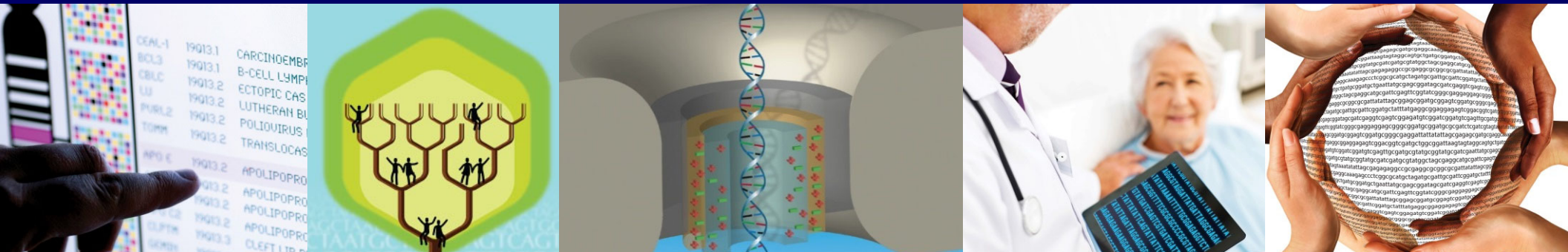


—  
The **Forefront**  
of **Genomics**  
—

# DIRECTOR'S REPORT

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

**February 2024**



# 100<sup>th</sup> NACHGR Meeting





# 101<sup>st</sup> NACHGR Meeting



## Director's Report-Related Documents

February 2024

Director's Report 

Director's Report 

No.	Relevant Documents
1	Finally!!!: The 17th NIH Director
2	New Director, National Cancer Institute
3	New Director, NIH Division of Program Coordination, Planning, and Strategic Initiatives
4	New Director, NIH Office of Nutrition Research
5	New Director, NIH Office of Science Policy
6	New Director, NIH Office of Legislative Policy and Analysis

[genome.gov/DirectorsReport](https://genome.gov/DirectorsReport)

Document # 



# Open Session Agenda

## Presentation:

**Research at the Intersection of Translational Science and Genomics**

**Joni Rutter**

## Concept Clearances:

**Building Partnerships and Broadening Perspectives to Advance Ethical, Legal, and Social Implications (ELSI) Research**

**Rene Sterling**

**NHGRI Predoctoral to Postdoctoral Transition Award for Ethical, Legal and Social Implications (ELSI) Research (F99/K00)**

**Rene Sterling**

# Open Session Agenda

## Concept Clearances:

Population Screening in Primary Care

**Simona Volpi**

ELSI Congress Research Conference

**Nicole Lockhart**

Enhancing Reuse of NHGRI Data Assets (ERND)

**Shurjo Sen**

Investigator-Initiated Research in Computational Genomics and  
Data Science

**Dan Gilchrist**

# Open Session Agenda

## Presentations:

**Evaluation of the Centers of Excellence in Genomic Science  
(CEGS) Program**

**Chris Gunter**

**Advancing Genomic Medicine through Genome-Informed Risk  
Assessment – eMERGE Network Update**

**Eimear Kenny**



# Open Session Agenda

## Reports:

**Genomic Data Science Working Group of Council Annual Report**  
**Shannon McWeeney & Anshul Kundaje**

**Genomic Medicine Working Group of Council Annual Report**  
**Teri Manolio**

# 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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# **New Extramural Program Director**



**Alanna Kulchak Rahm, Ph.D., M.S., C.G.C.**

# New Extramural Program Director



**Sheethal Jose, Ph.D.**

# New Scientific Review Officer



**Lori Bonnycastle, Ph.D.**



# New Data and Technology Advancement (DATA) National Service Scholar



**Arthur Ko, Ph.D.**

# Terry McAuliffe Visits NHGRI



# Director's Report Outline

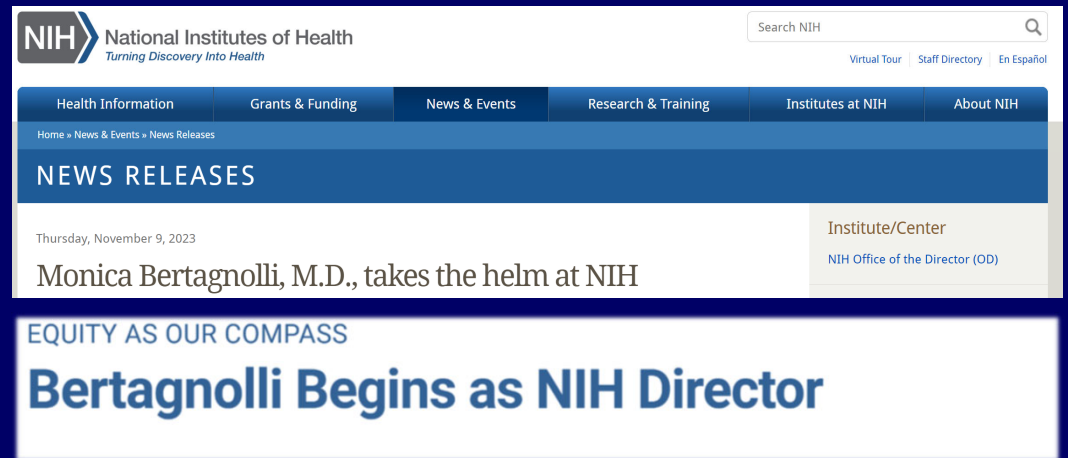
- I. General NHGRI Updates
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- III. General Genomics Updates
- IV. NHGRI Extramural Research Program
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- VI. NHGRI Communications, Policy, and Education
- VII. NHGRI Intramural Research Program



# Finally!!!: The 17<sup>th</sup> NIH Director



**Monica Bertagnolli, M.D.**



The screenshot shows the NIH website's news release page. At the top, the NIH logo and tagline "National Institutes of Health Turning Discovery Into Health" are visible. A search bar and navigation links for "Virtual Tour", "Staff Directory", and "En Español" are in the top right. A menu bar includes "Health Information", "Grants & Funding", "News & Events", "Research & Training", "Institutes at NIH", and "About NIH". Below the menu, the breadcrumb "Home » News & Events » News Releases" is shown. The main heading is "NEWS RELEASES". The date "Thursday, November 9, 2023" is on the left, and "Institute/Center NIH Office of the Director (OD)" is on the right. The main title of the release is "Bertagnolli Begins as NIH Director" with the subtitle "EQUITY AS OUR COMPASS".



