## Director's Report-Related Documents

### February 2021

<table>
<thead>
<tr>
<th>No.</th>
<th>Relevant Documents</th>
</tr>
</thead>
<tbody>
<tr>
<td>2020 NHGRI Strategic Vision</td>
<td></td>
</tr>
</tbody>
</table>
  - Establishing a 2020 Vision for Genomics  
  - Nature - 2020 NHGRI Strategic Vision  
  - Scientific American - A Vision for the Next Decade of Human Genomics Research  
  - Forbes - How Human Genome Sequencing Went From $1 Billion A Pop To Under $1,000 |
| 1   | American Journal of Medical Genetics - Genome Research Institute Identifies Vision for the Future  
  - Genetic Engineering and Biotechnology News - NHGRI's Strategic Vision Builds Up Diversity, Breaks Down Barriers  
  - Genetics Society Podcast - The Past, Present and Future of the Human Genome Project  
  - The Future of Genomics: 10 Bold Predictions Video  
  - Bold Predictions for Human Genomics by 2030 Seminar Series |

[genome.gov/DirectorsReport](genome.gov/DirectorsReport)
Open Session Presentations

Presentations:

Final NIH Policy on Data Management and Sharing
  Carrie Wolinetz

Building a Diverse Genomics Workforce: An NHGRI Action Agenda
  Vence Bonham
Open Session Presentations

Concept Clearances:

Genome Research Experiences to Attract Talented Undergraduates into the Genomics Field to Promote Diversity (GREAT Program)

Tina Gatlin

Grants for New Investigators to Promote Diversity in Genomics Research

Jyoti Dayal
Open Session Presentations

Concept Clearances:

Non-Human Primate dGTEx
Jennifer Troyer

Computational Genomics and Data Science
Daniel Gilchrist

Molecular Phenotypes of Null Alleles in Cells Pilot Project
Adam Felsenfeld
Open Session Presentations

Presentation:

Phenotypes and Exposures (PhenX) Toolkit

Erin Ramos
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
Director’s Report Outline

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2020 NHGRI Strategic Vision

Strategic vision for improving human health at The Forefront of Genomics

Perspective

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 the clinical care. In the future, the digitization and advances in genomics, biopharmaceuticals, biologic imaging, and clinical application (among others) will lead to a widespread integration of genomics into all areas of medical and public health practice, and an increasing relevance of genomics to every level of research. As a result, the National Human Genome Research Institute has recently completed a multi-year process of strategic engagement to identify future research priorities and opportunities in biomedical and human genomics, with an emphasis on health applications. Here we describe the highest priority elements envisioned for the cutting-edge human genomics going forward — that is, at The Forefront of Genomics.
2020 NHGRI Strategic Vision

Outreach and Media Coverage

A Vision for the Next Decade of Human Genomics Research

An article in Nature lays out bold predictions for a field whose extraordinary achievements are just the beginning of what could be possible.

By Eric D. Green on October 28, 2020

READ THIS NEXT

SPONSORED
Your Next House May Pop Out of a 3D Printer

NATURAL DISASTERS
Miniature Sandstorms Reveal Cause of Deadly Unexplained Flood That Devastated Hydroelectric Dam

PUBLIC HEALTH
Coronavirus News Roundup, February 6 – February 12
2020 NHGRI Strategic Vision

Box 5

Bold predictions for human genomics by 2030

Some of the most impressive genomics achievements, when viewed in retrospect, could hardly have been imagined ten years earlier. Here are ten bold predictions for human genomics that might come true by 2030. Although most are unlikely to be fully attained, achieving one or more of these would require individuals to strive for something that currently seems out of reach. These predictions were crafted to be both inspirational and aspirational in nature, provoking discussions about what might be possible at the Forefront of Genomics in the coming decade.

