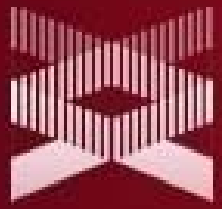


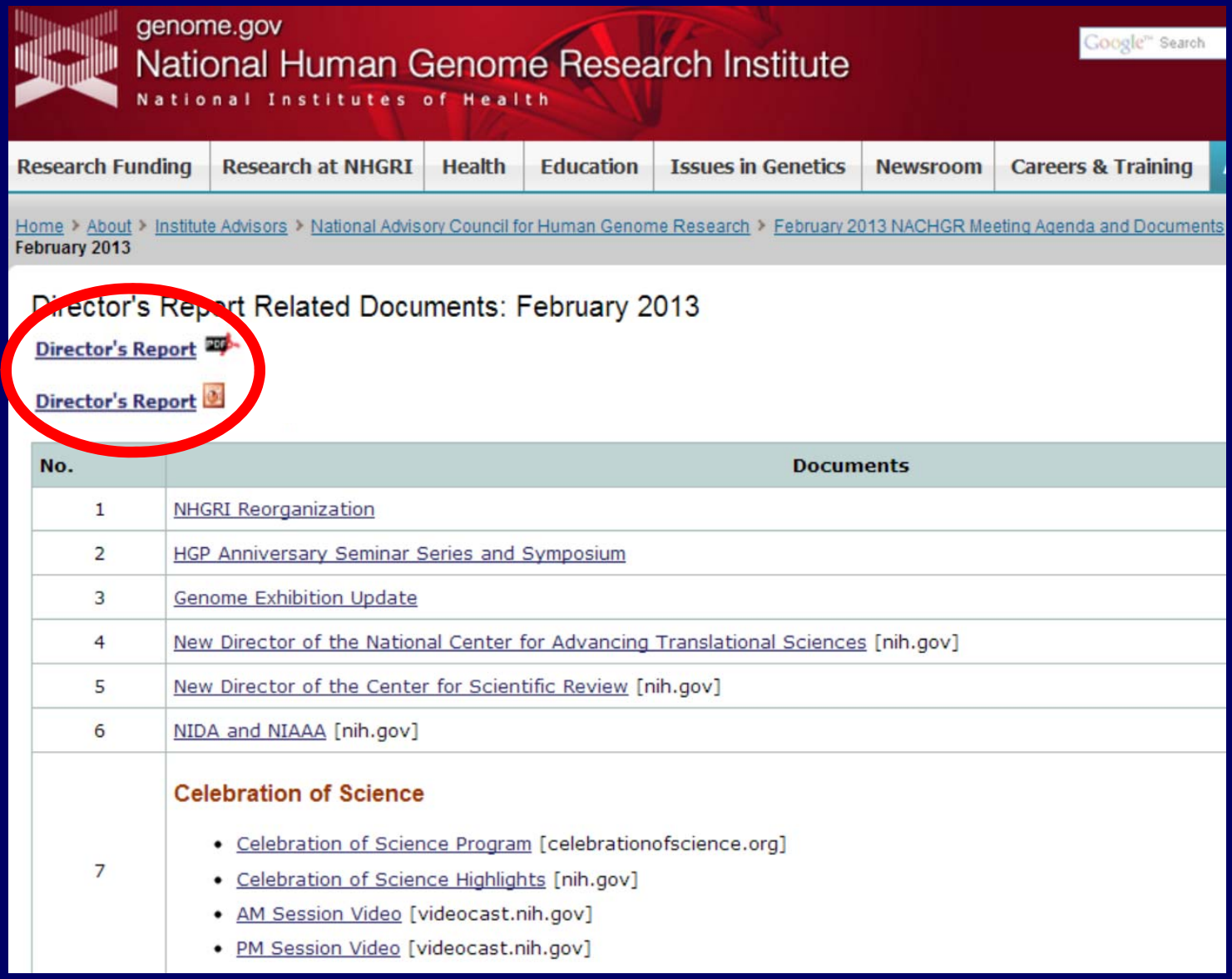
genome.gov
National Human Genome Research Institute
National Institutes of Health

DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

February 2013

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



genome.gov/DirectorsReport

Document #

Open Session Presentations

Presentation by NHLBI Director:

Gary Gibbons

Project Updates:

- **The Cancer Genome Atlas (TCGA)**

  Brad Ozenberger

- **Genotype-Tissue Expression (GTEx) Project**

  Simona Volpi

- **Human Heredity and Health in Africa (H3Africa)**

  Jane Peterson

Open Session Presentations

Biennial Report on Inclusion of Women and Minorities in NHGRI-Supported Research

Rongling Li

Centers of Excellence in Genomic Science (CEGS) Program Update:

Jeff Schloss

David Kingsley

Deidre Meldrum

Annual Review of the Statement of Understanding between NACHGR and NHGRI

Rudy Pozzatti

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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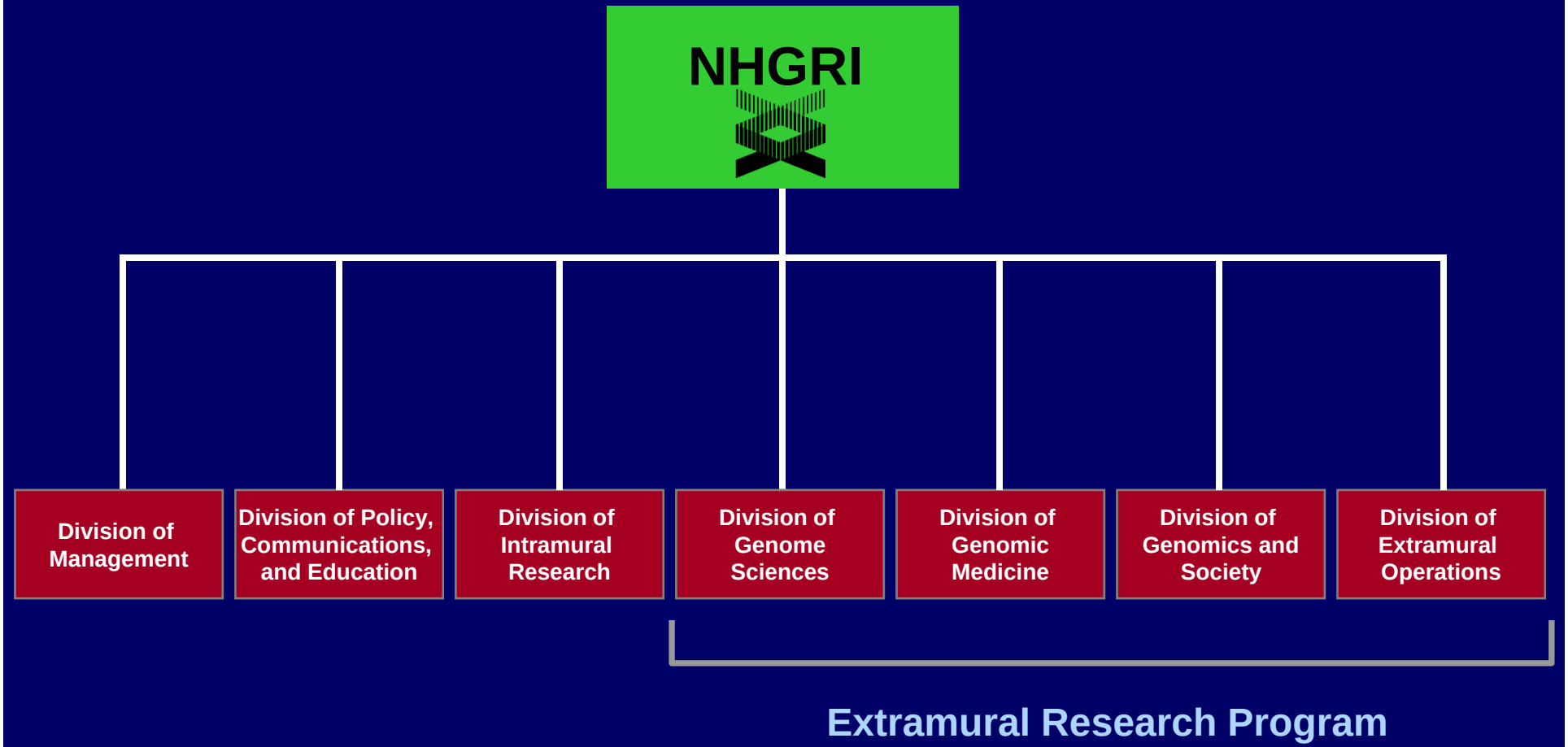
NHGRI Reorganization

