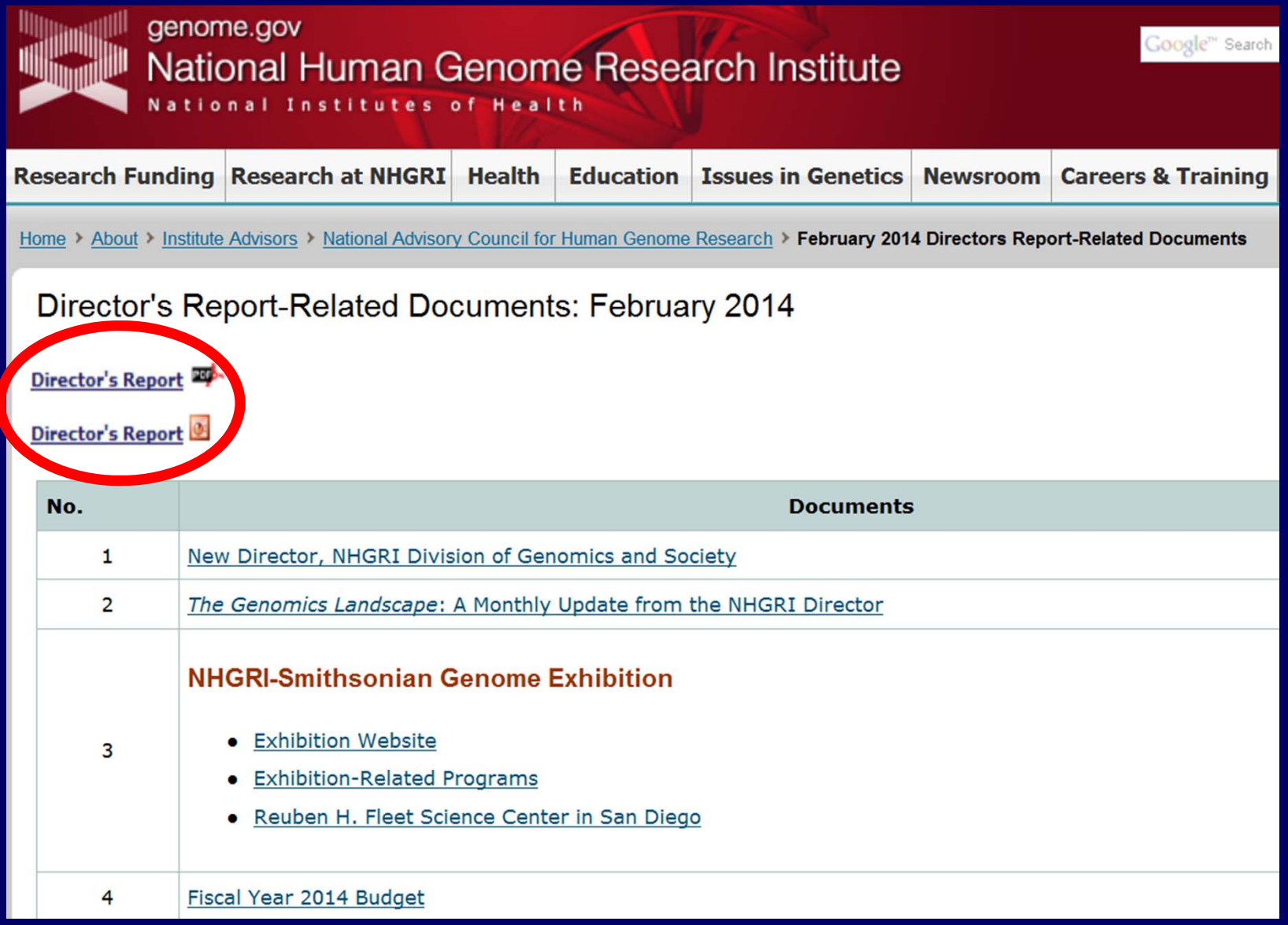
NIH National Human Genome Research Institute

DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

February 2014

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



genome.gov/DirectorsReport

Document #

Open Session Presentations

Major Presentations:

- **NHGRI Intramural Research Program**

  Dan Kastner

- **National Center for Advancing Translational Sciences (NCATS)**

  Chris Austin

Recent NHGRI Meeting:

- **Genomic Medicine VI Meeting**

  Teri Manolio

Open Session Presentations

Project Update:

- **H3Africa Initiative**

  Jane Peterson

Major Presentation:

- **NHGRI Large-Scale Sequencing and Analysis Centers**

  Richard Gibbs

  Eric Lander

  Richard Wilson

Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

IV. NHGRI Extramural Research Program

V. NIH Common Fund/Trans-NIH

VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

Director's Report Outline

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

Extramural Staff Departure

Jane Peterson, Ph.D.

Extramural Staff Departure

Carson Loomis, Ph.D.

New Director,
NHGRI Division of Genomics and Society

Larry Brody, Ph.D.

Document 1

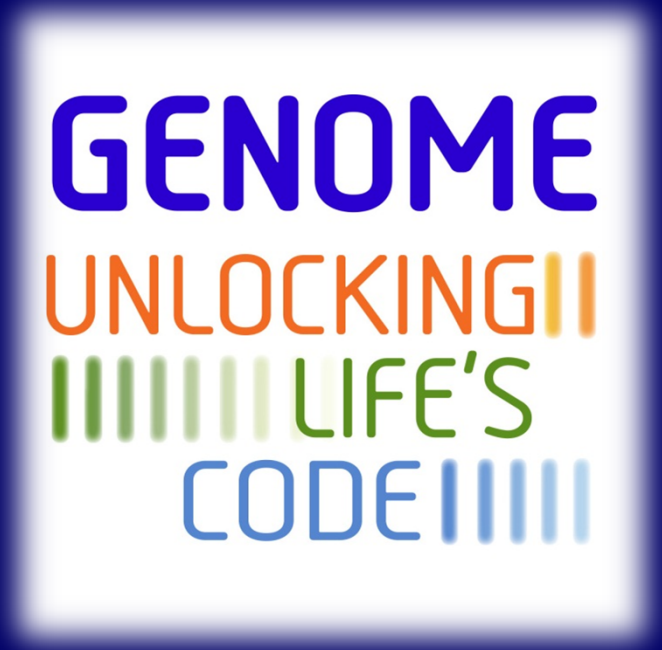
The Genomics Landscape

A monthly update from the NHGRI Director

October 2013:	**Welcome Fiscal Year 2014! - Or Not?**
November 2013:	**Announcing the First Director of NHGRI's Division of Genomics and Society**
December 2013:	**Jumping into the Deep End of Genomic Medicine**
January 2014:	**A Decade of the NHGRI Social and Behavioral Research Branch**
February 2014:	**The Big Data to Knowledge Initiative**

Document 2

Genome: Unlocking Life's Code Exhibition

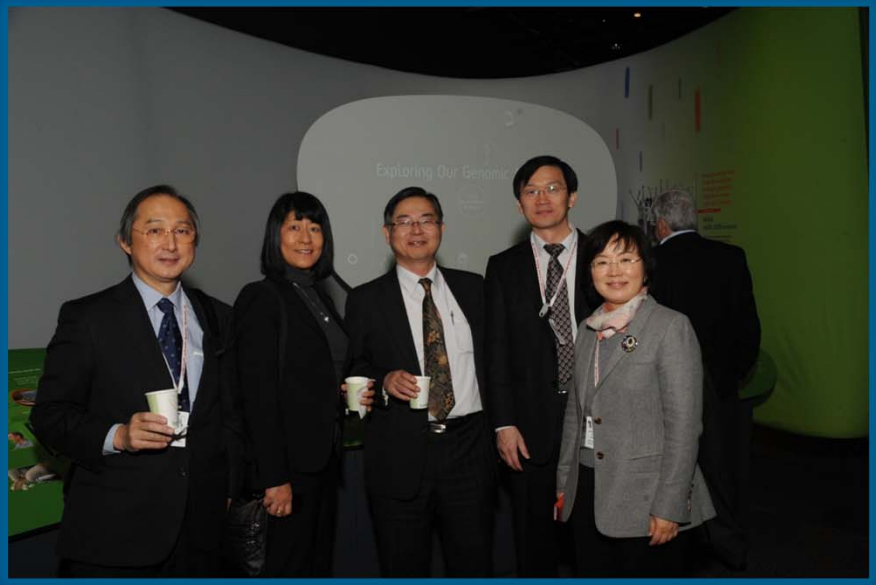


- More than 1.6 million visitors have seen the exhibition since its opening in June
- More than 70 NIH scientists have volunteered within the exhibition

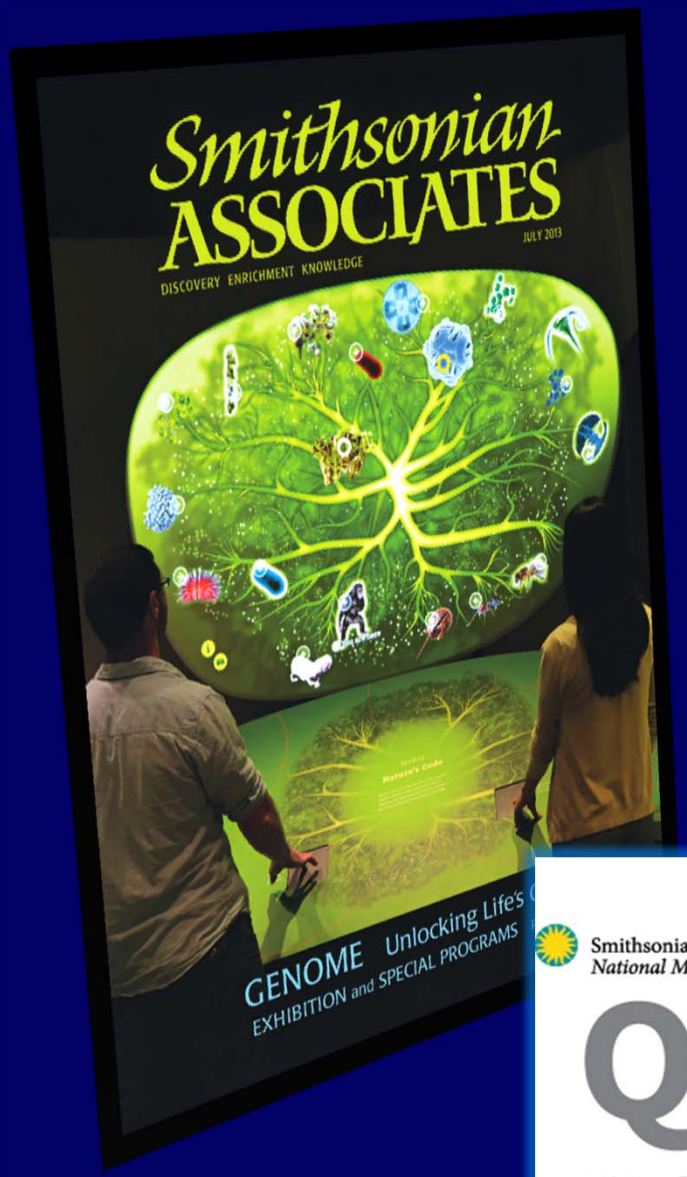
Document 3

Genome Exhibition Attracting VIP Visitors

Genome Exhibition Attracting VIP Visitors



Genome Exhibition Programs



Smithsonian
National Museum of Natural History

Q?rius

Unlock your world.

Document 3

Genome Exhibition Travel

Reuben H. Fleet Science Center

September 24, 2014 to January 4, 2015

Document 3

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

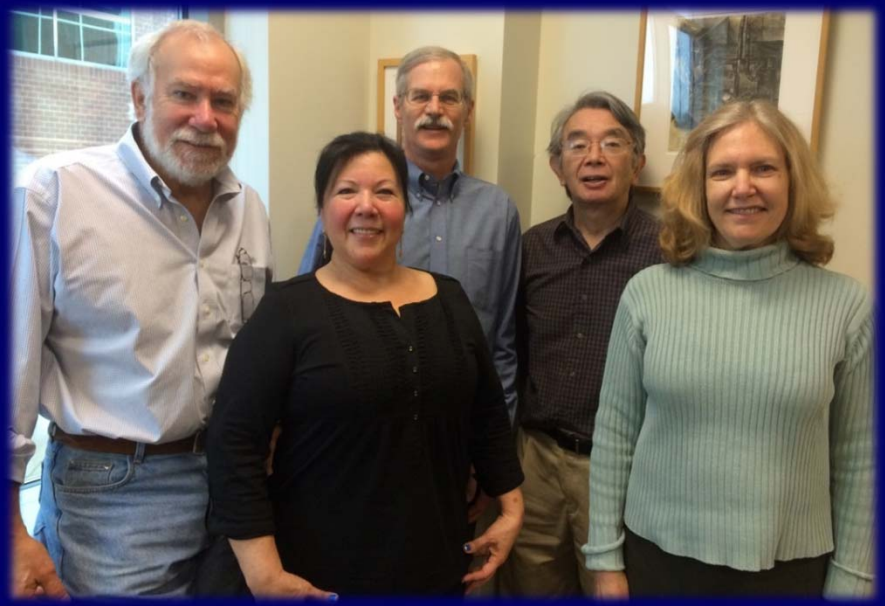
VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

Government Shutdown 2013

Special ‘Shout Out’ to NIH Scientific Review Officers

- Government shutdown required rescheduling of many review meetings
- Center for Scientific Review and institute Scientific Review Officers quickly rescheduled meetings
- Summary statements prepared over holiday season



NIH National Human Genome Research Institute

NIH Center for Scientific Review

Fiscal Year 2014 Budget

- Congressional deal avoided Fiscal Year 2014 sequestration
- Congress passed Fiscal Year 2014 omnibus bill in January

	FY2011	FY2012	FY2013	FY2014
NIH	$30.7 B	$30.7 B	$29.2 B	$30.1 B (3.4%)
NHGRI	$511 M	$513 M	$483 M	$498 M (3.0%)

Document 4

Francis Collins Editorial

The Washington Post

Back to previous page

Investing in the nation's health

By Francis S. Collins, Published: December 24

Francis S. Collins is director of the National Institutes of Health.

Biomedical research is at a critical juncture — a moment of exceptional opportunities that demand exceptional attention if their promise is to be fully realized. Many of the most exciting possibilities stem from the convergence of several factors: innovative tools and technologies arising from the Human Genome Project; advances in computers and biomedical imaging that are fueling a generation of complex digital data sets known as "big data"; and increased interest by both public and private sectors in finding ways to accelerate the rate at which research findings are turned into treatments and cures.