# New Director, National Cancer Institute



**W. Kimryn Rathmell,  
M.D., Ph.D.**

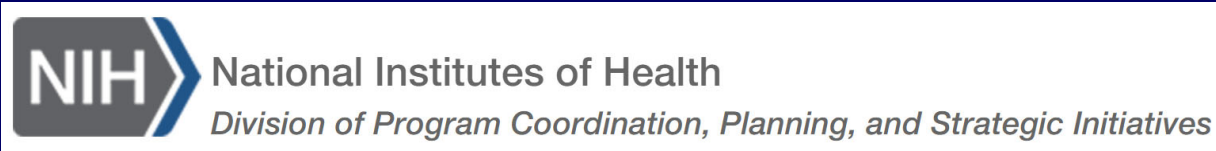


National Cancer Institute

# New Director, NIH Division of Program Coordination, Planning, and Strategic Initiatives



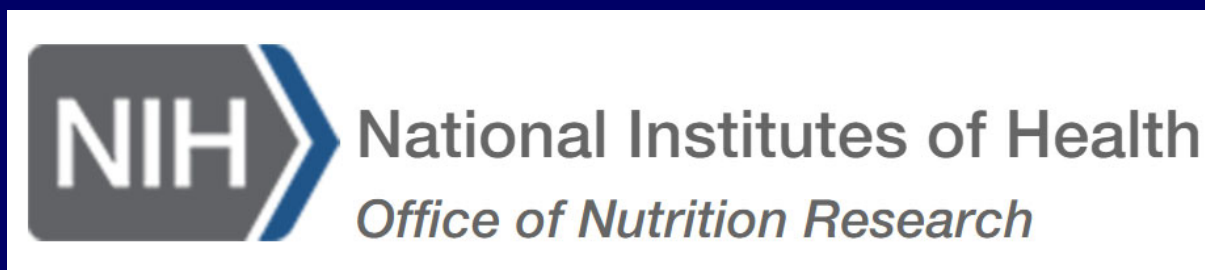
**Tara Schwetz, Ph.D.**



# New Director, NIH Office of Nutrition Research



**Andrew Bremer,  
M.D., Ph.D., M.A.S.**



# New Director, NIH Office of Science Policy



**Lyric Jorgenson, Ph.D.**



**National Institutes of Health**  
*Office of Science Policy*



# New Director, NIH Office of Legislative Policy and Analysis



**Kate Klimczak**



**National Institutes of Health**  
*Turning Discovery Into Health*



# New Director, NIH Center for Information Technology (CIT)

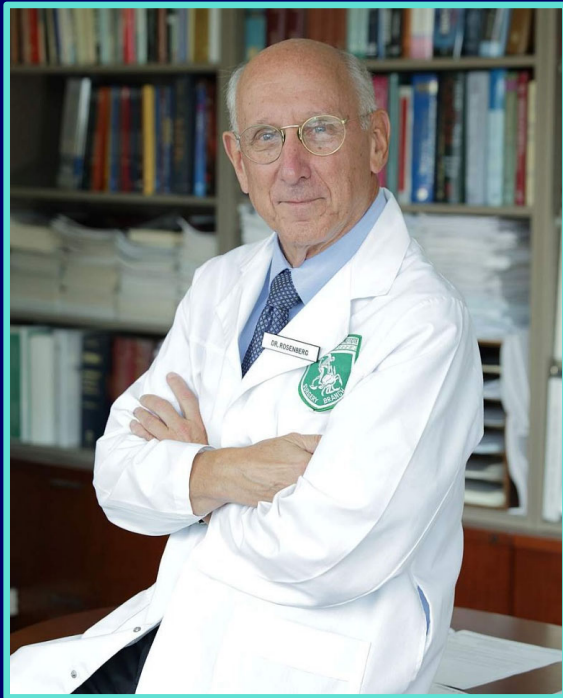


**Sean Mooney, Ph.D.**



*Breaking News*

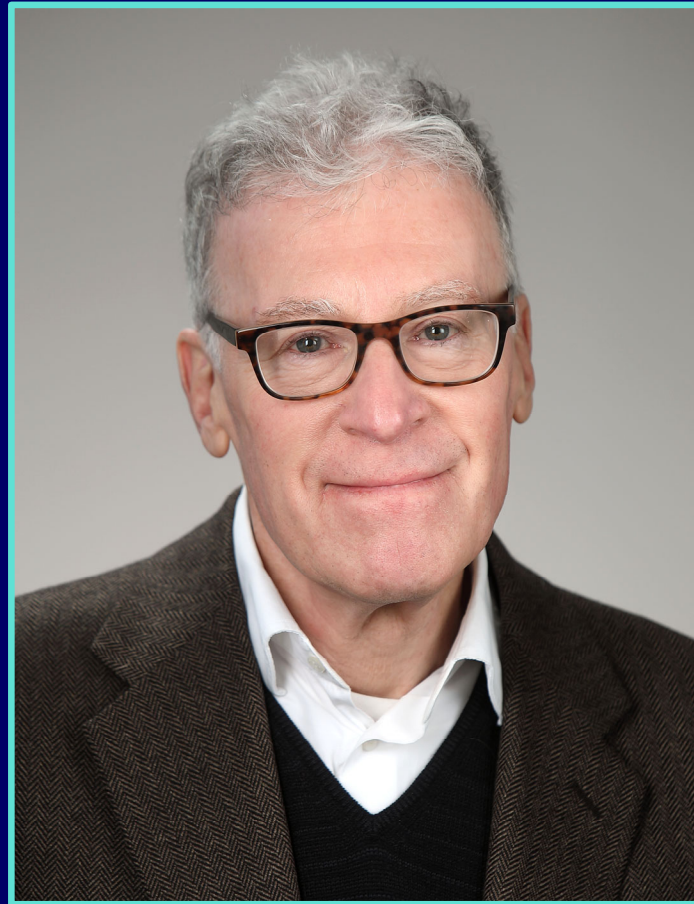
# National Medal of Technology and Innovation



**Steven Rosenberg,  
M.D., Ph.D.**



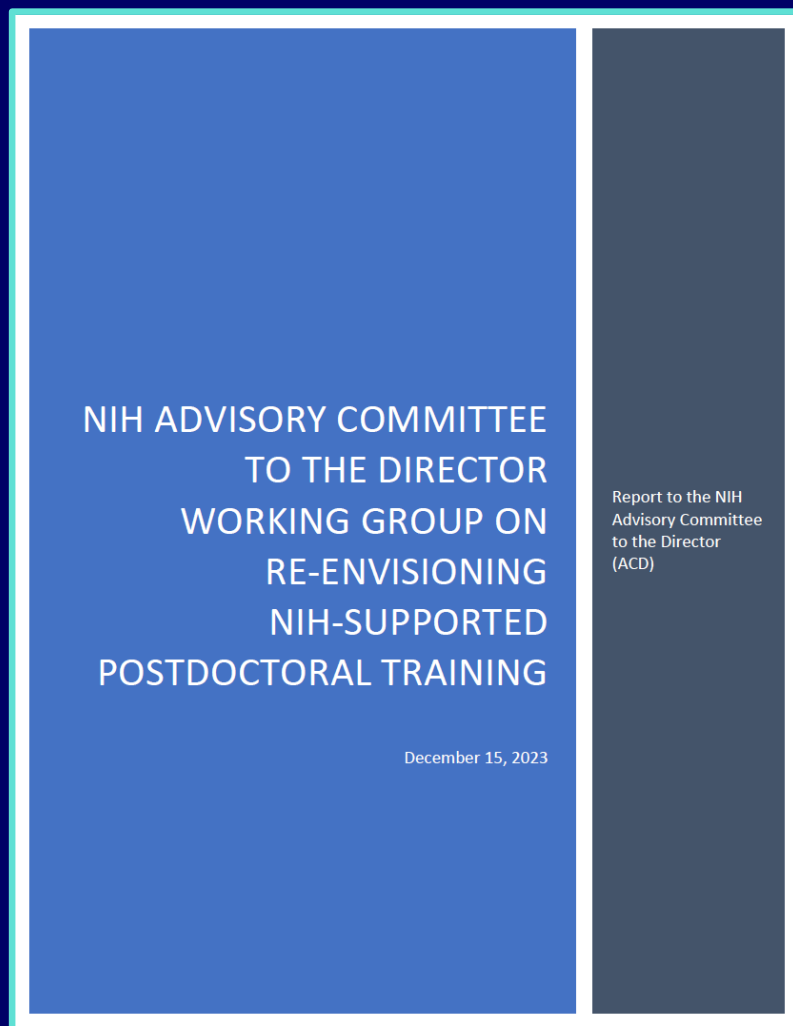
# Mourning the Loss of Robert Eiss



# NIH Designates People with Disabilities as a Population with Health Disparities

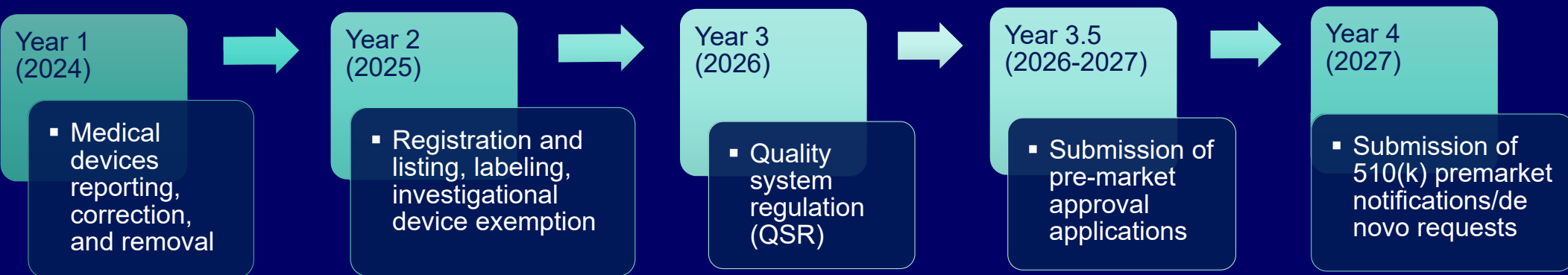


# Re-Envisioning NIH-Supported Postdoctoral Training



# FDA Proposed Rule for LDT Oversight and Regulation

- Re-classifies LDTs as medical devices
- 5-year phase-out of enforcement discretion policy
- Implementation by April 2024
- Phase-out timeline:



- Likely legal challenges



# Fiscal Year 2024 Appropriations

- **Currently operating under a Continuing Resolution**

**Expires March 8, 2024**

- **Agreement from Congressional leadership:**

**Discretionary spending (includes NIH): \$772.7 billion**

**Chambers must come to agreement on final budget breakdown**

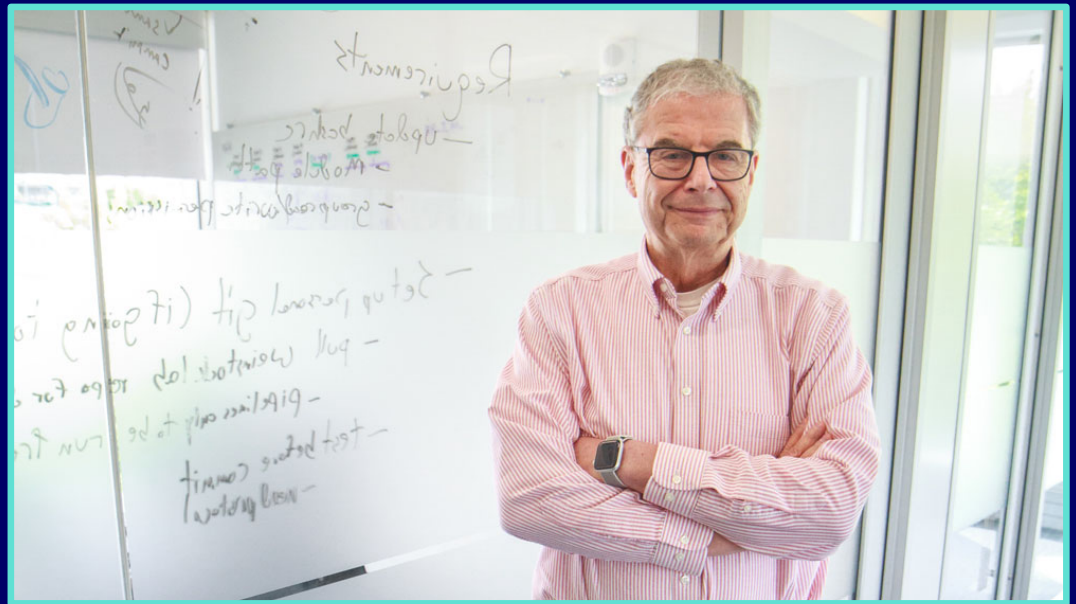
# Fiscal Year 2024 Draft Appropriations from July 2023

Chamber	Portion	Fiscal Year 2023 Enacted L-HHS Appropriation	Fiscal Year 2024 Draft L-HHS Appropriation	Fiscal Year 2024-2023 Difference \$	Fiscal Year 2024-2023 Difference %
<b>House</b> <small>(\$644.5 B top-line discretionary)</small>	NIH	\$46.8 B	\$44 B	-\$2.8 B	-6%
	<b>NHGRI</b>	\$660.51 M	\$663.2 M	\$2.69 M	+0.4%
<b>Senate</b> <small>(\$703.7 B top-line discretionary)</small>	NIH	\$46.8 B	\$47.8 B	\$943 M	+2%
	<b>NHGRI</b>	\$660.51 M	\$663.2 M	\$2.69 M	+0.4%

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# Mourning the Loss of George Weinstock



