

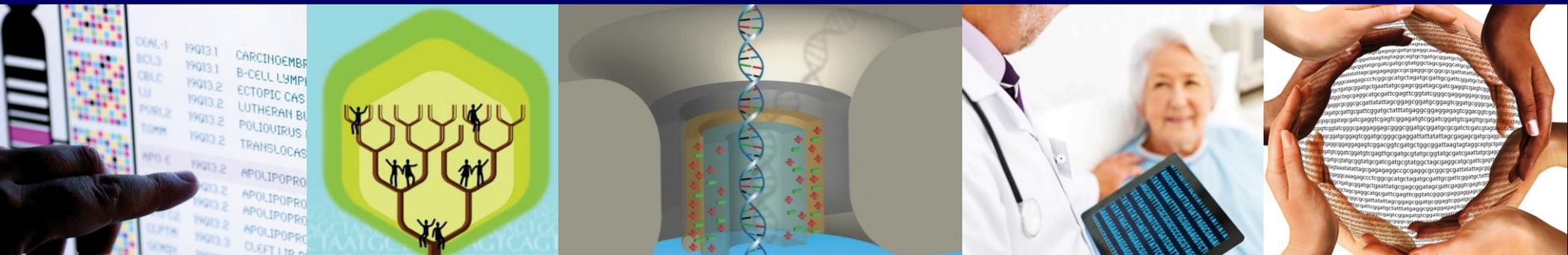


—  
The **Forefront**  
of **Genomics**  
—

# DIRECTOR'S REPORT

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

**May 2021**





## Director's Report-Related Documents

May 2021

Director's Report

Director's Report

| No. | Relevant Documents  |
|-----|---|
| 1   | New NHGRI Brochure  |
| 2   | <a href="#">Bold Predictions for Human Genomics by 2030: NHGRI Seminar Series</a> |
| 3   | <a href="#">NHGRI Strategic Plan for Congress</a>                                 |

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

Document #



# Open Session Presentations

## Presentations:

**Genomics & Society Working Group of NACHGR Annual Report**  
**Steven Joffe**

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

# Open Session Presentations

## Concept Clearances:

**Supporting Talented Early Career Researchers in Genomics**

**Lisa Chadwick**

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

**Adam Felsenfeld**

**The Knockout Mouse Phenotyping Program**

**Colin Fletcher**

**Curriculum Development in Genomics, Genetics, or Genomic Informatics for Medical Students**

**Heather Colley**

# 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 NHGRI Brochure



- NHGRI's history, organization, values, and major research areas
- PDF and 'flipbook' versions available at [genome.gov/brochure](http://genome.gov/brochure)
- Printed copies will be available on request

# Bold Predictions for Human Genomics by 2030

## NHGRI Seminar Series

### Bold Predictions for Human Genomics

A National Human Genome Research Institute (NHGRI) Seminar Series

#### Bold Prediction #1

February 1, 2021; 3 p.m. - 4:30 p.m. ET

Generating and analyzing a complete human genome sequence will be routine for any research laboratory, becoming as straightforward as carrying out a DNA purification.

Evan Eichler, Ph.D., University of Washington  
Karen Miga, Ph.D., University of California, Santa Cruz  
Moderator: Eric Green, M.D., Ph.D., NHGRI

#### Bold Prediction #2

March 8, 2021; 3 p.m. - 4:30 p.m. ET

The biological function(s) of every human gene will be known; for non-coding elements in the human genome, such knowledge will be the rule rather than the exception.

Nancy Cox, Ph.D., Vanderbilt University  
Neville Sanjana, Ph.D., New York Genome Center  
Moderator: Carolyn Hutter, Ph.D., NHGRI

#### Bold Prediction #3

April 12, 2021; 3 p.m. - 4:30 p.m. ET

The general features of the epigenetic landscape and transcriptional output will be routinely incorporated into predictive models of the impact of genotype on phenotype.

Tom Gingeras, Ph.D., Cold Spring Harbor Laboratory  
Tuuli Lappalainen, Ph.D., New York Genome Center  
Moderator: Paul Liu, M.D., Ph.D., NHGRI

#### Bold Prediction #4

May 25, 2021; 3 p.m. - 4:30 p.m. ET

Research in human genomics will have moved beyond population descriptors based on historic social constructs such as race.

Charmaine Royal, Ph.D., Duke University  
Genevieve Wojcik, Ph.D., Johns Hopkins University  
Moderator: Vence Bonham, Jr., J.D., NHGRI

#### Bold Prediction #5

June 7, 2021; 3 p.m. - 4:30 p.m. ET

Studies involving analyses of genome sequences and associated phenotypic information for millions of human participants will be regularly featured at school science fairs.

Neil Lamb, Ph.D., HudsonAlpha Institute for Biotechnology  
Chanda Jefferson, Albert Einstein Distinguished Educator Fellow  
Office of Mark DeSaulniers, California's 11th Congressional District  
Moderator: Larry Brody, Ph.D., NHGRI

### Bold Predictions for Human Genomics by 2030

A National Human Genome Research Institute (NHGRI) Seminar Series

#### Bold Prediction #6

July 12, 2021; 3 p.m. - 4:30 p.m. ET

The regular use of genomic information will have transitioned from boutique to mainstream in at clinical settings, making genomic testing as routine as complete blood counts (CBCs).

Jennifer Posny, M.D., Ph.D., Baylor College of Medicine  
Kathrina Armstrong, M.D., Massachusetts General Hospital and Harvard Medical School  
Moderator: Teri Manolio, M.D., Ph.D., NHGRI

#### Bold Prediction #7

September 16, 2021; 3 p.m. - 4:30 p.m. ET

The clinical relevance of all encountered genomic variants will be readily predictable, rendering the diagnostic designation "variant of uncertain significance (VUS)" obsolete.

