



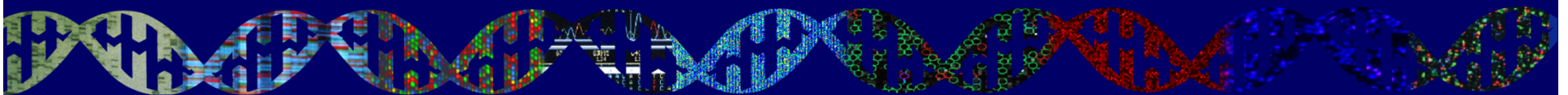
National Human Genome
Research Institute

DIRECTOR'S REPORT

**National Advisory Council
for Human Genome Research**

February 2014

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





genome.gov

National Human Genome Research Institute

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Director's Report-Related Documents: February 2014

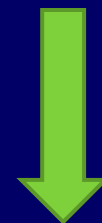
[Director's Report](#) 

[Director's Report](#) 

No.	Documents
1	New Director, NHGRI Division of Genomics and Society
2	The Genomics Landscape: A Monthly Update from the NHGRI Director
3	NHGRI-Smithsonian Genome Exhibition <ul style="list-style-type: none">• Exhibition Website• Exhibition-Related Programs• Reuben H. Fleet Science Center in San Diego
4	Fiscal Year 2014 Budget

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

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- II. General NIH Updates
- III. General Genomics Updates
- IV. NHGRI Extramural Research Program
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- VI. NHGRI Division of Policy,
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**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.

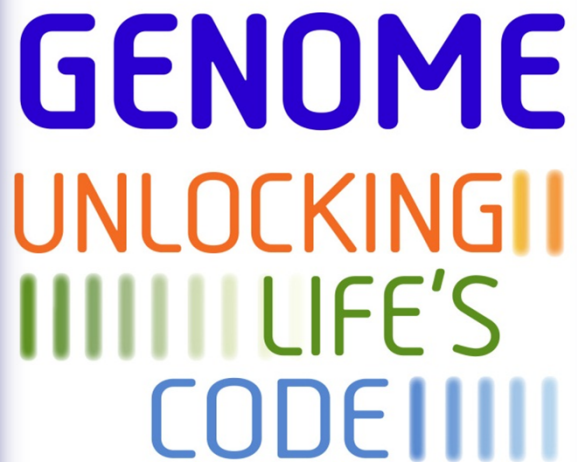
The Genomics Landscape

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**

Genome: Unlocking Life's Code Exhibition



GENOME
UNLOCKING
LIFE'S
CODE

- 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

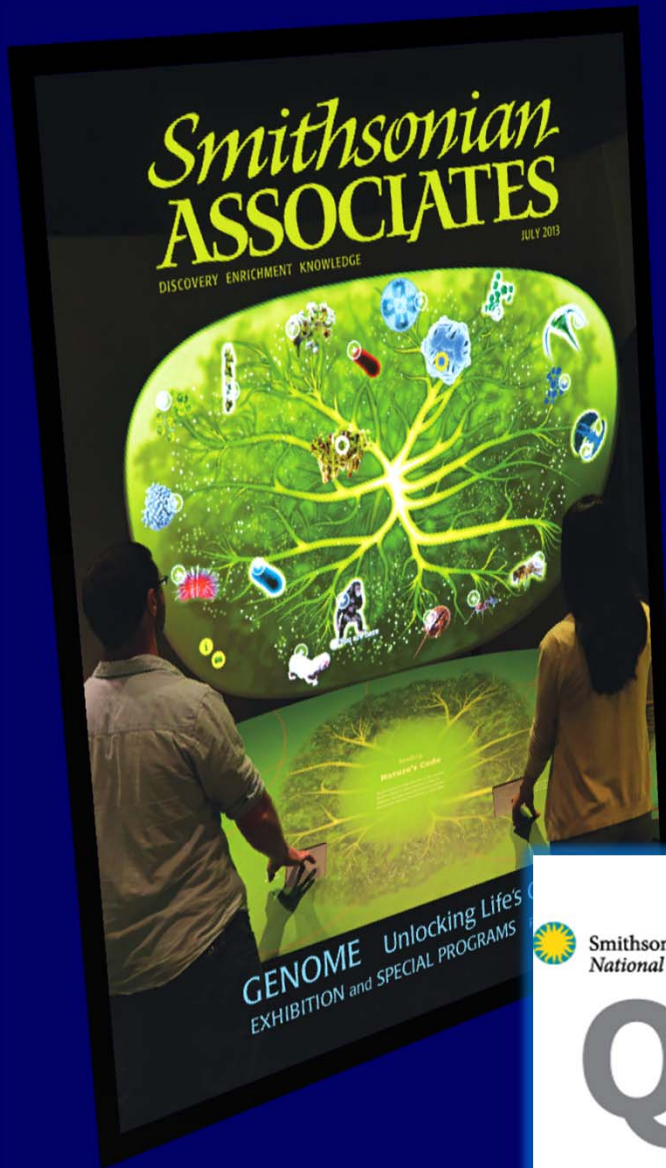
Genome Exhibition Attracting VIP Visitors




Genome Exhibition Attracting VIP Visitors



Genome Exhibition Programs




Smithsonian
 National Museum of Natural History

Q?rius

Unlock **your** world.

science + nature

UPCOMING PROGRAMS in the ongoing series complementing the exhibition *GENOME: Unlocking Life's Code* at the Natural History Museum. Programming is made possible thanks to generous grants and gifts made through the Foundation for the National Institutes of Health.

Finding Our Inner Neanderthal
 Evolutionary Geneticist Svante Paabo's DNA Quest

In 2010, Svante Paabo shook the scientific world with the results from a groundbreaking genetic study. Paabo, a director at the Max Planck Institute for Evolutionary Anthropology in Leipzig, Germany, headed a team whose Neanderthals, who disappeared 30,000 years ago, mated with members of our own species. "Neanderthals are not totally extinct, says the scientist. "In some of us they live on, a little bit" in the DNA of many people today.

Through extraction of ancient DNA from a variety of sources and geographic locations, Paabo's team studied genetic data across time and continents. He discusses the study's methods and findings and is later joined by Rick Potts, director of the Human Origins Program of the Natural History Museum, for a conversation about how paleogenetics can deepen our understanding of our species' long history.

Tues, March 25, 7-8:30 p.m.; Baird Auditorium, Natural History Museum; CODE 1M2-713; \$20-\$25

Is Genetic Information Different?

The increase in genetic testing is giving rise to new ethical and professional issues. For example, should you be able to control what genetic tests are performed on you? While conducting a specific test, should your doctor look for additional, unrelated genetic risk factors? Do you have responsibility to members who might not want to know the possible disease risks they share? Two experts in genetics and genomics address these questions in a lively, interactive event. After the opening discussion, enjoy a glass of wine while developing questions to ask the debaters.

Susan M. Wolf is the McLaughlin presidential professor of law, medicine, and public policy and the Frayre Baker Distinguished professor of law at the University of Minnesota. Robert C. Green is associate professor of medicine at the division of Hospital and Health Services at the University of Washington.

Thurs, April 10, 7-9 p.m.; Baird Auditorium, Natural History Museum; CODE 1P0-385; \$20-\$25; students with valid ID \$15

The Drama of DNA
 Genomics on Stage

Uncovering the human genome more than a decade ago led to questions about its complexities, realities, and implications. A number of contemporary playwrights used our lives, as well as to stimulate conversations about biomedical research.

Cassandra Medley (*Relativity*), Dorothy Fortenberry (*Good Egg*), Lisa Loamer (*Distraction*), and Anna Ziegler (*Photograph 51*) examine why and how they transpired complex topics rooted in genetics—including issues of identity, family dynamics around health decisions, and the power that genetic information holds—into compelling theater. Staged readings of scenes from their plays are presented by Ari Both, artistic director and director of Washington's Theater 1, where *Photograph 51* was produced in 2011.

The program is moderated by Karen Reibenberg, senior adviser to the director of Genomics and Society at the National Human Genome Research Institute and co-author of *The Drama of DNA*. She and the playwrights are joined by James Evans, Bryan Distinguished Professor of Genetics and Medicine at the University of North Carolina.

The program is presented in partnership with the National Museum of Natural History and the National Human Genome Research Institute with support from the Foundation for the National Institutes of Health.

Genome Exhibition Travel

Reuben H. Fleet Science Center

September 24, 2014 to January 4, 2015



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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



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%)

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.

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
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32 minutes
495 views

Tags: Medicine



Cancer Research at the National Institutes of Health
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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

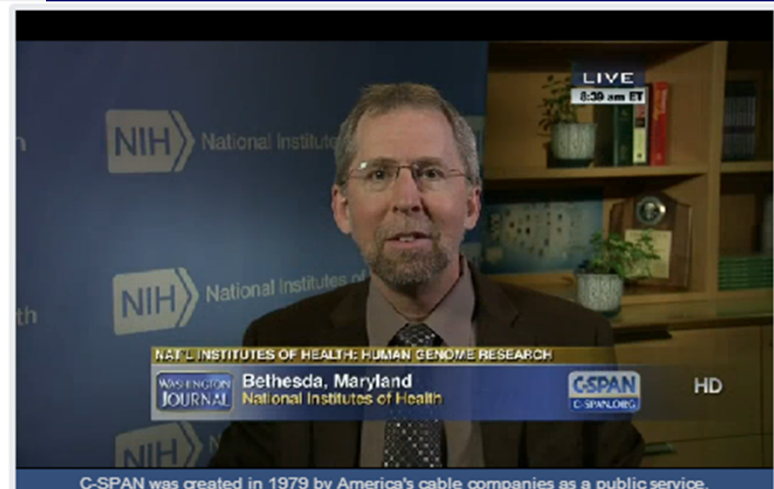


C-SPAN | Washington Journal

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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](#)



