



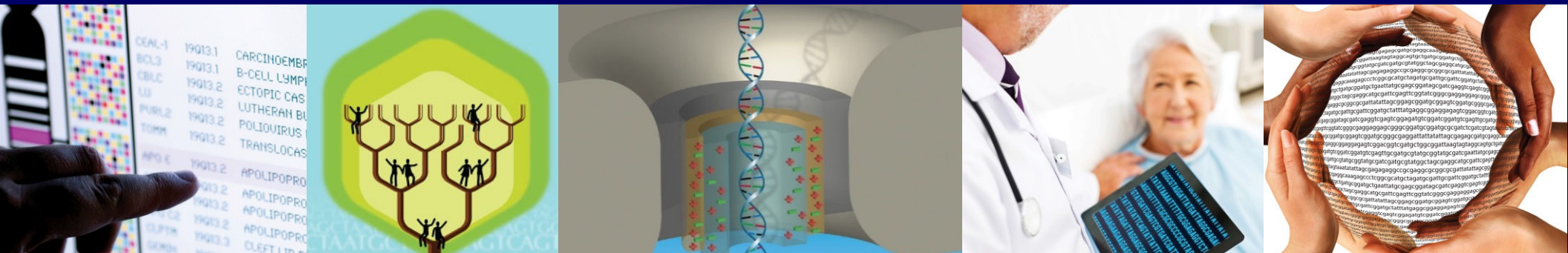
National Human Genome
Research Institute


—
The **Forefront**
of **Genomics**
—

DIRECTOR'S REPORT

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

September 2020




National Human Genome Research Institute

Begin your search here


f
t
d


About Genomics
Research Funding
Research at NHGRI
Health
Careers & Training
News & Events
About NHGRI

Home / News & Events / Calendar of Events / 91st Meeting of the National Advisory Council for Human Genome Research / Director's Report-Related Documents: September 2020

Director's Report-Related Documents

September 2020

Director's Report


Director's Report


No.	Relevant Documents
1	New ASHG-NHGRI Fellows
	Genetics and Public Policy Fellow
	Genetics and Education Fellow
2	30 th Anniversary of Human Genome Project Launch
	Interviews with Genomics Leaders
	Video Testimonials - TBD
3	2020 NHGRI Strategic Vision

genome.gov/DirectorsReport

Document #



Open Session Presentations

Presentation:

Foreign Interference on Research Integrity: NIH Experience and Advice

Michael Lauer

Concept Clearances:

Predoctoral to Postdoctoral Transition Award (F99/K00) to Promote Diversity

Luis Cubano

Short-Term Research Career Enhancement Award for Established Investigators (K18) to Promote Diversity

Luis Cubano

Open Session Presentations

Presentations:

Perspectives in Comparative Genomics and Evolution

Elinor Karlsson

ELSI Research Activities across NIH

Lawrence Brody

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

- 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**

New Deputy Director, NHGRI Division of Genomic Medicine



Erin M. Ramos, Ph.D., M.P.H.

New Extramural Program Director



Rene Sterling, Ph.D., M.H.A.

New ASHG-NHGRI Fellows

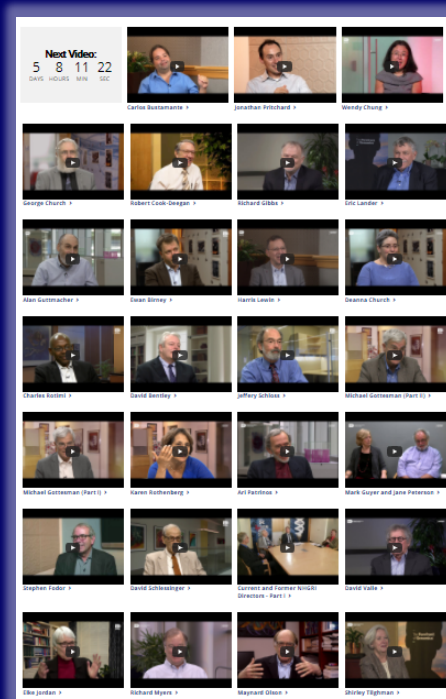


Emma Alme, Ph.D.
Genetics & Public Policy Fellow



Tiffany Rolle, Ph.D.
Genetics & Education Fellow

30th Anniversary of Human Genome Project Launch



2020 NHGRI Strategic Vision

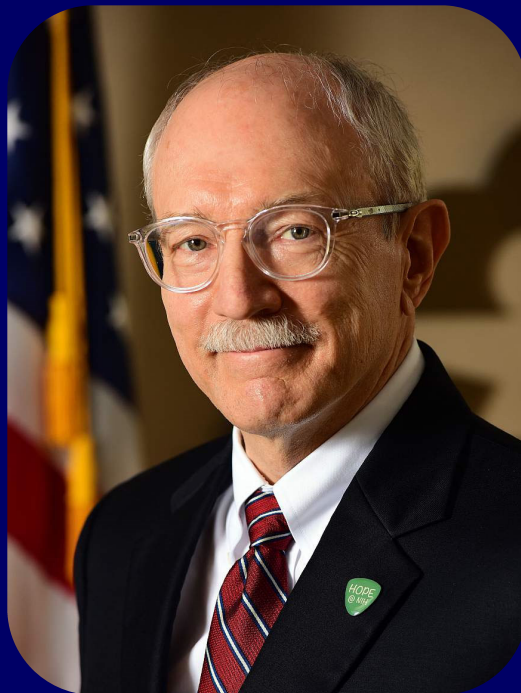


**Strategic Vision for Improving Human Health
at *The Forefront of Genomics***

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

New Director, National Institute of Environmental Health Sciences



**Rick Woychik,
M.D., Ph.D.**



New Director, National Institute of Nursing Research



**Shannon Zenk,
Ph.D., M.P.H., R.N., F.A.A.N.**

New Director, National Eye Institute



**Michael Chiang,
M.D.**



New Director, National Institute of Arthritis and Musculoskeletal and Skin Diseases