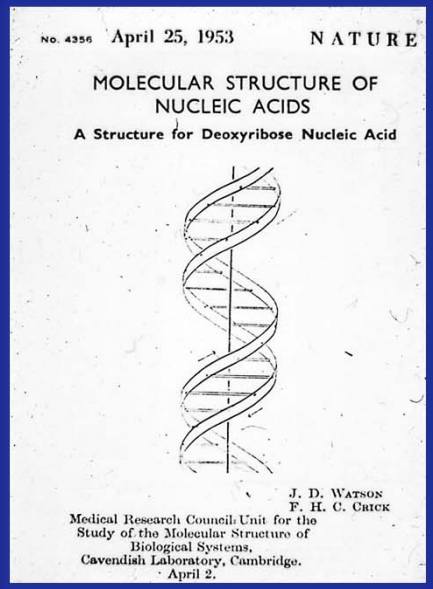
October 1, 2012: Implementation of New Organizational Structure for NHGRI

Document 1

New NHGRI Organizational Structure



2013: A Celebratory Year for Genomics



No. 4356 April 25, 1953 NATURE

MOLECULAR STRUCTURE OF NUCLEIC ACIDS

A Structure for Deoxyribose Nucleic Acid

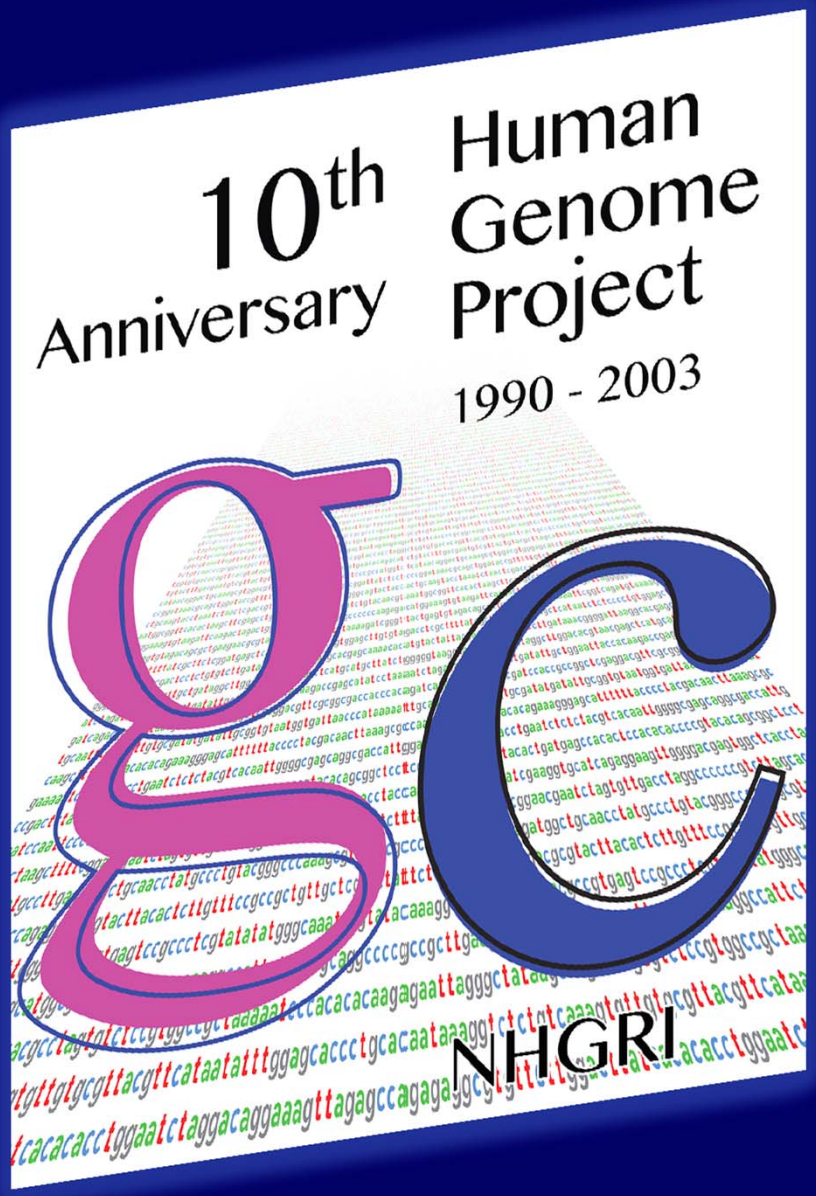
J. D. WATSON
F. H. C. CRICK

Medical Research Council Unit for the Study of the Molecular Structure of Biological Systems, Cavendish Laboratory, Cambridge.

April 2.