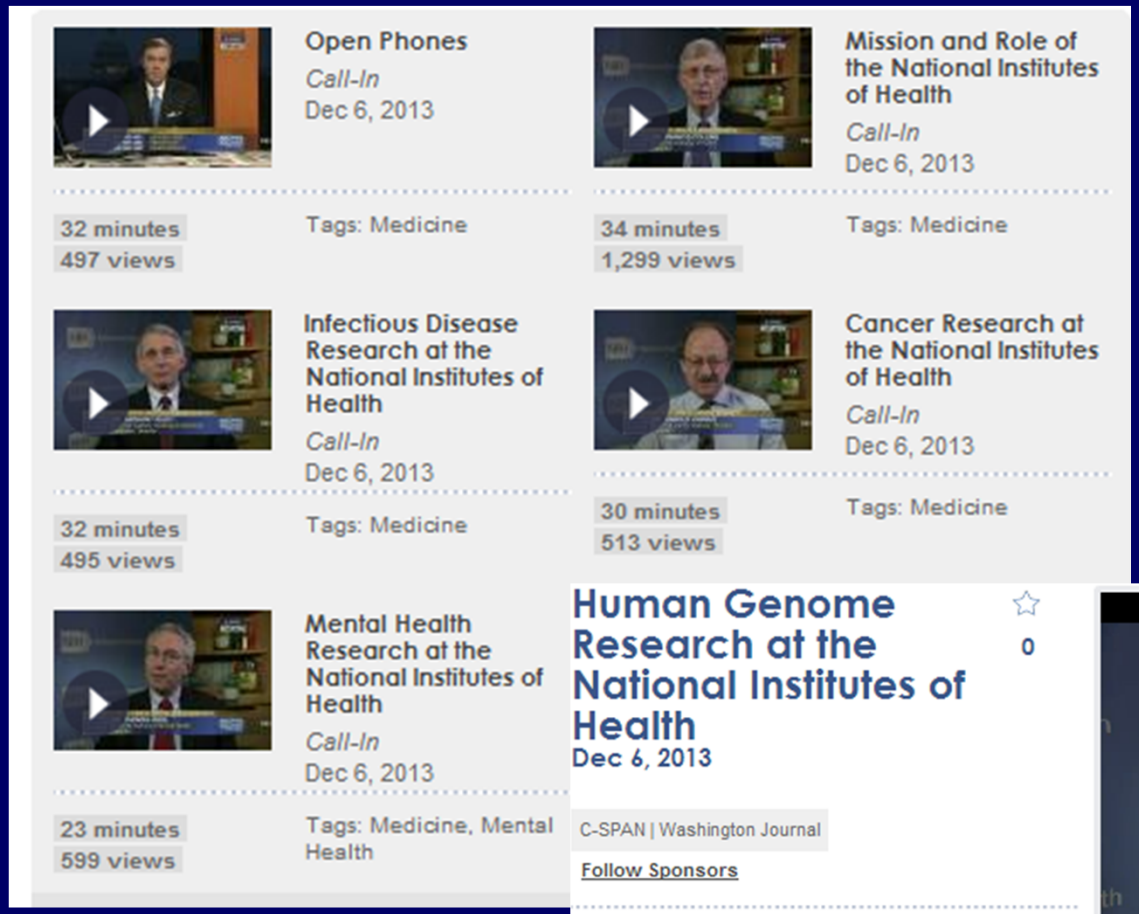
The National Institutes of Health (NIH) is responsible for turning scientific discoveries into better health, but a combination of sequestration-mandated spending cuts and budgets that have not kept pace with biomedical research inflation over the past 10 years has weakened NIH's ability to carry out its mission.

One transformative program that hangs in the balance is the Brain Research through Advancing Innovative Neurotechnologies (BRAIN) Initiative, part of a focus aimed at revolutionizing understanding of the human brain. By catalyzing development and application of new technologies, researchers plan to produce a dynamic picture of the brain that shows, for the first time, how individual cells and complex neural circuits interact in both time and space. This could mean enormous advances for efforts to treat and possibly even prevent epilepsy, schizophrenia, Alzheimer's disease, autism, Parkinson's disease, traumatic brain injury and many other neurological conditions.

Another area of great promise is the ability to assemble very large data sets of medical research information. The advent of electronic health records will dramatically accelerate the "big data revolution." Details can be understood by focusing on one disease: cancer. Taking advantage of breathtaking advances in DNA-sequencing technology, NIH-funded researchers working on the Cancer Genome Atlas have conducted comprehensive analyses of more than 20 different types of cancer and plan to study dozens more. Besides expanding understanding of the molecular roots of different cancers, this pioneering work has opened doors to new therapeutic targets, as well as to new and more precise uses of chemotherapy drugs. Unfortunately, this mountain of data will be of limited use to cancer patients if researchers and clinicians lack the tools necessary to manipulate and mine it effectively.

Document 5

C-SPAN's Washington Journal: NIH Feature



Open Phones
Call-In
Dec 6, 2013
32 minutes
497 views
Tags: Medicine

Mission and Role of the National Institutes of Health
Call-In
Dec 6, 2013
34 minutes
1,299 views
Tags: Medicine

Infectious Disease Research at the National Institutes of Health
Call-In
Dec 6, 2013
32 minutes
495 views
Tags: Medicine

Cancer Research at the National Institutes of Health
Call-In
Dec 6, 2013
30 minutes
513 views
Tags: Medicine

Mental Health Research at the National Institutes of Health
Call-In
Dec 6, 2013
23 minutes
599 views
Tags: Medicine, Mental Health

Human Genome Research at the National Institutes of Health

Dec 6, 2013

0

C-SPAN | Washington Journal

Follow Sponsors

Dr. Eric Green talked about the state of human genome research and the changes it was bringing to medical treatment in the U.S.

The majority of the December 6, 2013, "Washington Journal" focused on .. Read More

Document 6

Representative Michael Burgess, M.D. (R-TX)

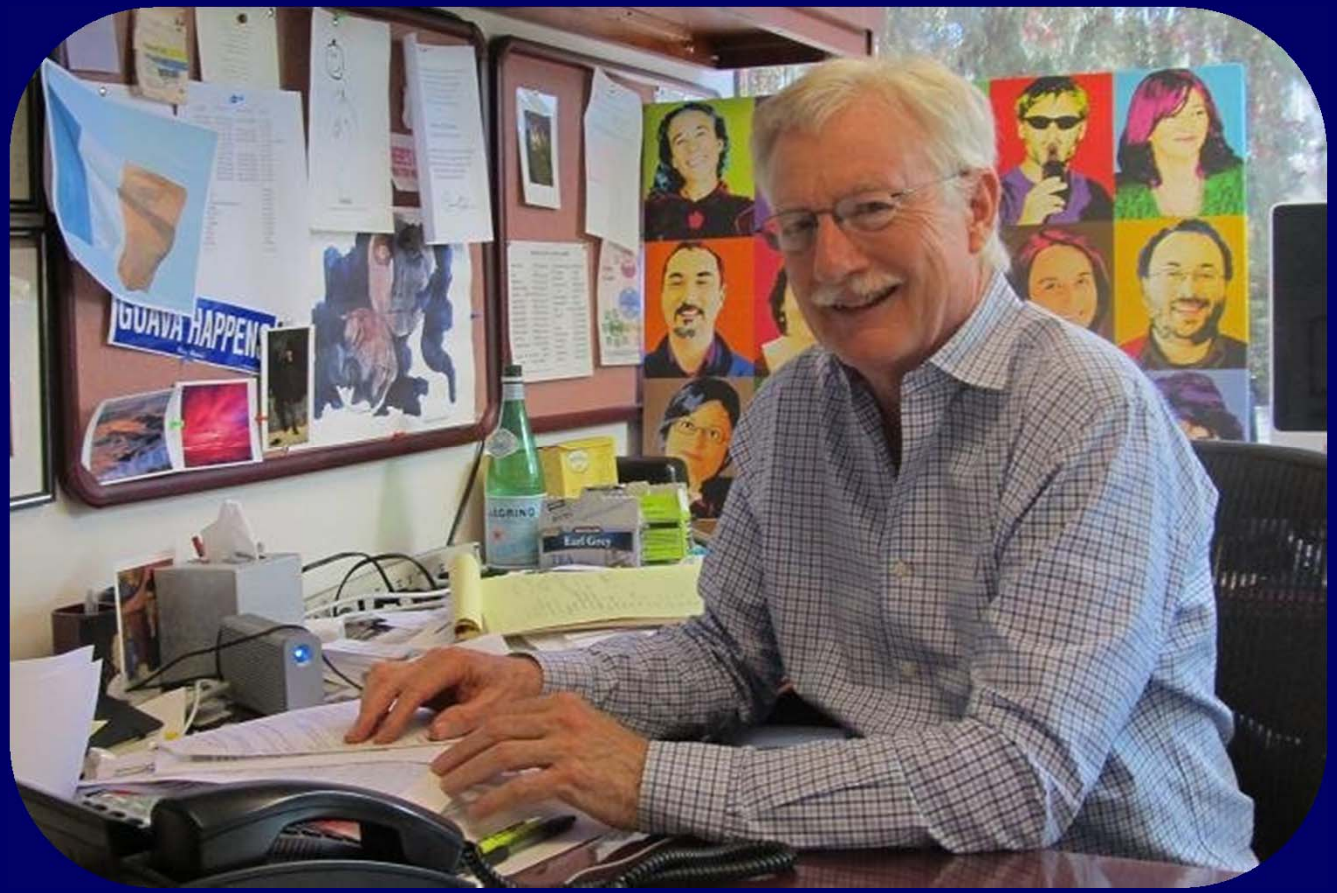
November, 2013

THE HILL

"The true transformation comes with how these [genome] sequences are starting to change the way that doctors treat patients and the extraordinary therapies that could result… This is just the start of a new golden age in medicine.

Document 7

New Director, National Institute of Alcohol Abuse and Alcoholism



George Koob, Ph.D.

Document 8

First NIH Associate Director for Data Science

Philip Bourne, Ph.D.

First NIH Chief Officer for Scientific Workforce Diversity

Hanna Valantine, M.D.

Surgeon General Nomination

Vivek Murthy, M.D.

Document 11

NIH Genomic Data Sharing Policy

DEPARTMENT OF HEALTH AND HUMAN SERVICES

National Institutes of Health

Draft NIH Genomic Data Sharing Policy Request for Public Comments

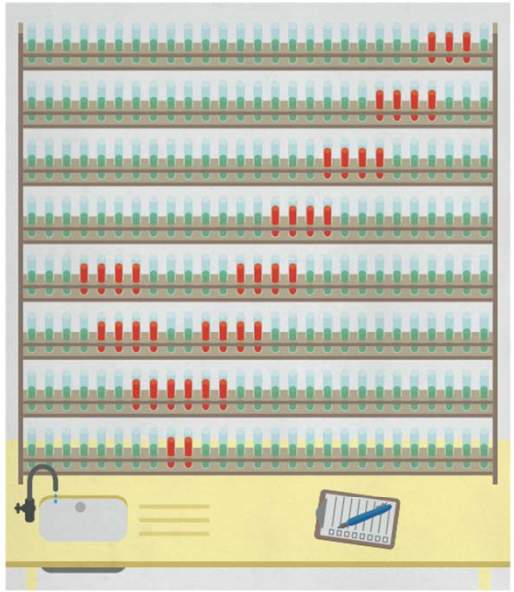
SUMMARY: The National Institutes of Health (NIH) is seeking public comments on the draft Genomic Data Sharing (GDS) Policy that promotes sharing, for research purposes, of large-scale human and nonhuman genomic [1] data generated from NIH-supported and NIH-conducted research.

DATES: To ensure that your comments will be considered, please submit your response to this Request for Comments no later than 60 days after publication of this notice.

Document 12

NIH Plan to Enhance Reproducibility

COMMENT



CHRIS RYAN/NATURE

NIH plans to enhance reproducibility

Francis S. Collins and **Lawrence A. Tabak** discuss initiatives that the US National Institutes of Health is exploring to restore the self-correcting nature of preclinical research.

A growing chorus of concern, from scientists and laypeople, contends that the complex system for ensuring the reproducibility of biomedical research is failing and is in need of restructuring[1,2]. As leaders of the US National Institutes of Health (NIH), we share this concern and here explore some of the significant interventions that we are planning.

Science has long been regarded as 'self-correcting', given that it is founded on the replication of earlier work. Over the long term, that principle remains true. In the shorter term, however, the checks and balances that once ensured scientific fidelity have been hobbled. This has compromised the ability of today's researchers to reproduce others' findings.

Let's be clear: with rare exceptions, we have no evidence to suggest that irreproducibility is caused by scientific misconduct. In 2011, the Office of Research Integrity of the US Department of Health and Human Services pursued only 12 such cases[3]. Even if this represents only a fraction of the actual problem, fraudulent papers are vastly outnumbered by the hundreds of thousands published each year in good faith.

Instead, a complex array of other factors seems to have contributed to the lack of reproducibility. Factors include poor training of researchers in experimental design; increased emphasis on making provocative statements rather than presenting technical details; and publications that do not report basic elements of experimental design[4]. Crucial experimental design elements that are all too frequently ignored include blinding, randomization, replication, sample-size calculation and the effect of sex differences. And some scientists reputedly use a 'secret sauce' to make their experiments work — and withhold details from publication or describe them only vaguely to retain a competitive edge[5]. What hope is there that other scientists will be able to build on such work to further biomedical progress?

Exacerbating this situation are the policies and attitudes of funding agencies, academic centres and scientific publishers. Funding agencies often uncritically encourage the overvaluation of research published in high-profile journals. Some academic centres also provide incentives for publications in such journals, including promotion and tenure, and in extreme circumstances, cash rewards[6].

Then there is the problem of what is not published. There are few venues for researchers to publish negative data or papers that point out scientific flaws in previously published work. Further compounding the problem is the difficulty of accessing unpublished data — and the failure of funding agencies to establish or enforce policies that insist on data access.

PRECLINICAL PROBLEMS

Reproducibility is potentially a problem in all scientific disciplines. However, human clinical trials seem to be less at risk because they are already governed by various regulations that stipulate rigorous design and independent oversight — including randomization, blinding, power estimates, pre-registration of outcome measures in standardized, public databases such as ClinicalTrials.gov and oversight by institutional review boards and data safety monitoring boards. Furthermore, the clinical trials community has taken important steps towards adopting standard reporting elements[7].

Preclinical research, especially work that uses animal models[1], seems to be the area that is currently most susceptible to reproducibility issues. Many of these failures have simple and practical explanations: different animal strains, different lab environments or subtle changes in protocol. Some irreproducible reports are probably the result of coincidental findings that happen to reach statistical significance, coupled with publication bias.