# Mourning the Loss of Eta Berner



# Mourning the Loss of Sandra Day O'Connor





# 2023 American Society of Human Genetics Awards

2023 ASHG Lifetime Achievement Award

**Neil Risch, PhD**

University of California, San Francisco



2023 ASHG Early-Career Award

**Jennifer Posey, MD, PhD**

Baylor College of Medicine



2023 ASHG Leadership Award

**Nancy Cox, PhD**

Vanderbilt University Medical Center



2023 ASHG Early-Career Award

**Karen Miga, PhD**

University of California, Santa Cruz



2023 ASHG Scientific Achievement Award

**Molly Przeworski, PhD**

Columbia University



# 2023 MacArthur Fellow



**Jason Buenrostro,  
Ph.D.**



# Elected to National Academy of Medicine

**Bradley Bernstein**

**John Carpten**

**Michael Chiang**

**Daniele Fallin**

**Guoping Feng**

**Eric Green**

**Christian Happi**

**Jeannie Lee**

**Siddhartha Mukherjee**

**Rudolph Tanzi**

**Phillip Zamore**



**NATIONAL ACADEMY OF MEDICINE**

## Elected to AAAS

**Emmanuelle Charpentier**

**Franklin Costantini**

**Laurence Fishburne**

**W. Kimryn Rathmell**

**Beth Shapiro**

**Barbara Wold**

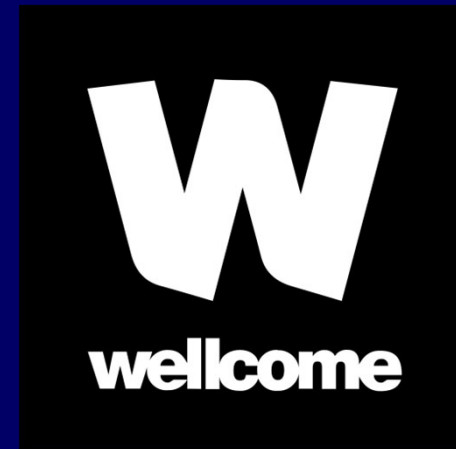
**Phillip Zamore**



# New Chief Executive Officer, Wellcome



**John-Arne Røttingen, Ph.D.**



# Global Alliance for Genomics & Health (GA4GH)

- Heidi Rehm new Chair of the GA4GH Board of Directors
- Agreement to support responsible sharing of genomic and related health data on a global scale through developments in technology standards, tools, and policy frameworks



**Heidi Rehm,  
Ph.D.**





# Global Alliance for Genomics & Health (GA4GH)



**2023 GA4GH Driver Project**

# Together for CHANGE



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- VII. NHGRI Intramural Research Program

# Genomics Research to Elucidate the Genetics of Rare Diseases (GREGoR)



- **GREGoR v1 Dataset released September 2023**
- **First “joint call set” will be released in 2024**
- ***Beyond the Exome: What’s Next in Diagnostic Testing for Mendelian Conditions* published in *AJHG* in August 2023**

# Impact of Genomic Variation on Function (IGVF)

The screenshot shows the IGVF website home page. The navigation bar includes links for Home, About, Overview, Affiliate Membership, Publications, Outreach, and Wiki. A 'Data Portal' button is also visible. The main heading reads 'Welcome to the IGVF Consortium!'. Below this, a paragraph describes the consortium's mission: 'The IGVF (Impact of Genomic Variation on Function) Consortium aims to understand how genomic variation affects genome function, which in turn impacts phenotype. The NHGRI is funding this collaborative program that brings together teams of investigators who will use state-of-the-art experimental and computational approaches to model, predict, characterize and map genome function, how genome function shapes phenotype, and how these processes are affected by genomic variation. These joint efforts will produce a catalog of the impact of genomic variants on genome function and phenotypes.' A video thumbnail is shown with the title 'MISSING Function of Variant X' and a play button icon. A caption below the video states: 'This video can be found on the NHGRI website.'

The screenshot shows a bioRxiv article page. The bioRxiv logo and 'THE PREPRINT SERVER FOR BIOLOGY' are at the top. The article title is 'Transcription factor stoichiometry, motif affinity and syntax regulate single-cell chromatin dynamics during fibroblast reprogramming to pluripotency'. The authors listed are Nair, S., Ameen, M., Sundaram, L., Pampari, A., Schreiber, J., Balsubramani, A., Wang, Y. X., Burns, D., Blau, H. M., Karakikes, I., Wang, K. C., and Kundaje, A. The article ID is 10.1101/2023.10.04.560808 and it was posted on 2023-10-21. Below this, another article is partially visible: 'A machine-readable specification for genomics assays' by Boeshaghi, A. S., Chen, X., and Pachter, L., with article ID 10.1101/2023.03.17.533215, posted on 2023-07-18.

The screenshot shows a 'My NCBI Collection - IGVF Publications' page. It displays 71 results. The first result is 'Demographic and Viral-Genetic Analyses of COVID-19 Severity in Bahrain Identify Local Risk Factors and a Protective Effect of Polymerase Mutations.' by Koch EM, Du J, Dressner M, Alwasti HE, Al Taif Z, Shehab F, Mohamed AM, Ghanem A, Alhajeri A, Alawadhi A, Almoamen N, Ashoor K, Hasan S, Haghighi A, Sunyaev S, Farhat M. The preprint is dated Oct 12, 2022, with medRxiv ID 2022.08.13.22278740 and doi: 10.1101/2022.08.13.22278740. The PMID is 36032980 and it is a free PMC article. The second result is 'Powerful, scalable and resource-efficient meta-analysis of rare variant associations in large whole genome sequencing studies.' by Li X, Quick C, Zhou H, Gaynor SM, Liu Y, Chen H, Selvaraj MS, Sun R, Dey R, Arnett DK, Bielak LF, Bis JC, Blangero J, Boerwinkle E, Bowden DW, Brody JA, Cade BE, Correa A, Cupples LA, Curran JE, de Vries PS, Duggirala R, Freedman BI, Göring HHH, Guo X, Haessler J, Kalyani RR, Kooperberg C, Kraljic L, Lange LA, Manichaikul A, Martin LW, McGarvey ST, Mitchell BD, Montasser ME, Morrison

The screenshot shows an arXiv article page. The arXiv logo and 'q-bio > arXiv:2307.13708' are at the top. The article is categorized under 'Quantitative Biology > Other Quantitative Biology'. It was submitted on 24 Jul 2023. The article title is 'The Impact of Genomic Variation on Function (IGVF) Consortium' and the author is IGVF Consortium.

# Centers of Excellence in Genomic Science (CEGS)

Article | [Open access](#) | Published: 01 November 2023

## Mouse genome rewriting and tailoring of three important disease loci

[Weimin Zhang](#), [Ilona Golynker](#), [Ran Brosh](#), [Alvaro Fajardo](#), [Yinan Zhu](#), [Aleksandra M. Wudzinska](#), [Raquel](#)

[Ordoñez](#), [André M. Ribeiro](#)

[Khairallah](#), [Antonio Vela-G](#)

[Matthew T. Maurano](#), [Sang](#)

*Nature* **623**, 423–431 (2023)

## The Center for Synthetic Regulatory Genomics (SyRGE)

Article | [Open access](#) | Published: 09 November 2023

## Transcriptional and epigenetic regulators of human CD8<sup>+</sup> T cell function identified through orthogonal CRISPR screens

[Sean R. McCutcheon](#)

[Keith Siklenka](#), [Luca](#)

[Smita K. Nair](#), [Scott](#)

*Nature Genetics* **55**

RESOURCE | VOLUME 30, ISSUE 9, P1262–1281 E8, SEPTEMBER 07, 2023 [Download Full Issue](#)

Single-cell multi-omics defines the cell-type-specific impact of splicing aberrations in human hematopoietic clonal outgrowths

[Mariela Cortés-López](#)<sup>19</sup> • [Paulina Chamelv](#)<sup>19</sup> • [Allegra G. Hawkins](#)<sup>19</sup> • [Omar Abdel-Wahab](#) •

[Federico Gaiti](#)<sup>20</sup> • [Dan A. Lan](#)

Published: August 14, 2023 • DOI: [10.1016/j.cel.2023.08.011](#)

Article | Published: 02 January 2024

## Programmable protein expression using a genetically encoded m<sup>6</sup>A sensor

[Bahjat F. Marayati](#), [Matthew G. Thompson](#), [Chuan He](#)

*Nature Biotechnology* (2024) | [Cite this article](#)

## The Duke Center for Combinatorial Gene Regulation

## The Center for Integrated Cellular Analysis

## The Center for Dynamic RNA Epitranscriptomes

## The Center for Genomic Information Encoded by RNA Nucleotide Modifications

Article | [Open access](#) | Published: 02 January 2024

## Ultrafast bisulfite sequencing detection of 5-methylcytosine in DNA and RNA

[Qing Dai](#) ✉, [Chang Ye](#), [Iryna Irklyienko](#), [Yiding Wang](#), [Hui-Lung Sun](#), [Yun Gao](#), [Yushuai Liu](#), [Alana Beadell](#), [José Perea](#), [Ajay Goel](#) & [Chuan He](#) ✉

*Nature Biotechnology* (2024) | [Cite this article](#)



# Cross-Consortia Day 2023



- Washington University School of Medicine
- IGVF, HPRC, GREGoR, ClinGen, & AVE

# Genetic Architecture of Complex Traits Workshop

- **Workshop held November 2023**
  - >60 participants and >900 webinar registrations
  - Recordings available
  - Meeting report available



- **Developing Novel Theory and Methods for Understanding the Genetic Architecture of Complex Human Traits (R01/R21)**
  - PAR-23-302 and PAR-23-301
  - Standard receipt dates

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

AnVIL Data Explorer

Help & Documentation

Filters Clear All

Search all filters...

Dataset (160)

Identifier (37)

Anatomical Site (30)

BioSample Type (6)

Consent Group (55)

Data Modality (1)

Diagnosis (862)

File Format (85)

Organism Type (2)

Explore Data 10.2k BioSamples • 318.2k Files Export

Datasets Donors BioSamples Activities Files

Results 1 - 25 of 160

Dataset ↑	Access	Consent Group
ANVIL_1000G_high_coverage_2019	Granted	NRES
ANVIL_African_American_Seq_HGV	Required	HGV

# DUOS

## Data Use Oversight System

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

- AnVIL Data Explorer launch
- Data Use Oversight System (DUOS) enabled

# NHGRI Genomic Data Science Working Group


## Cell Genomics

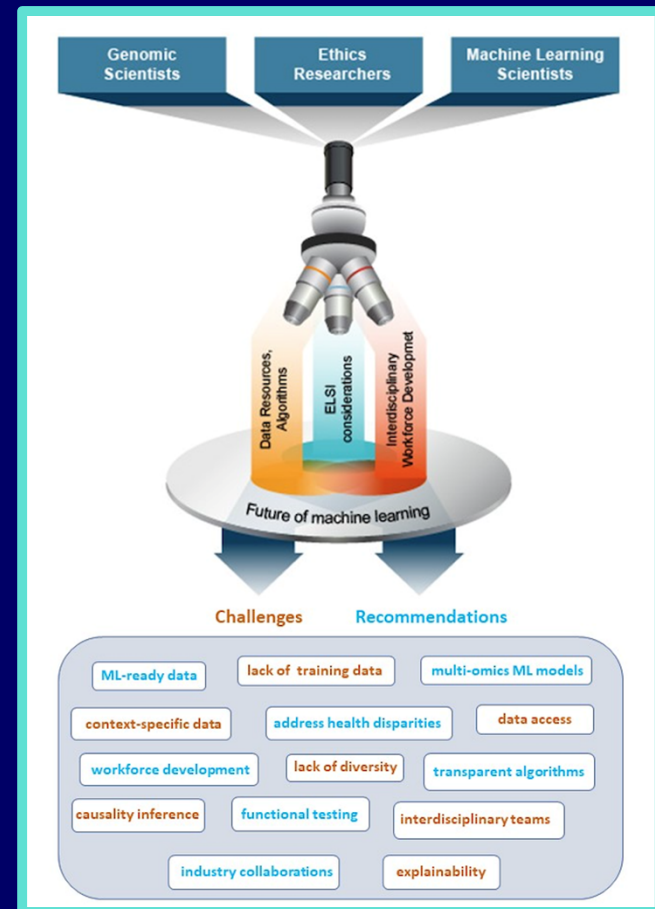


Volume 4, Issue 1, 10 January 2024, 100466

Perspective

### Opportunities for basic, clinical, and bioethics research at the intersection of machine learning and genomics

Shurjo K. Sen<sup>1</sup>  , Eric D. Green<sup>1</sup>, Carolyn M. Hutter<sup>1</sup>, Mark Craven<sup>2,3</sup>, Trey Ideker<sup>4</sup>,  
Valentina Di Francesco<sup>1</sup>  