60th Anniversary

10th Anniversary


10th
Anniversary
Human
Genome
Project
1990 - 2003
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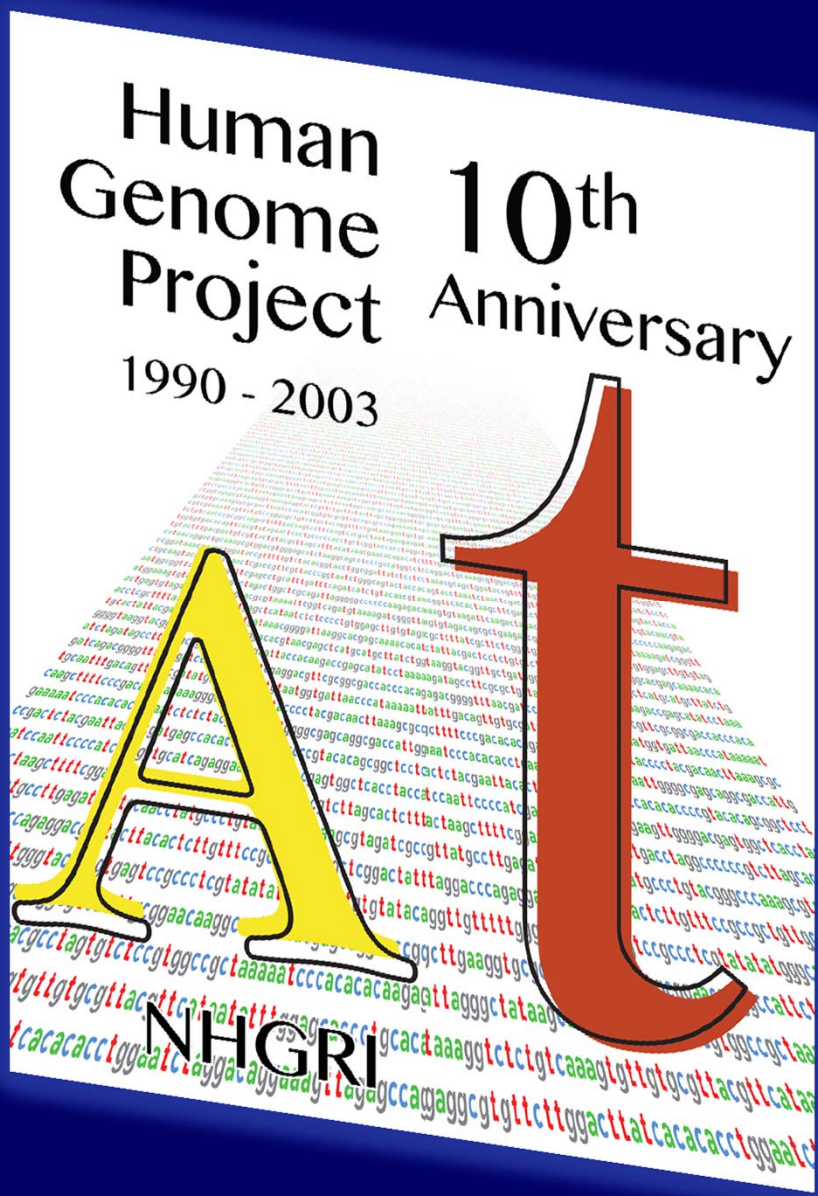
NHGRI


Human
Genome
Project
10th
Anniversary
1990 - 2003
A
t
NHGRI

Commemorative HGP 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

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).

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).

Document 2

Smithsonian NHGRI Genome Exhibition
Exhibition Advisory Board
Exhibition Announcement
HGP10

The Genomics Landscape a Decade After the Human Genome Project

Share Print

In April 2003, the International Human Genome Project (HGP), led in the United States by the National Institutes of Health, was completed ahead of schedule and under budget. For the first time, anyone could freely read the fundamental instruction set needed to make a human body. But much more still must be learned about life's operating system in order for it to be fully applied to human health.

"The Human Genome Project has had an incalculable impact on science over the past decade," said Eric D. Green, M.D., Ph.D., director of NHGRI, who was recruited to NIH early on in the 13-year project. "I am especially pleased that our varied events highlight genomics in so many ways-because the ongoing work of human genetics and genomics benefits all of us."

Beginning in February 2013, the National Human Genome Research Institute (NHGRI), the NIH institute that spearheaded the HGP, will celebrate the 10th anniversary of the completion of the Human Genome Project with a series of stimulating talks, a thought-provoking symposium and a fascinating interactive exhibit to mark the project's 10-year anniversary and to reflect on the HGP's revolutionary influence on biomedicine.

Upcoming Events

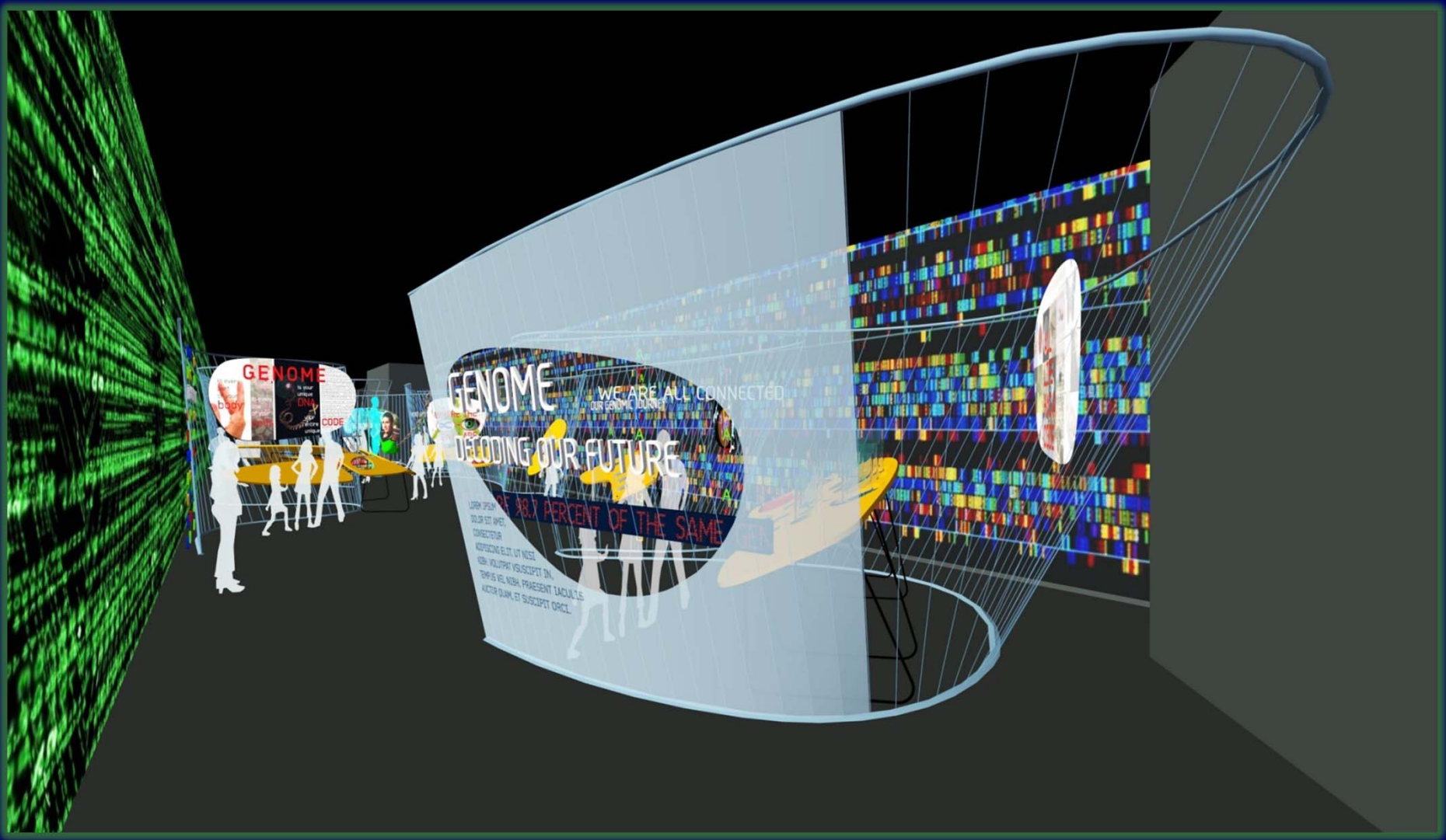
- HGP 10th Anniversary Seminar Series
  February - March, 2013
- HGP 10th Anniversary Symposium Agenda
  April 25th, 2013
- Smithsonian NHGRI Genome Exhibition
  NHGRI's official exhibition page for the HGP 10th Anniversary