**Lindsey Criswell,
M.D., M.P.H., D.Sc.**



New Director, National Institute of Dental and Craniofacial Research



**Rena D'Souza,
D.D.S., M.S., Ph.D.**



Hannah Valentine Departs as Chief Officer for Scientific Workforce Diversity



**Hannah Valentine,
M.D.**



**Marie Bernard,
M.D.**

PUBLICATIONS OF THE CANCER GENOME ATLAS TCGA

The chart displays the following categories and publications:

- CELL OF ORIGIN**
 - Thymoma / 2018-02-17
 - TCGA Sigmoid / 2018-04-05
 - Gastric / 2018-04-05
 - Flagship / 2018-04-05
 - Stomach Pathways / 2018-04-05
 - Gynecologic & Breast / 2018-04-05
 - Gastrointestinal Tract / 2018-04-09
 - Squamous Cell / 2018-04-09
 - Esophageal Cancer / 2018-04-09
 - Flagship / 2018-04-05
 - Genitourinary / 2018-04-05
 - Driver Genes & Mutations / 2018-04-05
 - Driver Genes / 2018-04-05
 - Immune Landscape / 2018-04-05
 - Tumor-Infiltrating Lymphocytes / 2018-04-05
 - Enhancer Expression / 2018-04-05
 - EPIC1 In RNA / 2018-04-05
 - miRNA Regulation / 2018-04-05
 - Splice Site Creation / 2018-03-28
 - Mutation Calling of Tumor Exomes / 2018-04-03
 - Clinical Data Resource / 2018-08-13
 - Alternative Splicing / 2018-08-13
 - Flagship / 2018-04-05
 - MYC Pathway / 2018-04-03
 - Ras Pathway / 2018-04-03
 - DNA Damage Repair / 2018-04-03
 - Metabolic Pathways / 2018-04-03
 - Ubiquitin Pathway / 2018-04-03
 - Splice Factors / 2018-04-03
 - TGF- β Superfamily / 2018-10-24
 - Hippo Signaling / 2018-10-31
 - Testicular Germ Cell / 2018-06-12
 - ATAC-seq / 2018-10-26
 - Mesothelioma / 2018-12-01
- PAN-CANCER ATLAS**
 - TCGA Sigmoid / 2018-04-05
 - Gastric / 2018-04-05
 - Flagship / 2018-04-05
 - Stomach Pathways / 2018-04-05
 - Gynecologic & Breast / 2018-04-05
 - Gastrointestinal Tract / 2018-04-09
 - Squamous Cell / 2018-04-09
 - Esophageal Cancer / 2018-04-09
 - Flagship / 2018-04-05
 - Genitourinary / 2018-04-05
 - Driver Genes & Mutations / 2018-04-05
 - Driver Genes / 2018-04-05
 - Immune Landscape / 2018-04-05
 - Tumor-Infiltrating Lymphocytes / 2018-04-05
 - Enhancer Expression / 2018-04-05
 - EPIC1 In RNA / 2018-04-05
 - miRNA Regulation / 2018-04-05
 - Splice Site Creation / 2018-03-28
 - Mutation Calling of Tumor Exomes / 2018-04-03
 - Clinical Data Resource / 2018-08-13
 - Alternative Splicing / 2018-08-13
 - Flagship / 2018-04-05
 - MYC Pathway / 2018-04-03
 - Ras Pathway / 2018-04-03
 - DNA Damage Repair / 2018-04-03
 - Metabolic Pathways / 2018-04-03
 - Ubiquitin Pathway / 2018-04-03
 - Splice Factors / 2018-04-03
 - TGF- β Superfamily / 2018-10-24
 - Hippo Signaling / 2018-10-31
 - Testicular Germ Cell / 2018-06-12
 - ATAC-seq / 2018-10-26
 - Mesothelioma / 2018-12-01
- ONCOGENIC PROCESSES**
 - TCGA Sigmoid / 2018-04-05
 - Gastric / 2018-04-05
 - Flagship / 2018-04-05
 - Stomach Pathways / 2018-04-05
 - Gynecologic & Breast / 2018-04-05
 - Gastrointestinal Tract / 2018-04-09
 - Squamous Cell / 2018-04-09
 - Esophageal Cancer / 2018-04-09
 - Flagship / 2018-04-05
 - Genitourinary / 2018-04-05
 - Driver Genes & Mutations / 2018-04-05
 - Driver Genes / 2018-04-05
 - Immune Landscape / 2018-04-05
 - Tumor-Infiltrating Lymphocytes / 2018-04-05
 - Enhancer Expression / 2018-04-05
 - EPIC1 In RNA / 2018-04-05
 - miRNA Regulation / 2018-04-05
 - Splice Site Creation / 2018-03-28
 - Mutation Calling of Tumor Exomes / 2018-04-03
 - Clinical Data Resource / 2018-08-13
 - Alternative Splicing / 2018-08-13
 - Flagship / 2018-04-05
 - MYC Pathway / 2018-04-03
 - Ras Pathway / 2018-04-03
 - DNA Damage Repair / 2018-04-03
 - Metabolic Pathways / 2018-04-03
 - Ubiquitin Pathway / 2018-04-03
 - Splice Factors / 2018-04-03
 - TGF- β Superfamily / 2018-10-24
 - Hippo Signaling / 2018-10-31
 - Testicular Germ Cell / 2018-06-12
 - ATAC-seq / 2018-10-26
 - Mesothelioma / 2018-12-01
- ONCOGENIC SIGNALING**
 - TCGA Sigmoid / 2018-04-05
 - Gastric / 2018-04-05
 - Flagship / 2018-04-05
 - Stomach Pathways / 2018-04-05
 - Gynecologic & Breast / 2018-04-05
 - Gastrointestinal Tract / 2018-04-09
 - Squamous Cell / 2018-04-09
 - Esophageal Cancer / 2018-04-09
 - Flagship / 2018-04-05
 - Genitourinary / 2018-04-05
 - Driver Genes & Mutations / 2018-04-05
 - Driver Genes / 2018-04-05
 - Immune Landscape / 2018-04-05
 - Tumor-Infiltrating Lymphocytes / 2018-04-05
 - Enhancer Expression / 2018-04-05
 - EPIC1 In RNA / 2018-04-05
 - miRNA Regulation / 2018-04-05
 - Splice Site Creation / 2018-03-28
 - Mutation Calling of Tumor Exomes / 2018-04-03
 - Clinical Data Resource / 2018-08-13
 - Alternative Splicing / 2018-08-13
 - Flagship / 2018-04-05
 - MYC Pathway / 2018-04-03
 - Ras Pathway / 2018-04-03
 - DNA Damage Repair / 2018-04-03
 - Metabolic Pathways / 2018-04-03
 - Ubiquitin Pathway / 2018-04-03
 - Splice Factors / 2018-04-03
 - TGF- β Superfamily / 2018-10-24
 - Hippo Signaling / 2018-10-31
 - Testicular Germ Cell / 2018-06-12
 - ATAC-seq / 2018-10-26
 - Mesothelioma / 2018-12-01