Heidi Rehm, Ph.D., Broad Institute, Harvard Medical School and Massachusetts General Hospital  
Douglas Fowler, Ph.D., University of Washington  
Moderator: Gus Stolton, M.D., NHGRI

#### Bold Prediction #8

October 4, 2021; 3 p.m. - 4:30 p.m. ET

A person's complete genome sequence along with informative annotations can be securely and readily accessible on their smartphone.

Michael Schatz, Ph.D., Johns Hopkins University and Cold Spring Harbor Laboratory  
Gillian Hooker, Ph.D., ScM, LCGC, Concert Genetics  
Moderator: Sarah Bates, M.S., NHGRI

#### Bold Prediction #9

November 1, 2021; 3 p.m. - 4:30 p.m. ET

Individuals from ancestrally diverse backgrounds will benefit equitably from advances in human genomics.

Robert Wirtz, M.D., Virginia Commonwealth University  
Alison M. Hahn, Ph.D., Massachusetts General Hospital, Harvard Medical School, and Broad Institute  
Moderator: Chris Gunter, Ph.D., NHGRI

#### Bold Prediction #10

January 10, 2022; 3 p.m. - 4:30 p.m. ET

Genomic discoveries will lead to curative therapies involving genomic modifications for dozens of genetic diseases.

Timothy Yu, M.D., Ph.D., Boston Children's Hospital and Harvard Medical School  
Alexis Thompson, M.D., MPH, Ann and Robert H. Lurie Children's Hospital of Chicago and Northwestern University Feinberg School of Medicine  
Moderator: Dan Kastner, M.D., Ph.D., NHGRI

## Bold Prediction #4

### May 25, 2021; 3 p.m. - 4:30 p.m. ET

Research in human genomics will have moved beyond population descriptors based on historic social constructs such as race.

Charmaine Royal, Ph.D., Duke University  
Genevieve Wojcik, Ph.D., Johns Hopkins University  
Moderator: Vence Bonham, Jr., J.D., NHGRI



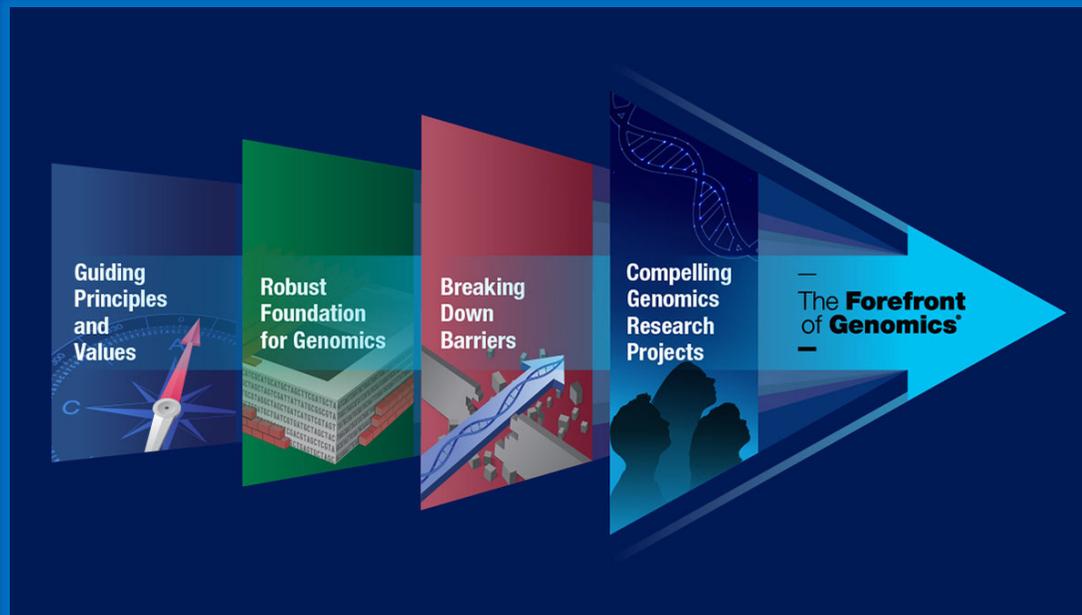
See our bold predictions at: [genome.gov/bold-predictions](https://genome.gov/bold-predictions)



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# NHGRI Strategic Plan for Congress



**Perspective**  
**Strategic vision for improving human health at The Forefront of Genomics**

<https://doi.org/10.1038/s41586-020-2017-4>  
Received: 30 June 2020  
Accepted: 4 September 2020  
Published online: 28 October 2020  
Check for updates

Erik D. Green<sup>1</sup>, Chris Gunter<sup>1</sup>, Leslie G. Besserker<sup>1</sup>, Valentin D. Franconi<sup>1</sup>, Carla L. Eastler<sup>1</sup>, Elise A. Fergolito<sup>1</sup>, Adam L. Fehon<sup>1,2</sup>, David J. Kaufman<sup>1</sup>, Elaine A. Ottensmeyer<sup>1</sup>, William J. Peaver<sup>1</sup>, Adam M. Phillips<sup>1</sup>, Anastasia L. Wasi<sup>1</sup>, Joel Gupta David<sup>1</sup>, Shelby J. Khan<sup>1</sup>, Allison Mandel<sup>1</sup>, Christopher B. Wiedinger<sup>1</sup>, Kris A. Wettenstein<sup>1</sup>, Sarah A. Bate<sup>1</sup>, Darryl Lipp<sup>1</sup>, Susan Vazquez<sup>1</sup>, William A. Gahl<sup>1</sup>, Bette J. Grunham<sup>1</sup>, Daniel L. Kastner<sup>1</sup>, Paul Liu<sup>1</sup>, Laura Lynn Rodriguez<sup>1</sup>, Benjamin D. Solomon<sup>1</sup>, Wenzel L. Bonham<sup>1</sup>, Lawrence C. Brody<sup>1</sup>, Carolyn M. Hutten<sup>1</sup> & Teri A. Manolio<sup>1</sup>