Background Resources

genome.gov/HGP10

Document 2

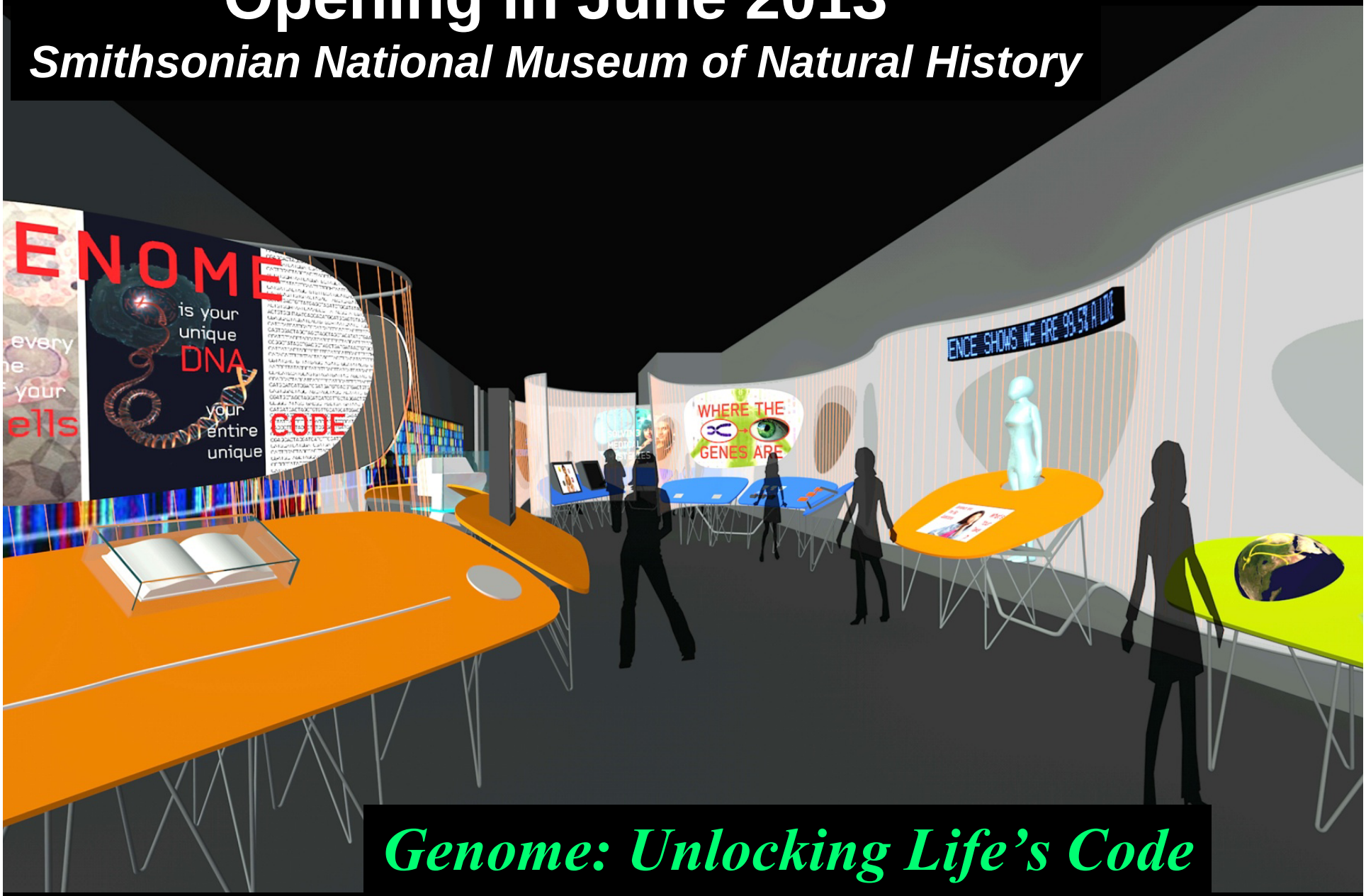
Genome Exhibition Update



Document 3

Opening in June 2013

Smithsonian National Museum of Natural History



Genome: Unlocking Life's Code

LISTINGS

Hot tickets for 2013 in science and art

Human Genome exhibition
Smithsonian National Museum of Natural History, Washington DC
June 2013 to June 2014

In a year that sees both the 60th anniversary of Francis Crick and James Watson's elucidation of DNA's structure and the 10th anniversary of the human genome's complete decoding, the Smithsonian Institution in is pulling out all the stops. For this exhibition, its natural history museum joins forces with the National Human Genome Research Institute in Bethesda, Maryland, to explore what the genome is, what it tells us and how this information could revolutionize health care and our understanding of our place in the world. After its time on the National Mall, the show will travel around North America.

***Nature* 2013**

NHGRI Historical Archiving Efforts

Director's Report Outline

I. General NHGRI Updates

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III. General Genomics Updates

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VII. NHGRI Intramural Research Program

New Director of the National Center for Advancing Translational Sciences

Christopher Austin, M.D.

Document 4

New Director of the Center for Scientific Review

center for scientific review

Richard Nakamura, Ph.D.

Document 5

NIDA and NIAAA: No Merger

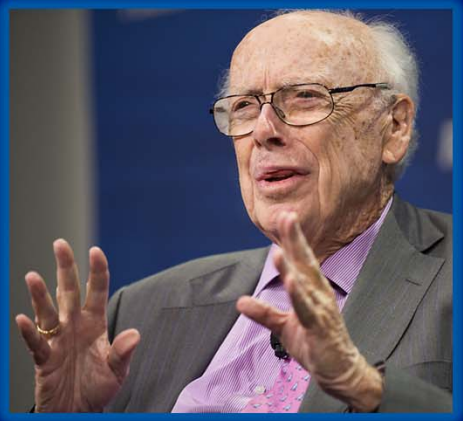
Statement by NIH Director Dr. Francis Collins on the future of substance use, abuse, and addiction-related research at NIH

FOR IMMEDIATE RELEASE
Friday, November 16, 2012

NIDA NATIONAL INSTITUTE ON DRUG ABUSE
The Science of Drug Abuse & Addiction

Document 6

A Celebration of Science



A CELEBRATION OF SCIENCE
Renewing Our Commitment to the Future
Washington, DC • September 7-9, 2012

Document 7

NIH Director's Blog

NIH DIRECTOR'S BLOG

Post archives

Select Month

Top Posts for the Month

- New Weapon Targets Ancient Foe
- How Influenza Pandemics Occur
- A Brain Pacemaker for Alzheimer's Disease?
- The Symphony Inside Your Brain
- Mobile Health: Figuring Out What Works