Years shown: 2006, 2007, 2008, 2009, 2010, 2011, 2012, 2013, 2014, 2015, 2016, 2017, 2018.

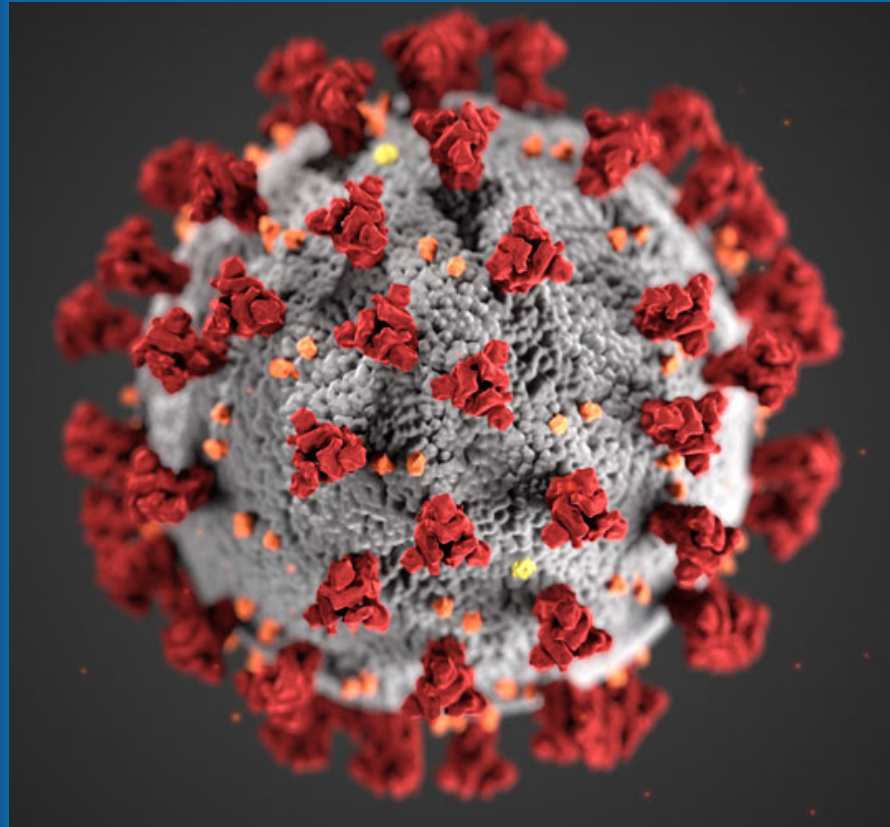
Templeton Prize 2020



Francis Collins, M.D., Ph.D.



NIH COVID-19 Information Resources



Fiscal Year 2021 Appropriations

	Fiscal Year 2020 Labor-HHS Appropriation	House Proposed Fiscal Year 2021 Labor-HHS Appropriation	\$ Increase	% Increase
NIH	~\$41.7 B	~\$42 B	~\$275 M	~0.65%
NHGRI	~\$606 M	~\$612 M	~\$6 M	~1.0%

House appropriations bill: Additional \$5 billion of emergency funds to be spent by Fiscal Year 2025 (providing each institute/center a ~7% increase for Fiscal Year 2021)

