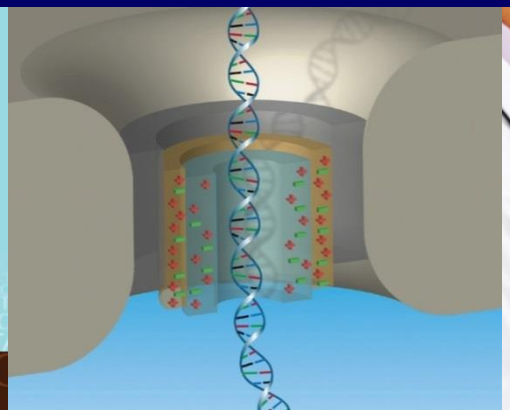
DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

September 2017

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





Director's Report-Related Documents: September 2017

Director's Report

Director's Report

No.	Relevant Documents
1	NIH-ACMG Fellowship in Genomic Medicine Program Management
2	Genomics and Health Disparities Lecture Series
3	Genomics and Health Disparities Scientific Interest Group
4	Francis Collins 'Retained' as NIH Director
5	New NIH Deputy Director for Management
6	Naming of New Director, National Cancer Institute
7	First Director, NIH Tribal Health Research Office

genome.gov/DirectorsReport

Document #

Open Session Presentations

- Next Generation Researchers Initiative

  Larry Tabak

- Report: Update on the eMERGE Network

  Rex Chisholm

- Report: The Cancer Genome Atlas (TCGA): A Decade of Discovery

  Carolyn Hutter

Open Session Presentations

- **Report: NAM Report on Evidence Framework for Genetic Testing**
  Wendy Chung

- **Report: Genomic Medicine Working Group Activities in 2017**
  Teri Manolio

- **Report: Update on the NSIGHT Program**
  Anastasia Wise

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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Retirement of NHGRI Program Director

Jean McEwen, J.D., Ph.D.

NIH-ACMG Fellowship in Genomic Medicine Program Management

NIH National Institutes of Health

ACMG American College of Medical Genetics and Genomics
Translating Genes into Health®

- Increase pool of physicians who can manage research and implementation programs in genomic medicine
- First fellow (Jennifer Krupp) started this month
- Applications for 2018 fellowship due December 1; two-year fellowship begins July 2018

Document 1

Genomics and Health Disparities Lecture Series

Herman Taylor, Jr., M.D., M.P.H
September 19, 2017
Director, Cardiovascular Research Institute
Morehouse School of Medicine

Richard Cooper, M.D.
November 7, 2017
Professor and Chair, Public Health Sciences
Loyola University Medical School

Jose Florez, M.D., Ph.D.
February 22, 2018
Chief, Diabetes Unit and Investigator, Center for Genomic Medicine
Massachusetts General Hospital
Harvard Medical School

Document 2

Genomics and Health Disparities Scientific Interest Group

Vence Bonham, J.D.
NHGRI

Hannah Valantine, M.D.
NIH OD and NHLBI

Sara Hull, Ph.D.
NIH Clinical Center and NHGRI

Gary Gibbons, M.D.
NHLBI

Rasika Mathias, Ph.D.
Johns Hopkins University

Timothy Thornton, Ph.D.
University of Washington

October 11, 2017 at 3:00 pm EST

Wylie Burke, M.D., Ph.D.
University of Washington

Charles Rotimi, Ph.D.
NHGRI

Document 3

Genomic Data Science Working Group

NIH National Human Genome Research Institute

Eric Boerwinkle	*Current Council Member*
Lon Cardon	*Former Council Member*
George Hripcsak	
Trey Ideker	*Current Council Member*
Gail Jarvik	
Mark Johnston	*Current Council Member*
Nancy Cox	
Michael Boehnke	*Former Council Member*
Anthony Philippakis	

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Francis Collins ‘Retained’ as NIH Director

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

NIH
National Institutes of Health

Document 4

New NIH Deputy Director for Management

Alfred Johnson, Ph.D.

NIH
National Institutes of Health

Document 5

Naming of New Director, National Cancer Institute

Norman Sharpless, M.D.

NIH

NATIONAL CANCER INSTITUTE

Document 6

First Director,
NIH Tribal Health Research Office

NIH National Institutes of Health
Tribal Health Research Office

David Wilson, Ph.D.

Josie Briggs Departs as Director, National Center for Complementary and Integrative Health

Josephine Briggs, M.D.

NIH NCCIH

New Commissioner, U.S. Food and Drug Administration

Scott Gottlieb, M.D.

New Director, U.S. Centers for Disease Control and Prevention

CDC

CENTERS FOR DISEASE CONTROL AND PREVENTION

Brenda Fitzgerald, M.D.

New U.S. Surgeon General

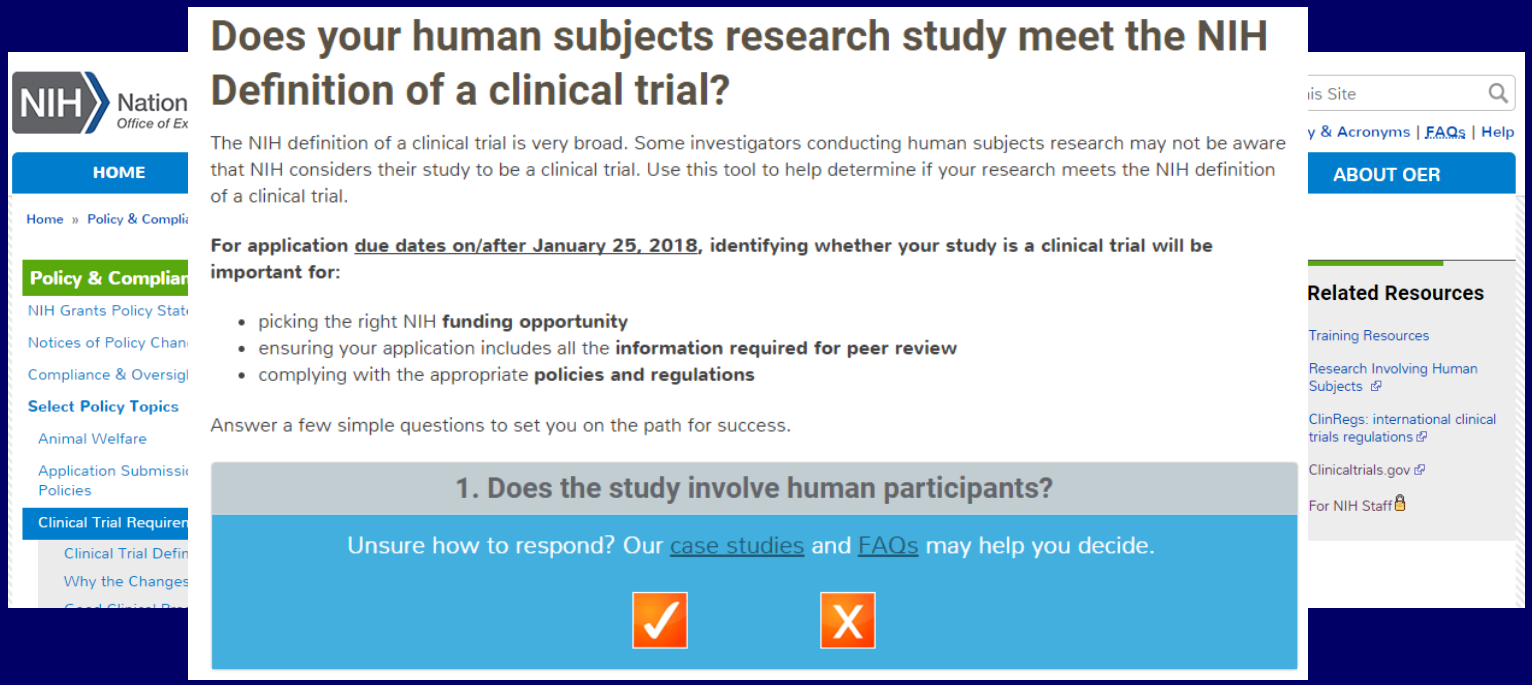
Jerome Adams, M.D.

Alberto Gutierrez Retiring as Director, FDA Office of In Vitro Diagnostics and Radiological Health

Alberto Gutierrez, Ph.D.