## Genome-informed risk assessment (GIRA)

- Polygenic risk
- Monogenic risk
- Family health history
- Clinical covariates



Atrial fibrillation  
Hypercholesterolemia  
Coronary heart disease



Type 1 and 2 diabetes



Chronic kidney disease



Obesity



Breast cancer  
Prostate cancer

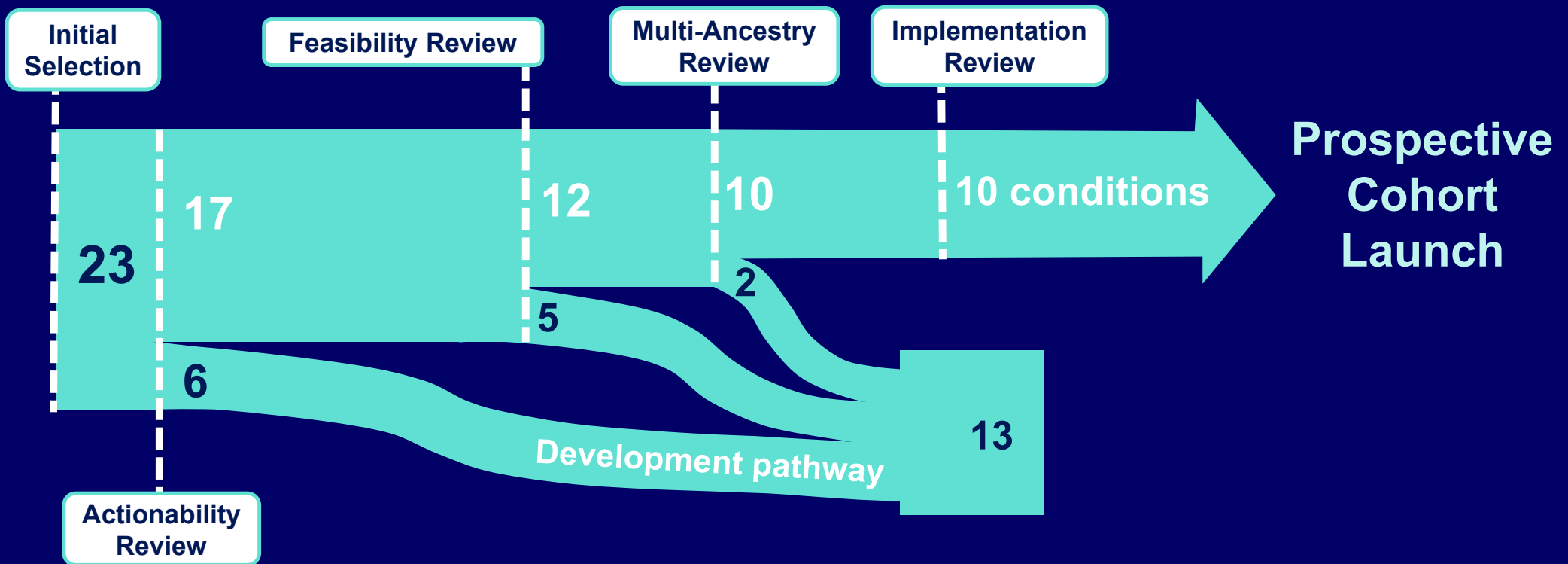


Asthma

**Selection, optimization, and validation of ten chronic disease polygenic risk scores for clinical implementation in diverse populations**



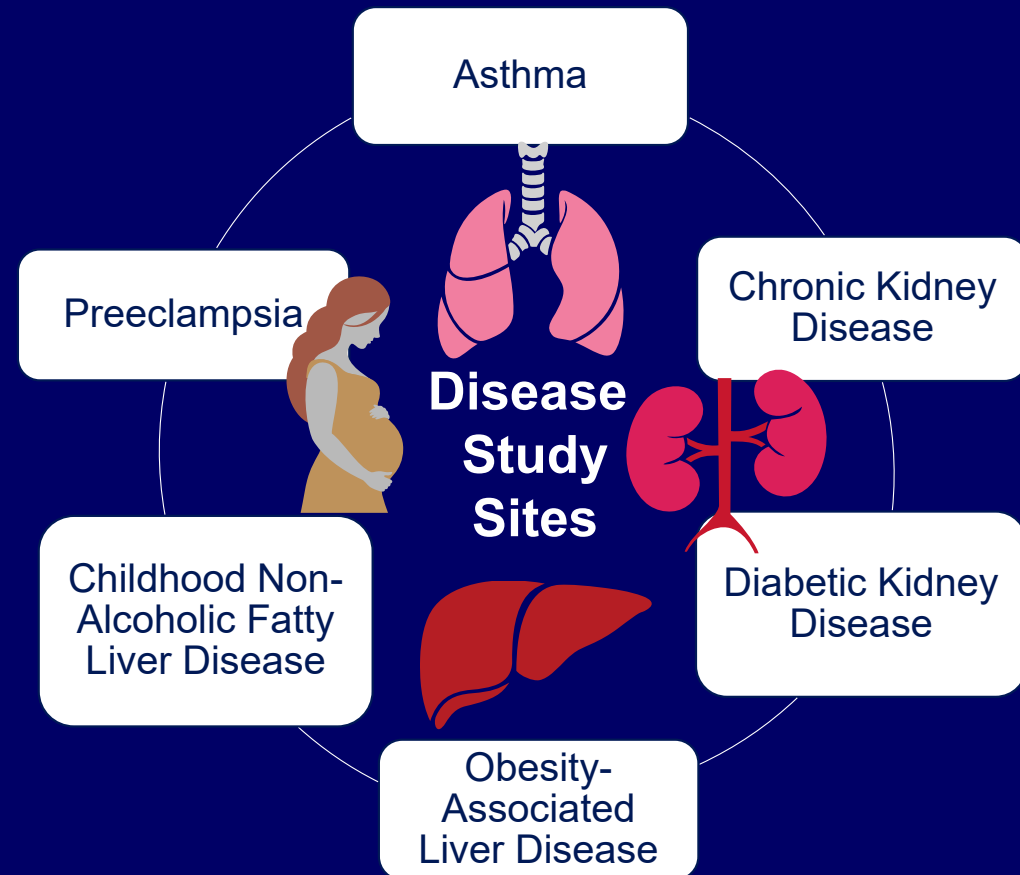
## Condition Selection Process



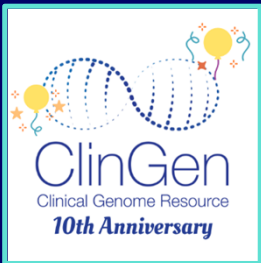




# Multi-Omics for Health and Disease (MOHD) Consortium



First in-person meeting: January 2024

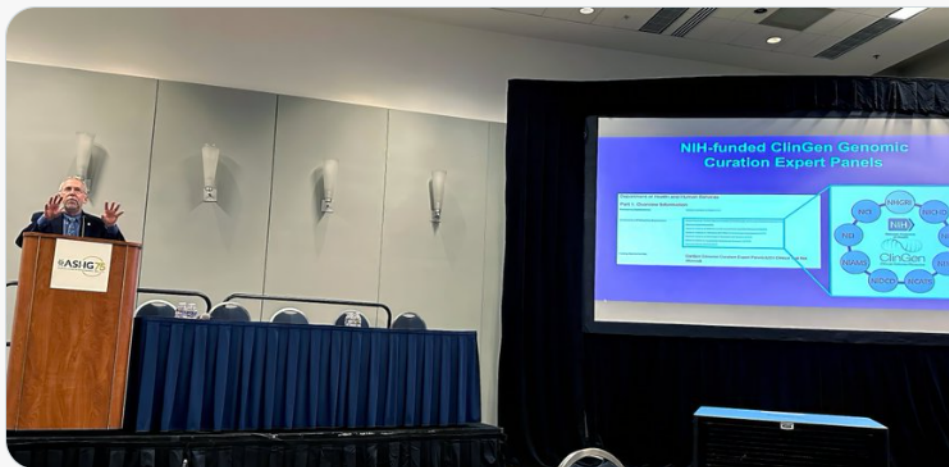


# Clinical Genome Resource (ClinGen) 10<sup>th</sup> Anniversary Event



**Eric Green** @NHGRI\_Director · Nov 3

I really enjoyed speaking to the **ClinGen** Consortium at their 10th Anniversary Celebratory Lunch yesterday at [#ASHG2023](#). What an amazing group – they have already accomplished so much for genomic medicine!

