DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

May 2015

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

NIH National Human Genome Research Institute

Director's Report-Related Documents: May 2015

Director's Report

Director's Report

No.	Relevant Documents
1	**Harold Varmus Departs as NCI Director** Varmus Stepping Down as Director of NIH's National Cancer Institute Douglas Lowy Named Acting Director of the National Cancer Institute
2	Jack Whitescarver Steps Down as Director, NIH Office of AIDS Research
3	New Director, National Institute on Minority Health and Health Disparities
4	New NIH Associate Director for Legislative Policy and Analysis
5	**NIH Genomic Data in the Cloud Policy** NIH Statement: Use of Cloud Computing Services for Storage and Analysis of Controlled-Access Data Subject to the NIH Genomic Data Sharing Policy Input | Output Blog Post: The Cloud, dbGaP and the NIH

genome.gov/DirectorsReport

Document #

Open Session Presentations

Concept Clearance:

- **Workshop Report: "From Genome Function to Biomedical Insight— ENCODE and Beyond"**

  Mike Pazin

- **Concept Presentation: Functional Genomics**

  Elise Feingold

Open Session Presentations

- **Clinical Sequencing Exploratory Research Program**

  Gail Jarvik

- **NHGRI ClinSeq Project**

  Les Biesecker

Open Session Presentations

Concept Clearances:

- **Concept Presentation: Data Analysis and Coordinating Center for Diversity Action Plans and Institutional Training Grants**

  Tina Gatlin

- **Workshop Report: “Genomic Technology Development”**
- **Concept Presentation: Genomic Technology Development**

  Mike Smith

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 Chief Grants Management Officer

Cheryl Chick

Departure of Senior Advisor to the Director

Karen Rothenberg, J.D., M.P.A.

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Harold Varmus Departs as NCI Director

Harold Varmus, M.D.

Doug Lowy, M.D.

NATIONAL® CANCER INSTITUTE

Document 1

Jack Whitescarver Steps Down as Director, NIH Office of AIDS Research

Jack Whitescarver, Ph.D.

NIH National Institutes of Health
Office of AIDS Research

Document 2

New Director, National Institute on Minority Health and Health Disparities

Eliseo Pérez-Stable, M.D.

NIH National Institute on Minority Health and Health Disparities

Document 3

New NIH Associate Director for Legislative Policy and Analysis

Adrienne Hallett, M.T.S.

National Institutes of Health

Document 4

NIH Genomic Data in the Cloud Policy

- NIH now allows use of public or private cloud systems for storing and analyzing controlled-access genomic data
- NIH expects Institutions to ensure that local and cloud computing systems meet data use and security standards

Document 5

NIH Web Portal on Rigor and Reproducibility

Document 6

Battelle Report on NIH Innovation

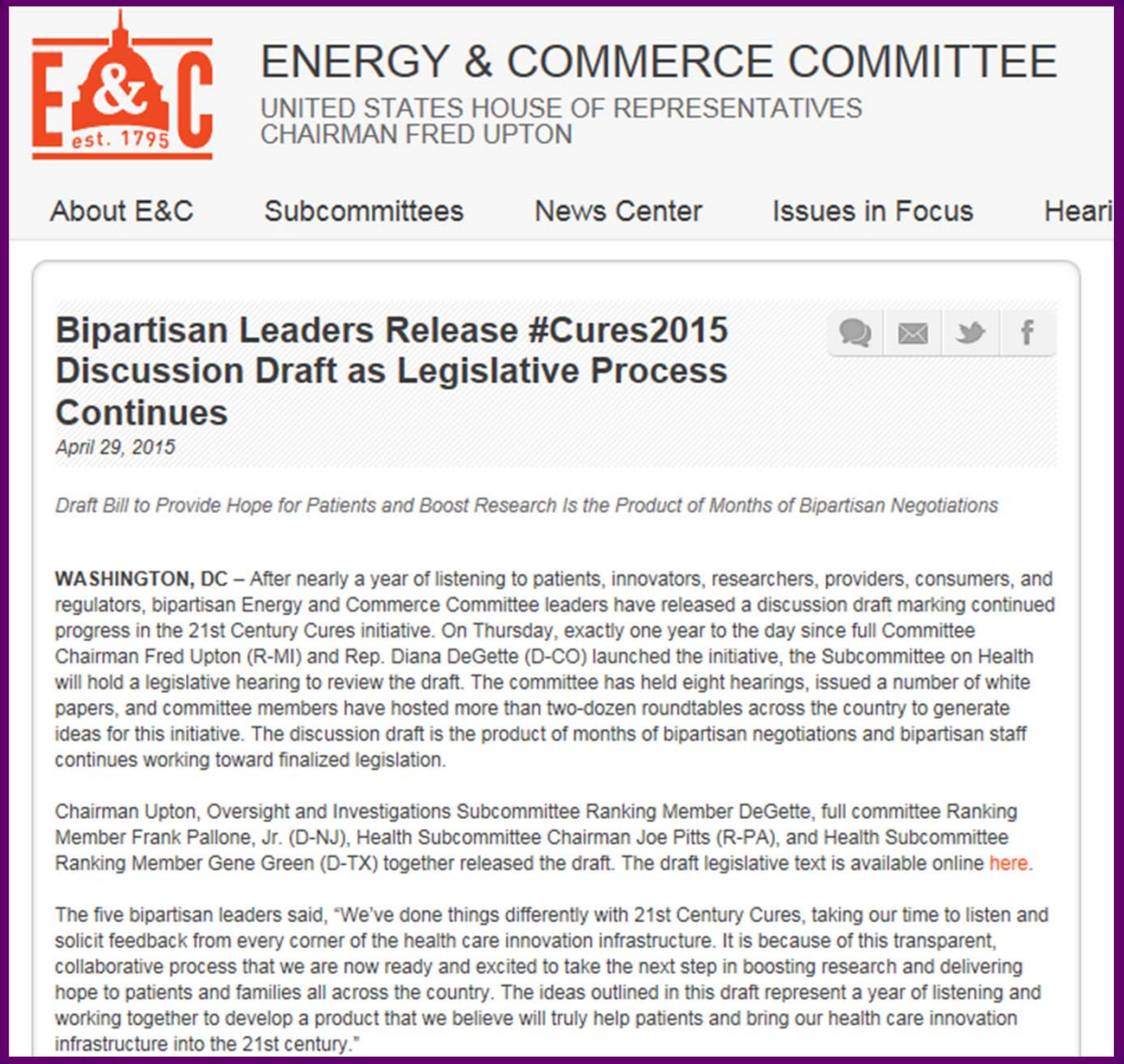
Document 7

NIH and NHGRI Appropriations

	FY 2014	*FY 2015*	*FY 2016 President's Budget*
NIH	**$30.2 B**	**$30.3 B**	**$31.1 B**
NHGRI	**$498 M**	**$499 M (+ 0.3%)**	**$515 M (+ 3.2%)**

- **President's proposed Fiscal Year (FY) 2016 budget sent to Congress in February**
- **House and Senate hold NIH funding hearings**

New Draft of 21st Century Cures Act



ENERGY & COMMERCE COMMITTEE
UNITED STATES HOUSE OF REPRESENTATIVES
CHAIRMAN FRED UPTON

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Bipartisan Leaders Release #Cures2015 Discussion Draft as Legislative Process Continues

April 29, 2015

Draft Bill to Provide Hope for Patients and Boost Research Is the Product of Months of Bipartisan Negotiations

WASHINGTON, DC – After nearly a year of listening to patients, innovators, researchers, providers, consumers, and regulators, bipartisan Energy and Commerce Committee leaders have released a discussion draft marking continued progress in the 21st Century Cures initiative. On Thursday, exactly one year to the day since full Committee Chairman Fred Upton (R-MI) and Rep. Diana DeGette (D-CO) launched the initiative, the Subcommittee on Health will hold a legislative hearing to review the draft. The committee has held eight hearings, issued a number of white papers, and committee members have hosted more than two-dozen roundtables across the country to generate ideas for this initiative. The discussion draft is the product of months of bipartisan negotiations and bipartisan staff continues working toward finalized legislation.

Chairman Upton, Oversight and Investigations Subcommittee Ranking Member DeGette, full committee Ranking Member Frank Pallone, Jr. (D-NJ), Health Subcommittee Chairman Joe Pitts (R-PA), and Health Subcommittee Ranking Member Gene Green (D-TX) together released the draft. The draft legislative text is available online here.

The five bipartisan leaders said, "We've done things differently with 21st Century Cures, taking our time to listen and solicit feedback from every corner of the health care innovation infrastructure. It is because of this transparent, collaborative process that we are now ready and excited to take the next step in boosting research and delivering hope to patients and families all across the country. The ideas outlined in this draft represent a year of listening and working together to develop a product that we believe will truly help patients and bring our health care innovation infrastructure into the 21st century."

Document 8

Senate Hearing on the Precision Medicine Initiative

Document 8

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Dan David Prize 2015
Future – Bioinformatics

David Haussler, Ph.D.

Elected to NAS

Marianne Bronner
Aravinda Chakravarti
Scott Edwards
John Lis
Joachim Messing
Rodney Rothstein

NATIONAL ACADEMY OF SCIENCES

Elected to AIMBE

American Institute for Medical and Biological Engineering

Deidre Meldrum, Ph.D.

Executive VP for Medical Genomics and Chief Medical Genomics Officer, HudsonAlpha

HUDSONALPHA
INSTITUTE FOR BIOTECHNOLOGY

Howard Jacob, Ph.D.

Document 12

New Chief Medical Officer, Invitae

INVITAE

Robert Nussbaum, M.D.

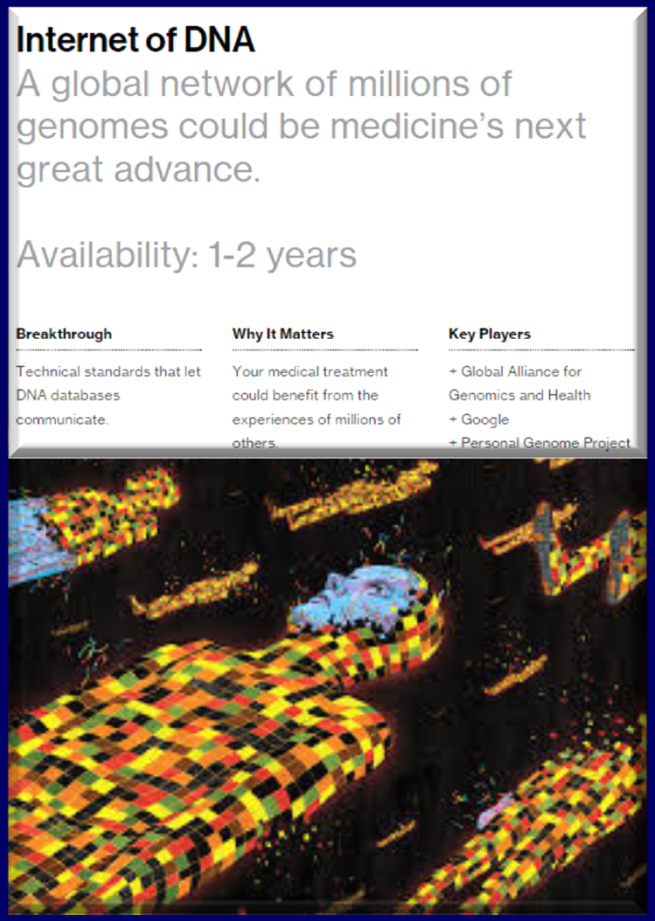
New Joint Directors of EMBL-EBI

Rolf Apweiler, Ph.D.