Document 12

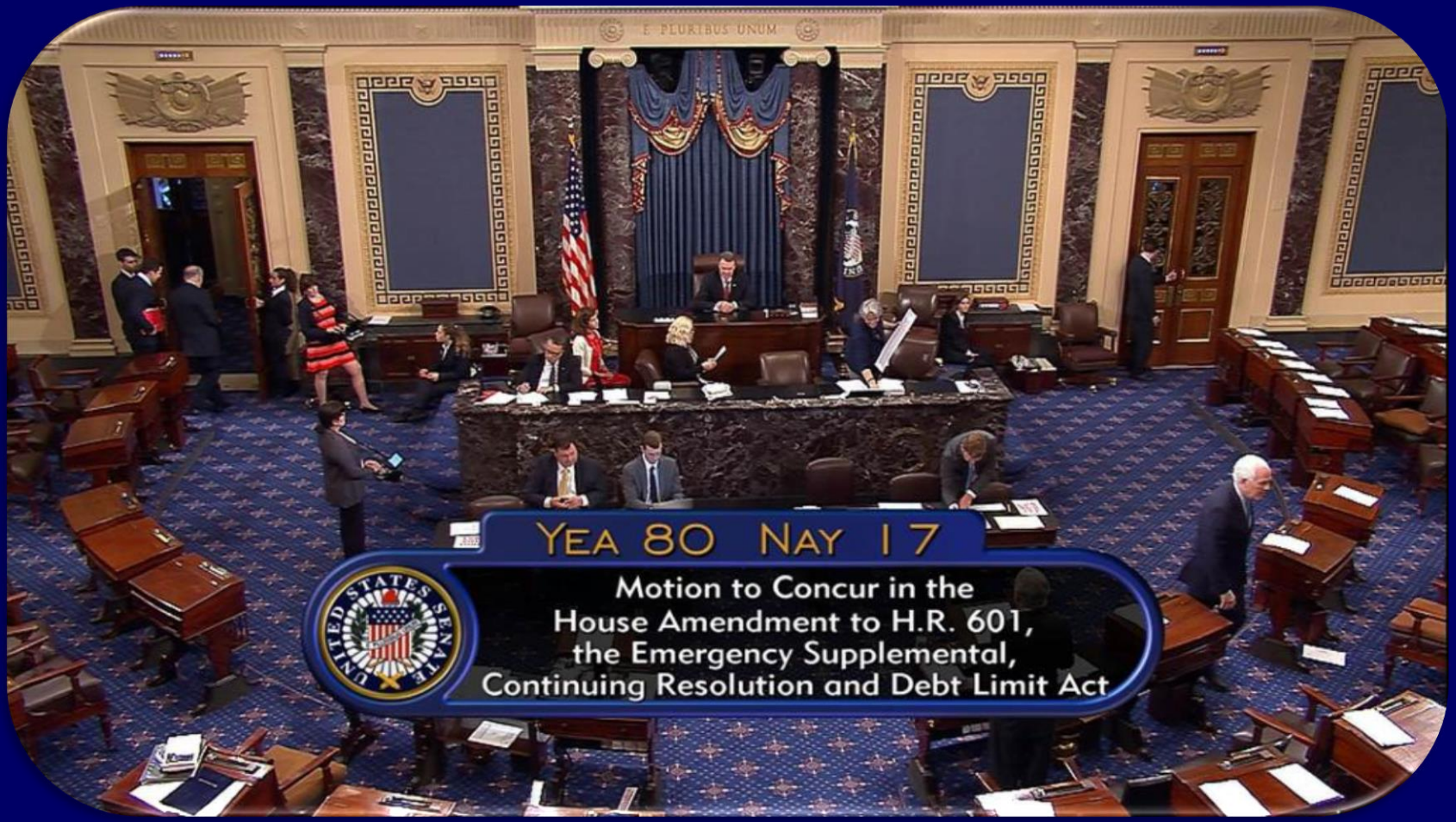
Upcoming Changes to NIH Clinical Trials Policies



- Upcoming policy changes aim to improve stewardship of NIH-funded clinical trials
- New requirements go into effect for applications due on or after January 25, 2018

Document 13

Budget Update



NIH Appropriations

	Fiscal Year 2017 Budget	Fiscal Year 2018 Senate Appropriations Labor-HHS Spending Bill
NIH	$34.1 B	$36.1 B
NHGRI	$528 M	$547 M

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Global Genomic Medicine Collaborative (G2MC)

Athens, April 2017

- Leading effort to convene major large-scale cohort studies and promote interoperability and sharing
- Meeting planned for early 2018

Document 15

New Executive Director, American Society of Human Genetics

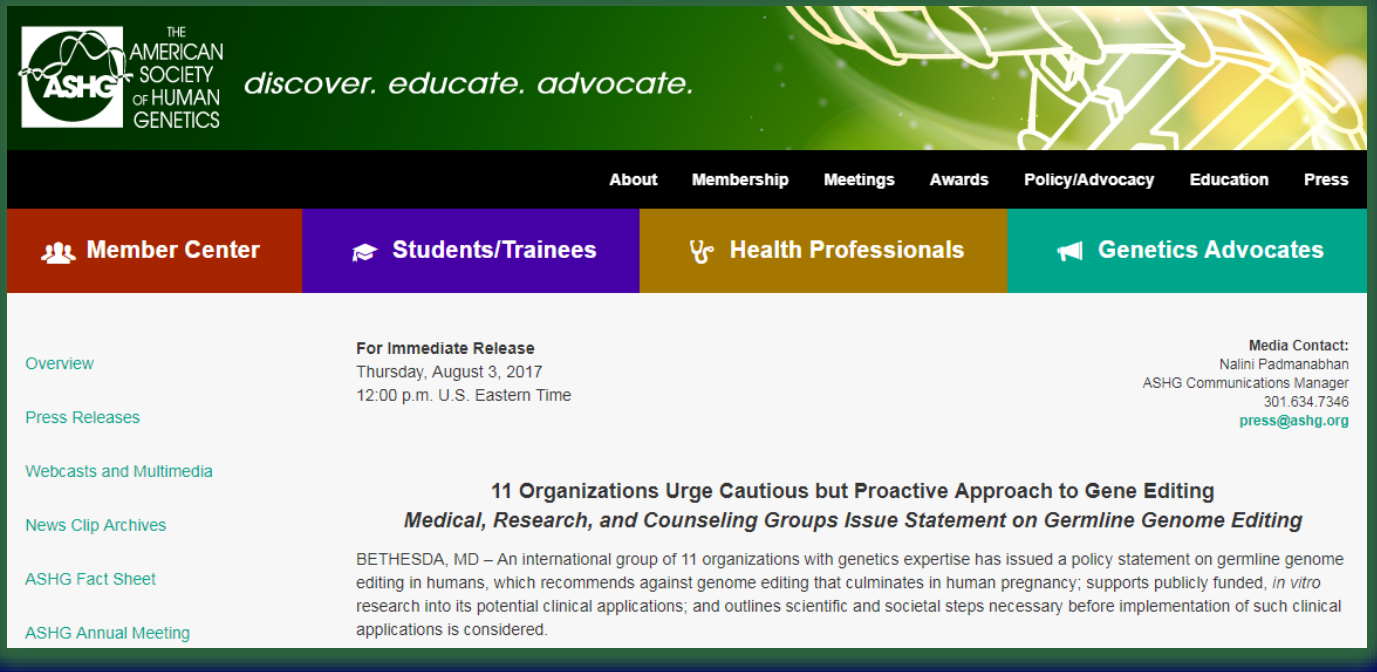
Mona Miller, M.P.P.

THE AMERICAN SOCIETY OF HUMAN GENETICS

ASHG

Document 16

ASHG Position Statement: Human Germline Genome Editing



THE AMERICAN SOCIETY OF HUMAN GENETICS

discover. educate. advocate.

About | Membership | Meetings | Awards | Policy/Advocacy | Education | Press

Member Center | Students/Trainees | Health Professionals | Genetics Advocates

Overview
Press Releases
Webcasts and Multimedia
News Clip Archives
ASHG Fact Sheet
ASHG Annual Meeting

For Immediate Release
Thursday, August 3, 2017
12:00 p.m. U.S. Eastern Time

Media Contact:
Nalini Padmanabhan
ASHG Communications Manager
301.634.7346
press@ashg.org

11 Organizations Urge Cautious but Proactive Approach to Gene Editing

Medical, Research, and Counseling Groups Issue Statement on Germline Genome Editing

BETHESDA, MD – An international group of 11 organizations with genetics expertise has issued a policy statement on germline genome editing in humans, which recommends against genome editing that culminates in human pregnancy; supports publicly funded, *in vitro* research into its potential clinical applications; and outlines scientific and societal steps necessary before implementation of such clinical applications is considered.