Document 13

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

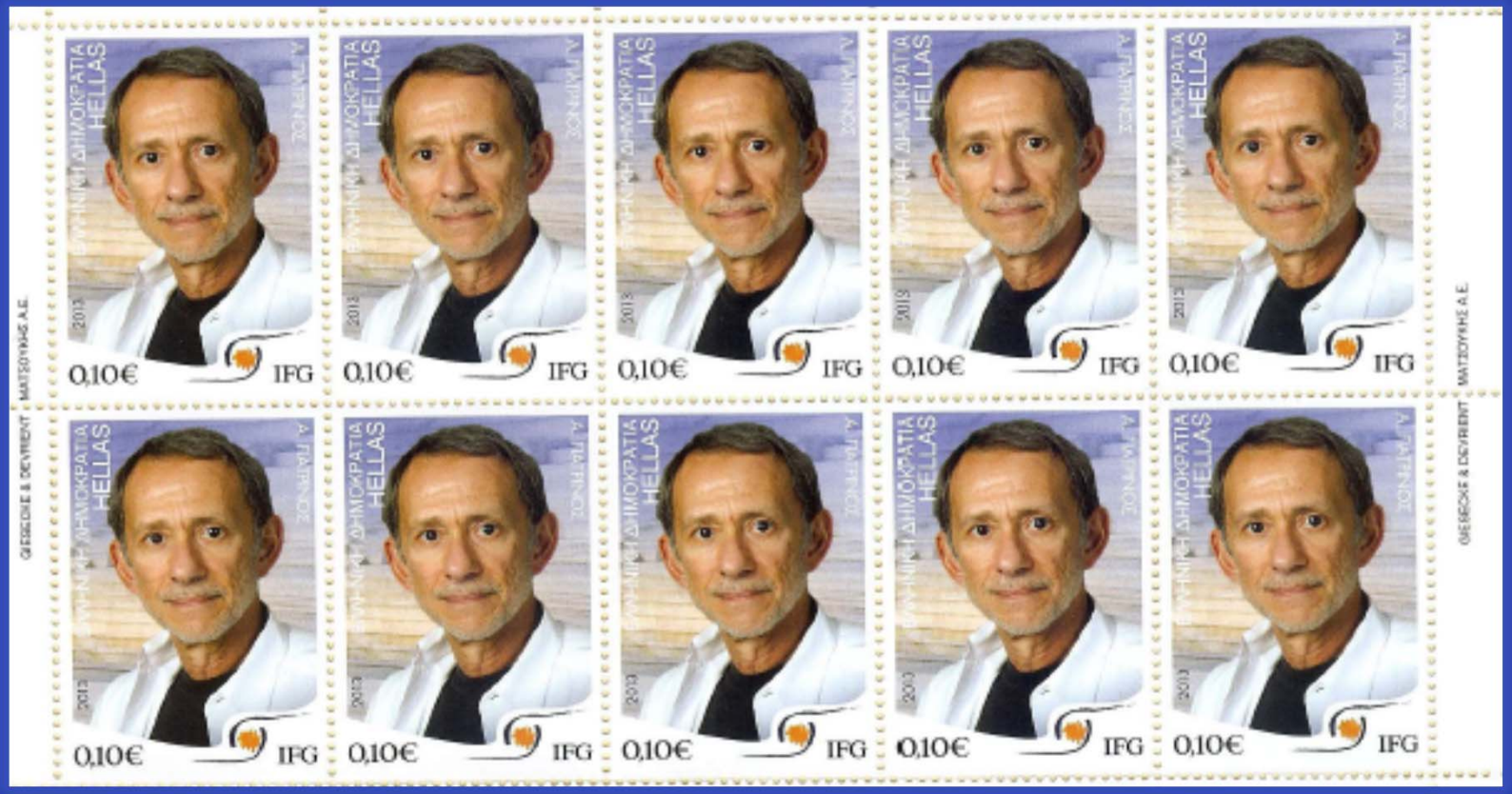
Mourning the Loss of Fred Sanger

Document 14

Mourning the Loss of Janet Rowley

Document 15

The International Foundation for Greece Greek Postage Stamp

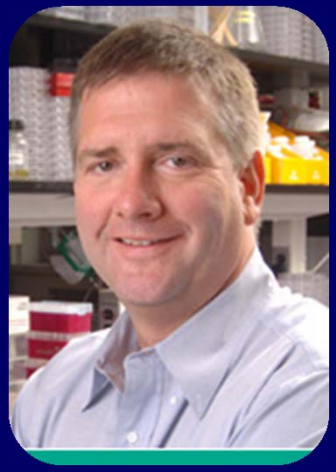


Ari Patrinos, Ph.D.

Document 16

Awards at 2013 ASHG Annual Meeting

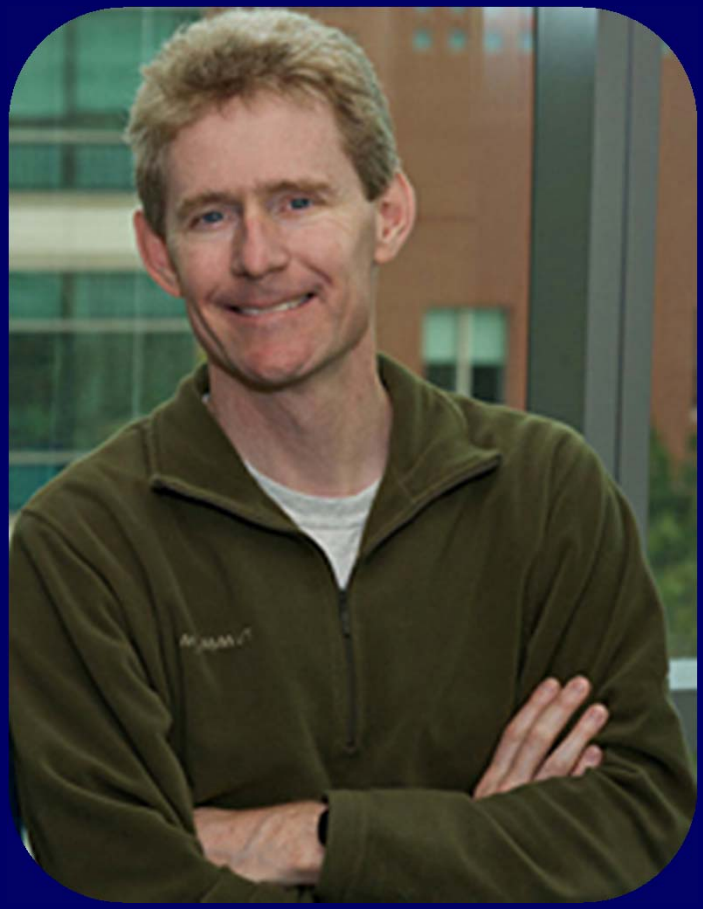
Aravinda Chakravarti, Ph.D.
William Allan Award



John Moran, Ph.D.
Curt Stern Award

Document 17

2014 NAS John J. Carty Award for the Advancement of Science



Joe DeRisi, Ph.D.

Document 18

2014 Breakthrough Prize in Life Sciences

Rick Lifton, M.D., Ph.D.

Document 19

Elected to the Institute of Medicine

David DeMets
Judy Garber
Richard Kolodner
Brendan Lee
Pamela Sklar
Chris Walsh

Document 20

Elected to AAAS

James Broach
Frederic Bushman
Peter Cherbas
Harold Craighead
Job Dekker
David Goldstein
Robert Grossman
Kent Lloyd
Carole Ober
Bing Ren
Shankar Subramaniam

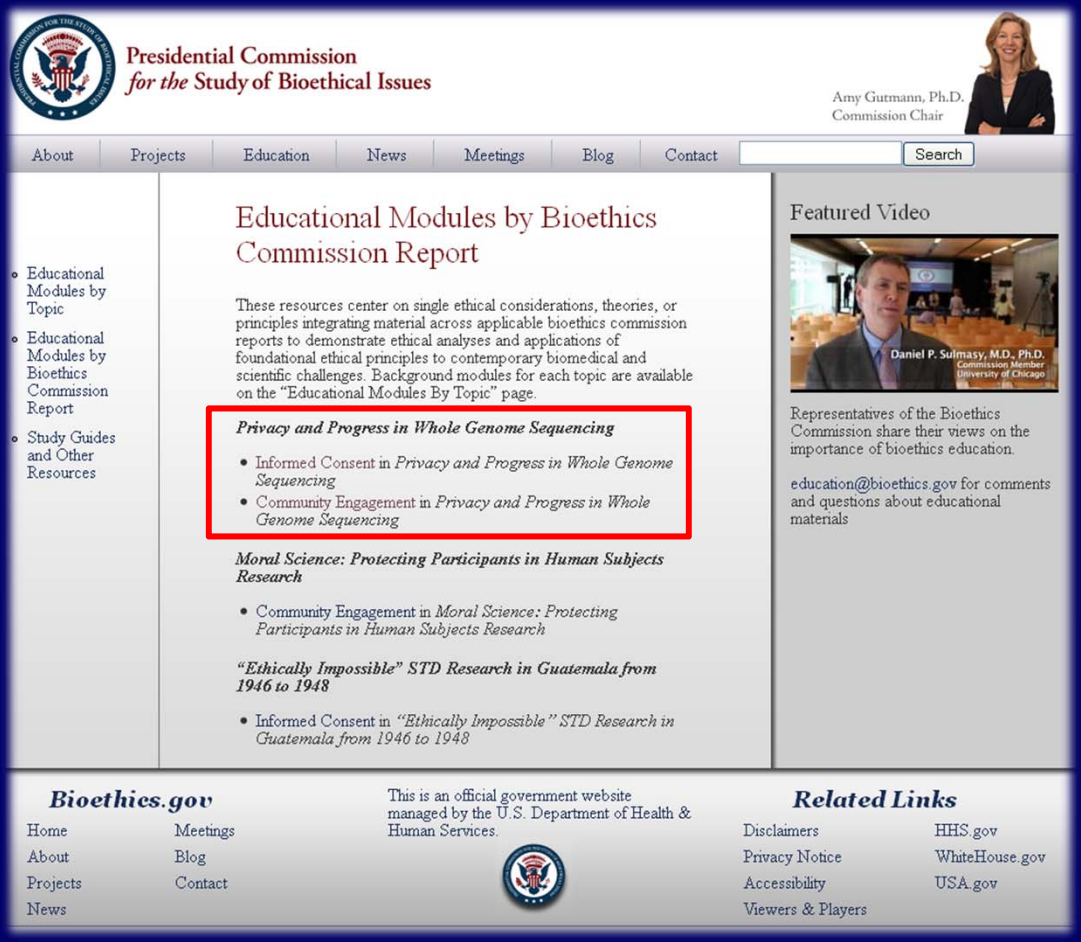
Document 21

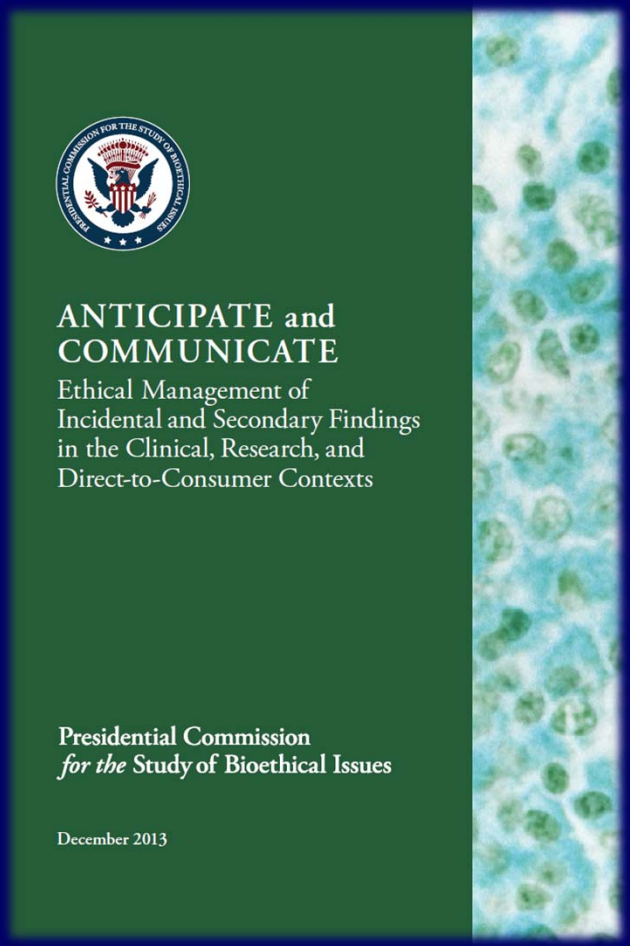
NCHPEG Transitions to The Jackson Laboratory

- The Jackson Laboratory plans to expand healthcare professional continuing education
- Shared opportunity with key former NCHPEG members moving to JAX to create new curricula and programs
- Will temporarily maintain NCHPEG education programs and website at nchpeg.org

Document 22

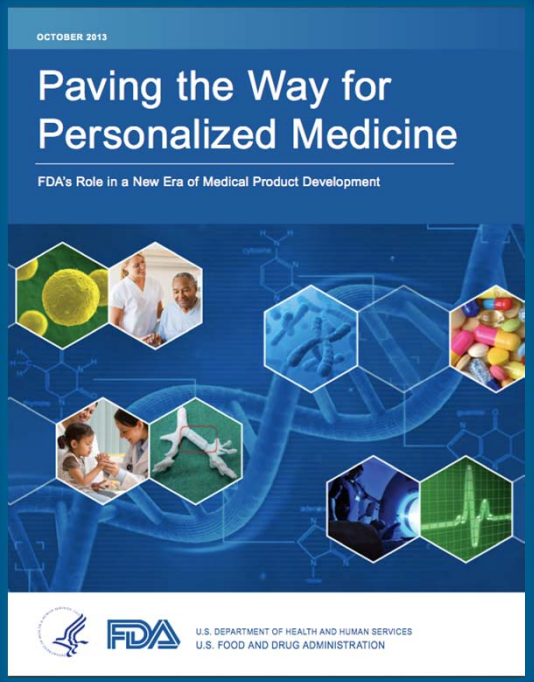
Presidential Commission for the Study of Bioethical Issues





Document 23

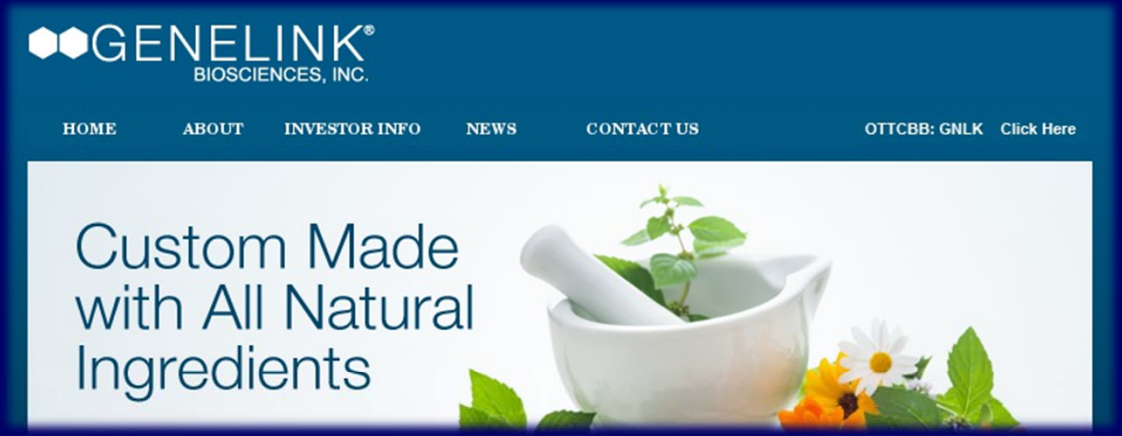
FDA and Genomics



Document 24

FDA and FTC Focus on Genomics

23andMe



Document 25

NHGRI Genome Advance of the Month

The X and Y of human origins: Using Y chromosome sequencing data to explore human evolution

By Shannon Biello
Scientific Program Analyst, NHGRI

To sequence the exome or the genome: that is the question

By Elizabeth Burke, Ph.D.
Intramural Postdoctoral Fellow, NHGRI

Pilot study eyes implications of ACMG stance on incidental findings

By Anh Quynh Nguyen
NHGRI Scientific Program Analyst

Multi-tasking DNA: Dual-use codons in the human genome

By Kris A. Wetterstrand, M.S.
Scientific Liaison to the Director for Extramural Activities, NHGRI

We know that the human genome is the molecular instruction book for building the human body, but exactly what are all the intricacies of how it functions? In 2003, the Human Genome Project (HGP) reached completion, comprehensively sequencing the 3 billion base pairs that make up a full human genome. Yet, having the complete human genome sequence did not mean a complete understanding of what all those As, Cs, Ts and Gs meant in terms of our biology.