Ewan Birney, Ph.D.

EMBL-EBI

MIT Technology Review
Breakthrough Technologies 2015



Document 15

NHGRI Genome Advance of the Month

CRISPR probes the inner workings of the genome in real time

By Kyle Davis
ScM Candidate, Genetic Counseling, JHU/NHGRI

genes in a
DNA seque

Scientists create a new "roadmap" for the human epigenome

By Sara Cassidy, M.S., Ph.D.
Postdoctoral Fellow, NHGRI

our genes
the Month
this way,

Iceland study provides insights into disease, paves way for large-scale genomic studies

By Yekaterina Vaydylevich
Scientific Program Analyst, NHGRI

For some, Iceland conjures thoughts of geothermal spas like the Blue Lagoon, moonlike landscapes and literary sagas peopled with Huldufólk, elfin creatures. Most people do not, however, think of large-scale genomic studies.

But maybe they should. Last month, Kári Stefánsson, M.D., Daníel Fannar Guðbjartsson, Ph.D., and their research team at deCODE genetics announced findings based on the whole genome sequence information of 2,636 Icelanders and the genotypic information of 104,220 other Icelanders. Dr. Stefánsson is also chief executive officer of deCODE genetics, a biopharmaceutical company in Reykjavík, Iceland. The March Genome Advance of the Month focuses on some of the interesting results published as a collection of papers in last month's Nature Genetics.

Iceland is well-suited for genomics research for a few reasons. The island's relative isolation has resulted in a population of approximately 320,000 that is almost entirely descended from a single family tree. Data about the population's genealogy has been extensively recorded since 740 AD, and sits in an accessible database called Íslendingabók or book of Icelanders. In population genomics, this is called the founder effect, in which the lack of diversity limits the number of genomic variants (differences between people), and allows usually rare variants to become common enough in the population to be more easily noticed and studied. (To avoid awkward situations like accidentally dating your cousin, Íslendingabók was recently connected to an application that quickly identifies the degree of relatedness between any two people.)

Document 16

Genomics In The News…

The New York Times

A Never-Ending Genetic Quest

Mary-Claire King's Pioneering Gene Work, From Breast Cancer to Human Rights

FEB. 9, 2015

Claudia Dreifus
A CONVERSATION WITH

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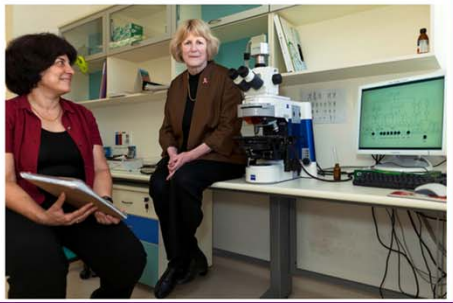
There has never been a scientific career quite like Mary-Claire King's. Years ago, her doctoral thesis concluded that humans and chimpanzees were, genetically speaking, 99 percent the same — a revolutionary thought. Her later work on human cancers resulted in the discovery of the so-called breast cancer gene, BRCA1, which transformed the diagnosis and treatment of the disease.

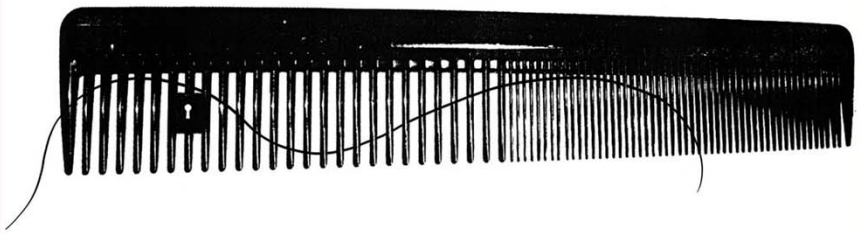
Besides her traditional scientific pursuits, Dr. King created genetic tests to help ascertain the identities of victims of political violence in places like Rwanda and El Salvador. And she did all this as a single mother raising a daughter.

Dr. King, 68, is now a geneticist at the



Fix the Flaws in Forensic Science

By ERIC S. LANDER APRIL 21, 2015



Mike McQuade

Newt Gingrich: Double the N.I.H. Budget

By NEWT GINGRICH APRIL 22, 2015

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FAR FROM THE MADDING CROWD
NOW PLAYING

MCLEAN, Va. — NO one who lived through the 1990s would have suspected that one day people would look back on the period as a golden age of bipartisan cooperation. But in some important ways, it was. Amid the policy fights that followed the Republican victories of 1994, President Bill Clinton and the new majorities in Congress reached one particularly good deal: doubling the budget for the National Institutes of Health.

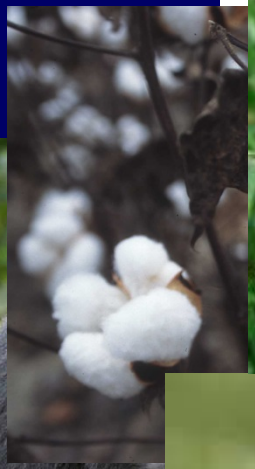
The decision was bipartisan, because health is both a moral and financial issue. Government spends more on health care than any other area. Taxpayers spend more than $1 trillion a year for Medicare and Medicaid alone, and even more when you add in programs like Veterans Affairs, the Children's Health Insurance Program and the Indian Health Service.

Unfortunately, since the end of the five-year effort that roughly doubled the N.I.H. budget by 2003, funding for the institutes has been flat. The N.I.H. budget (about $30 billion last year) has effectively been reduced by more than 20 percent since then. As 92 percent of the N.I.H. budget goes directly to research, one result is that the institutes awarded 12.5 percent fewer grants last year than in 2003. Grant applications, over the same period, increased by almost 50 percent.

Even as we've let financing for basic scientific and medical research stagnate, government spending on health care has grown significa should trouble every fiscal con conservative myself, I'm often government "investments." Bu comes to breakthroughs that c not just treat — the most exper

Document 17

Genomes In The News…



Document 18

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Genome Sequencing Program

- **Applications for CCDGs and CMGs in review**
- **Coordinating Center RFA released**
  - **Genome Sequencing Program Coordinating Center (U24): RFA-HG-15-019**
- **FOAs for remaining concepts in process:**
  - **Genome Sequencing Program Analysis Centers**
  - **High-Quality (“Gold”) Reference Genomes**
  - **Comparative Genomics**

Sequencing Opportunity: Gabriella Miller Kids First Pediatric Research Program

- Facilitate discovering the genomic basis of structural birth defects and childhood cancers
- Access to whole-genome sequencing for available samples
- Applications due July 27

NIH
The Common Fund

One Hundred Thirteenth Congress
of the
United States of America

AT THE SECOND SESSION

Begun and held at the City of Washington on Friday, the third day of January, two thousand and fourteen

An Act

To eliminate taxpayer financing of political party conventions and reprogram savings to provide for a 10-year pediatric research initiative through the Common Fund administered by the National Institutes of Health, and for other purposes.

Be it enacted by the Senate and House of Representatives of the United States of America in Congress assembled,

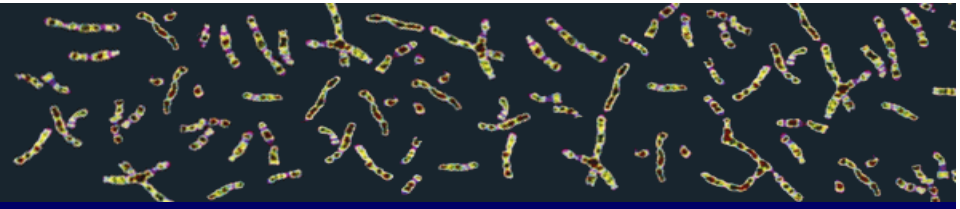
SECTION 1. SHORT TITLE.

This Act may be cited as the "Gabriella Miller Kids First Research Act".

Document 19

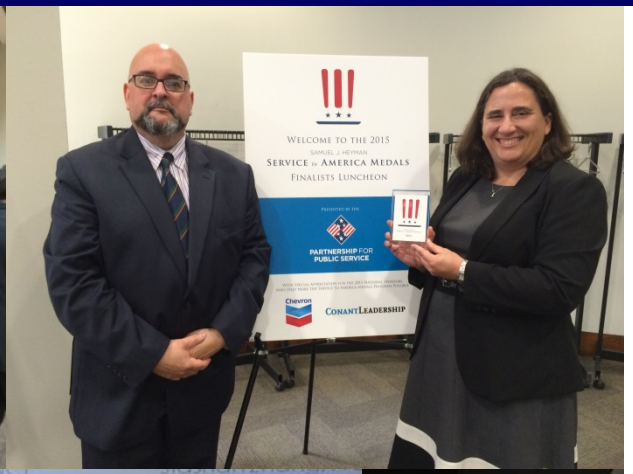
1000 Genomes

A Deep Catalog of Human Genetic Variation



- **Goal: Identify 95% of common variants**
- **Final dataset: >99% of common variants**
  - **Based on 2,504 people from 26 populations**
- **>84 million variant sites:**
  - **81M SNPs**
  - **3.4M indels (insertions/deletions)**
  - **68K structural variants**
- **Final paper published by October**