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

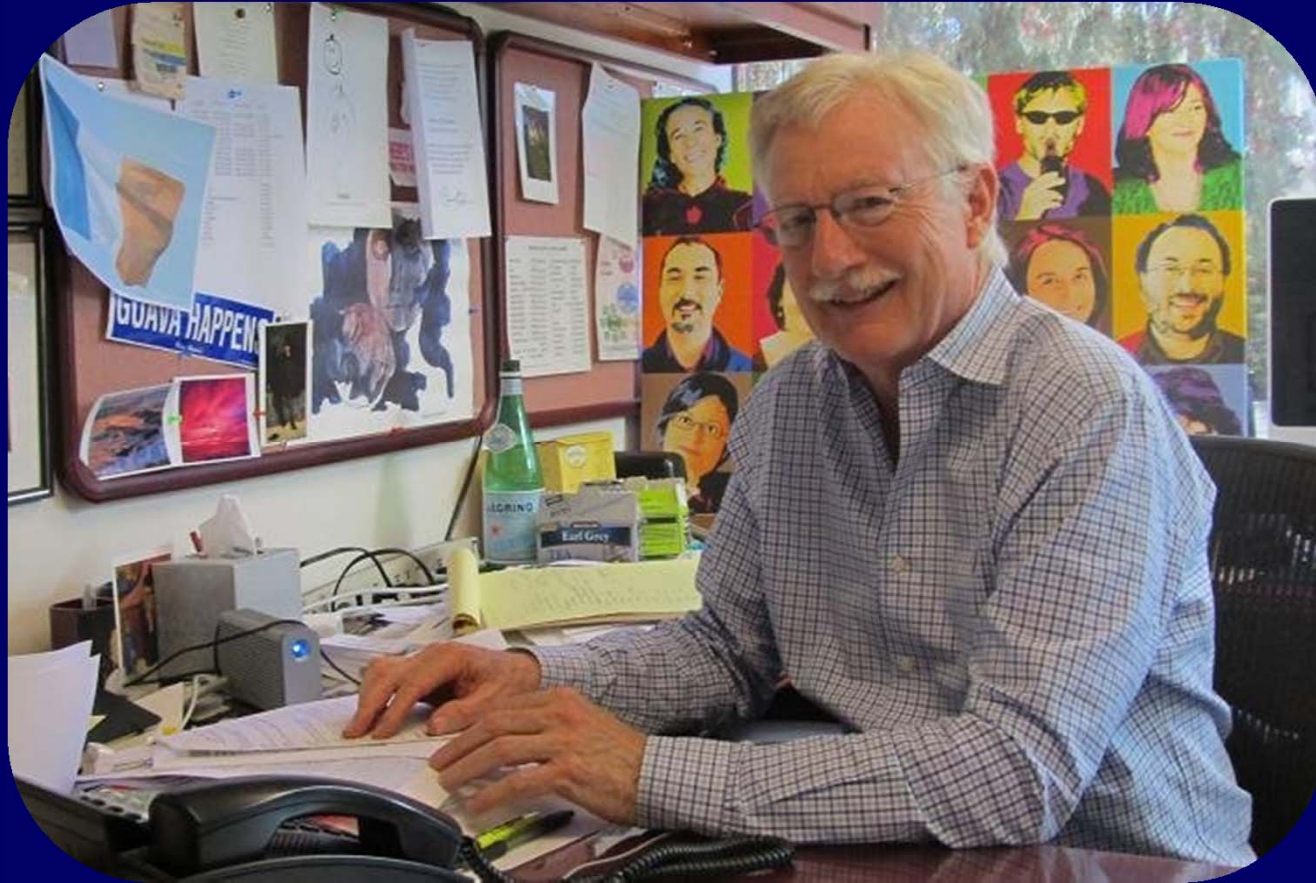


November, 2013

“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.

First NIH Associate Director for Data Science



Philip Bourne, Ph.D.

First NIH Chief Officer for Scientific Workforce Diversity



Hanna Valentine, M.D.

Surgeon General Nomination



Vivek Murthy, M.D.



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 genomic¹ 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.

U.S. Department of Health & Human Services www.hhs.gov
www.nih.gov

NIH Genomic Data Sharing (GDS)

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- Data Repositories
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Introduction

Genomic research advances our understanding of factors that influence health and disease. In January 2008, NIH established expectations for sharing data obtained through NIH-funded genome-wide association studies (GWAS) with the implementation of the [GWAS Policy](#). [GWAS research](#) compares DNA markers across the genome (an individual's complete genetic material) in people with a disease or particular trait to people without the disease or trait.

Information and resources related to the GWAS Policy can be found on this website. Any questions about the Policy can be e-mailed to GWAS@mail.nih.gov.

In the Spotlight

Advances in DNA sequencing technologies, as well as a steep drop in sequencing costs, have enabled NIH to fund research that generates a greater volume and wide range of genomic data. In light of these developments, NIH decided to extend the current GWAS Policy to encompass data from a broader spectrum of human and non-human genomic research.

On September 20, 2013, NIH released a draft *Genomic Data Sharing Policy* (GDS Policy) for a 60-day public comment period that closed November 20, 2013.

The draft GDS Policy is available at <http://www.federalregister.gov/a/2013-22941> and the public comments can be viewed at http://gds.nih.gov/pdf/GDS_Policy_Public_Comments.pdf.

On November 6, 2013, NIH hosted a webinar to provide an overview of the draft Genomic Data Sharing Policy and answer questions about its scope and expectations.

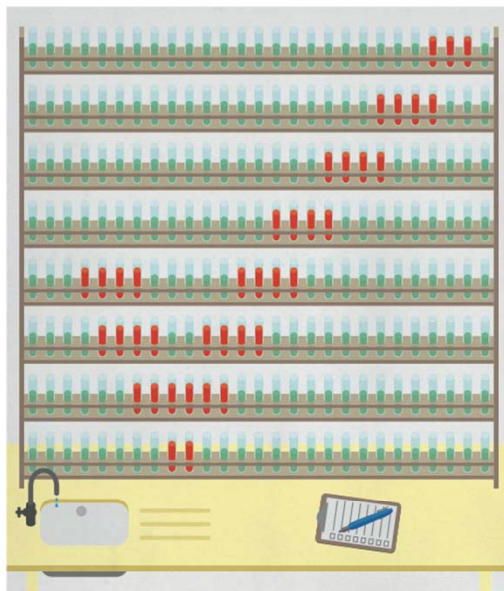
You can watch the archived webinar at <https://webmeeting.nih.gov/p7sqp5ayp6j/>. The webinar agenda is available at http://gds.nih.gov/pdf/webinar_agenda.pdf and the presentation slides are available at http://gds.nih.gov/pdf/webinar_slides.pdf.

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NIH Plan to Enhance Reproducibility

COMMENT



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 cases³. 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 design⁴. 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 edge⁵. 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 rewards⁶.

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 elements⁷.

Preclinical research, especially work that uses animal models⁸, 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.

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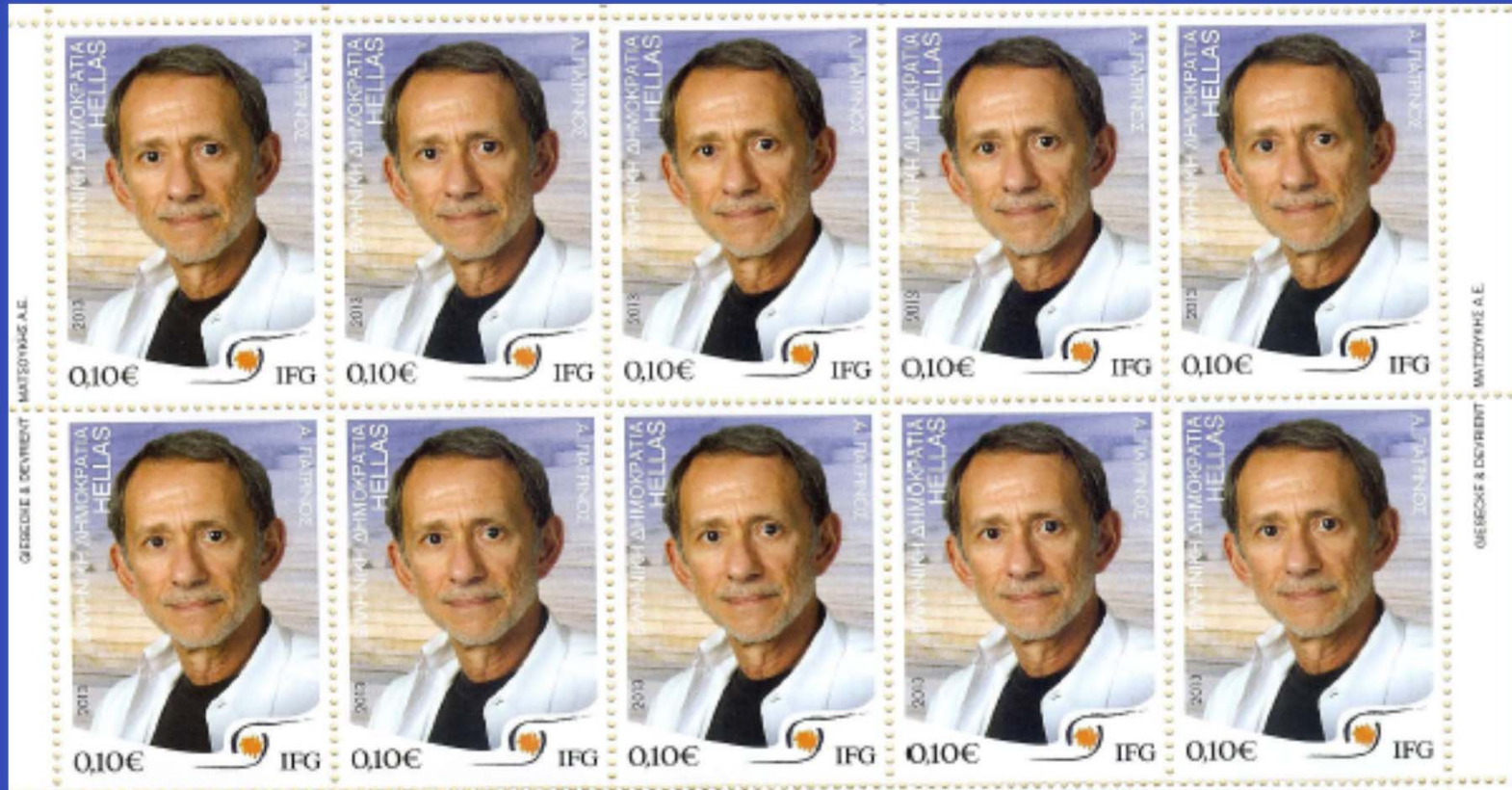
Mourning the Loss of Fred Sanger



Mourning the Loss of Janet Rowley



The International Foundation for Greece Greek Postage Stamp



Ari Patrinos, Ph.D.