Starting with the launch of the Human Genome Project three decades ago, and continuing after its completion in 2003, genomics has progressively come to have a central and catalytic role in basic and translational research. In addition, studies increasingly demonstrate how genomic information can be effectively used in clinical care. In the future, the anticipated advances in technology development, biological insights, and clinical applications (among others) will lead to more widespread integration of genomics into almost all areas of biomedical research, the adoption of genomics into mainstream medical and public health practices, and an increasing relevance of genomics for everyday life. On behalf of the research community, the National Human Genome Research Institute recently completed a multi-year process of strategic engagement to identify future research priorities and opportunities in human genomics, with an emphasis on health applications. Here we describe the highest priority elements envisioned for the cutting edge of human genomics going forward—that is, at ‘The Forefront of Genomics’.

Beginning in October 1990, a pioneering group of international researchers began an audacious journey to generate the first map and sequence of the human genome, marking the start of a 13-year odyssey called the Human Genome Project<sup>1</sup>. The successful and early completion of the Project in 2003, which included parallel studies of a set of model organism genomes, catalysed enormous progress in genomics research. Leading the signature advances has been a greater than one million-fold reduction in the cost of DNA sequencing<sup>2</sup>. This decrease has allowed the generation of immenseable genome sequences, including hundreds of thousands of human genome sequences both in research and clinical settings, and the continuous development of assays to identify and characterize functional genomic elements<sup>3</sup>. These new tools, together with increasingly sophisticated statistical and computational methods, have enabled researchers to create rich catalogues of human genome variants<sup>4</sup>. As a gain-in-depth understanding of the functional complexities of the human genome<sup>5</sup>, and to determine the genomic bases of thousands of human diseases<sup>6,7</sup>. In turn, the past decade has brought the initial realization of genomic medicine<sup>8</sup>—as research successes have been converted into powerful tools for use in healthcare, including somatic genome analysis for cancer (enabling development of targeted therapeutic agents)<sup>9</sup>, non-invasive prenatal genetic screening<sup>10</sup>, and genomics-based tests for a growing set of paediatric conditions and rare disorders<sup>11</sup>, among others.

In essence, with growing insights about the structure and function of the human genome and ever-improving laboratory and computational technologies, genomics has become increasingly woven into the fabric of biomedical research, medical practice, and society. The scope, scale, and pace of genomic advances so far were nearly unimaginable when the Human Genome Project began over 30 years ago, such advances are yielding scientific and clinical opportunities beyond our initial expectations, with many more anticipated in the next decade.

Embracing its leadership role in genomics, the National Human Genome Research Institute (NHGRI) has developed strategic visions for the field by reflection notes, in particular at the end of the Human Genome Project in 2003<sup>12</sup> and then again at the beginning of the last decade in 2012<sup>13</sup>. These visions outlined the most compelling opportunities for human genomics research, in each case informed by a multi-year engagement process. NHGRI endeavored to start the new decade with an updated strategic vision for human genomics research. Through a planning process that involved more than 50 events (such as dedicated workshops, conference sessions, and webinars) over the past two years (see <http://genome.gov/genomics2020>), the Institute collected input from a large number of stakeholders, with the resulting report catalogued and synthesized using the framework depicted in Fig. 1.

Unlike the past, this round of strategic planning was greatly influenced by the now widely disseminated nature of genomics across biomedicine. A representative glimpse into this historic phenomenon is illustrated in Fig. 2, tracing the Human Genome Project, NHGRI was the primary funder of human genomics research at the US National Institutes of Health (NIH), but the past two decades have brought a greater than tenfold increase in the relative fraction of funding coming from other parts of the NIH.

National Human Genome Research Institute, National Institutes of Health, Bethesda, MD, USA. \*[terman@nhgri.nih.gov](mailto:terman@nhgri.nih.gov)

Nature | Vol 586 | 28 October 2020 | 683

- NIH Institutes/Centers required to submit a plan every 5 years
- Companion document to go with 2020 NHGRI Strategic Vision
- Develop a framework for crafting next plan in 5 years

Document 3

# NHGRI Staff Volunteer in Support of Unaccompanied Children's Program



# Director's Report Outline

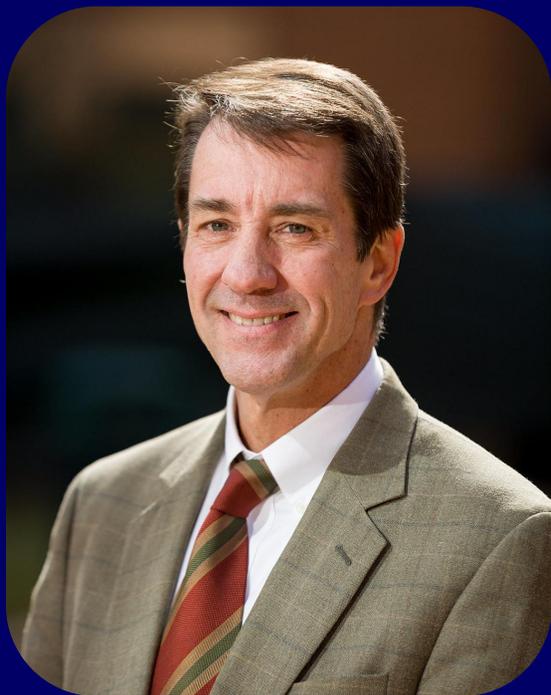
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# New Secretary, Department of Health and Human Services