TCGA Project Team Named as Finalist for Service to America Medal



The Samuel J. Heyman
SERVICE to AMERICA
MEDALS

Document 21

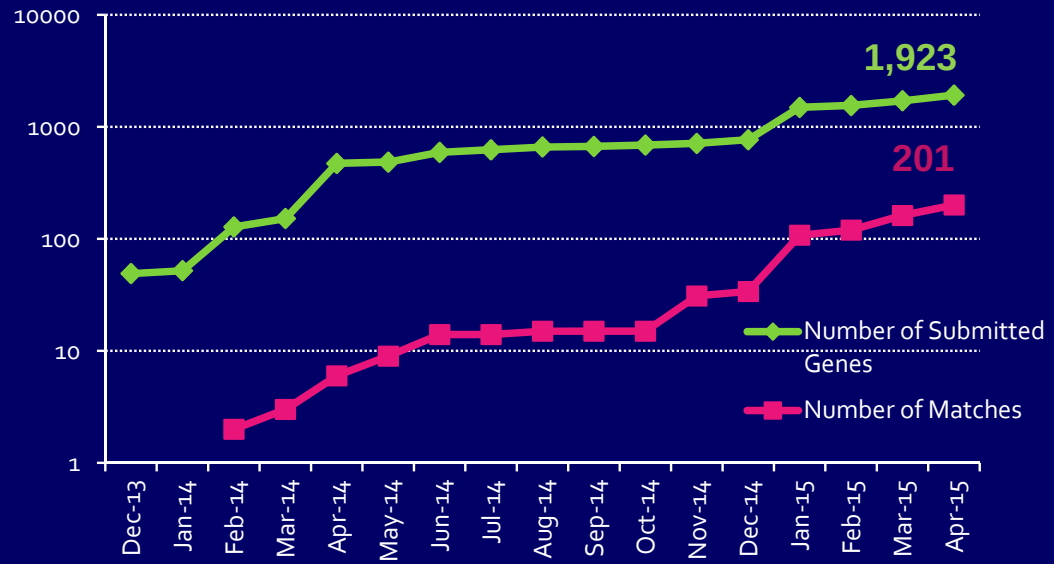
Centers for Mendelian Genomics

Finding the genes underlying human Mendelian conditions

Discovery

- Discovered >1,100 causal genes
- 453 novel causal genes
- >170 publications

GeneMatcher



Resource Sharing

- PhenoDB (>216 downloads)
- ALoFT source code
- Data analysis workshops
- dbGaP data deposition

Document 22

Clinical Sequencing Exploratory Research Program

- **Enrolled 3,638 adults, 796 children**
- **149 publications, 10 working group publications**

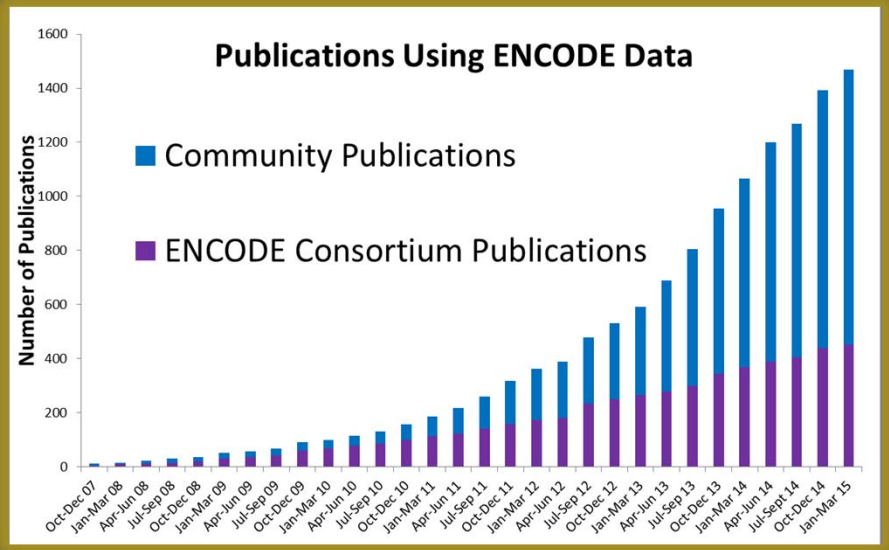
Document 23

Encyclopedia of DNA Elements (ENCODE)

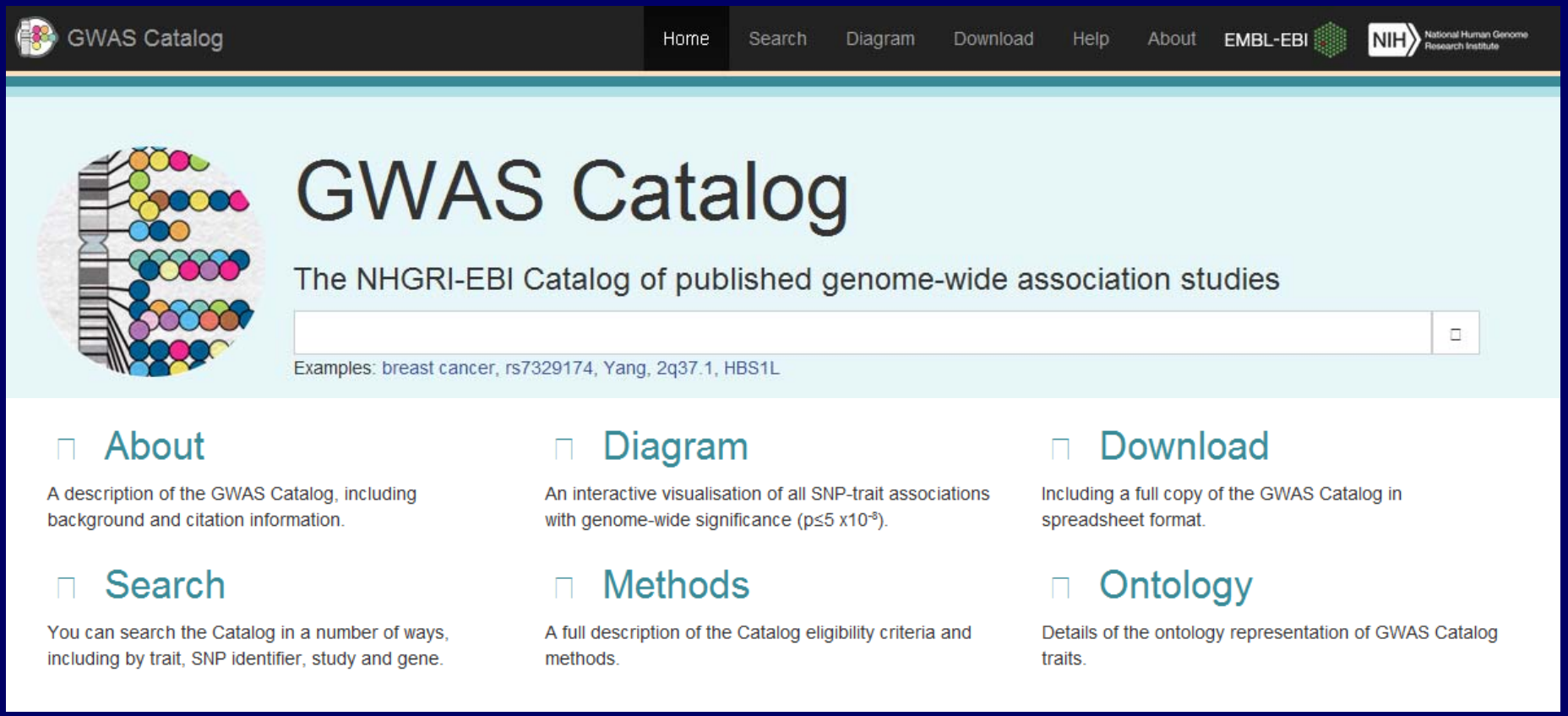
- **Planning Workshop: "From Genome Function to Biomedical Insight— ENCODE and Beyond"**
- **ENCODE Community User's Meeting**

  June 29 to July 1 in Potomac, Maryland

  Hands-on workshops using ENCODE data



Genome-Wide Association Studies (GWAS) Catalog



The eMERGE Network
electronic Medical Records & Genomics

Summit on Clinical Research Informatics

Chunhua Weng, PhD, MS, *Chair*, 2015 CRI Scientific Program Committee

Distinguished Paper Award

A Prototype for Executable and Portable Electronic Clinical Quality Measures Using the KNIME Analytics Platform

Huan Mo, MD, Jennifer A. Pacheco, Luke V. Rasmussen, Peter Speltz, Jyotishman Pathak, PhD, Joshua C. Denny, MD, William K. Thompson, PhD

Student Paper Award

Phenotyping Adverse Drug Reactions: Statin-related Myotoxicity

Laura K. Wiley, Jeremy D. Moretz, PharmD, Joshua C. Denny, MD, MS, Josh F. Peterson, MD, William S. Bush, PhD

- **Clinical validity classification system**
- **ClinGen-DECIPHER public meeting in May**

2015
ClinGen/DECIPHER Meeting
May 27-28, 2015 ~ Renaissance Washington DC Downtown

ClinGen
Clinical Genome Resource

DECIPHER
GRCh37

Implementing Genomics in Practice (IGNITE) Network

- **Marker paper submitted to *Genetics in Medicine***
- **Affiliate members:**

MOFFITT CANCER CENTER

UIC UNIVERSITY OF ILLINOIS AT CHICAGO

Nemours. Children's Health System

Precision Medicine Initiative Working Group of the Advisory Committee to the Director

Document 28

Newborn Sequencing in Genomic Medicine and Public Health (NSIGHT)

NSIGHT video interviews

Interviews with NSIGHT experts

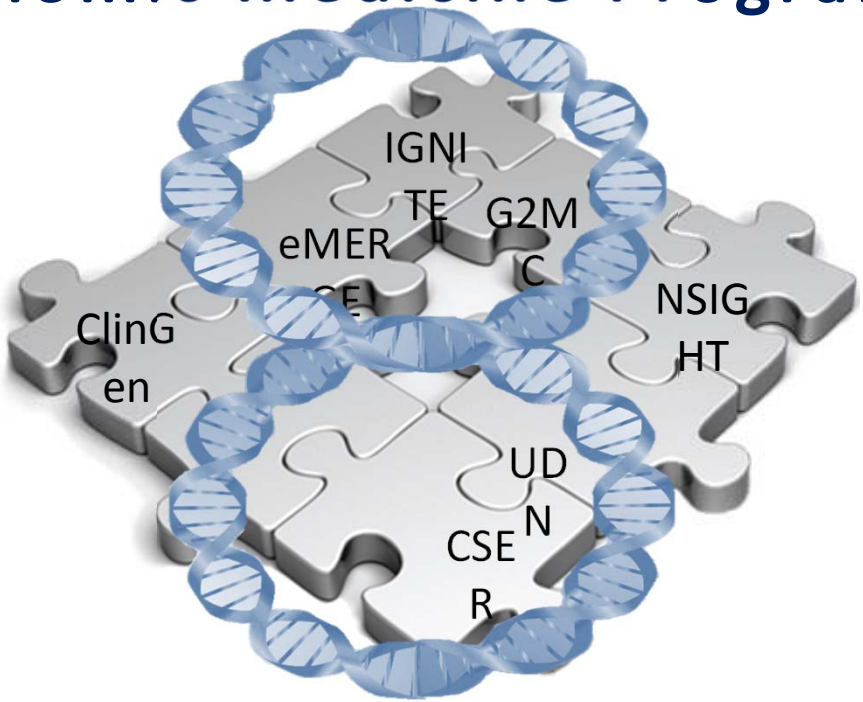
Click here to watch the playlist of videos. Throughout the conference, we will be adding new interviews after each presentation.

