National Human Genome Research Institute

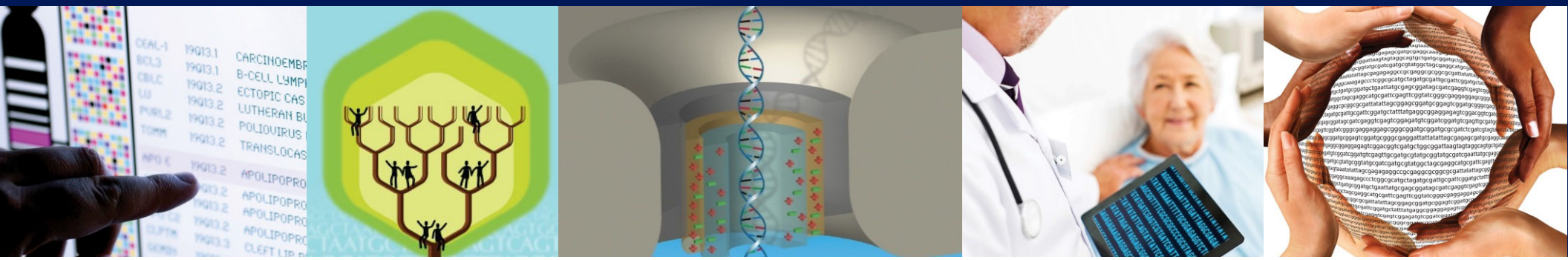
The Forefront of Genomics®

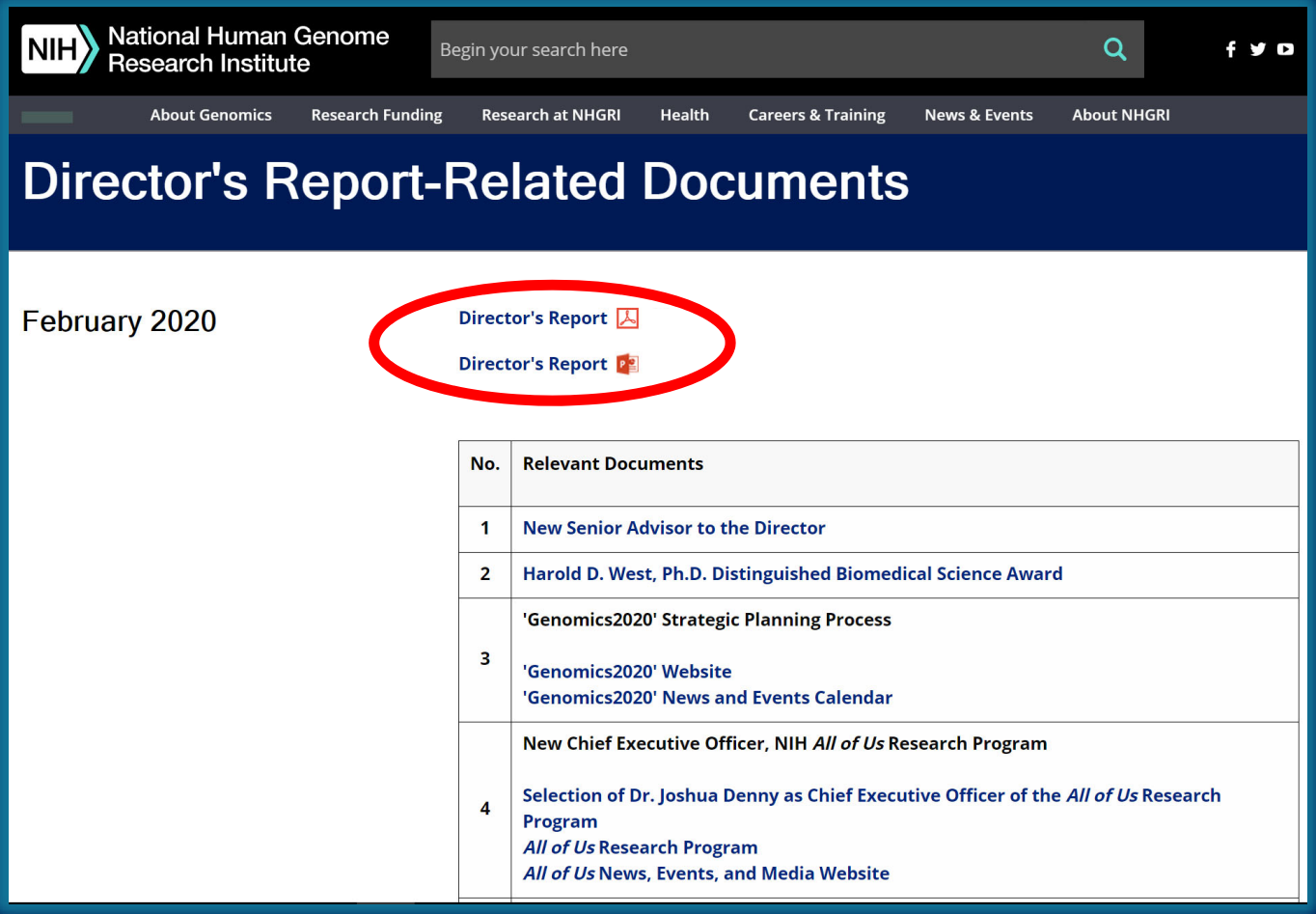
DIRECTOR'S REPORT

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

Director, NHGRI

February 2020





genome.gov/DirectorsReport

Document #

Open Session Presentations

- **Presentation: NHGRI Training Task Force Report**

  Wendy Chung

- **Concept Clearances:**

  Consortium for Understanding the Impact of Genomic Variation on Genome Function

  Mike Pazin & Dan Gilchrist

  Developmental Genotype-Tissue Expression

  Jyoti Dayal

  Advancing Genomic Medicine Research

  Christine Chang

Open Session Presentations

- **Updates:**

  Genomic Medicine Working Group Update

  Teri Manolio

  Genomics and Society Working Group Update

  Jeff Botkin

- **Public Hearing: Proposed NHGRI Organizational Changes**

  Ellen Rolfes & Eric Green

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 Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

Director's Report Outline

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VII. NHGRI Intramural Research Program

Retirement of Grants Management Specialist

Diane Patterson

Departure of Extramural Program Director

Anastasia Wise, Ph.D.

New Senior Advisor to the Director

Chris Gunter, Ph.D.

New Extramural Program Director

Stephanie Morris, Ph.D.

New Extramural Program Director

Luis Cubano, Ph.D.

New Extramural Genomic Program Administrator and Program Director

Jennifer Strasburger, M.S.

Harold D. West, Ph.D. Distinguished Biomedical Science Award

Ken Wiley Jr., Ph.D.

‘Genomics2020’ Strategic Planning Process

- Workshop: Equity, Diversity, and Data Science in Genomics
- Workshop: Genomics in Medicine and Health
- Workshop: Integrating the Past and Future of ELSI Research (American Society of Bioethics and Humanities Annual Meeting)
- NHGRI-NIMH Strategic Planning Session (World Congress of Psychiatric Genetics)
- Two Genomic Data Science Virtual Town Halls
- Upcoming ‘Genomics2020’ Strategic Planning Finale Meeting

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New Chief Executive Officer, NIH *All of Us* Research Program

All of Us RESEARCH PROGRAM

Josh Denny, M.D., M.S.

Document 4

New Director, NIH BRAIN Initiative

John Ngai, Ph.D.

Nominated Director,
National Science Foundation

Sethuraman Panchanathan, Ph.D.

Document 6

Leadership Shuffles at the National Cancer Institute & Food and Drug Administration

Ned Sharpless, M.D.

NIH NATIONAL CANCER INSTITUTE

FDA

Stephen Hahn, M.D.

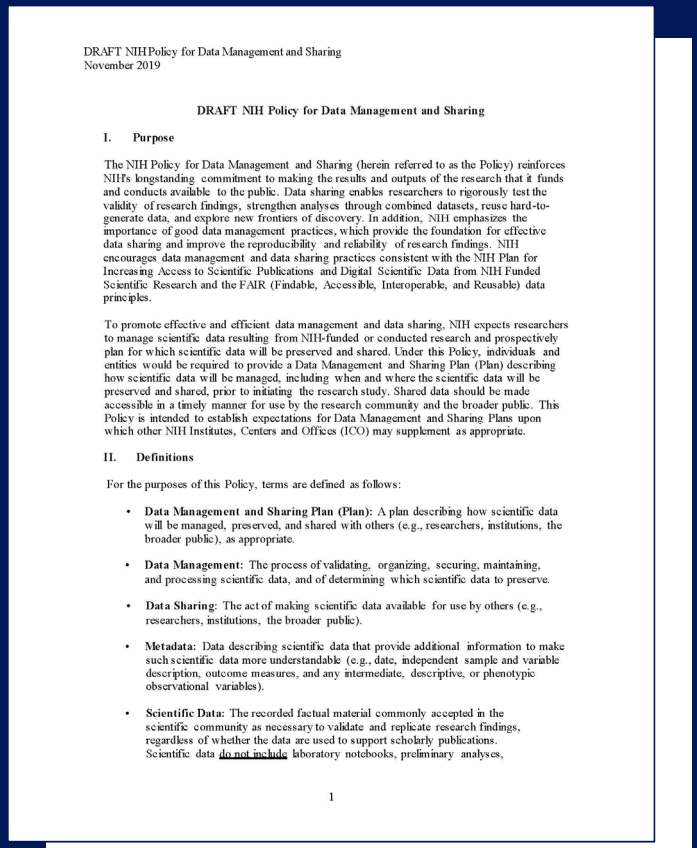
Document 7

Martha Somerman Retires as Director, National Institute of Dental and Craniofacial Research

Martha Somerman, D.D.S., Ph.D.

Lawrence Tabak, D.D.S., Ph.D.

Draft NIH Policy for Data Management and Sharing



DRAFT NIH Policy for Data Management and Sharing
November 2019

DRAFT NIH Policy for Data Management and Sharing

I. Purpose

The NIH Policy for Data Management and Sharing (herein referred to as the Policy) reinforces NIH's longstanding commitment to making the results and outputs of the research that it funds and conducts available to the public. Data sharing enables researchers to rigorously test the validity of research findings, strengthen analyses through combined datasets, reuse hard-to-generate data, and explore new frontiers of discovery. In addition, NIH emphasizes the importance of good data management practices, which provide the foundation for effective data sharing and improve the reproducibility and reliability of research findings. NIH encourages data management and data sharing practices consistent with the NIH Plan for Increasing Access to Scientific Publications and Digital Scientific Data from NIH Funded Scientific Research and the FAIR (Findable, Accessible, Interoperable, and Reusable) data principles.