Awards at 2013 ASHG Annual Meeting



Aravinda Chakravarti, Ph.D.
William Allan Award



John Moran, Ph.D.
Curt Stern Award

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



Joe DeRisi, Ph.D.

2014 Breakthrough Prize in Life Sciences



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

Elected to the Institute of Medicine

David DeMets

Judy Garber

Richard Kolodner

Brendan Lee

Pamela Sklar

Chris Walsh



INSTITUTE OF MEDICINE
OF THE NATIONAL ACADEMIES

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

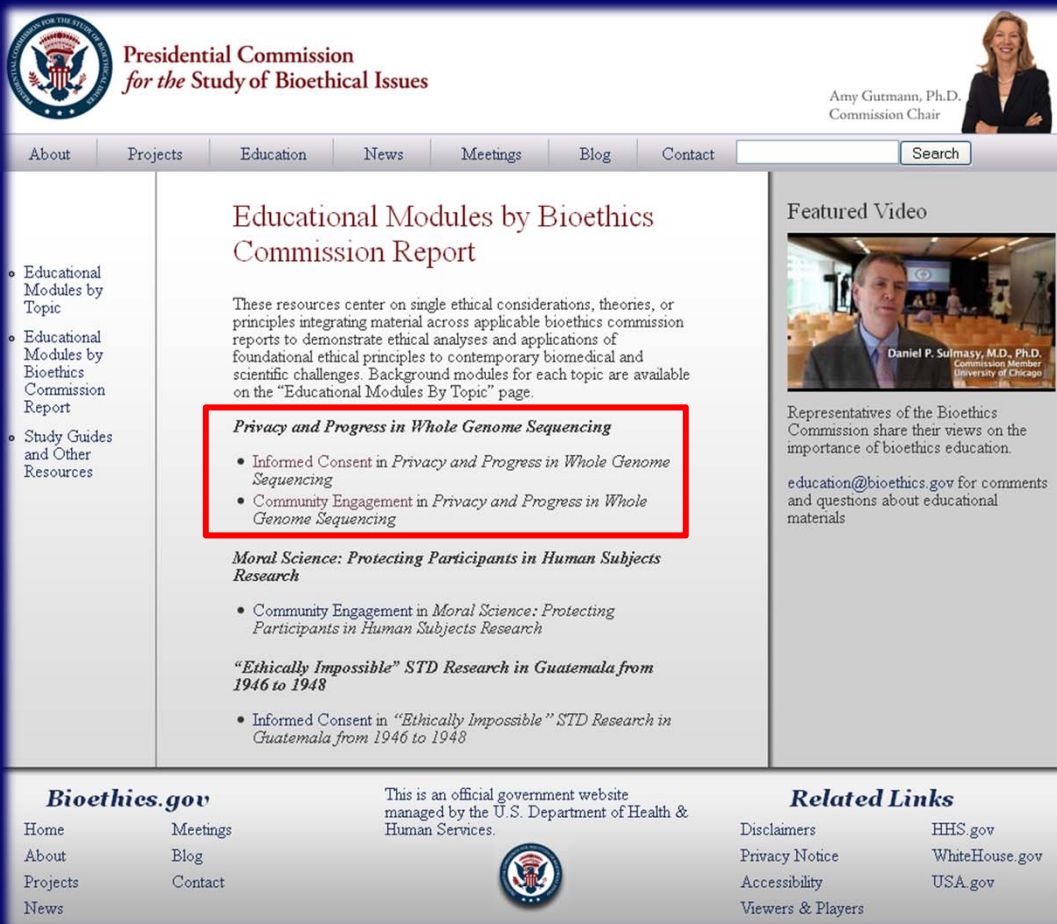



NCHPEG Transitions to The Jackson Laboratory



- **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**

Presidential Commission for the Study of Bioethical Issues



 Presidential Commission
for the Study of Bioethical Issues

Amy Gutmann, Ph.D.
Commission Chair

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Educational Modules by Bioethics Commission Report

These resources center on single ethical considerations, theories, or principles integrating material across applicable bioethics commission reports to demonstrate ethical analyses and applications of foundational ethical principles to contemporary biomedical and scientific challenges. Background modules for each topic are available on the "Educational Modules By Topic" page.

Privacy and Progress in Whole Genome Sequencing

- Informed Consent in *Privacy and Progress in Whole Genome Sequencing*
- Community Engagement in *Privacy and Progress in Whole Genome Sequencing*


Moral Science: Protecting Participants in Human Subjects Research

- Community Engagement in *Moral Science: Protecting Participants in Human Subjects Research*

"Ethically Impossible" STD Research in Guatemala from 1946 to 1948

- Informed Consent in *"Ethically Impossible" STD Research in Guatemala from 1946 to 1948*

Featured Video



Daniel P. Sulmasy, M.D., Ph.D.
Commission Member
University of Chicago

Representatives of the Bioethics Commission share their views on the importance of bioethics education.

education@bioethics.gov for comments and questions about educational materials


Bioethics.gov

This is an official government website managed by the U.S. Department of Health & Human Services.

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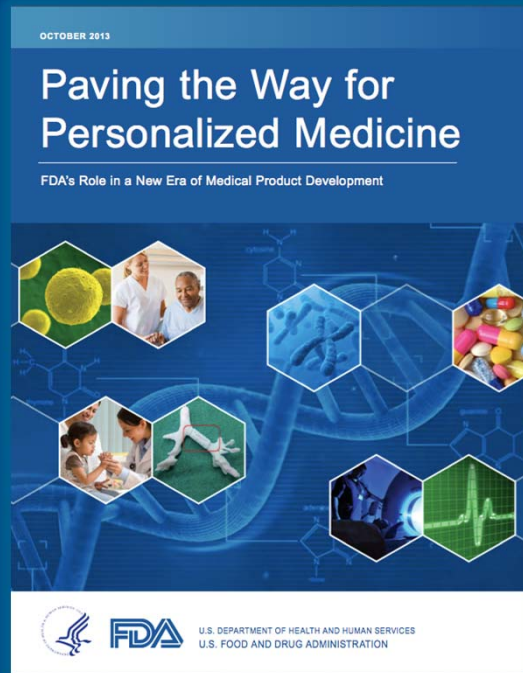
ANTICIPATE and COMMUNICATE

Ethical Management of Incidental and Secondary Findings in the Clinical, Research, and Direct-to-Consumer Contexts

Presidential Commission
for the Study of Bioethical Issues

December 2013

FDA and Genomics



The NEW ENGLAND JOURNAL *of* MEDICINE

First FDA Authorization for Next-Generation Sequencer

Francis S. Collins, M.D., Ph.D., and Margaret A. Hamburg, M.D.

NEJM (2013)

FDA and FTC Focus on Genomics



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A promotional banner for 'forU ACCELERATION'. It features a sleek black sports car on the left, a checkered racing flag at the top, and several purple and black diagonal banners on the right with the text: 'Turbo Your Success', 'Drive New Business', 'Accelerate Your Wealth', and 'Drive Your Goals'. The main text reads 'forU | ACCELERATION' and 'Life. Personalized. | What Drives Your DNA?'. A purple 'REGISTER' button and the text 'Do Not Miss This Event.' are at the bottom.

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

found
2013

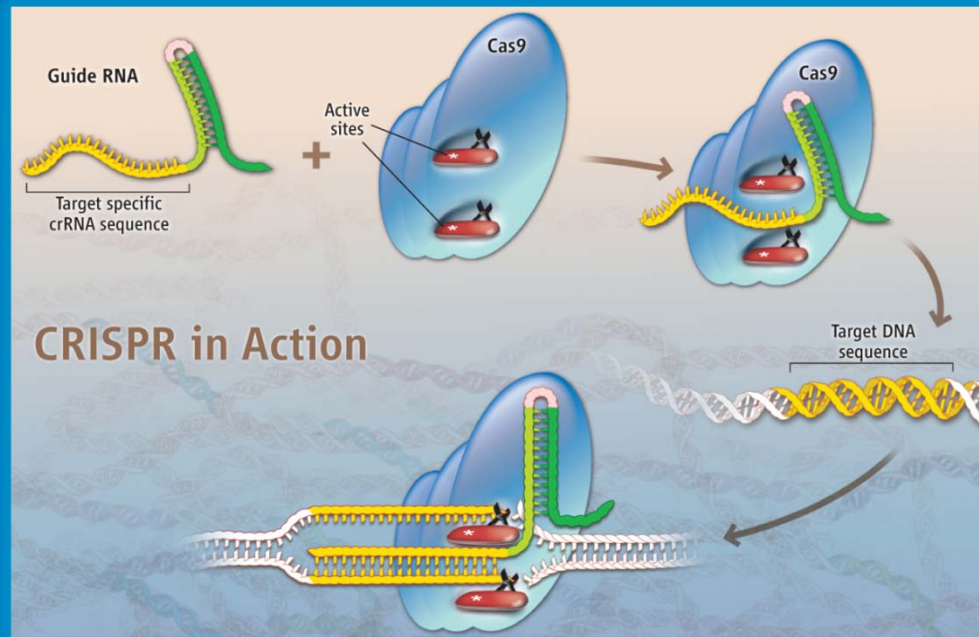


We know that the human genome is the molecular instruction book for building the human body, but exactly what are all the intricacies of how it functions? In 2003, the Human Genome Project (HGP) reached completion, comprehensively sequencing the 3 billion base pairs that make up a full human genome. 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.



Two groups fostering this revolution:

Centers of Excellence in
Genomic Science
George Church

Large Scale Sequencing
& Analysis Center
Eric Lander

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...