Watch the NSIGHT press briefing

Document 29

Genomic Medicine Working Group

NHGRI
Genomic Medicine Programs



Genomic Medicine #8
Bethesda, MD
June 8-9, 2015

Document 30

Genomics and Society Working Group

- Recent in-person meeting
- Topics discussed:
  - Normative and conceptual research
  - Portfolio balance
  - Health services research
  - ELSI and precision medicine
- Update planned for September Council meeting

Document 31

Centers of Excellence in ELSI Research (CEER)

- First Regional CEER Networking Meeting in February
- Annual CEER Investigator Meeting in March
- CEER RFA released, with applications due July 15

Document 32

NHGRI Large Genomics Resources Meeting: May 20-21

- **Goal: Develop strategic vision for NHGRI-funded large genomics resources**
- **Attendees:**
  - **PIs and Advisors**
  - **NIH staff (including NHGRI, NIGMS, and ADDS)**
- **Key discussion topics:**
  - **Efficiency**
  - **Impact on genomic research**
  - **Long-term sustainability**

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Integrative HMP (iHMP)

- **Longitudinal research projects**
  - **Inflammatory bowel disease**
  - **Type 2 diabetes**
  - **Preterm birth**
- **Data Coordination Center**
- **2nd iHMP Consortium Meeting**
  - **June 2015, Bethesda**
- **International Human Microbiome Consortium Congress**
  - **5th: March 2015, Luxembourg**
  - **6th: October 2016, Houston**

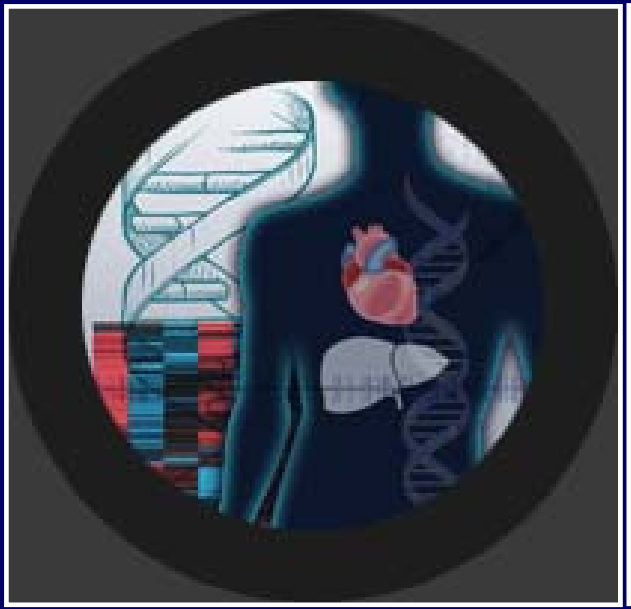
Fast-Track Action Committee: Mapping the Microbiome

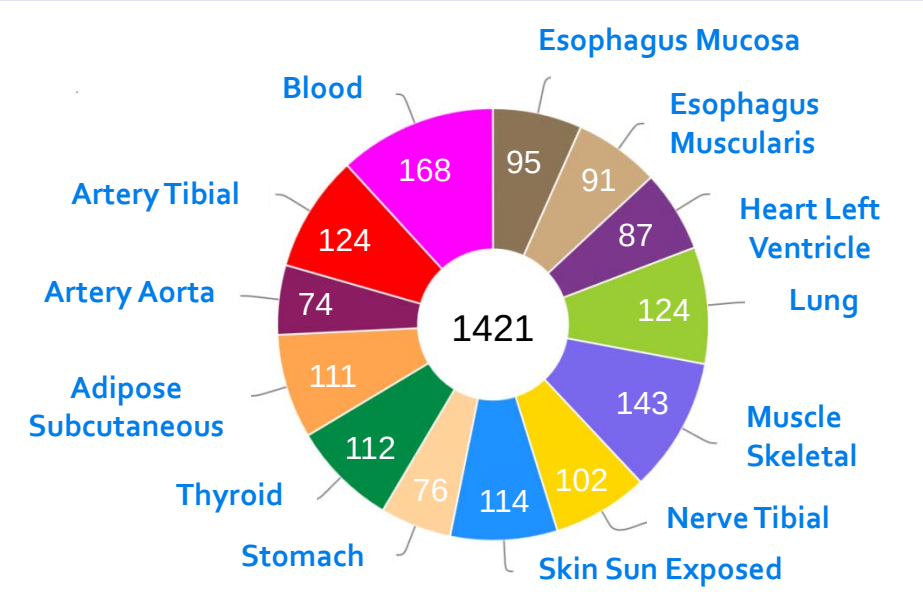
- **Chartered by OSTP**

- **Data call:**
  - **Portfolio analysis**
  - **11 departments and agencies**
  - **Host- and ecosystem-associated microbiomes**

- **Report to OSTP:**
  - **Identify gaps, needs, and challenges**
  - **Outline a Federal coordinated plan to support President's Fiscal Year 2017 budget request**

Genotype-Tissue Expression Project (GTEx)





- Donor recruitment to conclude by end of summer (n=900)
- Main manuscript and two companion papers published in *Science*

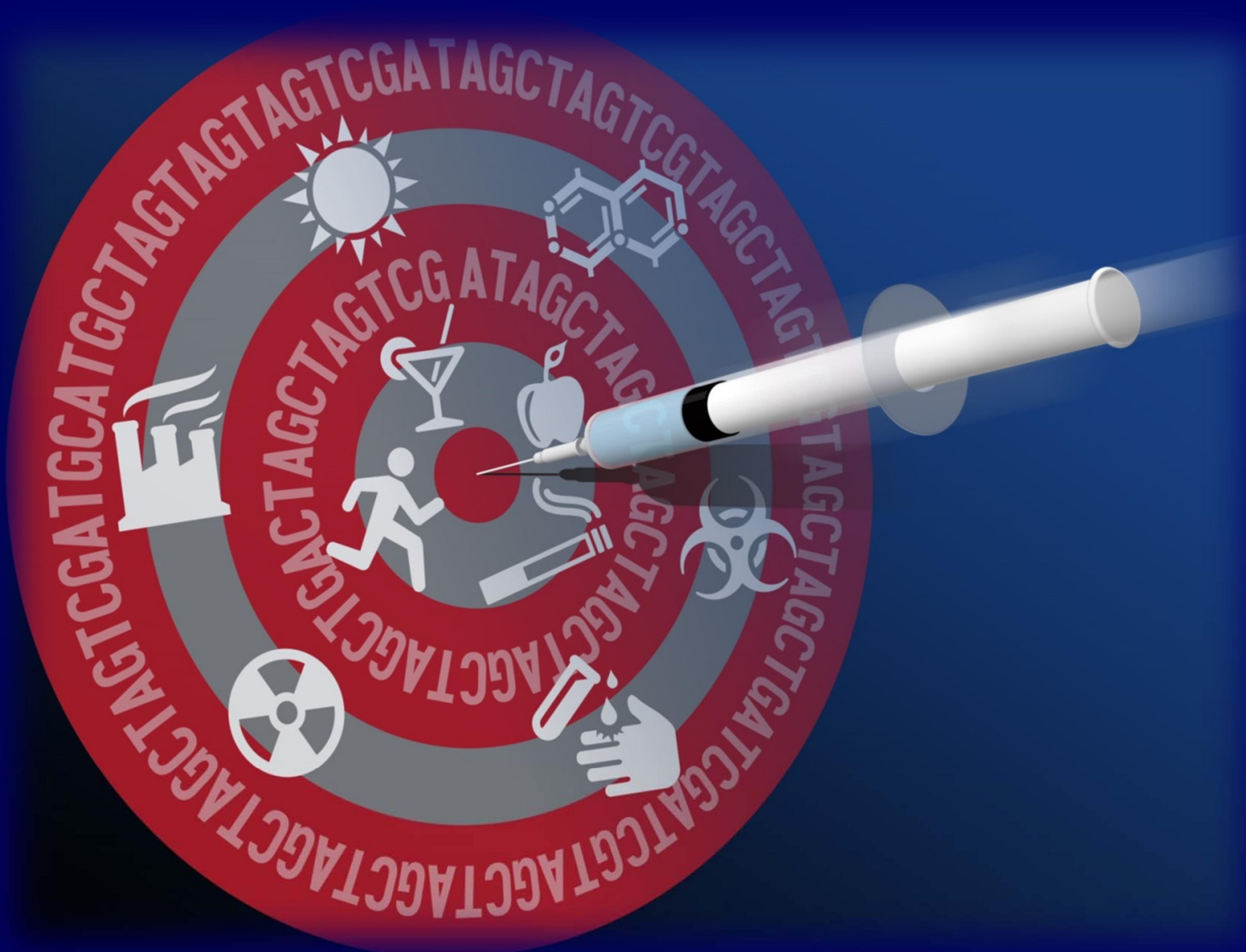
Human Heredity and Health in Africa (H3Africa)

- 6th Consortium Meeting in May (Zambia)
  - Grants Management Workshop
  - Community Engagement Training Workshop
  - 2nd Ethics Consultation Meeting

- H3Africa and South African ethics guidelines
- 3 New ELSI awards
- Alliance for Accelerating Excellence in Science in Africa (AESA)

Document 35

Precision Medicine Initiative



Document 36

Precision Medicine Initiative: Building a Large U.S. Research Cohort

February 11-12, 2015

Porter Neuroscience Building
NIH Campus

Advisory Committee to the NIH Director
Working Group on the Precision Medicine Initiative

Co-Chairs:

Richard Lifton, MD, PhD
Bray Patrick-Lake, MFS
Kathy Hudson, PhD

- Esteban Gonzalez Burchard, MD, MPH
- Tony Coles, MD, MPH
- Rory Collins, FMedSci
- Andrew Conrad, PhD
- Josh Denny, MD
- Susan Desmond-Hellmann, MD, MPH
- Eric Dishman
- Kathy Giusti, MBA
- Sekar Kathiresan, MD
- Sachin Kheterpal, MD, MBA
- Shiriki Kumanyika, PhD, MPH
- Spero M. Manson, PhD
- P. Pearl O'Rourke, MD
- Richard Platt, MD
- Jay Shendure, MD, PhD
- Sue Siegel

Precision Medicine Initiative: Next Steps

- **Working Group of Advisory Committee to the Director**
  - Intense planning for next ~3-4 months
  - Interim report in September
- **Past and future meetings/workshops**
- **Implementation**
  - Trans-NIH implementation model
  - Fall 2015: Initial funding opportunities announced
  - Fiscal Year 2016: Funding begins
  - Coordination with other U.S. government agencies