1. Generating and analyzing a complete human genome sequence will be routine for any research laboratory, becoming as straightforward as carrying out a DNA purification.
2. 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.
3. The general features of the epigenetic landscape and transcriptional output will be routinely incorporated into predictive models of the effect of genotype on phenotype.
4. Research in human genomics will have moved beyond population descriptors based on historic social constructs such as race.
5. Studies that involve analyses of genome sequences and associated phenotypic information for millions of human participants will be regularly featured at school science fairs.
6. The regular use of genomic information will have transitioned from boutique to mainstream in all clinical settings, making genomic testing as routine as complete blood counts.
7. The clinical relevance of all encountered genomics variants will be readily predictable, rendering the diagnostic designation "variant of uncertain significance (VUS)" obsolete.
8. An individual's complete genome sequence along with informative annotations will, if desired, be securely and readily accessible on their smartphone.
9. Individuals from ancestrally diverse backgrounds will benefit equitably from advances in human genomics.
10. Breakthrough discoveries will lead to curative therapies involving genomic modifications for dozens of genetic diseases.
Bold Predictions for Human Genomics by 2030

NHGRI Seminar Series

- **Bold Prediction #1: February 1**
  Evan Eichler, University of Washington
  Karen Miga, UC, Santa Cruz

- **Bold Prediction #2: March 8**
  Nancy Cox, Vanderbilt University
  Neville Sanjana, NY Genome Center
Departure of Chief, Education and Community Involvement Branch

Carla Easter, Ph.D.
New Extramural Program Director

Joannella Morales, Ph.D.
20th Anniversary of Publications Reporting Draft Human Genome Sequence
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Leadership Changes at Centers for Disease Control (CDC) and Food and Drug Administration (FDA)

New Director, CDC
Rochelle Walensky M.D., M.P.H.

Acting Commissioner, FDA
Janet Woodcock, M.D.
VIPs Visit NIH

Vice President Kamala Harris

President Joe Biden

First Lady Dr. Jill Biden
2020 Federal Employee of the Year

FEDERAL EMPLOYEE OF THE YEAR

ANTHONY S. FAUCI, M.D.
Director, National Institute of Allergy and Infectious Diseases
National Institutes of Health
Judith Greenberg Retires as Deputy Director, National Institute of General Medical Sciences

Judith Greenberg, M.D., Ph.D.
<table>
<thead>
<tr>
<th></th>
<th>Fiscal Year 2020 Labor-HHS Appropriation</th>
<th>Fiscal Year 2021 Labor-HHS Appropriation</th>
<th>$ Increase</th>
<th>% Increase</th>
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<tbody>
<tr>
<td>NIH</td>
<td>$41.68 B</td>
<td>$42.93 B</td>
<td>$1.25 B</td>
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<td>NHGRI</td>
<td>$606.35 M</td>
<td>$615.78 M</td>
<td>$9.43 M</td>
<td>1.56%</td>
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</table>
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Mourning the Loss of Marvin Frazier
Mourning the Loss of Gertjan van Ommen
2020 American Society of Human Genetics Awards

William Allan Awardee
Mary-Claire King, PhD
University of Washington

Education Awardee
Kenneth Lange, PhD
University of California
Los Angeles

Early-Career Awardee
Benjamin Neale, PhD
Massachusetts General Hospital
Elected to National Academy of Medicine

Judy Cho  Alondra Nelson
Wendy Chung  Aviv Regev
Levi Garraway  Pardis Sabeti
Joel Hirschhorn  Louis Staudt
David Liu  Hannah Valantine
Consuelo Hopkins Wilkins
Elected to AAAS

Lisa Brooks  Pui-Yan Kwok
R. Alta Charo  Matthew Meyerson
Dana Crawford  William Murphy
Ronald Davis  Len Pennacchio
International Common Disease Alliance (ICDA)
Science 2020 Breakthrough of the Year Runners Up

First CRISPR cures?