To promote effective and efficient data management and data sharing, NIH expects researchers to manage scientific data resulting from NIH-funded or conducted research and prospectively plan for which scientific data will be preserved and shared. Under this Policy, individuals and entities would be required to provide a Data Management and Sharing Plan (Plan) describing how scientific data will be managed, including when and where the scientific data will be preserved and shared, prior to initiating the research study. Shared data should be made accessible in a timely manner for use by the research community and the broader public. This Policy is intended to establish expectations for Data Management and Sharing Plans upon which other NIH Institutes, Centers and Offices (ICO) may supplement as appropriate.

II. Definitions

For the purposes of this Policy, terms are defined as follows:

- **Data Management and Sharing Plan (Plan):** A plan describing how scientific data will be managed, preserved, and shared with others (e.g., researchers, institutions, the broader public), as appropriate.
- **Data Management:** The process of validating, organizing, securing, maintaining, and processing scientific data, and of determining which scientific data to preserve.
- **Data Sharing:** The act of making scientific data available for use by others (e.g., researchers, institutions, the broader public).
- **Metadata:** Data describing scientific data that provide additional information to make such scientific data more understandable (e.g., date, independent sample and variable description, outcome measures, and any intermediate, descriptive, or phenotypic observational variables).
- **Scientific Data:** The recorded factual material commonly accepted in the scientific community as necessary to validate and replicate research findings, regardless of whether the data are used to support scholarly publications. Scientific data do not include laboratory notebooks, preliminary analyses,

1

- **Draft NIH policy proposes that researchers tell NIH how they plan to manage and share data**

  All data MUST be managed, but not all data necessarily need to be shared

- **Comment period closed on January 10, 2020**

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Fiscal Year 2020 Appropriations

	Fiscal Year 2019 Labor-HHS Appropriation	Fiscal Year 2020 Labor-HHS Appropriation	$ Increase	% Increase
NIH	$39.1 B	$41.7 B	**~$2.6 B**	~6.6%
NHGRI	$575 M	$604 M	**~$29 M**	~5.0%

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Mourning the Loss of Jo Messing

Document 11

Mourning the Loss of David Hogness

Document 12

Mourning the Loss of Phil Leder

2019 American Society of Human Genetics Awards

Stylianos Antonarakis

Charles Rotimi

Sarah Tishkoff

Huda Zoghbi

Hal Dietz

ASHG 2019 ANNUAL MEETING
HOUSTON • OCTOBER 15-19

Document 13

Schottenstein Prize &
Oscar B. Hunter Career Award

Dan Roden, M.D.

Elected to National Academy of Medicine

James Eberwine
Michael Lenardo
Elaine Mardis
Julie Segre
Richard Young

Elected to AAAS

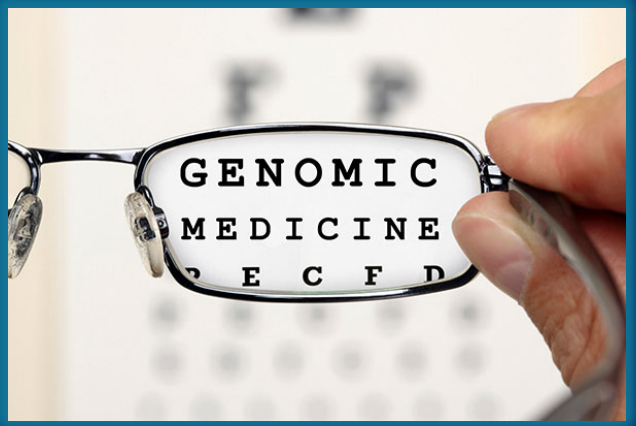
Gary Churchill
Shirley Tilghman

Document 16

International Common Disease Alliance (ICDA)

INTERNATIONAL COMMON DISEASE ALLIANCE | from Maps to Mechanisms to Medicine

Genomic Medicine Working Group Publishes "Year in Review: 2019"



YEAR IN REVIEW

Genomic Medicine Year in Review: 2019

Teri A. Manolio,[1,*] Carol J. Bult,[2] Rex L. Chisholm,[3] Patricia A. Deverka,[4] Geoffrey S. Ginsburg,[5] Gail P. Jarvik,[6] Howard L. McLeod,[7] George A. Mensah,[8] Mary V. Relling,[9] Dan M. Roden,[10] Robb Rowley,[1] Cecelia Tamburro,[1] Marc S. Williams,[11] and Eric D. Green[1]

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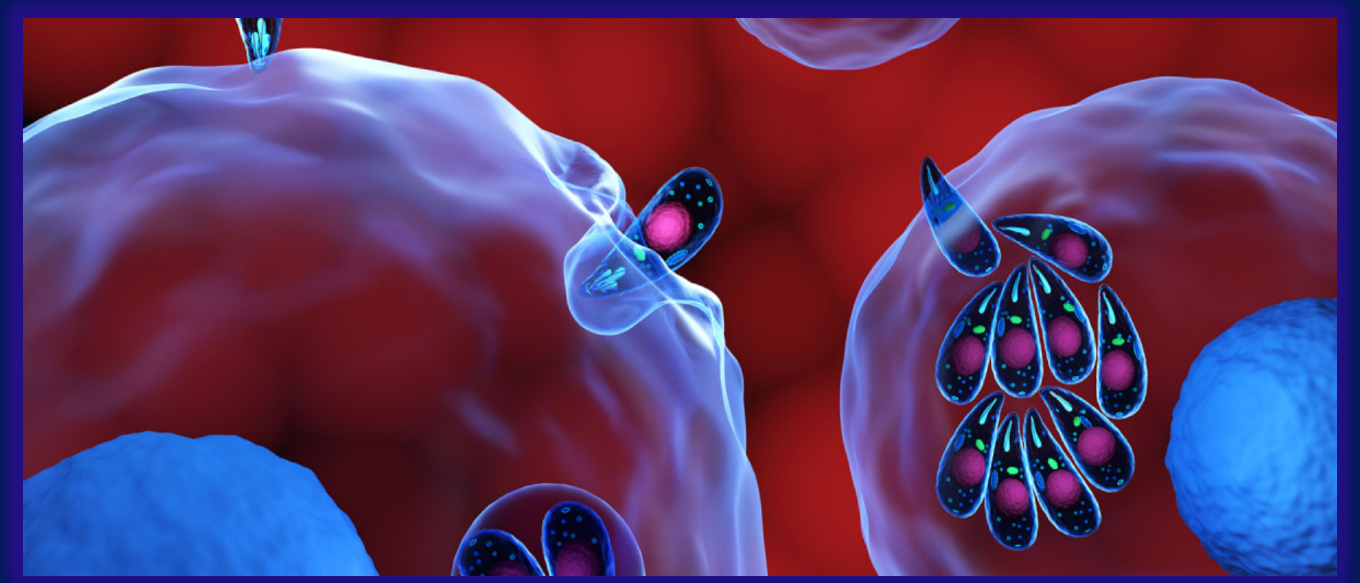
Science's 2019 Breakthrough of the Year Runners Up

Face-to-face with the Denisovans

A 'missing link' microbe emerges

Drug treats most cases of cystic fibrosis

The Scientist's Top Technical Advances of 2019



Artificial intelligence tackles life science
Gene editing spies on cells
DNA on a chip
A boost for CRISPR accuracy

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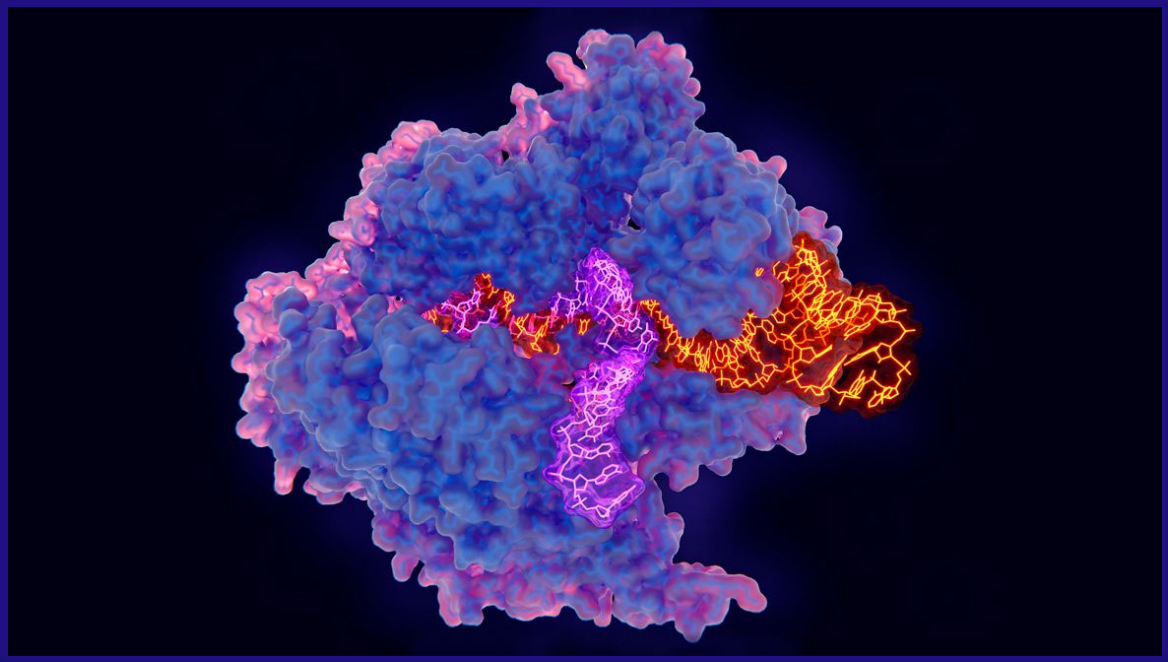
Nature's Science of the Decade