MIND & BODY



Crack Your Own Code

Having your DNA analyzed doesn't involve a million-dollar machine or even your doctor. Just order a test, spit, mail it back, and learn how to optimize your nutrition, exercise, and health.

by JOSEPH HOOPER

Illustration by BRIAN STAUFFER | OCTOBER 2013 | 67 MEN'S JOURNAL

JONATHAN ZITRAIN, a 43-year-old Harvard law professor, went to the hospital suffering from a mysterious fever when his doctors found a mass in his liver that they couldn't explain. To find out just what was wrong, Zitrain turned to a test he had ordered online previously from 23andMe, the largest company in an expanding field of "personal genomics" which tests out your genetic info from a saliva sample as if you were a pop star. It estimates your risk for more than 100 diseases and conditions. A very ill Zitrain found that he was at high risk for blood clots — "venous thromboembolism" — a clot that helped his doctors figure out which they removed. He soon recovered completely. "It's a data-driven guy," says Zitrain, "and I saw the test as more than just a curiosity."

Two years ago, the Human Genome Project deciphered the entire human DNA code at a cost of about \$3 billion. What no one dreamed at the time was that the technology would quickly become so accessible and so cheap. 23andMe, for one, recently slashed the price of its product to \$99 and aims to serve a million people by the end of this year. The genomic panels offered by 23andMe and its competitors can arm us with data about disease risk that we can use to help make lifestyle decisions or, as in Zitrain's case, pull out of our pocket in an emergency. And while doctors are not yet routinely sending off samples to personal genomic companies, this simple online test is one of the most all-encompassing — and high-tech — ways to take health care into your own hands.

What Our Genes Tell Us
Unlike blood work or an MRI, a genetic test is neither invasive nor time-consuming. The drill goes something like this: You order a test off the website of 23andMe or MyHeritage-based Inherent Health, and when it arrives in the mail, you produce the DNA sample — a vial of saliva for 23andMe and a cheek swab for Inherent — and mail it back. In about

TECH JANUARY/FEBRUARY 2014

When Will Genomics Cure Cancer?

A conversation with the biogeneticist Eric S. Lander about how genetic advances are transforming medical treatment

JAMES FALLOWS | DEC 22, 2013, 9:25 PM ET

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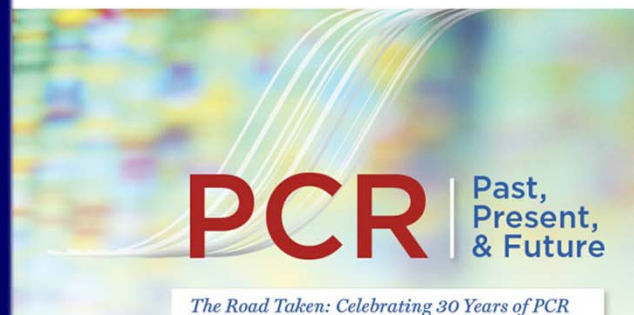
The Scientist » Magazine » Special Section

PCR: Past, Present, & Future

Highlights from a webinar held by *The Scientist* to celebrate 30 years of PCR: the technique's invention, quantitative real-time PCR, and digital PCR

By Jeffrey M. Perkel | December 1, 2013

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GENETIC TESTING: PROMISES & PROBLEMS

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Consumers Digest

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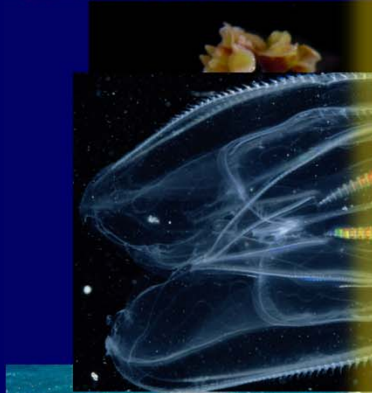
Genomes In The News...



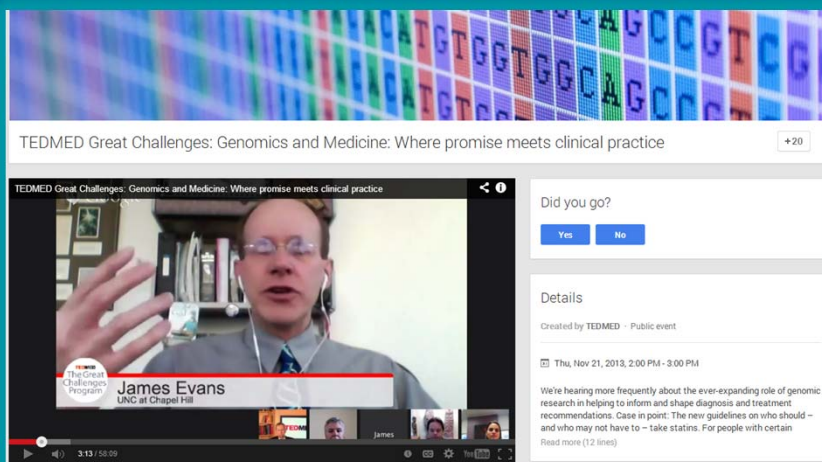
Genomes of the Year

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.



Genomics In Video...



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

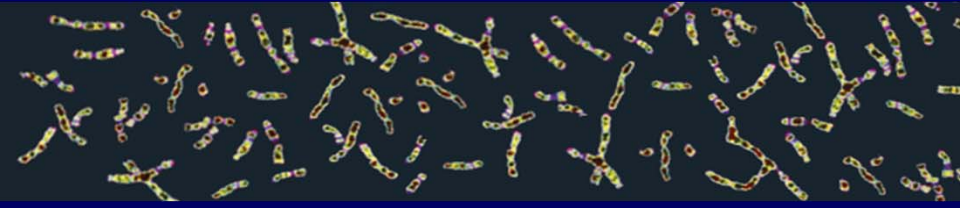
Large-Scale Genome Sequencing and Analysis Centers



- **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)

1000 Genomes

A Deep Catalog of Human Genetic Variation

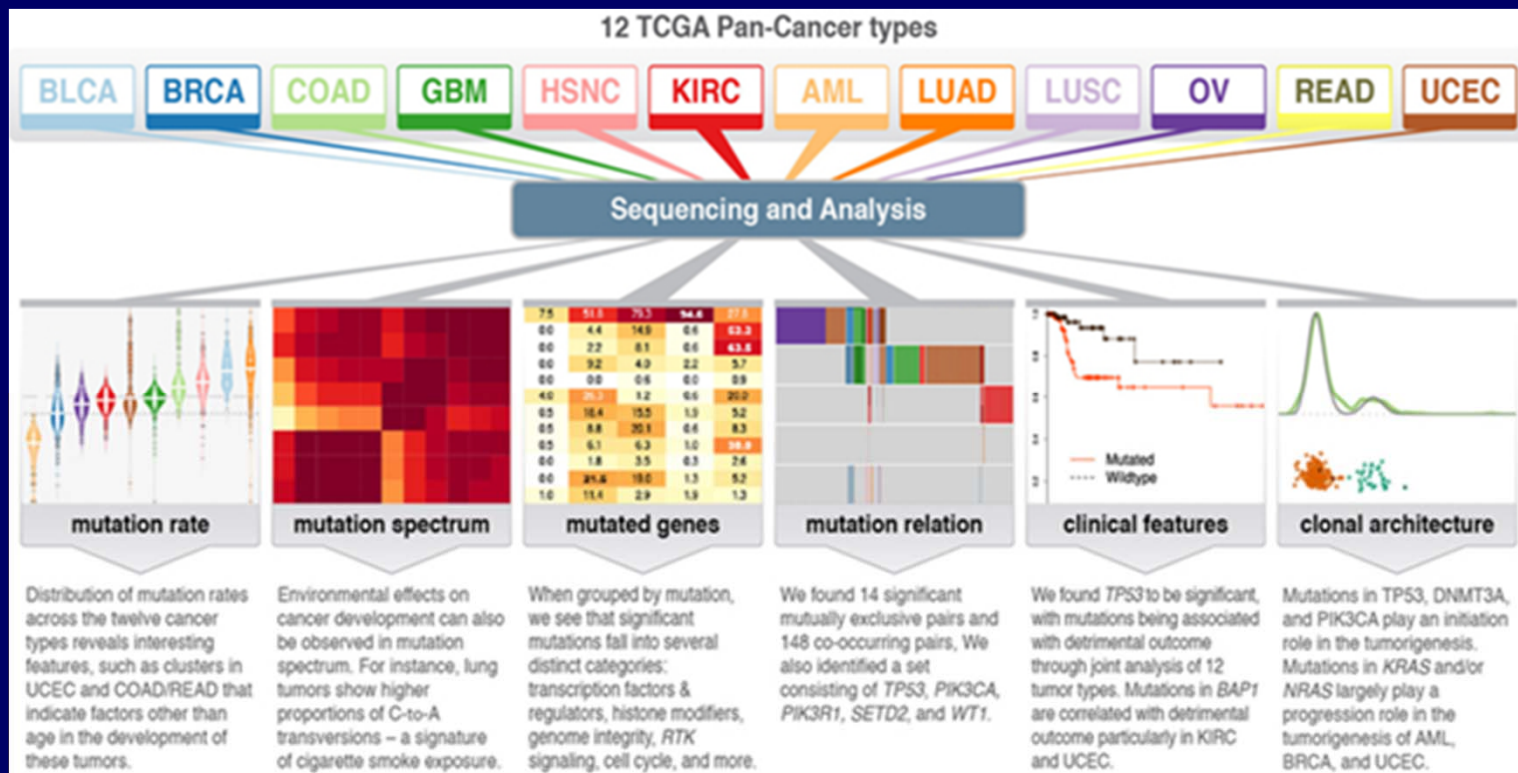


- **Currently ~90 million variants in 2535 samples**
 - ~76 million SNPs
 - ~7 million indels
 - ~7 million structural and other complex variants
- **Final release of variants by Fall 2014**
- **Final 1000 Genomes Project Meeting and Community Meeting: June 24-26 in the U.K.**



■ Pan-cancer analysis

Nature Genetics feature: 27 papers and 5 thematic threads
 ICGC/TCGA whole-genome pan-cancer analysis project