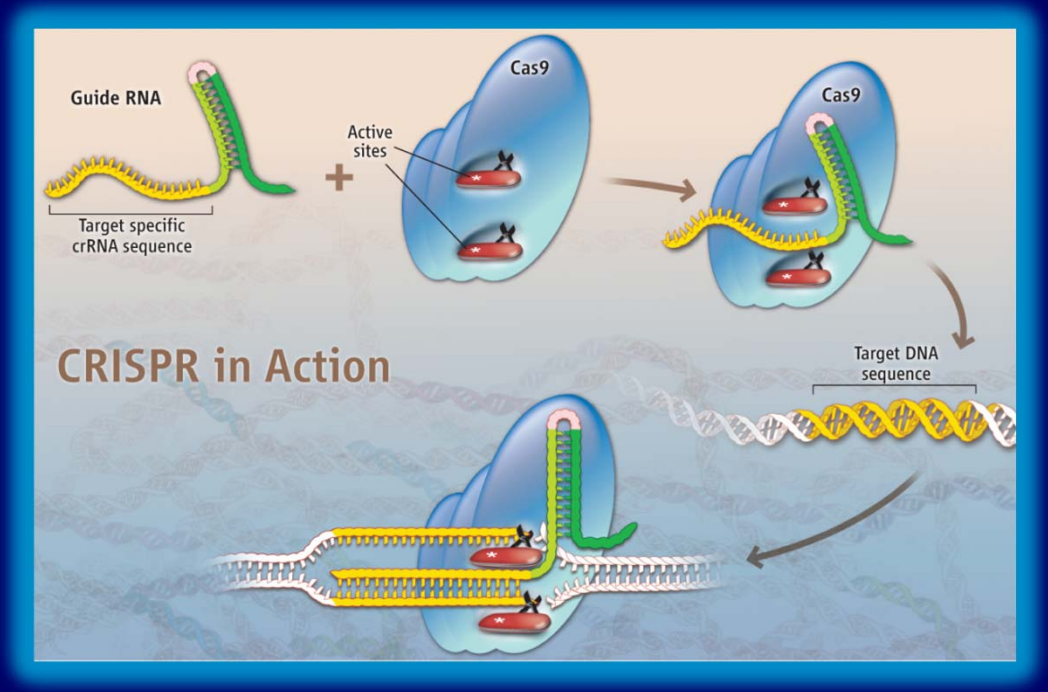
Researchers have been hard at work understanding how our genome works, how tiny differences account for the wide diversity among us, how slightly more differences explain why we're so different from our closest primate relatives, and how differences in our genomes contribute to health and disease. December's Genome Advance of the Month highlights a paper published by Andrew Stergachis, Ph.D., professor of epidemiology at the University of Washington and his colleagues, in the December 13, 2013, issue of Science. So what's all the fuss about?

A surprising finding from the HGP was just how few genes are in the human genome. Estimates were much higher than the eventual answer, which was 21,000 genes. These 21,000 genes produce different proteins at different times to generate myriad cell types (e.g., muscle cells, skin cells, brain cells, etc.) that make a human being. But the 21,000 genes in the human genome make up only one percent of the overall DNA sequence.

Document 26

Science
AAAS

Breakthrough of the Year 2013



Two groups fostering this revolution:

Centers of Excellence in Genomic Science
George Church

Large Scale Sequencing & Analysis Center
Eric Lander

Document 26

People and Things to Watch in 2014

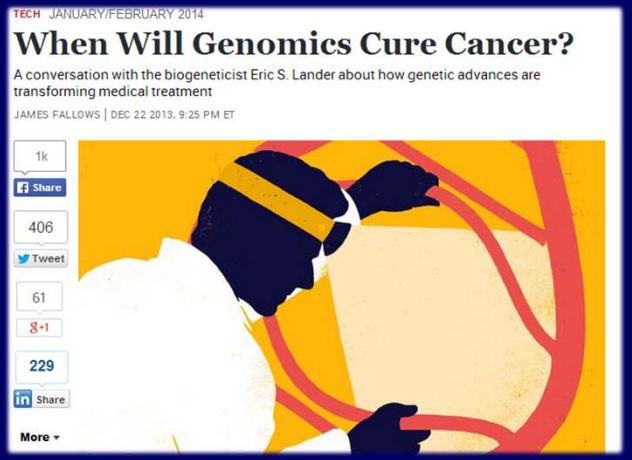
Science ...Areas to Watch in 2014

Clinical Genomics

nature FIVE TO WATCH 2014

Gordon Sanghera
Chief Executive, Oxford Nanopore

Genomics In The News…



Document 29

Genomes In The News…

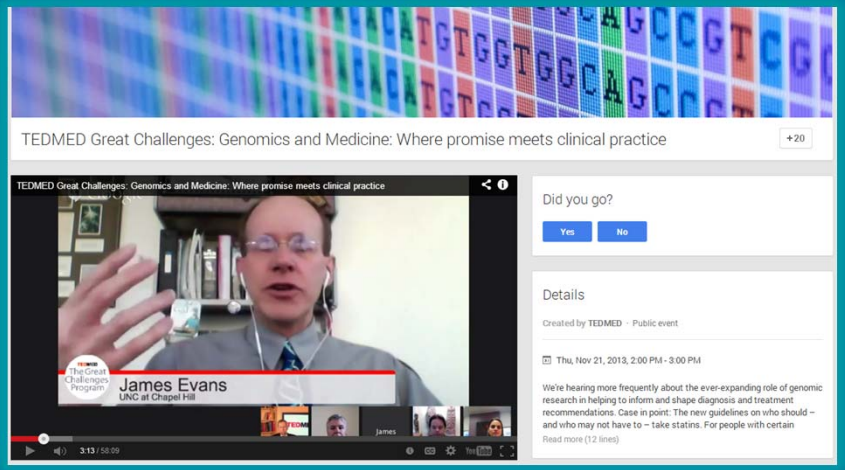
Genomes of the Year

Science

Notable sequences of 2013: The **oldest human mitochondrial DNA**, which comes from a 400,000-year-old Neandertal ancestor found in Spain but mysteriously resembles that of a different extinct human • The oldest organismal genome, from **a 700,000-year-old frozen horse hoof** • Other complete genomes came from the **comb jelly**, changing views of the animal tree of life • **Minke whale**, revealing how marine mammals cope with deep dives • ***Amborella*, sister to all flowering plants**, explaining the early days of angiosperms • Tiger, lion, and snow leopard, capturing **the genomic essence of big cats** • The **scorpion** *Mesobuthus martensii* (at right), which has 10,000 more genes than humans do • **Norway spruce** (*Picea abies*), **white spruce** (*Picea glauca*), and, soon, **loblolly pine** (*Pinus taeda*), each with genomes about seven times the size of a human's—a sequencing tour de force • The invaluable **HeLa cancer research cell line**, requiring permission from the family of Henrietta Lacks • **King cobra** and **Burmese python**, telling an evolutionary tale of extreme adaptations • **Four bats** which, when compared to dolphins, highlight a common core of echolocation genes • **Pigeon**, revealing the gene for crests • **Irish famine potato blight**, showing that this historic strain is extinct.

Document 30

Genomics In Video…



Document 31

Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

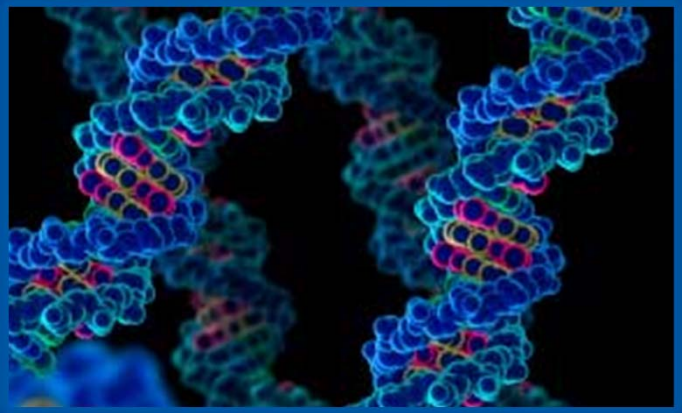
IV. NHGRI Extramural Research Program

V. NIH Common Fund/Trans-NIH

VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

Large-Scale Genome Sequencing and Analysis Centers



- 32 new papers in the most recent quarter
- Alzheimer's Disease Sequencing Project:

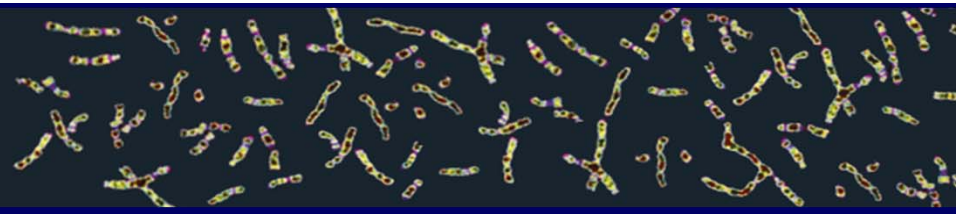
  Initial data freeze and release in December 2013

  Whole-genome sequence data from 410 individuals (89 families)

Document 32

1000 Genomes

A Deep Catalog of Human Genetic Variation

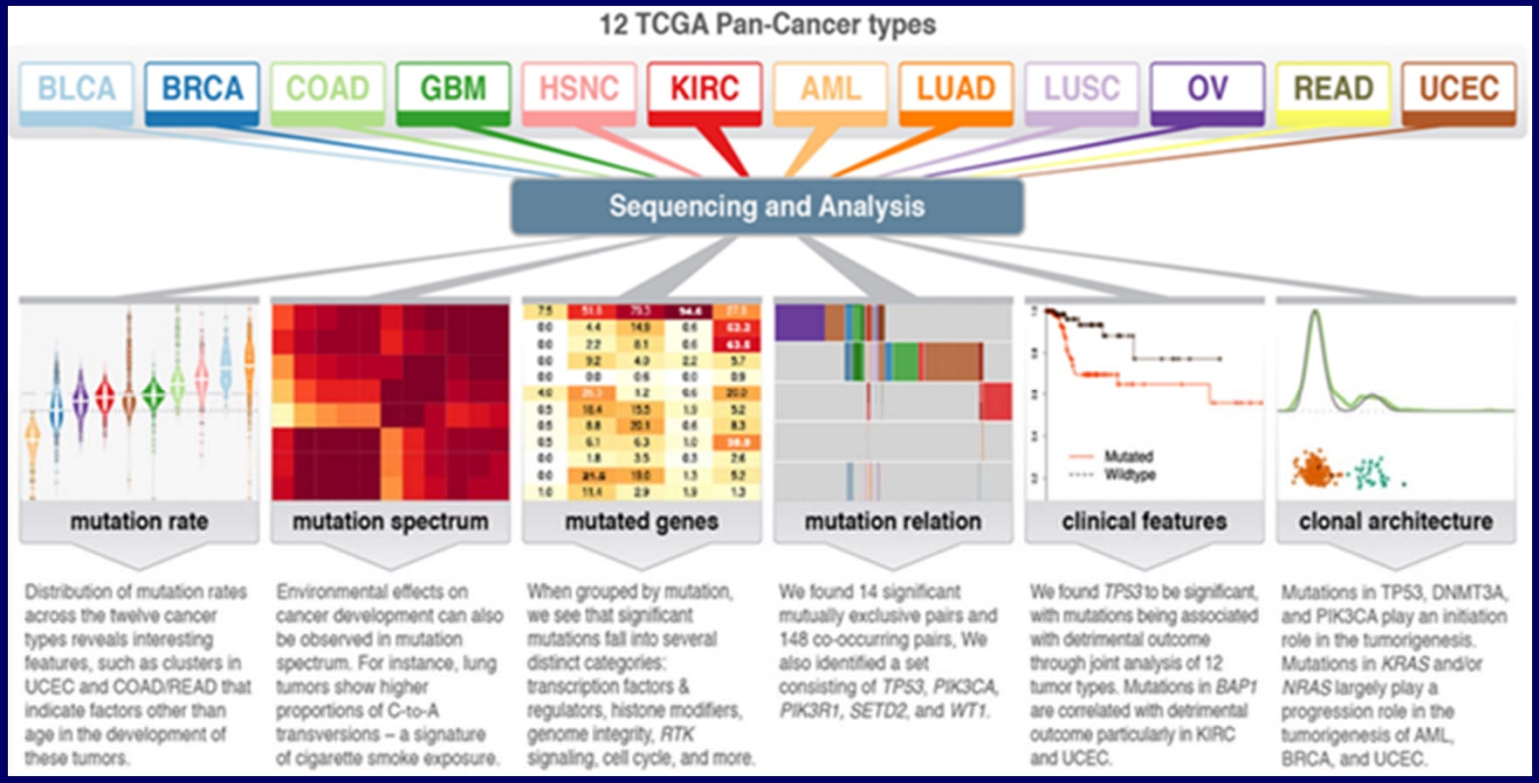


- **Currently ~90 million variants in 2535 samples**
  - **~76 million SNPs**
  - **~7 million indels**
  - **~7 million structural and other complex variants**
- **Final release of variants by Fall 2014**
- **Final 1000 Genomes Project Meeting and Community Meeting: June 24-26 in the U.K.**

- **Pan-cancer analysis**

  ***Nature Genetics* feature: 27 papers and 5 thematic threads**

  ICGC/TCGA whole-genome pan-cancer analysis project



- **Sample accrual is completed**

Centers for Mendelian Genomics

Finding the genes underlying human Mendelian conditions

- **>10,000 exome sequences, >862 diseases**
- **>467 disease genes (202 novel) for >273 diseases**
- **>70 publications: disease-gene discoveries, methods, resources, and practices of data sharing**
- **Disease-gene discovery publications**

  Novel disease genes/pathways and disease biology
  Phenotypic expansion
  Levels of penetrance
  Heterogeneity
  Other