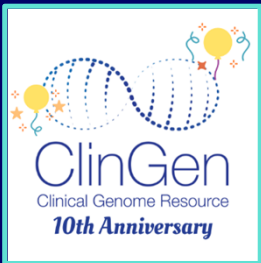


↻ 6

♥ 30

📊 3.4K



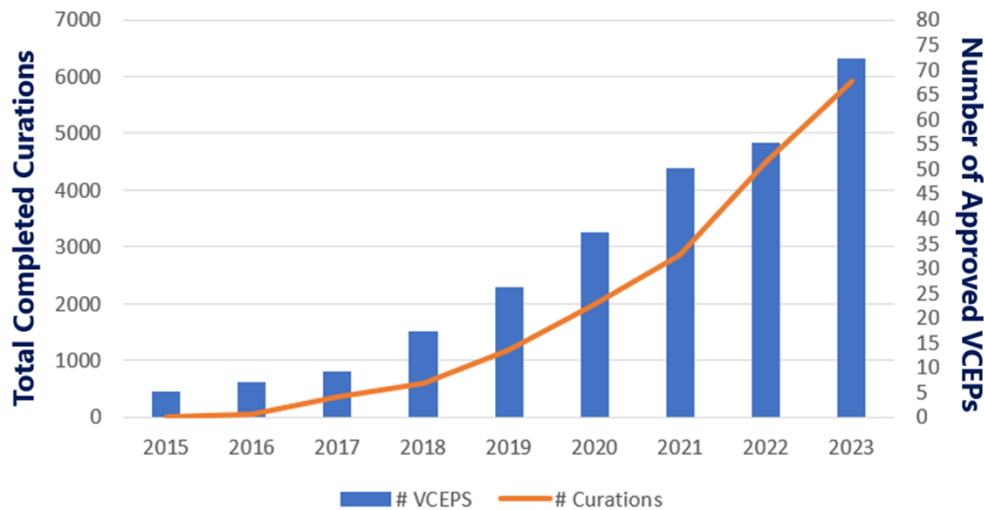


# Clinical Genome Resource (ClinGen)

## Progress with Genomic Variant Curation



**Variant Curation Growth**



### Variant Classification for Pompe disease; ACMG/AMP specifications from the ClinGen Lysosomal Diseases Variant Curation Expert Panel

Jennifer L Goldstein<sup>1</sup>, Jennifer McGlaughon<sup>2</sup>, Dona Kanavy<sup>3</sup>, Shelly Goomber<sup>3</sup>, Yinghong Pan<sup>4</sup>, Brett Deml<sup>5</sup>, Taraka Donti<sup>4</sup>, Liz Kearns<sup>6</sup>, Bryce A Seifert<sup>7</sup>, Miriam Schachter<sup>8</sup>, Rachel G Son<sup>9</sup>, Courtney Thaxton<sup>10</sup>, Rupa Udani<sup>11</sup>, Deeksha Bali<sup>3</sup>, Heather Baudet<sup>10</sup>, Michele Caggana<sup>12</sup>, Christina Hung<sup>2</sup>, Lianna Kyriakopoulou<sup>13</sup>, Lynne Rosenblum<sup>14</sup>, Robert Steiner<sup>15</sup>, Filippo Pinto E Vairo<sup>16</sup>, Yang Wang<sup>4</sup>, Michael Watson<sup>17</sup>, Raquel Fernandez<sup>17</sup>, Meredith Weaver<sup>17</sup>, Lorne Clarke<sup>18</sup>, Catherine Rehder<sup>3</sup>

Affiliations + expand

PMID: 37907381 DOI: 10.1016/j.ymgme.2023.107715

### Gene-specific ACMG/AMP classification criteria for germline APC variants: Recommendations from the ClinGen InSiGHT Hereditary Colorectal Cancer/ Polyposis Variant Curation Expert Panel



Isabel Spier<sup>1,2,3</sup>, Xiaoyu Yin<sup>3,4,5,\*</sup>, Marcy Richardson<sup>6</sup>, Marta Pineda<sup>3,7,8</sup>, Andreas Laner<sup>9</sup>, Deborah Ritter<sup>10,11</sup>, Julie Boyle<sup>12</sup>, Pilar Mur<sup>7,8</sup>, Thomas v O. Hansen<sup>13,14</sup>, Xuemei Shi<sup>15</sup>, Khalid Mahmood<sup>16,17</sup>, John-Paul Plazzer<sup>4</sup>, Elisabet Ognedal<sup>18</sup>, Margareta Nordling<sup>19,20</sup>, Susan M. Farrington<sup>21</sup>, Gou Yamamoto<sup>22</sup>, Stéphanie Baert-Desurmont<sup>23</sup>, Alexandra Martins<sup>23</sup>, Ester Borrás<sup>24</sup>, Carli Tops<sup>25</sup>, Erica Webb<sup>26</sup>, Victoria Beshay<sup>27</sup>, Maurizio Genuardi<sup>28</sup>, Tina Pesaran<sup>6</sup>, Gabriel Capellá<sup>3,7,8</sup>, Sean V. Tavtigian<sup>12,29</sup>, Andrew Latchford<sup>30,31</sup>, Ian M. Frayling<sup>30,32</sup>, Sharon E. Plon<sup>10,11</sup>, Marc Greenblatt<sup>33</sup>, Finlay A. Macrae<sup>4,5</sup>, Stefan Aretz<sup>1,2,3</sup>, on behalf of the InSiGHT-ClinGen Hereditary Colon Cancer/Polyposis Variant Curation Expert Panel



# Phenotypes and Exposures (PhenX) Toolkit



PhenX Toolkit

Home Protocols COVID-19 Search Resources News Help About Cite PhenX Contact

Search: Search all protocols in the Toolkit using keywords (e.g. diabetes) or PhenX ID (e.g. 011502)

 [New Social Determinants of Health protocols](#)

### PhenX Steering Committee Recommendations

- Use Social Determinants of Health: Core collection for all study designs
- Review Investigator Guidance for conducting research with human participants
- PhenX Citation
  - How to cite the PhenX Toolkit
  - How to cite use of PhenX protocols

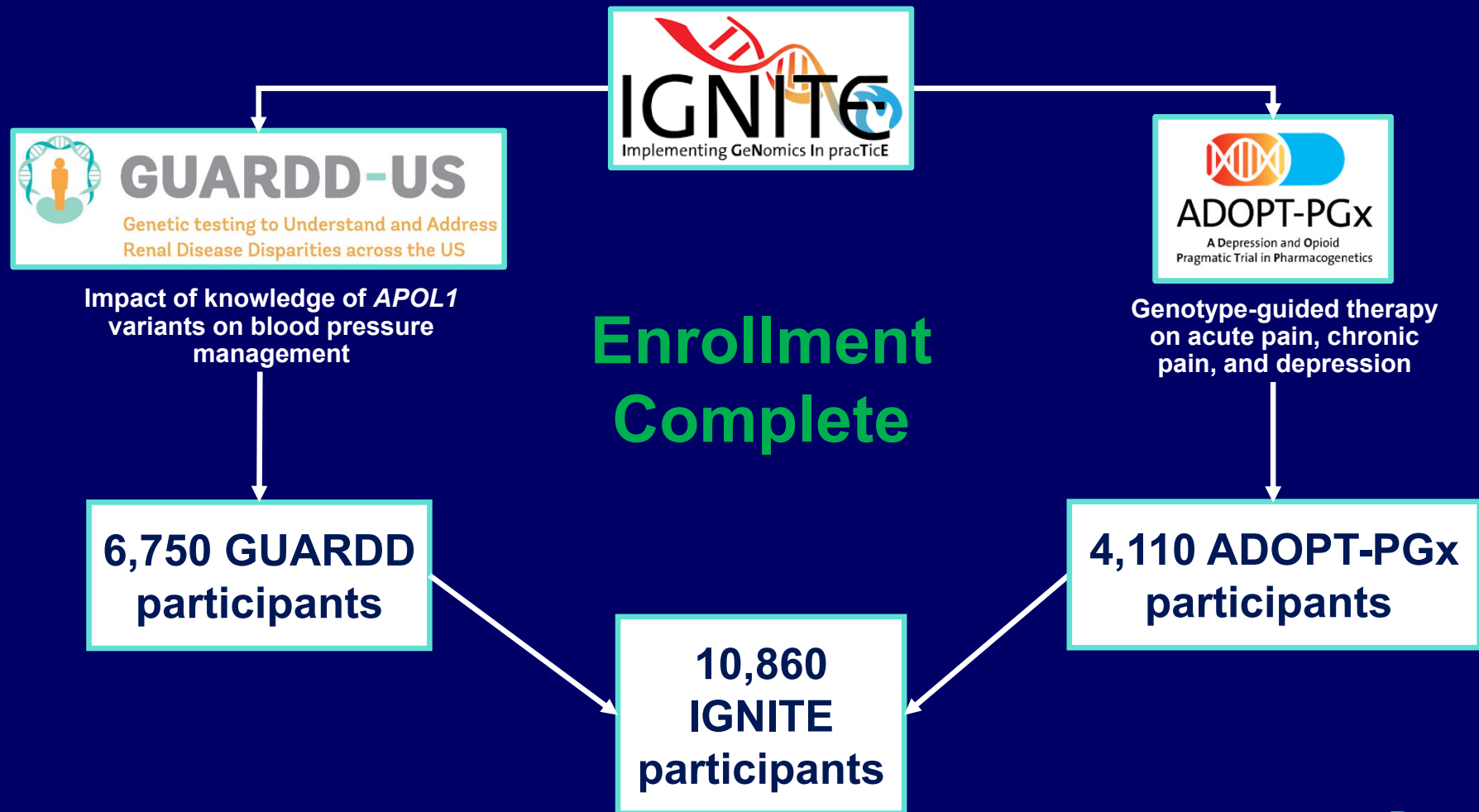
## Steering Committee Recommendation:

*All PhenX Toolkit users should consider adding Social Determinants of Health (SDOH) Core measures to their studies*



National Institute  
on Minority Health  
and Health Disparities

# Implementing Genomics in Practice (IGNITE)



# Regional Genomic Medicine eConsult Services

Department of Health and Human Services

## Part 1. Overview Information

Participating Organization(s) National Institutes of Health (NIH)

Components

Department of Health and Human Services

## Part 1. Overview Information

Participating Organization(s) National Institutes of Health (NIH)

Component

Department of Health and Human Services

## Part 1. Overview Information

Participating Organization(s) National Institutes of Health (NIH)

Components of Participating Organizations National Human Genome Research Institute (NHGRI)

Funding Opportunity Title Solutions to Enable Regional Genomic Medicine eConsult Services (R41/R42 Clinical Trial Optional)

Activity Code R41/R42 Small Business Technology Transfer (STTR) Grant - Phase I, and Fast-Track



