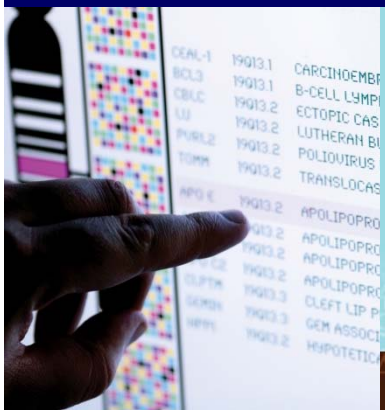
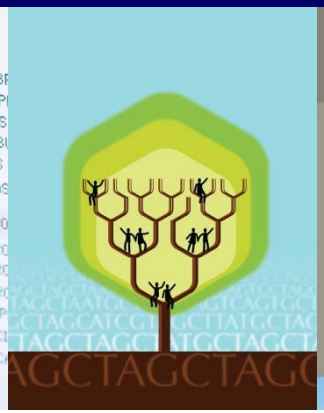
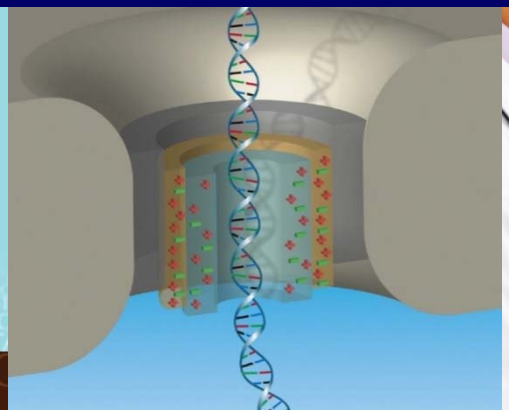
DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

May 2017

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

Director's Report-Related Documents: May 2017

Director's Report

Director's Report

No.	Relevant Documents
1	New Secretary, Department of Health and Human Services
2	Nominated Commissioner, Food and Drug Administration
3	H.R. 1313: Preserving Employee Wellness Programs Act
4	NIH Appropriations: Fiscal Year 2017
5	NIH Appropriations: Fiscal Year 2018
6	American Society for Microbiology William A. Hinton Research Training Award

genome.gov/DirectorsReport

Document #

Open Session Presentations

- Update from the National Institute of Mental Health

  Josh Gordon

- DataScience@NIH: Current State, Future Directions

  Patti Brennan

- Report: IGNITE and Beyond Workshop

  Chanita Hughes-Halbert

- Concept Clearance: IGNITE II

  Ebony Madden

Open Session Presentations

- Concept Clearance: Centers of Excellence in ELSI Research (CEER)

  Joy Boyer

- Report: Computational Genomics and Data Science Workshop

  Ajay Pillai

- NHGRI History of Genomics Program

  Chris Donohue

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

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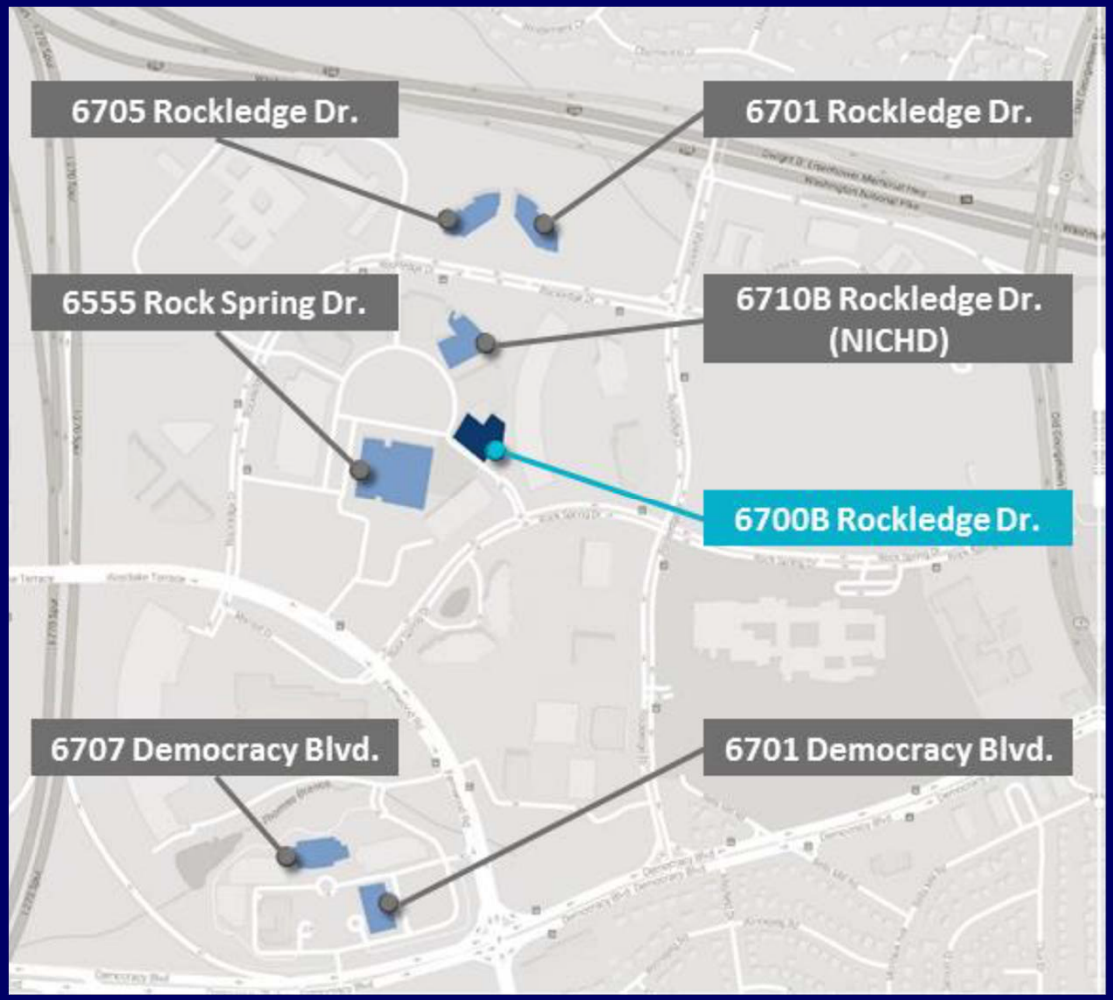
(Some of) NHGRI will be Moving

"Rock Spring Park Cluster"



NIH Components:

NHGRI	NIAAA
NEI	OD
ORS	NICHD
CSR	NLM
NHLBI	CIT
NCATS	NIAMS
NIDCR	NINR
NCCIH	NIBIB
NIDDK	NIMHD

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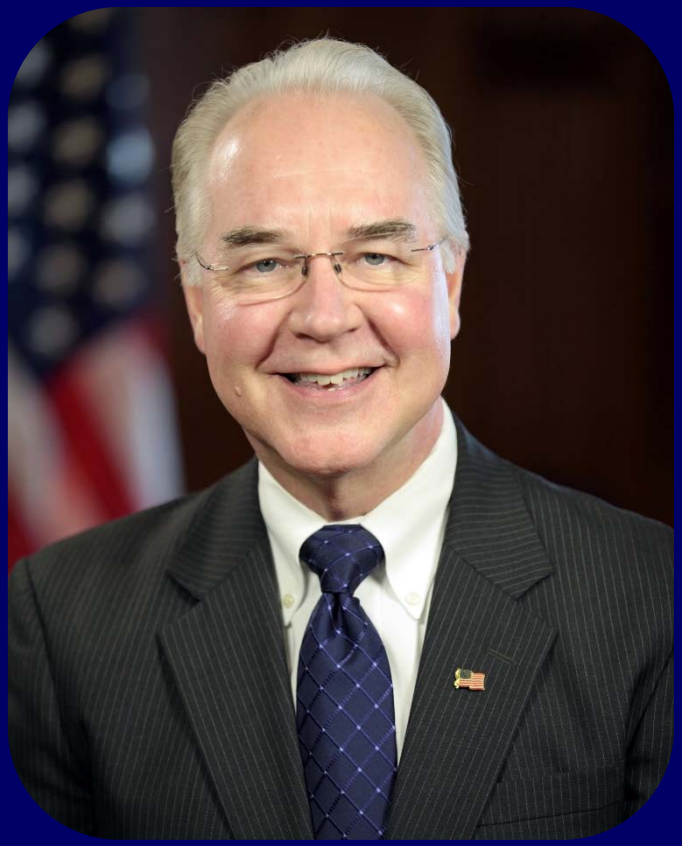
New Secretary, Department of Health and Human Services



Tom Price, M.D.

Document 1

Nominated Commissioner, Food and Drug Administration

Scott Gottlieb, M.D.

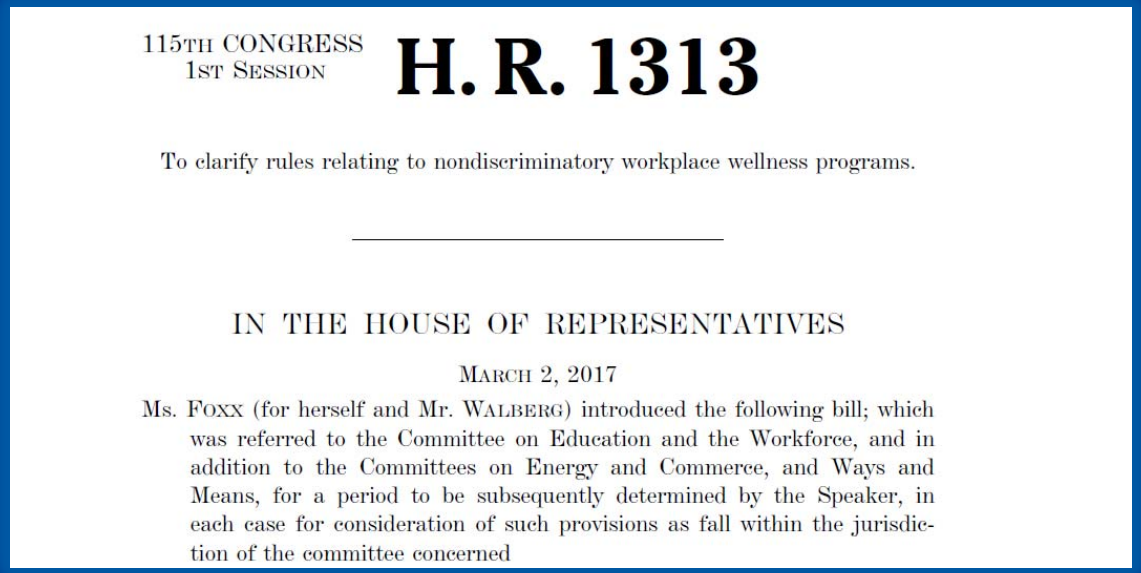
Document 2

David Lipman Departing as Director, National Center for Biotechnology Information

David Lipman, M.D.

Jim Ostell, Ph.D.

H.R. 1313: Preserving Employee Wellness Programs Act



115TH CONGRESS
1ST SESSION

H. R. 1313

To clarify rules relating to nondiscriminatory workplace wellness programs.

IN THE HOUSE OF REPRESENTATIVES

MARCH 2, 2017

Ms. FOXX (for herself and Mr. WALBERG) introduced the following bill; which was referred to the Committee on Education and the Workforce, and in addition to the Committees on Energy and Commerce, and Ways and Means, for a period to be subsequently determined by the Speaker, in each case for consideration of such provisions as fall within the jurisdiction of the committee concerned

- House sponsor: Virginia Foxx (R-NC)
- Would exempt workplace wellness programs from provisions of Americans with Disabilities Act and Genetic Information Nondiscrimination Act

Document 3

NIH Appropriations: Fiscal Year 2017

Entity	FY 16 Budget	FY 17 Budget
NIH	$32.1 B	$34.1 B
NHGRI	$512 M	$528 M

Document 4

NIH Appropriations: Fiscal Year 2018

	FY17 Budget	FY18 President's Budget
NIH	$34.1 B	$25.9 B

Document 5

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American Society for Microbiology
William A. Hinton Research Training Award

Steven Finkel, Ph.D.

AMERICAN SOCIETY FOR MICROBIOLOGY

Document 6

Acting President, Canadian Institute for Health Research

CIHR IRSC
Canadian Institutes of Health Research
Instituts de recherche en santé du Canada

Rod McInnes, M.D., Ph.D.

Document 7

Elected to National Academy of Sciences

Stephen Baylin

Karen Nelson

Sarah Tishkoff

NATIONAL ACADEMY
OF SCIENCES

Document 8

BioGenomics2017: Global Biodiversity Genomics Conference

- Diversity of species
- Diversity of biological questions
- Multiple data types
- Synergy with work in human genomics

Document 9

MIT Technology Review
Breakthrough Technologies for 2017

10 Breakthrough Technologies

2017

These technologies all have staying power. They will affect the economy and our politics, improve medicine, or influence our culture. Some are unfolding now; others will take a decade or more to develop. But you should know about all of them right now.

The Cell Atlas

Biology's next mega-project will find out what we're really made of.

Gene Therapy 2.0

Scientists have solved fundamental problems that were holding back cures for rare hereditary disorders. Next we'll see if the same approach can take on cancer, heart disease, and other common illnesses.

Document 10

Genomics In The News…

PUTTING GENOMES TO WORK IN AFRICA

Investment promises to bring precision medicine to Africans. But will it help?

BY LINDA NORDLING

It took a public-health disaster for the Zimbabwean government to recognize the power of precision medicine. In 2015, the country switched from a standard three-drug cocktail for HIV to a single-pill combination therapy that was cheaper and easier for people to take every day. The new drug followed a World Health Organization recommendation to incorporate the antiretroviral drug efavirenz as a first-line therapy for public-health programmes. But as tens of thousands of Zimbabweans were put onto the drug, reports soon followed about people quitting it in droves.

Collen Masimirembwa, a geneticist and founding director of the African Institute of Biomedical Science and Technology in Harare, was not surprised. In 2007, he had shown that a gene variant carried by many Zimbabweans slows their ability to break down efavirenz1. For those with two copies of the variant — about 20% of the population — the drug accumulates in the bloodstream, leading to hallucinations, depression and suicidal tendencies. He had tried to communicate this to his government, but at the time efavirenz was not a staple of the country's HIV programme, and so the health ministry ignored his warnings.

Masimirembwa continued to publish his research, but the authorities took no heed until there was trouble. A lot of confusion could have been avoided if the government had listened, he says. "It's not a bad drug. We just know it can be improved in Africa."

Masimirembwa is a rare breed. Although scientists worldwide have been pushing for ways to improve health care by tailoring diagnostics and treatment to the environment, lifestyle and genes of individual patients, few researchers have taken this precision-medicine approach in Africa.