- Recommends against the use of germline gene editing that would result in pregnancy
- Asserts that in vitro studies of germline gene editing should continue without restriction of public funds

Document 17

Albany Medical Center Prize in Medicine and Biomedical Research

Document 18

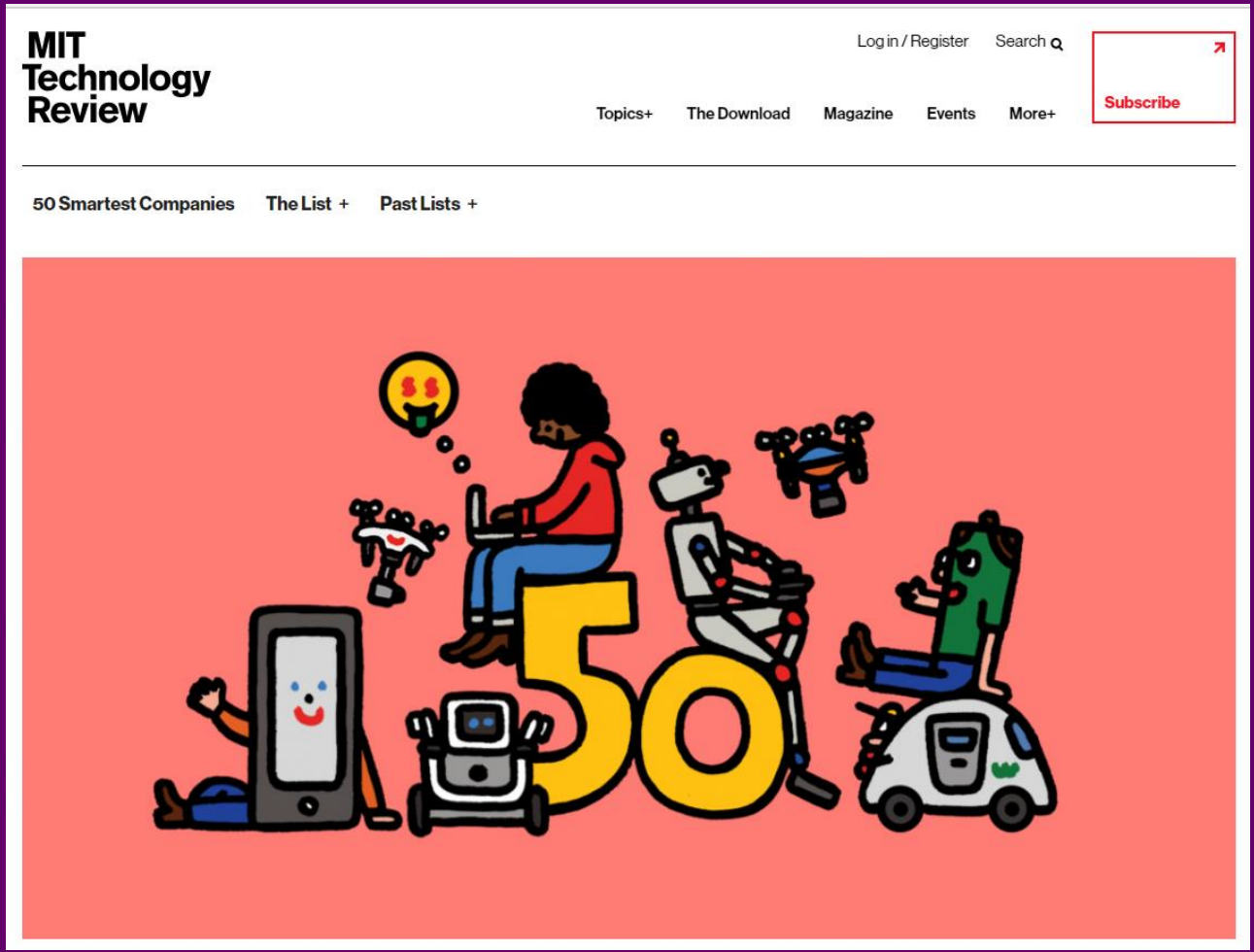
Allen Distinguished Investigator Awards

Fei Chen, Ph.D.

Jason Buenrostro, Ph.D.

THE
PAUL G. ALLEN
FRONTIERS GROUP

MIT Technology Review:
50 Smartest Companies



Document 20

Genomics In The News…

The JAMA Network

JN

Genomics and Precision Health

Genomics | Series | Glossary | Editors' Selections

JAMA Insights: Genomics and Precision Health

Precision medicine is a rapidly evolving approach to disease treatment and prevention that matches treatments to patients based on individual genetic variability. To help clinicians understand the latest developments in precision medicine so they can make the most informed decisions for their patients *JAMA* in 2017 is publishing a series of essays to explain the state of the field, its concepts, and technologies.

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Genomes In The News…

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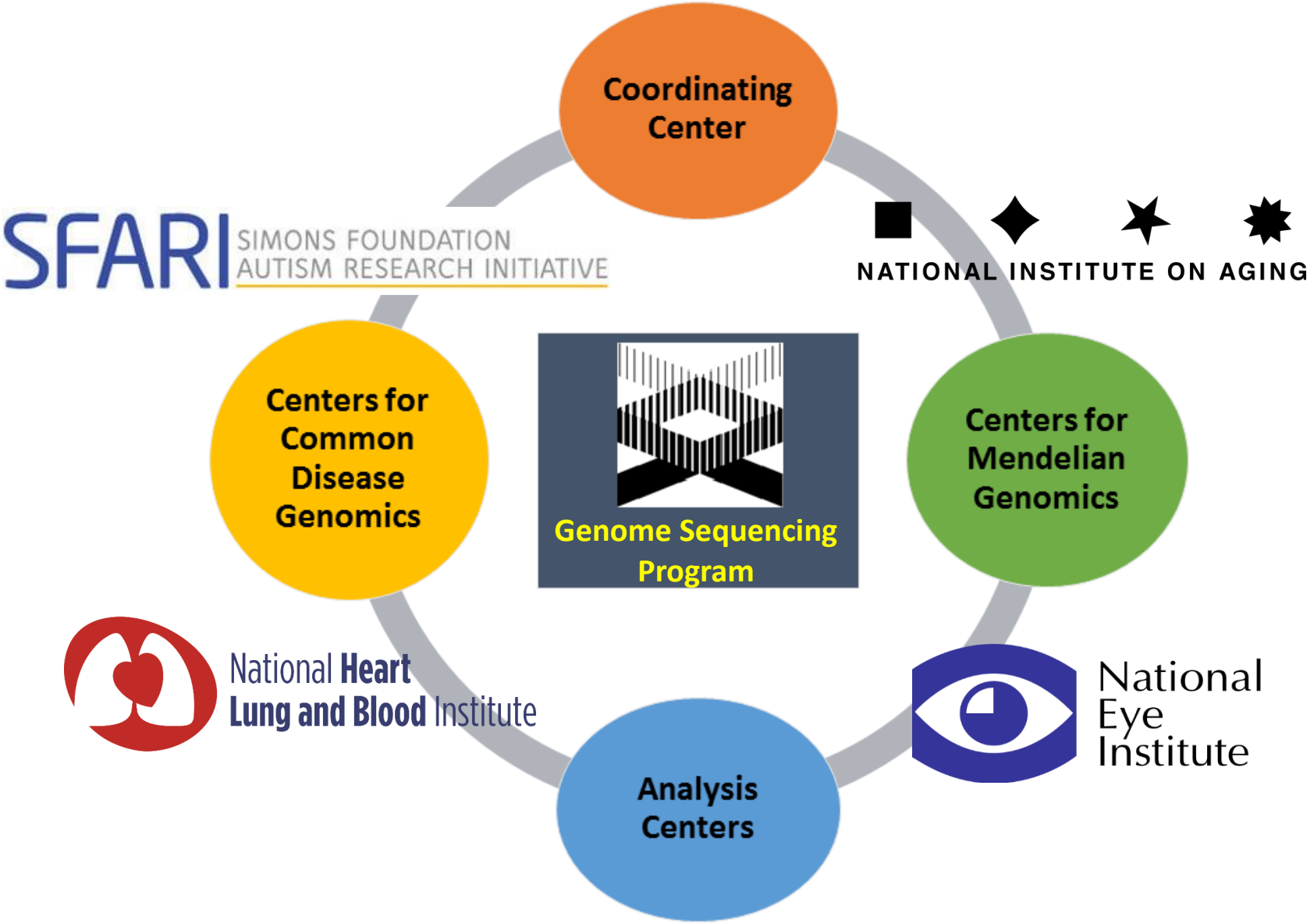
Genome Sequencing Program