Applications due on May 30, 2024

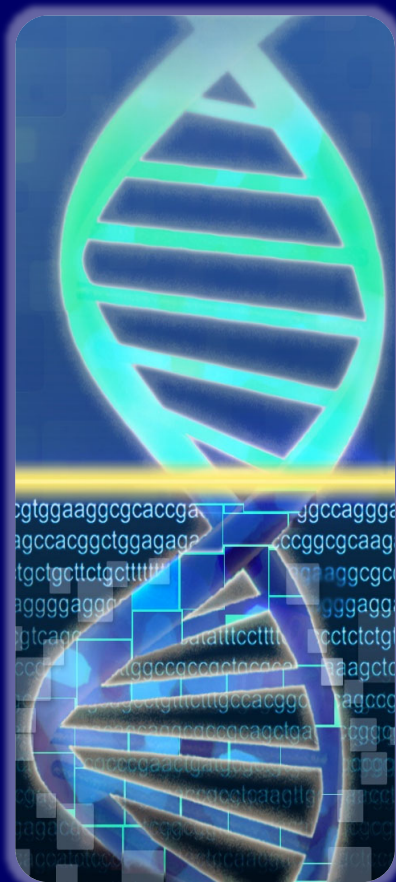


# Genomic Medicine XV: Genomics and Population Screening

- **Develop probabilistic models for conditions to be screened**
- **Establish pathways for clinical hand-off and follow-up of positive tests to clinical providers**
- **Ensure active community engagement and co-creation of screening and follow-up programs**
- **Pilot population screening for conditions that are ready for implementation in clinical settings**



# Genome Technology Program



- **Notice of Special Interest (NOSI): Advancing Genomic Technology Development for Research and Clinical Application**  
**NOT-HG-24-012 (R01, R21, R41, R42, R43, R44)**  
**Standard receipt dates**
- **NHGRI Technology Development Coordinating Center (U24, Clinical Trial Not Allowed)**  
**RFA-HG-24-013**  
**Applications due on April 2, 2024**

# Genome Technology Program



## Publication Highlight


nature chemical biology

Article <https://doi.org/10.1038/s41589-023-01384-1>

### Direct enzymatic sequencing of 5-methylcytosine at single-base resolution

Featuring:  
Rahul M. Kohli, M.D., Ph.D.  
University of Pennsylvania  
Tong Wang, M.D., Ph.D.  
Stanford University

  
@genomeTDCC




## Publication Highlight

PNAS RESEARCH ARTICLE | BIOCHEMISTRY

### A snoRNA-tRNA modification network governs codon-biased cellular states

Minjie Zhang<sup>1,2</sup>, Kongpan Li<sup>1</sup>, Jianhui Bai<sup>1</sup>, Ryan Van Damme<sup>2</sup>, Wei Zhang<sup>2</sup>, Mario Albar<sup>2</sup>, Bangyan L. Stalek<sup>2</sup>, Jian-Fu Chen<sup>2</sup>, and Zhipeng Lu<sup>1,2</sup>

Featuring:  
Zhipeng Lu, Ph.D.  
University of Southern California  
Minjie Zhang, Ph.D.  
Tianjin Medical University  
Kongpan Li, Ph.D.  
University of Southern California

  
@genomeTDCC



## Publication Highlight

nature biotechnology

Article <https://doi.org/10.1038/s41587-023-01854-x>

### Translocation of linearized full-length proteins through an engineered nanopore under opposing electrophoretic force

Featuring:  
Giovanni Maglia, Ph.D.  
University of Groningen  
Adina Sauciuc, MSc.  
University of Groningen

  
@genomeTDCC



## Publication Highlight

Nucleic Acids Research, 2023, 51  
<https://doi.org/10.1093/nar/gkz669>

### Observing inhibition of the SARS-CoV-2 helicase at single-nucleotide resolution

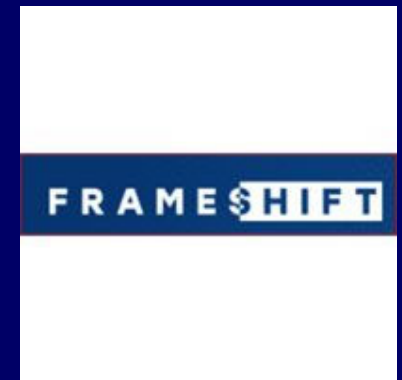
Sinduja K. Marx<sup>1</sup>, Keith J. Mickolajczyk<sup>1,3</sup>, Jonathan M. Craig<sup>1,1</sup>, Christopher A. Thomas<sup>1,1</sup>, Akira M. Pfeffer<sup>1</sup>, Sarah J. Abell<sup>1</sup>, Jessica D. Carrasco<sup>1</sup>, Michaela C. Franzl<sup>1</sup>, Jesse R. Huang<sup>1,1</sup>, Hwanhee C. Kim<sup>1,1</sup>, Henry Brinkerhoff<sup>1,1</sup>, Tarun M. Kapoor<sup>2</sup>, Jens H. Gundlach<sup>1</sup> and Andrew H. Laszlo<sup>1,1</sup>

Featuring:  
Andrew H. Laszlo Ph.D.  
University of Washington  
Sinduja K. Marx Ph.D.  
Seattle Children's Research Institute

  
@genomeTDCC

## Small Business Program

- ~\$17.9 million in Fiscal Year 2023
- 17 Phase I Proof-of-principle awards
- 13 Phase II Pre-commercialization awards



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

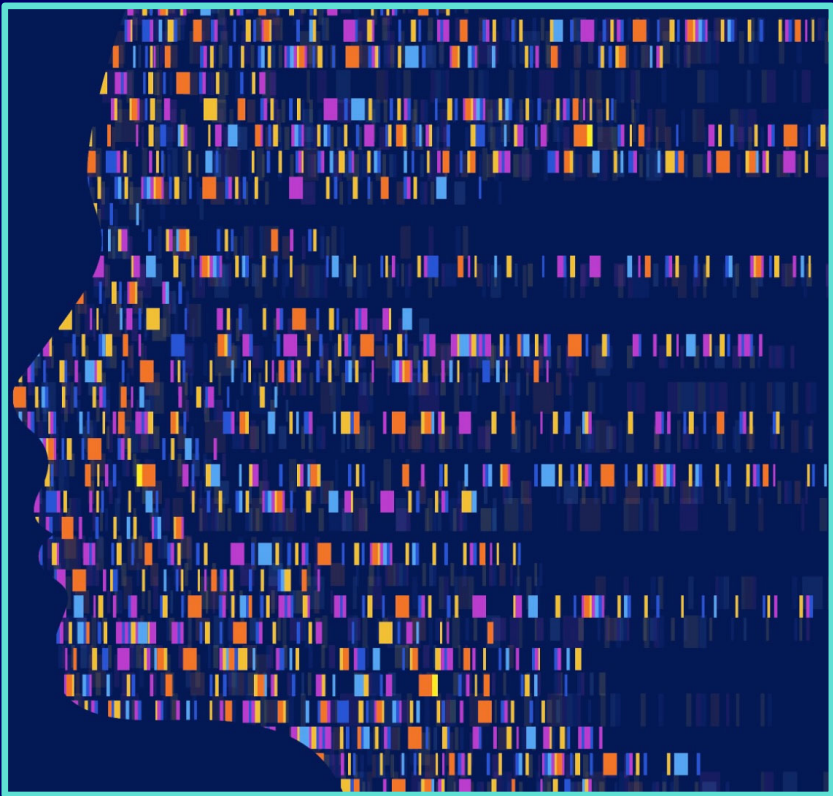
## Renewed Notices of Funding Opportunity

NOFO	Features
R01 <a href="#">PAR-23-293</a>	\$500K per year; up to 5 years Requires Plan for Enhancing Diverse Perspectives ( <a href="#">PEDP</a> )
R21 <a href="#">PAR-23-294</a>	\$275K total; Up to 3 years
R03 <a href="#">PAR-23-295</a>	\$50K per year; Up to 2 years

**Provide rationale for population descriptors**

# Training, Diversity, and Health Equity (TiDHE) Office

## Identifying and Evaluating Trainee Outcomes



- New evidence-based, online toolkit for genomics training programs
- Aids programs to implement streamlined, data-driven approaches to evaluate trainee outcomes and success
- Support trainee career goals and contribute to enhancing genomics workforce diversity



# Training, Diversity, and Health Equity (TiDHE) Office Outreach Activities



**SACNAS Conference: October 2023**

**ABRCMS Conference: November 2023**

# Training, Diversity, and Health Equity (TiDHE) Office

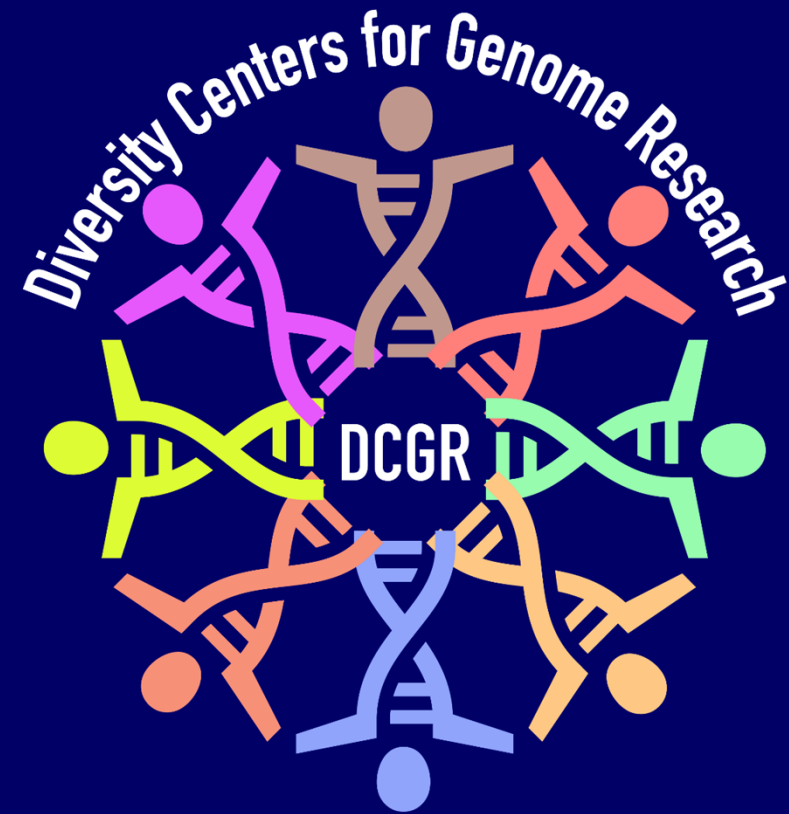
## New R25 Program Awardees

<b>Program</b>	<b>Principal Investigator</b>	<b>Institution</b>
<b>GREAT</b>	<b>Harriette Block</b>	<b>Prairie View A&amp;M University and Texas A&amp;M University</b>
	<b>Kristy Duran</b>	<b>Metropolitan State University of Denver</b>
<b>R25 Genetic Counselors</b>	<b>Nancy Cox</b>	<b>Vanderbilt University Medical Center</b>
<b>R25 Data Scientist</b>	<b>Audrey Hendricks</b>	<b>University of Colorado Denver</b>
	<b>Amanda Marie Larracuenta</b>	<b>University of Rochester</b>

# Training, Diversity, and Health Equity (TiDHE) Office

## Diversity Centers for Genome Research

- Support innovative genomics research, form interdisciplinary teams, build genomics research infrastructure
- Support research capabilities and enhance a diverse genomics workforce
- Increase diverse participation in genomics research



# Training, Diversity, and Health Equity (TiDHE) Office

## Diversity Centers for Genome Research



Anil Shanker, Ph.D.

