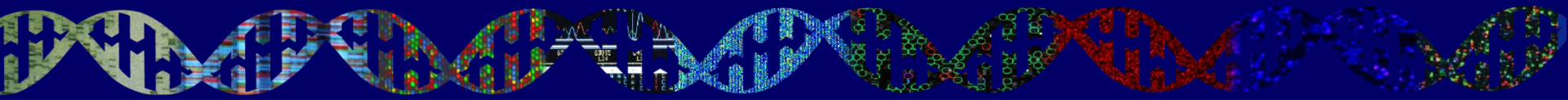
NIH National Human Genome Research Institute

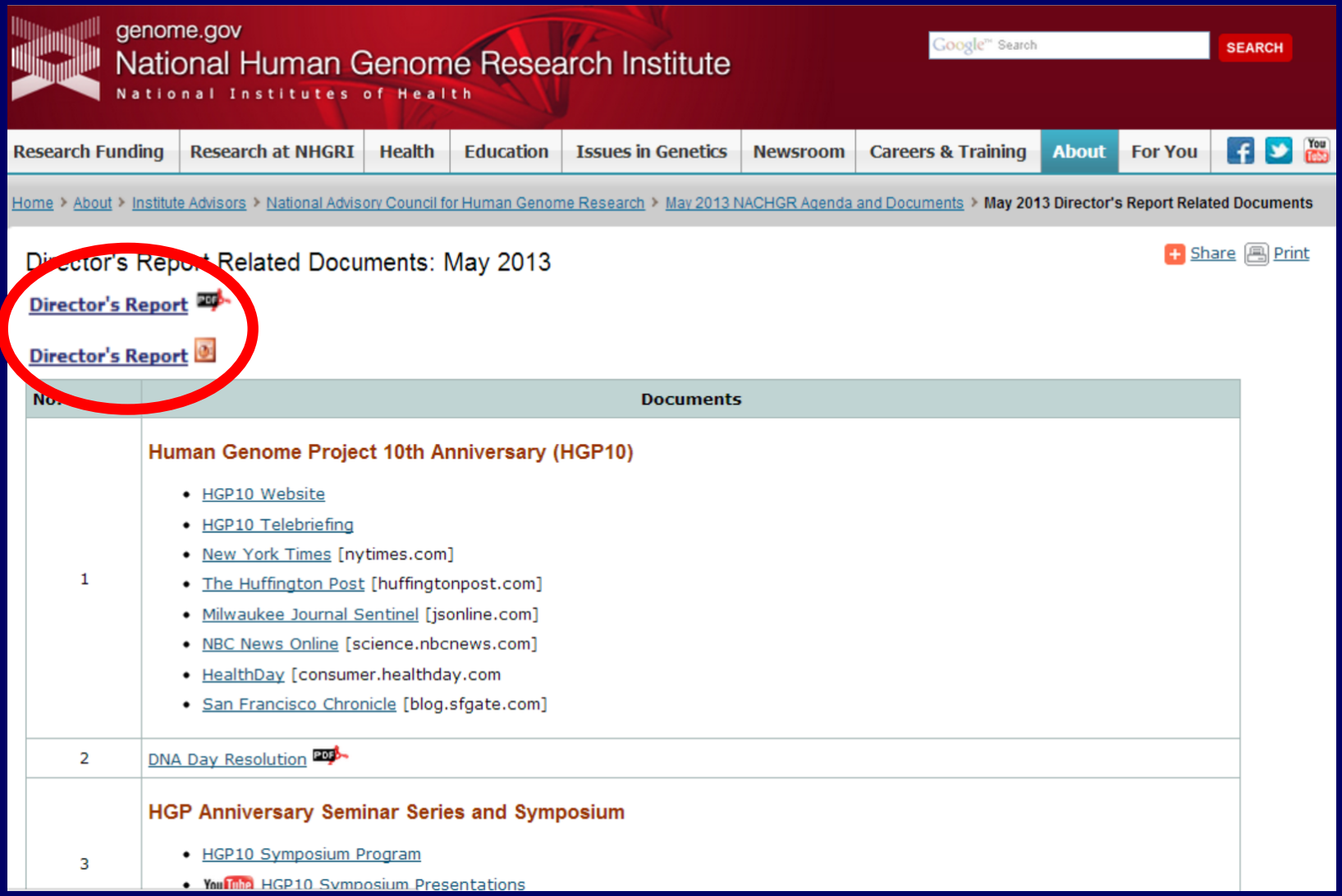
DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

May 2013

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





genome.gov/DirectorsReport

Document #

Open Session Presentations

NIH Center for Scientific Review

Richard Nakamura

ACMG Recommendations for Reporting Incidental Findings

Bob Nussbaum

Recent NHGRI Meetings:

- **Genomics and Society Working Group**

  Pamela Sankar

- **NHGRI Training and Career Development**

  Bettie Graham

Open Session Presentations

Program Update:

Genome Sequencing Program Update: Disease 2020

Adam Felsenfeld

Concept Clearance:

Interpreting Variants in Non-Coding Regions of the Genome

Lisa Brooks

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 Programs

VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

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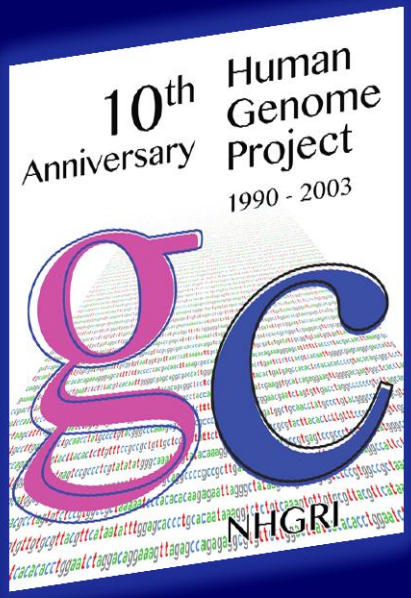
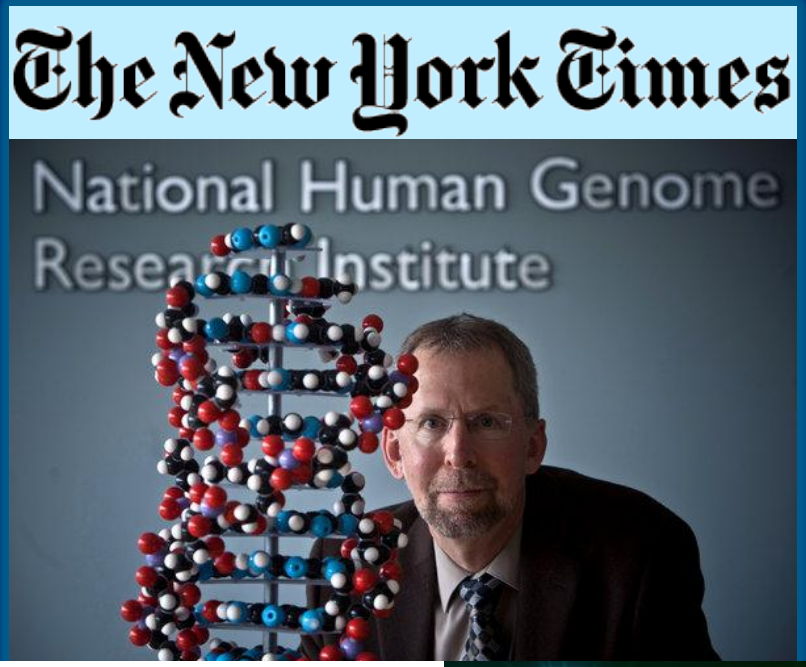
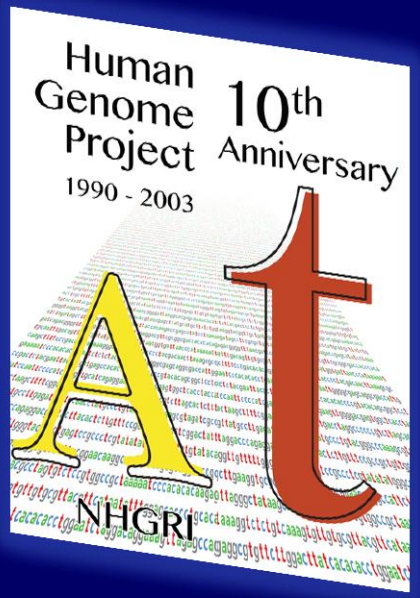
10th Anniversary of Human Genome Project







MILWAUKEE

Science on NBC NEWS

HealthDay®
News for Healthier Living

10 Years On, Still Much To Be Learned From Human Genome Map

Advances made in genetics of disease, but creating new drugs more complex than first thought

San Francisco Chronicle

science

« Who should have responsibility for naming exoplanets? | Main | Apollo-era NASA officials say climate change research 'corrupted' by politics and special interests »

Ten years ago: Human Genome Project completed

Document 1

Happy 10th Anniversary from U.S. Congress

113TH CONGRESS
1ST SESSION

H. RES. 180

Recognizing the sequencing of the human genome as one of the most significant scientific accomplishments of the past 100 years and expressing support for the designation of April 25, 2013, as "DNA Day".

IN THE HOUSE OF REPRESENTATIVES

APRIL 25, 2013

Ms. SLAUGHTER (for herself, Mr. BURGESS, Ms. SCHAKOWSKY, and Ms. SPEIER) submitted the following resolution; which was referred to the Committee on Energy and Commerce

---

RESOLUTION

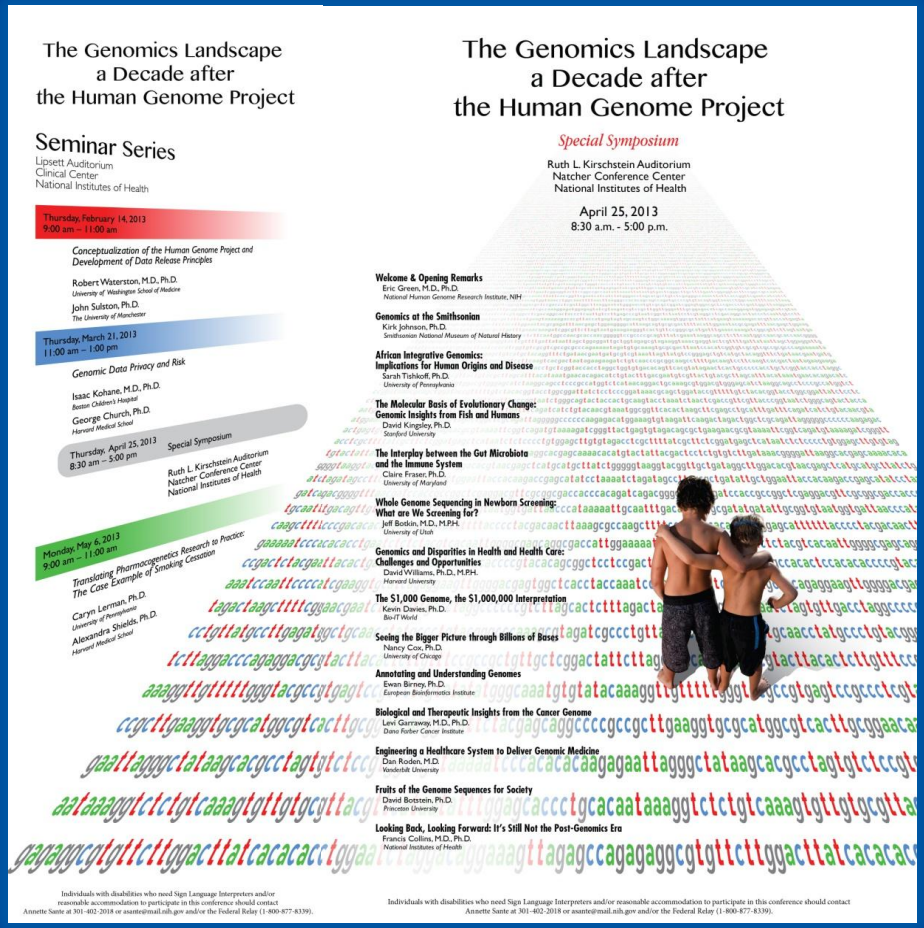
Recognizing the sequencing of the human genome as one of the most significant scientific accomplishments of the past 100 years and expressing support for the designation of April 25, 2013, as "DNA Day".

Whereas April 25, 2013, is the 60th anniversary of the publication of the description of the double-helical structure of deoxyribonucleic acid (DNA) in the scientific journal Nature by James D. Watson and Francis H.C. Crick, which is considered by many to be one of the most significant scientific discoveries of the 20th century;

Whereas their discovery launched a field of inquiry that explained how DNA encoded biological information and how this information is duplicated and passed from generation

Document 2

HGP 10th Anniversary Seminar Series and Symposium



The Genomics Landscape
a Decade after
the Human Genome Project

Seminar Series
Lipsett Auditorium
Clinical Center
National Institutes of Health

Thursday, February 14, 2013
9:00 am – 11:00 am
Conceptualization of the Human Genome Project and Development of Data Release Principles
Robert Waterston, M.D., Ph.D.
University of Washington School of Medicine
John Sulston, Ph.D.
The University of Manchester

Thursday, March 21, 2013
11:00 am – 1:00 pm
Genomic Data Privacy and Risk
Isaac Kohane, M.D., Ph.D.
Boston Children's Hospital
George Church, Ph.D.
Harvard Medical School

Thursday, April 25, 2013
8:30 am – 5:00 pm
Special Symposium
Ruth L. Kirschstein Auditorium
Natcher Conference Center
National Institutes of Health

Monday, May 6, 2013
9:00 am – 11:00 am
Translating Pharmacogenetics Research to Practice: The Case Example of Smoking Cessation
Caryn Lerman, Ph.D.
University of Pennsylvania
Alexandra Shields, Ph.D.
Harvard Medical School

The Genomics Landscape
a Decade after
the Human Genome Project

Special Symposium

Ruth L. Kirschstein Auditorium
Natcher Conference Center
National Institutes of Health

April 25, 2013
8:30 a.m. - 5:00 p.m.