Blog Info

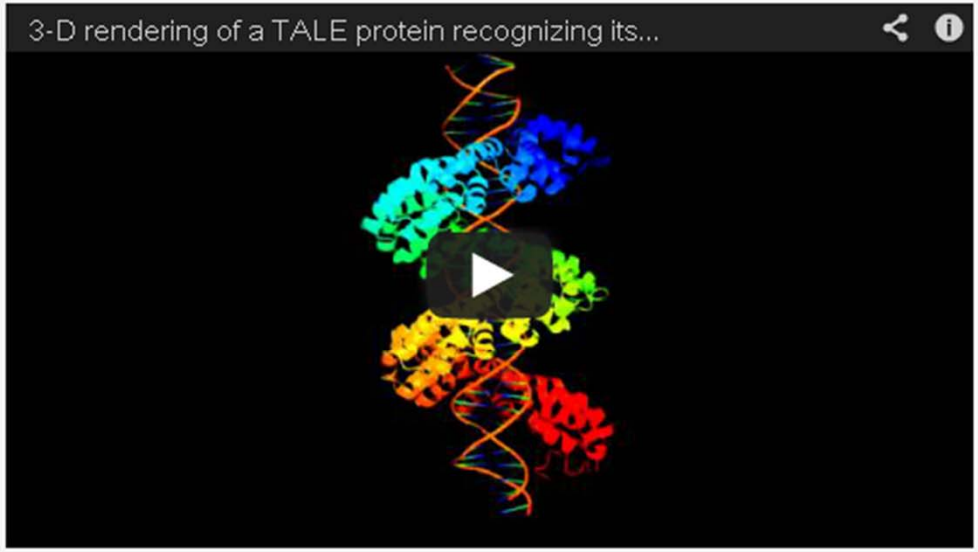
Editor: Bijal Trivedi

If you have comments or questions not related to the current discussions, please direct them to olib@od.nih.gov

« MRSA in a New Light The Science of Stuttering »

Copy-editing the Genome: Extreme Personalized Medicine?

By Dr. Francis Collins, on January 22nd, 2013



COOL TOOL. See how the TALE protein (rainbow colored) recognizes the target DNA site and wraps around the double helix. When this TALE protein is fused to a nuclease (the scissors), creating a TALEN, the hybrid protein will clip the DNA at the target site. Credit: Jeffry D. Sander, Massachusetts General Hospital

Document 8

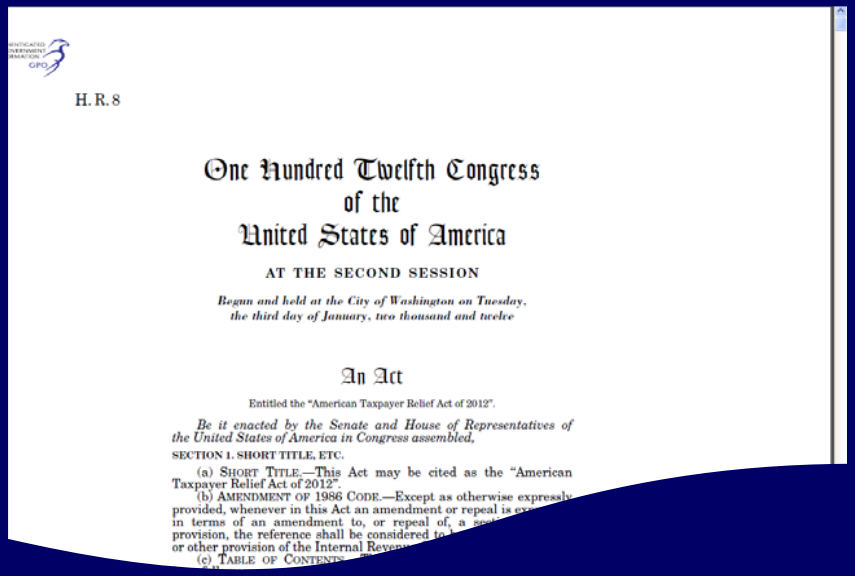
Use of Chimpanzees in NIH-Supported Research

Document 9

Fiscal Year 2013 Appropriations Update

- Continuing Resolution expires on March 27
- Congress must still determine appropriations for the rest of Fiscal Year 2013

Other Budgetary Complexities



H. R. 8

One Hundred Twelfth Congress
of the
United States of America

AT THE SECOND SESSION

Begun and held at the City of Washington on Tuesday, the third day of January, two thousand and twelve

An Act

Entitled the "American Taxpayer Relief Act of 2012".

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

SECTION 1. SHORT TITLE, ETC.

(a) SHORT TITLE.—This Act may be cited as the "American Taxpayer Relief Act of 2012".

(b) AMENDMENT OF 1986 CODE.—Except as otherwise expressly provided, whenever in this Act an amendment or repeal is ex... in terms of an amendment to, or repeal of, a ... provision, the reference shall be considered to ... or other provision of the Internal Reven...

(c) TABLE OF CONTENTS.—...

- Immediate threat of 8.2% cut averted
- If no resolution, a new sequester will be ordered by the President on March 1; implemented on March 27
- Potential reduction in Fiscal Year 2013 NIH budget would be less than 8.2%

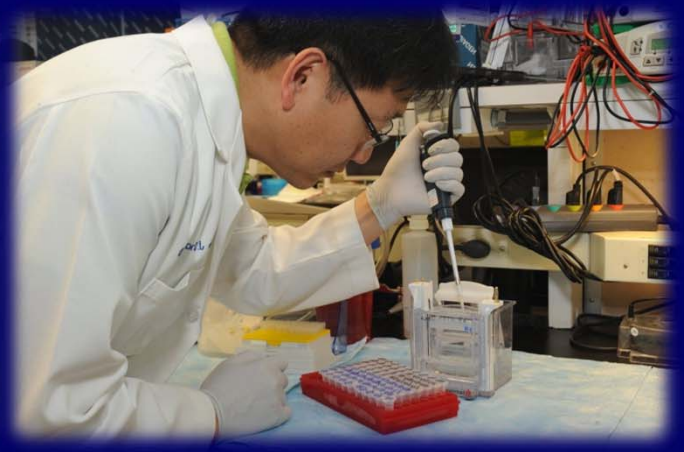
Diversity in the Biomedical Research Workforce Working Group

- NIH Building Infrastructure Leading to Diversity (BUILD) Program
- National Research Mentoring Network (NRMN)
- BUILD and NRMN Coordinating and Evaluation Centers

Document 10

Biomedical Workforce Working Group

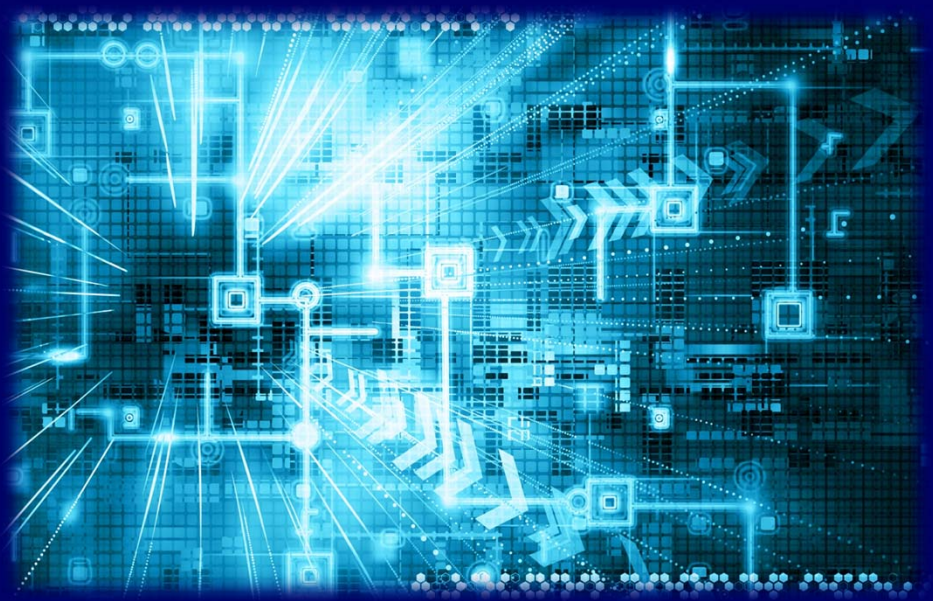
- Establish a grant program
- Improve graduate student/postdoc training
- Create an NIH ‘functional unit’ to assess the biomedical research workforce