UNIVERSITY  
*of* HAWAII  
MĀNOA

Lang Wu, Ph.D.



Sarah Williams-Blangero, Ph.D.

**Meharry One of Three Institutions for New NIH- Established Diversity Centers for Genome Research**

**\$10.9M to establish UH Pacific Center for Genome Research**

[UH News](#) » [Research](#) » [\\$10.9M to establish UH...](#)

UTRGV STDIOI investigators receive \$10M grant to establish an NIH Diversity Center for Genome Research

# Extramural Investigator-Initiated Highlights

**Improved sequence mapping using a complete reference genome and lift-over**

[Nae-Chy](#)

**Integrative single-cell characterization of a frugivorous and an insectivorous bat kidney and pancreas**

[Wei E. Gor](#)

[Kim, Alex C](#)

[Ilias Georg](#)

**Clinically Indicated Genomic Sequencing of Children in Foster Care: Legal and Ethical Issues**

[Hadley Stevens Smith, PhD](#)

[Stacey Pereira, PhD](#) • [Thor](#)

**Provision and availability of genomic medicine services in Level IV neonatal intensive care units**

[Monica H. Wojcik](#)<sup>1 2</sup>  , [Katharine P. Callahan](#)<sup>3 4</sup>, [Austin Antoniou](#)<sup>5</sup>, [Maya C. del Rosario](#)<sup>2</sup>, [Luca Brunelli](#)<sup>6</sup>, [Nahed O. ElHassan](#)<sup>7</sup>, [Semsu Gogcu](#)<sup>8</sup>, [Karna Murthy](#)<sup>9 10</sup>, [Jennifer A. Rumpel](#)<sup>7</sup>, [Jennifer A. Wambach](#)<sup>11</sup>, [Kristen Suhrie](#)<sup>12</sup>, [Kristen Fishler](#)<sup>13</sup>, [Bimal P. Chaudhari](#)<sup>5 14</sup>

# Resources for New Grant Writers



**NIH** National Human Genome  
Research Institute

**Inside the black box of the  
NIH funding system**

**Lisa Helbling Chadwick, Ph.D.**  
Program Director, NHGRI  
lisa.chadwick@nih.gov

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

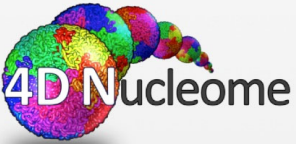
**View on YouTube: Inside the black box of the NIH funding system**

- **What funding announcement is right for me?**
- **How do I find a Program Officer to talk to?**
- **What happens in a grant review?**



# 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



# 4D Nucleome (4DN)

**CellPress** OPEN ACCESS **Cell**

Article  
**Spatial organization of the genome**  
Thomas A. Maniatis, Ashley L. Maniatis, Chunming Chen, Eric F. J. Velthuis

**RES** nature reviews genetics <https://doi.org/10.1038/s41576-023-00638-1>

**EPIGE**  
**Design**  
Review article  
Jeremy A. Boyce

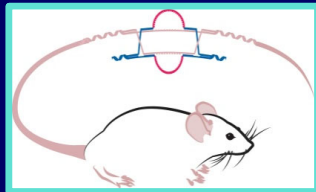
**CellPress** **Molecular Cell**

**Perspective**  
**Spatial and temporal organization of the genome: Current state and future aims of the 4D nucleome project**

Job Dekker,<sup>1,2,\*</sup> Frank Alber,<sup>3</sup> Sarah Aufmkolk,<sup>4</sup> Brian J. Beliveau,<sup>5</sup> Benoit G. Bruneau,<sup>6,12</sup> Andrew S. Belmont,<sup>7</sup> Lacramioara Bintu,<sup>8</sup> Alistair Boettiger,<sup>8</sup> Riccardo Calandrelli,<sup>9</sup> Christine M. Disteche,<sup>5</sup> David M. Gilbert,<sup>22</sup> Thomas Gregor,<sup>10</sup> Anders S. Hansen,<sup>11</sup> Bo Huang,<sup>12</sup> Danwei Huangfu,<sup>13</sup> Reza Kalhor,<sup>14</sup> Christina S. Leslie,<sup>13</sup> Wenbo Li,<sup>15</sup> Yun Li,<sup>16</sup> Jian Ma,<sup>17</sup> William S. Noble,<sup>5</sup> Peter J. Park,<sup>4</sup> Jennifer E. Phillips-Cremins,<sup>18</sup> Katherine S. Pollard,<sup>6,12,23</sup> Susanne M. Rafelski,<sup>19</sup> Bing Ren,<sup>9</sup> Yijun Ruan,<sup>20</sup> Yaron Shav-Tal,<sup>21</sup> Yin Shen,<sup>12</sup> Jay Shendure,<sup>5</sup> Xiaokun Shu,<sup>12</sup> Caterina Strambio-De-Castilla,<sup>1</sup> Anastassiia Vertii,<sup>1</sup> Huaiying Zhang,<sup>17</sup> and Sheng Zhong<sup>9,\*</sup>

Yang Zhang<sup>1</sup>, L...

# Knockout Mouse Phenotyping Project (KOMP2)



- **KOMP2 & IMPC Fall Meeting – November 2023**  
Hosted by Baylor College of Medicine
- **Booth at The Allied Genetics Conference – March 2024**
- **IMPC Virtual Workshop – March 2024**  
Neurodegenerative consequences of genetic defects in taste and olfactory genes in IMPC mice

# All of Us

## RESEARCH PROGRAM

- **\$1.5 Million in awards to institutions partnering with American Indian, Alaska Native, and Indigenous communities**
- **\$9.7 Million in new awards to advance the use of the *All of Us* Dataset**



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- VI. NHGRI Communications, Policy, and Education**
- VII. NHGRI Intramural Research Program

# NHGRI Exhibits at Annual Meetings



**American Society of Human Genetics, November 2023**

**American College of Medical Genetics and Genomics, March 2024**



# New Instagram Accounts for NHGRI Director & NHGRI

nhgri\_director Following Message ...

9 posts 172 followers 175 following

**Eric Green** he/him/his  
Government Official  
@nhgri\_nih Director, Genomicist. @cardinals fan. He/Him/His  
🌐 [www.genome.gov/About-NHGRI/Director](http://www.genome.gov/About-NHGRI/Director) + 2

Followed by e.dietz, nhgri\_nih, and lifeafterfiftysomething

POSTS REELS TAGGED

Grid of posts: 1. Chromosomes. 2. Genomic map. 3. Portrait of Eric Green.

nhgri\_director ...

View insights Boost post

Liked by abbeygreen22 and 22 others

nhgri\_director I am officially kicking off a new weekly feature — called #WildlifeWednesday! For my... more

nhgri\_director ...

View insights Boost post

Liked by abbeygreen22 and 7 others

nhgri\_director We are back with another #WildlifeWednesday, this time with the first of what will be a number of... more

nhgri\_director ...

View insights Boost post

Liked by josh\_green\_redhead and 4 others

nhgri\_director Since I am attending the #AGBT2024 this week, I thought it would be appropriate to use an AGBT-themed ... more

nhgri\_nih Following Message ...

54 posts 2,325 followers 378 following

**National Human Genome Research Institute**  
Government organization  
Funding research at the forefront of genomics to improve human health. Formerly the Human Genome Project. Part of @nihgov  
🌐 [genome.gov](http://genome.gov) + 4

Followed by nshelter8, jenK0\_jeans, lifeafterfiftysomething + 12 more

POSTS REELS TAGGED

# NHGRI History of Genomics Program

## Archival Spotlight

National Institutes of Health  
AD HOC PROGRAM ADVISORY COMMITTEE  
 ON  
 COMPLEX GENOMES  
Draft Agenda

Monday, February 29

Morning Session

9:00 Opening Remarks . . . . . Dr. Wyngaarden  
 9:10 Legislative Activities . . . . . Dr. Moskowitz

Status of NIH Effort: Report of the NIH Working Group on the Human Genome

9:20 Chairman's Comments . . . . . Dr. Wyngaarden  
 9:30 National Institute of General Medical Sciences . . . . . Dr. Kirschstein  
 9:40 National Library of Medicine . . . . . Dr. Lindberg  
 9:50 Division of Research Resources . . . . . Dr. Pickett  
 9:55 National Institute of Child Health and  
 Human Development . . . . . Dr. Alexander  
 10:00 Division of Research Grants . . . . . Dr. Green

10:10 Discussion

10:30 Coffee Break

10:45 Science Working Groups

- A. Mapping
- B. Sequencing
- C. Information

11:30 Lunch

Afternoon Session

1:00 Science Working Groups (Cont.)

3:00 Coffee Break

Science Working Group Presentations

3:15 A. Mapping . . . . . Drs. Botstein/Cantor  
 3:30 B. Sequencing . . . . . Drs. Hood/Ayala  
 3:45 C. Information . . . . . Drs. Lander/Smith

4:00 Discussion

5:30 Adjourn

6:00 Dinner

Evening Session

7:30 Needs Working Groups

- 1. Scientific Needs
- 2. Resource Needs
- 3. Administrative Mechanisms

9:00 Adjourn

Tuesday, March 2

8:00 Continental Breakfast

Needs Working Groups Presentations

8:30 1. Scientific Needs . . . . . Drs. Collins/Olsen  
 8:45 2. Resource Needs . . . . . Drs. Caskey/White  
 9:00 3. Administrative Mechanisms . . . . . Drs. Baltimore/

9:15 Discussion

10:45 Coffee Break

11:00 Recommendations

Noon Adjourn/Lunch for Working Group Chairmen

1:00 Draft Report

3:00 Adjourn

AD HOC PROGRAM ADVISORY COMMITTEE ON COMPLEX GENOMES

Sheraton International Conference Center  
 Reston, Virginia

February 29-March 1, 1988

List of Committee Members

Chairman: David Baltimore, Ph.D.  
 Director  
 Whitehead Institute  
 9 Cambridge Center  
 Cambridge, Massachusetts 02142  
 (617) 258-5155

Francisco Ayala, Ph.D.  
 Distinguished Professor  
 of Biological Sciences  
 Department of Ecology and Evolutionary  
 Biology  
 University of California at Irvine  
 916 Engineering Building  
 Irvine, California 92717  
 (714) 856-8293