Document 35

Centers for Mendelian Genomics

AGTC

Finding the genes underlying human Mendelian conditions

- **CMG Network:**
  >414 investigators, 200 institutions, 33 countries
- **Coordination: public disorder list, GeneMatcher**
- **IRDiRC**

  Develop diagnostic tests for most rare disorders

  New treatments for 200 rare disorders by 2020
- **NHGRI participation in IRDiRC**

  Executive Committee

  Working groups on phenotypic data collection, sequencing standards, and disease prioritization

IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

Document 35

Clinical Sequencing Exploratory Research (CSER) Program

Processes and preliminary outputs for identification of actionable genes as incidental findings in genomic sequence data in the Clinical Sequencing Exploratory Research Consortium

Jonathan S. Berg, MD, PhD[1–4], Laura M. Amendola, MS[5], Christine Eng, MD[6], Eliezer Van Allen, MD[7–9], Stacy W. Gray, MD, AM[8,10,11], Nikhil Wagle, MD[8,11,12], Heidi L. Rehm, PhD[10,13,14], Elizabeth T. DeChene, MS[15,16], Matthew C. Dulik, PhD[15,16], Fuki M. Hisama, MD[5], Wylie Burke, MD, PhD[5,17], Nancy B. Spinner, PhD[15], Levi Garraway, MD, PhD[7,12,18], Robert C. Green, MD, MPH[12,19], Sharon Plon, MD, PhD[6,20], James P. Evans, MD, PhD[1–4] and Gail P. Jarvik, MD, PhD[5,21] and the members of the CSER Actionability and Return of Results Working Group

A survey of informatics approaches to whole-exome and whole-genome clinical reporting in the electronic health record

Peter Tarczy-Hornoch, MD[1,2], Laura Amendola, MS[1,2], Samuel J. Aronson, MA, ALM[1,3], Levi Garraway, MD, PhD[1,4,5], Stacy Gray, MD, AM[4–6], Robert W. Grundmeier, MD[1,5,7], Lucia A. Hindorff, PhD, MPH[1,8], Gail Jarvik, MD, PhD[2], Dean Karavite, MS[1,7], Matthew Lebo, PhD[3,5,9], Sharon E. Plon, MD, PhD[1,10], Eliezer Van Allen, MD[1,4,6], Karen E. Weck, MD[1,11], Peter S. White, PhD[1,5,7] and Yaping Yang, PhD[10]

Recommendations for returning genomic incidental findings? We need to talk!

Wylie Burke, MD, PhD[1], Armand H. Matheny Antommaria, MD, PhD[2], Robin Bennett, MS, CGC[3], Jeffrey Botkin, MD, MPH[4], Ellen Wright Clayton, MD, JD[5], Gail E. Henderson, PhD[6], Ingrid A. Holm, MD, MPH[7–9], Gail P. Jarvik, MD, PhD[3], Muin J. Khoury, MD, PhD[10], Bartha Maria Knoppers, JD, PhD[11], Nancy A. Press, PhD[12], Lainie Friedman Ross, MD, PhD[13], Mark A. Rothstein, JD[14], Howard Saal, MD[15], Wendy R. Uhlmann, MS, CGC[16], Benjamin Wilfond, MD[17], Susan M. Wolf, JD[18] and Ron Zimmern, FRCP, FFPHM[19]

Actionable, Pathogenic Incidental Findings in 1,000 Participants' Exomes

Michael O. Dorschner,[1,4,5] Laura M. Amendola,[2] Emily H. Turner,[1,5] Peggy D. Robertson,[1] Brian H. Shirts,[5] Carlos J. Gallego,[2] Robin L. Bennett,[2] Kelly L. Jones,[2] Mari J. Tokita,[2] James T. Bennett,[2,3] Jerry H. Kim,[6] Elisabeth A. Rosenthal,[2] Daniel S. Kim,[1] National Heart, Lung, and Blood Institute Grand Opportunity Exome Sequencing Project, Holly K. Tabor,[5,6] Michael J. Bamshad,[1,3] Arno G. Motulsky,[1,2] C. Ronald Scott,[2,3] Colin C. Pritchard,[5] Tom Walsh,[2] Wylie Burke,[2,6] Wendy H. Raskind,[2,4] Peter Byers,[2,7] Fuki M. Hisama,[2] Deborah A. Nickerson,[1] and Gail P. Jarvik[1,2,*]

Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders

Yaping Yang, Ph.D., Donna M. Muzny, M.Sc., Jeffrey G. Reid, Ph.D., Matthew N. Bainbridge, Ph.D., Alecia Willis, Ph.D., Patricia A. Ward, M.S., Alicia Braxton, M.S., Joke Beuten, Ph.D., Fan Xia, Ph.D., Zhiyv Niu, Ph.D., Matthew Hardison, Ph.D., Richard Person, Ph.D., Mir Reza Bekheirnia, M.D., Magalie S. Leduc, Ph.D., Amelia Kirby, M.D., Peter Pham, M.Sc., Jennifer Scull, Ph.D., Min Wang, Ph.D., Yan Ding, M.D., Sharon E. Plon, M.D., Ph.D., James R. Lupski, M.D., Ph.D., Arthur L. Beaudet, M.D., Richard A. Gibbs, Ph.D., and Christine M. Eng, M.D.

Activating *ESR1* mutations in hormone-resistant metastatic breast cancer

Dan R Robinson[1,2,12], Yi-Mi Wu[1,2,12], Pankaj Vats[1,2], Fengyun Su[1,2], Robert J Lonigro[1,3], Xuhong Cao[1,4], Shanker Kalyana-Sundaram[1,2], Rui Wang[1,2], Yu Ning[1,2], Lynda Hodges[1], Amy Gursky[1,2], Javed Siddiqui[1,2], Scott A Tomlins[1,2], Sameek Roychowdhury[5], Kenneth J Pienta[6], Scott Y Kim[7], J Scott Roberts[8], James M Rae[3,9], Catherine H Van Poznak[9], Daniel F Hayes[9], Rashmi Chugh[9], Lakshmi P Kunju[1,2], Moshe Talpaz[9], Anne F Schott[9] & Arul M Chinnaiyan[1–4,10,11]

- **Collaborations with other NHGRI consortia:**
  eMERGE, CMG, ClinGen

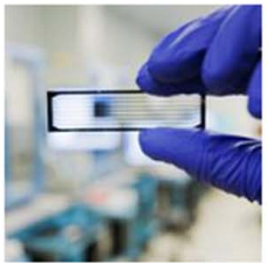
Clinical Sequencing Exploratory Research (CSER) Program

cser
Clinical Sequencing Exploratory Research

PROJECTS
PUBLICATIONS
RESOURCES
IMPACT

Log in

Moving the genome into the clinic

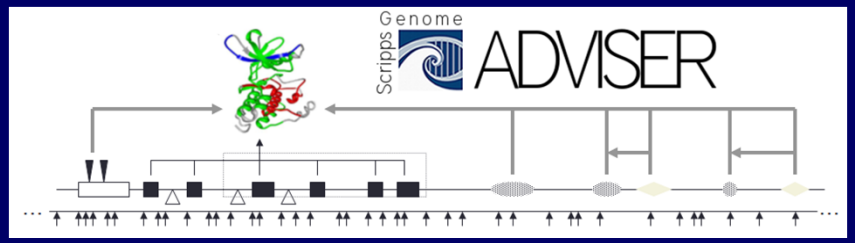


In the past, standard medical practice for genetic testing involved looking at one gene at a time. With new advances in our understanding of the genomic basis of health and disease and in technology, it is now possible to test all of our genes at once using tests called whole exome or whole genome sequencing. Medical uses of genome sequencing are being applied and adapted on a case-by-case basis, but research to study the optimal uses and implementation of these tests is needed.

To rapidly address these questions, the National Human Genome Research Institute (NHGRI) and the National Cancer Institute (NCI) have initiated a Clinical Sequencing Exploratory Research (CSER) program to support multidimensional research in this area. CSER is a national consortium of projects that bring together clinicians, scientists, laboratories, bioinformaticians, economists, legal scholars, ethicists, and patients working together to develop and share innovations and best practices in the integration of genomic sequencing into clinical care.

Document 36

Genome Sequencing Informatics Tools

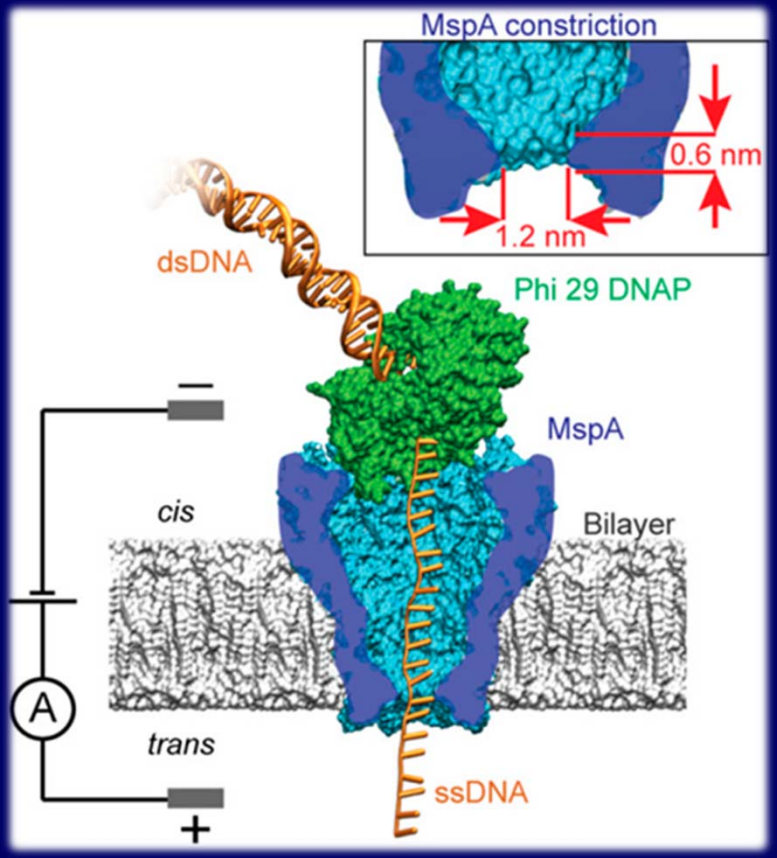


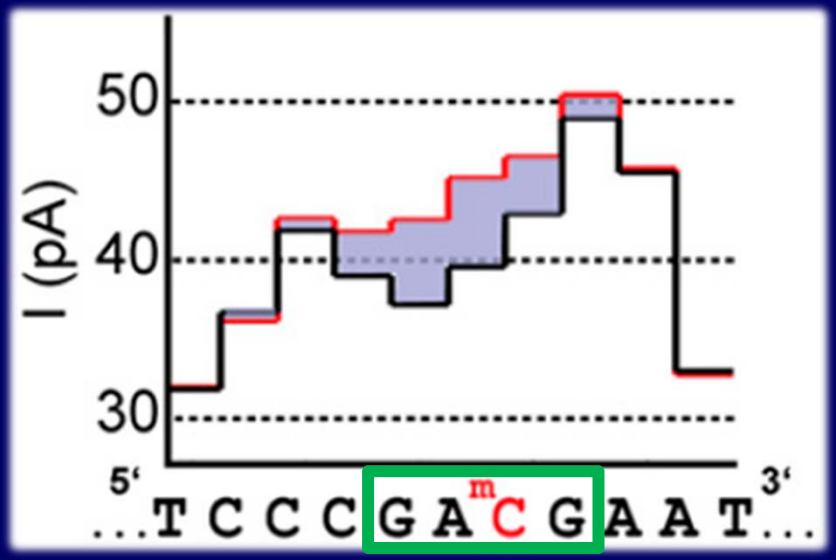
- GS-IT has released SG ADVISER to annotate and filter genomic variants
- Used to solve clinical case of 15-year-old girl
- iSeqTools Portal: “The search for what’s wrong with Lilly”

iSeqTools

DNA Sequencing Technology Development

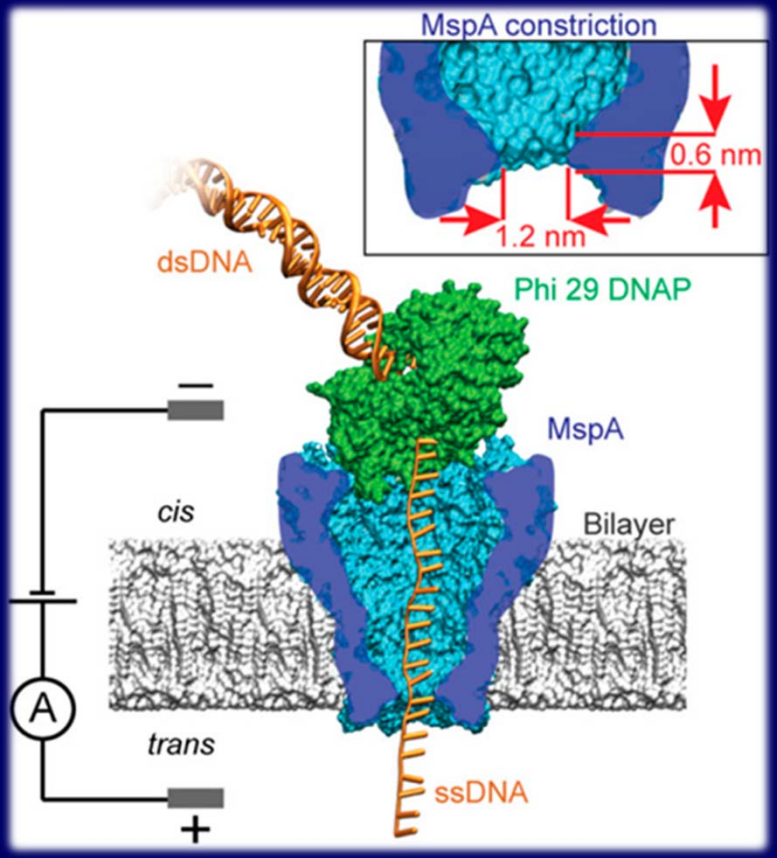
- Sequencing of epigenetic modifications using biological nanopores
- Developing error models for nanopore sequencing