Scientists speak up for diversity
DNA assembly and amplification
Single cell gene expression
Spatial gene expression
Digital genome engineering
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Human Genome Reference Program

TOWARDS A COMPLETE REFERENCE OF HUMAN GENOME DIVERSITY

- Generate ≥350 high-quality reference genome sequences
- Year 1 data released
- HGRP and T2T joint meeting (September 2020)
- Human pangenome video
Comparative Genomics

Notice of NIH Participation in the National Science Foundation Enabling Discovery through GEnomics (EDGE) Program

Notice Number:
NOT-HG-21-014

- Enabling Discovery Through GEnomics (EDGE)
  Functional Genomic Tools Track
  Complex Multigenic Traits Track

- Applications due: March 16, 2021
Technology Development Program

Funding Opportunities

Notice of Special Interest (NOSI): Advancing Genomic Technology Development for Research and Clinical Applications
NOT-HG-21-018
Standard due dates start February 5, 2021

Novel Synthetic Nucleic Acid Technology Development
RFA-HG-20-014 (R01, also linked R21 and R43/44)
Applications due March 9, 2021

Notice of Intent to Publish: Transformative Nucleic Acid Sequencing Technology Innovation and Early Development
NOT-HG-21-016
Technology Development Program
Advanced Genomic Technology Development Meetings

- New colloquia (Fall 2020)
- Virtual annual meeting
  May 25-27, 2021
  Northeastern University
High-Throughput Molecular and Cellular Phenotyping Opportunity

Notice of Special Interest (NOSI): High-throughput Molecular and Cellular Phenotyping

Notice Number:
NOT-HG-21-004

Key Dates

<table>
<thead>
<tr>
<th>Release Date:</th>
<th>December 3, 2020</th>
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<tbody>
<tr>
<td>First Available Due Date:</td>
<td>February 05, 2021</td>
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<tr>
<td>Expiration Date:</td>
<td>January 10, 2024</td>
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## ENCODE 2020 Research Applications & Users Meeting

Encode 2020 Research Applications & Users meeting is virtual  
September 30th - October 2nd

### Meeting materials available on the ENCODE portal

<table>
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<tr>
<th>No.</th>
<th>Presentation Title</th>
<th>Speaker(s)</th>
<th>Institute</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Aravinda Chakravati: Sequence-based studies of regulatory control</td>
<td>National Human Genome Research Institute</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>Stephanie Morris: ENCODE: The Encyclopedia of DNA Elements</td>
<td>National Human Genome Research Institute</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>Paul Flicek: Comparative regulatory genomics approaches to</td>
<td>National Human Genome Research Institute</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>John Stamatoyannopoulos: High-resolution maps of regulatory DNA:</td>
<td>National Human Genome Research Institute</td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>Neva Durand: Juicebox Hands-on Session - September 30, 2020</td>
<td>National Human Genome Research Institute</td>
<td></td>
</tr>
</tbody>
</table>
Algorithm development
Data and resource needs
ELSI
Clinical applications
Clinical Genome Resource (ClinGen)

ClinGen Validity Curations Integrated into Open Targets Platform

**Open Targets Platform**

*Introducing ClinGen's Gene Validity Curations*

Learn how the Open Targets Platform integrates and scores ClinGen's Gene-Disease Validity curation data to enhance rare disease associations.
Clinical Sequencing Evidence-generating Research (CSER) Program

- “Variant bake-off 2.0” of ACMG secondary findings genes
- Concordance rate of 84%
- CSER sites adapted to COVID-19 challenges through telemedicine
International 100K+ Cohort Consortium (IHCC)

- Exploring the Role of Genetically Determined BMI in Infancy, Childhood, and Early Adulthood on Colorectal Cancer Development in Later Life
- High-throughput Metabolomic Biomarker Measures in Diverse Ancestries
- Opioid Cohort Consortium (OPICO) to Investigate the Effects of Regular Opioid Use on Mortality and on Cancer Development
- Global Mental Health Impact of the COVID-19 Pandemic
- Novel coronavirus host susceptibility study in South Africa (COVIgen-SA)
Ethical, Legal, and Social Implications (ELSI) Research Program
Launch of ELSIhub