nature

Genomes In The News…

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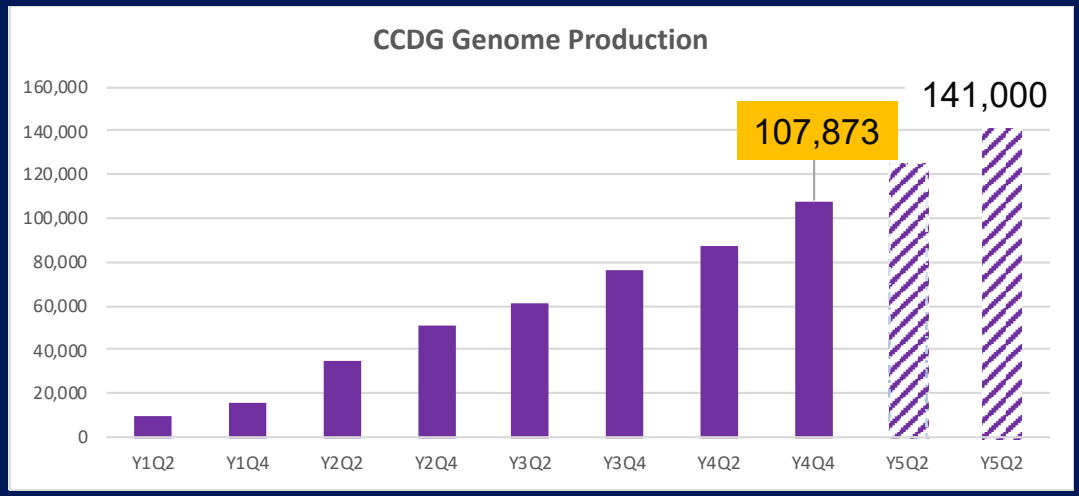
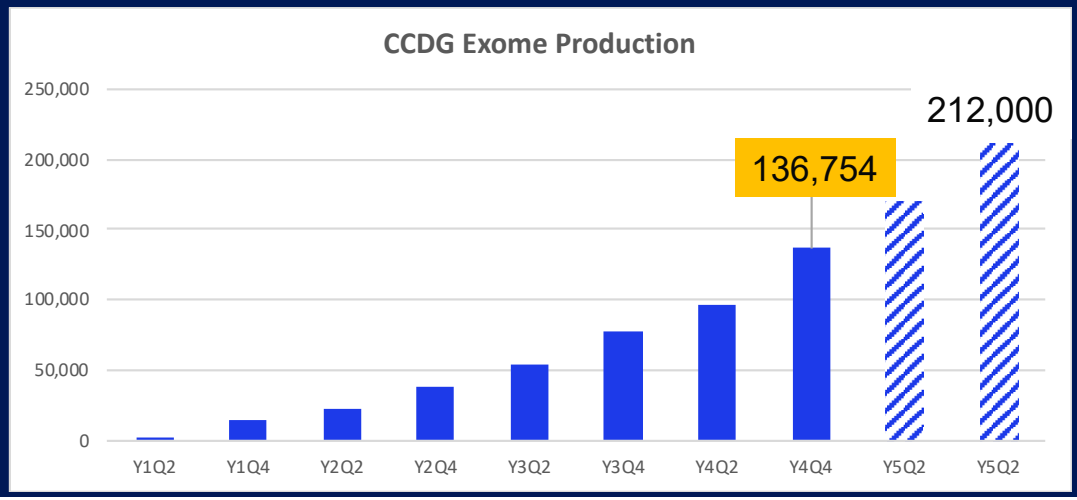
Genome Sequencing Program
Centers for Common Disease Genomics





Genome Sequencing Program

Genome Sequencing Program Analysis Centers

Abul-Husn *et al. Genome Medicine* (2020) 12:2
https://doi.org/10.1186/s13073-019-0691-1

Genome Medicine

Check for updates

Exome sequencing reveals a high prevalence of *BRCA1* and *BRCA2* founder variants in a diverse population-based biobank

Noura S. Abul-Husn[1,2,3,4*], Emily R. Soper[1,2†], Jacqueline A. Odgis[1,2†], Sinead Cullina[1,2], D
Arden Moscati[1,2], Jessica E. Rodriguez[1,2], CBIPM Genomics Team[1,2], Regeneron Genetics C
Judy H. Cho[2,3,4], Gillian M. Belbin[1,2,3], Sabrina A. Suckiel[1,2] and Eimear E. Kenny[1,2,3,4]

SCIENTIFIC REPORTS

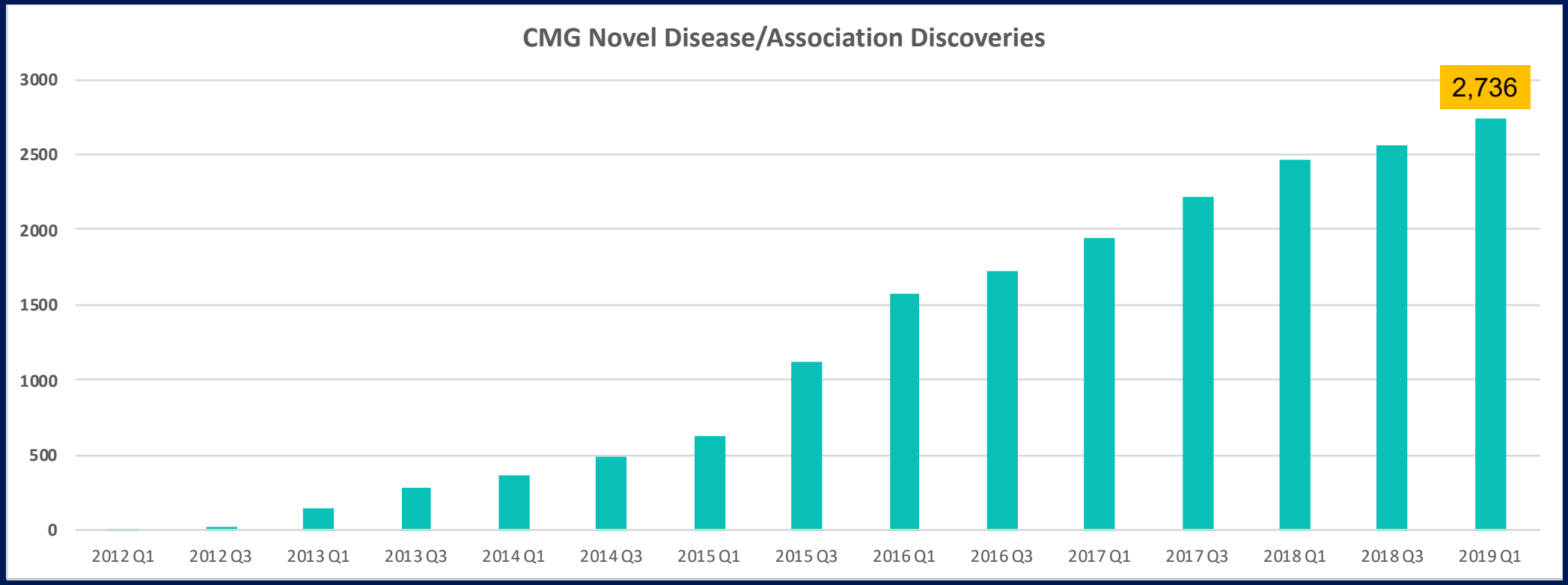
nature research

A Bootstrap Method for Goodness of Fit and Model Selection with a Single Observed Network

Sixing Chen & Jukka-Pekka Onnela*

Genome Sequencing Program

Centers for Mendelian Genomics



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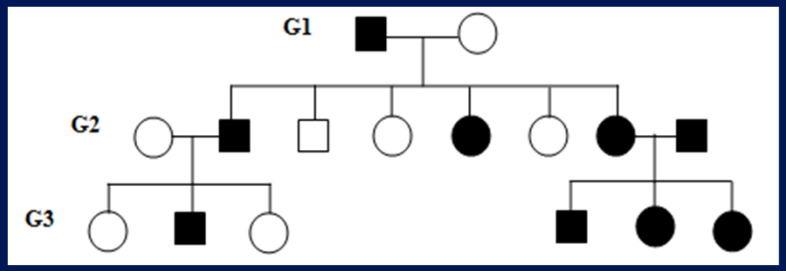
Mendelian Genomics Research Consortium Funding Opportunity Announcements

National Human Genome Research Institute @genome_gov · Jan 15

@genome_gov released two calls for applications to establish a Mendelian Genomics Research Consortium to expand the research done on Mendelian conditions with an identified genetic cause.

Research Centers: bit.ly/2NFHe0L

Data Coordinating Center: bit.ly/35YqW9y



Applications due April 15, 2020

Pre-Application webinar on Feb. 12, 2020

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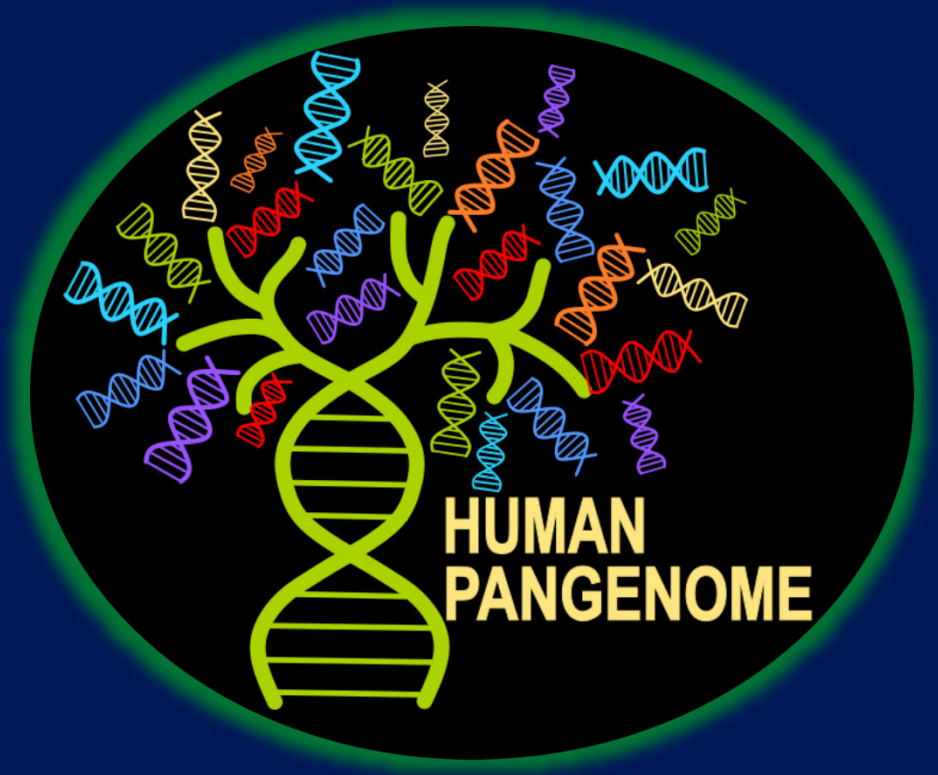
Human Genome Reference Program



Human Genome Reference Center

THE GENOME INSTITUTE at Washington University

High-Quality Human Reference Genomes Center

UNIVERSITY OF CALIFORNIA SANTA CRUZ Genomics Institute

Technology Development Program

- **Novel Nucleic Acid Sequencing Technology Development**
  - RFA-HG-18-001 (R01, also linked R21 and R43/44)
  - Applications due June 26, 2020
- **Advanced Genomic Technology Development Meeting**
  - Northeastern University
  - Grantee Session: May 27-28, 2020
  - Public Session: May 29, 2020