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

Mourning the Loss of Beverly Paigen



ASHG Podcast Series: *Genetically Speaking*



Host: Chris Gunter, PhD

Senior Advisor to the Director, NHGRI



Wendy Chung, MD, PhD

Columbia University, Irving Medical Center



Saumya Sisoudiya

Graduate Student at Baylor College of Medicine



Tiffany Oliver, PhD

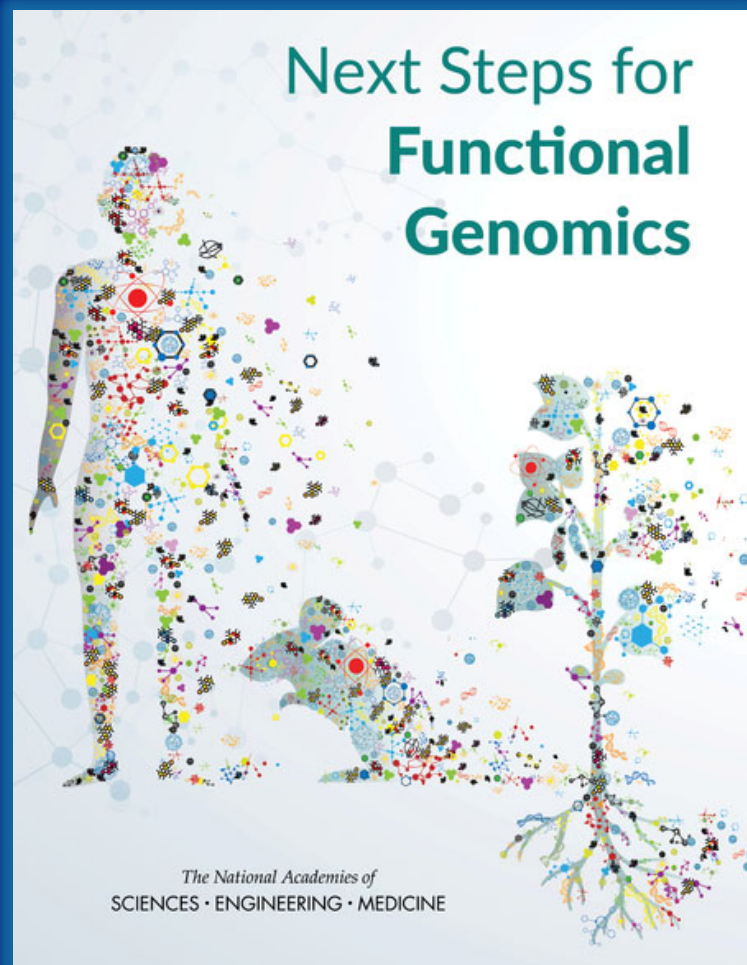
Associate Professor, Spelman College



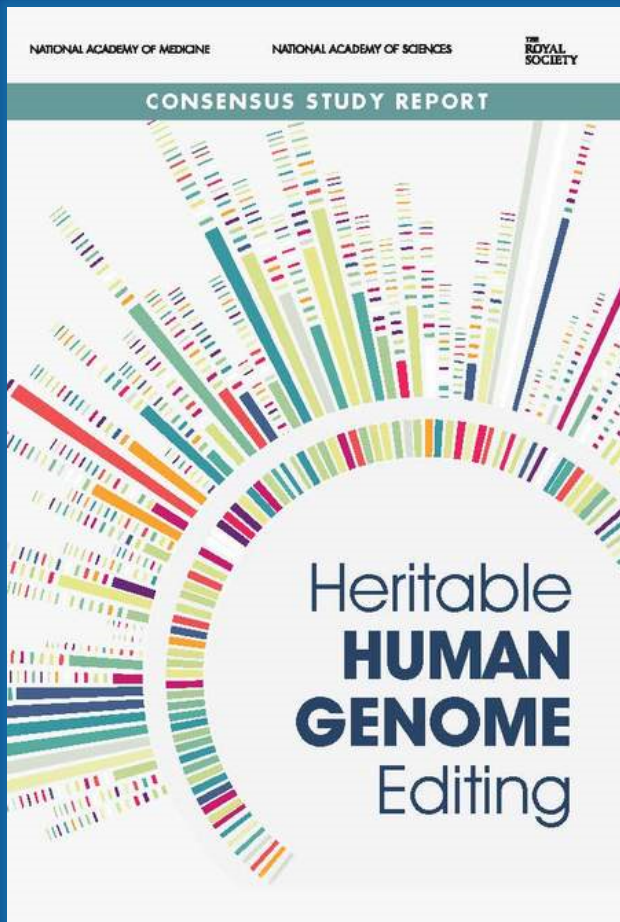
Kyle Brothers, MD, PhD

University of Louisville (PPSI, Co-chair)

Next Steps for Functional Genomics NASEM Report



Heritable Human Genome Editing Report



International Commission on the Clinical Use of Human Germline Genome Editing

SHARE [f](#) [t](#) [in](#) [✉](#)



Release of Heritable Human Genome Editing Report

The International Commission on the Clinical Use of Human Germline Genome Editing released its report, *Heritable Human Genome Editing*, at a public webinar on Thursday, September 3, 2020 at 10:15 a.m. EDT.

FNIH Lurie Prize in Biomedical Sciences 2020 Winner



Aviv Regev, Ph.D.



20th Anniversary of White House Event Announcing Draft Human Genome Sequence



Bill Clinton and Francis Collins at the White House.

The completion of the first draft sequence of the entire human genome was announced 20 years ago on June 26, 2000.

“

We couldn't be doing the biology that is possible today without the foundation of the human sequence and the revolutionary technology we created.

Jane Peterson, Ph.D.

The Human Genome Project showed that creative, impactful science can take many forms.

Eric Lander, Ph.D.

The completely free, total availability of the draft sequence has catalyzed research in ways I could only dream of in 2000.

Robert Waterston, M.D., Ph.D.

Seeing the Human Genome Project change biology and our thinking more than ever has been greatly satisfying to watch.

Elke Jordan, Ph.D.

We had developed the tools and the infrastructure to have completed a draft of the human genome in barely more than a decade!

Richard Wilson, Ph.D.

It was an absolute game-changer, like the A-Bomb or the Moonshot, for the ages.

George Weinstock, Ph.D.

The attention given to the completion of the genome draft was a worldwide phenomenon.

Jeffery Schloss, Ph.D.

As the endless stream of As, Cs, Gs and Ts cascaded across my computer screen, tears welled up in my eyes.

David Haussler, Ph.D.

The high praise and high expectations for the positive impact of the human genome on medicine established that day set a high bar

Mark Adams, Ph.D.

I was profoundly affected by the sense of crossing a bridge into new territory. We had a sense that this was real history.

Francis Collins, M.D., Ph.D.