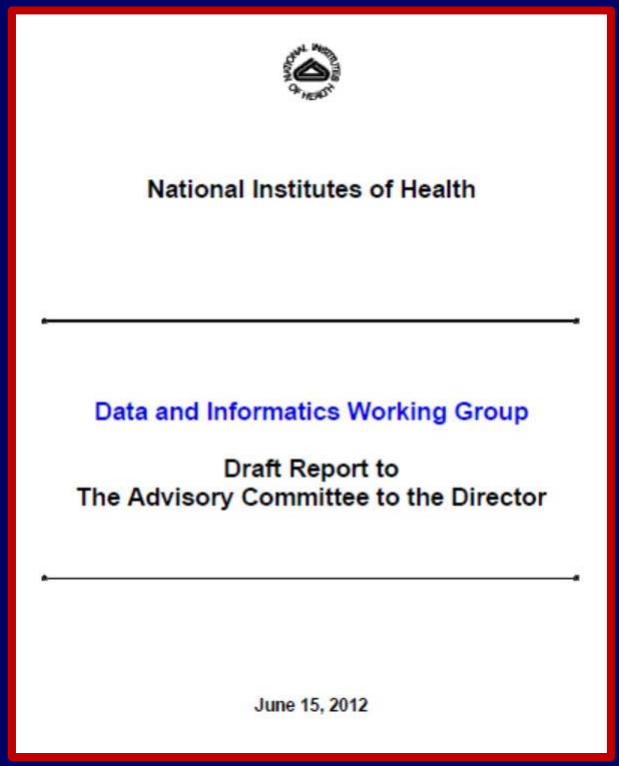


Document 10

Data and Informatics Working Group

- InfrastructurePlus
- Scientific Big Data: Three-pronged plan





National Institutes of Health

Data and Informatics Working Group

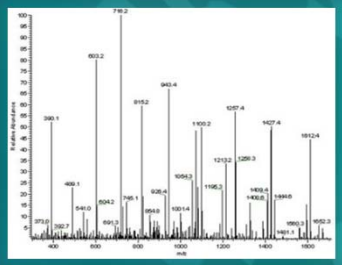
Draft Report to
The Advisory Committee to the Director

June 15, 2012

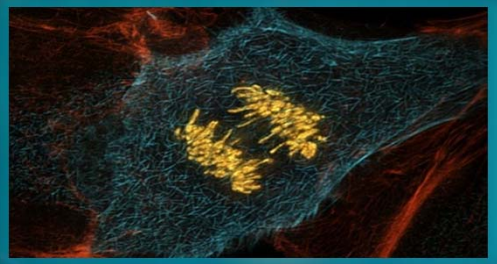
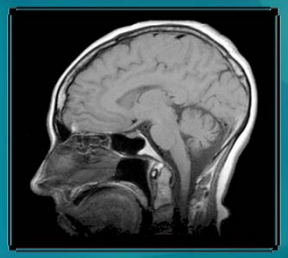
Document 10

Myriad Data Types


Genomic




Other ‘Omic

Imaging


Phenotypic


Exposure


Clinical

NIH is Tackling the 'Big Data' Problem

1. New NIH Leadership Position:

   Associate Director for Data Science

2. New Internal NIH Governing/Oversight Body:

   Scientific Data Council

3. New Trans-NIH Initiative:

   Big Data to Knowledge (BD2K)

Big Data to Knowledge (BD2K): Overview

- Major trans-NIH initiative addressing an NIH imperative and key roadblock
- Transformational, catalytic, and synergistic
- Overarching goal:

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

BD2K: Four Programmatic Areas

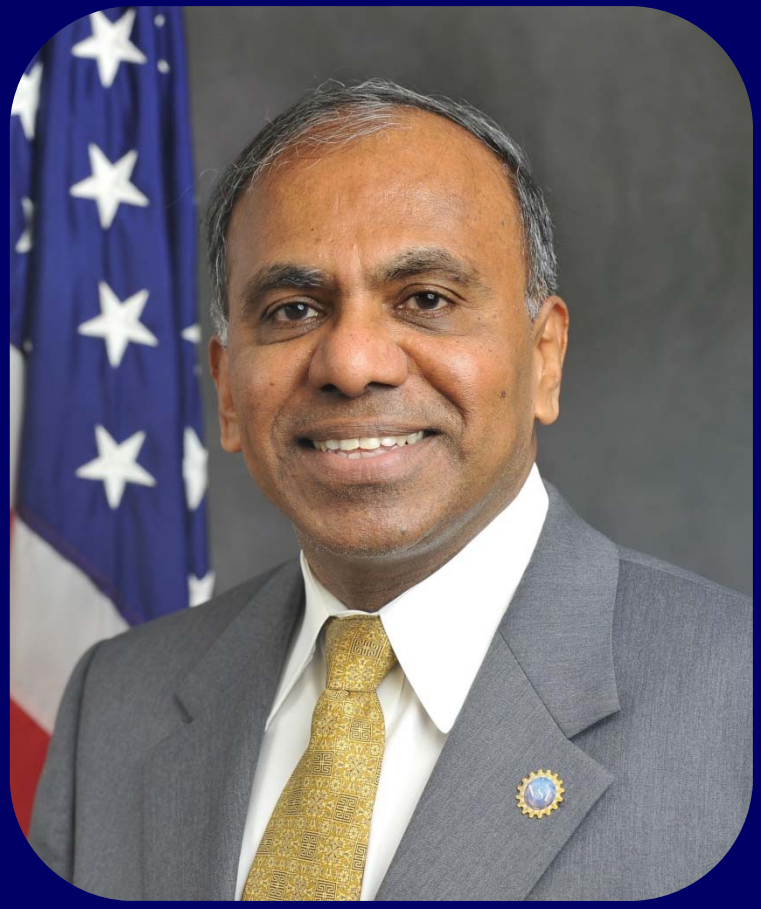
I. Facilitating Broad Use of Biomedical Big Data

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

NSF Director to Depart for Presidency of Carnegie Mellon University



Subra Suresh, Sc.D.

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Mourning the Loss of David Cox