Document 26

Technology Development Program

nature methods

ARTICLES

https://doi.org/10.1038/s41592-019-0617-2

Nanopore native RNA sequencing of a human poly(A) transcriptome

Rachael E. Workman[1,9], Alison D. Tang[2,3,9], Paul S. Tang[4,9], Miten Jain[2,3,9], John R. Tyson[5,9], Roham Razaghi[1,9], Philip C. Zuzarte[4], Timothy Gilpatrick[1], Alexander Payne[6], Joshua Quick[7], Norah Sadowski[1], Nadine Holmes[6], Jaqueline Goes de Jesus[7], Karen L. Jones[5], Cameron M. Soulette[2,3], Terrance P. Snutch[5], Nicholas Loman[7], Benedict Paten[2,3], Matthew Loose[6], Jared T. Simpson[4,8], Hugh E. Olsen[2,3,10], Angela N. Brooks[2,3,10], Mark Akeson[2,3,10]* and Winston Timp[1,10]*

ENCyclopedia of DNA Elements (ENCODE)

- Jamboree Meeting at Duke University – Oct. 2019
- ENCODE Consortium Meeting – Dec. 2019

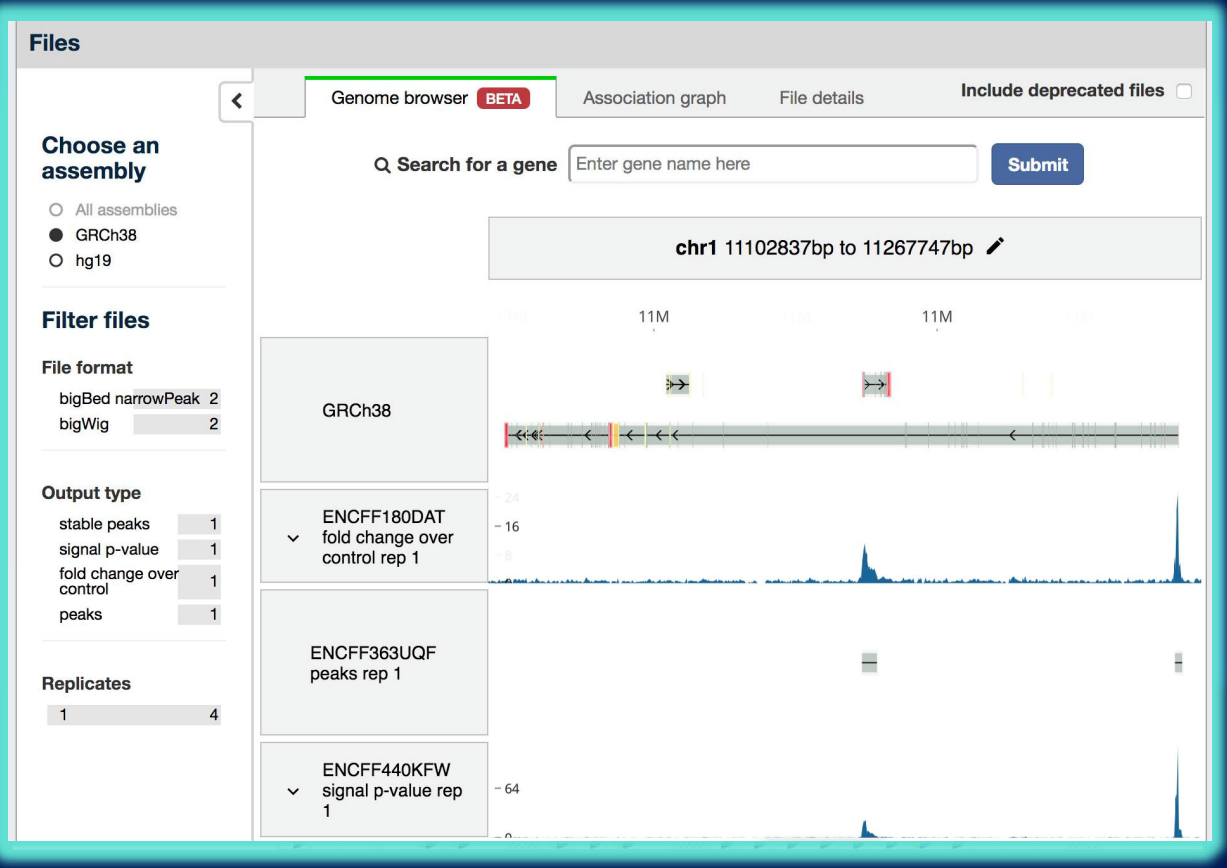
- Users & Applications Meeting in Barcelona, Spain – Oct. 2020

First Public Meeting for ENCODE Users in Europe

ENCyclopedia of DNA Elements (ENCODE)

ENCODE Portal Updates

- Data Collections: EN-TEx
- Genome Browser



Centers of Excellence in Genomic Science (CEGS)

Emerging Centers of Excellence

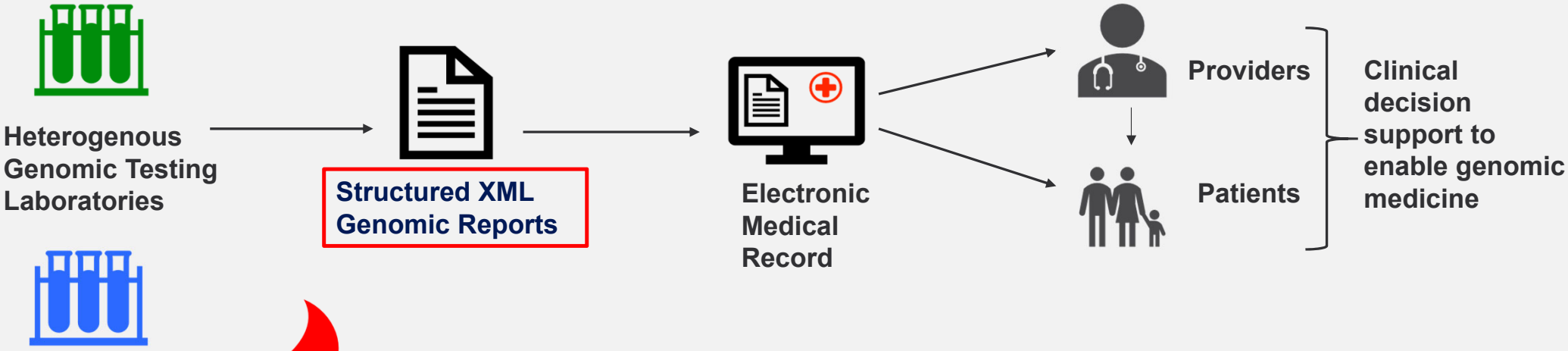
- $10M in Fiscal Year 2020
- To fund Centers at institutions that are not previous CEGS grantees

Notice of Change: NOT-HG-20-013

- Amends PAR-19-204 (CEGS PAR)
- Updated selection criteria

New receipt date for Emerging Centers:
March 26, 2020
Standard CEGS receipt dates unaffected:
May 18, 2020; May 20, 2021

eMERGE network
ELECTRONIC MEDICAL RECORDS AND GENOMICS

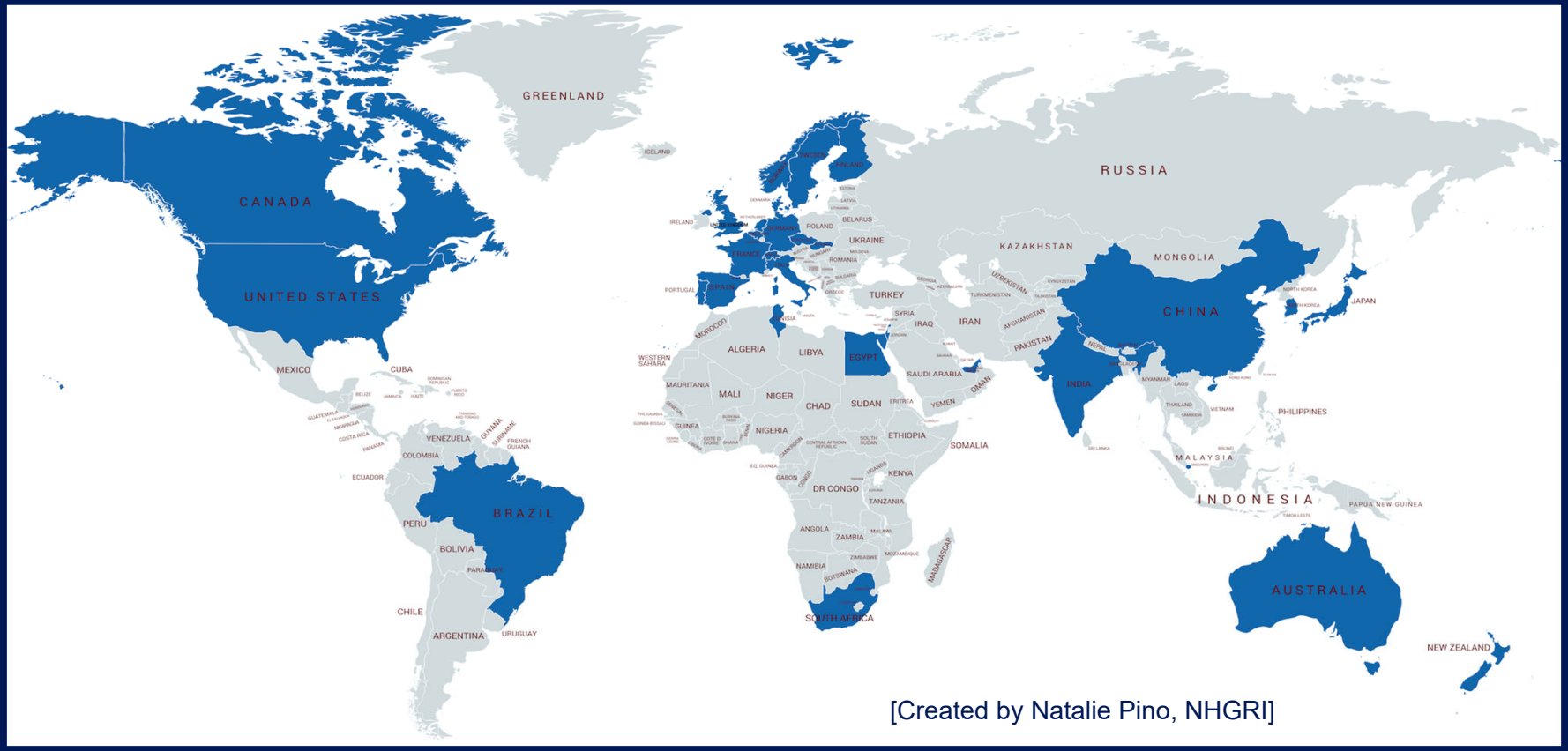


Helping to define a **Fast Healthcare Interoperability Resources (FHIR)** standard

- **Problem: Genomic testing results from heterogenous sources in non-structured pdf formats are not easily shareable**
- **Solution: Harmonized DNA-capture panels and structured Extensible Markup Language (XML) schema, which offers flexibility for manipulating and updating diverse information in the electronic health record**

Clinical Genome Resource (ClinGen)