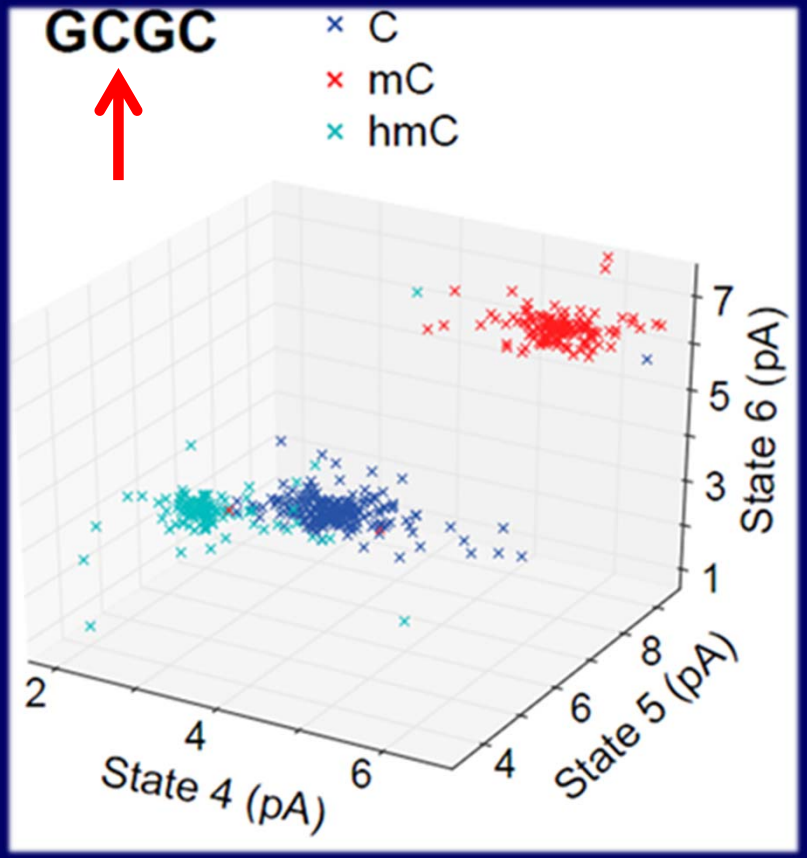




DNA Sequencing Technology Development

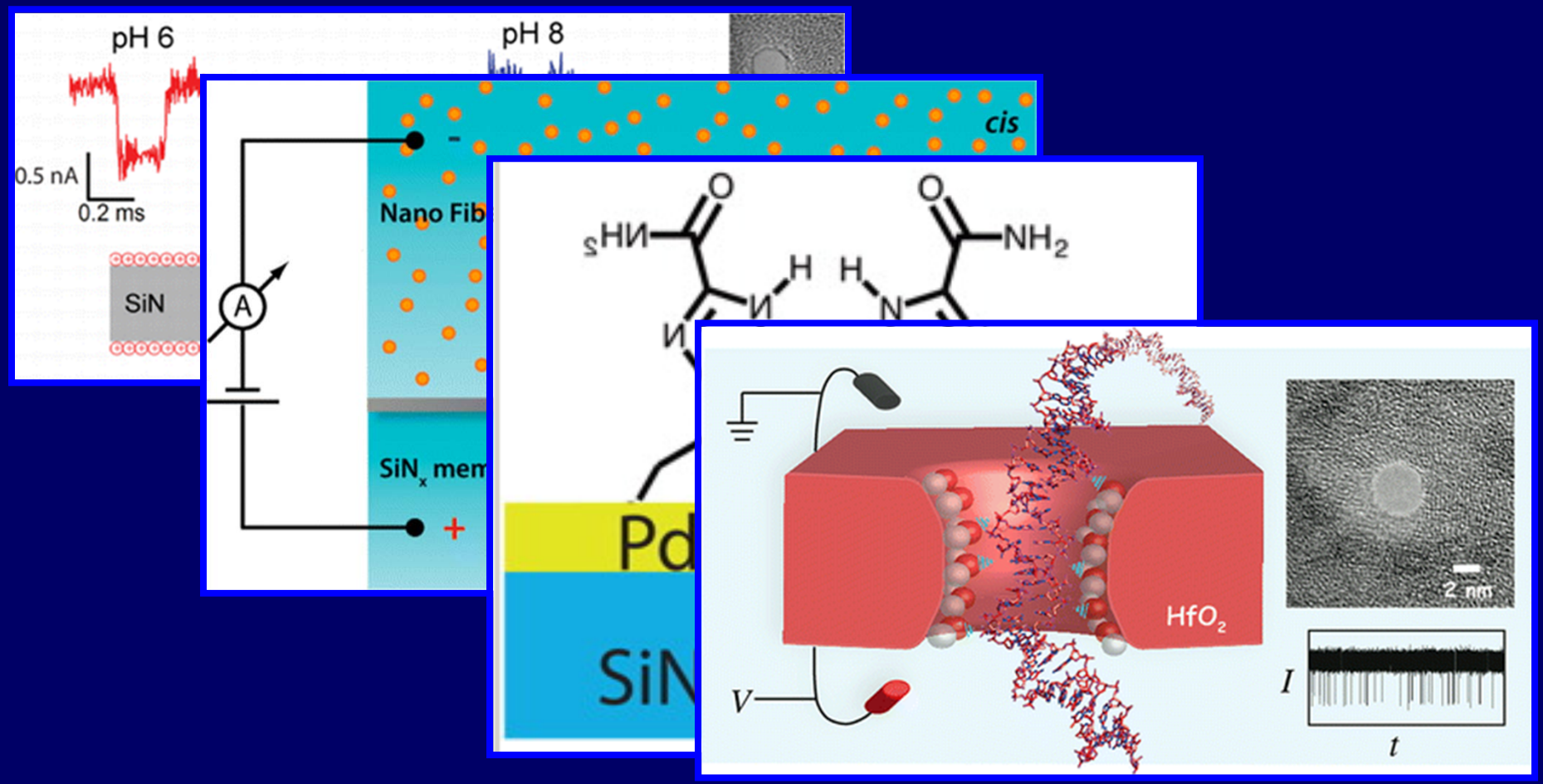
- Sequencing of epigenetic modifications using biological nanopores
- Developing error models for nanopore sequencing





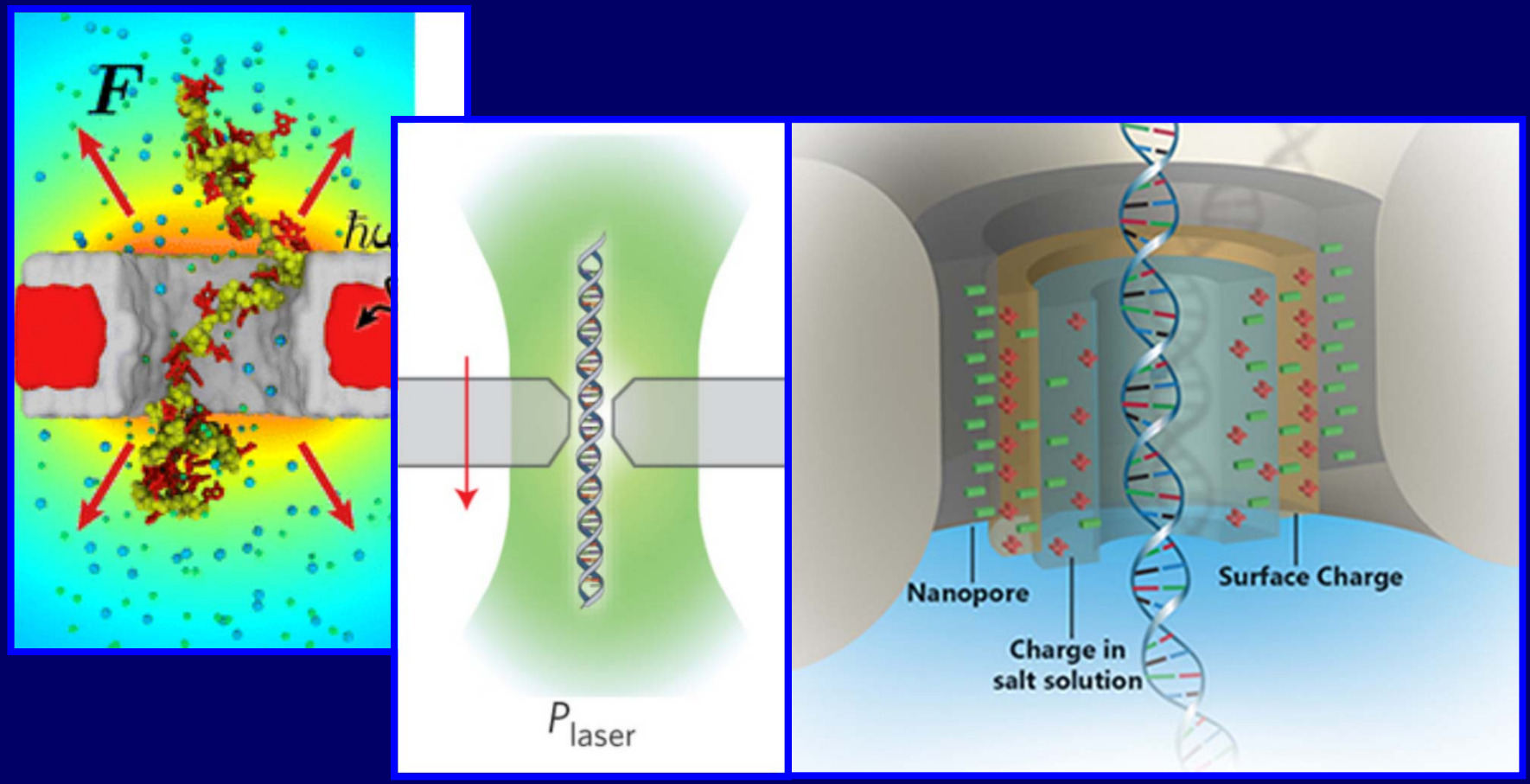
DNA Sequencing Technology Development

- Need to control DNA translocation rate in solid-state nanopores

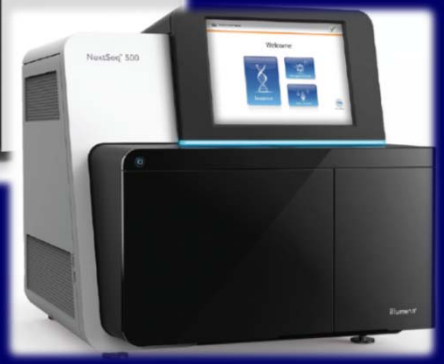


DNA Sequencing Technology Development

- Need to control DNA translocation rate in solid-state nanopores



DNA Sequencing Technology Development



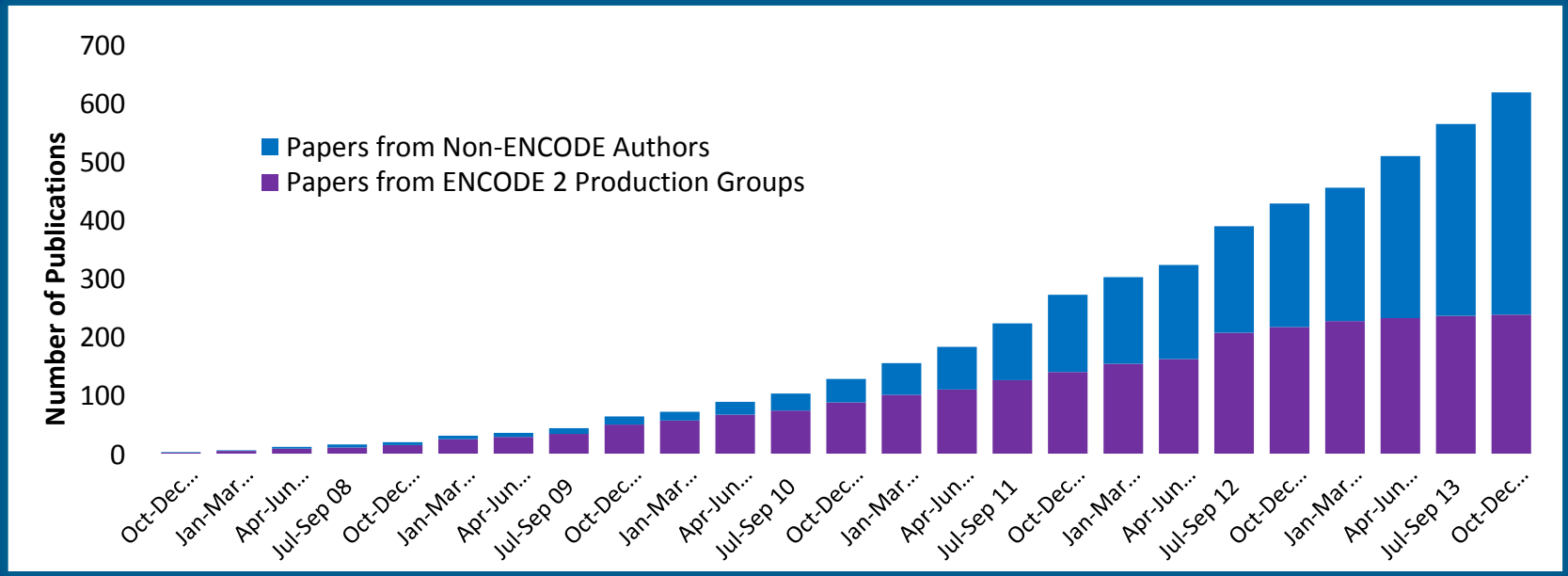
华大基因 BGI

Complete genomics
A BGI Company

Document 38

ENCODE

- **ENCODE Outreach Activities:**
  - **ASHG Tutorial**
  - **Tutorial at CHARGE Consortium Meeting**
- **Publications using ENCODE Data**



Centers of Excellence in Genomic Science (CEGS) Program

- CEGS investigators met in October
- CEGS renewed at Dana Farber Cancer Institute