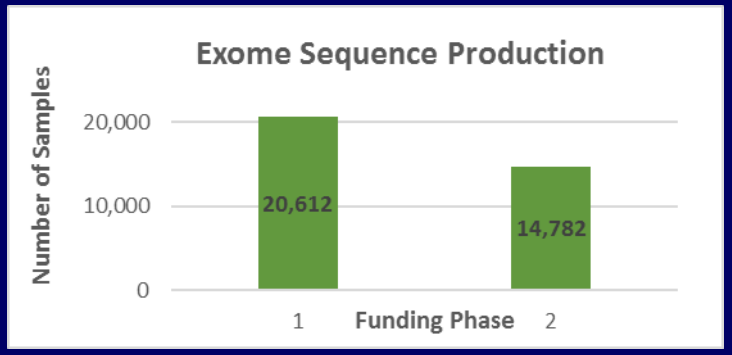
Genome Sequencing Program

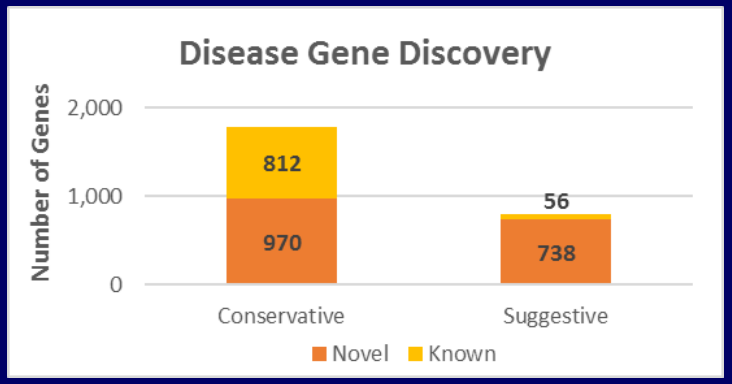
Centers for Common Disease Genomics

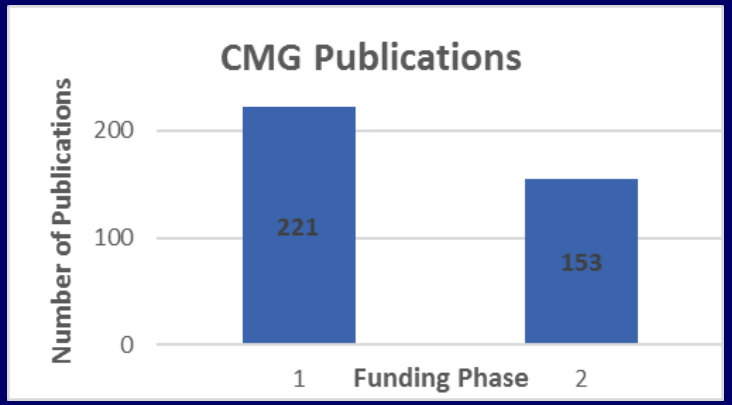
Disease Category	Sequence Type	Sequenced Samples	Approved Samples
Cardiovascular	Genomes	22,729	46,300
	Exomes	10,861	21,000
Immune-Mediated	Genomes	5,383	25,000
	Exomes	0	2,000
Neuropsychiatric	Genomes	9,366	15,300
	Exomes	11,450	32,000
TOTAL		**59,789**	**141,600**

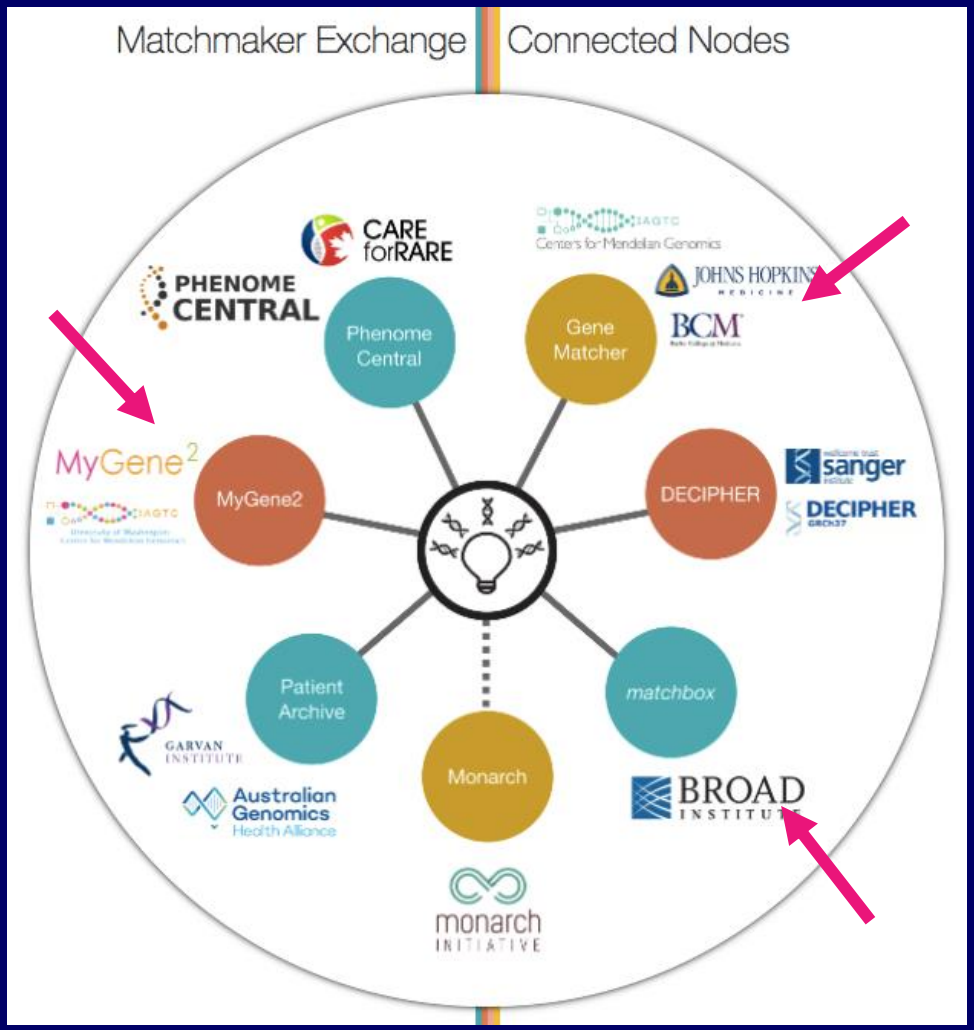
Genome Sequencing Program

Centers for Mendelian Genomics









Document 23

Technology Development Program

- **Advanced Genomic Technology Development Meeting - May 2017**
- **Novel Genomic Technology Development**
  - **PAR-16-14 (R01, also linked R21 and R43/44)**
  - **Next due date: October 31, 2017**

ENCyclopedia Of DNA Elements (ENCODE)

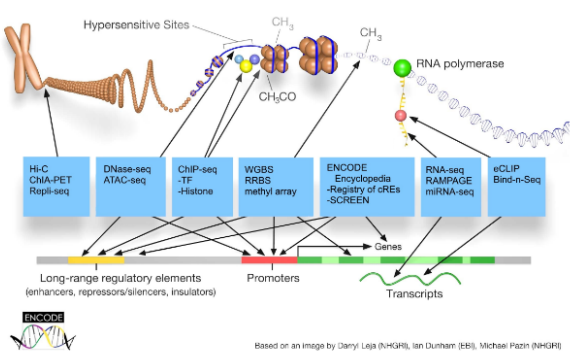
ASHG2017

ORLANDO • OCTOBER 17-21, 2017

SHARING DISCOVERIES. SHAPING OUR FUTURE.