■ Sample accrual is completed

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

- **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



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, PhD¹⁻⁴, Laura M. Amendola, MS⁵, Christine Eng, MD⁶, Eliezer Van Allen, MD⁷⁻⁹, 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, MD⁵, Wylie Burke, MD, PhD^{5,17}, Nancy B. Spinner, PhD¹⁵, Levi Garraway, MD, PhD^{7,12,18}, Robert C. Green, MD, MPH^{12,19}, Sharon Plon, MD, PhD^{8,20}, James P. Evans, MD, PhD¹⁻⁴ 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, PhD², Dean Karavite, MS^{1,7}, Matthew Lebo, PhD^{1,5,9}, 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, PhD¹⁰

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

Wylie Burke, MD, PhD¹, Armand H. Matheny Antommaria, MD, PhD², Robin Bennett, MS, CGC³, Jeffrey Botkin, MD, MPH⁴, Ellen Wright Clayton, MD, JD⁵, Gail E. Henderson, PhD⁶, Ingrid A. Holm, MD, MPH⁷⁻⁹, Gail P. Jarvik, MD, PhD³, Muin J. Khoury, MD, PhD¹⁰, Bartha Maria Knoppers, JD, PhD¹¹, Nancy A. Press, PhD¹², Lainie Friedman Ross, MD, PhD¹³, Mark A. Rothstein, JD¹⁴, Howard Saal, MD¹⁵, Wendy R. Uhlmann, MS, CGC¹⁶, Benjamin Wilfond, MD¹⁷, Susan M. Wolf, JD¹⁸ and Ron Zimmern, FRCP, FFPHM¹⁹

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

Michael O. Dorschner^{1,4,5}, Laura M. Amendola², Emily H. Turner^{1,5}, Peggy D. Robertson¹, Brian H. Shirts⁵, Carlos J. Gallego², Robin L. Bennett², Kelly L. Jones², Mari J. Tokita², James T. Bennett^{2,3}, Jerry H. Kim⁸, Elisabeth A. Rosenthal², Daniel S. Kim¹, National Heart, Lung, and Blood Institute Grand Opportunity Exome Sequencing Project, Holly K. Tabor^{5,6}, Michael J. Bamshad^{1,2}, Arno G. Motulsky^{1,2}, C. Ronald Scott^{2,3}, Colin C. Pritchard⁵, Tom Walsh², Wylie Burke^{2,6}, Wendy H. Raskind^{2,4}, Peter Byers^{2,7}, Fuki M. Hisama², Deborah A. Nickerson¹ and Gail P. Jarvik^{1,2,9}

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., Zhiyi 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 Hodges¹, Amy Gursky^{1,2}, Javed Siddiqui^{1,2}, Scott A Tomlins^{1,2}, Sameek Roychowdhury⁵, Kenneth J Pienta⁶, Scott Y Kim⁷, J Scott Roberts⁸, James M Rae^{3,9}, Catherine H Van Poznak⁹, Daniel F Hayes⁹, Rashmi Chugh⁹, Lakshmi P Kunju^{1,2}, Moshe Talpaz⁹, Anne F Schott⁹ & Arul M Chinnaiyan^{1-4,10,11}

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

Clinical Sequencing Exploratory Research (CSER) Program



PROJECTS

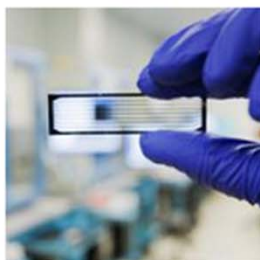
PUBLICATIONS

RESOURCES

IMPACT

Log in

Moving the genome into the clinic

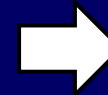
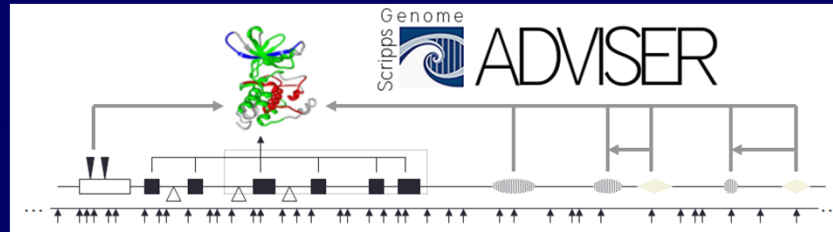
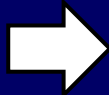


In the past, standard medical practice for genetic testing involved looking at one gene at a time. With new advances in our understanding of the genomic basis of health and disease and in technology, it is now possible to test all of our genes at once using tests called whole exome or whole genome sequencing. 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.



Genome Sequencing Informatics Tools

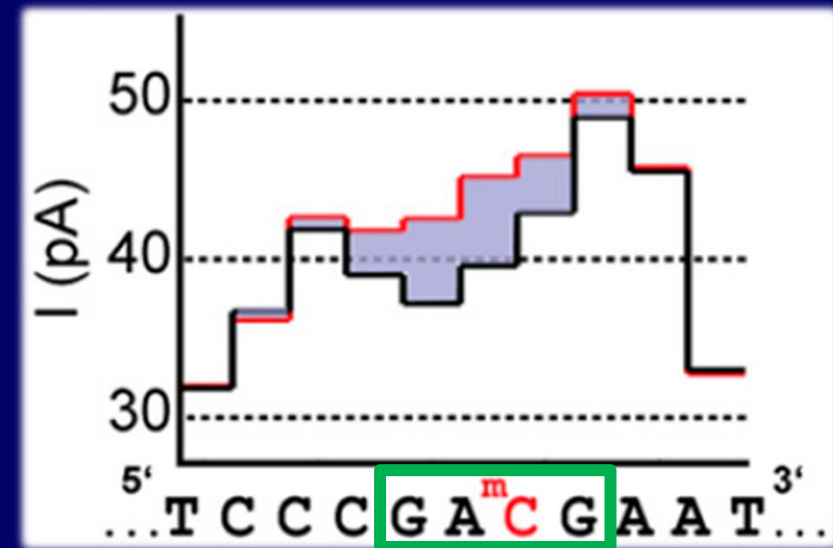
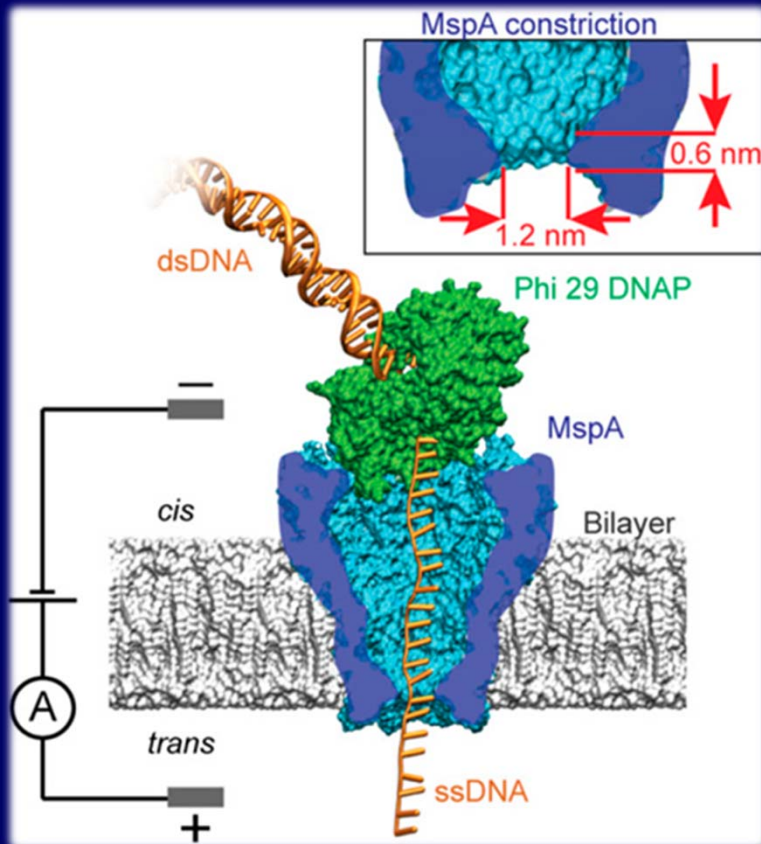


- **GS-IT has released SG ADVISER to annotate and filter genomic variants**
- **Used to solve clinical case of 15-year-old girl**
- **iSeqTools Portal: “The search for what’s wrong with Lilly”**