1125 investigators from 30 countries



[Created by Natalie Pino, NHGRI]

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Clinical Genome Resource (ClinGen)

1125 investigators from 30 countries

Topics of ClinGen Genomic Variant and Gene Curation Expert Panels

- *ACADVL*
- Aminoacidopathy
- Arrhythmogenic RV Cardiomyopathy
- Brain Malformations
- Breast/Ovarian Cancer
- Brugada Syndrome
- Cardiomyopathy
- *CDH1*
- Cerebral Creatine Deficiency Syndromes
- Coagulation Factor Deficiency
- Colon Cancer
- Congenital Myopathies
- DICER1 and miRNA-Processing
- Epilepsy
- Familial Hypercholesterolemia
- Familial Thoracic Aortic Aneurysm and Dissection
- Fatty Acid Oxidation Disorders
- *FBN1*
- Glaucoma
- Hearing Loss
- Hemostasis/Thrombosis
- Hereditary Breast, Ovarian and Pancreatic Cancer
- Hereditary Cancer
- Hereditary Hemorrhagic Telangiectasia
- Hypertrophic Cardiomyopathy
- Intellectual Disability and Autism
- *KCNQ1*
- Long QT Syndrome
- Lysosomal Storage Disorders
- Malignant Hyperthermia Susceptibility
- Mitochondrial Diseases
- Monogenic Diabetes
- Myeloid Malignancy
- Peroxisomal Disorders
- Phenylketonuria
- Platelet Disorder
- *PTEN*
- RASopathy
- Rett and Angelman-like Disorders
- *TP53*
- *VHL*

Clinical Genome Resource (ClinGen)

ACMG & ClinGen Publish CNV Interpretation Guidelines

ACMG American College of Medical Genetics and Genomics

ACMG TECHNICAL STANDARDS | Genetics inMedicine

Technical standards for the interpretation and reporting of constitutional copy-number variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics (ACMG) and the Clinical Genome Resource (ClinGen)

Erin Rooney Riggs, MS, CGC[1], Erica F. Andersen, PhD[2,3], Athena M. Cherry, PhD[4], Sibel Kantarci, PhD[5], Hutton Kearney, PhD[6], Ankita Patel, PhD[7], Gordana Raca, MD, PhD[8], Deborah I. Ritter, PhD[9], Sarah T. South, PhD[10], Erik C. Thorland, PhD[6], Daniel Pineda-Alvarez, MD[11], Swaroop Aradhya, PhD[4,11] and Christa Lese Martin, PhD[1]

ACMG/ClinGen Technical Standards for CNVs

Our in-depth, multi-part overview of the new ACMG/ClinGen technical standards for interpretation and reporting of constiutional CNVs begins January 16, 2020. Learn more and register today!

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Clinical Genome Resource (ClinGen)

Cancer Activities

COLD SPRING HARBOR
Molecular Case Studies

COMMENTARY

A case for expert curation: an overview of cancer curation in the Clinical Genome Resource (ClinGen)

Deborah I. Ritter,[1,2] Shruti Rao,[3] Shashikant Kulkarni,[1,4] Subha Madhavan,[3] Kenneth Offit,[5,6] and Sharon E. Plon[1,2]

REGULAR ARTICLE

blood advances

Check for updates

ClinGen Myeloid Malignancy Variant Curation Expert Panel recommendations for germline *RUNX1* variants

Xi Luo,[1,*] Simone Feurstein,[2,*] Shruthi Mohan,[3] Christopher C. Porter,[4] Sarah A. Jackson,[5] Sioban Keel,[6] Michael Chicka,[7] Anna L. Brown,[8] Chimene Kesserwan,[9] Anupriya Agarwal,[10] Minjie Luo,[11] Zejuan Li,[12,13] Justyne E. Ross,[3] Panagiotis Baliakas,[14] Daniel Pineda-Alvarez,[15] Courtney D. DiNardo,[16] Alison A. Bertuch,[1] Nikita Mehta,[17] Tom Vulliamy,[18] Ying Wang,[19] Kim E. Nichols,[9] Luca Malcovati,[20] Michael F. Walsh,[21] Lesley H. Rawlings,[22] Shannon K. McWeeney,[23] Jean Soulier,[24] Anna Raimbault,[24] Mark J. Routbort,[25] Liying Zhang,[26] Gabriella Ryan,[27] Nancy A. Speck,[28] Sharon E. Plon,[1] David Wu,[29,†] and Lucy A. Godley[2,†]

Clinical Sequencing Evidence-Generating Research Program

Currently provide or recommend coverage? N = 14 Payers	
Pediatric Exome Sequencing	71% (10/14)
Prenatal Exome Sequencing	0% (0/14)

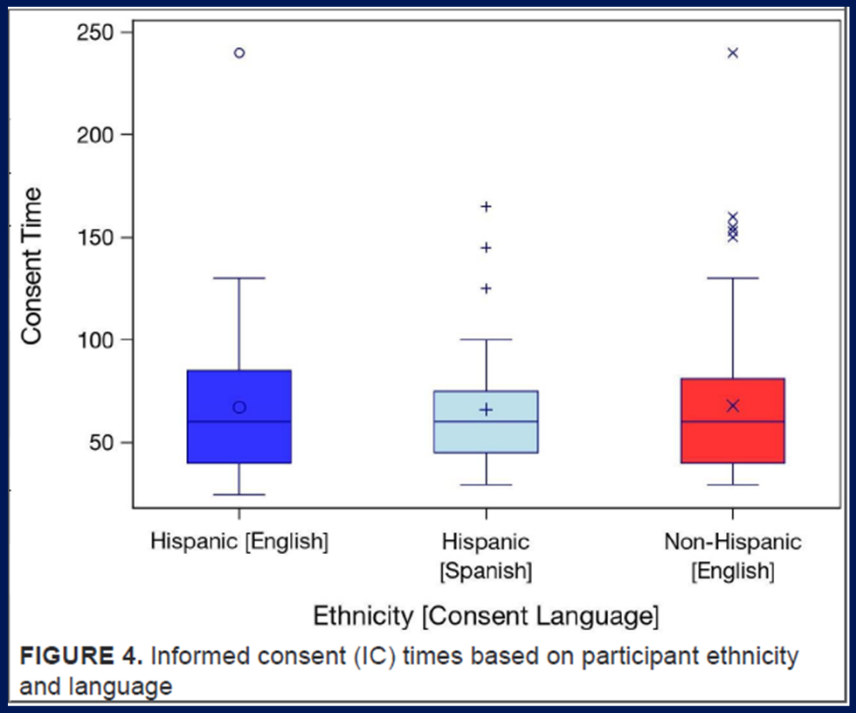


FIGURE 4. Informed consent (IC) times based on participant ethnicity and language

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

- **Involvement in trans-NIH interoperability efforts:**
  - NIH Workshop on Cloud-Based Platforms Interoperability
  - Research Authorization & Authentication Service (RAS)

- **Upcoming AnVIL datasets:**

cser
Clinical Sequencing Evidence-Generating Research

emerge network
ELECTRONIC MEDICAL RECORDS & GENOMICS

NIH Biomedical Knowledgebase and Biomedical Data Repository FOAs

- **New NIH-wide FOAs for Biomedical Data Resources:**
  - **Biomedical Data Repository U24 (PAR-20-089)**
  - **Biomedical Knowledgebase U24 (PAR-20-097)**
  - **NHGRI participating (NOT-HG-20-017)**
  - **First due date: Sept. 25, 2020**
- **Renewal of the NHGRI Genomic Community Resources U24 (PAR-20-100)**
  - **Next due date: May 25, 2020**

6th iDASH Genomic Privacy Challenge

- **Challenge Tasks:**
  - Secure genotype imputation using homomorphic encryption
  - Secure training of machine learning models
  - Sharing gene-drug interaction data with blockchain
  - Deep learning as a service with secure hardware

- **Meet challenges of new privacy regulations in Europe (for GDPR) and in US (for possible federal law)**

ELSI Research Program

Notice of Special Interest: Administrative Supplement for Research on Bioethical Issues (Admin Supp Clinical Trial Optional)

Notice Number:

NOT-OD-20-038

- **Supplement bioethics-focused awards OR add bioethics to non-bioethics awards**
- **Areas of interest can be found in the FOA**
- **Applications are due March 20, 2020**

ELSI Research Program

5th ELSI Congress

- June 15-17, 2020 at Columbia University
- Plenary session topics include:
  - Past, present, and future of ELSI and genomics
  - CRISPR and gene editing
  - Research collaboration with indigenous communities
  - Polygenic risk scores
- 229 abstracts submitted

Small Business Program

SBIR · STTR
America's Seed Fund™

- ~$15 million dollars in Fiscal Year 2019
- 19 Phase I Proof of Principle awards
- 11 Phase II Pre-Commercialization awards, including:

  Ultima Genomics: New DNA sequencing platform
  Electronic Biosciences: Exonuclease-based DNA sequencing
  Arima Genomics: HiC analysis service
  Kromatid: Whole-genome chromosome painting
  Active Motif: Multiplexed ChIP-seq technology
  Somagenics: Accurate single-cell small RNA sequencing

Extramural Training and Career Development

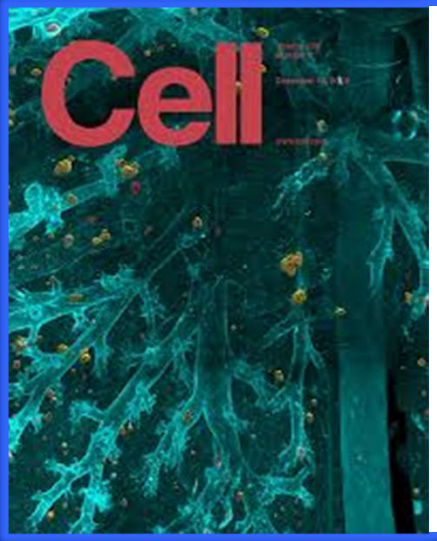
Diversity Action Plan (DAP)

- Re-issued under PAR-19-380
- $300K per year, up to 5 years
- One annual receipt date: January 25
- New reporting requirements

NHGRI Investigator-Initiated Research

- NHGRI welcomes novel and innovative investigator-initiated applications
- Investigators encouraged to reach out to program staff during development of an application

Extramural Investigator-Initiated Highlights



Article

Exon-Mediated Activation of Transcription Starts

Ana Fiszbein [1], Keegan S. Krick [1], Bridget E. Begg [1], Christopher B. Burge [1,2]