That may be changing. In the past five years, international research-funding organizations have invested more than US$100 million in projects to boost genetic research on people in Africa. These studies could lead to improved treatments for Africans as well as for people of recent African descent in Europe and the Americas, who tend to experience more ill health than other ethnicities — a situation that is often attributed to socioeconomic challenges, but which some scientists say could also have genetic roots.

Although few would question the importance of African genomics, opinions differ on whether this will translate into better care. Globally, precision medicine has failed to live up to its promise, even in countries that spend lots of money on health. And some argue that the money spent on investigating genes should instead be used to improve basic health care on the continent.

Many African scientists bristle at that simple calculus. They are frustrated that they have been left out of research on everything from health to human origins — a field that has particularly benefited from African genome data — and they want Africans to gain from the work. For Masimirembwa and others, the money presents an opportunity to take control of how genetic data are collected and used. "Unless capacity is built on the continent, Africans won't have a chance to participate," he says.

POPULATION-SCALE PRECISION

There's a big problem, however. Precision medicine is expensive. For a continent that, for the most part, struggles to provide even basic health care, tailor-made treatments for individual patients may seem like an unaffordable luxury.

Enter 'precision public health' — a new approach to precision medicine that bases health decisions on populations and communities rather than on individuals. It would use genomic insights into a country's population to inform general treatment programmes. For instance, a country might tweak its essential medicines list that specifies the drugs it buys in bulk at reduced rates from pharmaceutical companies, to avoid medicines that are known to cause problems in its population.

This is already happening in some places. Botswana — a middle-income country — stopped using the three-in-one drug containing efavirenz in 2016, opting instead for a newer and better-performing, but more expensive, drug called dolutegravir. The gene variant that causes problems with efavirenz is common in Botswana — around 13.5% of the population has two copies of it. And in 2015, Ethiopia banned the use of the painkiller codeine, because a high proportion of people in

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Genomes In The News…

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Genome Sequencing Program

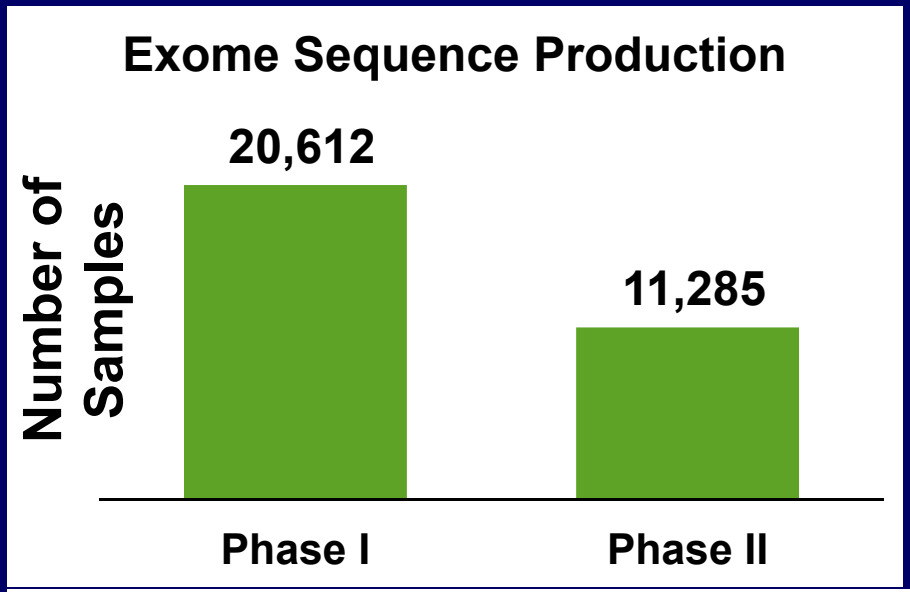
Centers for Common Disease Genomics

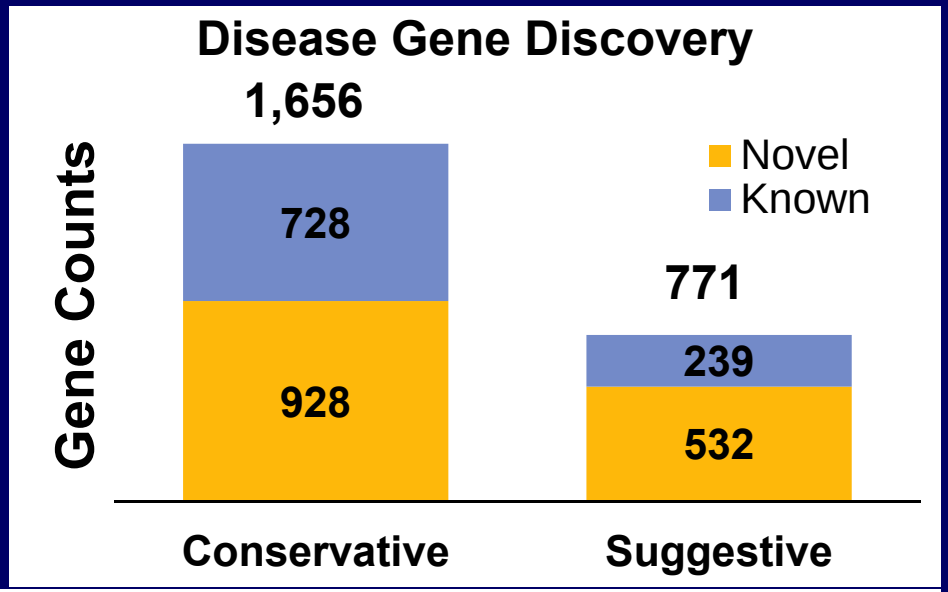
Disease Category	Sample Type	Sequenced Samples	Approved Samples
Cardiovascular	Genomes	16,096	46,300
	Exomes	10,861	21,000
Immune-Mediated	Genomes	4,718	25,000
	Exomes	0	2,000
Neuropsychiatric	Genomes	8,670	15,300
	Exomes	11,374	32,000
TOTAL		51,719	141,600

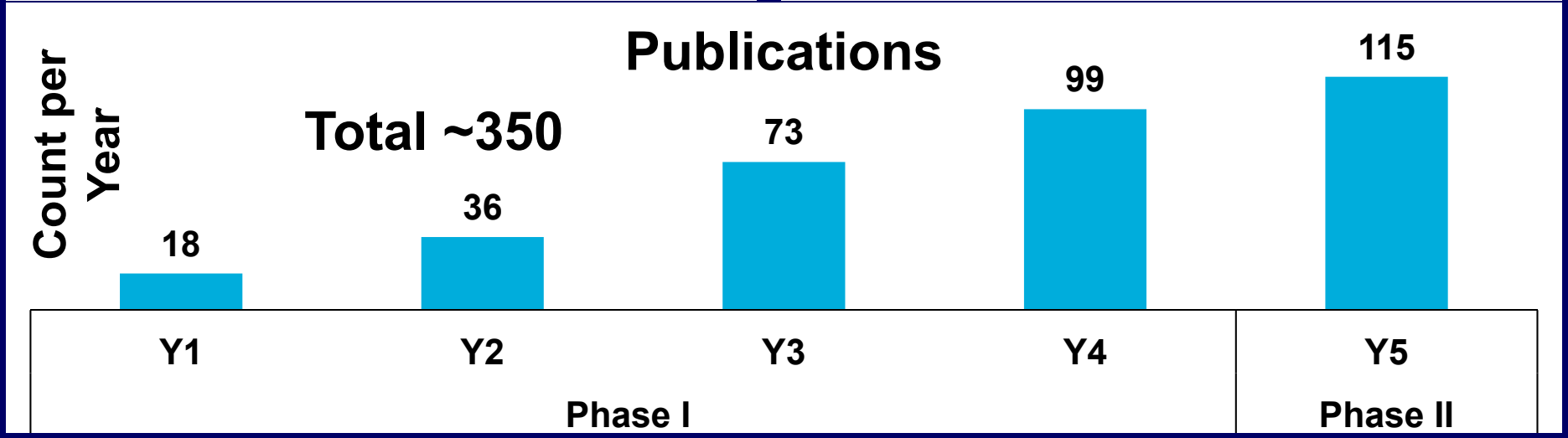
Document 13

Genome Sequencing Program

Centers for Mendelian Genomics







Document 13

Genome Sequencing Program

2017 Consortium Meeting

- Progress updates
- Collaborative analysis plans
- Data sharing and distribution
- New research directions