SCREEN: Search Candidate Regulatory Elements by ENCODE

Overview | About | Tutorial | Files

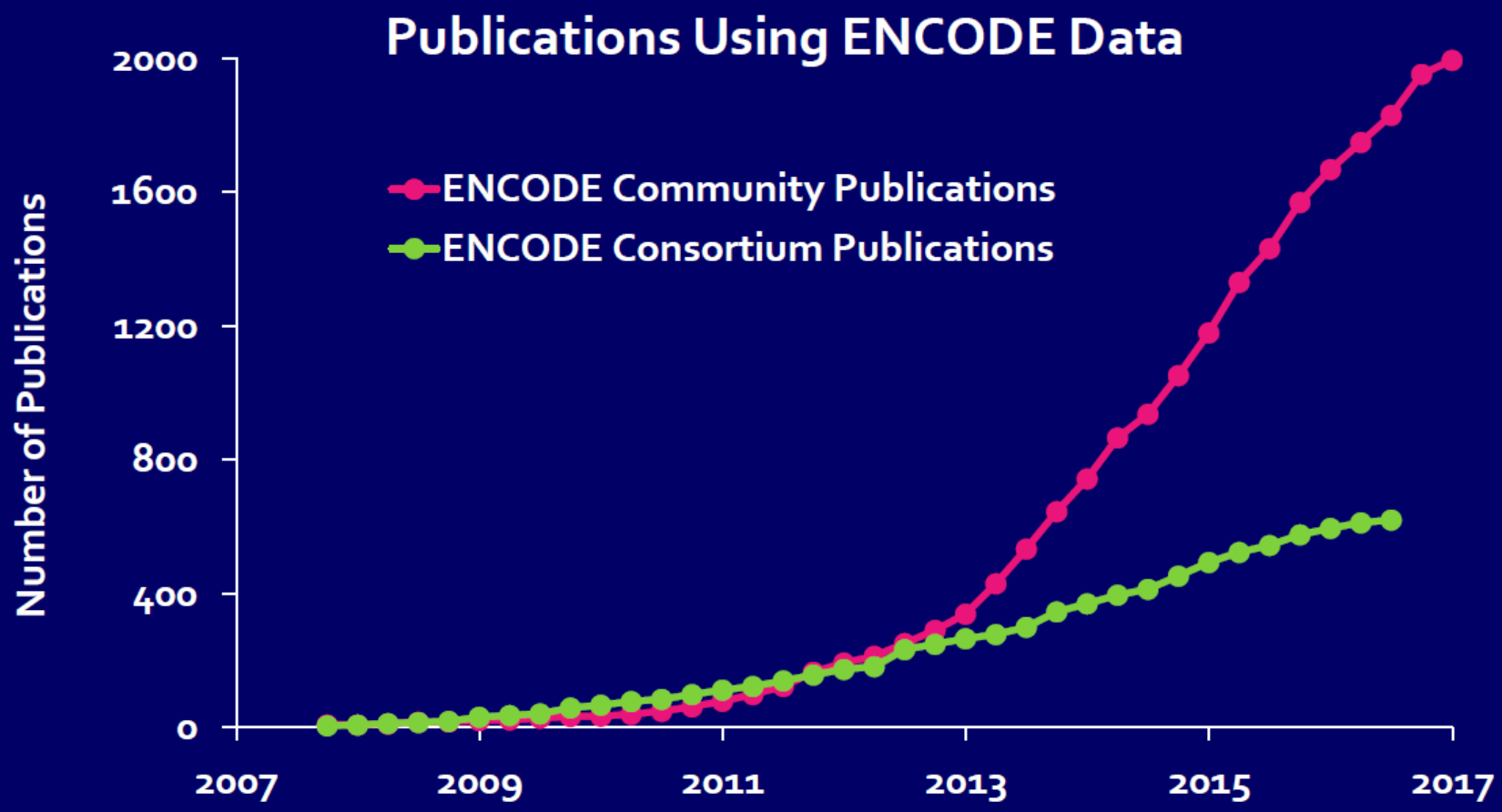


SCREEN is a web interface for searching and visualizing the Registry of candidate Regulatory Elements (cREs) derived from ENCODE data. The Registry contains 1.31M human cREs in hg19 and 0.52M mouse cREs in mm10, with orthologous cREs cross-referenced. SCREEN presents the data that support biochemical activities of the cREs and the expression of nearby genes in specific cell and tissue types.

You may launch SCREEN using the search box below or browse a curated list of SNPs from the NHGRI-EBI Genome Wide Association Study (GWAS) catalog to annotate genetic variants using cREs.

Browse GWAS

ENCyclopedia Of DNA Elements (ENCODE)



Document 25

Centers of Excellence in Genomic Science (CEGS) Program

- Center for Genome Editing and Recording

- PI - Jennifer Doudna

- Create methods to detect, alter, and record the sequence and output of the genome in individual cells and tissues

Document 26

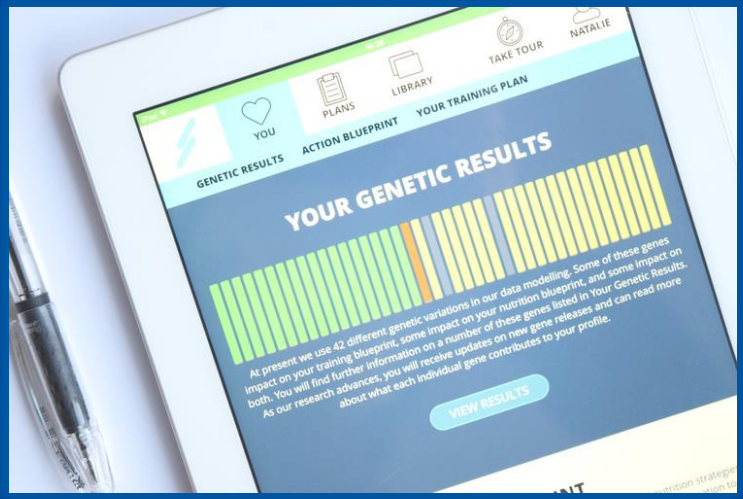
emerge network

ELECTRONIC MEDICAL RECORDS AND GENOMICS

eMERGE and Beyond: The Future of EHR and Genomics Workshop

October 30, 2017

- **Evidence generation for genomic medicine**
- **Identification of novel & disruptive opportunities**
- **Electronic phenotyping**
- **EMR integration of genomic results**



Document 27

Clinical Genome Resource (ClinGen)

Clinical Laboratories Meeting Minimum Data Sharing Requirements

- Ambry ✓
- ARUP ✓
- Athena ✓
- U. Medical Centre Ljubljana ✓
- Children's Mercy Hospital ✓
- Counsyl ✓
- EGL Genetics ✓
- GeneDx ✓
- Illumina ✓
- Invitae ✓
- Partners Healthcare LMM ✓
- Quest Diagnostics ✓
- University of Chicago ✓

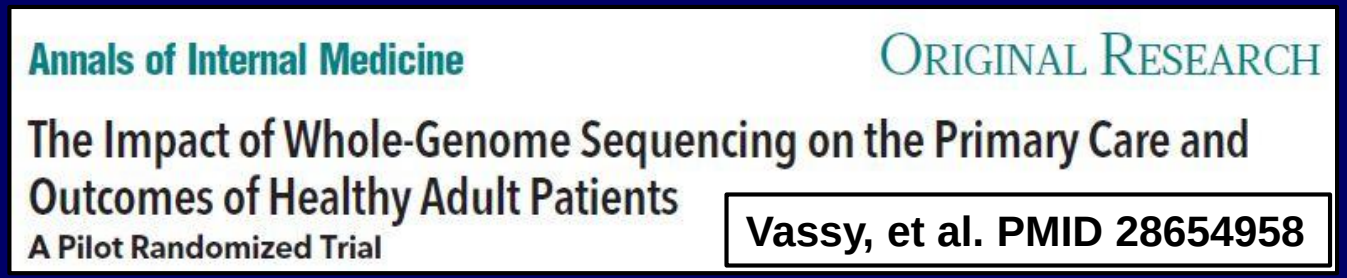
List as of Sept. 5, 2017

Save the Date!
May 23-25, 2018

Wellcome Genome Campus
Hinxton, England

Clinical Sequencing Exploratory Research Program

- Enrolled 5,477 adults and 1,434 children
- 345 total publications, 21 working group publications



- Demonstrated feasibility of returning genomic results by primary care providers
- Discovered monogenic disease risk in 22% of patients
- Recommended new clinical actions for 34% of patients

Clinical Sequencing Evidence-Generating Research Program

Phase II of CSER: Clinical Sequencing Evidence-generating Research Program

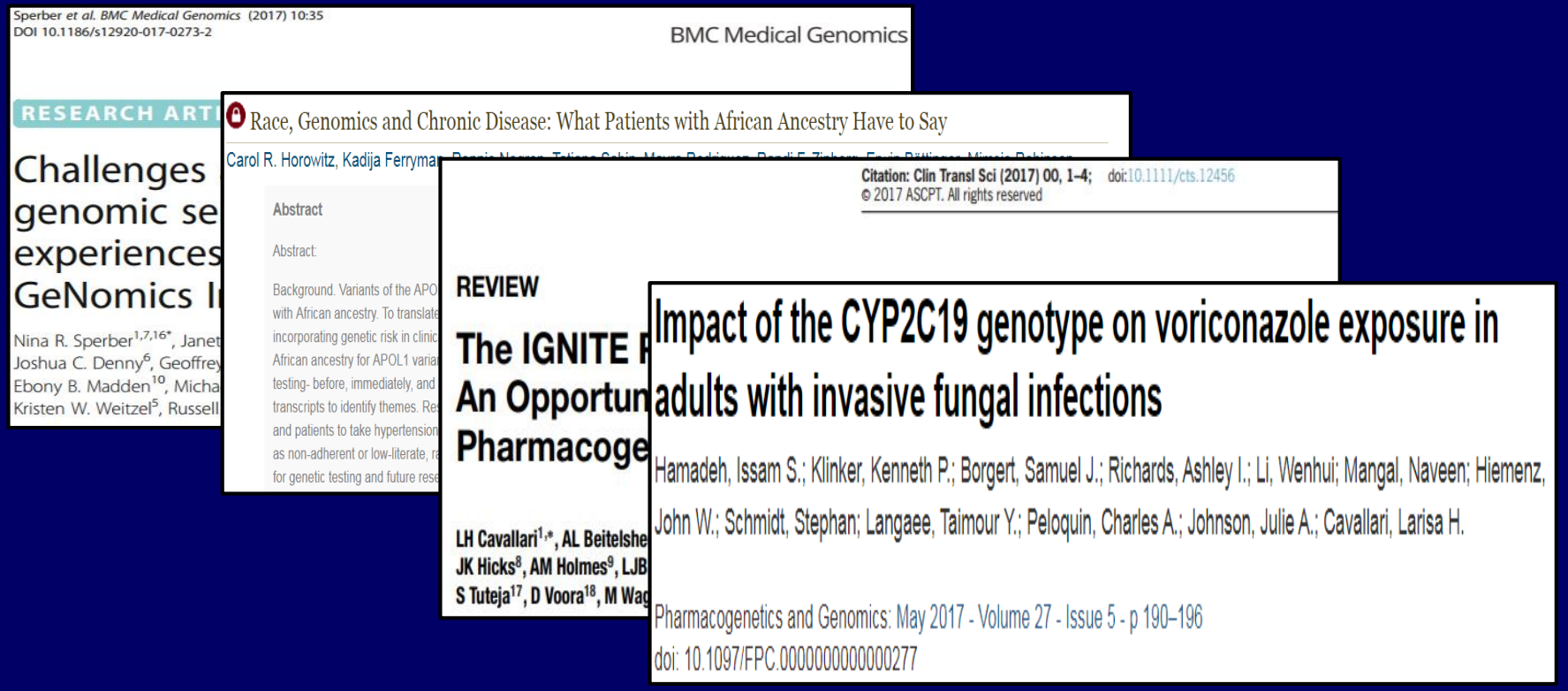
Clinical Sites:

- Baylor College of Medicine
- HudsonAlpha Institute of Biotechnology
- Kaiser Foundation Research Institute
- Icahn School of Medicine at Mount Sinai
- University of California, San Francisco
- University of North Carolina, Chapel Hill

Coordinating Center:

- University of Washington

Implementing Genomics In Practice (IGNITE) Network

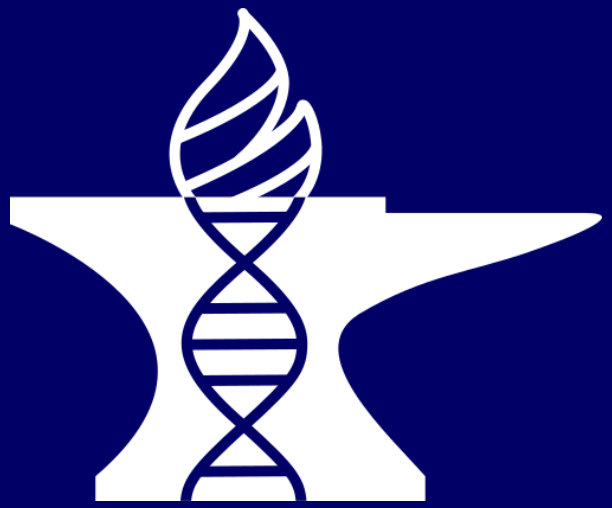


IGNITE RFAs released: RFA HG-17-008, -009, -010

Receipt date: November 3, 2017

Document 30

Computational Genomics and Data Science Program



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

- **RFA-HG-17-011 release: July 17, 2017**
- **Application due date: November 9, 2017**

Ethical, Legal, and Social Implications (ELSI) Research Program

- Genomics and Society Working Group annual meeting
- ELSI Program Announcements:

  Genomic Research, Genomic Healthcare, and Broader Legal, Policy, and Societal Issues

  Participating Institutes and Centers: FIC, NCI, NIA, NIAID, NICHD, NIDCD, NIEHS, NIMHD, NINDS

- Centers of Excellence in ELSI Research (CEER) RFA:

  Letter of Intent Due: September 30, 2017

  Application Due Date: October 31, 2017

4th ELSI Congress

Expanding the ELSI Universe #ELSICon

- June 2017
- Jackson Laboratory, CT
- 300+ attendees
- Videos of plenary presentations available on ELSICon website

Training and Career Development

2017 Meeting in St. Louis

2018 Meeting: March 18-20 in Los Angeles

Training and Career Development

Three new T32 programs:

Duke University (PI: Geoffrey Ginsburg)
Genomic Medicine

U. of Pennsylvania (PI: Katherine Nathanson)
Genomic Medicine

U. of Pennsylvania (PI: Steven Joffe)
ELSI Research

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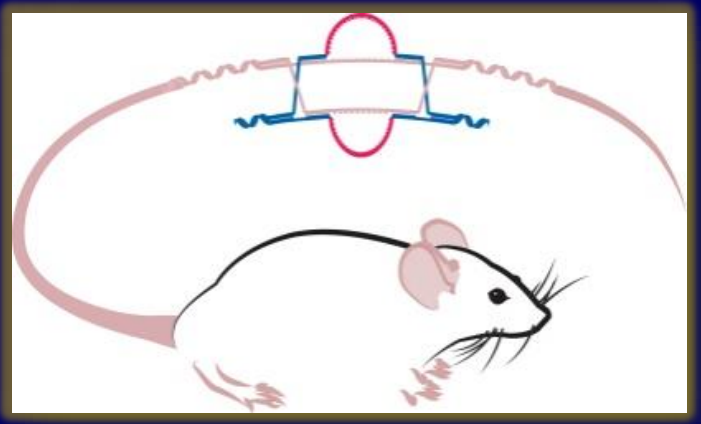
Knockout Mouse Phenotyping Project (KOMP2)



IMPC
International Mouse Phenotyping Consortium

- Annual IMPC Meeting – Athens, November 2017
- Sexual dimorphism paper published in *Nature Communications*
- Human disease models paper published in *Nature Genetics*
- Deafness paper in press in *Nature*

Human Heredity and Health in Africa (H3Africa)

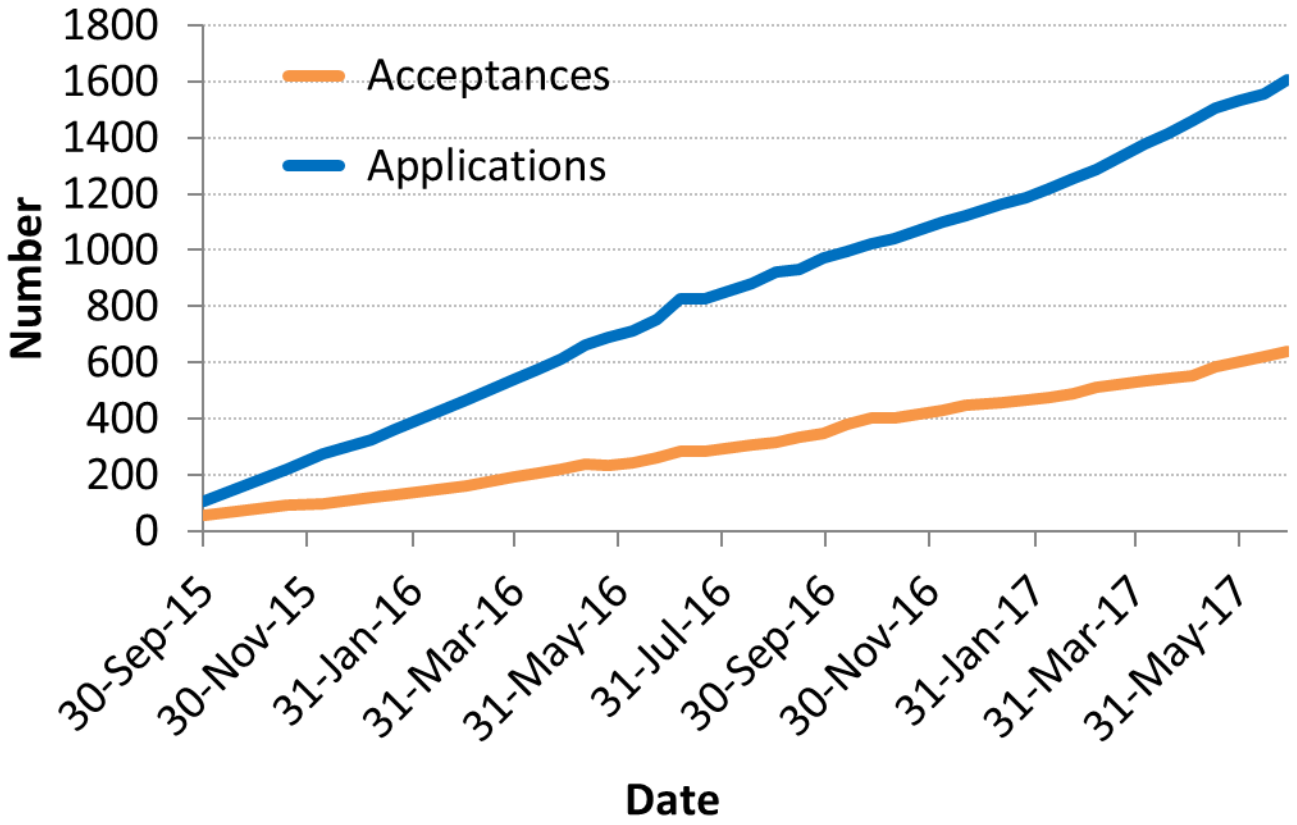
- Last Consortium meeting for Stage I in May 2017
  - Guest speaker: Eric Green, NHGRI Director

- Awards for Stage II anticipated soon
- First Consortium meeting for Stage II in March 2018

Document 35

Undiagnosed Diseases Network (UDN)

Undiagnosed Diseases Network | UDN Applications and Acceptances



1,606 applications, ~71 per month

639 acceptances, ~28 per month

~50% Accepted

~25% Diagnosed

APPLY

93 Diagnoses

Document 36

Undiagnosed Diseases Network (UDN)

Phase II FOAs

- **Clinical Sites:** 8-10, RFA-RM-17-019
- **Coordinating Center:** 1, RFA-RM-17-018
- **Model Organisms Screening Center(s):** 1-2, RFA-RM-17-017
- **Sequencing Core(s):** 1-2, RFA-RM-17-016
- **Metabolomics Core(s):** 1-2, RFA-RM-17-015

All Applications Due November 2, 2017

Gabriella Miller Kids First Pediatric Research Program

- 23 cohorts (>18,000 samples) available
- Genome sequencing:
  - BCM & WashU ($12.6M, 2015)
  - Broad Institute & HudsonAlpha/St. Jude ($31M, 2016-2019)
  - ~9,000 samples sequenced to date
- Data resource center:
  - CHOP ($15M, 2017-2022)
- Data analysis