**Xavier Becerra, J.D.**

# Departure of Director, National Center for Advancing Translational Sciences



**Chris Austin, M.D.**



**Joni Rutter, Ph.D.**

# NIH UNITE Initiative



- U — Understanding stakeholder experiences through listening and learning**
- N — New research on health disparities, minority health, and health equity**
- I — Improving the NIH culture and structure for equity, inclusion, and excellence**
- T — Transparency, communication, and accountability with our internal and external stakeholders**
- E — Extramural research ecosystem: changing policy, culture, and structure to promote workforce diversity**

# NIH COVID-19 Information Resources

- **NOT-OD-21-106: Continuation of Temporary Extension of Eligibility for the NIH K99/R00 Pathway to Independence Award During the COVID-19 Pandemic**
- **RFA-OD-21-009: RADx-UP - Social, Ethical, and Behavioral Implications Research on Disparities in COVID-19 Testing among Underserved and Vulnerable Populations**



**OPEN MIKE**

*Helping connect you with the NIH perspective, and helping connect us with yours.*

## Fiscal Year 2022 Appropriations

|              | Fiscal Year<br>2021<br>Enacted | Fiscal Year<br>2022<br>President's<br>Budget<br>Outline | \$<br>Increase | %<br>Increase |
|--------------|--------------------------------|---|----------------|---------------|
| <b>HHS</b>   | \$108.6 B                      | \$133.7 B   | \$25.1 B       | 23.1%         |
| <b>NIH</b>   | \$41.7 B                       | \$51.0 B  | \$9.0 B *      | 21.6%         |
| <b>NHGRI</b> | \$606.3 M                      | TBD   | TBD            | TBD           |

\* ARPA-H would receive \$6.5 billion of the \$9 billion increase for NIH

# NIH Feline-in-Chief and Head NIH Zoom Bomber



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# Mourning the Loss of Mark Boguski



# Elected to National Academy of Sciences

**Katherine High**

**Kenneth Lange**

**David Liu**

**Olufunmilayo Olopade**



**NATIONAL  
ACADEMY  
OF SCIENCES**

# Elected to American Academy of Arts & Sciences

**Charles Rotimi**

**Sarah Tishkoff**



# 2021 ACMG David L. Rimoin Lifetime Achievement Award in Medical Genetics



**Ada Hamosh, M.D., M.P.H., FACMG**

# Director's Report Outline

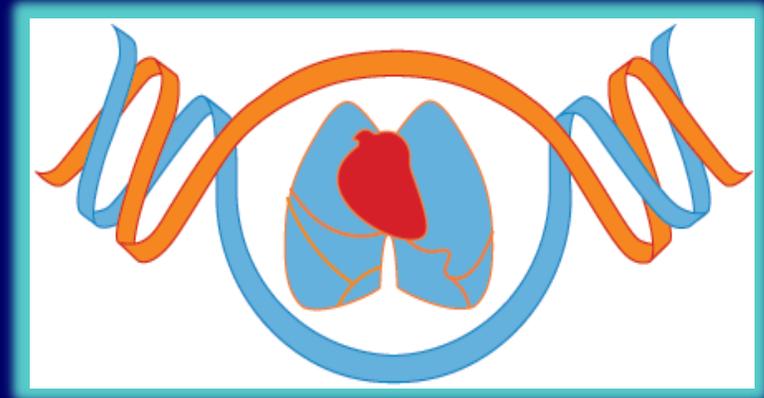
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# Genome Sequencing Program

**Final GSP datasets in AnVIL**

**Joint GSP-TOPMed Workshop**

- Data sharing
- Data analysis



# Comparative Genomics

Notice of Intent to Publish a Funding Opportunity  
Announcement for Non-Human Primate Developmental  
Genotype-Tissue Expression (NHP dGTEx) Project  
(U24 Clinical Trials Not Allowed)

Notice Number:

NOT-HG-21-027

- **Study gene-expression patterns in non-human primates and compare to humans**
- **Expected due date: Summer 2021**

# Technology Development Program



## Funding Opportunities:

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

- **NOT-HG-21-018**

### Transformative Nucleic Acid Sequencing Technology Innovation and Early Development

- **RFA-HG-21-007 (R01, also linked R21 and R43/44)**
- **First due date: June 25, 2021**

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

## Genomic Data Science Community Network

- Partnerships from diverse spectrum of institutions
- Develop curricula for undergraduate students



## Virtual Applied Data Science Training Institute

- Free eight-week training series
- AnVIL-led session on cloud computing

Data Science Approaches to Better Understand Clinical and Genomic Informatics

Virtual Applied Data Science Training  
Institute (VADSTI)

February 11 – April 30, 2021 | An 8-week data science training series in a virtual setting

# NHGRI Genomic Data Science Working Group



- Define key areas in genomics for machine learning analysis and NHGRI's role in machine learning research for both genomic medicine and genomic sciences
- Nearly 1,800 participants
- Video recordings and presentations available on workshop webpage