C. Thomas Caskey, M.D.  
 Investigator  
 Howard Hughes Medical Institute  
 Director and Professor  
 Institute for Molecular Genetics  
 Baylor College of Medicine  
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 Houston, Texas 77030  
 (713) 799-4774

Frederick R. Blattner, Ph.D.  
 Professor  
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 Madison, Wisconsin 53706  
 (608) 262-2534

Francis S. Collins, M.D., Ph.D.  
 Assistant Investigator  
 Howard Hughes Medical Institute  
 Chief  
 Division of Medical Genetics  
 University of Michigan  
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 Ann Arbor, Michigan 48109-0618  
 (313) 747-3414

David Botstein, Ph.D.  
 Vice President of Science  
 Genentech, Inc.  
 460 Point San Bruno Boulevard  
 South San Francisco, California 94080  
 (415) 266-2199

Leroy Hood, M.D., Ph.D.  
 Bowles Professor and Chairman  
 Division of Biology, 156-29  
 California Institute of Technology  
 Pasadena, California 91125  
 (818) 356-4951


Charles R. Cantor, Ph.D.  
 Professor and Chairman  
 Department of Genetics and Development  
 Columbia University  
 College of Physicians and Surgeons  
 Suite 1602  
 701 West 168th Street  
 New York, New York 10032  
 (212) 305-7915

Eric S. Lander, Ph.D.  
 Whitehead Fellow  
 Whitehead Institute  
 Room 467  
 9 Cambridge Center  
 Cambridge, Massachusetts 02142  
 (617) 258-5192

# Undergraduate Genetics Education Network (UGEN)

4TH ANNUAL  
**Undergraduate  
Genetics Education Network's  
Virtual Workshop**

Thursday, January 4, 2024 | 11 am - 4 pm EST



**UGEN**  
DNA


**Keynote speaker**  
**Chris Donohue, PhD**  
Historian, NHGRI History of Genomics

**Geneticists' Support for Eugenics  
Before and After WWII**

Interactive sessions on:

- Giving undergraduates agency over their own learning
- Including students in creating a more inclusive genetics curriculum
- Resources and implementation ideas

Stay tuned for more information  
and how to register

Co-hosted by:  
 **NIH** National Human Genome  
Research Institute



**Dina Newman, Ph.D.**  
**Rochester Institute  
of Technology**



**Rivka Glaser, Ph.D.**  
**Stevenson  
University**

# ISCC-PEG Scholars Program



**Swathi Balaji,  
M.P.H, M.S.  
Genetic Counselor  
University of Pittsburgh**



**Chase Beeman,  
M.S.  
Genetic Counselor  
Ohio State University**



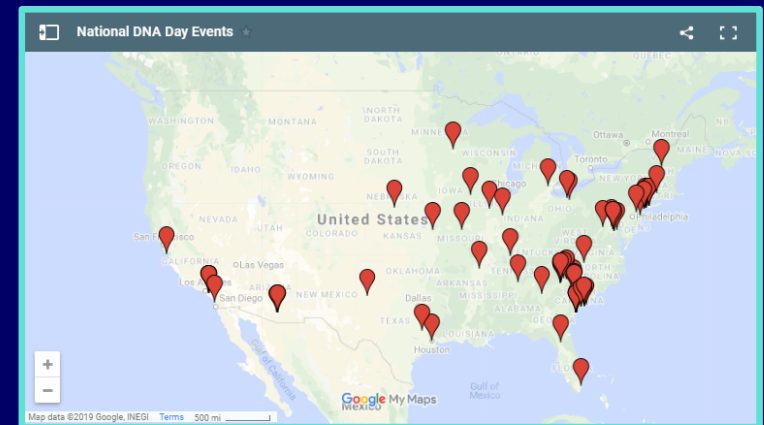
**Vanessa Smith,  
M.S.  
Genetic Counselor  
Vanderbilt University**



**Setareh Zandihaghighi,  
M.S.  
Genetic Counselor  
University of Michigan**



# Celebrate National DNA Day 2024!



# 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**



# Celebrating 30 Years of the NHGRI Intramural Research Program



## A conversation with the 4 NHGRI Scientific Directors

### Part 1



Recorded in July 2023

# Founding Director, NHGRI Center for Genomics and Data Science Research

—  
**NHGRI selects Adam Phillippy  
as first director of new Center  
for Genomics and Data  
Science Research**



# Gene Therapies for Sickle Cell Disease Community-based Research Project

NHLBI NEWS | Statement

## NIH statement on new FDA-approved gene therapies for sickle cell disease

December 8, 2023, 10:00 AM EST



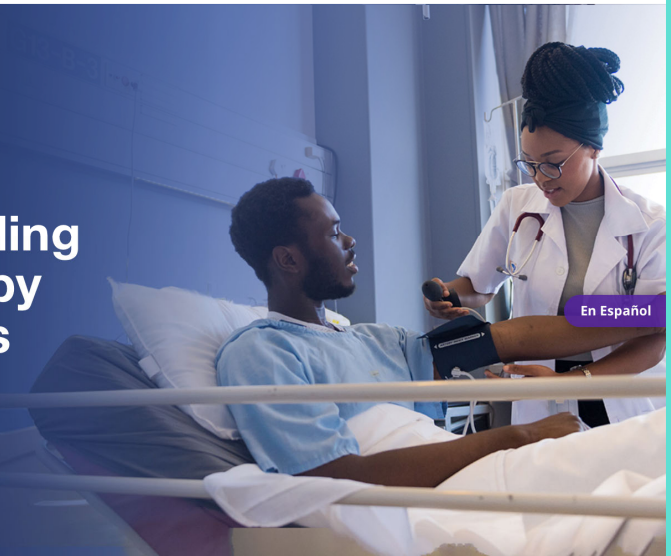
Today, the U.S. Food and Drug Administration (FDA) approved two gene therapies for the treatment of sickle cell disease in patients 12 years and older. About [100,000 Americans and millions of people](#) around the world have [sickle cell disease](#), a hereditary disease common among those whose ancestors come from sub-Saharan Africa, Mediterranean countries, India and the Middle East.

NIH National Human Genome  
Research Institute



Democratizing Education for  
Sickle Cell Disease Gene Therapy Project

## Understanding gene therapy approaches



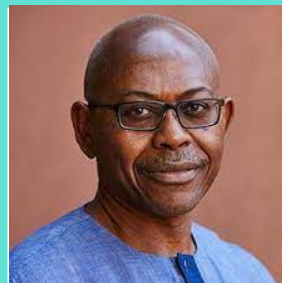
En Español

# NHGRI Intramural Research Highlights



## Science

**The history of Coast Salish “woolly dogs” revealed by ancient genomics and Indigenous Knowledge**



## European Journal of Human Genetics

**Genome, HLA and polygenic risk score analyses for prevalent and persistent cervical HPV infections**



BMC Part of Springer Nature

## Genome Biology

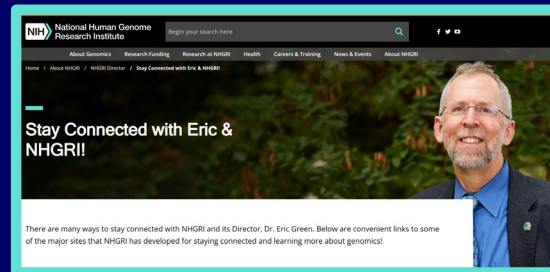
**A genome catalog of the early-life human skin microbiome**





# 'One-Stop-Shop' to Stay Connected

## genome.gov/stayconnected



**Genome.gov**  
Explore the official website of the National Human Genome Research Institute.

**NHGRI Director's Biography**  
Learn more about the NHGRI Director.

**The Genomics Landscape**  
Subscribe to Dr. Eric Green's monthly newsletter summarizing NHGRI and genomics highlights.

**Dr. Eric Green on LinkedIn**  
Follow Dr. Green, M.D., Ph.D. on LinkedIn.

**Dr. Eric Green on Instagram**  
Follow @eric\_green on Instagram.

**Dr. Eric Green on X**  
Follow @NHGRI\_Director on X.

**2020 NHGRI Strategic Vision**  
Read NHGRI's strategic vision for improving human health at the beginning of the decade.

**Building a Diverse Genomics Workforce**  
Read about NHGRI's plans to increase the number of individuals from diverse backgrounds in the genomics workforce.

**NHGRI Brochure**  
Learn more about NHGRI and the Institute's history, core activities and values, research portfolio, and various other programs.

**GenomeTV**  
Watch GenomeTV, a vast collection of HUGO videos, including research highlights, interviews, seminars, and educational films.

**NHGRI's Oral History Collection**  
View oral histories of those who have shaped and advanced the genomics program and other NHGRI endeavors.

**Talking Glossary of Genetic and Genomic Terms**  
Gain a better understanding of hundreds of genetic and genomic terms and concepts using the accessible NHGRI resources.

### Among the Best of Eric Green

#### Videos

**IBB Summit: "Creating the Fourth Chapter of Human Genomics"**  
October 2, 2022

**PGH Summit 2023: Genomics: Making Genomic Medicine a Reality**  
October 3, 2023

**Scale of the Human Genome (with animations)**  
January 21, 2020

**GMA K-12: 2020 NHGRI Strategic Vision**  
February 9, 2021

**Talking Glossary of Genetic and Genomic Terms**  
December 6, 2023

**20 Years After the Human Genome Project: GS Leaders Reunite**  
September 6, 2023

### Podcasts

**Genetics Wrapped with Eric Green and Brandon Lee (Episode #268)**

**Phenotype Population Genomics in Clinical Practice (Episode #269)**

**Genetics Wrapped with Eric Green (Episode #214)**

**Precision health and genomics with Dr. Eric Green**

**Eric Green on the Future of NHGRI**

**Eric Green on the Complete Human Genome Project**

**The Butterfly Effect**

**Genomics: Rewriting Our Health**

**The Past, Present and Future of the Human Genome Project**

### Published Features

- Green Day: An Interview with NHGRI Director Eric Green**  
*GEN Biotechnol*, April 18, 2023
- Completing the Human Genome Sequence (Again)**  
*Scientific American*, March 31, 2022
- An Anti-racist Action Plan for Studying the Human Genome**  
*The Hill*, September 16, 2021
- The Genomics Workforce Must Become More Diverse: A Strategic Imperative**  
*Am J Hum Genet*, January 7, 2021
- A Vision for the Next Decade of Human Genomics Research**  
*Scientific American*, October 28, 2020







National Human Genome  
Research Institute

**Thanks!**



**Special Thanks!**



The **Forefront**  
of **Genomics**<sup>®</sup>