”

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

Technology Development Program

Funding Opportunity Announcements

Novel Genomic Technology Development

- R01 (PAR-18-777) & linked R21, R43/R44
- Due date: Oct 2, 2020

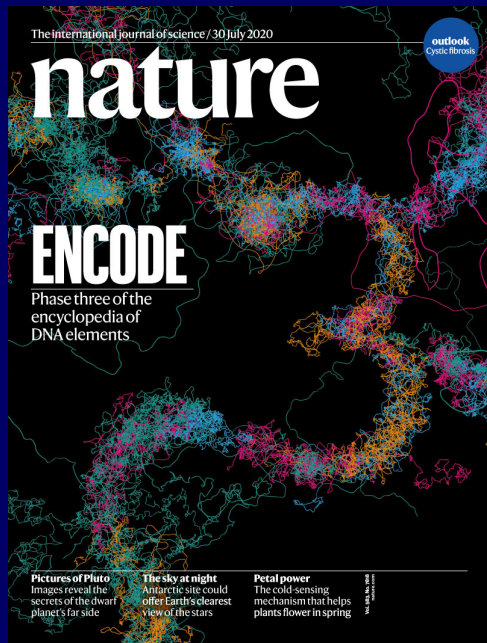
Novel Synthetic Nucleic Acid Technology Development

- R01 (RFA-HG-20-014) & linked R21, R43/R44
- Due date: Feb 1, 2021

Virtual Advanced Genomic Technology Development Meeting 2020



ENCyclopedia of DNA Elements (ENCODE)



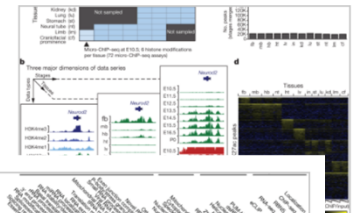
Expanded encyclopaedias of DNA elements in the human and mouse

Perspectives on ENCODE

The ENCODE Project Consortium, Michael P. Snyder , Thomas R. Gingeras, Jill E.

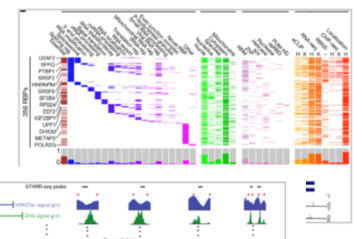
An atlas of dynamic chromatin landscapes in mouse fetal development

Analysis of chromatin state and accessibility in mouse tissues from twelve sites and eight developmental stages provides a comprehensive view of chromatin



A large-scale binding and functional map of human RNA-binding proteins

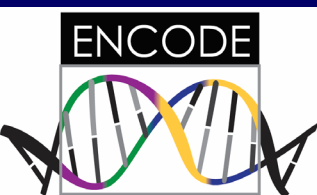
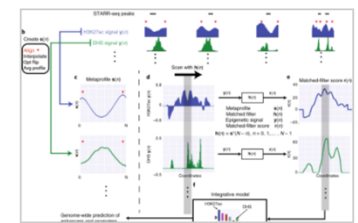
A combination of five assays is used to produce a catalogue of RNA elements to which RNA-binding proteins bind in human cells.



Supervised enhancer prediction with epigenetic pattern recognition and targeted validation

Supervised machine-learning models trained using *Drosophila* epigenetic and STARR-seq data can be transferred to predict mouse and human enhancers.

Anurag Sethi, Mengting Gu ... Mark Gerstein



ENCyclopedia of DNA Elements (ENCODE)



National Human Genome
Research Institute

ENCODE 2020 | Research Applications & Users Meeting

Encode 2020 Research Applications & Users meeting is virtual

September 30th - October 2nd

Workshops:

- **ENCODE Portal**
- **Navigating the ENCODE Encyclopedia with SCREEN**
- **An interactive introduction to Factorbook**

Impact of Genomic Variation on Function (IGVF) Program

- **Five new funding opportunities:**

Systematic Characterization of Genomic Variation on Genome Function and Phenotype (UM1 Clinical Trial Not Allowed)

Defining Genomic Influence on Gene Network Regulation (U01 Clinical Trial Not Allowed)

Single-cell Profiling of Regulatory Element and Gene Activity in Relationship to Genome Function (UM1 Clinical Trial Not Allowed)

Genomic Variation and Function Data and Administrative Coordinating Center (U24 Clinical Trial Not Allowed)

Developing Predictive Models of the Impact of Genomic Variation on Function (U01 Clinical Trial Not Allowed)

- **Applications due November 4, 2020**

Centers of Excellence in Genomic Science (CEGS)

Emerging Centers of Excellence

- **1st Notice of Change for Fiscal Year 2020**
 - **Opportunity for institutions that are not previous CEGS grantees**
-
- **2nd Notice of Change for Fiscal Year 2021**
 - **Additional Receipt Date**
 - **Emphasize interest in institutions that are not previous CEGS grantees**



National Human Genome Research Institute
@genome_gov

Big news for researchers interested in funding opportunities for Centers Of Excellence in Genomic Studies (CEGS)! @genome_gov seeks applications from institutions that have not previously been funded under the CEGS program. bit.ly/2FCnSF9

11:33 AM · Jan 13, 2020 · Sprout Social

New receipt date for Emerging Centers:
November 20, 2020
Standard CEGS receipt dates unaffected:
May 20, 2021

Developmental Genotype-Tissue Expression (dGTEx)

Laboratory, Data Analysis, and Coordinating Center (LDACC) for the Developmental Genotype-Tissue Expression Project (U24 Clinical Trial Not Allowed)

RFA-HG-20-039

- **Applications due on November 13, 2020**

Pediatric Biospecimen Procurement Center (BPC) Supporting the Developmental Gene Expression (dGTEx) Project (U24 Clinical Trial Not Allowed)