Technology Development Program

- **Advanced Genomic Technology Development Meeting – May 22-25**
- **Novel Nucleic Acid Sequencing Technology Development**

  RFA-HG-15-031 (R01, also linked R21 and R43/44)

  Next due date: June 15, 2017
- **Novel Genomic Technology Development**

  PAR-16-14 (R01, also linked R21 and R43/44)

  Next due date: October 31, 2017

Document 14

ENCyclopedia Of DNA Elements (ENCODE)

PsychENCODE

NIH National Institute of Mental Health

Program Announcements

Announcement Number	Primary IC	Title	Release Date	Expiration Date
PAR-17-258	NIMH	PsychENCODE: Non-coding Functional Elements in the Human Brain and their Role in the Development of Psychiatric Disorders (Collaborative U01)	2017-04-18	2019-06-07
PAR-17-257	NIMH	PsychENCODE: Non-coding Functional Elements in the Human Brain and their Role in the Development of Psychiatric Disorders (U01)	2017-04-18	2019-06-07

Centers of Excellence in Genomic Science (CEGS)

Two recent CEGS awards:

Center for Cell Circuits
Broad Institute – Aviv Regev

Center for Dynamic RNA Epitranscriptomes
University of Chicago – Chuan He

ALLIANCE of GENOME RESOURCES

AGR Community Meeting

- Share AGR plans with the community and review progress
- Participants:
  - Model organism researchers
  - AGR investigators and key personnel
  - Scientific Advisory Board members
  - NIH staff

Document 17

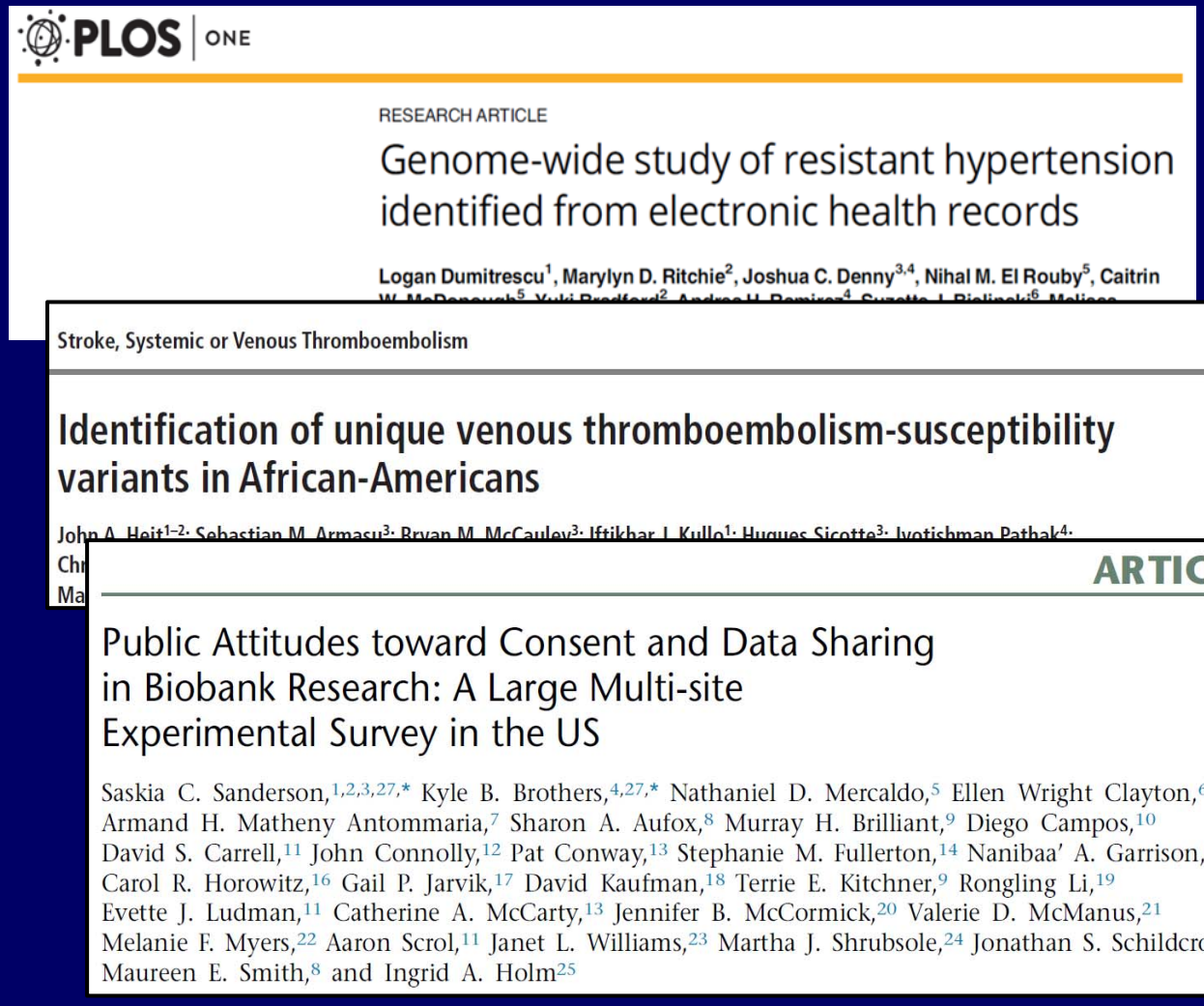
NHGRI 'Sandbox' Update

- Resource to democratize genomic data access, sharing, and computing
- Cooperative agreement funding mechanism
- $5M per year for 5 years

Document 18

eMERGE network

ELECTRONIC MEDICAL RECORDS AND GENOMICS



- **35** abstracts accepted for 2017 ACMG
- **16** abstracts accepted for 2017 AMIA

Clinical Genome Resource (ClinGen)

Evaluating the clinical validity of gene-disease associations: an evidence-based framework developed by the Clinical Genome Resource

Natasha T Strande, Erin Rooney Riggs, Adam H. Buchanan, Ozge Ceyhan-Birsoy, Selina T. Dwight, Selina S. Dwight, Jennifer L. Goldstein, Rajarshi Ghosh, Bryce A. Seifert, Tam P Sneddon, Matt W. Wright, Laura V. Milko, Monica A. Giovanni, Michael F. Murray, Julianne M. O'Daniel, Erin M. Ramos, Avni B. Santani, Alan F. Scott, J. Michael Cherry, Sharon E. Plon, Heidi L. Rehm

doi: https://doi.o

This article is a pre

bioRxiv beta

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Genetics inMedicine Official Journal of the American College of Medical Genetics

Home | Current Issue | Archive | Podcasts | For Authors & Referees | About the journal

Advance Online Publication > Article

GENETICS IN MEDICINE | ORIGINAL RESEARCH ARTICLE

Clinical laboratories collaborate to resolve differences in variant interpretations submitted to ClinVar