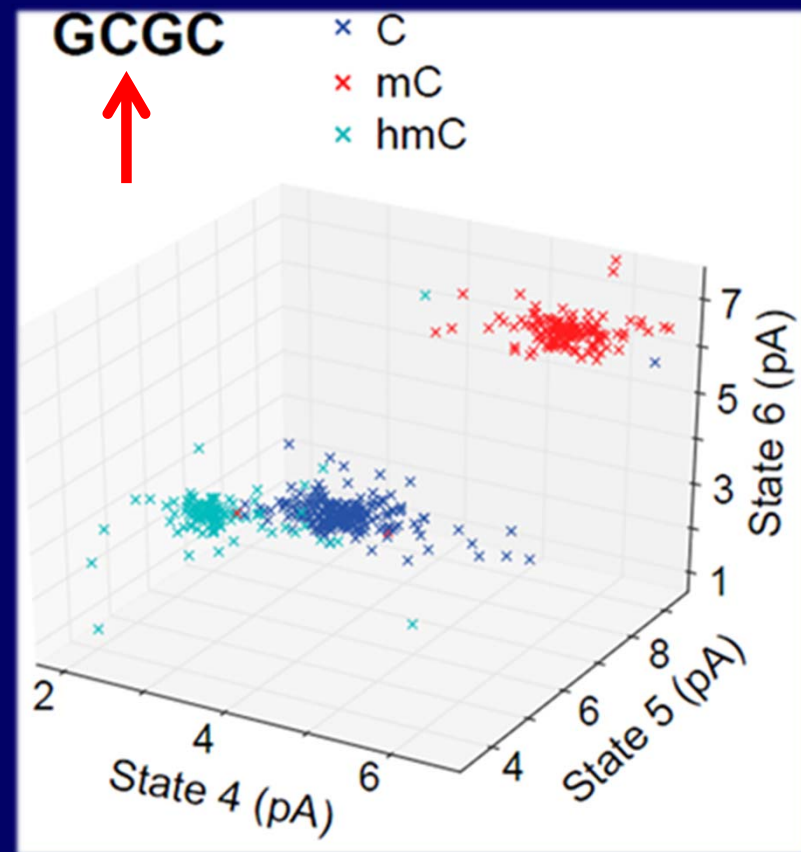
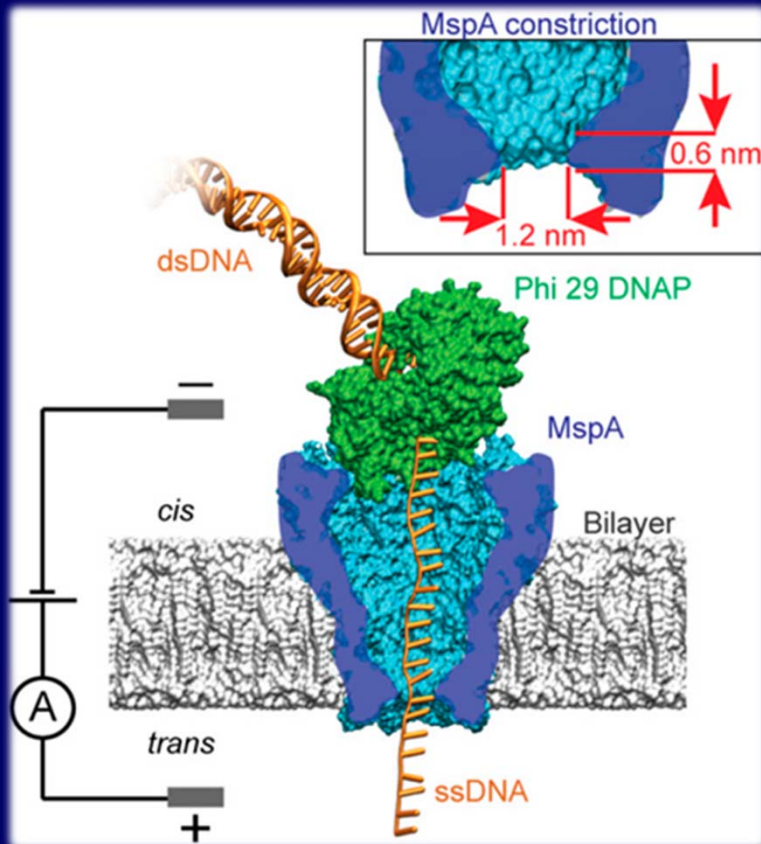
DNA Sequencing Technology Development

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



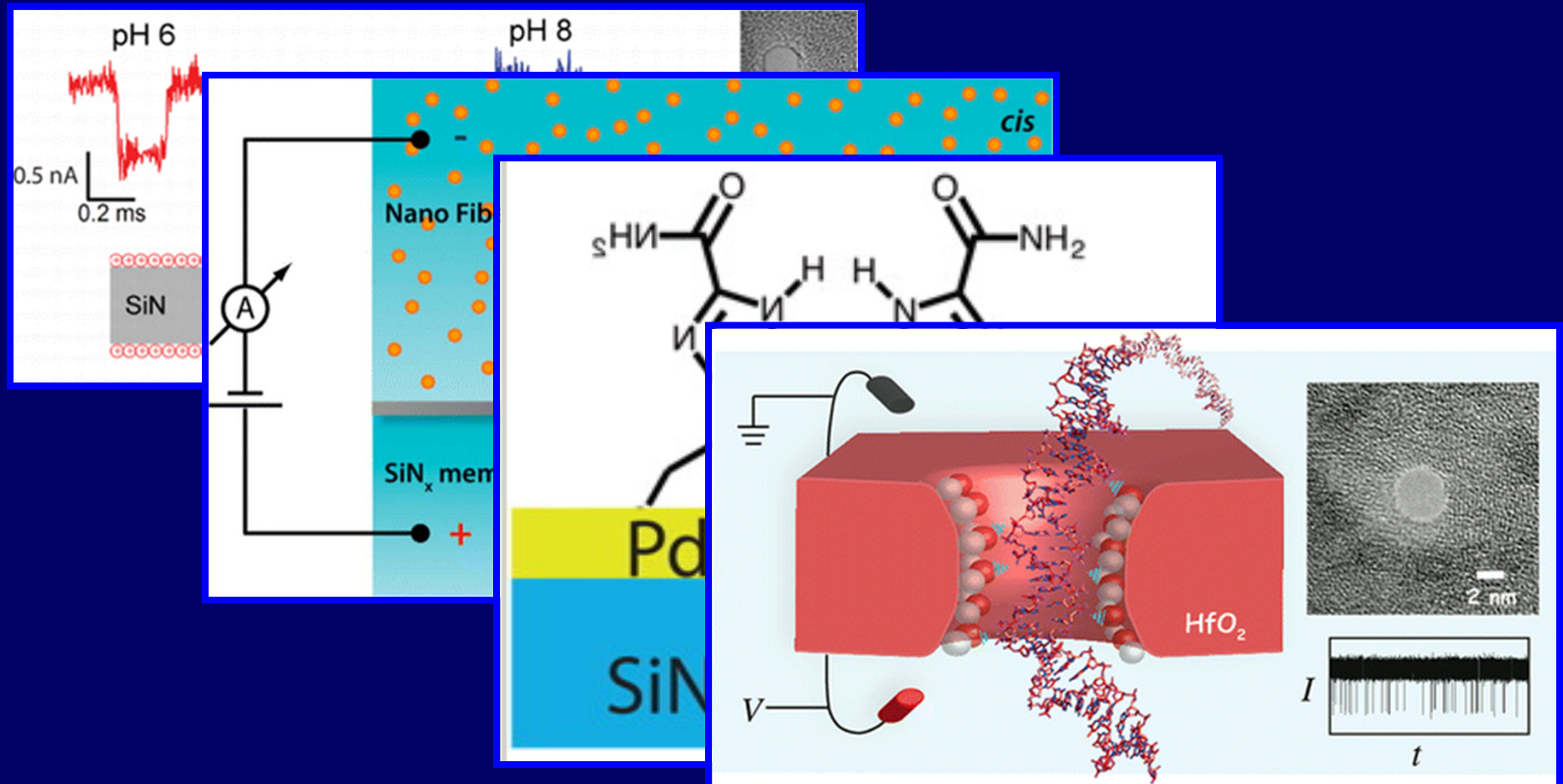
DNA Sequencing Technology Development

- Sequencing of epigenetic modifications using biological nanopores
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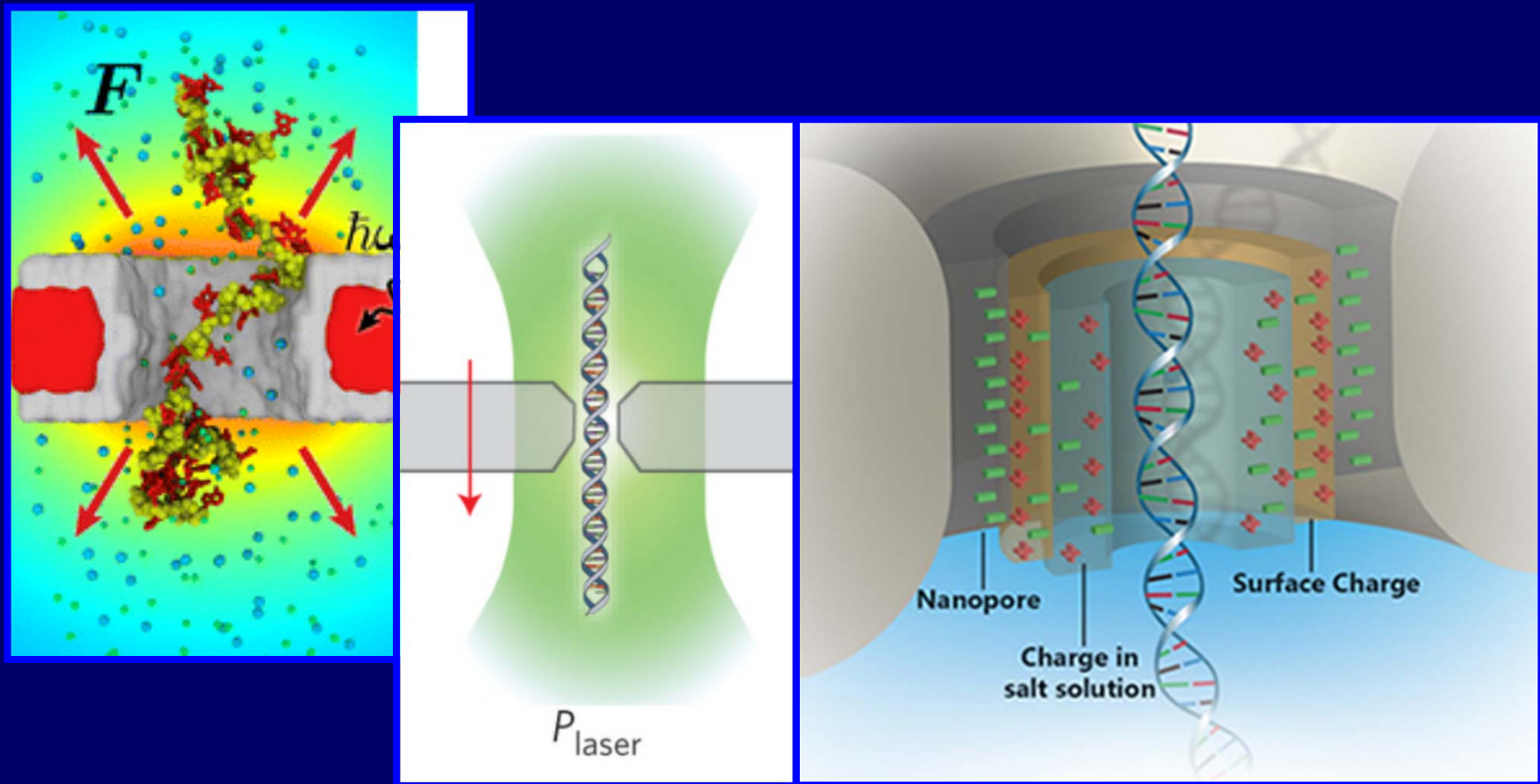
DNA Sequencing Technology Development