National Institutes of Health
Office of Extramural Research

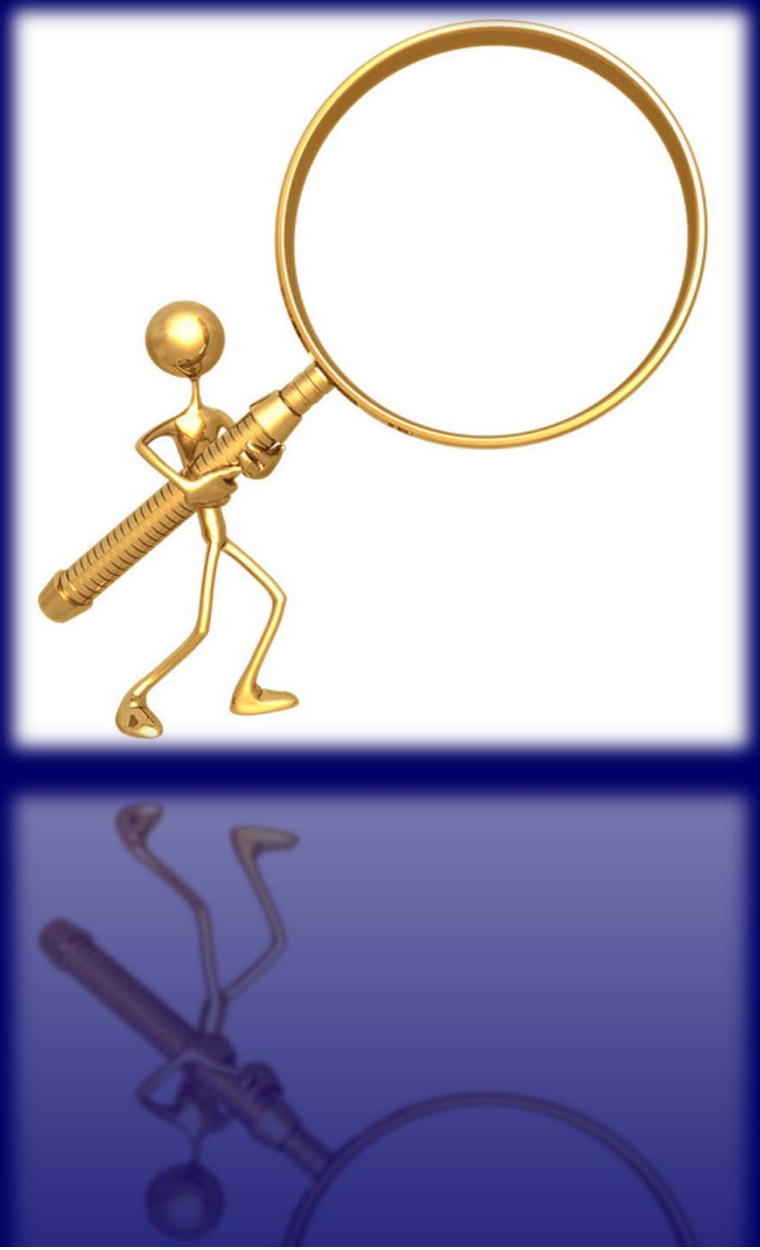
Request for Information: NIH Precision Medicine Cohort

Purpose

This Request for Information (RFI) seeks feedback to help guide the National Institutes of Health (NIH) in creating a longitudinal cohort of 1 million or more Americans who have volunteered to participate in research as part of the President's proposed Precision Medicine Initiative. Participants will be asked to give consent for extensive characterization of biologic specimens (potentially including cell populations, proteins, metabolites, RNA, and DNA whole-genome sequencing, if/when costs permit) and behavioral and environmental data, all linked to their electronic health records (EHRs). Qualified researchers from many organizations will, with appropriate protection of participant confidentiality, have access to the cohort's de-identified data for research and analysis.

Due Date: May 7, 2015

Emphasis on Transparency



PRECISION MEDICINE INITIATIVE

Precision Medicine Initiative

- Near-term Goals
- Longer-term Goals
- Scale and Scope
- Participation
- PMI Working Group
- Events
- Advisory Committees
- PMI in the News

The Precision Medicine Initiative: Infographic
View larger (PDF - 163KB)

Read blogs by the NIH Director about precision medicine

Senate hearing: Continuing America's Leadership: Realizing the Promise of Precision Medicine for Patients

ABOUT THE PRECISION MEDICINE INITIATIVE

Far too many diseases do not have a proven means of prevention or effective treatments. We must gain better insights into the biology of these diseases to make a difference for the millions of Americans who suffer from them. Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person. While significant advances in precision medicine have been made for select cancers, the practice is not currently in use for most diseases. Many efforts are underway to help make precision medicine the norm rather than the exception. To accelerate the pace, President Obama unveiled the Precision Medicine Initiative (PMI) – a bold new enterprise to revolutionize medicine and generate the scientific evidence needed to move the concept of precision medicine into every day clinical practice.

Read about the scientific justification of the Precision Medicine Initiative in a *New England Journal of Medicine* Perspective by NIH Director Dr. Francis S. Collins and former NCI director Dr. Harold Varmus.

Email Updates

To sign up for updates please enter your e-mail address.

Submit

Related Links

- NEJM Perspective: A New Initiative on Precision Medicine
- White House Precision Medicine Web Page
- White House Fact Sheet: President Obama's Precision Medicine Initiative
- Precision Medicine Initiative and Cancer Research
- Storify: The Precision Medicine Initiative
- Precision Medicine Initiative YouTube Channel

www.nih.gov/precisionmedicine

Document 36

PRECISION MEDICINE INITIATIVE

Precision Medicine Initiative

- Near-term Goals
- Longer-term Goals
- Scale and Scope
- Participation
- PMI Working Group
- Events
- Announcements
- PMI in the News

The Precision Medicine Initiative: Infographic
View larger (PDF - 163KB)

Events

The following are upcoming and past events held by the NIH to gather input from participant, scientific, and other stakeholder groups as it plans the development of the President's Precision Medicine Initiative and the vision for building the national participant group. Please sign up for updates or check back frequently for additional meeting information.

All public workshops will be videocast live.

UPCOMING EVENTS

- Public workshop on cohorts and electronic health records
  Thursday, May 28 - Friday, May 29, 2015, Time to be Determined
  Vanderbilt University, Nashville, Tenn.
- Public workshop on participants and community engagement
  Wednesday, July 1 - Thursday, July 2, 2015, Time to be Determined
  National Institutes of Health, Bethesda, Md.
- Public workshop on mHealth
  Monday, July 27 - Tuesday, July 28, 2015
  Intel Corp., Santa Clara, Calif.

PAST EVENTS

2015

- Public workshop on unique scientific opportunities for the national research cohort
  Tuesday, April 28 - Wednesday, April 29, 2015
  National Institutes of Health, Bethesda, Md.
- Public Workshop on Building a Precision Medicine Research Cohort
  Wednesday, February 11 - Thursday, February 12, 2015
  National Institutes of Health, Porter Neuroscience Building, Bethesda, Md.

Email Updates

To sign up for updates please enter your e-mail address.

Submit

Related Links

- NEJM Perspective: A New Initiative on Precision Medicine
- White House Precision Medicine Web Page
- White House Fact Sheet: President Obama's Precision Medicine Initiative
- Precision Medicine Initiative and Cancer Research
- Storify: The Precision Medicine Initiative
- Precision Medicine Initiative YouTube Channel

PRECISION MEDICINE INITIATIVE

Precision Medicine Initiative

- Near-term Goals
- Longer-term Goals
- Scale and Scope
- Participation
- PMI Working Group
- Events
- Announcements
- PMI in the News

The Precision Medicine Initiative: Infographic
View larger (PDF - 163KB)

ACD Precision Medicine Initiative Working Group Public Workshop

Unique Scientific Opportunities for the Precision Medicine Initiative National Research Cohort

NIH hosted a public workshop on the NIH campus in Bethesda, Maryland, April 28-29, 2015, to consider visionary biomedical questions that could be addressed by the proposed national research cohort of one million or more volunteer participants. The workshop will result in a series of use cases describing the distinctive science that the cohort could enable in the near term and longer term.

This workshop is one of four that is being convened by the Precision Medicine Initiative Working Group of the Advisory Committee to the (NIH) Director to help inform the vision for building the PMI national participant group that they have been tasked to develop.

Workshop Videos

- Day 1 Videocast
- Day 2 Videocast

Follow the Conversation on Twitter (#PMINetwork)

Workshop Agenda (PDF - 115KB)

Speaker Biographies

PMI ACD Working Group Biographies (PDF - 451KB)

Email Updates

To sign up for updates please enter your e-mail address.

Submit

Related Links

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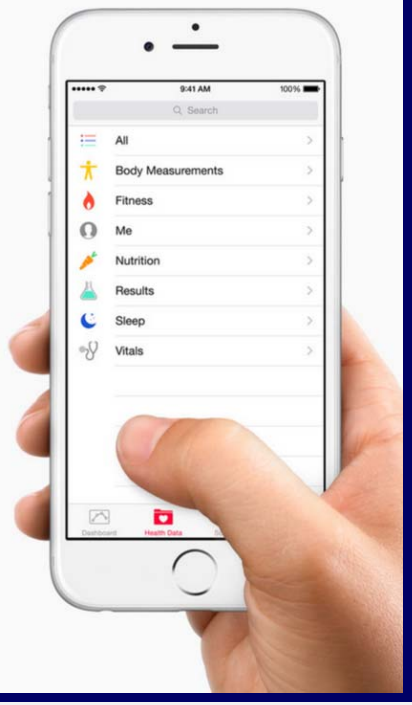
- Precision Medicine Initiative and Cancer Research
- Storify: The Precision Medicine Initiative
- Precision Medicine Initiative YouTube Channel

Apple Announces ResearchKit

You're already carrying a powerful medical research tool.

Hundreds of millions of people around the world have an iPhone in their pocket. Each one is equipped with powerful processors and advanced sensors that can track movement, take measurements, and record information — functions that are perfect for medical studies. The sheer number of them being used across the globe opens up new possibilities for researchers. With ResearchKit, researchers can easily create apps that take advantage of iPhone features to gather new types of data on a scale never available before.



Taking research out of the lab and into the real world.

Until now, taking part in a medical study has usually required traveling to a hospital or facility to complete tasks and fill out questionnaires. With ResearchKit, you can use your iPhone to perform activities and generate data wherever you are, providing a source of information that is more objective than ever possible before. This is invaluable to the progress of medical research — and we can all have a hand in it.

What's more, many of the apps built with ResearchKit will enable you to track your own data and potentially discover correlations between symptoms and daily actions such as diet or exercise.

Share your data. Keep your privacy.

We know how much you value the privacy of your information, and ResearchKit has been designed with that in mind. You choose what studies you want to join, you are in control of what information you provide to which apps, and you can see the data you're sharing.