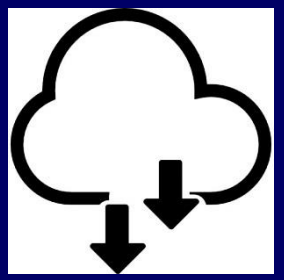
NIH Data Commons Pilot Phase

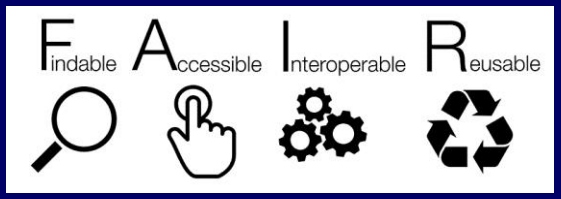
Other Transactions (OT) Research Opportunity Announcement (RM-17-026)

GTEx

TOPMed

ALLIANCE of GENOME RESOURCES





- **Data Commons Pilot Phase Consortium**
- **Datasets: GTEx, TOPMed, AGR**
- **Stage 1: Develop prototypes & implementation plan**

  Kickoff meeting: Fall 2017 (Bethesda)

- **Stage 2: Full implementation**

All of Us RESEARCH PROGRAM | The Future of Health Begins With You

All of Us Research Program

Fifty Forward
Love life at 50+

National Alliance for Hispanic Health

DELTA RESEARCH AND EDUCATIONAL FOUNDATION
CREATING POSSIBILITIES ... IMPROVING LIVES

SAN FRANCISCO GENERAL HOSPITAL FOUNDATION
Supporting the Heart of Our City

All of Us RESEARCH PROGRAM | The Future of Health Begins With You

Protocol Title	***All of Us* Research Program**[1]
Sponsor	National Institutes of Health (NIH)
Protocol Version	Core Protocol V1

[1] Precision Medicine Initiative, PMI, *All of Us*, the *All of Us* logo, and "The Future of Health Begins with You" are service marks of the U.S. Department of Health and Human Services.

Document 39

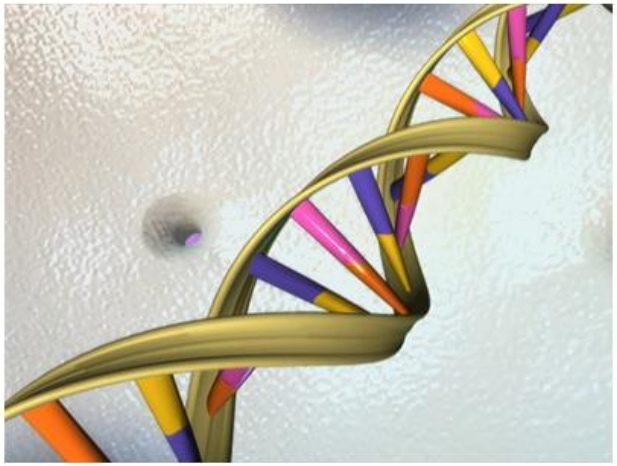
All of Us Research Program

All of Us Research Program Advisory Panel Launches Genomics Working Group

August 15, 2017

The *All of Us* Research Program Advisory Panel recently established a new working group to help inform the program's comprehensive genomics strategy. The group will consider various issues, including the evolving nature of genome sequencing technologies, the analysis of genomic data on a large scale, and the program's commitment to return information to participants. The group's work is expected to last approximately four months, during which time the group will prepare a final report for the advisory panel.

For more information, see the group's charge and roster.



DNA double helix
Credit: National Human Genome Research Institute, NIH.
www.genome.gov

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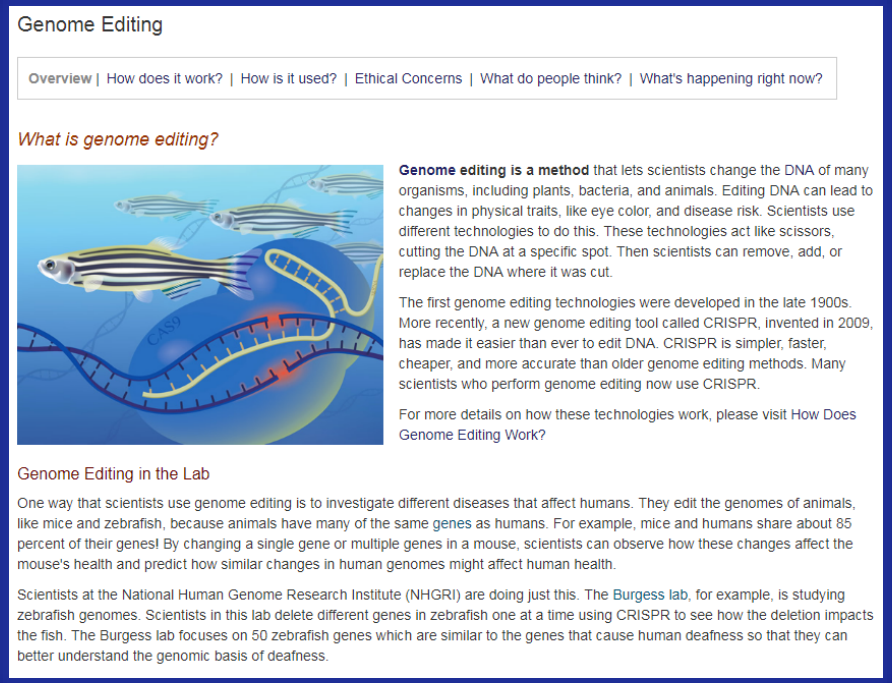
ASHG-NHGRI Genetics and Public Policy Fellow

Nikki Meadows, Ph.D.

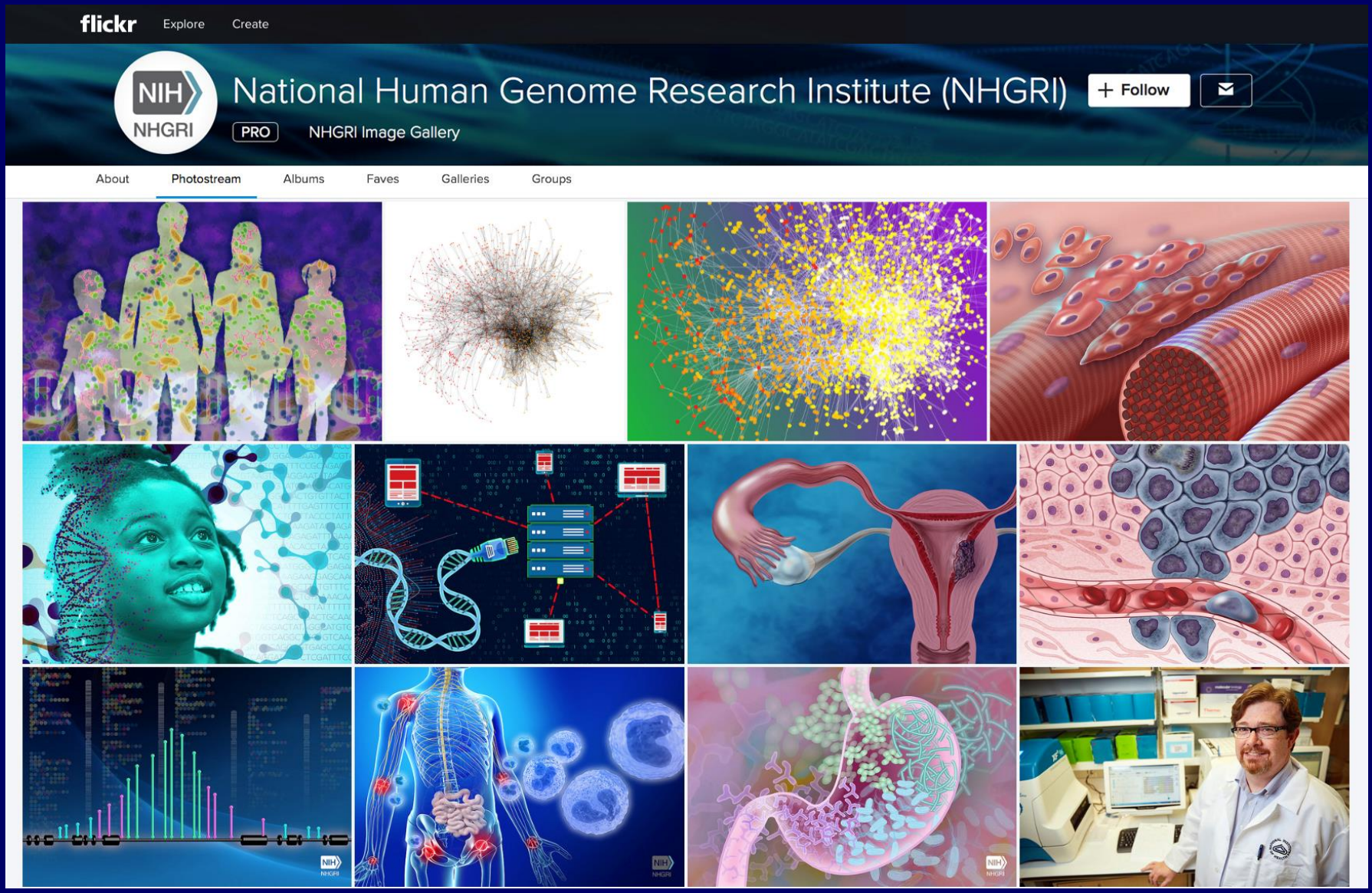
New Web Resources for Investigators and General Public