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



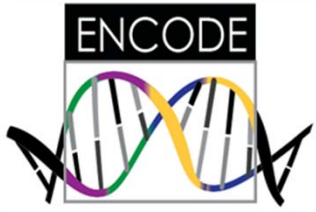
DNA Sequencing Technology Development

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



DNA Sequencing Technology Development

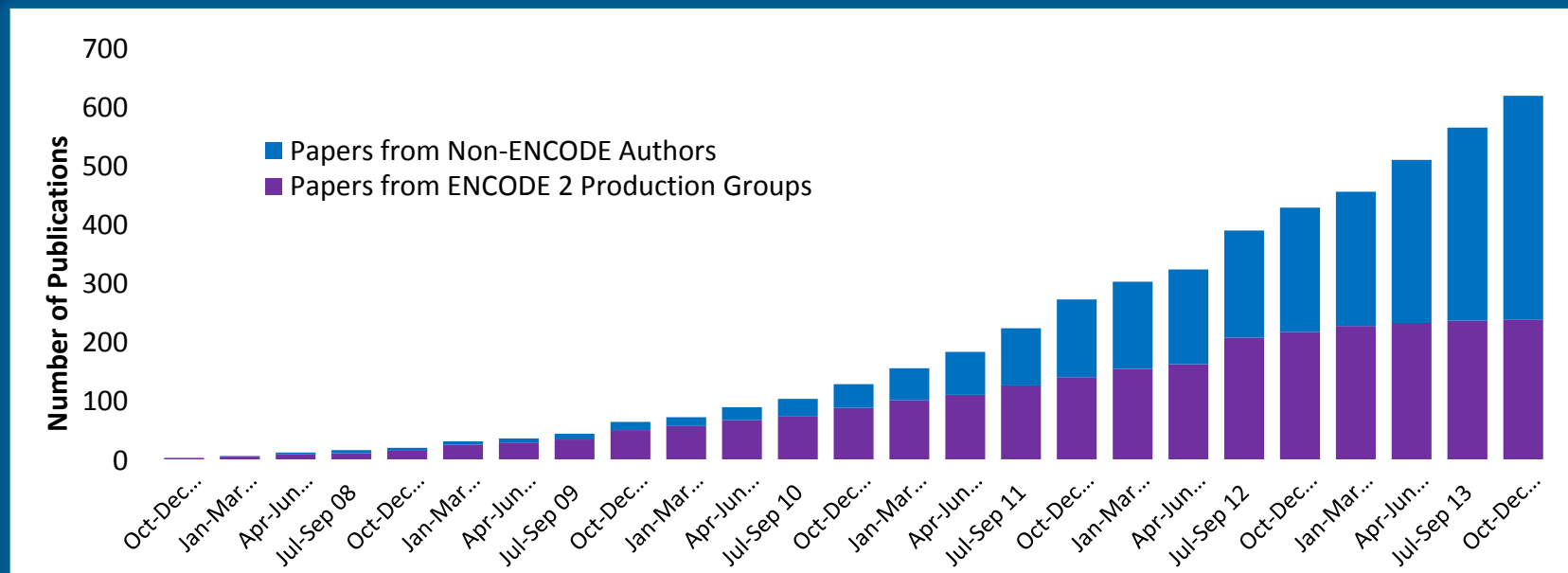




ENCODE



- **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

CEGS



DANA-FARBER
CANCER INSTITUTE

HARVARD
MEDICAL SCHOOL

Northeastern
UNIVERSITY

UNIVERSITY OF
TORONTO



CCSB Center of Excellence in Genomic Science

- 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**

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

Teri

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

Con

INTER-SOCIETY COORDINATING COMMITTEE, COMPETENCIES WORKGROUP

Fa

PROPOSED USE CASE TEMPLATE AND EXAMPLE

EP/

INTER-SOCIETY COORDINATING COMMITTEE, USE CASE WORKGROUP
(12/18/2013 draft)

Template

I. Specialty/Professional Society

- Next meeting April 2014

eMERGE Network

Genetics

October 2013
Volume 15 | Number 10
www.geneticsinmedicine.org

Nat Biotechnol • doi: 10.1038 • November 2013

Systematic comparison of phenome-wide association study of electronic medical record data and

MATTER

Linking Genes to Diseases by Sifting Through Electronic Medical Records

By CARL ZIMMER

Published: November 28, 2013

The eMERGE Network

electronic Medical Records & Genomics

Future Directions for the eMERGE Network

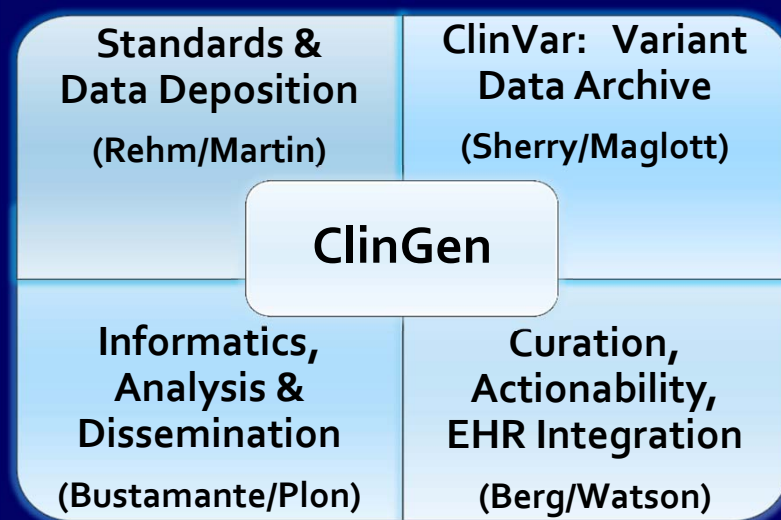
Wednesday, January 22, 2014,
8:00 a.m. - 5:30 p.m.

This event will be recorded and live streamed on
genome.gov/GenomeTVlive

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

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



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



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

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

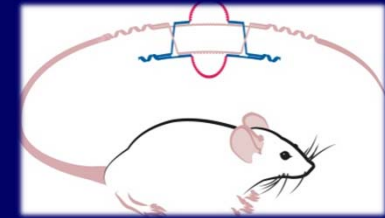
- I. General NHGRI Updates
- II. General NIH Updates
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Human Microbiome Project (HMP)

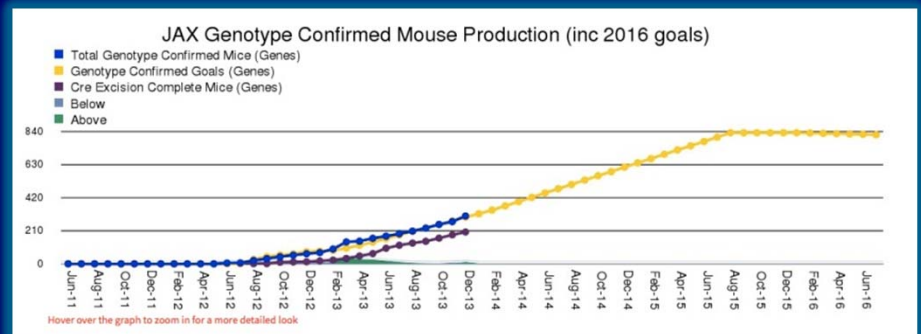
- Phase 2: Fiscal Year 2013 to 2015
- Create integrated multi-'omics dataset of microbiome and host properties
- A Common Fund initiative with co-funding from other ICs, \$22.1M total
(NIDDK, NCCAM, ORWH, ODS)
- HMP2 Consortium:
 - Preterm birth and microbiome: Buck
 - IBD and microbiome: Xavier/Huttenhower
 - Diabetes and microbiome: Snyder>Weinstock



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



phenodcc

Reporting Tracker Quality Control Colin Fletcher Sign Out

Centre	XML File Name	Phase	Status	Specimen Name	Baselin	Colony Name	Strain	Date of Birth	Active	Valid
MRC Harwell	MAPKBP1-TM1B-1C/1.1b_52...						MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	Valid	failed	MAPKBP1-TM1B-1C/1.1c_520...	✓		MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	Valid	failed	MAPKBP1-TM1B-1C/1.1d_52...	✓		MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/1.1e_52...	✓		MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/3.1a_52...	✓		MGI:2164831	26-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/4.1a_52...	✓		MGI:2164831	26-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/4.1b_52...	✗	MRC - Harwell-HEPD0507_9...	MGI:2164831	26-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/4.1e_52...	✓		MGI:2164831	26-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/5.1a_52...	✗	MRC - Harwell-HEPD0507_9...	MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/5.1c_520...	✗	MRC - Harwell-HEPD0507_9...	MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/5.1d_52...	✗	MRC - Harwell-HEPD0507_9...	MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/5.1h_52...	✓		MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/5.1i_520...	✗	MRC - Harwell-HEPD0507_9...	MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/6.1a_52...	✗	MRC - Harwell-HEPD0507_9...	MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/6.1e_520...	✗	MRC - Harwell-HEPD0507_9...	MGI:2164831	25-Dec-12	✗	✓
	H.2013-04-23...	over...	done	MAPKBP1-TM1B-1C/6.1f_520...	✗	MRC - Harwell-HEPD0507_9...	MGI:2164831	25-Dec-12	✗	✓

Submitted Files Specimens

Additional Tools

Filter Options

Start Date: 1-Jul-2012

Stop Date: 22-Jan-2014

Phase: download zip_r un zip_md5 xp

Watch Reset

Running Processes

Submitted Zip Files

Additional Information

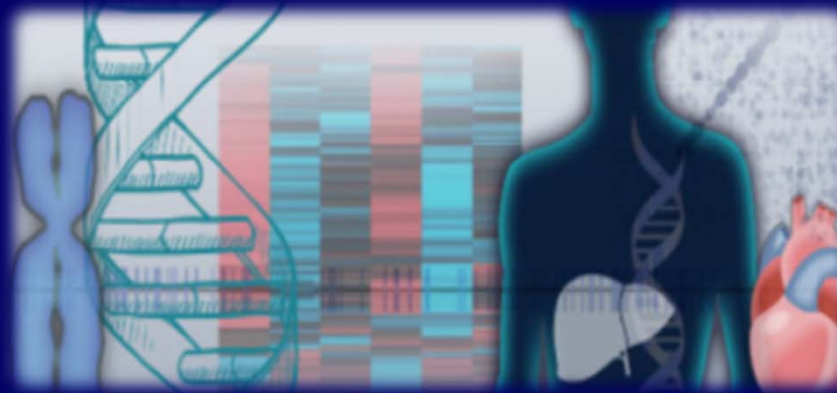
File Info File Errors Specimen Info Validation Issues