Document 11

Mourning the Loss of Arlen Specter

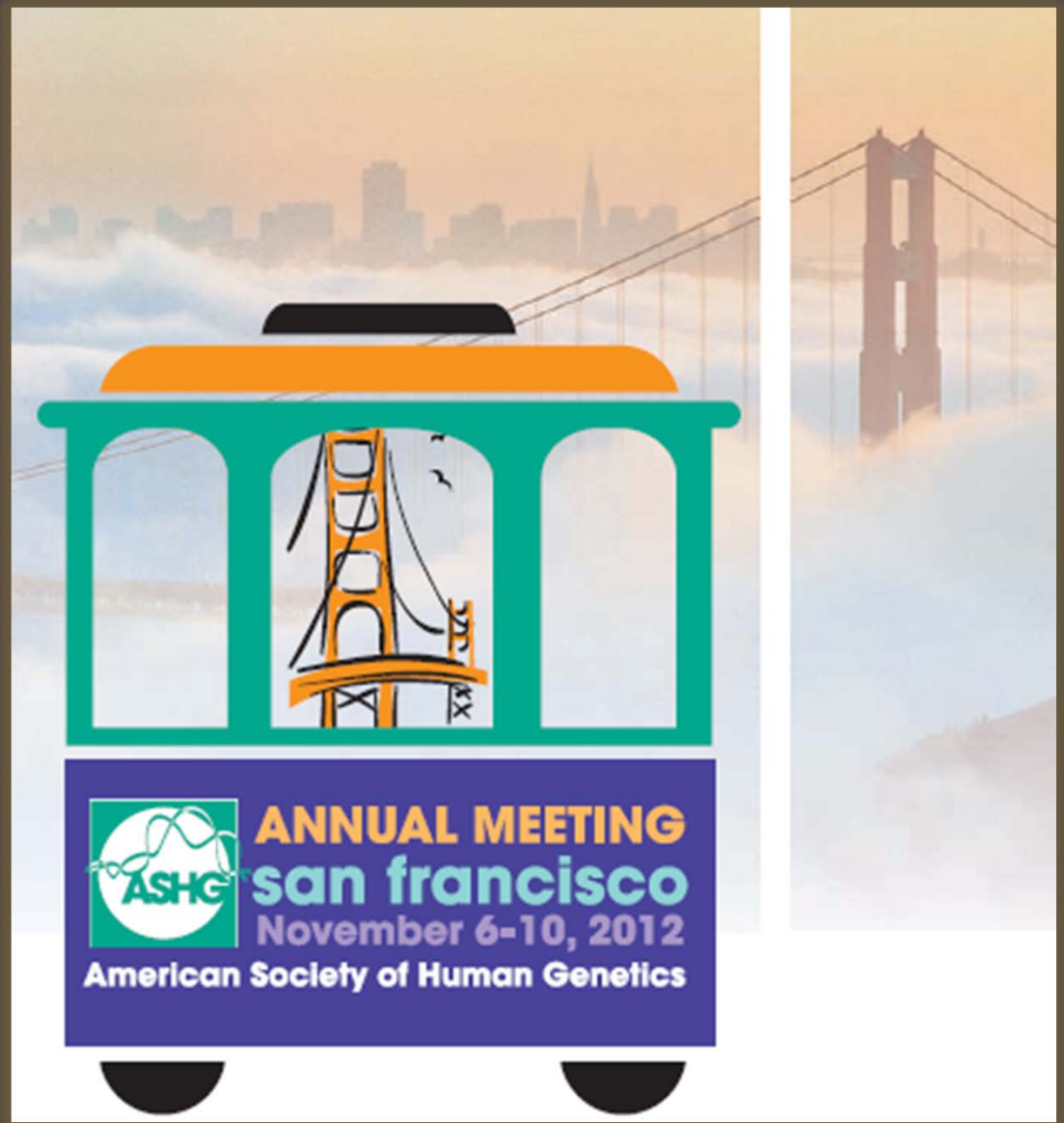
Document 12

National Medal of Science

Lee Hood, Ph.D.

Document 13

Awards at 2012 ASHG Annual Meeting

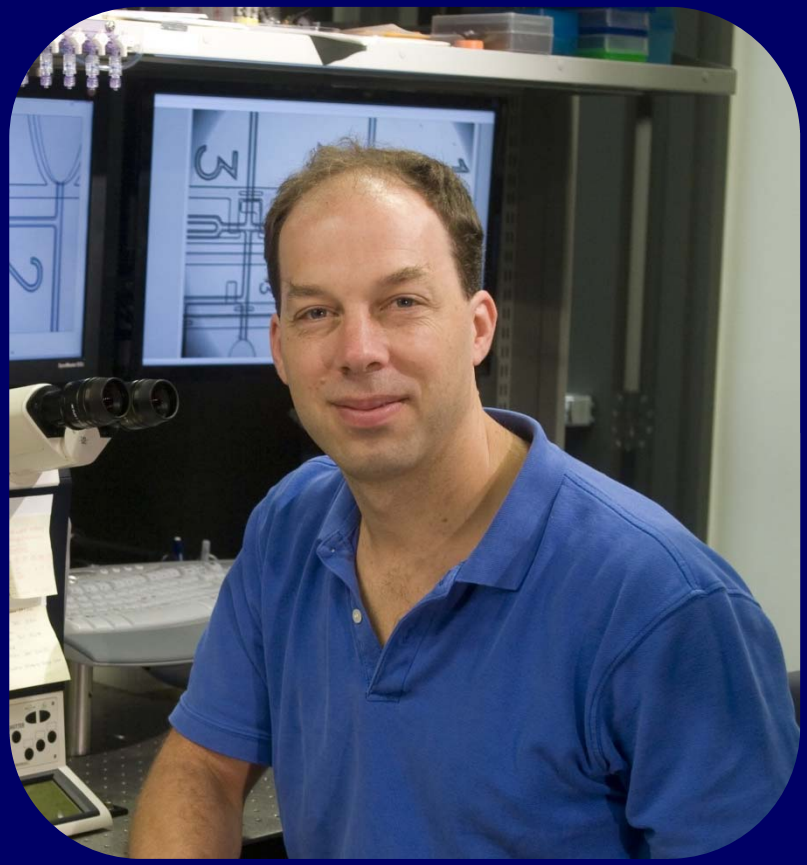


Francis Collins, M.D., Ph.D.

Jay Shendure, M.D., Ph.D.

Document 14

Human Frontier Science Program Organization 2013 Nakasone Award



HUMAN FRONTIER SCIENCE PROGRAM

FUNDING FRONTIER RESEARCH INTO COMPLEX BIOLOGICAL SYSTEMS

Stephen Quake, D.Phil.

Document 15

Taubman Prize for Excellence in Translational Medical Science

Hal Dietz, M.D.

Document 16

2013 Genetic Society of America Medal

Genetics Society of America

Elaine Ostrander, Ph.D.

Document 17

2013 Edward Novitski Prize

Genetics Society of America

Jonathan Pritchard, Ph.D.

Document 18

Elected to the Institute of Medicine

- Lynda Chin
- Daniel Kastner
- Stephen Quake

Elected to AAAS

- Michael Brent
- Susan Celniker
- Joseph Ecker
- Steven Henikoff
- Kathy Hudson
- Lynn Jorde
- Rob Knight
- Charles Lee
- Elaine Ostrander
- Reed Pyeritz
- Dan Roden
- Jane Silverthorne
- John Stamatoyannopoulos