Extramural Investigator-Initiated Highlights

EDITORIAL — Annals of Internal Medicine

Precision Medicine for Clinicians: The Future Begins Now

Until la… largel… personal … lying path…

Precision Medicine in Internal Medicine

Krzy… and …

Cases in Precision Medicine: When Patients Present With Dir…

Micha…

Cases in Precision Medicine: Genetic Assessment After a Sudden C…

Ro…

Cases in Precision Medicine: Concerns About Privacy and Di…

Deb…

Cases in Precision Medicine: The Role of Pharmacogenetics in Pr…

Boha…

Cases in Precision Medicine: A Personalized Approach to Stroke and C…

Na…

Cases in Precision Medicine: Should You Participate in a Study In…

Pau…

Cases in Precision Medicine: *APOL1* and Genetic Testing in the Ev…

Y. D…

Cases in Precision Medicine: The Role of Tumor and Germline Genetic Testing in Breast Cancer Management

Jonah Tischler, BA; Katherine D. Crew, MD, MS; and Wendy K. Chung, MD, PhD

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VII. NHGRI Intramural Research Program

Library of Integrated Network-based Cellular Signatures (LINCS)

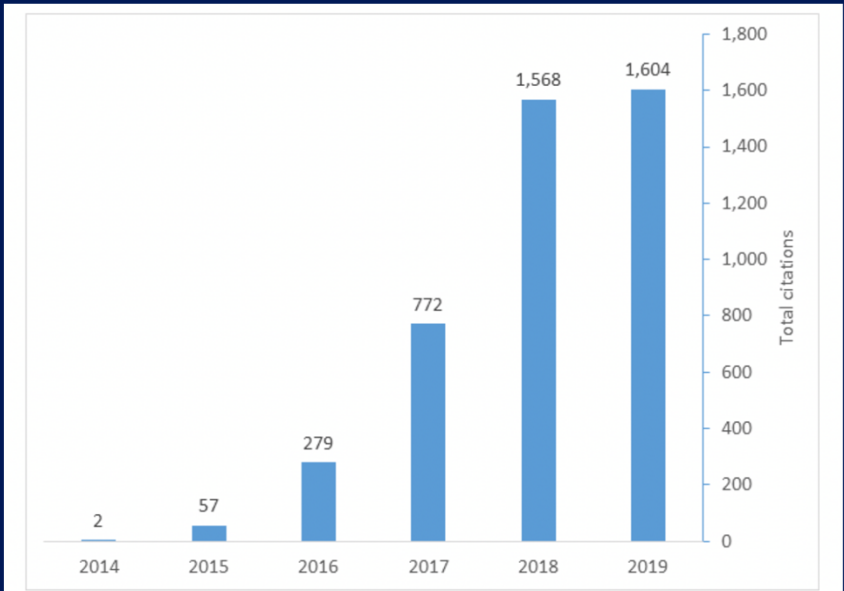


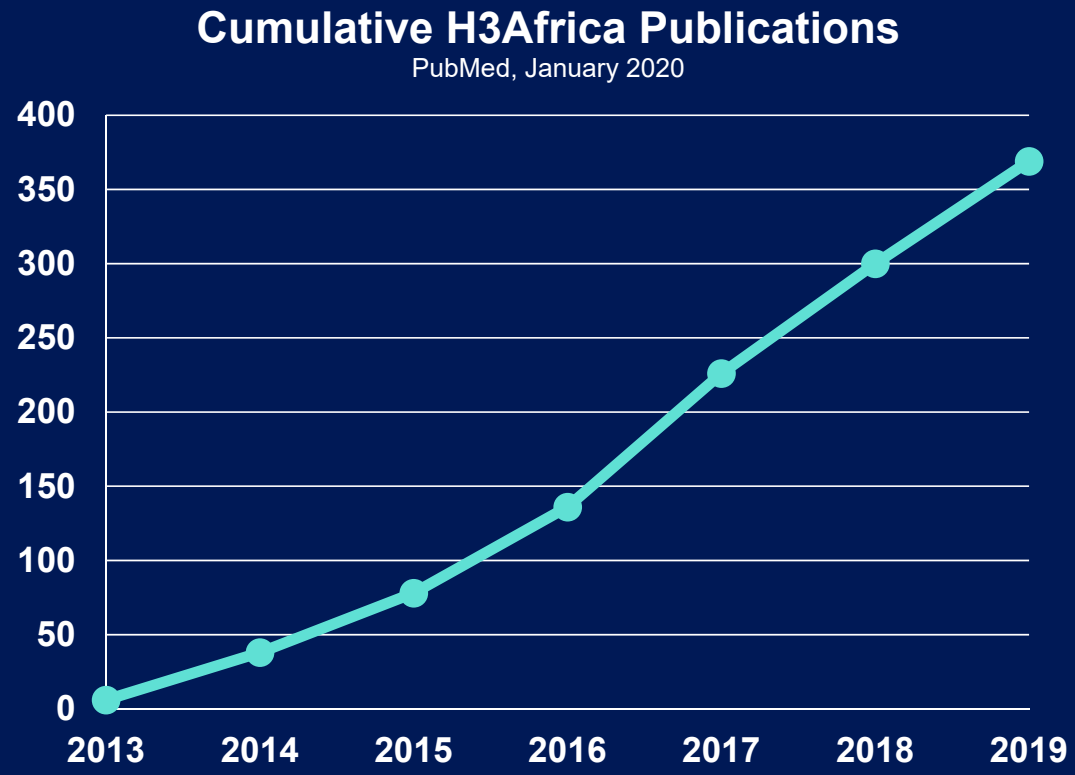
Fig. 1: This graph shows the number of times LINCS data has been cited in publications each calendar year.

- LINCS data citations continue to grow
- LINCS resources and data to be retained in Common Fund Data Ecosystem

Document 40

Human Heredity and Health in Africa (H3Africa)

51 projects, 34 countries

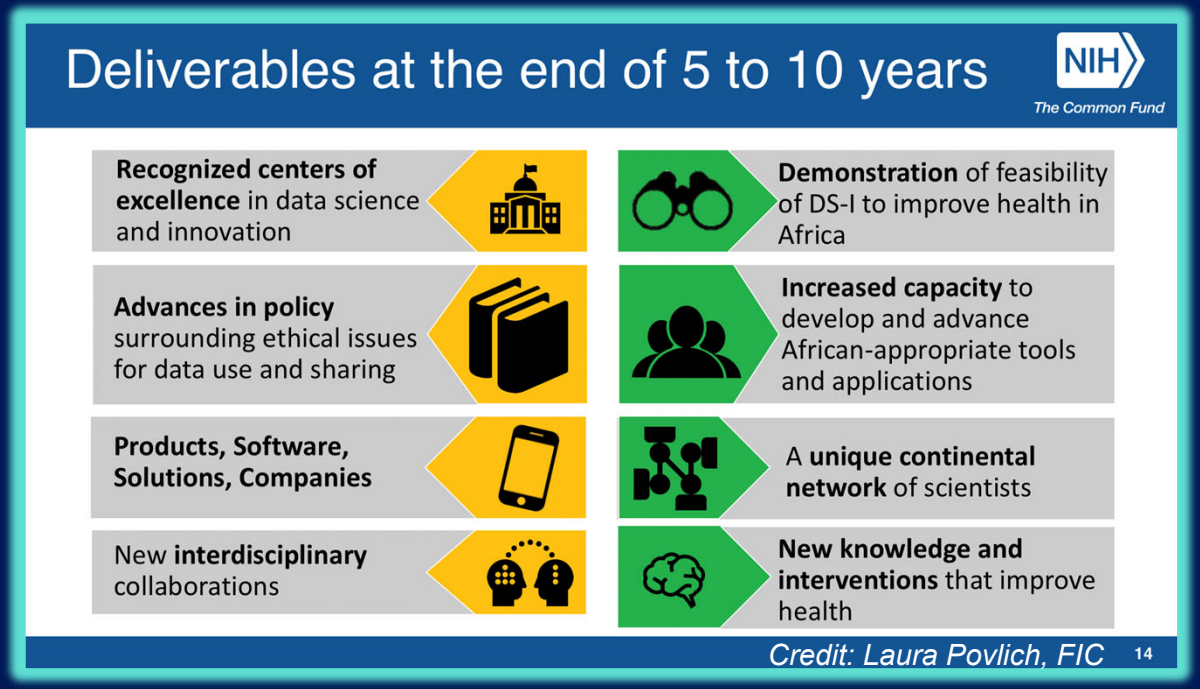


Document 41

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

Institute/Center Leads:

Roger Glass, FIC
Bruce Tromberg, NIBIB
Josh Gordon, NIMH
Patti Brennan, NLM



Undiagnosed Diseases Network (UDN)

Journal of Genetic Counseling

National Society of Genetic Counselors

ORIGINAL ARTICLE | Full Access

Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing

Chloe M. Reuter, Jennefer N. Kohler, Devon Bonner, Diane Zastrow, Liliana Fernandez, Annika Dries, Shruti Marwaha, Jean Davidson, Elly Brokamp, Matthew Herzog, Joyce Hong ... **See all authors**

First published: 03 September 2019 | **https://doi.org/10.1002/jgc4.1161**

- **Payer: 67% public, 26% private, 8% unknown**
- **Barrier: 64% documented denial, 29% payer does not cover, 8% denial other genetic testing**
- **Outcome: 35% diagnosed and 61% with clinical action**

UDN Undiagnosed Diseases Network

Human Biomolecular Atlas Program (HuBMAP)

PERSPECTIVE

OPEN

https://doi.org/10.1038/s41586-019-1629-x

The human body at cellular resolution: the NIH Human Biomolecular Atlas Program

HuBMAP Consortium*

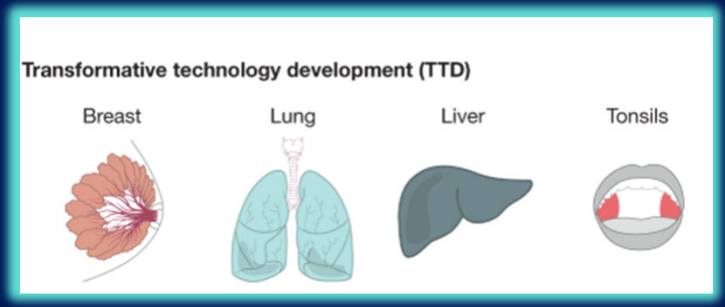
- Marker paper published October 2019
- First data release in Fall 2020
- NIH-HCA Joint Meeting in Spring 2020

Human Biomolecular Atlas Program (HuBMAP)

Two new funding opportunities:

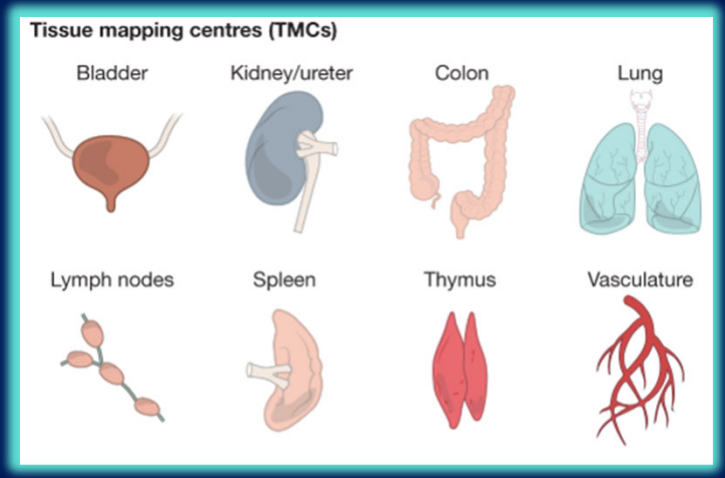
- **RFA-RM-20-001 – Transformative Technology Development (UG3/UH3)**