XML File Name: H.2013-04-23.03.specimen.impc.xml

Zip: H.2013-04-23.03.impc.zip

Processing Phase: overview

XML Status: done



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

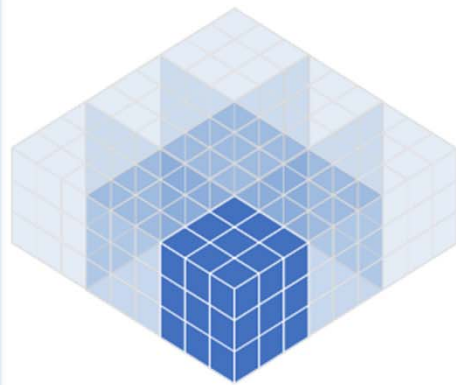


PROTEIN CAPTURE REAGENTS PROGRAM

- Protein Capture Data Portal went live in October

The screenshot shows the Protein Capture Data Portal interface. At the top, there is a navigation bar with the logo and text 'PROTEIN CAPTURE REAGENTS PROGRAM', and links for 'Home', 'About', and 'Download'. Below the navigation bar, the main content area is titled 'Data Portal' and includes a 'Having trouble? Try watching the tutorial' link. The interface features a sidebar with filters for 'ANTIGEN SOURCE LAB(S)' (set to 'None') and 'ONLY SHOW PROTEINS/ANTIGENS WITH BINDERS' (set to 'On'). The main content area has tabs for 'Binders', 'Antigens', 'Proteins', and 'Validations'. It includes a 'Show / hide columns' button, a search bar, and a pagination control showing 'Showing 1 to 50 of 150 entries'. A table is displayed with columns for 'HGNC Name', 'Antigen', 'Binder Name', 'Source Lab', 'Data Files', 'Passed Validations', and 'Distributors'. Each column has a dropdown menu for filtering.

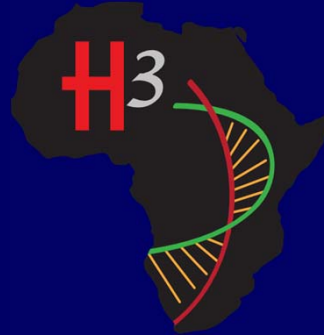
- 3rd Annual Consortium Meeting in December



NIH LINCS
PROGRAM

- **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



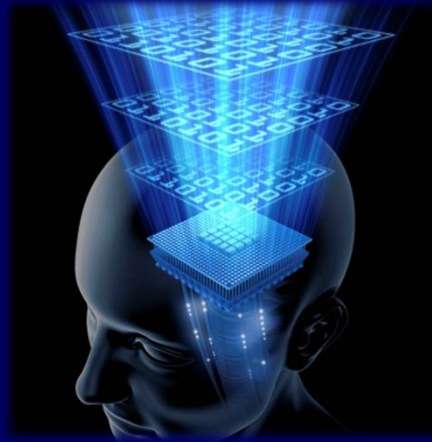
- **3rd Consortium Meeting held in Johannesburg in October (without NIH staff)**
- **Policy documents in final stages of approval**
- **H3Africa marker paper submitted**
- **Many grantees have received ethics approval**
- **Sample collection underway**
- **4th Consortium Meeting in Uganda in late May**

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**

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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

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.



Pharmacist Resources on G2C2



Genomic educational resources for pharmacists added to G2C2 website

The Genetics/Genomics Competency Center (G2C2), a free, online collection of materials for self-directed learning in genetics and genomics, now includes a new section on pharmacogenetics and pharmacogenomics. Geared specifically toward health care educators and practitioners, G2C2 was created in 2010 by NHGRI. [Read more](#)

Thanks To:

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

The screenshot shows the NHGRI website with the following elements:

- Header:** genome.gov National Human Genome Research Institute National Institutes of Health. Includes a Google search bar.
- Navigation:** Research Funding, Research at NHGRI, Health, Education, Issues in Genetics, Newsroom, Careers & Training, About, For You. Social media icons for Facebook, Twitter, and YouTube.
- Breadcrumbs:** 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
- Article Title:** Apply for NHGRI-ASHG's new education fellowship for genetics professionals
- Author:** By John Daniels, Assistant Public Affairs Specialist
- Image:** A logo for the National Human Genome Research Institute and The American Society of Human Genetics.
- Text:** 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.
- Section Header:** The 16-month program is divided into three parts:
- List-Group:**
 - 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.
- See Also:** [The Genetics and Education Fellowship \[ashp.org\]](#), [The Genetics and Public Policy Fellowship](#)

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

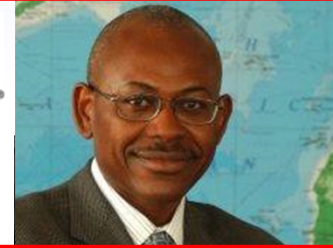
Federal Employee of the Year Award



NHGRI Intramural Research Highlights

Hypertension

Childhood Family Living Arrangements and Blood Pressure in Black Men



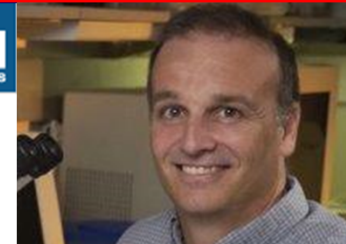
The genome of the ctenophore *Mnemiopsis leidyi* and its implications for cell type evolution

Science



Cell

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



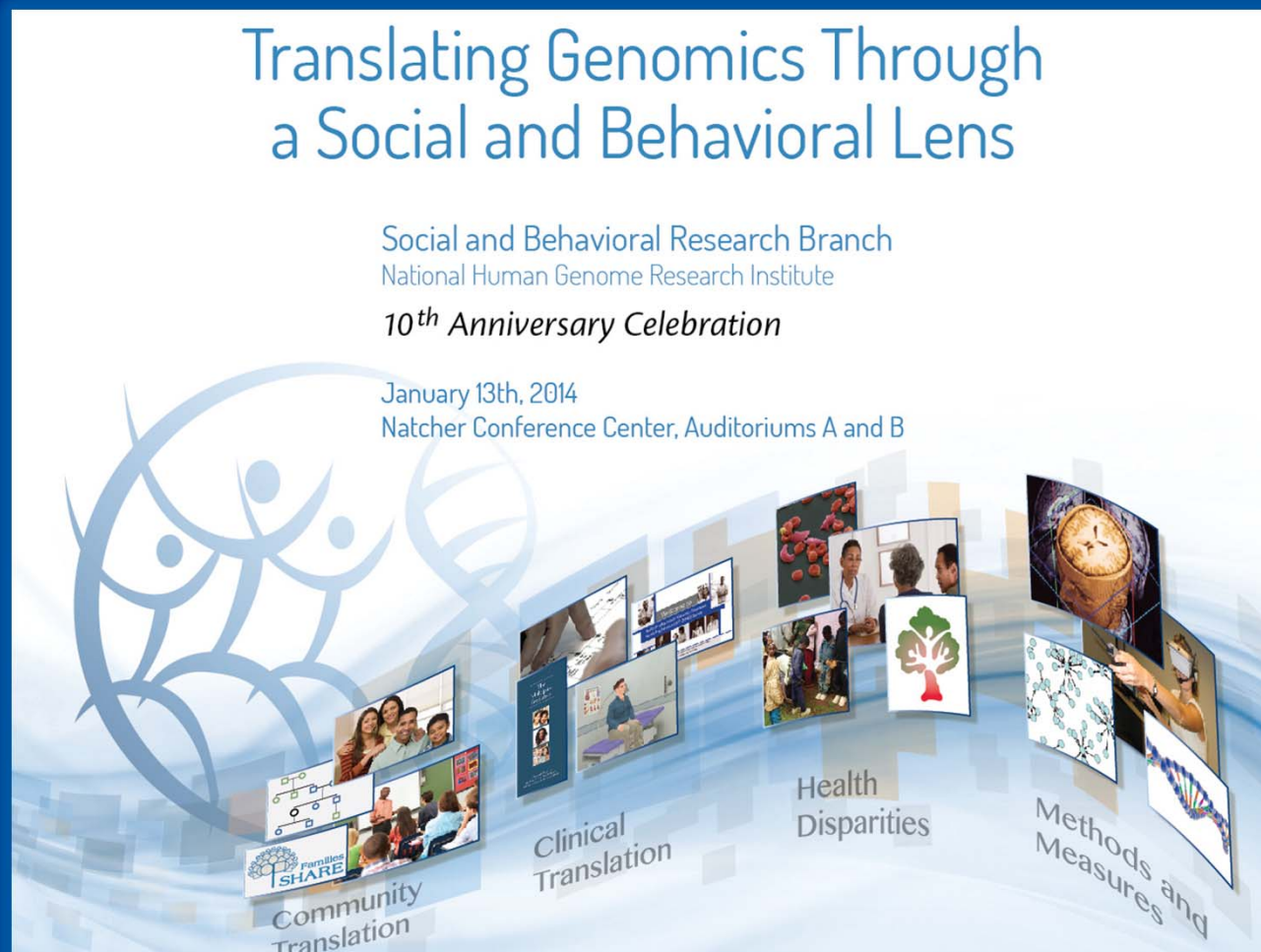
10th Anniversary of the NHGRI Social and Behavioral Research Branch

Translating Genomics Through a Social and Behavioral Lens

Social and Behavioral Research Branch
National Human Genome Research Institute

10th Anniversary Celebration

January 13th, 2014
Natcher Conference Center, Auditoriums A and B





genome.gov

National Human Genome Research Institute

National Institutes of Health



Special Thanks!



NATIONAL HUMAN GENOME RESEARCH INSTITUTE



***Advancing human health
through genomics research***