RFA-HD-21-008

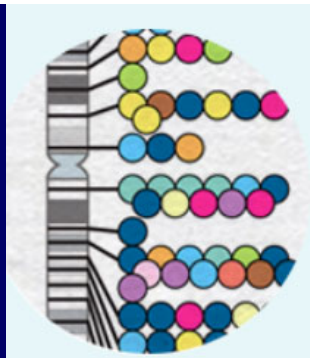
- **Applications due on December 3, 2020**

- **Create atlas of gene expression and biobank**
- **Analyze differences in gene expression**
- **Develop ELSI research projects**

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

- Massive Genome Informatics in the Cloud
(MaGIC) Jamboree
- Workshops ASHG 2020 Meeting:
GWAS analyses with Galaxy in AnVIL
Find and analyze cloud data with Gen3, Dockstore, & Terra
- Designated repository for COVID-19 HGI data





GWAS Catalog

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

Search the catalog

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

- **Recent GWAS Summary Statistics Standards and Sharing Workshop**
- **Attendees:** cohort representatives, summary statistics users, tools developers, resource providers, journal editors, and funders
- **Recommendations:**
 - Improve platform functionality
 - Standardize summary statistics reporting
 - Link summary statistics data to relevant datasets/resources

Phenotypes and Exposures (PhenX) Toolkit

PhenX Social Determinants of Health Assessments Collection

An open-access resource for standard social determinants of health measures.



NIH National Institute on Minority Health and Health Disparities



- Social Determinants of Health Assessments Collection (SDOH)

- Rapid Acceleration of Diagnostics – Underserved Populations (RADx-UP)

RAPID ACCELERATION OF DIAGNOSTICS (RADx)



 **PhenX Toolkit**

eMERGE Genomic Risk Assessment and Management Network (2020-2024)



Validate PRS
in diverse
populations



Combine with
family history,
clinical covariates



Return results
using EMR



Assess uptake of risk-reduction recommendations



Clinical Genome Resource (ClinGen)

Ancestry and Diversity Working Group Efforts

ARTICLE

Clinical Genetics Lacks Standard Definitions and Protocols for the Collection and Use of Diversity Measures

Alice B. Popejoy,^{1,2,*} Kristy R. Crooks,³ Stephanie M. Fullerton,⁴ Lucia A. Hindorff,⁵ Gillian W. Hooker,⁶ Barbara A. Koenig,⁷ Natalie Pino,⁵ Erin M. Ramos,⁵ Deborah I. Ritter,⁸ Hannah Wand,^{9,10} Matt W. Wright,¹ Michael Yudell,¹¹ James Y. Zou,¹ Sharon E. Plon,⁸ Carlos D. Bustamante,^{1,13} and Kelly E. Ormond,^{2,12,13} Clinical Genome Resource (ClinGen) Ancestry and Diversity Working Group

Clinical Genome Resource (ClinGen)

GenomeConnect Patient Registry

GenomeConnect Participation Update



>2900 participants



from 43 countries



have shared 1462
genetic changes!



ENVISION THE CURE
EMBRACE THE CHALLENGE

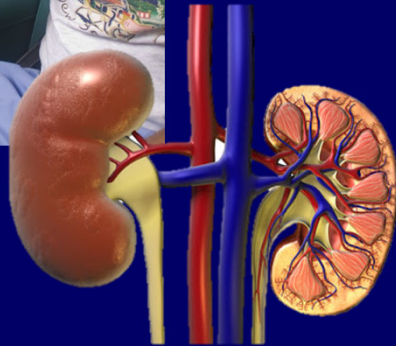


Implementing Genomics in Practice (IGNITE)



GUARDD-US

- 4 clinical sites activated
- First patient randomized in July
- >100 participants enrolled



Advancing Genomic Medicine Research and Genetic Counseling Research

- New R01 and R21 funding opportunities

Funding Opportunity Title	Advancing Genomic Medicine Research (R21 Clinical Trial Optional)		
Activity Code	Funding Opportunity Title	Advancing Genomic Medicine Research (R01 Clinical Trial Optional)	
Announcement	Activity Code	Funding Opportunity Title	Investigator-Initiated Research on Genetic Counseling Processes and Practices (R01 Clinical Trial Optional)
Related Notices	Announcement	Funding Opportunity Title	Investigator-Initiated Research on Genetic Counseling Processes and Practices (R21 Clinical Trial Optional)
Funding Opportunity	Related Notices	Activity Code	R21 Exploratory/Developmental Research Grant
	Funding Opportunity	Announcement	
		Related Notices	
		Funding Opportunity	
		Activity Code	
		Announcement Type	New
		Related Notices	None
		Funding Opportunity Announcement (FOA) Number	RFA-HG-20-049

- Stimulate innovation and advance understanding of when, where, and how to implement genomic information and technologies in clinical care
- Assess strategies to optimize the counseling process in the context of limited resources

ELSI Research Program

Program Announcements

- **Large multi-disciplinary studies:**
PAR-20-254 – R01
- **Small projects:**
PAR-20-255 – R21
- **Very small projects:**
PAR-20-257 – R03

**Due Dates
2021-2023:**

Oct. 19

Feb. 17

Jun. 17

Training and Career Development

Fiscal Year 2020 T32 Training Awards

Principal Investigator	Institution	Starting Year	Scientific Area
Michael Boehnke	U. Michigan at Ann Arbor	1995	Genomic Science
Evan Eichler	U. Washington	1995	Genomic Science
Michael Brent, Barak Cohen	Washington University	1997	Genomic Science
Maja Bucan, Junhyong Kim	U. Pennsylvania	1999	Genomic Science
John Storey, John Akey	Princeton University	2004	Genomic Science
Dan Rokhsar Michael Eisen	U. California Berkeley	2004	Genomic Science
Brent Graveley Charles Lee	U. Connecticut	2020	Genomic Science