- New CEGS applications are under consideration

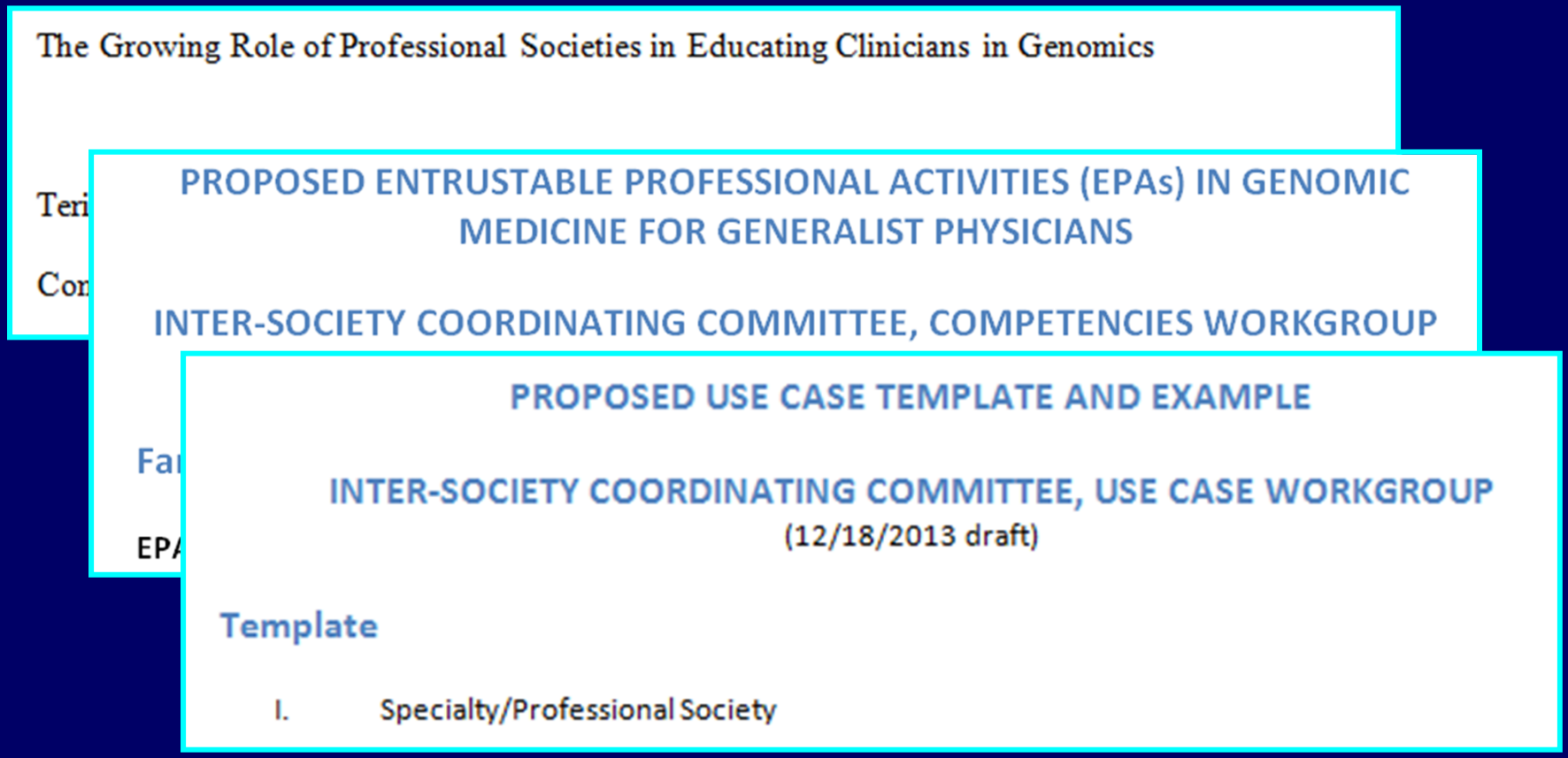
Global Leaders in Genomic Medicine Meeting

- 6th Genomic Medicine Meeting
- Focused on international projects and potential collaborations
- Several fertile areas identified
- Meeting summary to be presented by Teri Manolio

Document 41

Inter-Society Coordinating Committee for Practitioner Education in Genomics

- Met in person in September


The Growing Role of Professional Societies in Educating Clinicians in Genomics

PROPOSED ENTRUSTABLE PROFESSIONAL ACTIVITIES (EPAs) IN GENOMIC MEDICINE FOR GENERALIST PHYSICIANS

INTER-SOCIETY COORDINATING COMMITTEE, COMPETENCIES WORKGROUP

PROPOSED USE CASE TEMPLATE AND EXAMPLE

INTER-SOCIETY COORDINATING COMMITTEE, USE CASE WORKGROUP

(12/18/2013 draft)

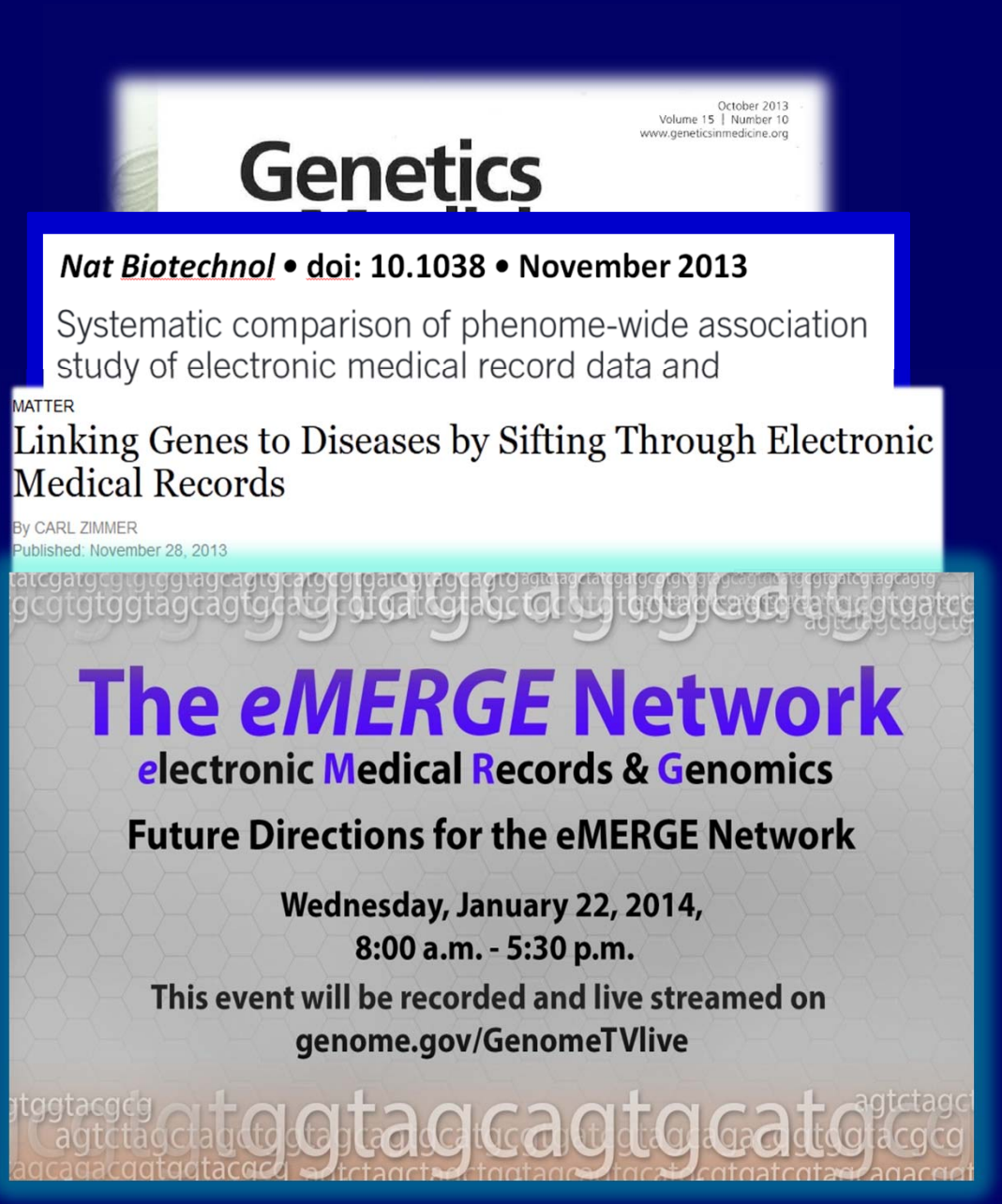
Template

I. Specialty/Professional Society

- Next meeting April 2014

Document 42

eMERGE Network

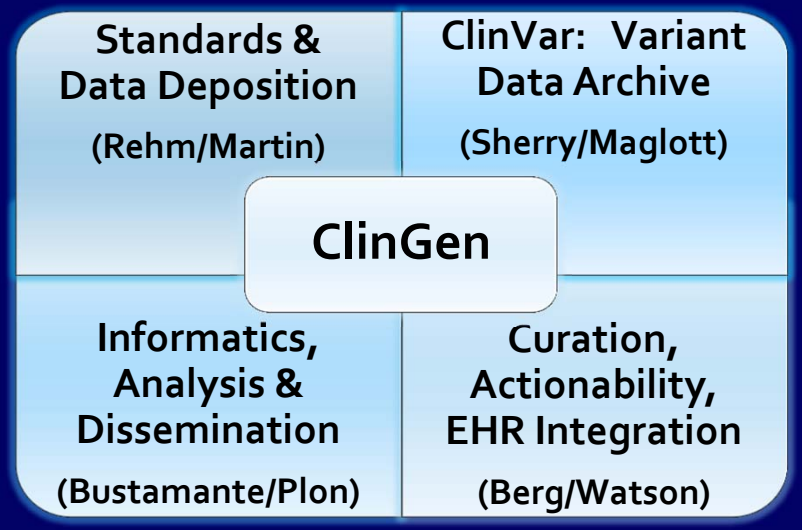


- *Genetics in Medicine* Special Issue
- Methods development and discovery
- eMERGE in the news
- eMERGE workshop: Future of eMERGE

Document 43

Clinical Genome (ClinGen) Resource

- Goal: Create a centralized repository and inter-connected resources of clinically relevant variants
- Standardize clinical assessment of variants & deposition into ClinVar
- Develop a consensus approach to identify clinically relevant variants
- Facilitate use of genomic information in clinical care and research



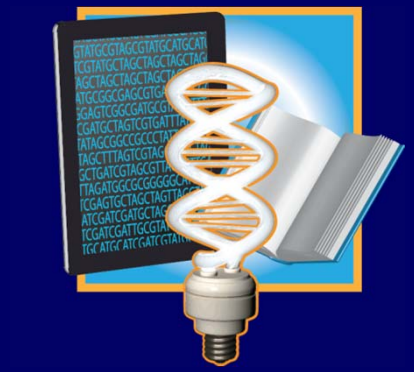
Document 44

Genomics and Society Working Group

- 2nd in-person meeting: November 2013
- Topics discussed included:
  - Priority Setting
  - Funding Mechanisms
  - Boundaries of ELSI Research
- Next in-person meeting: April 2014

Document 45

New NHGRI Training Notices



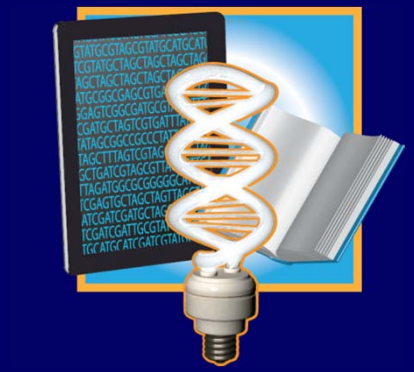
- **T32 Programs Focused on Genomic Science (NOT-HG-14-016)**

- **T32 Postdoctoral Training Programs Focused on Genomic Medicine (NOT-HG-14-017)**

New

Document 46

New NHGRI Training Notices



- **Research Scientist** Development K01 Award (NOT-HG-14-018)

- **Clinical Investigator** K08 Award (NOT-HG-14-019)

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

Human Microbiome Project (HMP)

- Phase 2: Fiscal Year 2013 to 2015
- Create integrated multi-'omics dataset of microbiome and host properties
- A Common Fund initiative with co-funding from other ICs, $22.1M total

  (NIDDK, NCCAM, ORWH, ODS)
- HMP2 Consortium:

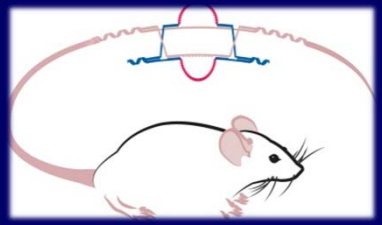
  Preterm birth and microbiome: Buck

  IBD and microbiome: Xavier/Huttenhower

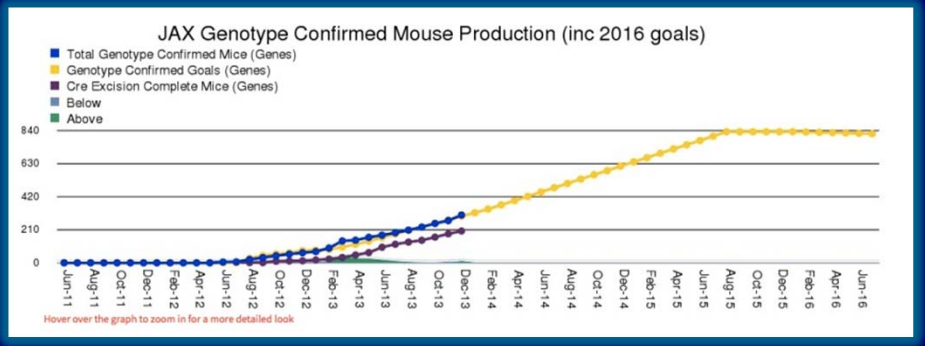
  Diabetes and microbiome: Snyder/Weinstock

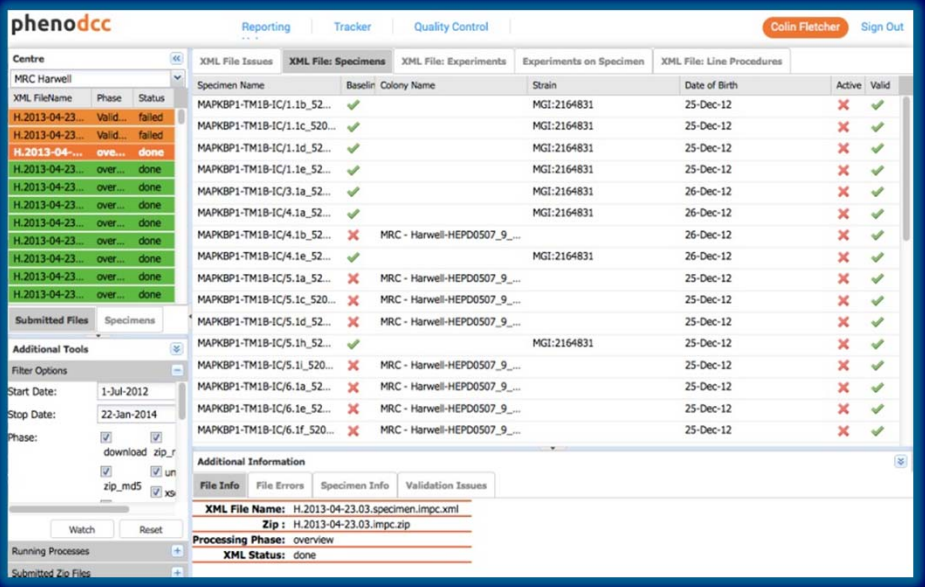
Document 47

Knockout Mouse Phenotyping Project (KOMP2)



- Halfway into the project and production is on track
- Major focus on phenotype data upload and quality control; roll out of PhenoDCC web interface
- IMPC launched pilot to evaluate the application of CRISPR technology