Welcome & Opening Remarks
Eric Green, M.D., Ph.D.
National Human Genome Research Institute, NIH

Genomics at the Smithsonian
Kirk Johnson, Ph.D.
Smithsonian National Museum of Natural History

African Integrative Genomics: Implications for Human Origins and Disease
Sarah Tishkoff, Ph.D.
University of Pennsylvania

The Molecular Basis of Evolutionary Change: Genomic Insights from Fish and Humans
David Kingsley, Ph.D.
Stanford University

The Interplay between the Gut Microbiota and the Immune System
Claire Fraser, Ph.D.
University of Maryland

Whole Genome Sequencing in Newborn Screening: What are We Screening for?
Jeff Botkin, M.D., M.P.H.
University of Utah

Genomics and Disparities in Health and Health Care: Challenges and Opportunities
David Williams, Ph.D., M.P.H.
Harvard University

The $1,000 Genome, the $1,000,000 Interpretation
Kevin Davies, Ph.D.
Bio-IT World

Seeing the Bigger Picture through Billions of Bases
Nancy Cox, Ph.D.
University of Chicago

Annotating and Understanding Genomes
Ewan Birney, Ph.D.
European Bioinformatics Institute

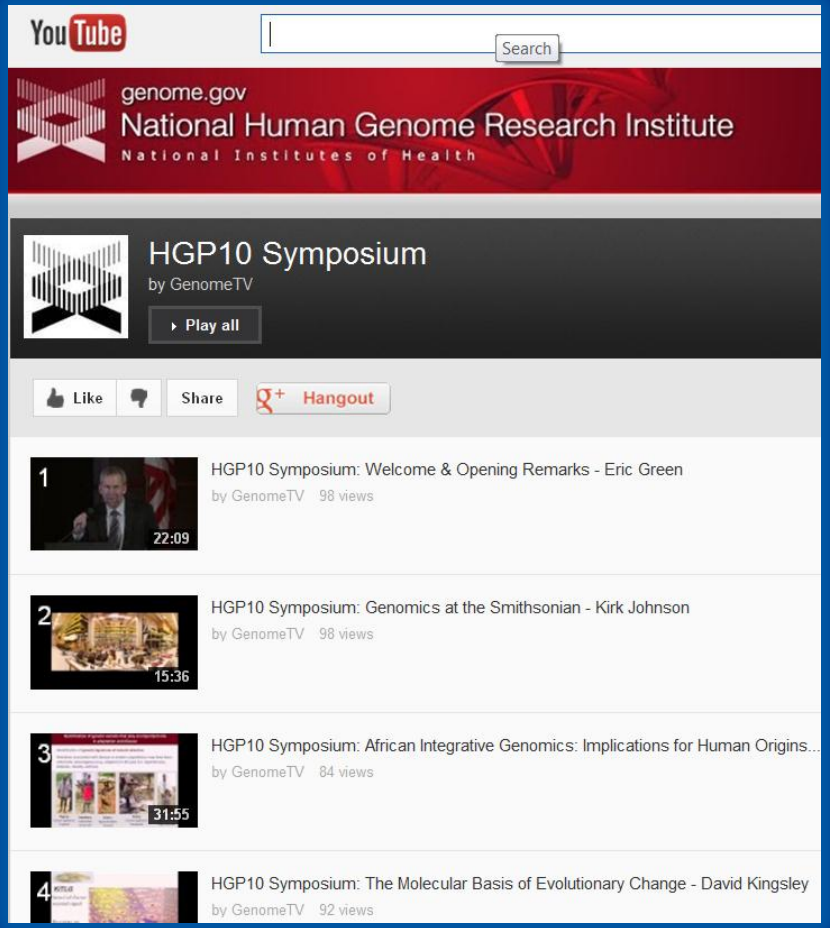
Biological and Therapeutic Insights from the Cancer Genome
Levi Garraway, M.D., Ph.D.
Dana Farber Cancer Institute

Engineering a Healthcare System to Deliver Genomic Medicine
Dan Roden, M.D.
Vanderbilt University

Fruits of the Genome Sequences for Society
David Botstein, Ph.D.
Princeton University

Looking Back, Looking Forward: It's Still Not the Post-Genomics Era
Francis Collins, M.D., Ph.D.
National Institutes of Health

Individuals with disabilities who need Sign Language Interpreters and/or reasonable accommodation to participate in this conference should contact Annette Sante at 301-402-2018 or asante@mail.nih.gov and/or the Federal Relay (1-800-877-8339).



YouTube Search

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

HGP10 Symposium
by GenomeTV
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Like Share Hangout

1 — 22:09 — HGP10 Symposium: Welcome & Opening Remarks - Eric Green
by GenomeTV 98 views

2 — 15:36 — HGP10 Symposium: Genomics at the Smithsonian - Kirk Johnson
by GenomeTV 98 views

3 — 31:55 — HGP10 Symposium: African Integrative Genomics: Implications for Human Origins...
by GenomeTV 84 views

4 — HGP10 Symposium: The Molecular Basis of Evolutionary Change - David Kingsley
by GenomeTV 92 views

Document 3

Smithsonian Exhibition: June 2013 Opening

Smithsonian
National Museum of Natural History

GENOME
UNLOCKING LIFE'S CODE

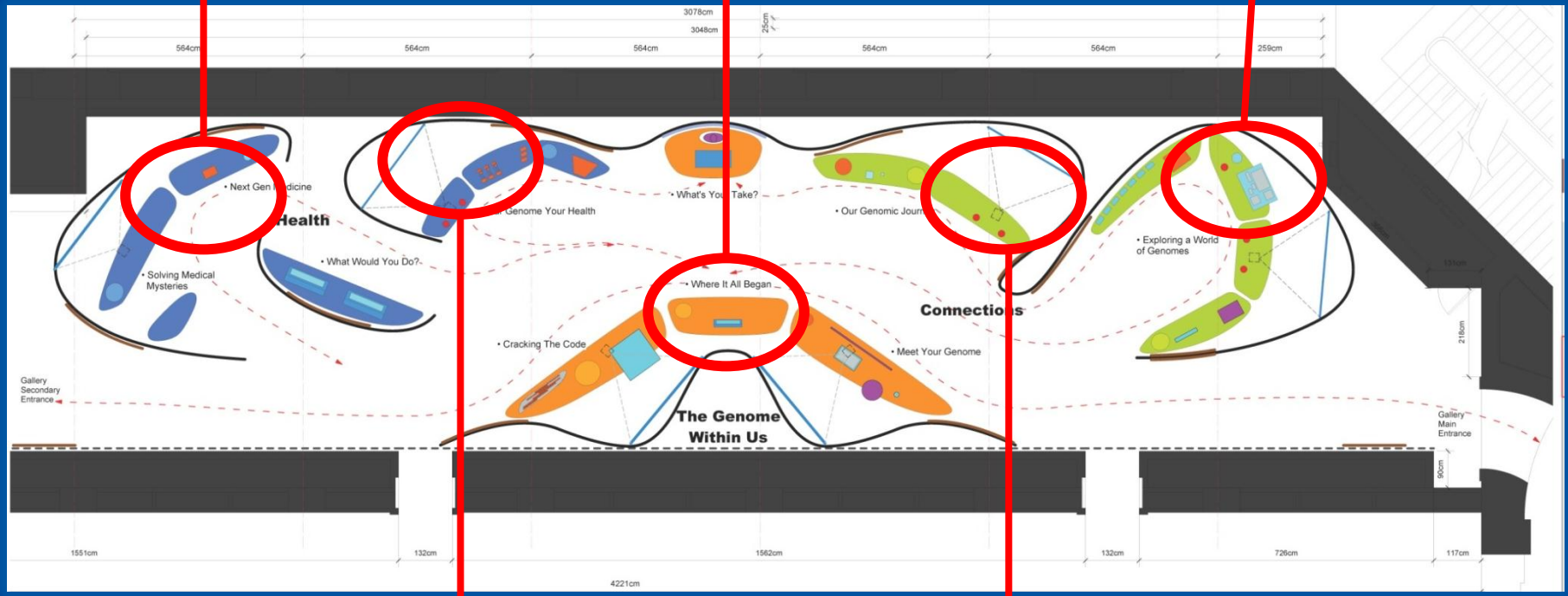
- ~2900 square foot exhibition
- Significant associated programming and outreach
- Resident in NMNH for ~1 year
- Subsequently will tour North America for 4-5 years

Smithsonian Exhibition: Layout By Theme

Next Gen Medicine

Natural World

Genome Within



Your Genome, Your Health

Genomic Journey

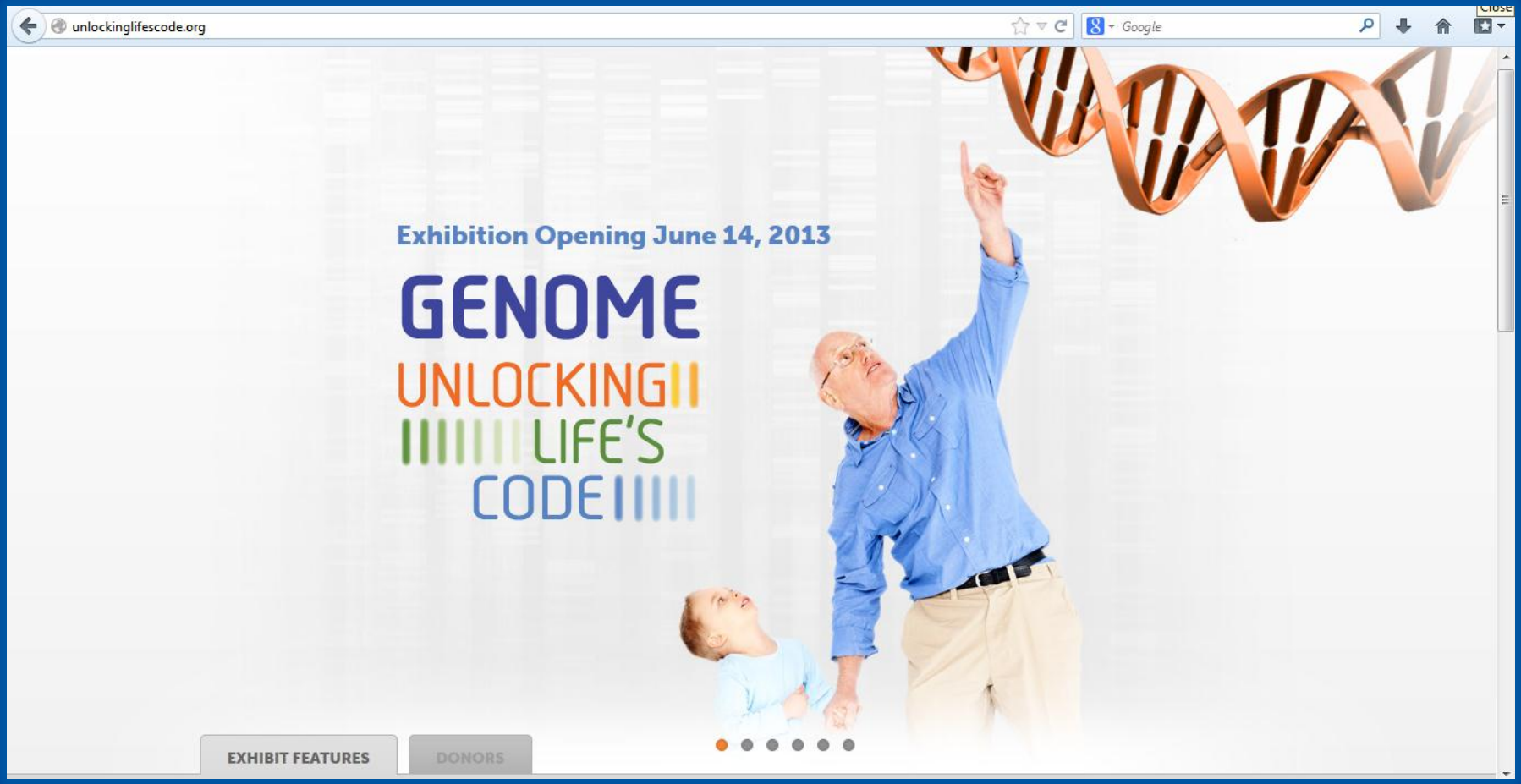
Document 4

Smithsonian Exhibition: Public Programs

- Ancestry and genetic testing
- Genomic themes in plays
- Consumer-based testing
- Debate on an ethical, legal, and social question
- Evolutionary genetics
- Genomics in pop culture

Document 4

Smithsonian Exhibition: Website



www.unlockinglifescode.org

Document 4

Acting Executive Officer at NHGRI

Ellen Rolfes, M.A.