ResearchKit is already being used today.

Several of the world's leading medical institutions are already using ResearchKit to gain further insight into some of our most serious diseases. Learn more about their stories below. You can also download these apps from the App Store ›

Asthma | Parkinson's Disease | Diabetes | Breast Cancer | Cardiovascular Disease

Document 36

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

Inter-Society Coordinating Committee for Practitioner Education in Genomics

- **New Co-Chairs:**

Bob Wildin, M.D.
NHGRI

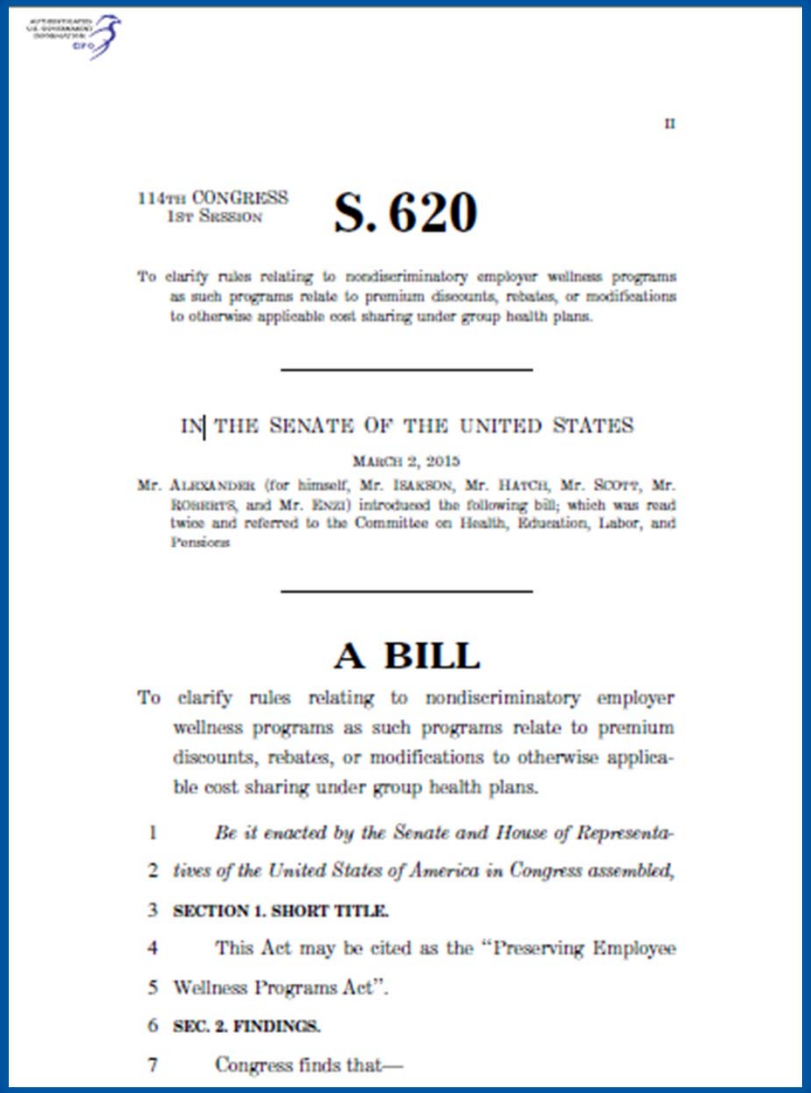
Ann Karty, M.D.
American Academy of Family Physicians

- **In-person meeting in May:**

  Patient Safety and Genomic Medicine
  Genetic Counselors' Roles in Education
  Point-of-Care Education Modes
  Inter-Professional Education Collaborative (IPEC)

Document 37

Preserving Employee Wellness Programs Act (S.620/H.R.1189)



II

114TH CONGRESS
1ST SESSION

S. 620

To clarify rules relating to nondiscriminatory employer wellness programs as such programs relate to premium discounts, rebates, or modifications to otherwise applicable cost sharing under group health plans.

IN THE SENATE OF THE UNITED STATES

MARCH 2, 2015

Mr. ALEXANDER (for himself, Mr. ISAKSON, Mr. HATCH, Mr. SCOTT, Mr. ROBERTS, and Mr. ENZI) introduced the following bill; which was read twice and referred to the Committee on Health, Education, Labor, and Pensions

---

A BILL

To clarify rules relating to nondiscriminatory employer wellness programs as such programs relate to premium discounts, rebates, or modifications to otherwise applicable cost sharing under group health plans.

Be it enacted by the Senate and House of Representatives of the United States of America in Congress assembled,

SECTION 1. SHORT TITLE.

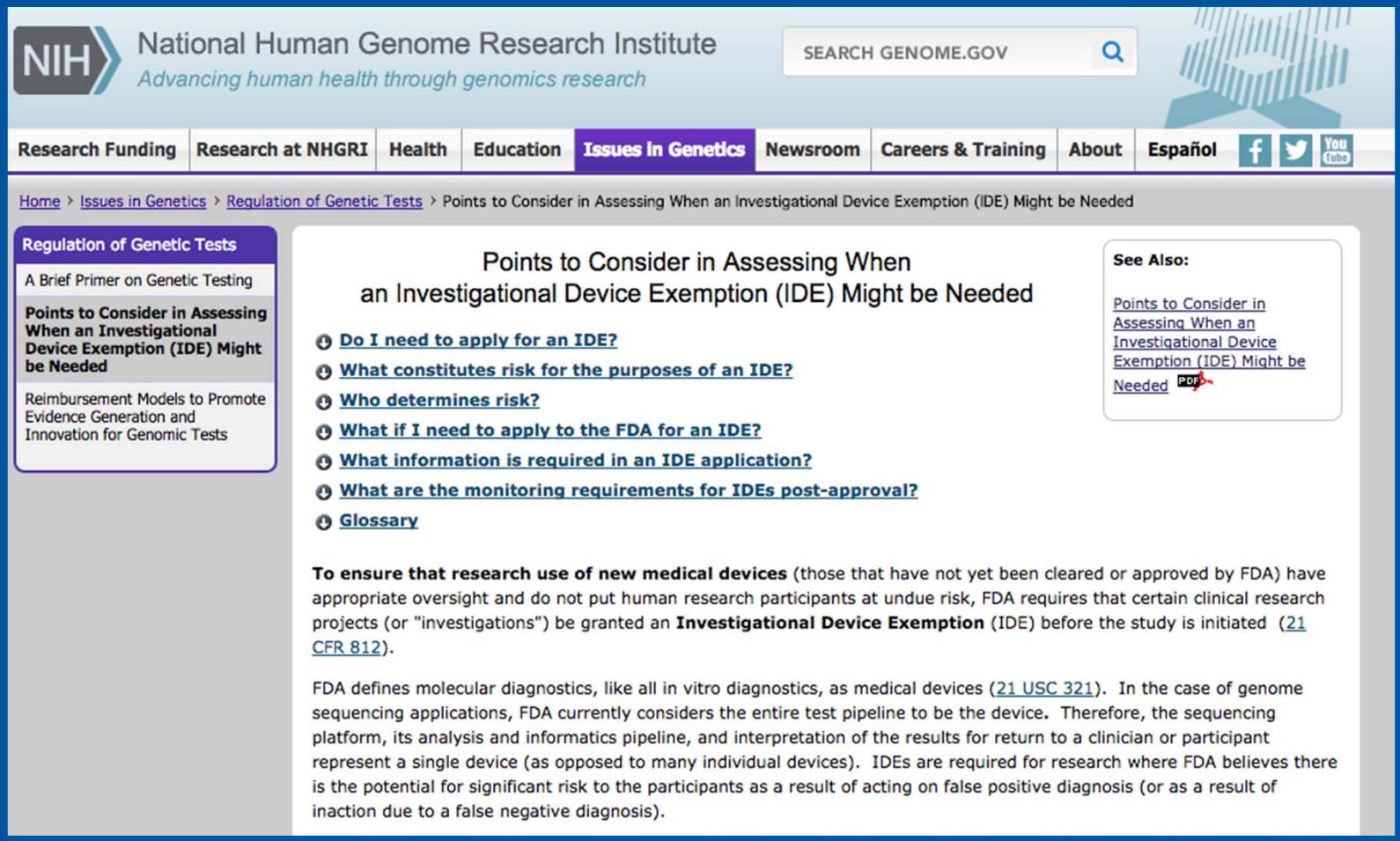
This Act may be cited as the "Preserving Employee Wellness Programs Act".

SEC. 2. FINDINGS.

Congress finds that—

Document 38

NHGRI IDE Resources for Research Community



National Human Genome Research Institute
Advancing human health through genomics research

SEARCH GENOME.GOV

Research Funding | Research at NHGRI | Health | Education | Issues in Genetics | Newsroom | Careers & Training | About | Español

Home › Issues in Genetics › Regulation of Genetic Tests › Points to Consider in Assessing When an Investigational Device Exemption (IDE) Might be Needed

Regulation of Genetic Tests

- A Brief Primer on Genetic Testing
- **Points to Consider in Assessing When an Investigational Device Exemption (IDE) Might be Needed**
- Reimbursement Models to Promote Evidence Generation and Innovation for Genomic Tests

Points to Consider in Assessing When an Investigational Device Exemption (IDE) Might be Needed

See Also:

Points to Consider in Assessing When an Investigational Device Exemption (IDE) Might be Needed PDF

- **Do I need to apply for an IDE?**
- **What constitutes risk for the purposes of an IDE?**
- **Who determines risk?**
- **What if I need to apply to the FDA for an IDE?**
- **What information is required in an IDE application?**
- **What are the monitoring requirements for IDEs post-approval?**
- **Glossary**

To ensure that research use of new medical devices (those that have not yet been cleared or approved by FDA) have appropriate oversight and do not put human research participants at undue risk, FDA requires that certain clinical research projects (or "investigations") be granted an **Investigational Device Exemption** (IDE) before the study is initiated (21 CFR 812).

FDA defines molecular diagnostics, like all in vitro diagnostics, as medical devices (21 USC 321). In the case of genome sequencing applications, FDA currently considers the entire test pipeline to be the device. Therefore, the sequencing platform, its analysis and informatics pipeline, and interpretation of the results for return to a clinician or participant represent a single device (as opposed to many individual devices). IDEs are required for research where FDA believes there is the potential for significant risk to the participants as a result of acting on false positive diagnosis (or as a result of inaction due to a false negative diagnosis).