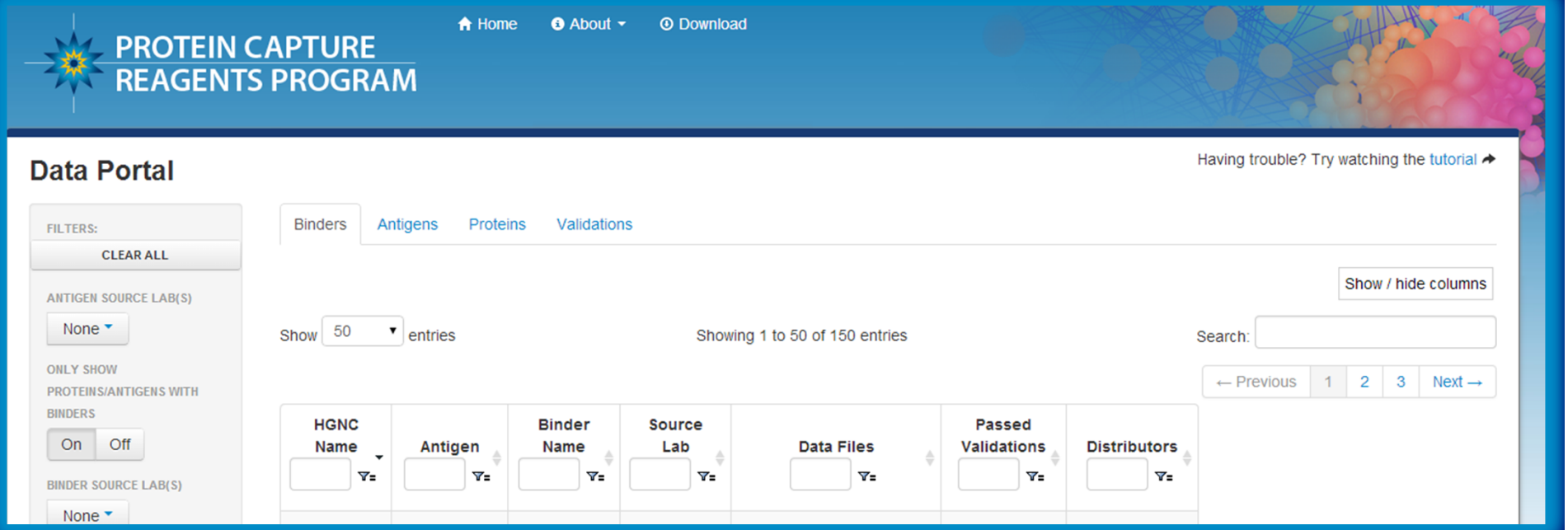


GTEx
Genotype-Tissue Expression Project

- **Scale-up phase underway**
  - **600/900 donors enrolled**
  - **10K/25K RNA-Seq studies**
- **Seven "Enhancing GTEx" U01 awards made**
- **Biospecimen Access Policy live**
- **2nd GTEx Community Scientific Meeting in June 2014 in Boston**

PROTEIN CAPTURE REAGENTS PROGRAM

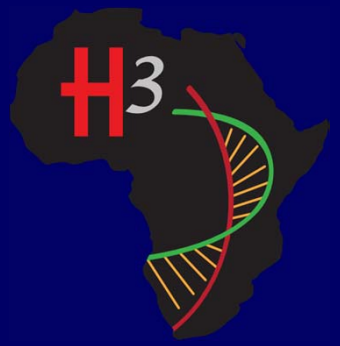
- **Protein Capture Data Portal went live in October**



- **3rd Annual Consortium Meeting in December**

- Applications for Data and Signature Generation Centers (RM13-013) received; review in April
- BD2K-LINCS Data Integration and Coordination Center RFA (HG-14-001) released; due in March
- LINCS Symposium held at Broad Institute in November

H3Africa Initiative



- 3rd Consortium Meeting held in Johannesburg in October (without NIH staff)
- Policy documents in final stages of approval
- H3Africa marker paper submitted
- Many grantees have received ethics approval
- Sample collection underway
- 4th Consortium Meeting in Uganda in late May

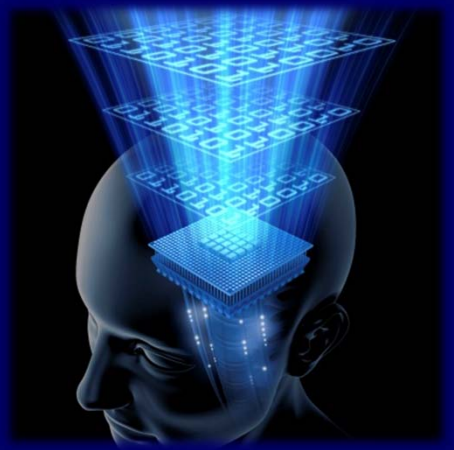
Document 52

Undiagnosed Diseases Network (UDN)

- Harvard Medical School awarded the UDN Coordinating Center (RFA-RM-12-020)

- Clinical Site (RFA-RM-13-004) applications have been received and are under review

Big Data to Knowledge (BD2K)



- Several FOAs issued following:
  - RFIs – data catalog, training, software needs
  - Workshops – training, data catalog, frameworks for standards, research use of clinical data
- Three workshops planned in Fiscal Year 2014

Big Data to Knowledge (BD2K)

Investigator-Initiated BD2K Centers of Excellence

- First-round applications submitted November 2013
- April 2014 review; May 2014 Council
- Awards September 2014
- Second-round applications in Fiscal Year 2015

BD2K-LINCS-Perturbation Data Coordination and Integration Center

- Released December 2013; due in March 2014
- Co-funded by BD2K and NIH Common Fund
- Awards late Fiscal Year 2014

Big Data to Knowledge (BD2K)

Data Discovery Index

- RFA "Development of an NIH BD2K Data Discovery Index Coordination Consortium (U24)"
- RFA issued in December 2013
- Applications due in early March 2014
- Awards in late Fiscal Year 2014
- Co-funded by BD2K and NIH Common Fund

Big Data to Knowledge (BD2K)

Training

- **Three training FOAs issued:**
  - **K01 for mentored career development awards**
  - **R25 for short courses for skills development**
  - **R25 for open educational resources**
  - **Applications due in early April**
  - **Awards in Fiscal Year 2014**
- **Three other FOAs directed at long-term training opportunities; Fiscal Year 2015 funding**

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

2014 USA Science and Engineering Festival (USASEF)

- April 25-27 at the Washington Convention Center
- 250,000 attendees in 2012
- Expect to exceed this number in 2014
- >300 new organizations participating in 2014
- NHGRI will have a booth

Document 55

Genomics in Medicine Lecture Series

Series culminates in 2014 with a focus on genomics and neurology & psychiatry

Maximilian Muenke, M.D.

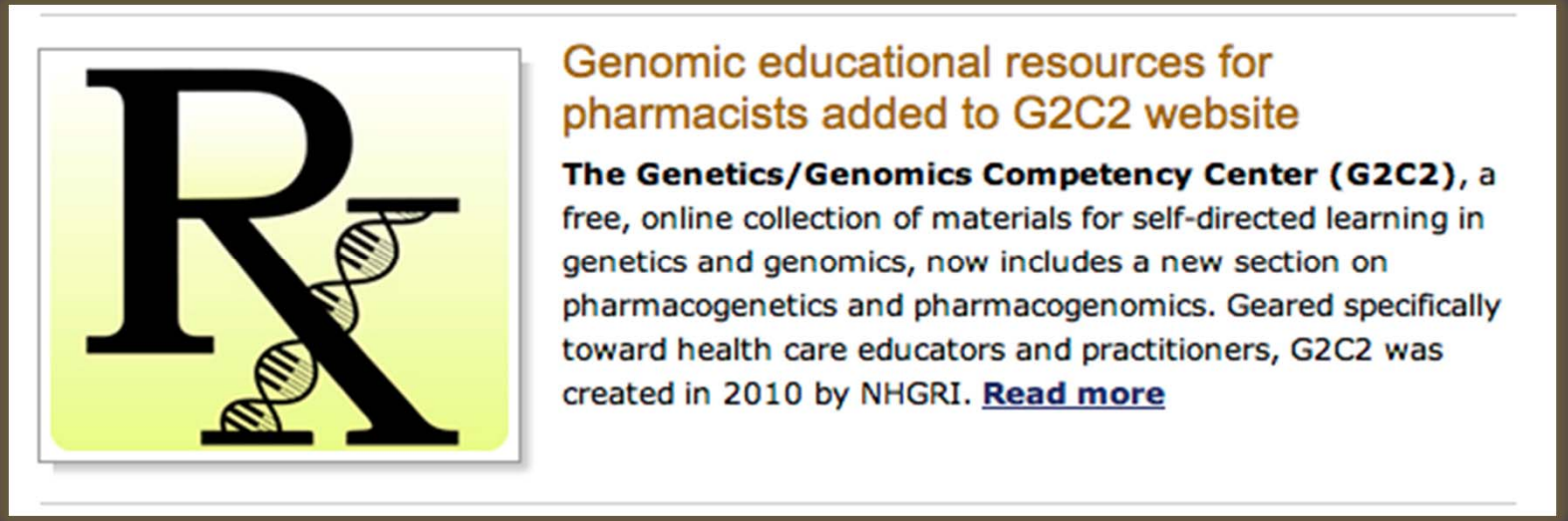
Kenneth H. Fischbeck, M.D.

Francis J. McMahon, M.D.

Kathleen R. Merikangas, Ph.D.

Document 56

Pharmacist Resources on G2C2



Thanks To:

Grace M. Kuo, PharmD, MPH, PhD, FCCP
Mary W. Roederer, Pharm.D., BCPS
James M. Hoffman, PharmD, MS, BCPS
Reginald F. Frye, Pharm.D., Ph.D., FCCP

ASHG-NHGRI Genetics & Public Policy and Genetics & Education Fellowships

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

Google™ Search SEARCH

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

Home › About › Organization › Office of the Director › News Features from the Office of the Director › Apply for NHGRI-ASHG's new education fellowship for genetics professionals

Share Print

Apply for NHGRI-ASHG's new education fellowship for genetics professionals

By John Daniels
Assistant Public Affairs Specialist

See Also:
The Genetics and Education Fellowship [ashp.org]
The Genetics and Public Polidy Fellowship

National Human Genome Research Institute
The American Society of Human Genetics

Realizing the benefits of genomics will require an educated public who can understand the implications of genomics for their health care and evaluate the relevant public policy issues, according to the National Human Genome Research Institute's (NHGRI) 2011 strategic plan.

To help cultivate an educated citizenry, the American Society of Human Genetics (ASHG) and NHGRI have teamed up to sponsor the new Genetics and Education Fellowship. Every year, one genetics professional will receive comprehensive training and experience to help prepare him or her for a career in genetics and genomics education.

The 16-month program is divided into three parts:

- From September 2014 to December 2014, the fellow will work in the Education and Community Involvement Branch, within NHGRI's Division of Policy, Communications and Education. At meetings and events, the fellow will help educate community groups on genomics and genetics and develop programs with a similar goal.
- From January 2015 to June 2015, the fellow will move to ASHG to conduct educational research and develop educational programs for a wide range of audiences.
- From July 2015 to December 2015, the fellow will have an option to do a rotation with a public or private organization involved in genetics education. The fellow will have an opportunity to participate in educational program administration, grant oversight and evaluation, science policy development, curriculum development, or genomic literacy program development.

Document 58

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

Federal Employee of the Year Award

NHGRI Intramural Research Highlights

Hypertension

American Heart Association

Childhood Family Living Arrangements and Blood Pressure in Black Men

Science

AAAS

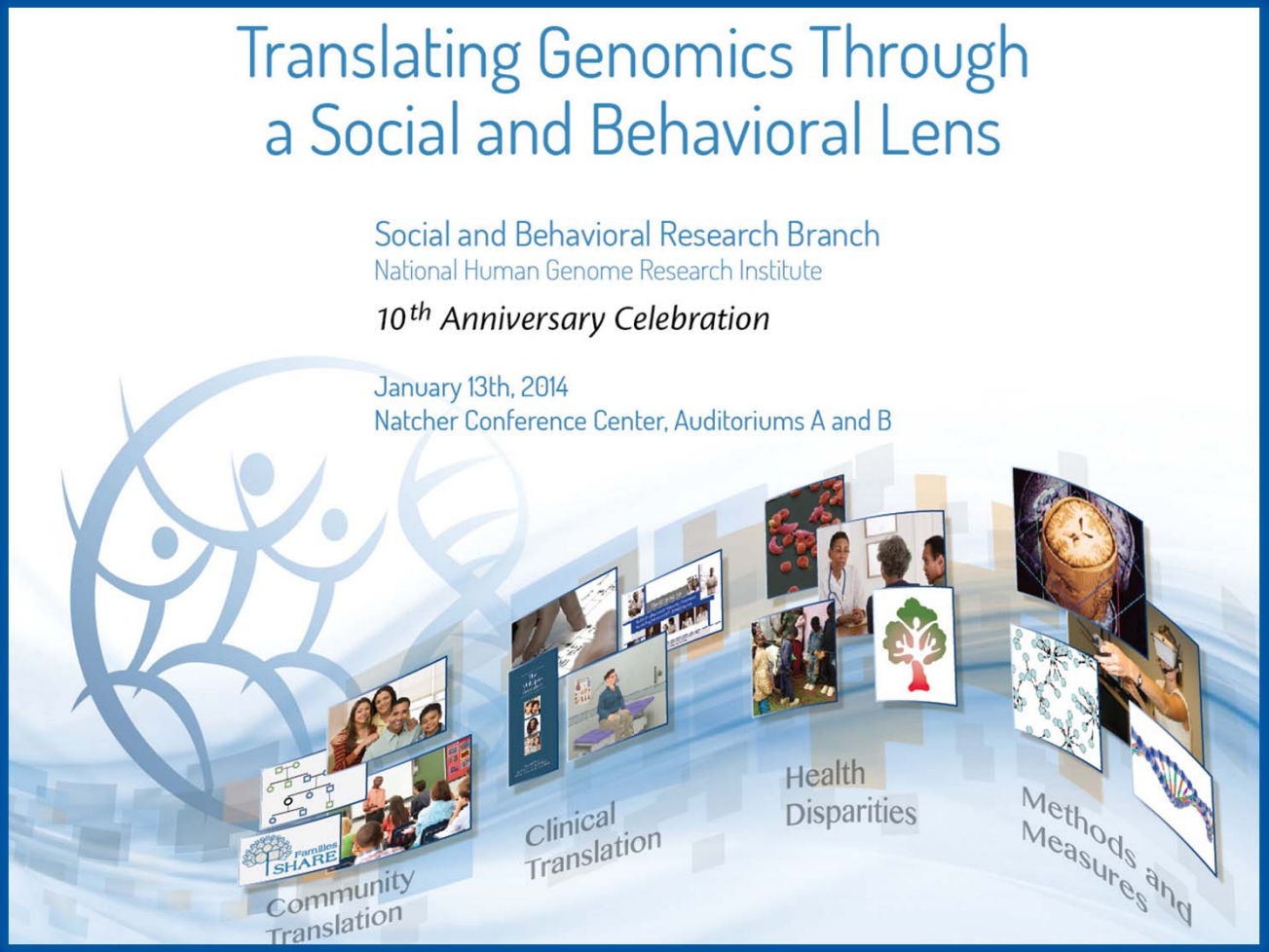
The genome of the ctenophore Mnemiopsis leidyi and its implications for cell type evolution

Cell

Cell Press

A Polymorphism in IRF4 Affects Human Pigmentation through a Tyrosinase-Dependent MITF/TFAP2A Pathway

10th Anniversary of the NHGRI Social and Behavioral Research Branch



Document 61

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

Special Thanks!


NATIONAL HUMAN GENOME
RESEARCH INSTITUTE
Advancing human health
through genomics research