NHGRI Recruitments

Director,
Division of Genomics and Society

Extramural Bioinformatics Program Directors

Physicians (Division of Genomic Medicine & Division of Policy, Communications, and Education)

Document 5

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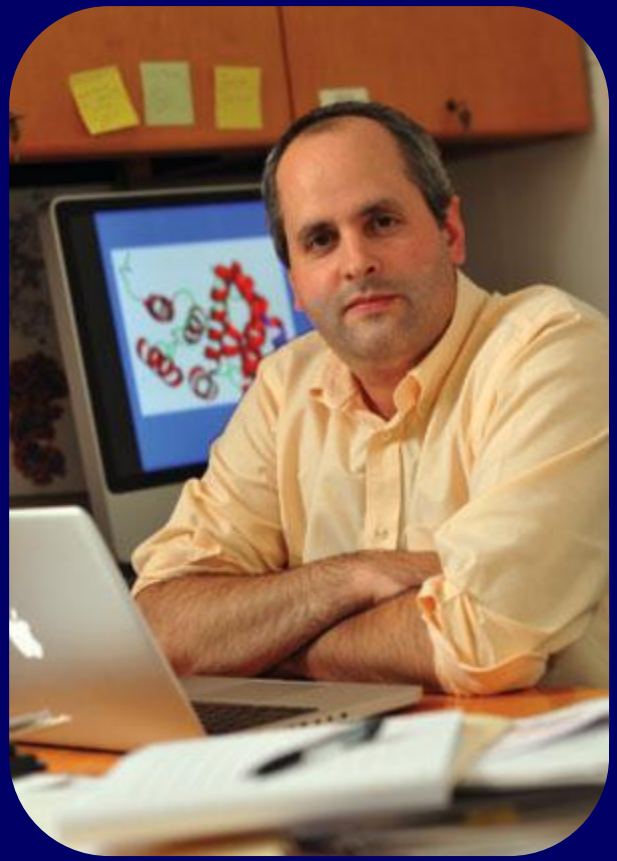
New Director, National Institute of General Medical Sciences

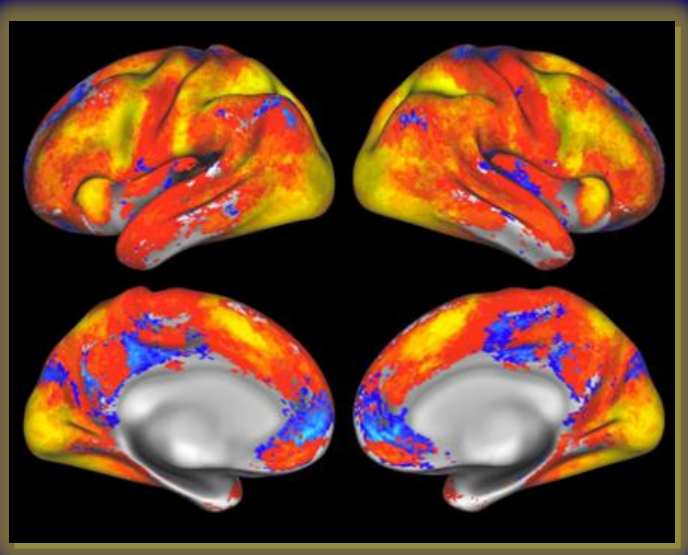


NIGMS

Jon Lorsch, Ph.D.

Document 6

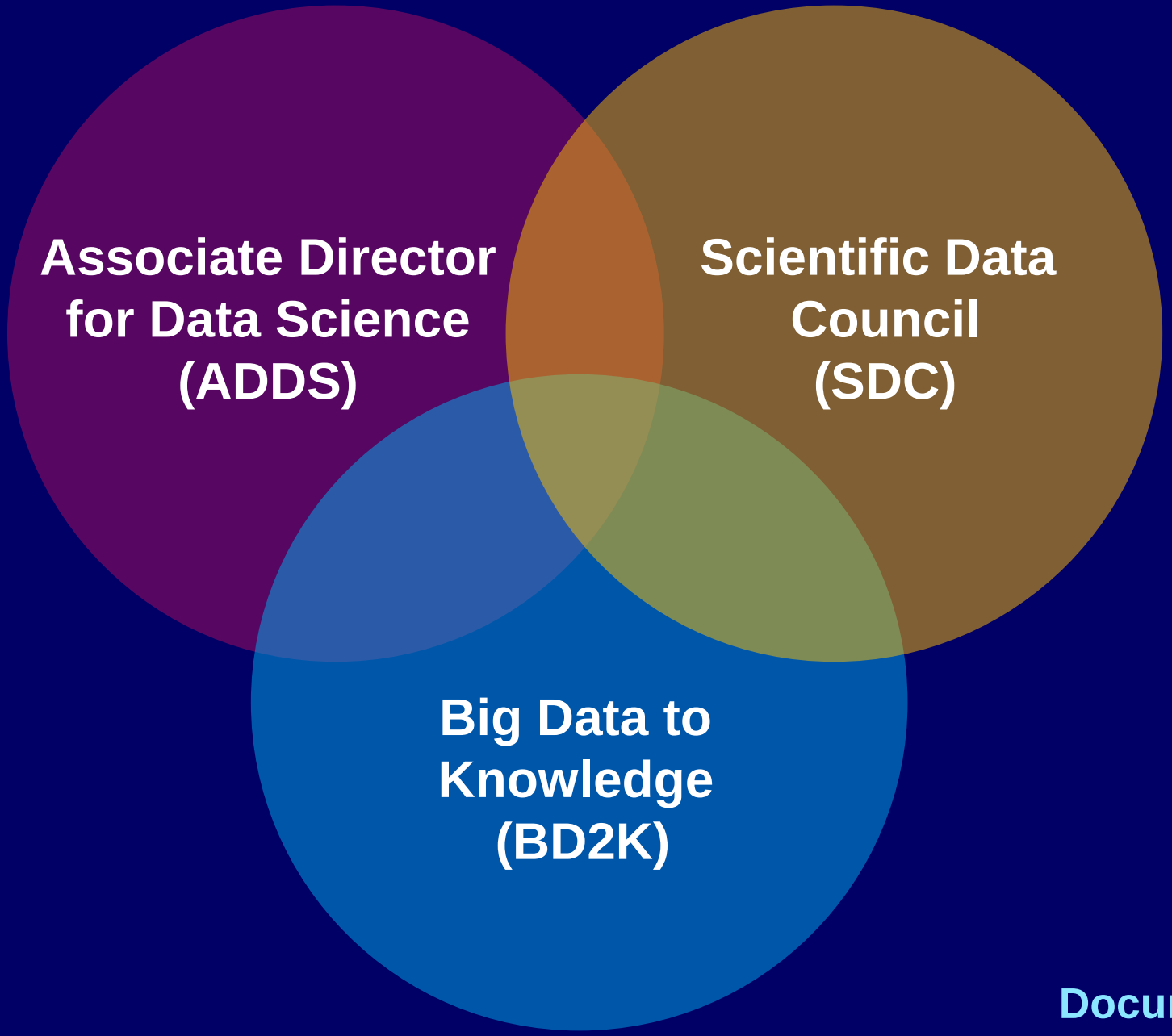
Brain Research through Advancing Innovative Neurotechnologies (BRAIN) Initiative



- $40M from the NIH to develop new tools, training opportunities, and other resources
- NIH Working Group co-chaired by Cori Bargmann and William Newsome

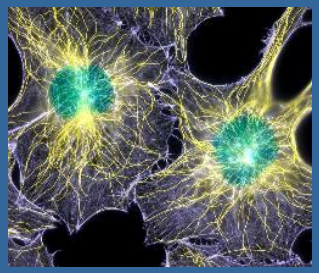
Document 7

NIH is Tackling the 'Big Data' Problem



Associate Director for Data Science
Office of the Director, National Institutes of Health, Department of Health and Human Services



The NIH is the center of medical and behavioral research for the Nation
----making essential medical discoveries that improve health and save lives.

Are you a top-level Scientific Researcher or Scientific Administrator seeking a career at the one of the preeminent biomedical research institutions in the Nation and the world? Are you at that point in your career where you're ready to "give back?" The position of Associate Director for Data Science (ADDS), Office of the Director (OD), National Institutes of Health (NIH), offers a unique and exciting opportunity to provide critical leadership for basic and translational research. The era of "Big Data" has arrived for the biomedical sciences. There is an urgent need and, with it, spectacular opportunities for NIH to enhance its programs in data science, such as those involving data emanating from different sources (e.g., genomics, imaging, and phenotypic information from electronic health records). The ADDS provides a vision for the utilization and extraction of knowledge from the data generated by, and relevant to, NIH research, and advises experts throughout the agency on a variety of complex, unique, and/or sensitive situations and issues in data science to ensure continual achievement of NIH's dynamic biomedical research mission.

We are looking for applicants with senior-level experience who have a commitment to excellence and the energy, enthusiasm, and innovative thinking necessary to lead a dynamic and diverse organization.

The successful candidate for this position will be appointed at a salary commensurate with his/her qualifications. Full Federal benefits will be provided including leave, health and life insurance, long-term care insurance, retirement, and savings plan (401k equivalent).

If you are ready for an exciting leadership opportunity, please see the detailed vacancy announcement at **http://www.jobs.nih.gov** ***(under Executive Careers).*** **Applications will be reviewed starting May 13, 2013, and will be accepted until the position is filled.**

THE NATIONAL INSTITUTES OF HEALTH AND THE DEPARTMENT OF HEALTH AND HUMAN SERVICES ARE EQUAL OPPORTUNITY EMPLOYERS

NIH

Big Data to Knowledge (BD2K): Overview

- **Trans-NIH effort with the overarching goal of:**

  By the end of the decade, enable a quantum leap in the ability of the research community to maximize the value of the growing volume and complexity of biomedical data

- **Strong support across NIH**

  - **Working group has about 125 members**
  - **Staff from 24 Institutes/Centers and several other offices involved**

BD2K: Four Programmatic Areas

I. Facilitating Broad Use of Biomedical Big Data

II. Developing and Disseminating Analysis Methods and Software for Biomedical Big Data

III. Enhancing Training for Biomedical Big Data

IV. Establishing Centers of Excellence for Biomedical Big Data

BD2K: Update

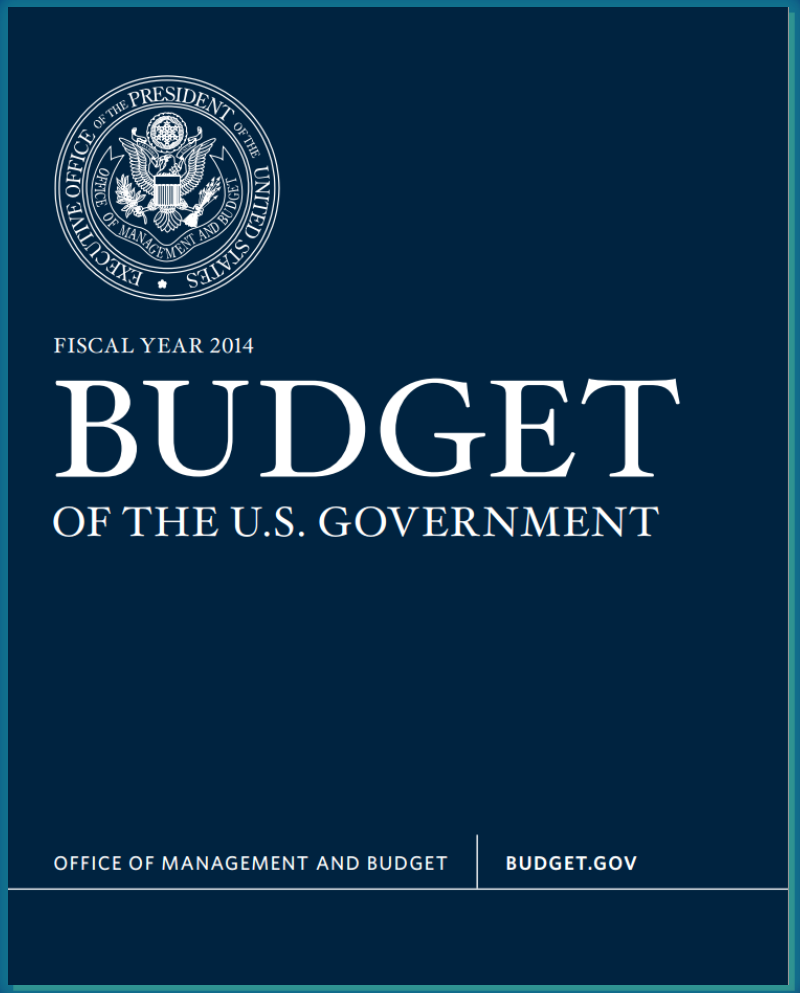
- **Timeline:**
  - Series of workshops, beginning this summer
  - Funding starts in Fiscal Year 2014
- **Unique funding model**

	FY14	FY15	FY16
Requested:	$64M	$96M	$109M
Available:	$27M	$80M	$99M

Fiscal Year 2013 Funding Finalized

- Federal government funded through continuing resolution for Fiscal Year 2013
- Sequester resulted in a 5.1% reduction
- Total NIH reduction of 5.8%
- NHGRI's final Fiscal Year 2013 budget: $483M

Fiscal Year 2014 Appropriations



NIH: $31.3 billion

NHGRI: $517 million

Document 9

Congressional Delegation Visits NIH

Reorganizing STEM Education

Prepare Students for STEM Careers in the 21st Century Economy. Our future competitiveness demands that we move American students from the middle or bottom to the top of the pack in science and mathematics…The Budget proposes a comprehensive reorganization of Federal STEM education programs to enable more strategic investment in STEM and more critical evaluation of outcomes… *(page 22)*