Document 39

LabTV

A website of mini-documentaries about scientists who are passionate about their work

Document 40

Capturing the Historical Legacy of the NHGRI: An Archival and Scholarly Initiative

Capturing the History of Genomics: A Workshop

Document 41

Genome: Unlocking Life's Code Exhibition

"Big Data, Genomics, and Precision Medicine"

Document 42

Genome: Unlocking Life's Code Exhibition

The Tech Museum of Innovation Family Day

The Tech
Museum of Innovation

TICKETS STORE MY ACCOUNT NEWSLETTER THE TECH TAG DONATE

Spirit of Silicon Valley

Visit | Programs | Educators | IMAX | Members | Support Us | About Us | Search

more

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of Events
New
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les
Needs
Store
Cafe

Epic Genetics Day

Saturday, April 18
10 a.m. to 5 p.m.
Presented by Pacific Biosciences
RSVP and share on Facebook

Join us for a day devoted to DNA. See the Smithsonian exhibition, Genome: Unlocking Life's Code, before it closes! Hands-on activities will explore and explain how DNA works, and you can even learn how the dog genome has provided deeper understanding of human disease.

Epic Genetics Day is free with museum admission on one day only, April 18.

Hands-on activities
11 a.m. to 4 p.m.

- Learn how natural selection works using the ability to drink milk as an adult as an example.
- Brush cells from inside your cheek, stain the cells, and look at them under a microscope. (You can even keep a picture of your cells!)
- Pull DNA from your cheek cells for examination, then isolate it in a necklace you can take home.
- Solve a murder mystery by looking at DNA patterns using the same tools as a forensic scientist.
- Make a bracelet that celebrates your unique collection of genetic traits. We've created custom, 3D printed beads to represent these traits, so you can be sure to go home with a bracelet as original as you are! (While supplies last; regular beads will be substituted once the custom 3D printed beads run out.)

More offerings
Play Go Extinct! The Game. Go Extinct! is a fun, strategic card game based on Go Fish, but played with animal cards that span the evolutionary tree. Sit down, play a game, and learn about evolution and the evidence used to understand how land animals are related.

Explore UC Santa Cruz's Genome Browser – an interactive look at a wide range of genome sequencing data – which was the original resource to distribute the research of the Human Genome Project.

Got a question? Ask a geneticist! Our resident expert Dr. Barry Starr, director of the Stanford @ The Tech Program, will be on hand from 10 a.m. to noon.

Help The Tech develop its new exhibit about synthetic biology and bio-design — check out our prototypes and give crucial feedback to our team of exhibit developers.

Special event!
Did you know the canine genome is remarkably similar to that of humans? Learn more about how research into the genetic makeup of our four-legged friends is shedding light on human disease. Dr. Elaine Ostrander of the National Human Genome Research Institute will speak at 1 p.m. in New Venture Hall, on the ground floor of the museum. RSVP and share on Facebook.

Document 42

Genome: Unlocking Life's Code Exhibition

Travel Schedule

2015

May 15 to September 10
Saint Louis Science Center (St. Louis, MO)

October 2 to January 3
Oregon Museum of Science and Industry (Portland, OR)

2016

January 18 to April 25
Discovery World Milwaukee (Milwaukee, WI)

Document 42

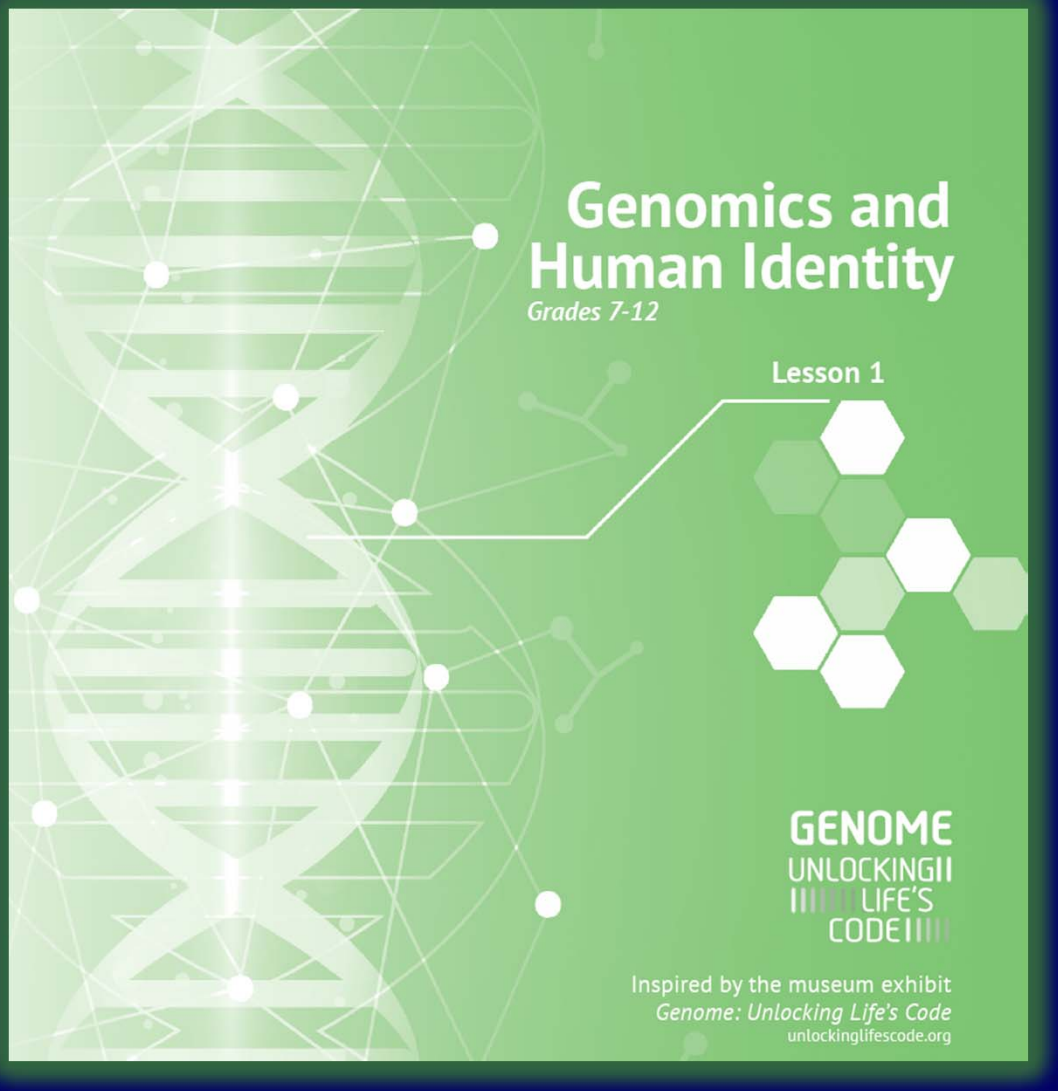
Genome: Unlocking Life's Code Exhibition

Website Interactive

Document 42

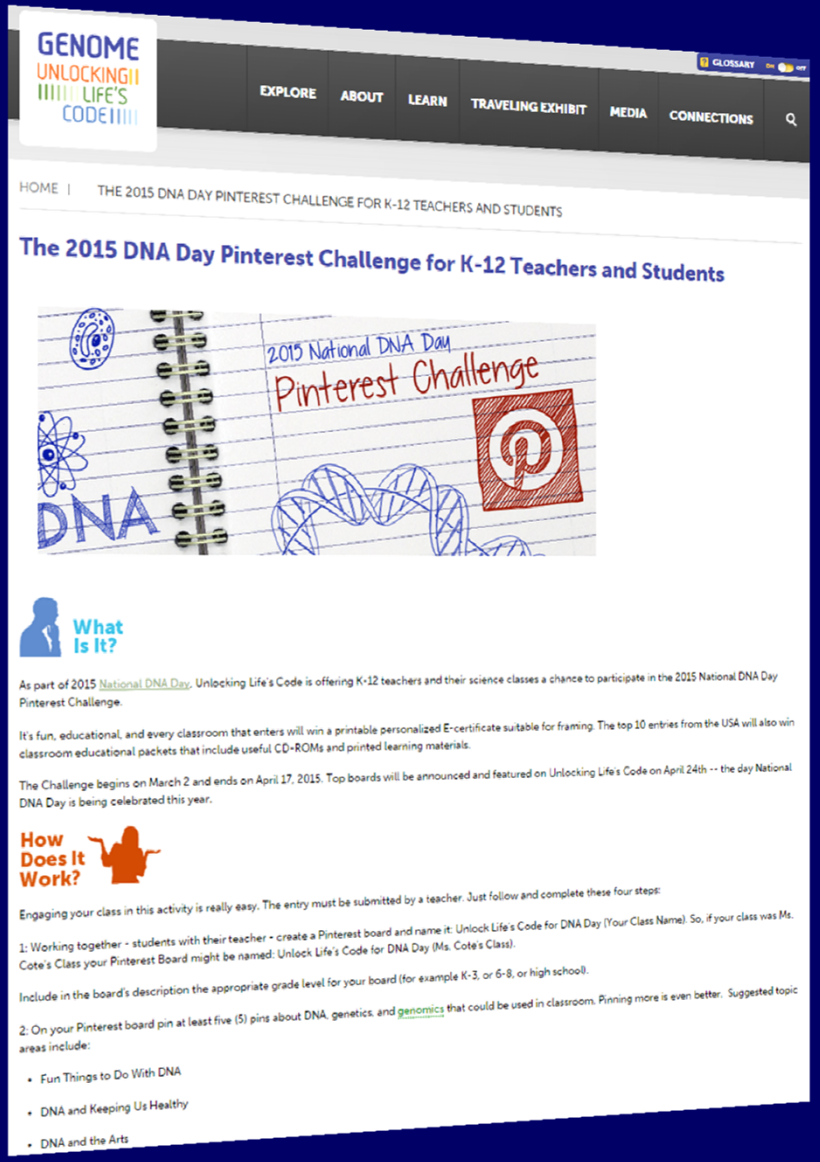
Genome: Unlocking Life's Code Exhibition