Steven M. Harrison PhD, Jill S. Dolinsky MS, Amy E. Knight Johnson MS, Tina Pesaran MA, MS, Danielle R. Azzariti MS, Sherri Bale PhD, Elizabeth C. Chao MD, Soma Das PhD, Lisa Vincent PhD & Heidi L. Rehm PhD

AMERICAN SOCIETY OF HEMATOLOGY

ClinGen Expert Panels in Hematology

Curating the Clinical Genome Meeting 2017

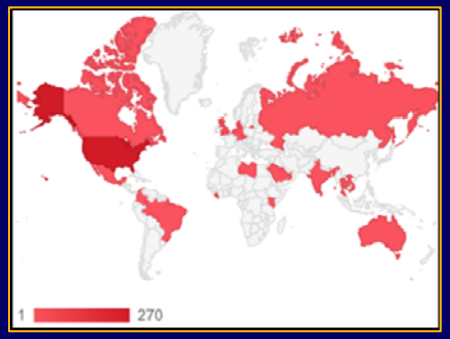
ClinGen Clinical Genome Resource

DECIPHER

June 28-30, Washington DC

Clinical Sequencing Exploratory Research Program

- Enrolled 5,241 adults and 1,357 children
- 312 publications, 20 working group publications



Document 21

Clinical Sequencing Exploratory Research Program

cser
Clinical Sequencing Exploratory Research

cser
Clinical Sequencing Exploratory Research

Guide to Interpreting Genomic Reports: A Genomics Toolkit

A guide to genomic test results for non-genetics providers

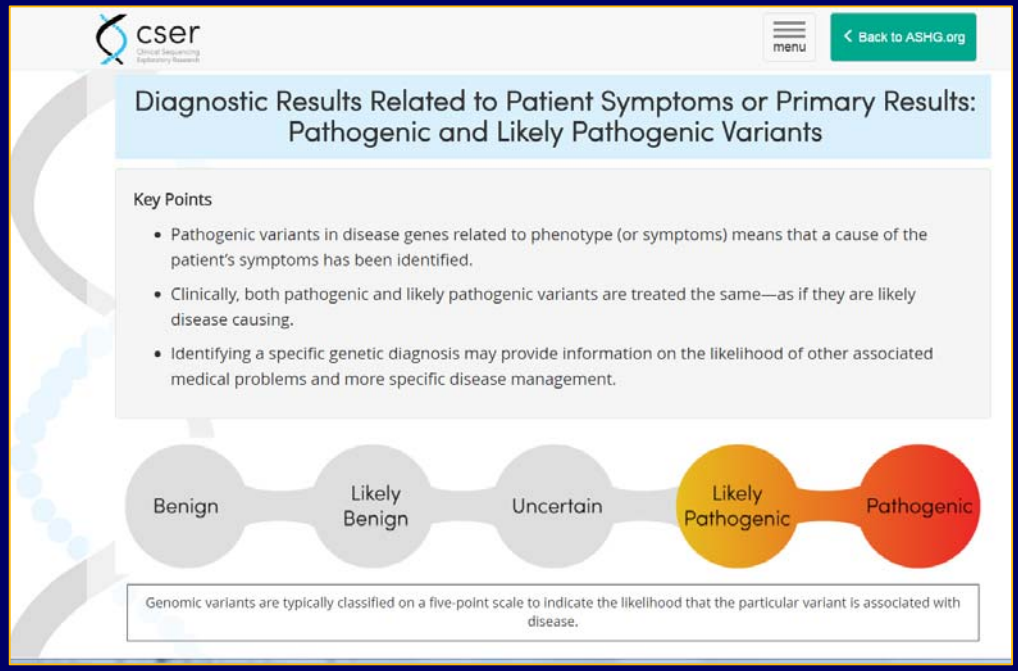
Created by the Practitioner Education Working Group of the Clinical Sequencing Exploratory Research (CSER) Consortium

Glossary

Autosomal recessive: genetic conditions that occur only when mutations are present in both copies of a given gene (i.e., the person is homozygous for a mutation, or carries two different mutations of the same gene, a state referred to as compound heterozygosity).
[Source – NCI Dictionary of Genetics Terms]

Benign (variant): an alteration in a gene distinct from the normal, wild-type allele that does.
[Source – Illustrated Glossary]

Carrier frequency: the proportion of individuals in a population who have a single copy of a specific recessive gene mutation; also sometimes applied to the prevalence of mutations in dominantly acting genes such as BRCA1 and BRCA2. Also called carrier rate.
[Source – NCI Dictionary of Genetics Terms]