Document 10

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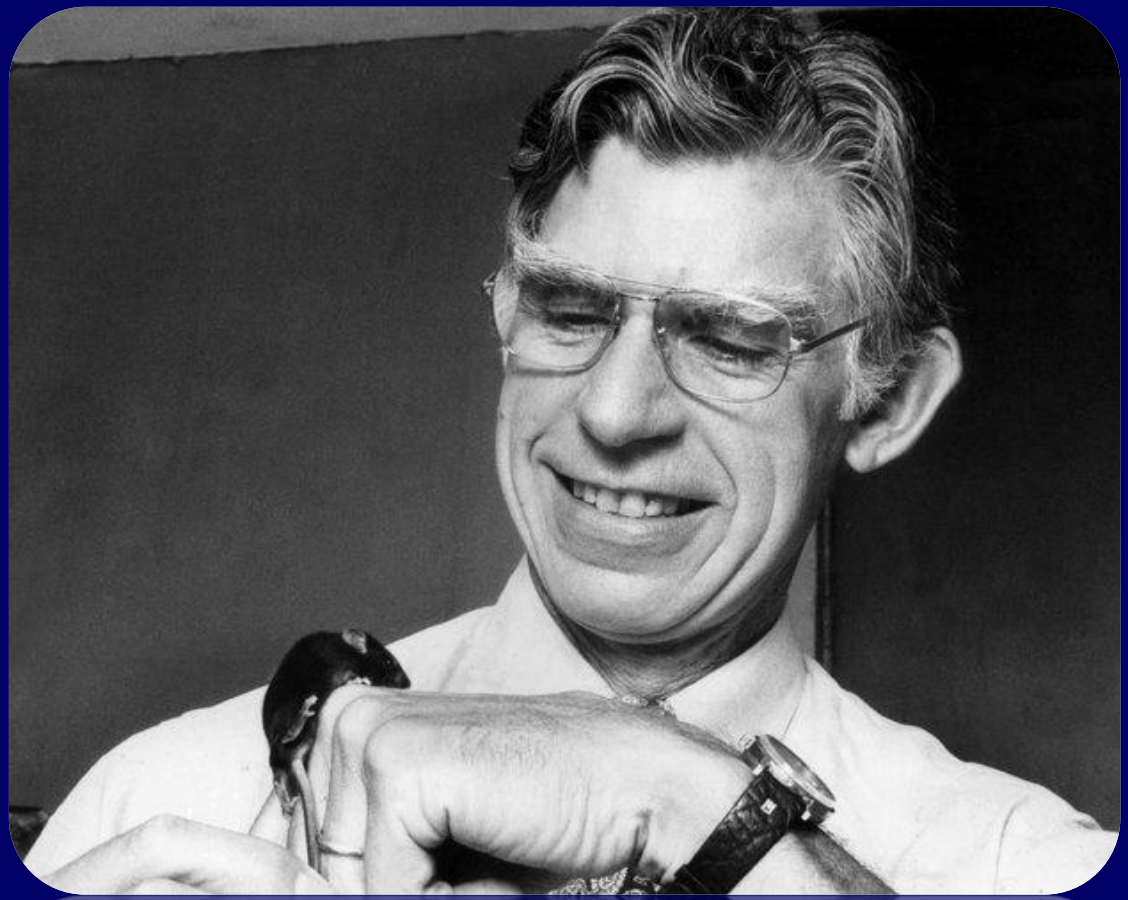
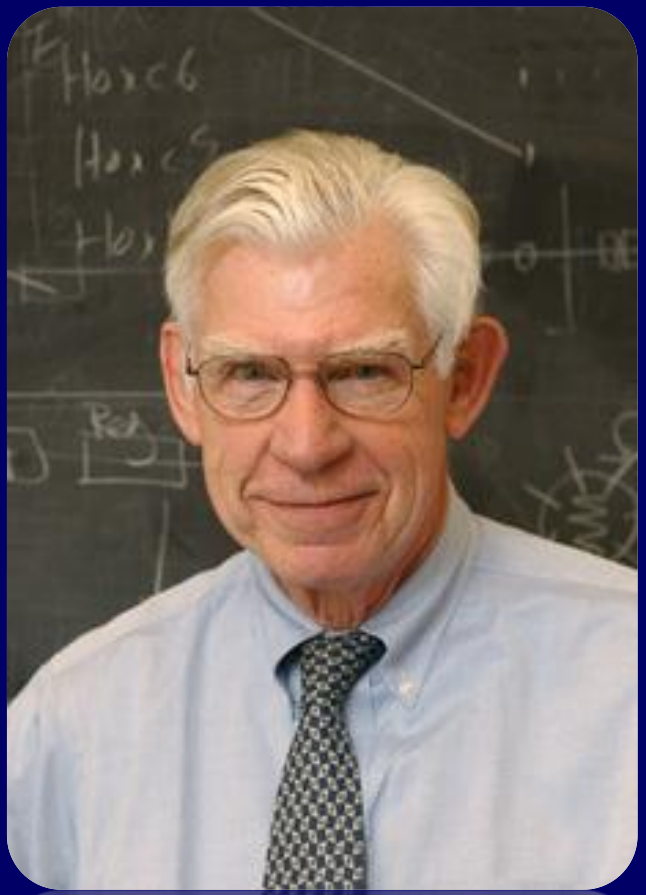
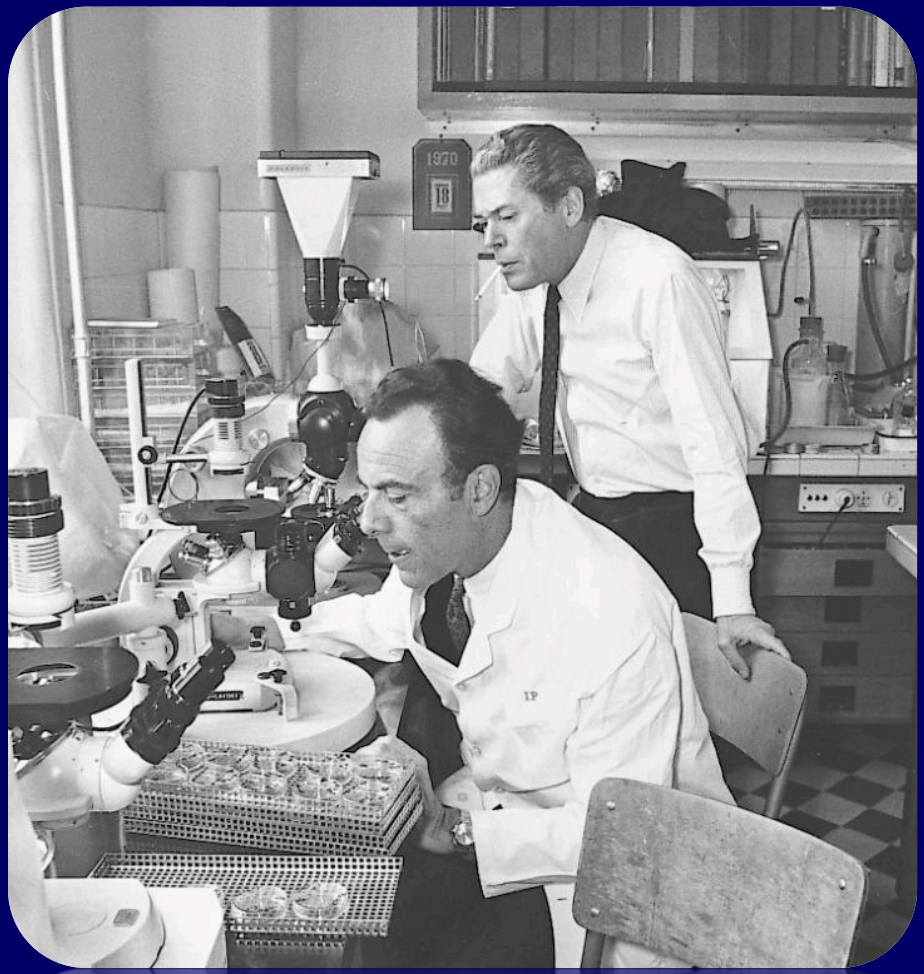
Mourning the Loss of Frank Ruddle





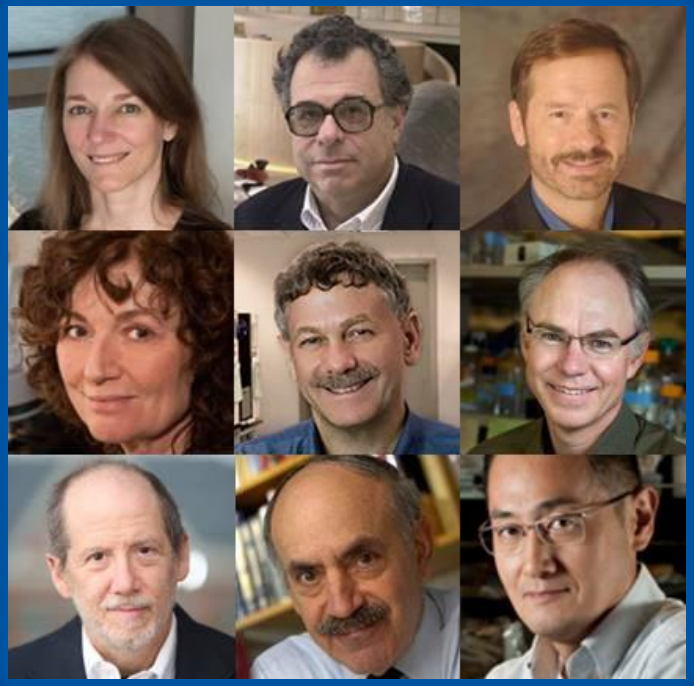
Document 11

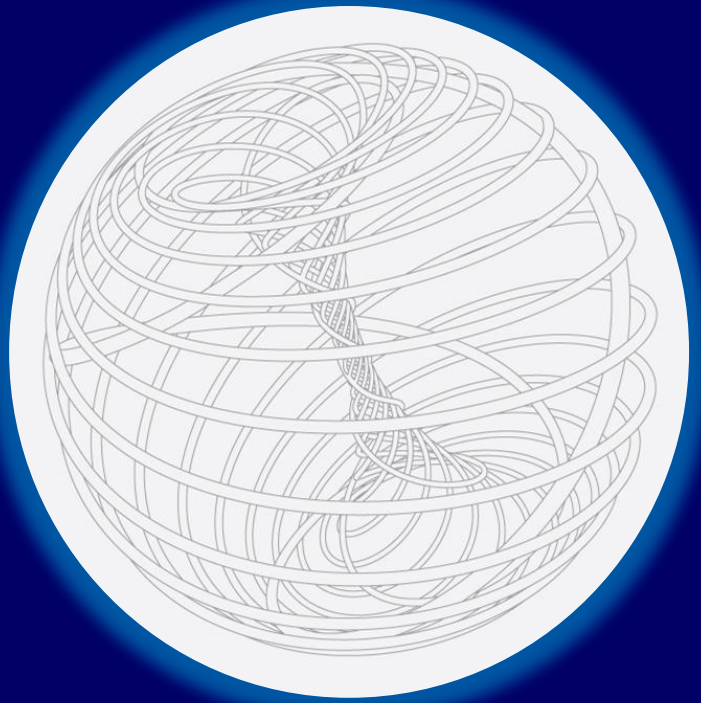
Mourning the Loss of François Jacob



Document 12

Breakthrough Prize in Life Sciences





David Botstein
Titia de Lange
Eric Lander

Charles Sawyers
Bert Vogelstein

Document 13

Newly Elected: National Academy of Sciences

Jef Boeke
Marcus Feldman
Michel Georges
Mary Lidstrom
Norbert Perrimon
Stephen Quake
Lou Staudt
Hunt Willard
Fred Winston

Document 14

Newly Elected: American Academy of Arts and Sciences

- David Altshuler
- Martin Blaser
- Eugene Koonin
- John Lis
- Jim Lupski
- Jonathan Pritchard