# Clinical Genome Resource (ClinGen)



**nature**

Perspective | Published: 10 March 2021

## Improving reporting standards for polygenic scores in risk prediction studies

Hannah Wand, Samuel A. Lambert, [Cecelia Tamburro](#), Michael A. Iacocca, Jack W. O'Sullivan, [Catherine Sillari](#), Iftikhar J. Kullo, Robb Rowley, Jacqueline S. Dron, Deanna Brockman, Eric Venner, Mark I. McCarthy, Antonis C. Antoniou, Douglas F. Easton, Robert A. Hegele, Amit V. Khera, Nilanjan Chatterjee, Charles Kooperberg, Karen Edwards, Katherine Vlessis, Kim Kinnear, John N. Danesh, Helen Parkinson, Erin M. Ramos, Megan C. Roberts, Kelly E. Ormond, Muin J. Khoury, A. Cecile J. W. Janssens, Katrina A. B. Goddard, Peter Kraft, Jaqueline A. L. MacArthur, Michael Inouye & Genevieve L. Wojcik  -Show fewer authors

*Nature* **591**, 211–219(2021) | [Cite this article](#)

**6887** Accesses | **322** Altmetric | [Metrics](#)

**Complex Disease Working Group publishes reporting standards for polygenic scores**

**Includes guidance for:**

- **Score development**
- **Describing study populations**
- **Score evaluation**
- **Clinical applications**



# Curating the Clinical Genome (CCG)

## ClinGen and DECIPHER Host Annual Conference



### Topics included:

- Polygenic risk scores across populations
- Estimating penetrance
- Non-invasive prenatal testing
- Precision oncology
- Sharing data and associated clinical phenotypes

# Clinical Sequencing Evidence-Generating Research Program



## Usual care minimal checklist

- ✓ Client-centered
- ✓ Respectful
- ✓ Culturally informed
- ✓ Empathetic
- ✓ Obtains personal and family history information
- ✓ Tailors education
- ✓ Discusses testing recommendations/option
- ✓ Provides guidance for follow-up
- ✓ Value-based decision making
- ✓ Identifies coping resources
- ✓ Promotes psychological well-being

- **Phillips et al.: affordability and value are key to precision medicine implementation**
- **Biesecker et al.: definition of “usual care” in genetic counseling**
- **ACMG 2021: challenges and strategies for reporting findings in young patients**

# Implementing Genomics in Practice (IGNITE)

**GUARDD-US assesses knowledge of *APOL1* variants on blood pressure management**

- Over 1,850 of 5,435 participants enrolled



**GUARDD-US**

Genetic testing to Understand and Address Renal Disease Disparities across the US

**ADOPT PGx investigates genotype-guided therapy on acute pain, chronic pain, and depression**

- Over 50 of 4,500 participants enrolled



**ADOPT-PGx**

A Depression and Opioid Pragmatic Trial in Pharmacogenetics

Genetics  
inMedicine

[www.nature.com/gim](http://www.nature.com/gim)

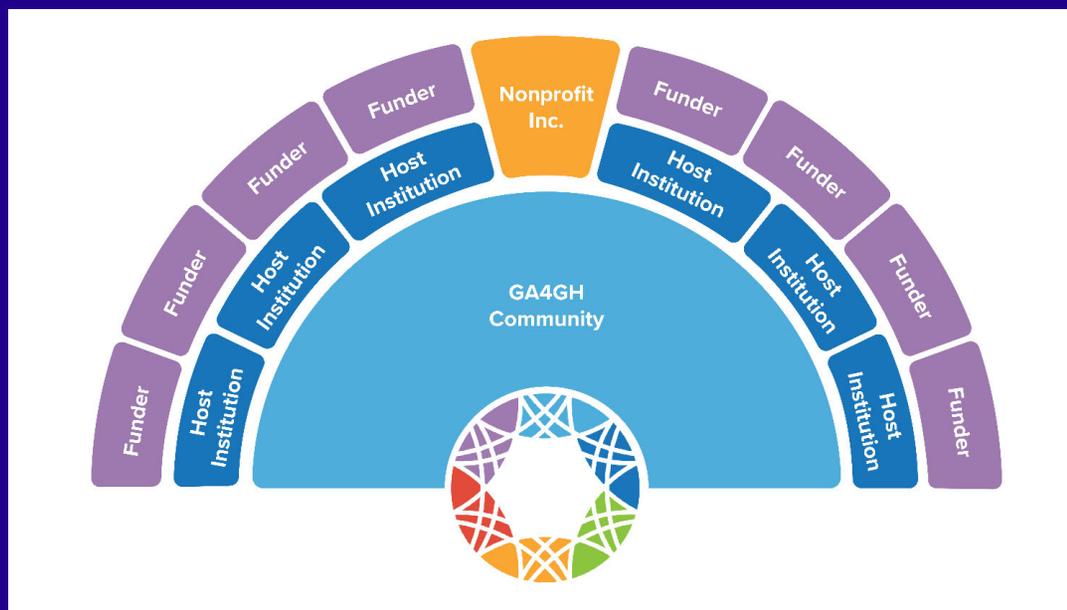


ARTICLE

Establishing the value of genomics in medicine: the IGNITE Pragmatic Trials Network