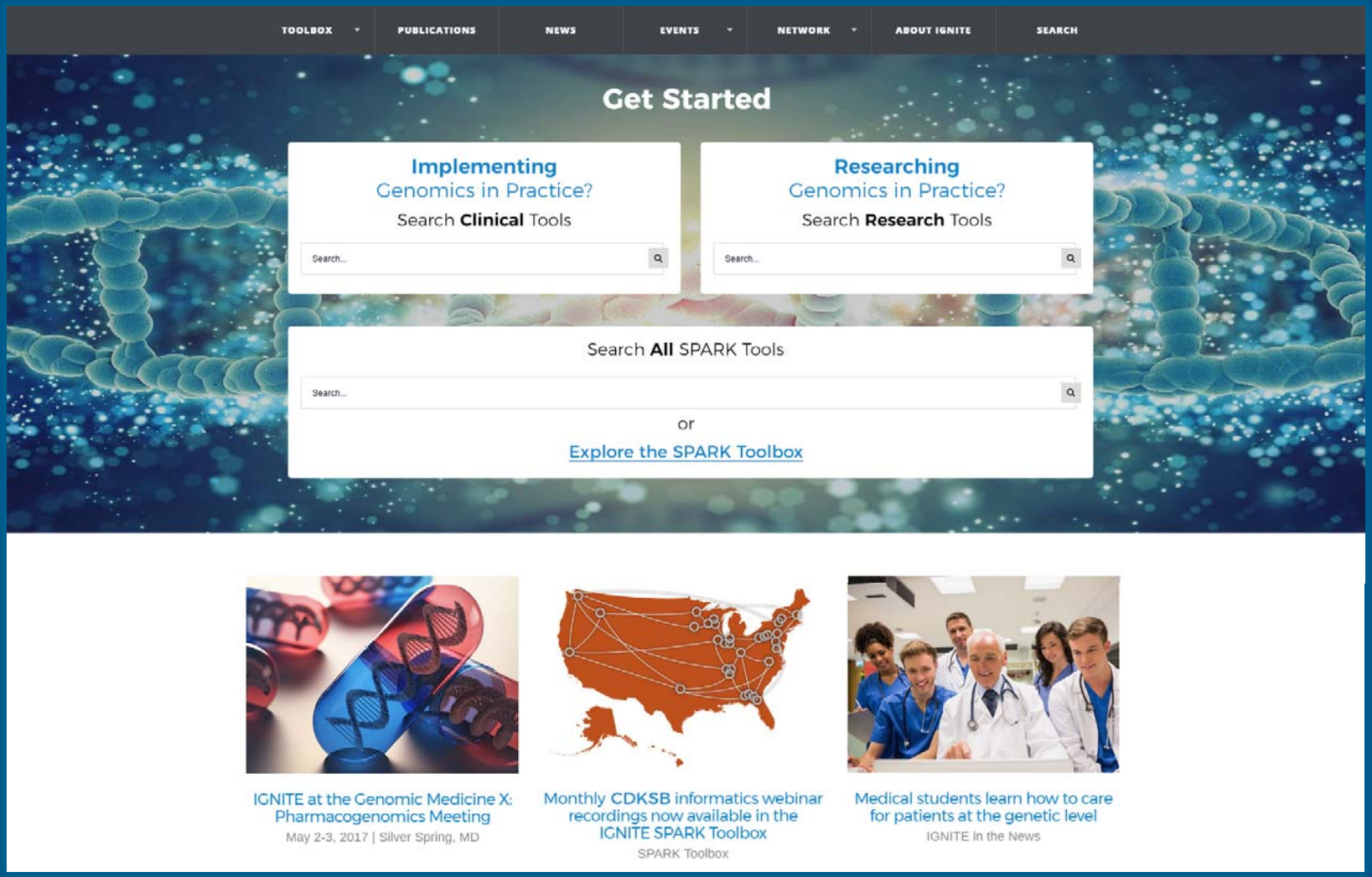
Document 21

Implementing Genomics In Practice (IGNITE) Network

92
publications

259
presentations

Implementing Genomics In Practice (IGNITE) Network



Document 22

4th ELSI Congress: Expanding the ELSI Universe

June 5-7, 2017 (Farmington, Connecticut)

- **Keynote and Plenary Speakers:** Eric Dishman, Alondra Nelson, Wylie Burke, James Evans, and Pearl O'Rourke
- **Topics:** Precision medicine; genome sequencing in the clinic; and genes, ancestry, and identity
- **Program and registration available online**

The Jackson Laboratory

Columbia University Medical Center

UCONN HEALTH

NIH National Human Genome Research Institute

Document 23

Genomics and Society Working Group

Outgoing Members:

- Arti Rai
- Lisa Parker

Current Members:

- Gail Henderson
- Chanita Hughes-Halbert
- Dave Veenstra

New Members:

Jeff Botkin

Steve Joffe

Max Mehlman

Melanie Myers

Sandra Soo-Jin Lee

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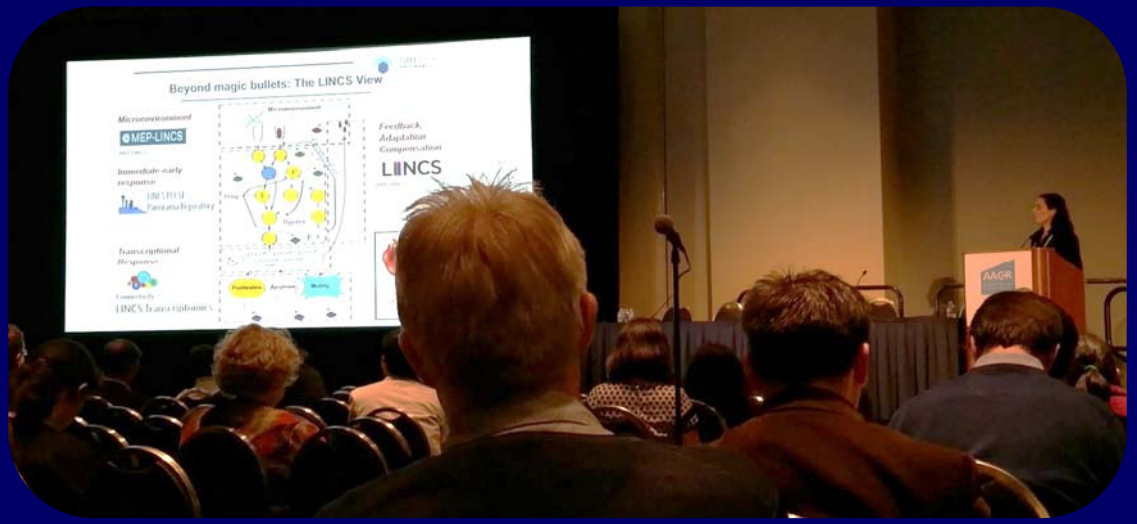
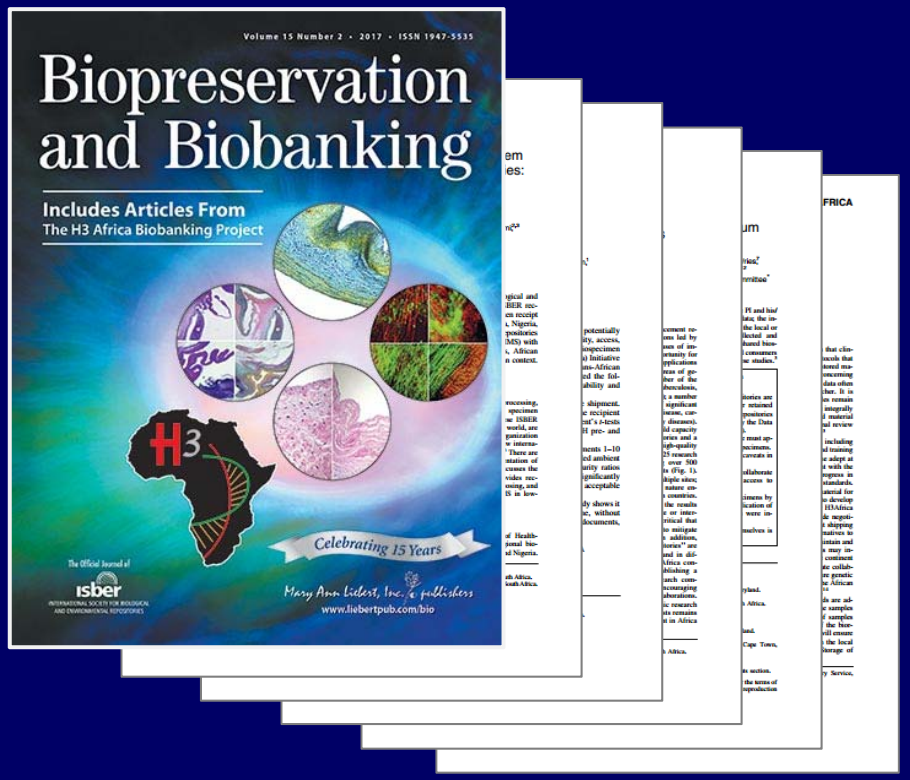
NIH LINCS PROGRAM

- Midcourse review in January
- Recommendations for enhancing utility of data:
  - Generate transcriptomics data at each center
  - Hire joint postdocs focused on integrative analysis
- Satellite session at April AACR meeting



Document 25

Human Heredity and Health in Africa (H3Africa)