New Investigators: HHMI

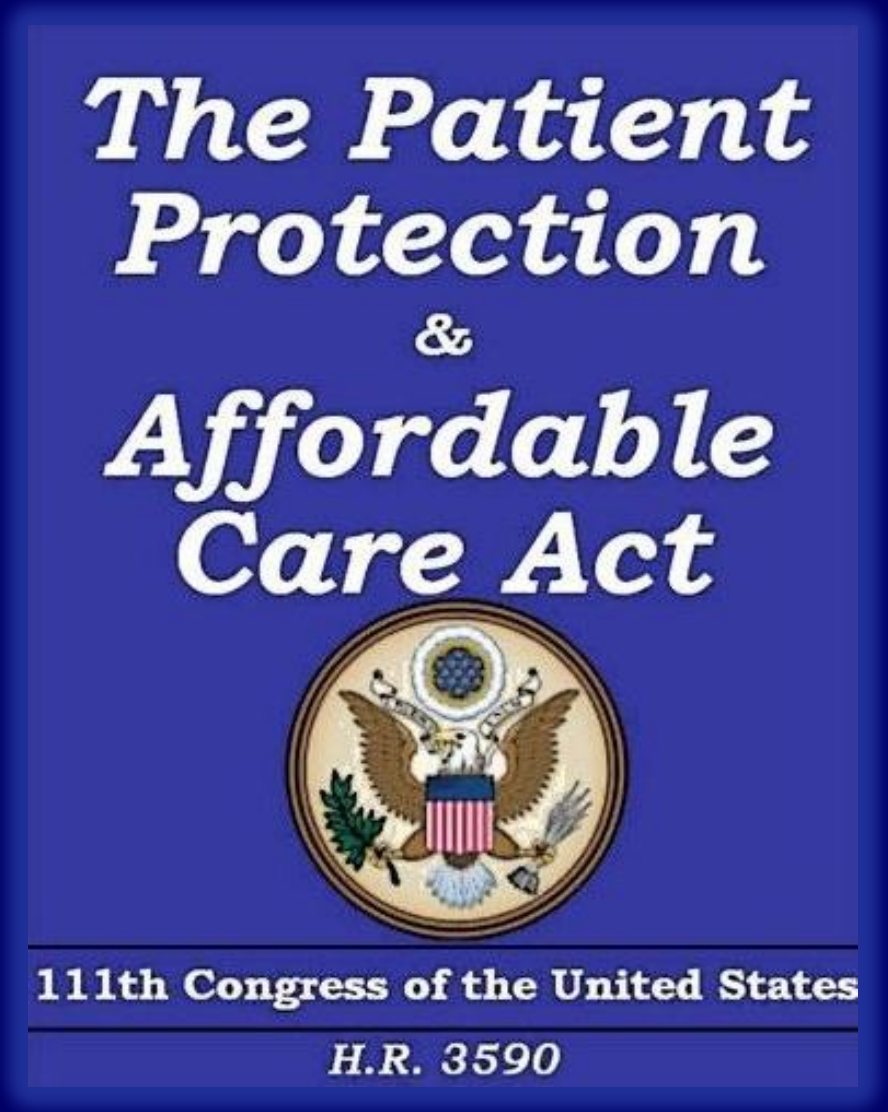
- Chuan He
- Vamsi Mootha
- Aviv Regev
- David Reich

Gene Patents and the Courts

MYRIAD®

Document 17

Affordable Care Act Covers *BRCA* Testing



Document 18

ACMG/AAP Statement on Testing and Screening in Children

Ethical and Policy Issues in Genetic Testing and Screening of Children

COMMITTEE ON BIOETHICS, COMMITTEE ON GENETICS, THE AMERICAN COLLEGE OF MEDICAL GENETICS, GENOMICS SOCIAL, ETHICAL and LEGAL ISSUES COMMITTEE

Pediatrics; originally published online February 21, 2013;
DOI: 10.1542/peds.2012-3680

Document 19

ACMG Recommendations for Clinical Genomic Incidental Findings

American College of Medical Genetics and Genomics

ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing

Robert C. Green, MD, MPH[1,2], Jonathan S. Berg, MD, PhD[3], Wayne W. Grody, MD, PhD[4-6], Sarah S. Kalia, ScM, CGC[1], Bruce R. Korf, MD, PhD[7], Christa L. Martin, PhD, FACMG[8], Amy McGuire, JD, PhD[9], Robert L. Nussbaum, MD[10], Julianne M. O Daniel, MS, CGC[11], Kelly E. Ormond, MS, CGC[12], Heidi L. Rehm, PhD, FACMG[2,13], Michael S. Watson, MS, PhD, FACMG[14], Marc S. Williams, MD, FACMG[15], Leslie G. Biesecker, MD[16]

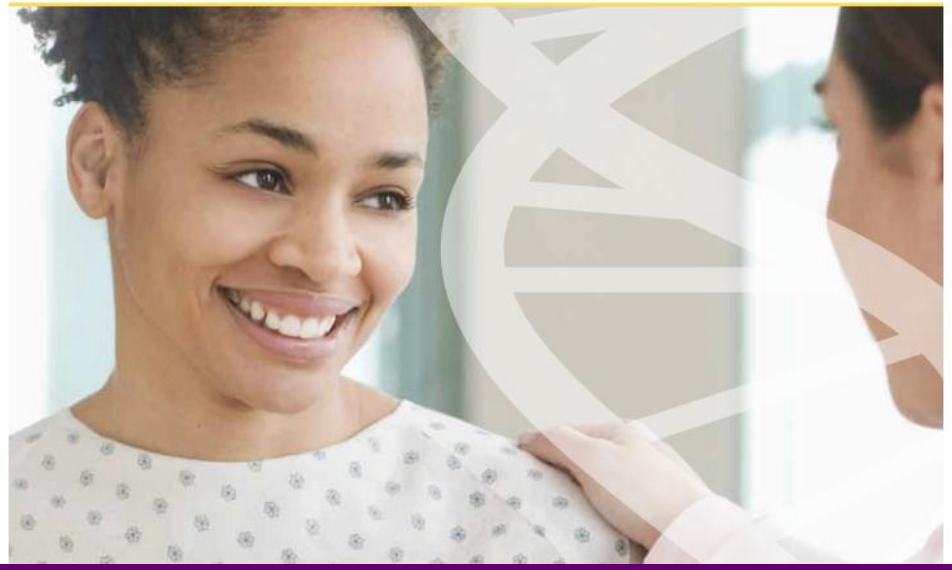
[1]Division of Genetics, Department of Medicine, Brigham and Women s Hospital and Harvard Medical School, Boston, Massachusetts, USA; [2]Partners Healthcare Center for Personalized Genetic Medicine, Boston, Massachusetts, USA; [3]Department of Genetics, University of North Carolina at Chapel Hill School of Medicine, Chapel Hill, North Carolina, USA; [4]Division of Medical Genetics, Department of Human Genetics, UCLA School of Medicine, Los Angeles, California, USA; [5]Division of Molecular Pathology, Department of Pathology & Laboratory Medicine, UCLA School of Medicine, Los Angeles, California, USA; [6]Division of Pediatric Genetics, Department of Pediatrics, UCLA School of Medicine, Los Angeles, California, USA; [7]Department of Genetics, University of Alabama, Birmingham, Alabama, USA; [8]Department of Human Genetics, Emory University School of Medicine, Atlanta, Georgia, USA; [9]Center for Medical Ethics and Health Policy, Baylor College of Medicine, Houston, Texas, USA; [10]Division of Genomic Medicine, Department of Medicine, and Institute for Human Genetics, University of California, San Francisco, San Francisco, California, USA; [11]Illumina, Inc., San Diego, California, USA; [12]Department of Genetics, Stanford University, Stanford, California, USA; [13]Department of Pathology, Brigham and Women s Hospital and Harvard Medical School, Boston, Massachusetts, USA; [14]American College of Medical Genetics and Genomics, Bethesda, Maryland, USA; [15]Genomic Medicine Institute, Geisinger Health System, Danville, Pennsylvania, USA; [16]National Human Genome Research Institute, National Institutes of Health, Bethesda, Maryland, USA

Document 20

AMA on Personalized Medicine

AMA
AMERICAN MEDICAL ASSOCIATION

Policy perspective on
personalized medicine



Document 21

New Journal:
Molecular Genetics & Genomic Medicine

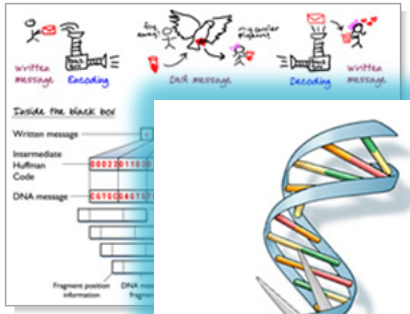
Max Muenke, M.D.

WILEY ONLINE LIBRARY

NHGRI Genome Advance of the Month

Fitting the National Archives in your pocket

By Joy Yang
Post-baccalaureate Fellow



This Genome Advance of the Month features an article that began as a somewhat facetious idea to address the issue of where to store big data.

Nick Goldman, Ph.D., and Ewan Birney, Ph.D., researchers at the European Bioinformatics Institute in Hinxton, UK,

DNA encoding. Vie

Editing the book of life with molecular scissors

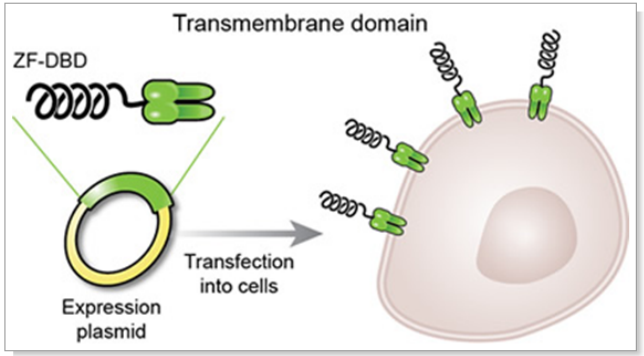
By Andrea Ramirez, M.D., M.S.
Clinical Fellow, NHGRI

Life's code is written in A's (adenine), T's (thymine), C's (cytosine) and G's (guanine), the letters representing the four nucleotides within the deoxyribonucleic acid (DNA) that direct the action of a cell from its nucleus. Three billion of these letters paired in two strands spell out the human genome sequence, a code scientists study every day looking for the causes of disease.

segment of le
Ph.D., worke
research fello
NHGRI Center
type of molec

Grabbing hold of cells and tissues with zinc fingers

By Ian L. Marpuri
NHGRI Scientific Program Analyst



The expression of zinc fingers on the surface of the cell. On the left is DNA engineered to express zinc finger DNA-binding domains (ZF-DBD). On the right are zinc fingers expressed on a cell's surface.

A uniquely named molecule called the zinc finger has frequently found itself in the news over the last few years. Zinc fingers are proteins found in human and animal cells that use zinc atoms to maintain their namesake "finger" shape. They bind to specific DNA sequences, which make zinc fingers ideal for targeting genes and other sections of the genome. They have become a potential tool in certain disease therapies because they could target defective copies of genes.

This month's Genome Advance of the Month describes new uses for zinc fingers to improve researchers' ability to study the processes of single cells and interactions between larger groups of cells.

Prashant Mali, Ph.D., and John Aach, Ph.D., both from the lab of George Church, Ph.D., at Harvard Medical School studied the interactions of zinc fingers and DNA. They noticed that both zinc fingers and DNA are highly customizable, creating an endless number of combinations between them. This led them to explore the use of zinc fingers to create structures using cells and other large molecules.

Genomics In The News…

"Every dollar we invested to map the human genome returned $140 to our economy -- every dollar."

– President Obama, February 2013

Document 24

Genomics In The News…

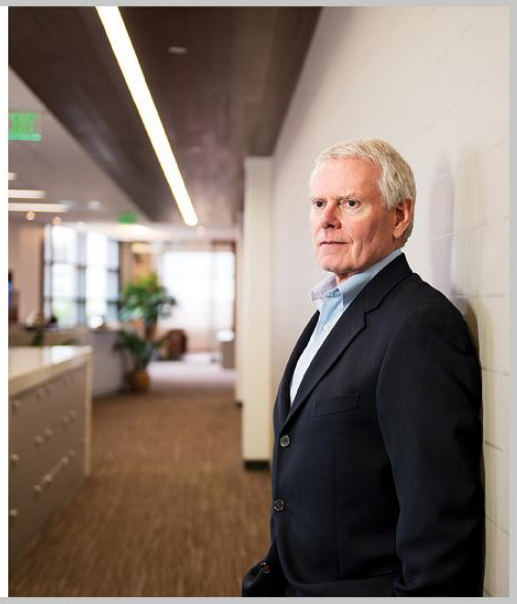
MIT Technology Review

10 BREAKTHROUGH TECHNOLOGIES 2013

Introduction | The 10 Technologies | Past Years

Prenatal DNA Sequencing

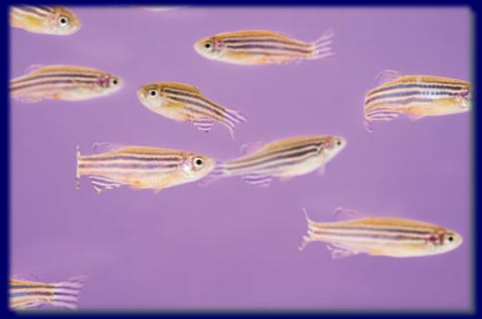
Reading the DNA of fetuses is the next frontier of the genome revolution. Do you really want to know the genetic destiny of your unborn child?