2020 Genomic Innovator Awards



Early Stage Investigators	
Jessica Chong	Billy Tsz Cheong Lau
Katrina Claw	Jie Liu
Lee Cong	Craig Lowe
Yarui Diao	Christine Lu
Jesse Engreitz	Wouter Meuleman
Audrey Hendricks	Casey Overby Taylor

Applications due October 30, 2020

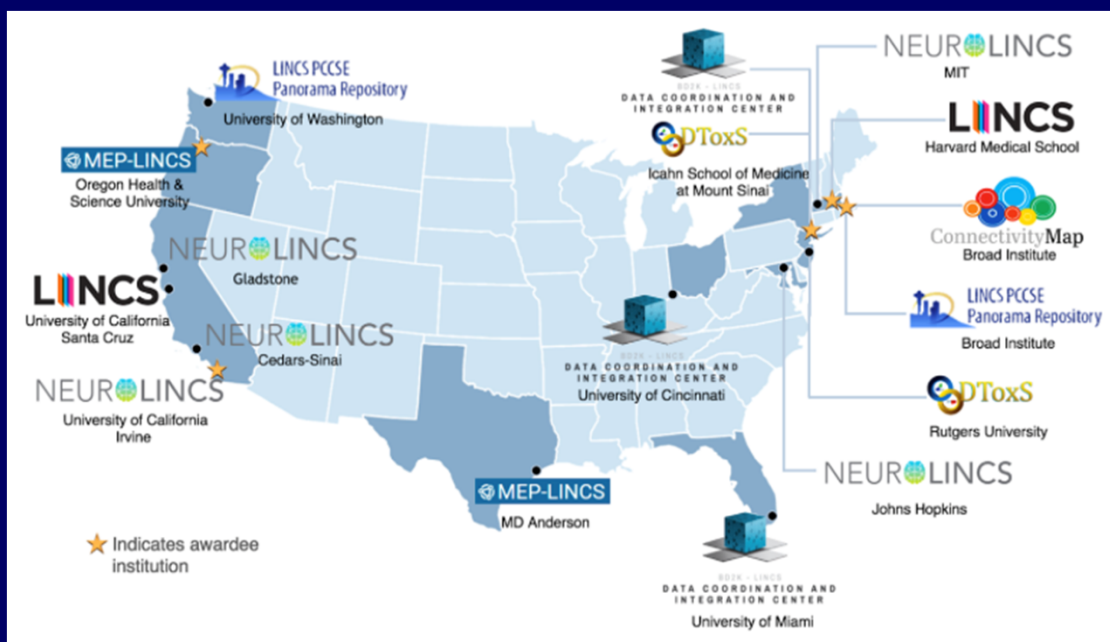
Director's Report Outline

- 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



Library of Integrated Network-based Cellular Signatures (LINCS)

LINCS Final Symposium: November 19-20, 2020



- Highlight achievements
- Focus on how community can access LINCS data and tools

Harnessing Data Science for Health Discovery and Innovation in Africa (DS-I Africa)

Funding Opportunity Announcements	FOA Number	Due Date
DS-I Africa Open Data Science Platform and Coordinating Center (U2C)	RFA-RM-20-018	December 3, 2020
DS-I Africa Research Hubs (U54)	RFA-RM-20-015	December 8, 2020
DS-I Africa Research Training Program (U2R)	RFA-RM-20-016	November 24, 2020
DS-I Africa Ethical, Legal, and Social Implications Research (U01)	RFA-RM-20-017	December 1, 2020

Human Biomolecular Atlas Program (HuBMAP)



HuBMAP Donors Samples Datasets Collections Previews CCF Documentation Login

Datasets

547 results found

Dataset ↓	Group ↓	Data Types ↓	Organ ↓	Status ↓	Last Modified ↓
HBM953.VVQN.686	Stanford TMC	Targeted Shotgun / Flow-injection LC-MS	Small Intestine	QA	2020-08-07 14:22:35
HBM873.RGWW.444	Stanford TMC	Targeted Shotgun / Flow-injection LC-MS	Small Intestine	QA	2020-08-07 14:20:44
HBM224.DLMP.923	Stanford TMC	Targeted Shotgun / Flow-injection LC-MS	Small Intestine	QA	2020-08-07 14:20:00
HBM233.DDWS.858	Stanford TMC	Targeted Shotgun / Flow-injection LC-MS	Small Intestine	QA	2020-08-07 14:19:04

Data Type

- ☐ SNARE-seq 120
- ☐ scATAC-seq (SNARE-seq) 48 [SnapATAC]
- ☐ Untargeted LC-MS 28
- ☐ CODEX 26
- ☐ CODEX [Cytokit + SPRM] 26

[View all](#)

Organ

- ☐ Kidney (Left) 168
- ☐ Kidney (Right) 168
- ☐ Spleen 48
- ☐ Small Intestine 45
- ☐ Large Intestine 42

[View all](#)

- HuBMAP's first data release in September
- Data generated with healthy tissues from seven organs
- Functional Proteomics Virtual Workshop
- New awards in October

Faculty Institutional Recruitment for Sustainable Transformation (FIRST) Program

- **Create cultures of inclusive excellence at NIH-funded institutions**
- **Notice of Intent to Publish two FOAs:**
Cohort hiring of diverse group of research faculty
Coordination and Evaluation Center
- **Estimated publication date: Sept. 9, 2020**
- **Estimated application due date: Nov. 9, 2020**