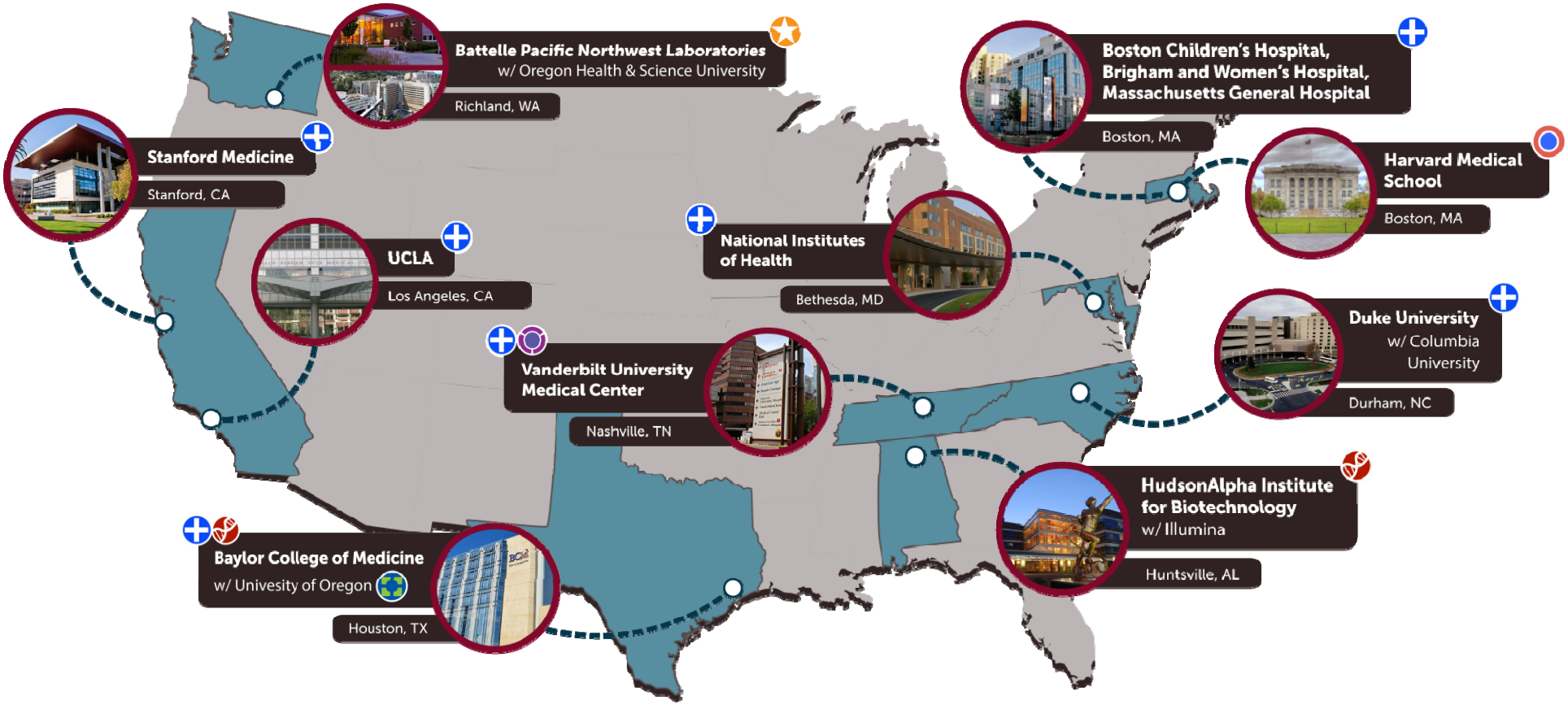


- 10th Consortium Meeting, May 12-15 (Botswana)

Document 26

Undiagnosed Diseases Network (UDN)

Undiagnosed Diseases Network | UDN Site Locations



1,387 Applications
545 Acceptances

APPLY

Document 27

Undiagnosed Diseases Network (UDN)

AJHG

COMMENTARY

(Feb 2, 2017)

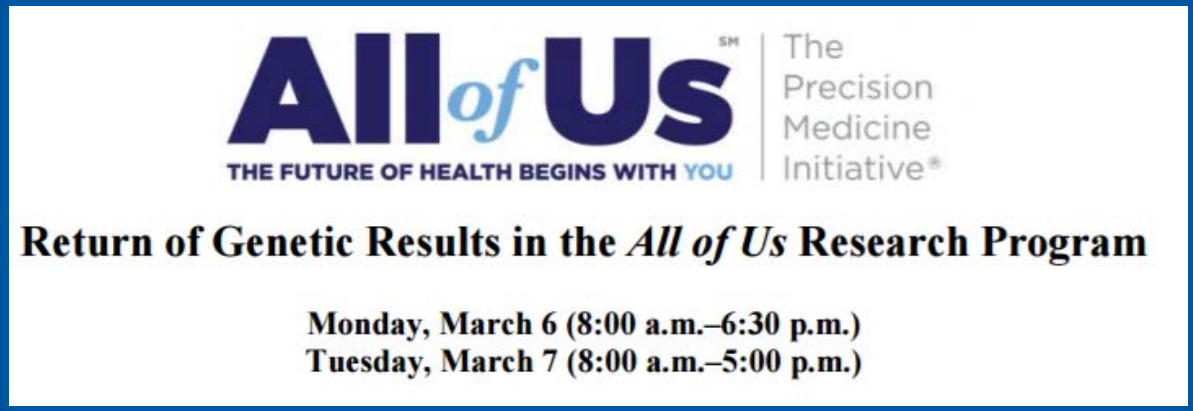
The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease

Rachel B. Ramoni,[1,2,*] John J. Mulvihill,[3] David R. Adams,[3] Patrick Allard,[4,5] Euan A. Ashley,[6] Jonathan A. Bernstein,[7] William A. Gahl,[3] Rizwan Hamid,[8] Joseph Loscalzo,[9] Alexa T. McCray,[1] Vandana Shashi,[10] Cynthia J. Tifft,[3] Undiagnosed Diseases Network, and Anastasia L. Wise[3]

Diagnosis at the edges of our knowledge calls upon clinicians to be data driven, cross-disciplinary, and collaborative in unprecedented ways. Exact disease recognition, an element of the concept of precision in medicine, requires new infrastructure that spans geography, institutional boundaries, and the divide between clinical care and research. The National Institutes of Health (NIH) Common Fund supports the Undiagnosed Diseases Network (UDN) as an exemplar of this model of precise diagnosis. Its goals are to forge a strategy to accelerate the diagnosis of rare or previously unrecognized diseases, to improve recommendations for clinical management, and to advance research, especially into disease mechanisms. The network will achieve these objectives by evaluating patients with undiagnosed diseases, fostering a breadth of expert collaborations, determining best practices for translating the strategy into medical centers nationwide, and sharing findings, data, specimens, and approaches with the scientific and medical communities. Building the UDN has already brought insights to human and medical geneticists. The initial focus has been on data sharing, establishing common protocols for institutional review boards and data sharing, creating protocols for referring and evaluating patients, and providing DNA sequencing, metabolomic analysis, and functional studies in model organisms. By extending this precision diagnostic model nationally, we strive to meld clinical and research objectives, improve patient outcomes, and contribute to medical science.

Document 27

All of Us Research Program
Return of Genetic Results Workshop



- Topics related to returning genomic results at scale
- Attendees represented broad range of perspectives
- Half the panelists were NHGRI grantees

Document 28

Chief Engagement Officer, *All of Us* Research Program

All of Us
THE FUTURE OF HEALTH BEGINS WITH YOU
The Precision Medicine Initiative

Dara Richardson-Heron, M.D.