Curated by the Center for ELSI Resources and Analysis (CERA)

- ELSI publications database
- Scholar directory
- Research tools database
- Policy resources
- ELSI funding opportunities
Mentored Research Experiences for Genetic Counselors (R25)  
PAR-21-074

Research Experience in Genomic Research for Data Scientists (R25)  
PAR-21-075

- Release date:  
  December 9, 2020

- Applications due:  
  May 25, 2021

FOAs Under Development:

- F99/K00 – NHGRI Predoctoral to Postdoctoral Transition Award to Promote Diversity
- K18 – Short-term Mentored Research Career Enhancement Award to Promote Diversity
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Genotype-Tissue Expression (GTEx) project

- September 2020: GTEx Version 8 data release
Library of Integrated Network-based Cellular Signatures (LINCS)

LINCS Virtual Symposium
LINCS Perturbation-Response Community Resources: Science, Utility, Challenges

Video recordings now available!
November 19-20, 2020

>700 attendees
Human Heredity and Health in Africa (H3Africa)

Growing Publication Record

- 440+ publications to date

**High-depth African genomes inform human migration and health**


*Show fewer authors*

*Nature* 586, 741–748 (2020) | Cite this article

27k Accesses | 2 Citations | 688 Altmetric | Metrics
Human Heredity and Health in Africa (H3Africa)

Continued International Collaborations

- Virtual Consortium Meeting: >150 attendees

- ClinGen and H3Africa Rare Disease Working Group
4D Nucleome (4DN)

30 Awards:
- Chromatin dynamics and function
- Data integration, modeling, and visualization
- Nuclear organization in human health and disease
- New investigator projects in human health and disease
- Data center and organizational hub

Phase II kickoff meeting in December 2020
Welcome to the All of Us Research Hub

The All of Us Research Program, part of the National Institutes of Health, is building one of the largest biomedical data resources of its kind. The All of Us Research Hub stores health data from a diverse group of participants from across the United States.

Approved researchers can access All of Us data and tools to conduct studies to help improve our understanding of health.

APPLY FOR ACCESS

EXPLORE THE NEW COVID-19 PARTICIPANT EXPERIENCE SURVEY
EXPLORE NEW DATA AVAILABLE IN THE DATA BROWSER
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Human Pangeneome Video

The Human Pangeneome
3.9K views • 2 months ago

National Human Genome Research Institute

In 2003, biologists created the first ever human genome sequence. Reference ...
Genome: Unlocking Life’s Code Exhibition’s Return to the Smithsonian
Today, NHGRI is celebrating Family Health History Day!

We encourage you to learn more about your family health history... by talking to your family! Swipe to learn more about how to talk to your family about their health.

FAMILY HEALTH
UNDERSTANDING FAMILY HEALTH HISTORY
New ISCC-PEG Scholars Program

A Genomics Education Opportunity

Samantha Bailey, Pharm.D., Ph.D.

Rene Begay, M.P.H.

Kelsey Ellis, M.S.

Katherine Robinson, Pharm.D.
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New Genomic Science and Health Equity Fellowship Program

- Use genetic, genomic, and pharmacogenomic approaches to advance minority health and health equity
- Research methodology and medical product development processes that facilitate delivery of drugs, biologics, and devices
- First fellow will start Summer/Fall 2021
Retirement of NHGRI Intramural Investigator

Alec Wilson, Ph.D.
New NHGRI Intramural Investigator

Neil Hanchard, M.D., Ph.D.
Royal Swedish Academy of Sciences’ Crafoord Prize

Dan Kastner, M.D., Ph.D.
NHGRI Scientific Director Search

- Dan Kastner will step down as NHGRI Scientific Director
- Search for the next NHGRI Scientific Director begins in mid-March
NHGRI Director on Twitter

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.

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Thanks!

Special Thanks!