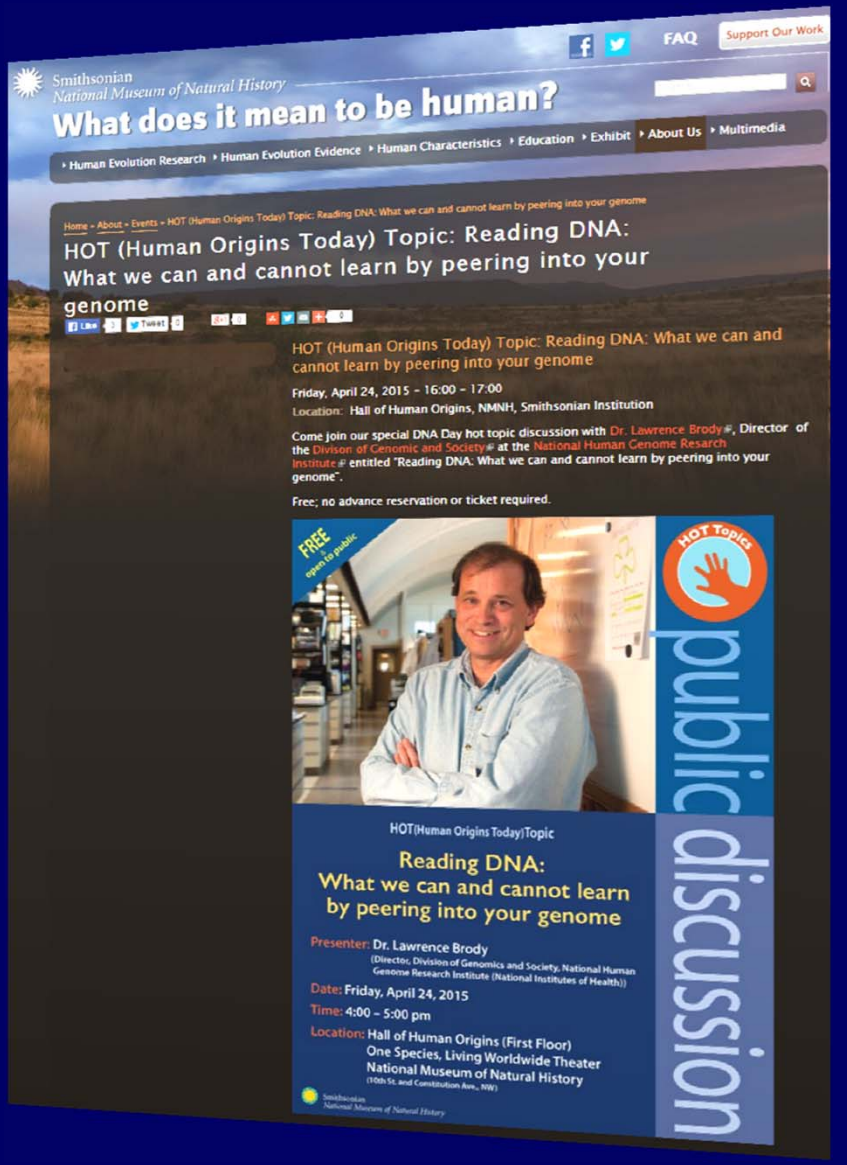
Human Identity Lesson Plan



Document 42

National DNA Day





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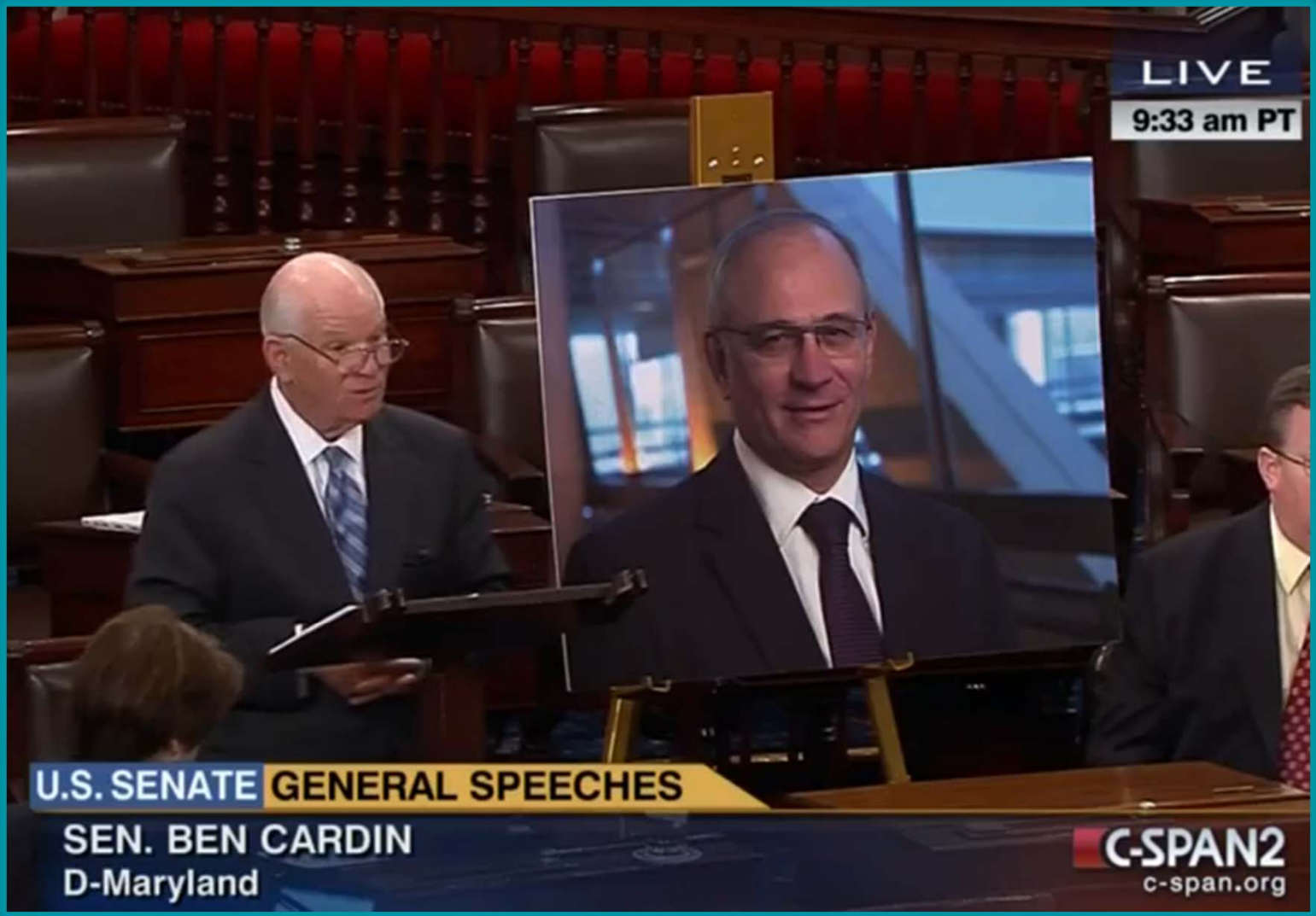
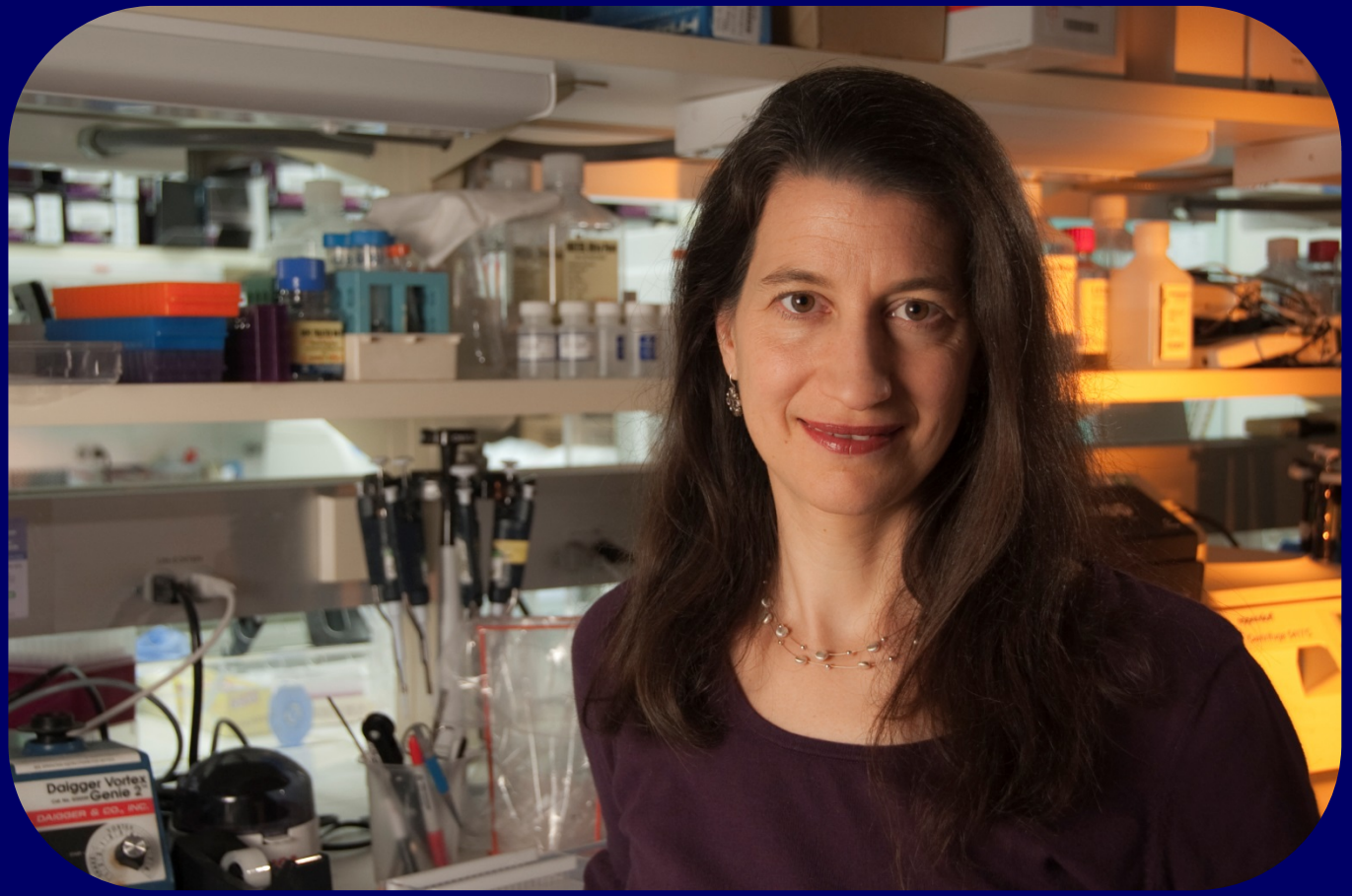
Senator Cardin Applauds NHGRI's Bill Gahl



Document 44

American Academy of Microbiology Fellow



Julie Segre, Ph.D.

NHGRI Intramural Research Highlights

Molecular Cancer Research

Homologous mutation to human BRAF V600E is common in naturally occurring canine bladder cancer-evidence for a relevant model system and urine-based diagnostic test

JHP Journal of Health Psychology

Mothers' guilt responses to children's obesity risk feedback

DMM Disease Models & Mechanisms

Phenotype –driven chemical screening in zebrafish for collective cell migration inhibitors identifies multiple potential pathways for targeting metastasis

The Genomics Landscape

A monthly update from the NHGRI Director

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

Search for NHGRILANDSCAPE

Past issues can be accessed at: genome.gov/27527308

genome.gov
National Human Genome Research Institute
National Institutes of Health

Thanks!

Special Thanks!