Geoffrey S. Ginsburg<sup>1</sup>✉, Larisa H. Cavallari<sup>2</sup>, Hrishikesh Chakraborty<sup>3</sup>, Rhonda M. Cooper-DeHoff<sup>2</sup>, Paul R. Dexter<sup>4</sup>, Michael T. Eadon<sup>5</sup>, Bart S. Ferket<sup>5</sup>, Carol R. Horowitz<sup>5</sup>, Julie A. Johnson<sup>2</sup>, Joseph Kannry<sup>5</sup>, Natalie Kucher<sup>7</sup>, Ebony B. Madden<sup>7</sup>, Lori A. Orlando<sup>1</sup>, Wanda Parker<sup>3</sup>, Josh Peterson<sup>8</sup>, Victoria M. Pratt<sup>9</sup>, Tejinder K. Rakhra-Burris<sup>1</sup>, Michelle A. Ramos<sup>6</sup>, Todd C. Skaar<sup>5</sup>, Nina Sperber<sup>1,10</sup>, Kady-Ann Steen-Burrell<sup>3</sup>, Sara L. Van Driest<sup>11</sup>, Deepak Voora<sup>1</sup>, Kristin Wiisanen<sup>2</sup>, Almut G. Winterstein<sup>12</sup>, Simona Volpi<sup>7</sup>, the IGNITE PTN\*

# Genomics in Healthcare: GA4GH in 2025



- Incorporated as not-for-profit “GA4GH, Inc.”
- U24 Community Resource (NHGRI, NCI, NHLBI, All of Us, ODSS)
- NIH Cloud Commons adopting GA4GH standards

# Genomic Medicine Meeting XIII

## Developing a Clinical Genomic Informatics Research Agenda

To develop a research strategy for using genomic-based clinical informatics tools and resources to improve the detection, treatment, and reporting of genetic disorders in clinical settings

### Key recommendations included:

- Incorporate an implementation component within the overall clinical informatics research framework
- Advance research to better understand the interface between human cognition and information technology
- Develop methods to identify and mitigate inherent and pervasive biases



# Training and Career Development

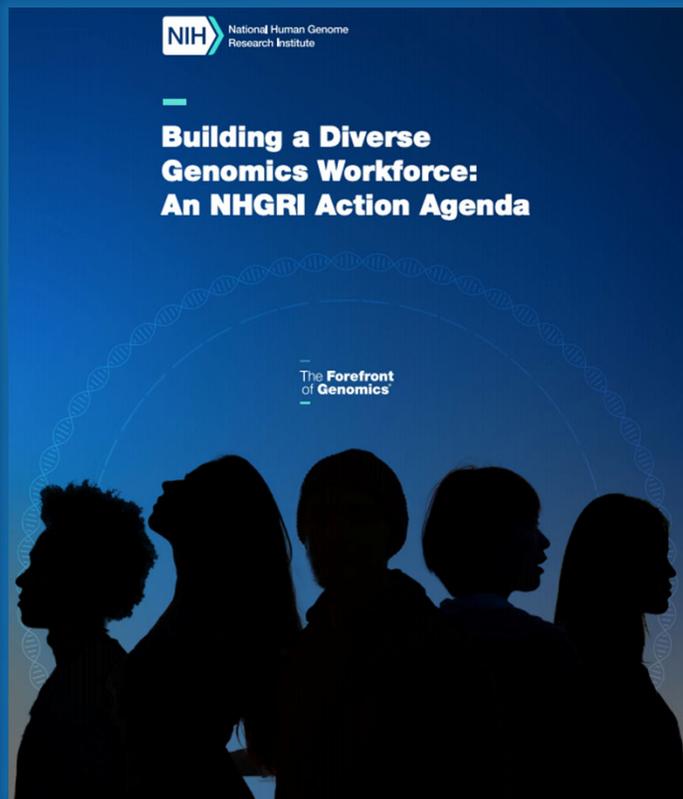
## Enhancing the Diversity of the Genomics Workforce

### New FOAs:

- **PAR-21-143**  
F99/K00 – NHGRI Predoctoral to Postdoctoral Transition Award to Promote Diversity
- **PAR-21-214**  
K18 – Short-term Mentored Research Career Enhancement Award to Promote Diversity

### FOAs Under Development:

- **R01 – Grants for New Investigators to Promote Diversity in Genomics Research**
- **R25 – Genome Research Experiences to Attract Talented Undergraduates into the Genomics Field to Promote Diversity (GREAT)**



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Article | [Open Access](#) | Published: 12 February 2021

## Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases

Shilpa Nadimpalli Kobren, Dustin Baldrige, Matt Velinder, Joel B. Krier, Kimberly LeBlanc, Cecilia Esteves, Barbara N. Pusey, Stephan Züchner, Elizabeth Blue, Hane Lee, Alden Huang, Lisa Bastarache, Anna Bican, Joy Cogan, Shruti Marwaha, Anna Alkelai, David R. Murdock, Pengfei Liu, Daniel J. Wegner, Alexander J. Paul, Undiagnosed Diseases Network, Shamil R. Sunyaev & Isaac S. Kohane 

*Genetics in Medicine* (2021) | [Cite this article](#)

- Investigated commonalities across genome-sequencing processing workflows
- Advances in structural variant detection, noncoding variant interpretation, and integration of additional biomedical data promising for solving undiagnosed cases

February 25, 2021

## Machine Learning of Patient Characteristics to Predict Admission Outcomes in the Undiagnosed Diseases Network

Hadi Amiri, PhD<sup>1,2</sup>; Isaac S. Kohane, MD, PhD<sup>1</sup>; for the Undiagnosed Diseases Network

[» Author Affiliations](#) | [Article Information](#)

*JAMA Netw Open.* 2021;4(2):e2036220. doi:10.1001/jamanetworkopen.2020.36220

- Developed and evaluated machine learning model to prioritize evaluation of patients with undiagnosed diseases by retrospective and prospective validation
- Predictive model may reduce the processing time for accepted applications by two thirds

# Human Biomolecular Atlas Program (HuBMAP)



UPDATED VIDEO TUTORIAL COMING SOON.