The Executive: *Illumina CEO Jay Flatley is looking to pregnancy as a new market for DNA sequencing.*

Document 24

Genomics In The News…



Document 24

Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

IV. NHGRI Extramural Research Program

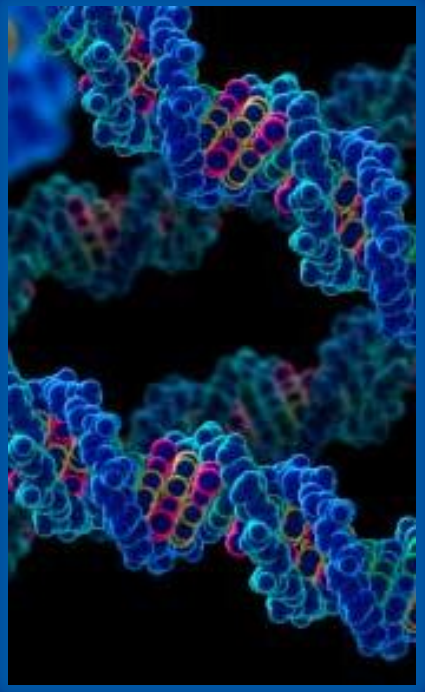
V. NIH Common Fund Programs

VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

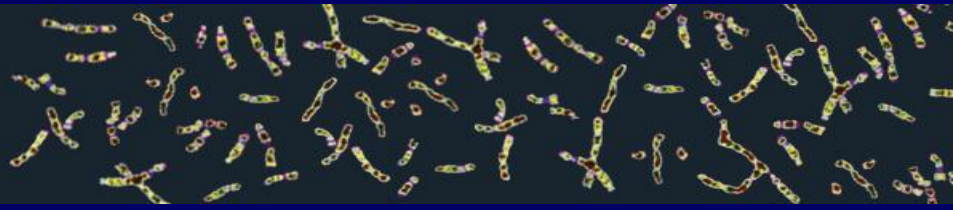
Large-Scale Genome Sequencing and Analysis Centers

- 101 Tb generated in last quarter
- >200 ongoing projects (e.g., cancer, complex disease, rare diseases, and comparative genomics)
- >25 papers published or in press this quarter



1000 Genomes

A Deep Catalog of Human Genetic Variation



- **Sequencing is now complete!**

  26 populations

  2683 samples: low-coverage whole-genome
  2658 samples: whole-exome
  2642 samples: whole-exome & low-coverage whole-genome
  ~465 samples: deep Complete Genomics data

- **1000 Genomes analysis meeting at CSHL**
- **Next 1000 Genomes analysis meeting prior to ASHG annual meeting**

- **TCGA meeting in early May**

- **Recent publications:**

  Acute Myeloid Leukemia

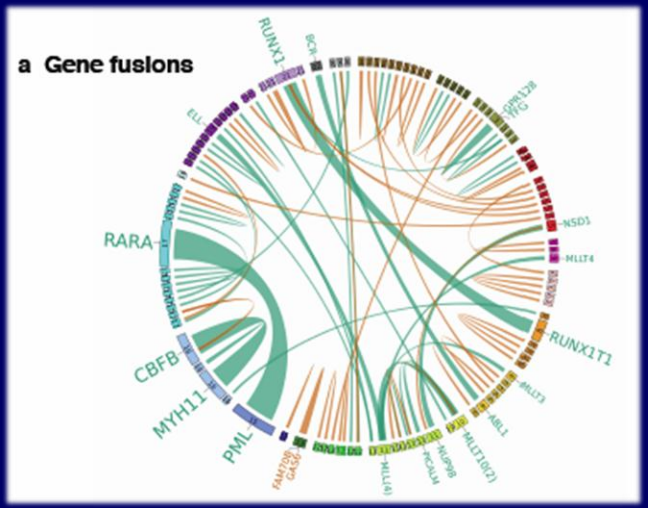
  (*NEJM*, 2013)

  Endometrial Carcinoma

  (*Nature*, 2013)

  Clear Cell Renal Cell Carcinoma

  (*In press*)



AML gene fusions: in-frame shown in green; out-of-frame in orange.

Centers for Mendelian Genomics

Finding the genes underlying human Mendelian conditions

Disease Gene Discovery:

- **>9000 whole-exome sequences have entered pipelines for studying 526 Mendelian disorders of all major organ systems**
- **64 disease genes and 116 candidate disease genes**

Collaborations and Outreach:

- **323 investigators, 189 institutions, 30 countries**
- **More than 100 presentations**

First Face-to-Face Meeting:

- **New working group on data analysis**
- **Potential collaborations**

International Rare Diseases Research Consortium

Goals by 2020:

- Diagnostic tests for most rare disorders
- New treatments for 200 rare disorders

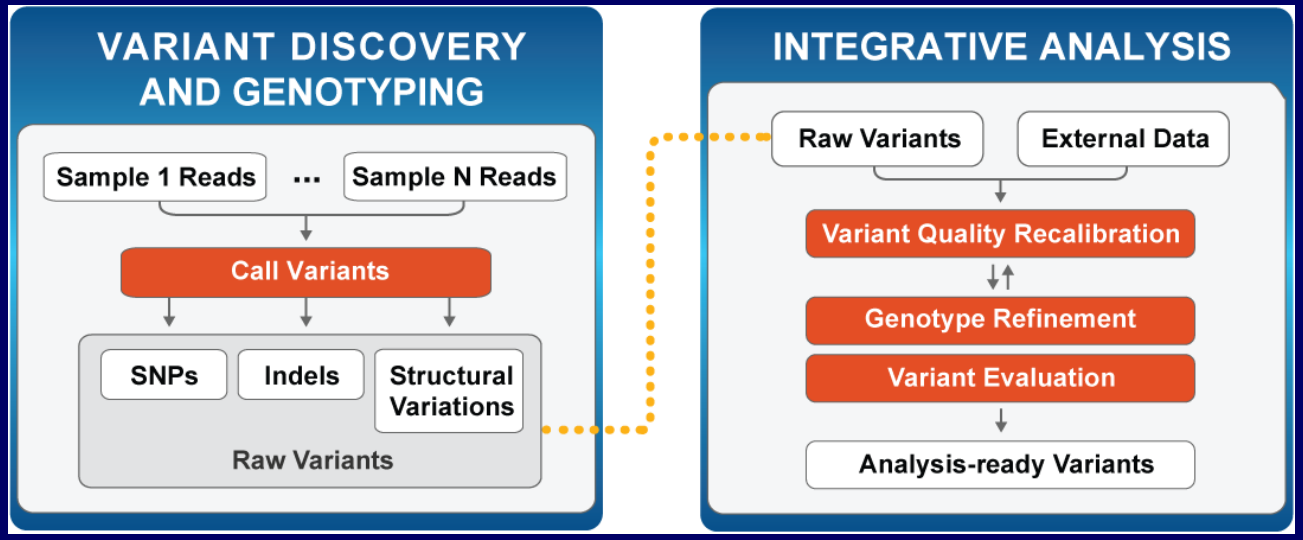
NHGRI and CMG Involvement:

- Executive Committee
- Diagnostic Committee
- First IRDiRC Scientific Meeting
- 2013 Bio International Convention

Genome Sequencing Informatics Tools

- GS-IT Program: “iSeqTools”

- iSeqTools projects hosting workshops for users



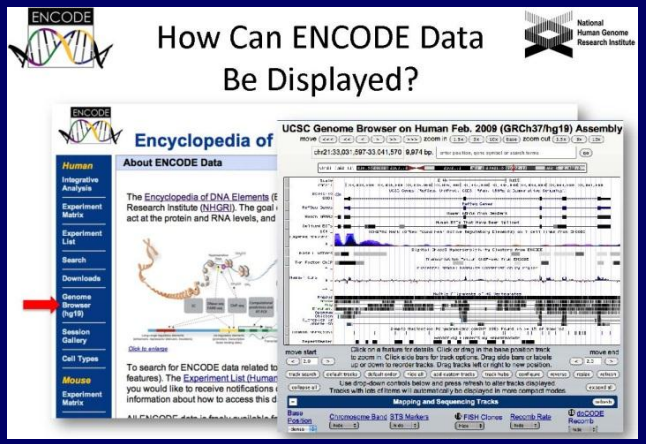
Broad Institute GATK Workflow

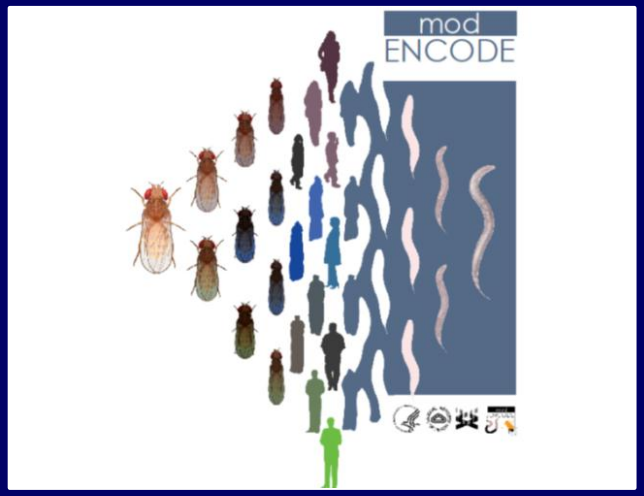
DNA Sequencing Technology Development

- **Grantee meeting April 29-May 2**
  - **Direct reading of modified cytosines**
  - **Fabrication of solid state nanopore arrays**
  - **Protein analysis**

- **New applications will be discussed in the Closed Session**

ENCODE





- CSHL 'Biology of Genomes' ENCODE Tutorial
- ENCODE Consortium Meeting on May 29-31
- Cross-species manuscripts submitted or in final preparations
  - modENCODE (fly/worm/human)
  - mouse ENCODE (mouse/human)

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ENCODE

- Mike Pazin gave ENCODE presentation at TEDMED 2013 in Washington, D.C.
- Two former ENCODE program analysts (Rebecca Lowdon and Judy Wexler) awarded NSF Graduate Research Fellowships

Rebecca Lowdon

Judy Wexler

Centers of Excellence in Genomic Science (CEGS) & Diversity Action Plan (DAP)

- **Program Announcement PAR-13-198**
  - **Letters of intent due June 8, 2013**
  - **Applications due July 9, 2013**

- **CEGS Grantee and DAP Meeting in October**

- 1,000th registered PhenX Toolkit user!
- 7 new FOAs recommend using PhenX measures (NIAAA, NIDA, NCI, DoD, etc.)

Posted on February 28, 2013 by NIH Staff

Improving Access to NIH-supported Common Data Element Initiatives

A growing number of NIH institutes and centers are enhancing opportunities for combining and comparing data from multiple studies by identifying sets of common data elements (CDEs). In January, NIH launched a web portal to improve access to information about NIH-supported CDE initiatives and assist investigators with tools and resources for developing protocols for data collection. The portal serves as both an entry point for NIH investigators seeking CDEs to use in their studies and as a means of coordinating work with other organizations that are interested in developing CDEs for their relevant research communities. Users can browse descriptive summaries of the CDE initiatives, identify the subject areas to which they apply, and link out to sources of additional information, including repositories of the data elements themselves. The portal currently contains information on 16 NIH-supported initiatives, tools, and resources, and will expand as additional initiatives are initiated and identified. For more information, visit http://cde.nih.gov.

GAIN Data Access Committee

COMMENTARY

A Mechanism for Controlled Access to GWAS Data: Experience of the GAIN Data Access Committee

Erin M. Ramos,[1,*] Corina Din-Lovinescu,[1] Ebony B. Bookman,[1] Lisa J. McNeil,[2] Carl C. Baker,[3] Georgy Godynskiy,[4] Emily L. Harris,[5] Thomas Lehner,[6] Catherine McKeon,[7] Joel Moss,[8] Vaurice

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Table 3. GAIN DAC Voting Decisions for Submitted PRs through 12/31/2011

Year	Number of PRs Submitted	PRs to DAC[a]	Average Data Sets per PR	Approved	Disapproved
2007	75	65 (87%)	1.75	57 (76%)	16 (21%)
2008	226	176 (78%)	2.92	190 (84%)	26 (12%)
2009	234	134 (57%)	3.62	159 (68%)	32 (14%)
2010	196	143 (73%)	3.05	129 (66%)	32 (16%)
2011	215	127 (59%)	2.97	136 (63%)	33 (15%)

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NHGRI GWAS Catalog

Published Genome-Wide Associations through 12/2012
Published GWA at $p \leq 5X10^{-8}$ for 17 trait categories

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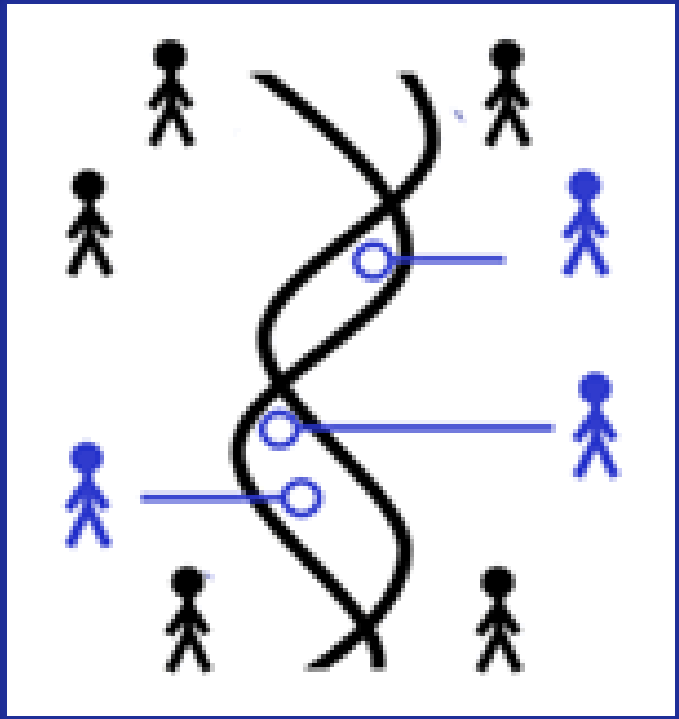
Population Architecture using Genomics and Epidemiology (PAGE)

- Metabochip genotyping completed on >60,000 non-European participants
- Analyses on >20 phenotypes underway



Population Architecture Using Genomics and Epidemiology (PAGE)