  Due March 3, 2020

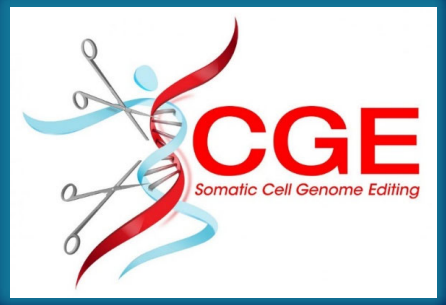


- **RFA-RM-20-002 – Tissue Mapping Centers (U54)**

  Due March 3, 2020

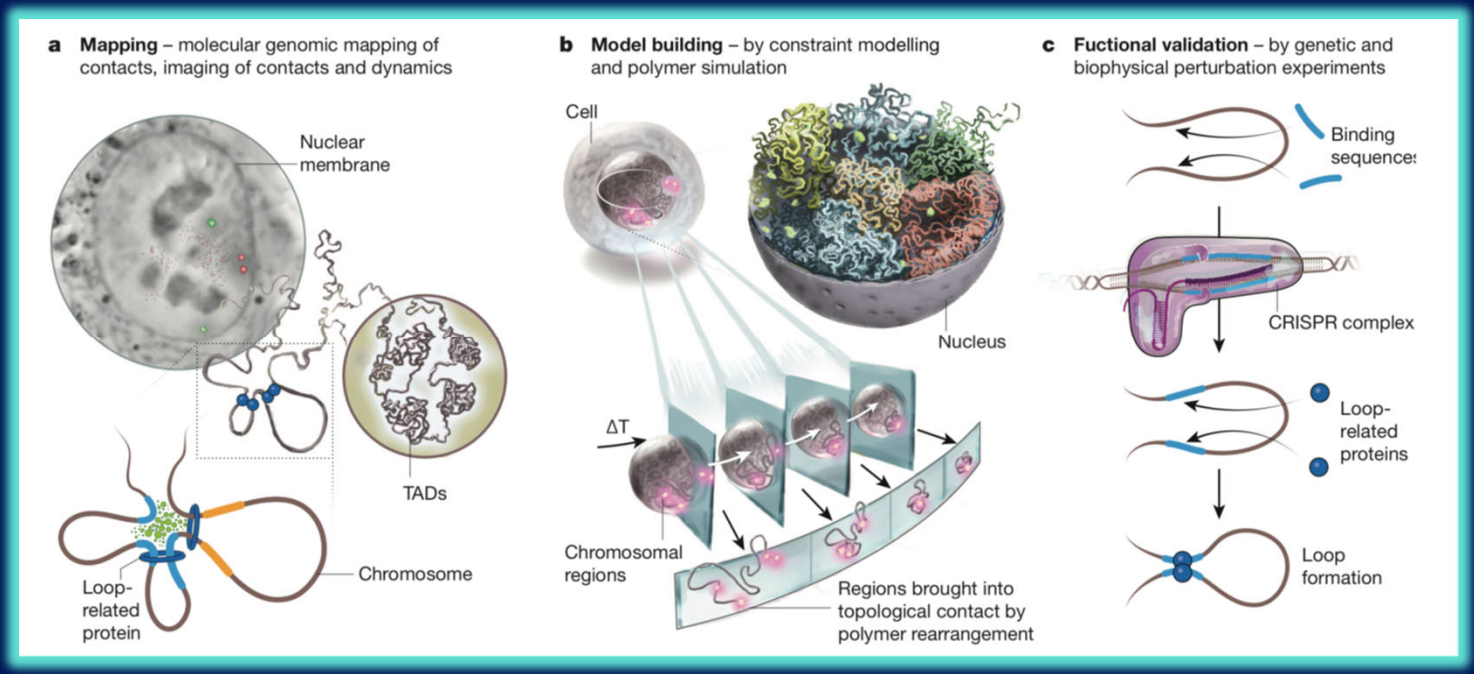


Somatic Cell Genome Editing



- **6 components:**
  - New Delivery Systems (20)
  - Animal Reporters (7)
  - Tissue Platforms (8)
  - *In Vivo* Tracking (4)
  - Novel Genome Editors (5)
  - Data Coordination Center (1)
- **45 awards now active**

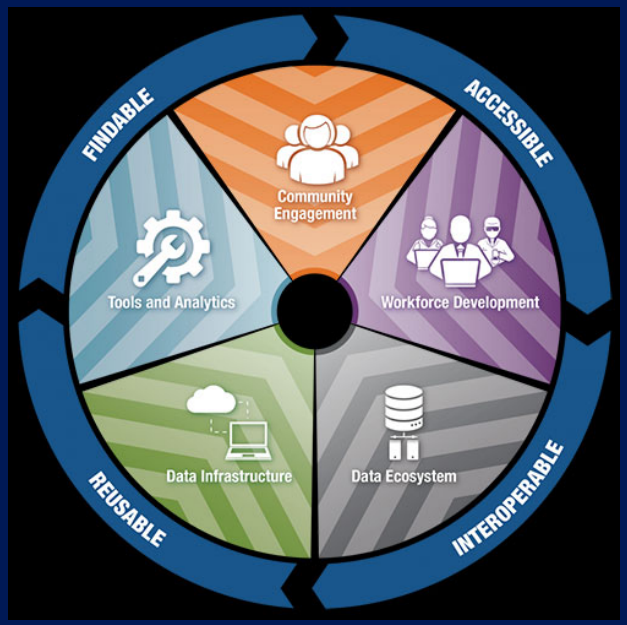
4D Nucleome Program (4DN)



- Applications for second stage due in March 2020
- NHGRI joined the leadership team

Data and Technology Advancement National Service Scholar (DATA Scholars)

- Attract experienced data scientists and engineers with interest in public service
- 1-2 year positions at NIH
- NHGRI position to focus on interoperability of cloud-based platforms
- Applications due April 30, 2020
- First cohort will start in summer 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 Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

Update to NHGRI's Implementation of the NIH Genomic Data Sharing Policy

- All human data generated by NHGRI-supported research must be derived from specimens or cell lines for which explicit consent for future research use and broad data sharing can be documented

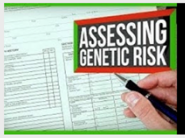
- Timeline for implementation:

  Relevant RFAs: After January 25, 2020

  All other NHGRI-supported research: After January 25, 2021

New NHGRI Polygenic Risk Score Resource

Featured video



SCISHOW

What your family history can't tell you ›

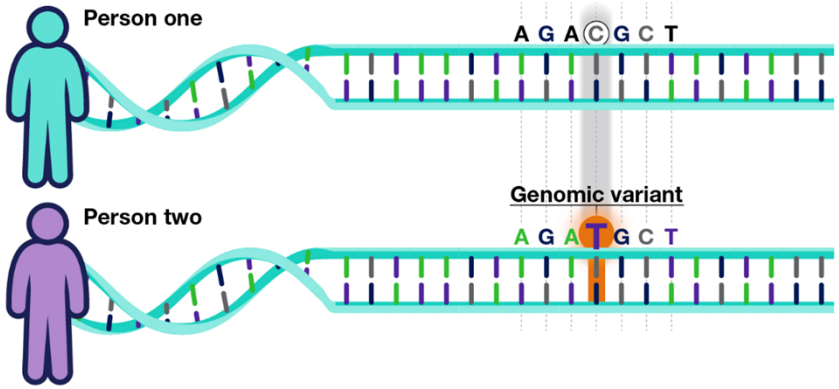
On this page

All humans have near-identical DNA sequences across the estimated 6 billion-letter code for their genome.

Slight differences exist between individuals, making each of us unique. These differences, called genomic variants, occur at specific locations within the DNA.

DNA is read like a code. This code is made up of four types of chemical building blocks - adenine, thymine, cytosine and guanine, abbreviated with the letters A, T, C and G. A genomic variant occurs in a location within the DNA where that code differs among people.

For example, in Person One below, the location shows a "C" base. But in the same location in Person Two, it is a "T."



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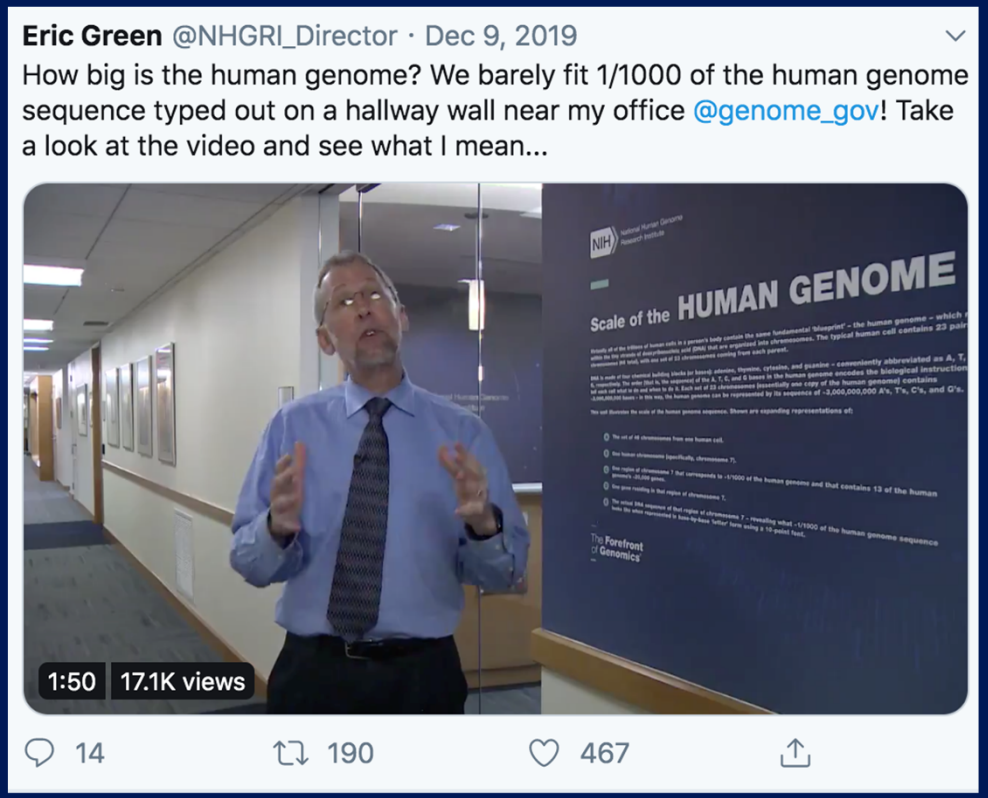
NHGRI Director's New Twitter Account @NHGRI_Director