## ABOUT

The ASCT+B Reporter is a state-of-the-art visualization tool. The video gives an overview of the functionalities listed below. Please use the selectors below to skip to any section you'd want to specifically know about.

### Introduction

Know about the Reporter, what it is, why was it built and by who.

### Getting Started

Know more about what how to use the buttons on the toolbar.

### Report Generator

The Reporter computes various statistics from the data - which can be viewed in the Report

### Debug Log

The Debug Log lists warnings and errors that occurred during data parsing and visualization creation

# HuBMAP CCF ASCT+B REPORTER

## ASCT+B REPORTER

### INSTRUCTIONAL VIDEO

0:00 / 3:34

**Community effort to relate anatomical structures and cell types to ontologies**

**Opportunities for trainees and early career investigators:**

- **Student Genome Internship Program for Undergraduates**
- **Jump Start Program for Junior Investigators**

# Bridge to Artificial Intelligence (Bridge2AI)

## New NIH Common Fund Program



Notice of Intent to Publish a Funding Opportunity  
Announcement for Research Opportunity Announcement for  
the Data Generation Projects of the NIH Bridge to Artificial  
Intelligence (Bridge2AI) Program (OT2)

Notice Number:

NOT-RM-21-022

Notice of Intent to Publish a Funding Opportunity  
Announcement for NIH Bridge2AI Integration, Dissemination,  
and Evaluation (BRIDGE) Center (U54 Clinical Trial Not  
Allowed)

Notice Number:

NOT-RM-21-021

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- VII. NHGRI Intramural Research Program

# NHGRI Guidance for Third-Party Involvement in Extramural Research

The screenshot shows the top navigation bar of the National Human Genome Research Institute (NHGRI) website. The header includes the NIH logo, the text 'National Human Genome Research Institute', a search bar with the placeholder 'Begin your search here', and social media icons for Facebook, Twitter, and YouTube. Below the header is a secondary navigation bar with links for 'About Genomics', 'Research Funding', 'Research at NHGRI', 'Health', 'Careers & Training', 'News & Events', and 'About NHGRI'. The main content area has a breadcrumb trail: 'Home / About NHGRI / NHGRI Policies and Guidance / Third-Party Involvement in NHGRI-Supported Extramural Projects'. The title of the page is 'Third-Party Involvement in NHGRI-Supported Extramural Projects'. The main text begins with an 'Effective Date' of July 1, 2021, followed by a paragraph explaining that NHGRI extramural grantees and contract-recipients sometimes seek or receive offers of support from third parties. It states that NHGRI is open to third-party involvement as long as the primary purpose is not compromised and NIH and NHGRI policies are followed. A second paragraph notes that NHGRI reviews potential agreements between grantees and third parties to promote transparency and prevent conflicts of interest. A section titled 'Who does this apply to?' is followed by a paragraph stating that the guidance applies to NHGRI-funded extramural investigators subject to the NIH Data Sharing Policy or the NIH Genomic Data Sharing (GDS) Policy, excluding grantees or contractors funded through the Small Business Innovation Research or Small Business Technology Transfer program.

**NIH** National Human Genome Research Institute

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Home / About NHGRI / NHGRI Policies and Guidance / Third-Party Involvement in NHGRI-Supported Extramural Projects

## Third-Party Involvement in NHGRI-Supported Extramural Projects

**Effective Date:** July 1, 2021

The National Human Genome Research Institute's (NHGRI) extramural grantees and contract-recipients sometimes seek or receive offers of support from companies or other third parties (e.g., nonprofits, other government agencies) for their studies, sub-studies, and ancillary studies after the grant or contract has been awarded. This type of third-party support, or "third-party involvement", may be valuable in advancing NHGRI-supported research. In general, NHGRI is open to third-party involvement in NHGRI-supported studies and contracts, so long as the primary purpose of the studies and contracts is not compromised and National Institutes of Health (NIH) and NHGRI policies are followed.

In some instances, NHGRI reviews potential agreements between grantees and third parties in order to promote transparency regarding third-party support, prevent potential conflicts of interest, and preserve scientific objectivity. This guidance describes NHGRI's position on appropriate third-party involvement. It applies to post-award interactions with third parties; pre-award interactions are reported and reviewed by NHGRI in the application process. This guidance describes to which grantees this guidance is applicable and lays out NHGRI's process for reviewing potential agreements with third parties.

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**Who does this apply to?**

This guidance applies to NHGRI-funded extramural investigators that are subject to the **NIH Data Sharing Policy** or the **NIH Genomic Data Sharing (GDS) Policy**, excluding grantees or contractors funded through the Small Business Innovation Research or Small Business Technology Transfer program.

# NHGRI Data Sharing Expectations: Sharing Quality Metadata and Phenotypic Data

**NHGRI-funded and -supported researchers are expected to:**

- **Share metadata and phenotypic data associated with the study**
- **Use standardized data-collection protocols and survey instruments for capturing data, as appropriate**
- **Use standardized notation for metadata to enable the harmonization of datasets for secondary research analyses**



# NHGRI Communications Video Wins Journalism Award

NHGRI science writer Prabarna Ganguly wins  
DCSWA Newsbrief Award for “The Human Pangenome” video