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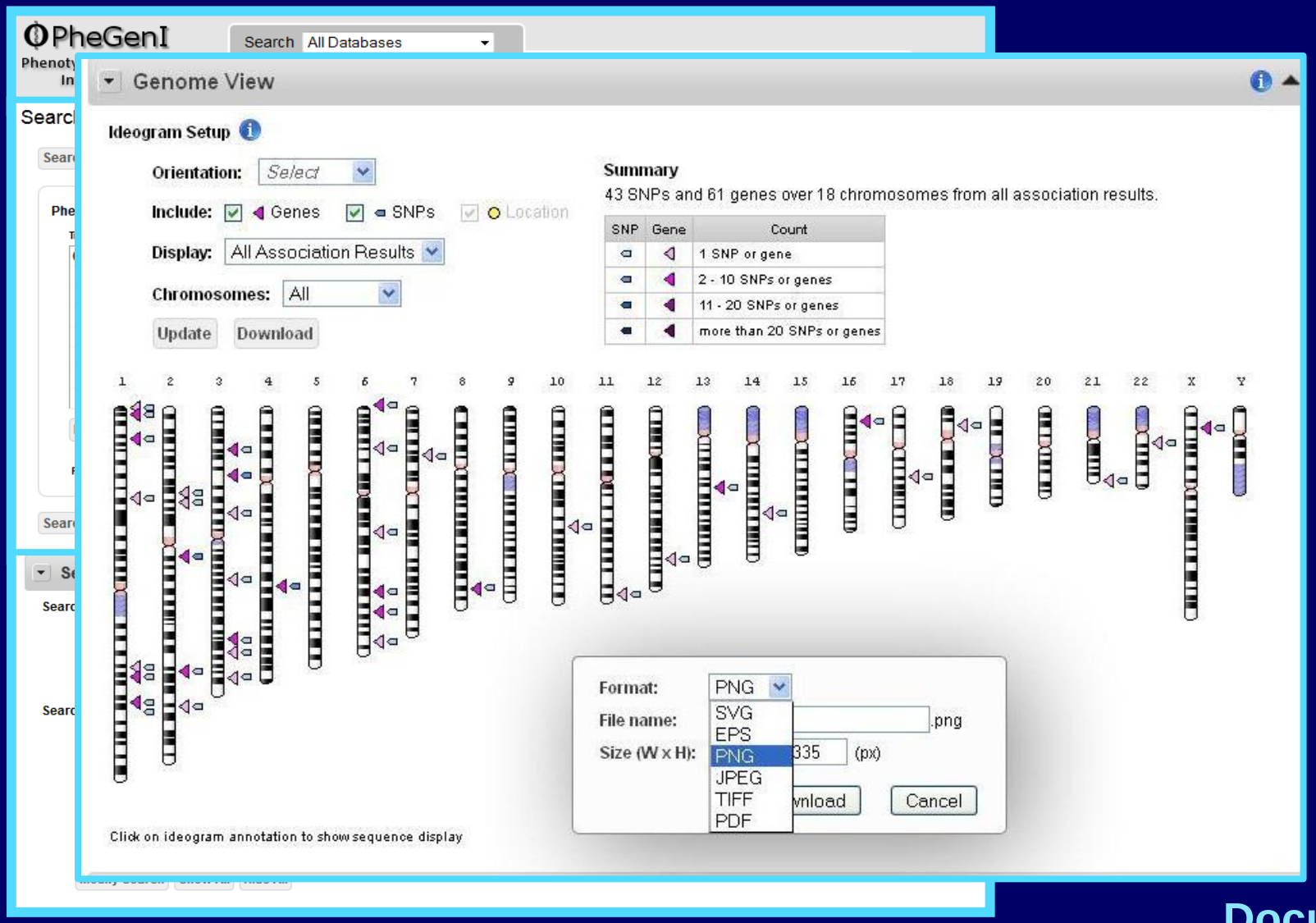
PheGenI
Phenotype-Genotype Integrator

Phenotype-Genotype Integrator (PheGenI)



Genomic Medicine Pilot Demonstration Projects

- Awards will be made summer 2013
- First Steering Committee meeting on July 9-10
- RFA re-issued– RFA-HG-13-004: Genomic Medicine Pilot Demonstration Projects (U01)

  Application Due Date: July 17, 2013

Genomic Medicine Working Group

- Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG) created
- ISCC-PEG Goal: To facilitate professional societies' efforts in educating physicians and other practitioners in the use of genomic medicine in clinical care

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

- Return of Results Consortium to hold joint meeting with CSER investigators on May 22-23
- Topics include:
  - ACMG recommendations on return of incidental findings
  - Liability issues
  - Legal issues related to CLIA
  - Implications of recent finding on identifiability of genomic data for data sharing

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Genomics and Society Working Group

- First meeting of the Genomics and Society Working Group held in April
- Next meeting in fall of 2013
- Presentation later in the Open Session

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NHGRI Training and Career Development Workshop

- ~20 experts in genomics, genomic medicine, and training met in April
- First review of NHGRI training programs since the Human Genome Project began
- Presentation later in the Open Session

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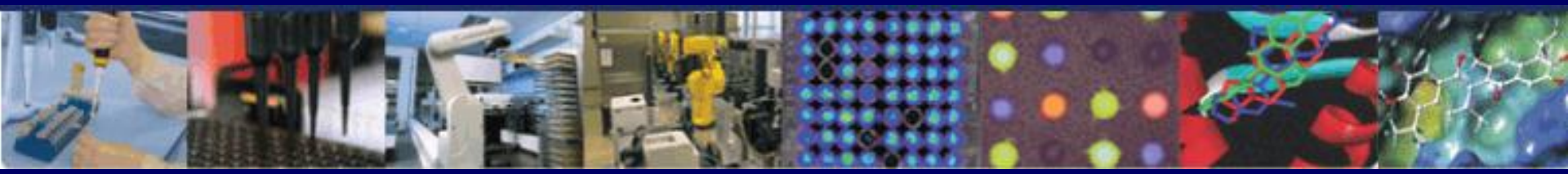
Molecular Libraries Program (MLP)

- Final year of the production phase
- Outcomes from past 5 years of production:

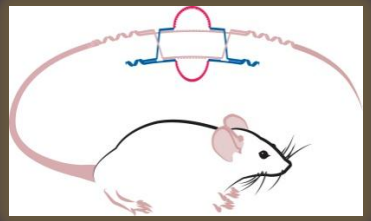
  Initiated 352 probe discovery projects

  Completed 340 high-throughput screens

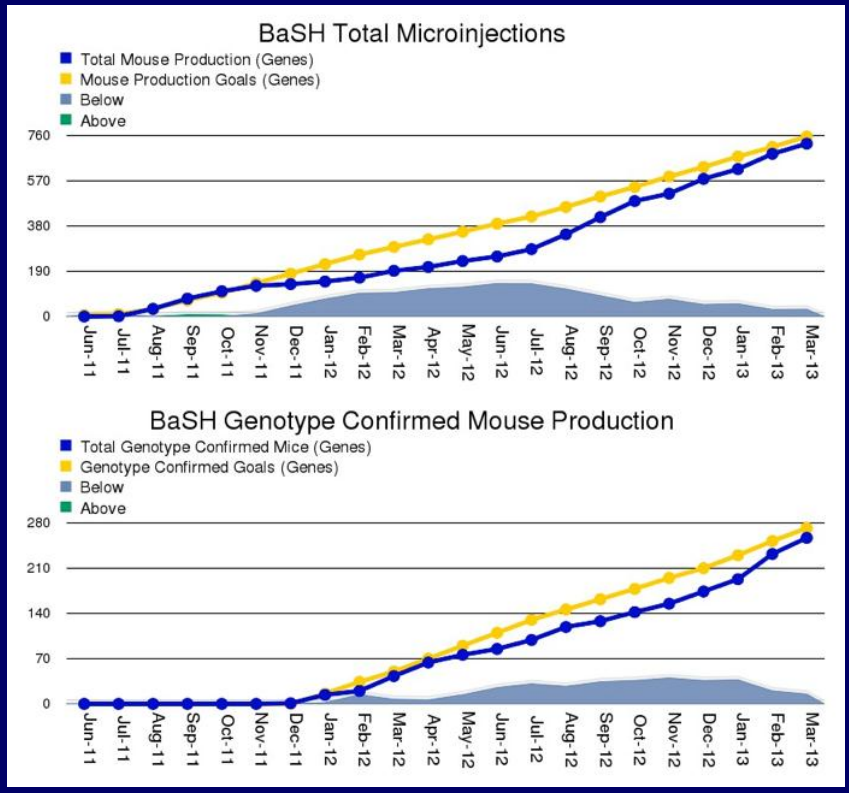
  Produced 348 small molecule probes



Knockout Mouse Phenotyping Project (KOMP^2)



- Launched in Fall, 2011
- Goal: make and phenotype 2,500 knockout strains
- Recent KOMP2 meeting
- Bloomsbury report on embryo phenotyping published

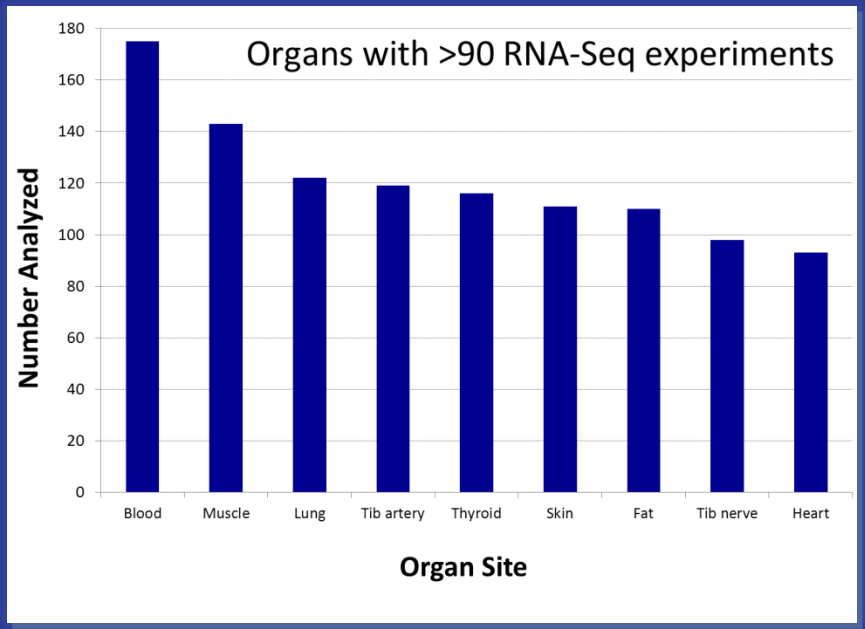


GTEx
Genotype-Tissue Expression Project

- GTEx pilot data in dbGaP

  182 post-mortem donors genotyped

  >1800 RNA-Seq studies



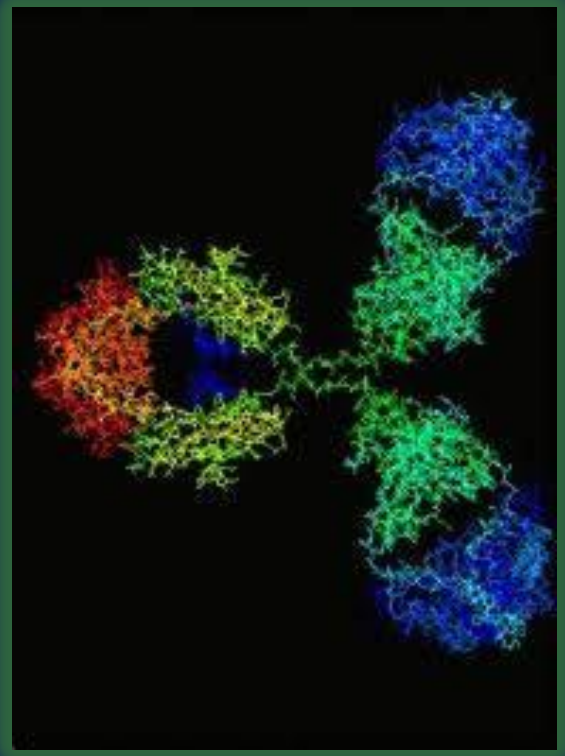
- 1st GTEx Community Meeting in Boston on June 18
- RFA "Enhancing GTEx with molecular analyses of stored biospecimens (U01)' will come to September Council meeting

Library of Integrated Network-based Cellular Signatures (LINCS)

- **LINCS Data Forum held in March**
  - **~150 total participants and >90 non-LINCS scientists (including 15 from pharma)**
  - **Included a joint LINCS-VIZBI session**
- **LINCS Phase 2 approved by NIH Common Fund**

Protein Capture Reagents Program

- Recent site visits of all 7 Centers
- Recent EU Affinomics Meeting
- First affinity reagents deposited in public repositories



H3Africa

Human Heredity and Health in Africa

- 2nd H3Africa Consortium meeting in Ghana
- Working groups met and presented policy recommendations to H3Africa Steering Committee
- Joint meeting with the AfSHG (May 19-21)

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Undiagnosed Diseases Network