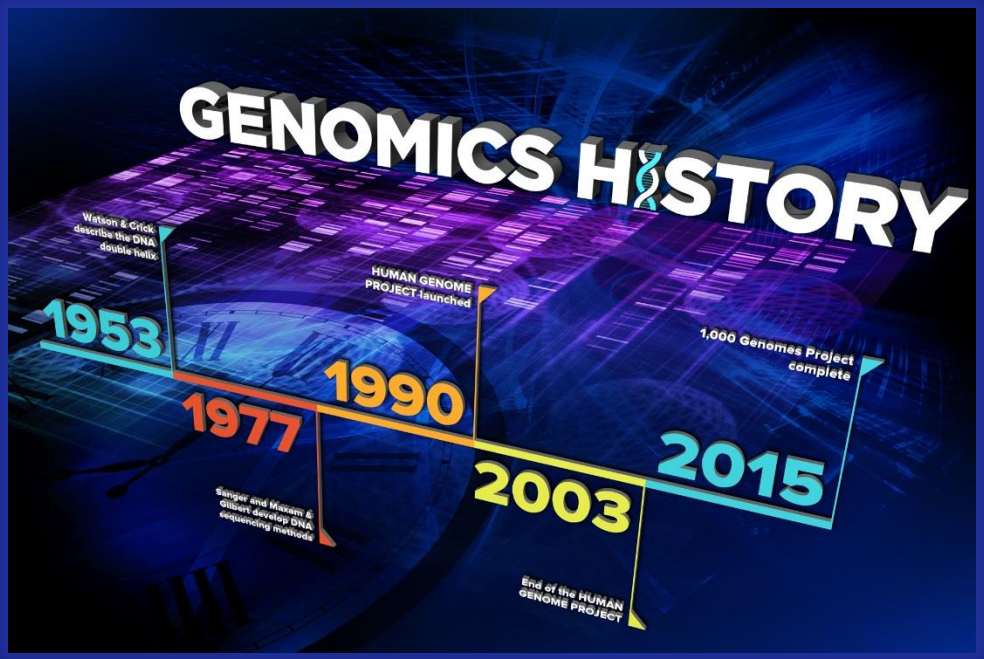
- **For Investigators:** Points to Consider Regarding the FDA's Investigational Device Exemption Regulations
- **For General Public:** Genome Editing

Document 40

Genomics Imagery on Flickr



NHGRI History of Genomics Program



- Database users meeting
- NHGRI History of Molecular Biology and Genomics Lecture Series

Genome: Unlocking Life's Code Exhibition

Travel Schedule

2017

June 12-September 11

Health Museum
Houston, TX

September 30-January 1

Science North

Sudbury, Ontario, Canada

2018

January 28-April 24

Rochester Museum and Science Center
Rochester, NY

NHGRI-Youth Career Connect High School Summer Program

The Immortal Henrietta Lacks Educator Workshop and Curriculum

NHGRI Short Course in Genomics

Middle/High School, Community College, and Tribal College Educators

National Human Genome Research Institute
Short Course in Genomics

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NHGRI Short Course in Genomics

Nurse, Physician Assistant, and Faculty

National Human Genome Research Institute
Short Course in Genomics
Nurse, Physician Assistant and Faculty Track

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Inter-Society Coordinating Committee for Practitioner Education (ISCC)

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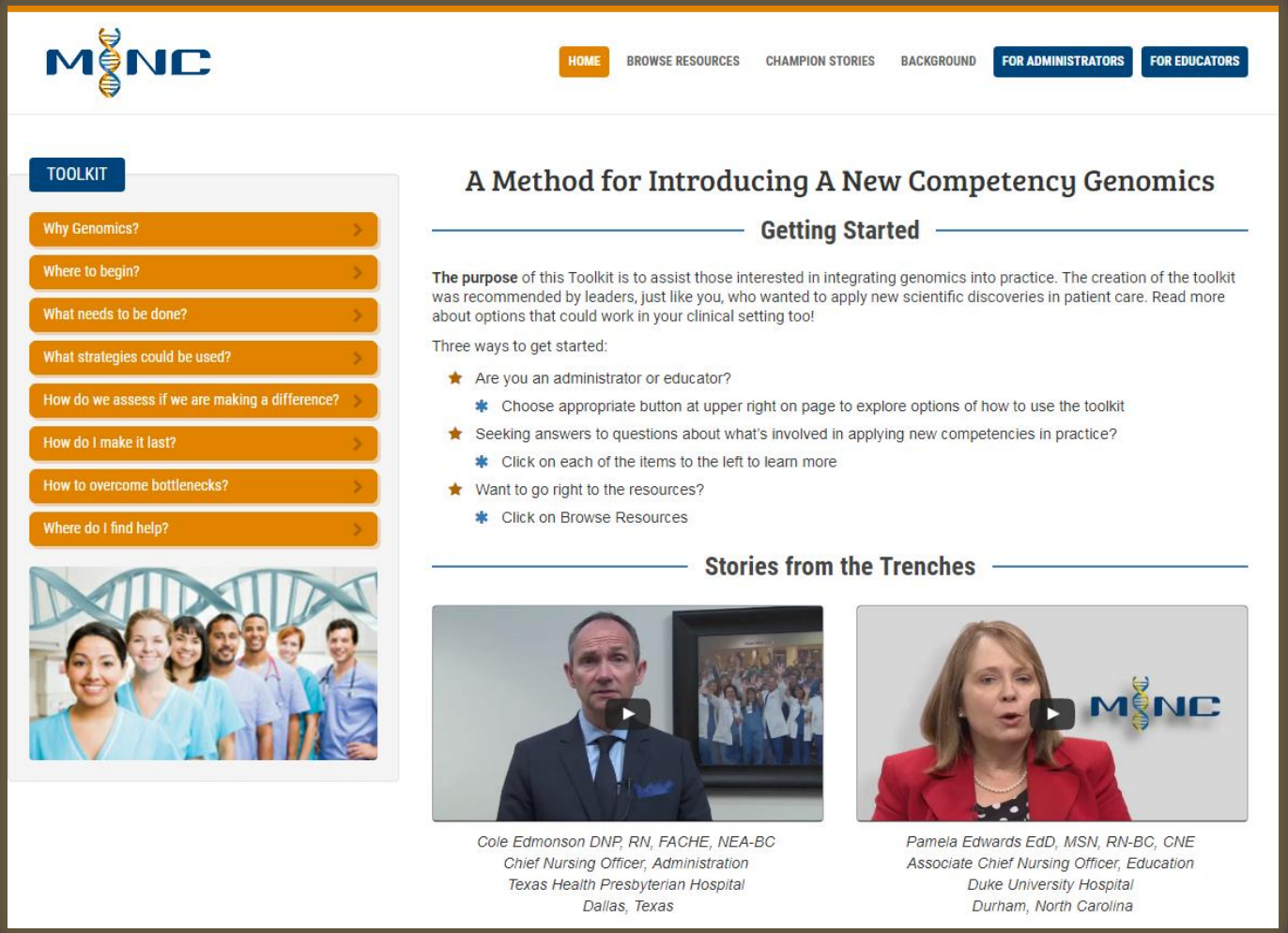
Method for Introducing a New Competency: Genomics (MINC)



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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

2017 Rare Impact Award, National Organization for Rare Disorders

NORD®
National Organization for Rare Disorders

Cynthia Tifft, M.D., Ph.D.

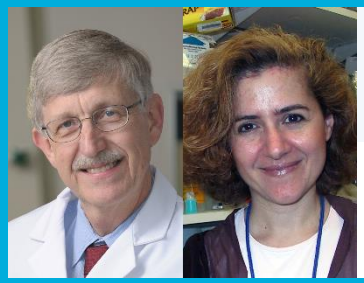
New President-Elect, American Society of Human Genetics

Les Biesecker, M.D.

NHGRI Intramural Research Highlights

Network Science

Social influence on 5-year survival in a longitudinal chemotherapy ward co-presence network



nature COMMUNICATIONS

A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome

Science Translational Medicine

***Staphylococcus aureus* and *Staphylococcus epidermidis* strain diversity underlying pediatric atopic dermatitis**

To receive *The Genomics Landscape*, go to list.nih.gov

Search for NHGRILANDSCAPE

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

National Human Genome Research Institute

Advancing human health through genomics research

Thanks!

Special Thanks!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Research Institute
Advancing human health
through genomics research