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23andMe Receives Approval to Return Risk Information for 10 Conditions

FDA News Release

FDA allows marketing of first direct-to-consumer tests that provide genetic risk information for certain conditions

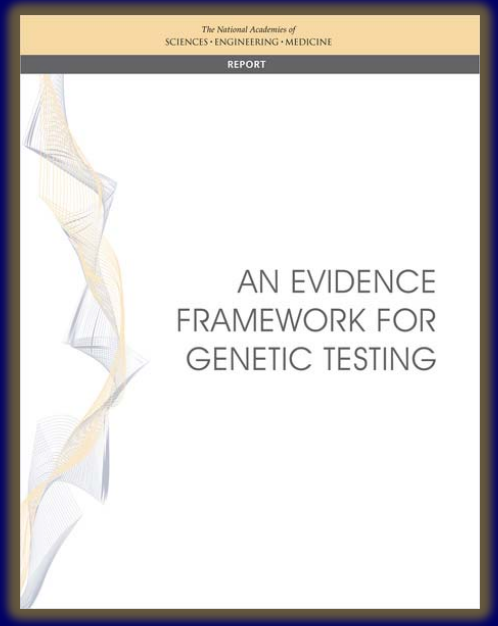
SHARE | TWEET | LINKEDIN | PIN IT | EMAIL | PRINT

For Immediate Release

April 6, 2017

Document 29

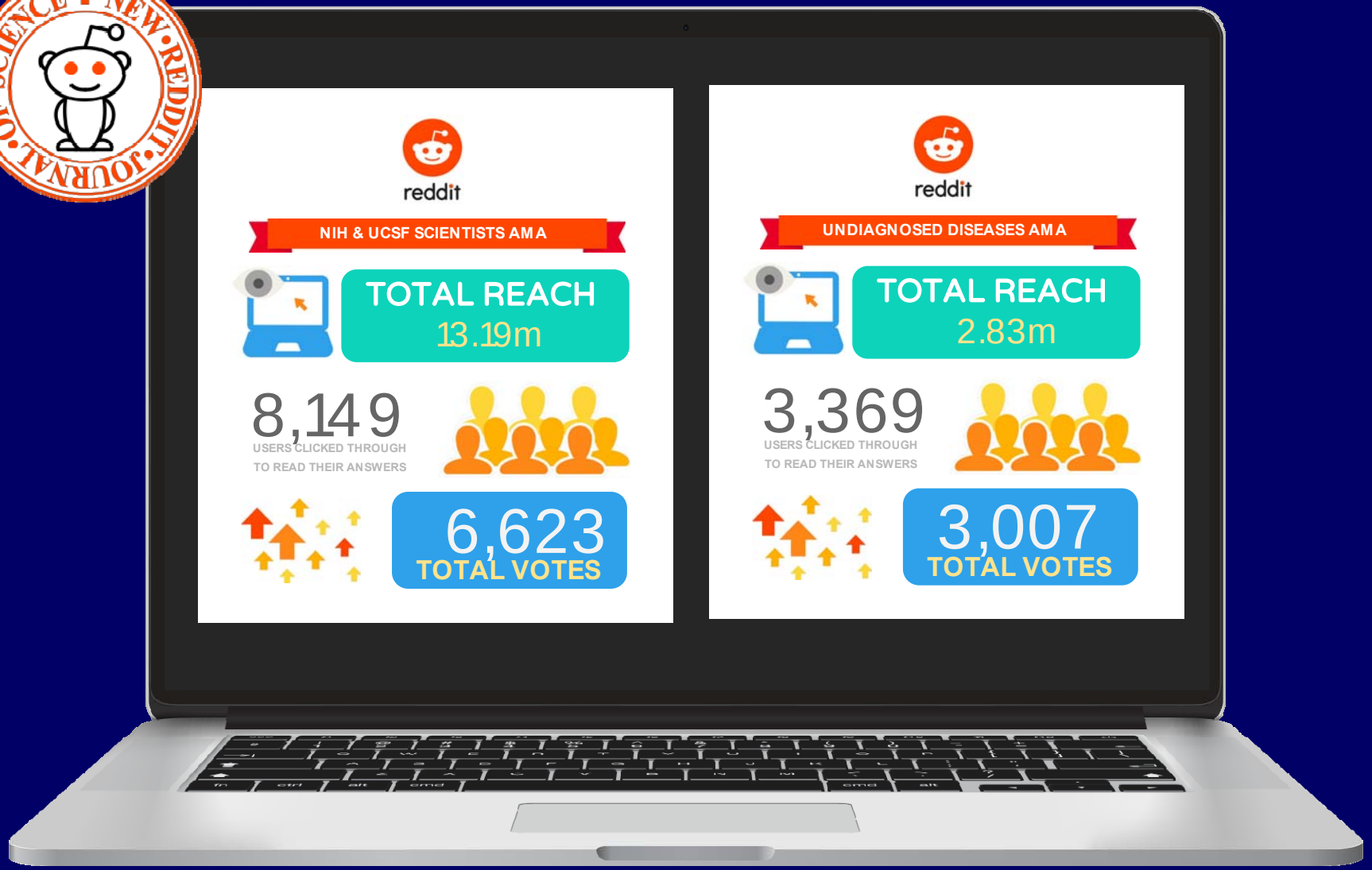
National Academies Report on Evidence for Genetic Testing



- Commissioned by the Department of Defense
- Provides recommendations for evidence-based decisions on coverage for genetic testing

Document 30

NHGRI Participates in Reddit's "Ask Me Anything" Series



Document 31

Genome: Unlocking Life's Code Exhibition

Travel Schedule

2017

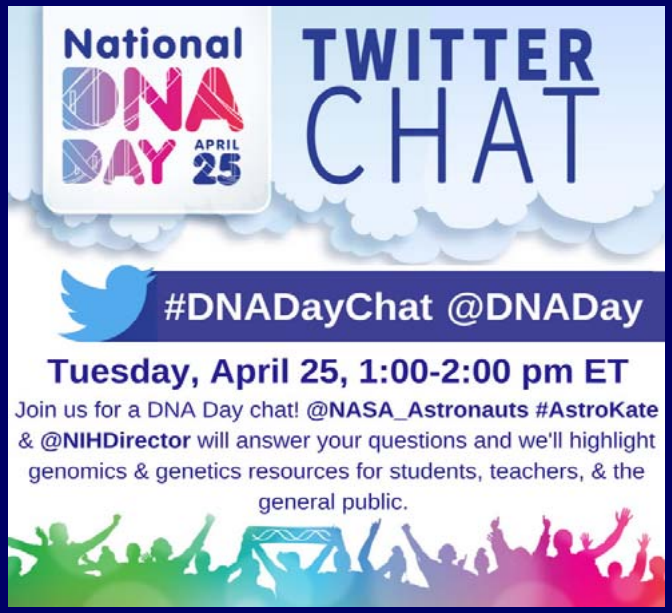
April 1-May 29
Peoria Riverfront Museum
Peoria, IL

June 12-September 11
Health Museum
Houston, TX

September 30-January 1
Science North
Sudbury, Ontario, Canada

Document 32

NHGRI & National DNA Day



Genomic Literacy, Education, and Engagement (GLEE) Initiative

Community Health Studio at Gallaudet University

GALLAUDET
UNIVERSITY

Inter-Society Coordinating Committee for Practitioner Education (ISCC)

6th In-Person Meeting

- Share members' genomic education activities, needs, and interests
- Learn about opportunities and needs for funding provider education activities
- Included presentations about activities in other countries
- Establish a new "Building Bridges" working group

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Elected to the Association of American Physicians

Ellen Sidransky, M.D.

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New Chief, Social and Behavioral Research Branch

Laura Koehly, Ph.D.

Social and Behavioral Research Branch
National Human Genome Research Institute

NHGRI Intramural Research Highlights

AJMG AMERICAN JOURNAL OF medical genetics

22q11.2 deletion syndrome in diverse populations

nature genetics

Single-molecule sequencing and chromatin conformation capture enable de novo reference assembly of the domestic goat genome

PIGMENT CELL & MELANOMA Research

Hypoxia-induced HIF1α targets in melanocytes reveal a molecular profile associated with poor melanoma prognosis

Science

Genetic structure of the purebred domestic dog

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Search for NHGRILANDSCAPE

Past issues can be accessed at: genome.gov/27541196

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National Human Genome Research Institute

Advancing human health through genomics research

Thanks!

Special Thanks!


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