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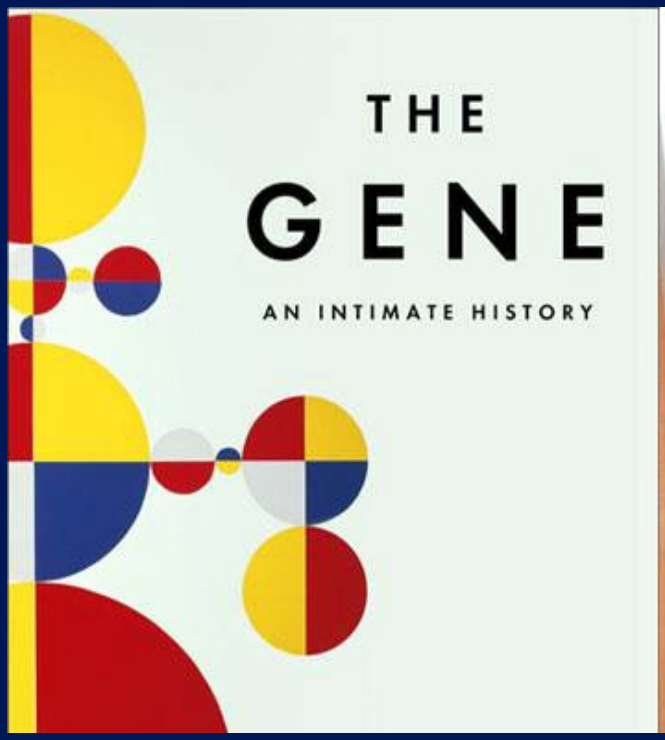
The Gene: An Intimate History WETA Documentary

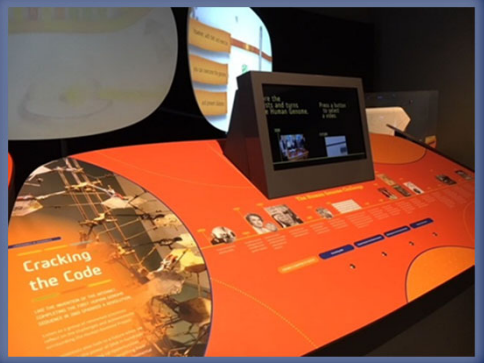


WETA

Document 51

Genome: Unlocking Life's Code Exhibition

Travel Schedule



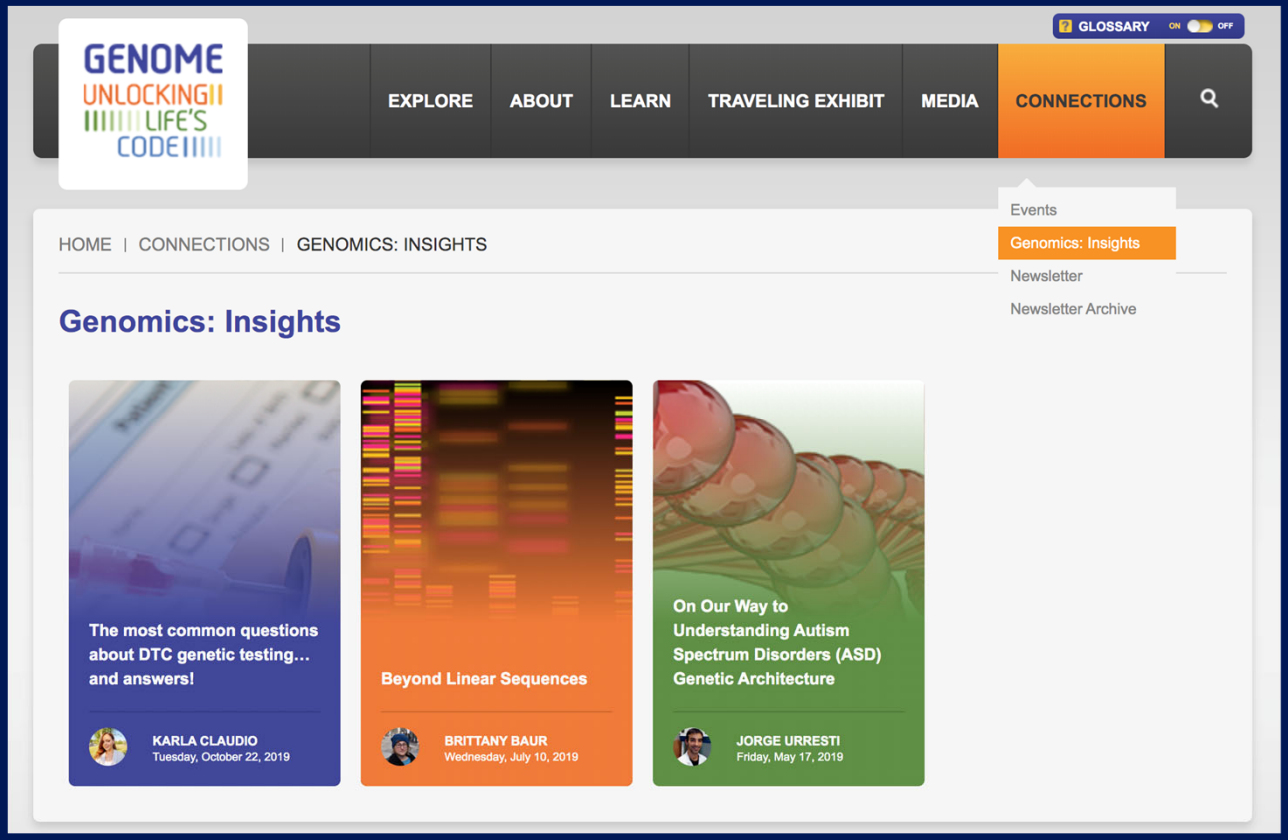
GENOME
UNLOCKING
LIFE'S
CODE

- **January 17 – April 12, 2020**

Museum of Science & History, Jacksonville, FL

Genome: Unlocking Life's Code Exhibition

New Website Feature "Genomics: Insights"



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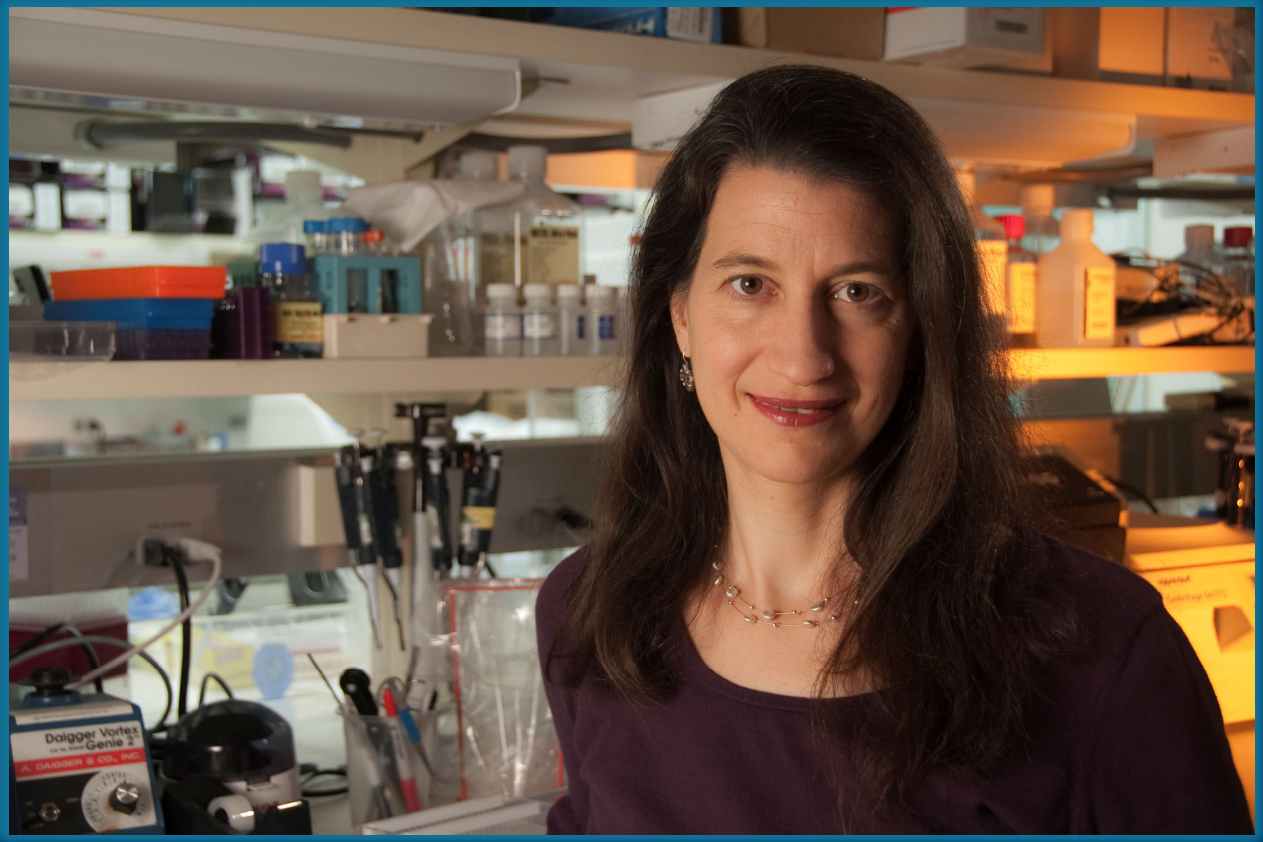
New NHGRI Clinical Director

Ben Solomon, M.D.

New NHGRI Intramural Precision Health Research Program

Les Biesecker, M.D.

Elected to National Academy of Medicine



Julie Segre, Ph.D.

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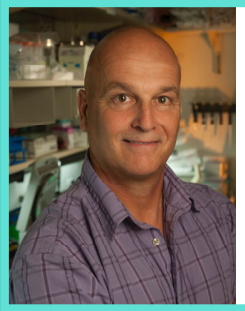
NHGRI Intramural Research Highlights

nature

Mutations that prevent caspase cleavage of RIPK1 cause autoinflammatory disease

Movement Disorders

Clinical evaluation of sibling pairs with Gaucher disease discordant for Parkinsonism



Journal of Clinical Medicine

NPC1 Deficiency in Mice is Associated with Fetal Growth Restriction, Neonatal Lethality and Abnormal Lung Pathology

The Genomics Landscape

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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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National Human Genome Research Institute

Thanks!

Special Thanks!


The Forefront
of Genomics®