*Credit: Massive Science/NHGRI*





## Genomics and the Media



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

May 20, 2021



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

November 4, 2021



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

May 25, 2022



**John Inglis**  
Executive Director, Cold Spring Harbor Laboratory Press; Co-founder, biorXiv, medrXiv

July 28, 2021



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

January 20, 2022



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

September 20, 2021



**Joe Palca**  
Science Correspondent, NPR

March 2022

Conversations with trailblazing science communicators

# National DNA Day 2021



**Do you know you have 10 billion different antibodies in your body?**



**Cathrine Scheepers, Ph.D.**  
March 25, 2021  
8:00 a.m. Eastern Daylight Time  
12:00 p.m. Coordinated Universal Time  
2:00 p.m. Central African Time



**Genomic Characterization and Surveillance of Microbial Threats in West Africa**



**Paul Oluniyi, Ph.D. Research Fellow**  
April 8, 2021  
11:00 a.m. Eastern Daylight Time  
3:00 p.m. Coordinated Universal Time  
4:00 p.m. West African Time



 **National DNA Day 25**

Louise M. Slaughter National DNA Day Lecture

**The Wildlife Detective:**  
A "DNA-driven" journey of koalas, cockatoos and wildlife forensics

**Rebecca Johnson, Ph.D.**  
Member of the Order of Australia

April 23, 2021 | 1:00 p.m. ET



 **Direct-to-Consumer Genetic Testing Adoptee Panel** 

Tuesday, May 4, 2021 | 6 p.m. - 7:30 p.m. ET



Kim Jacoby Morris    Damon Davis    Heewon Lee    Tim Holtan



# Inter-Society Coordinating Committee for Practitioner Education in Genomics

## 10<sup>th</sup> “In-Person” Meeting



# 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**

# Genome Sequencing on Prime-Time TV

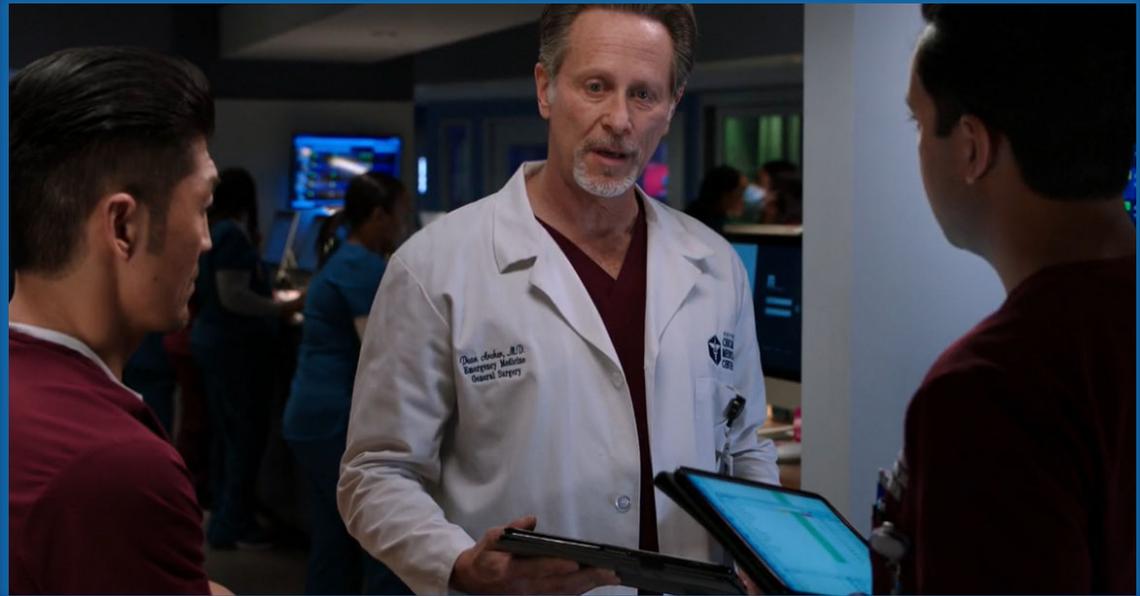
The NEW ENGLAND JOURNAL of MEDICINE

ORIGINAL ARTICLE

## Somatic Mutations in *UBA1* and Severe Adult-Onset Autoinflammatory Disease

D.B. Beck, M.A. Ferrada, K.A. Sikora, A.K. Ombrello, J.C. Collins, W. Pei, N. Balanda, D.L. Ross, D. Ospina Cardona, Z. Wu, B. Patel, K. Manthiram, E.M. Groarke, F. Gutierrez-Rodrigues, P. Hoffmann, S. Rosenzweig, S. Nakabo, L.W. Dillon, C.S. Hourigan, W.L. Tsai, S. Gupta, C. Carmona-Rivera, A.J. Asmar, L. Xu, H. Oda, W. Goodspeed, K.S. Barron, M. Nehrebecky, A. Jones, R.S. Laird, N. Deutch, D. Rowczenio, E. Rominger, K.V. Wells, C.-C.R. Lee, W. Wang, M. Trick, J. Mullikin, G. Wigerblad, S. Brooks, S. Dell'Orso, Z. Deng, J.J. Chae, A. Dulau-Florea, M.C.V. Malicdan, D. Novacic, R.A. Colbert, M.J. Kaplan, M. Gadina, S. Savic, H.J. Lachmann, M. Abu-Asab, B.D. Solomon, K. Retterer, W.A. Gahl, S.M. Burgess, I. Aksentijevich, N.S. Young, K.R. Calvo, A. Werner, D.L. Kastner, and P.C. Grayson

**NEJM 2020**



# 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 characters. 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.



# The Genomics Landscape

## Email Updates

Sign up to receive National Human Genome Research Institute (NHGRI) updates and stay informed about our latest science, research, news, upcoming events and website content.

Email Address



National Human Genome  
Research Institute

**Thanks!**



**Special Thanks!**



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