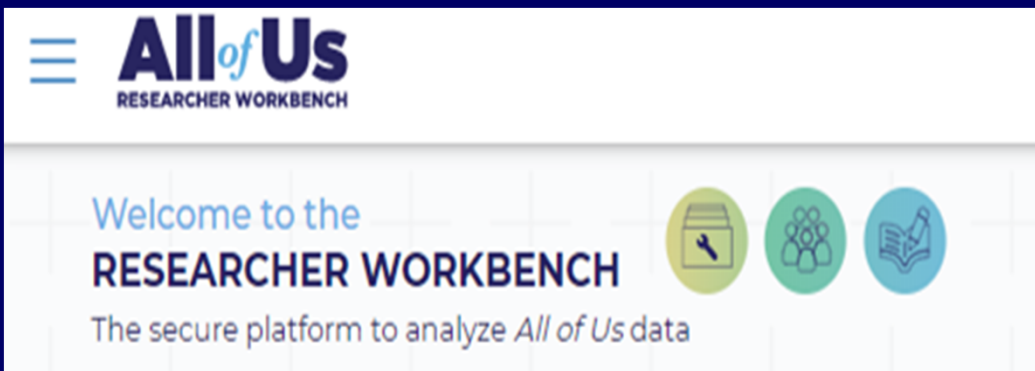
*Great minds
think differently...*

All of Us

RESEARCH PROGRAM

- Genomic data being generated
- Targets for the next year:
 - >200K with genotype arrays
 - ~100K with whole-genome sequencing
- Data Curation:
 - Data and Research Center
 - Data available in 2021
- Return of Information:
 - FDA approval of Investigational Device Exemption
 - Hereditary disease risk and pharmacogenomics





Data: physical measurements, survey data, and some EHR data - much more to come (e.g., genomics, digital health technology)



Tools: Cohort Builder, Dataset Builder, Concept Sets
-more to come



Help Resources: FAQs, sample notebooks and workspaces, documentation, Help Desk

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

NHGRI Postdoctoral Fellowship in History of Genomics



2020 Short Course in Genomics



Summer Virtual Engagement



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**

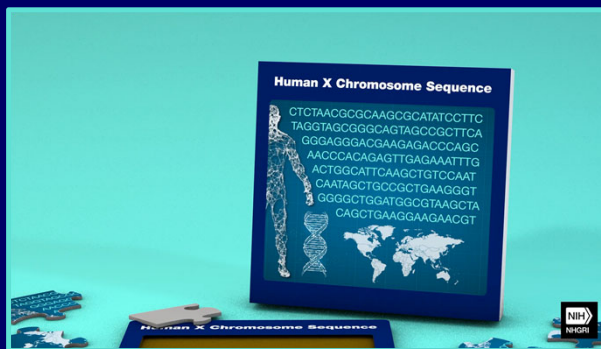
NHGRI Researchers Generate First Gapless Sequence of Human X Chromosome

Telomere-to-telomere assembly of a complete human X chromosome

Karen H. Miga , Sergey Koren, [...] Adam M. Phillippy 



YouTube



Social Media

News release

Press coverage

New President-Elect, American Society of Human Genetics



Charles Rotimi, Ph.D.



NHGRI Director on Twitter





The Genomics Landscape

Email Updates

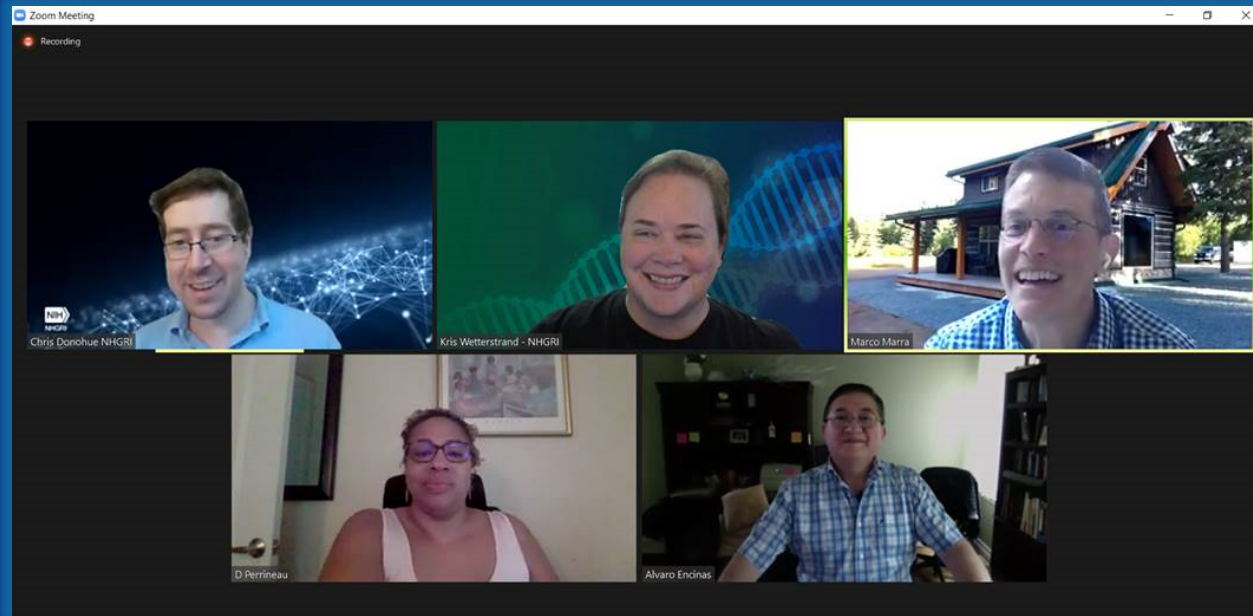
Sign up to receive National Human Genome Research Institute (NHGRI) updates and stay informed about our latest science, research, news, upcoming events and website content.

Email Address



National Human Genome
Research Institute

Thanks!



Special Thanks!

The background is a dark blue circle. In the center is a white circle. Surrounding the center are concentric circles containing the letters A, T, C, and G, representing DNA base pairs. The letters are arranged in a circular pattern, with some letters appearing more frequently than others. The overall design is clean and modern, with a focus on the central text.

The **Forefront**
of **Genomics®**