- **RFA-RM-13-003: Undiagnosed Diseases Gene Function Research (R21)**

  Application Due Date: June 14, 2013

- **RFA-RM-13-004: Clinical Sites for an Undiagnosed Diseases Network (U01)**

  Application Due Date: June 19, 2013

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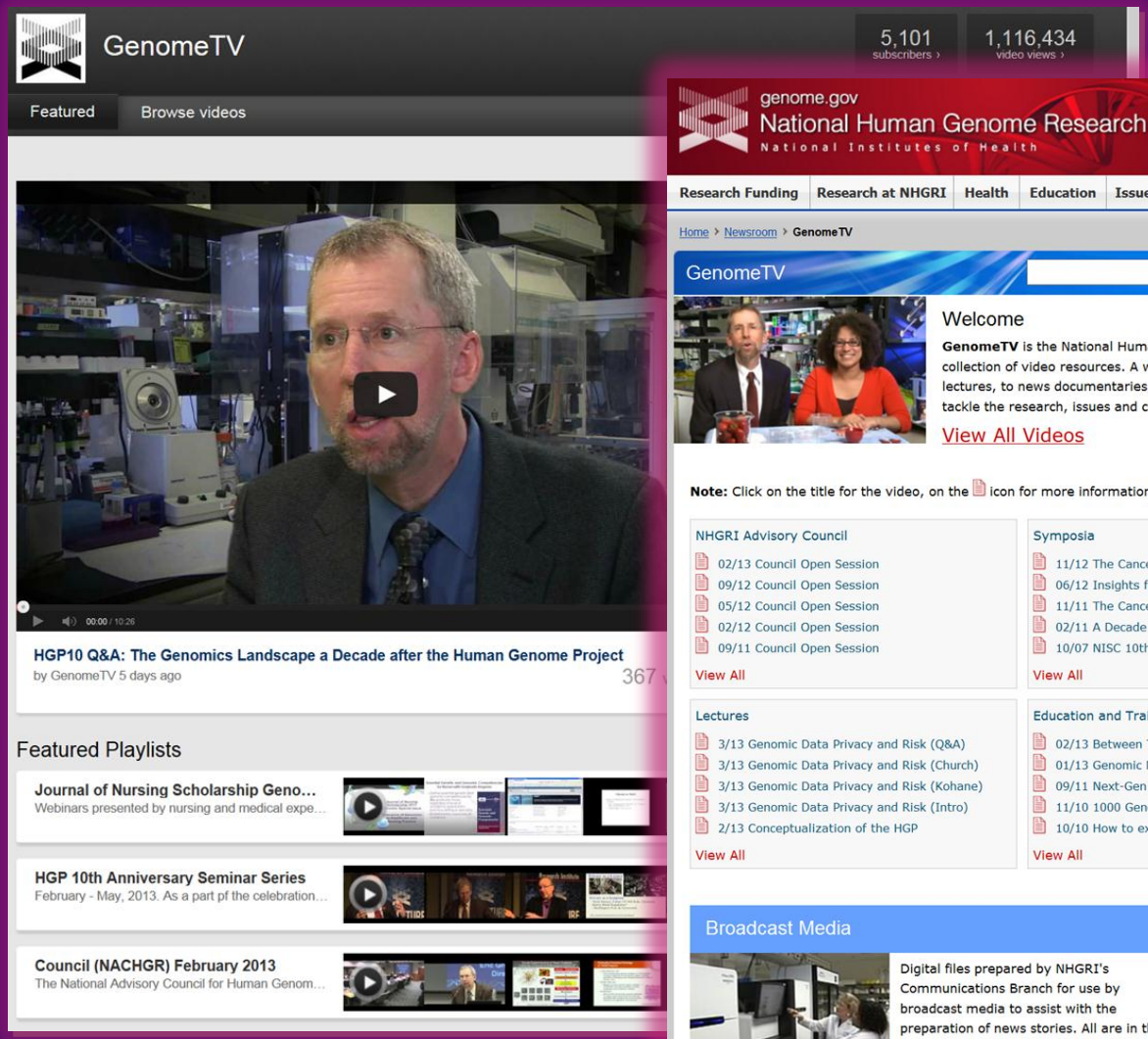
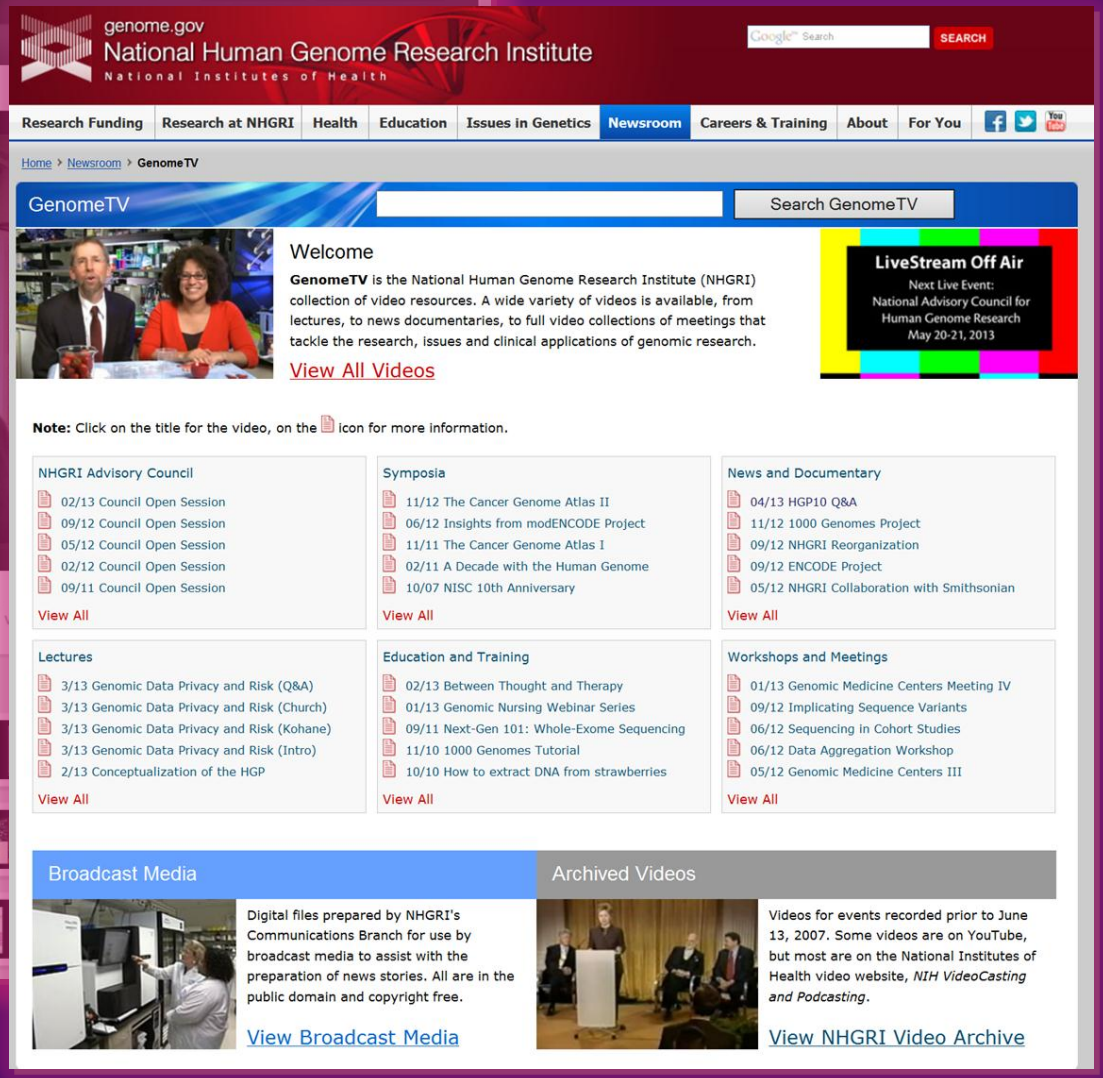
GenomeTV on YouTube





genome.gov/GenomeTV

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GenomeTV: Anecdotal Evidence of Outreach

From: khush bakhat <cool.khushi19@gmail.com>
Sent: Tue 3/19/2013 11:06 AM
To: Green, Eric (NIH/NHGRI) [E]
Cc:
Subject: Query of Strawbery DNA?

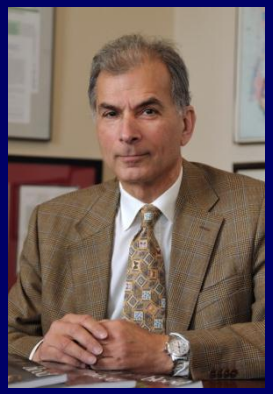
Respected Sir

i extract the strawbery DNA as u described in your video (http://www.genome.gov/genometv/search.cfm)
i perform all the steps same as u explain and spool out the DNA at the end in an eppendorf. i run it on 1 % agarose gel to confirm it but there was no band when i saw it in UV transilluminator?
Kindly explain me reason of this, if it was DNA then it should b visible in UV after running on Gel?

Regards
khush bakhat samreen
MS-II Biotechnology
Lahore College for Women University,
Lahore, Pakistan

Genomics in Medicine Lecture Series

2013 Focus on Genomics & Oncology



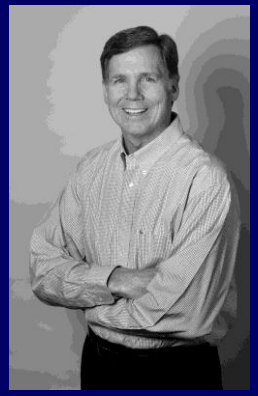
Neal Young, M.D.

Kathleen A. Calzone, Ph.D., R.N.

Thomas Ried, M.D.

Electron Kebebew, M.D.

Lee J. Helman, M.D.



W. Marston Linehan, M.D.

Louis M. Staudt, M.D., Ph.D.

Kenneth H. Kraemer, M.D.

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DNA Day 2013:
Washington, DC & Brooklyn, NY

>250 students at NMNH from 8 local high schools in Washington, DC

High school students from 5 partner schools in Brooklyn, NY

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International Canine Health Lifetime Achievement Award

THE KENNEL CLUB CHARITABLE TRUST
INTERNATIONAL CANINE HEALTH AWARDS
Supported by Vernon & Shirley Hill

Elaine Ostrander, Ph.D.

Social & Behavioral Research Branch (SBRB) 10th Anniversary: 2003-2013

- SBRB founded in December 2003
- Kick-off celebration coincided with last of the paired seminars commemorating 2013
- Upcoming celebration on October 30

Alexandra Shields, Eric Green, and Caryn Lerman

NHGRI Intramural Research Highlights

JAMA Neurology
Formerly *Archives of Neurology*

A Multicenter Study of Glucocerebrosidase Mutations in Dementia With Lewy Bodies ONLINE FIRST

nature genetics

A meta-analysis identifies new loci associated with body mass index in individuals of African ancestry

PLOS | GENETICS

A Copy Number Variant at the *KITLG* Locus Likely Confers Risk for Canine Squamous Cell Carcinoma of the Digit

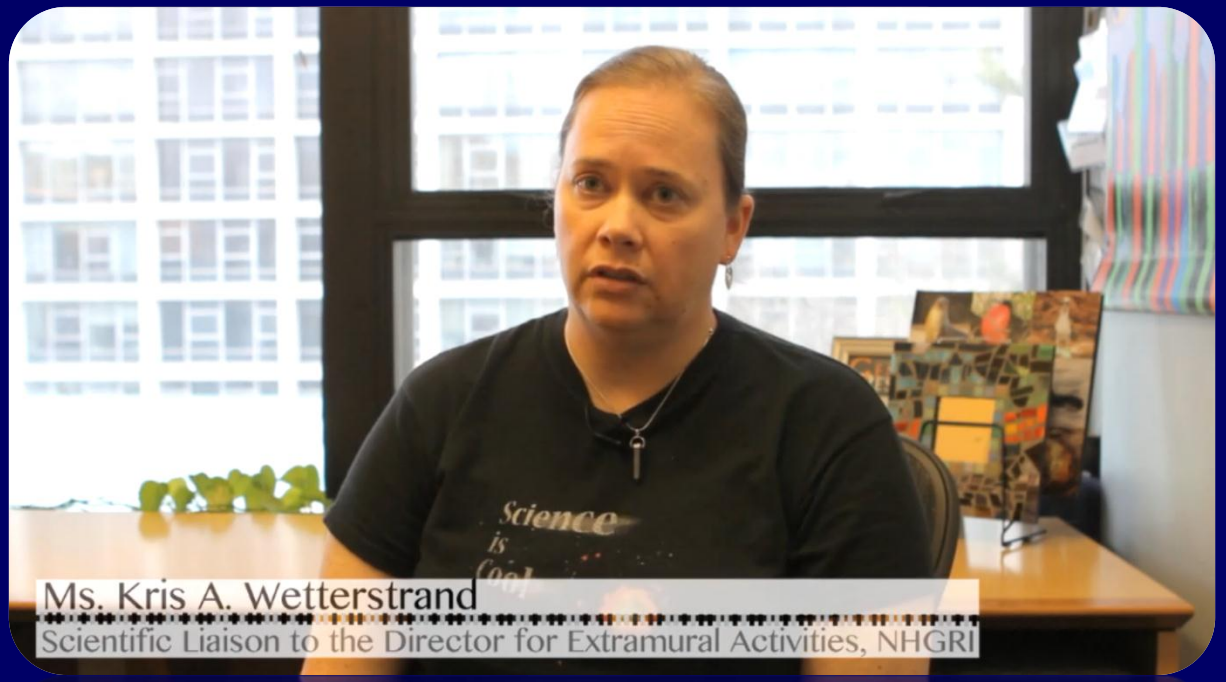
nature International weekly journal of science

Co-evolution of a broadly neutralizing HIV-1 antibody and founder virus

Document 53

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

Thanks!



Special Thanks!


National Human Genome
Research Institute
Advancing human health
through genomics research