Document 20

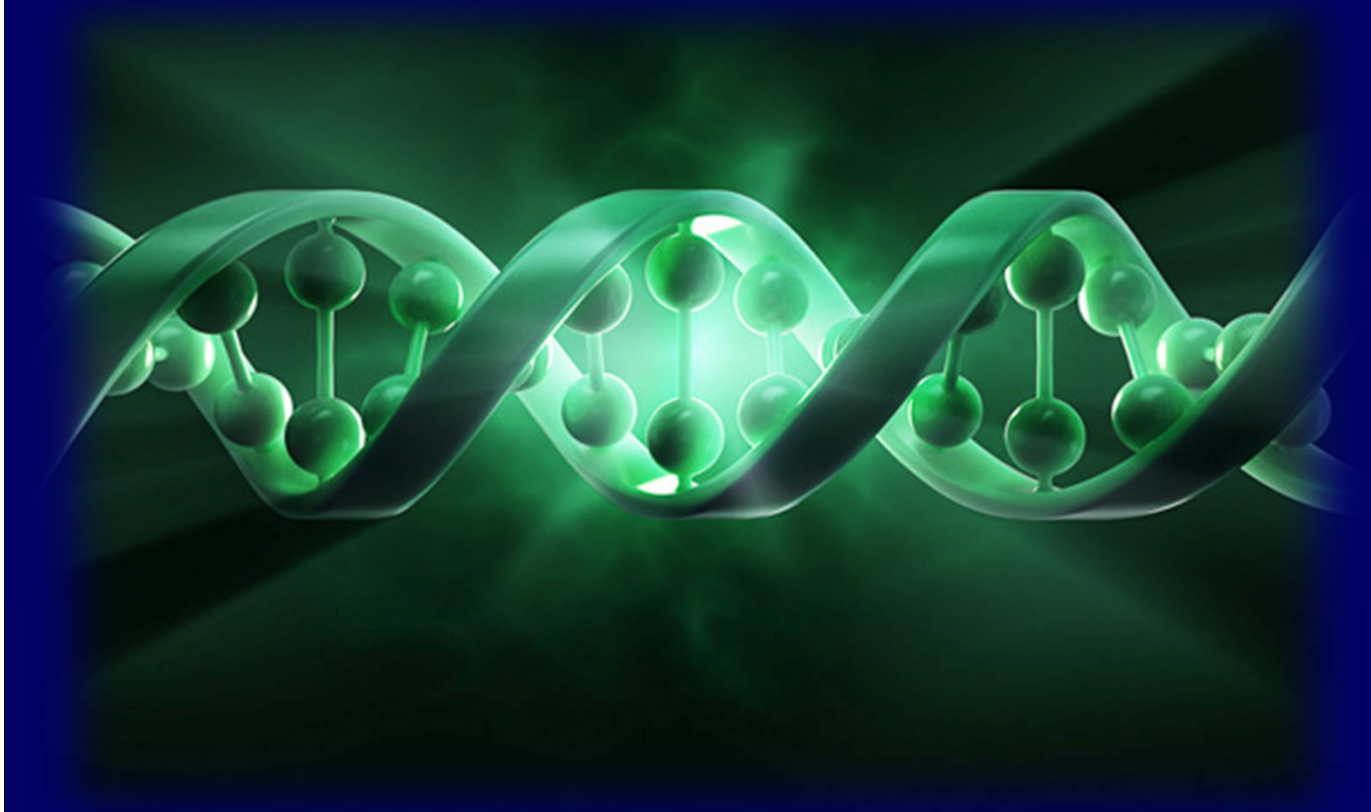
Director Named for the New York Genome Center

NEW YORK GENOME CENTER™

Bob Darnell, M.D., Ph.D.

Document 21

Launching of Stanford Center for Computational, Evolutionary, and Human Genomics



STANFORD UNIVERSITY
School of Humanities & Sciences

STANFORD
SCHOOL OF MEDICINE
Stanford University Medical Center

Document 22

Secretary's Advisory Committee on Human Research Protections: New Members

Jeffrey Botkin, M.D., M.P.H. (Chair Designate)

Pilar Ossorio, J.D., Ph.D.

Document 23

New Executive Vice President of ASHG

THE AMERICAN SOCIETY OF HUMAN GENETICS

Joe McInerney, M.S.

Genomic Identifiability

Identifying Personal Genomes by Surname Inference

Melissa Gymrek,[1,2,3,4] Amy L. McGuire,[5] David Golan,[6] Eran Halperin,[7,8,9] Yaniv Erlich[1]*

POLICYFORUM

RESEARCH ETHICS

The Complexities of Genomic Identifiability

Recent work reveals the need to re-examine the current paradigms for managing the potential identifiability of genomic and other "omic"-type data.

Laura L. Rodriguez,[1] Lisa D. Brooks,[1] Judith H. Greenberg,[2] Eric D. Green[1]*

Surnames of CEPH individuals identified using Y-STR data and publicly accessible Internet resources

Document 24

ACMG Statement on Variant Disclosure

ACMG
Translating Genes into Health®

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Advanced Search

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FOR IMMEDIATE RELEASE

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

Contact Kathy Beal, MBA
ACMG Media Relations Director
301-238-4582
kbeal@acmg.net

ACMG Releases New Position Statement on the Public Disclosure of Clinically Relevant Genome Variants

BETHESDA, MD—Nov. 7, 2012 | The American College of Medical Genetics and Genomics (ACMG) released a new official **Position Statement on the Public Disclosure of Clinically Relevant Genome Variants**. This important new statement addresses the problems resulting from gene patent monopolies that have allowed some to develop proprietary databases of the clinical meaning of the variants in particular genes.

Michael S. Watson, PhD, FACMG, executive director of the ACMG explained, "The next phase of the human genome project, which is to annotate the human genome sequence with the clinical and biological meaning of the sequences and variants, will require capturing information from a very large number of people from diverse populations across the US and internationally. Information that informs us about the meaning of genome sequences should be in the public domain where it can be used for the benefit of all."

The just-released Position Statement says, "The American College of Medical Genetics and Genomics (ACMG) believes that gene testing and the clinical data on which genetic data are interpreted must remain widely accessible and affordable, and that the development and improvement of safe and effective genetic tests should not be hindered. Monopolistic practices that limit a given genetic test to a single laboratory are inconsistent with ACMG's goals of broadly accessible and affordable genetic tests."

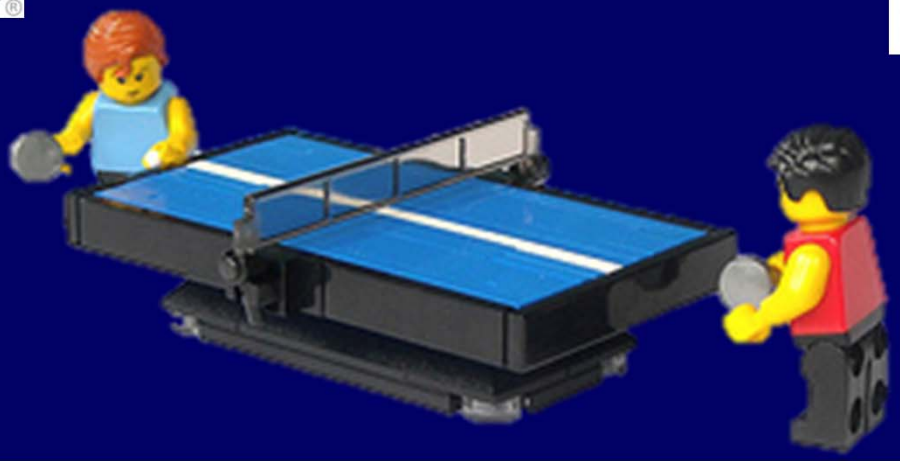
Position Statement on the Public Disclosure of Clinically Relevant Genome Variants
Genetic and genomic tests are playing an increasing role in medical practice, enabling prevention, diagnosis, and management of both rare and common disorders and the analysis of genetic changes associated with cancer. Testing is done in commercial, academic, and hospital laboratories throughout the world. The clinical interpretation of rare variants requires access to data on clinical annotation, but some laboratories have maintained private databases that are not publicly available. In some cases, these

"Monopolistic practices that limit a given genetic test to a single laboratory are inconsistent with ACMG's goals of broadly accessible and affordable genetic tests."

Document 25

Biotech Patents and the Courts

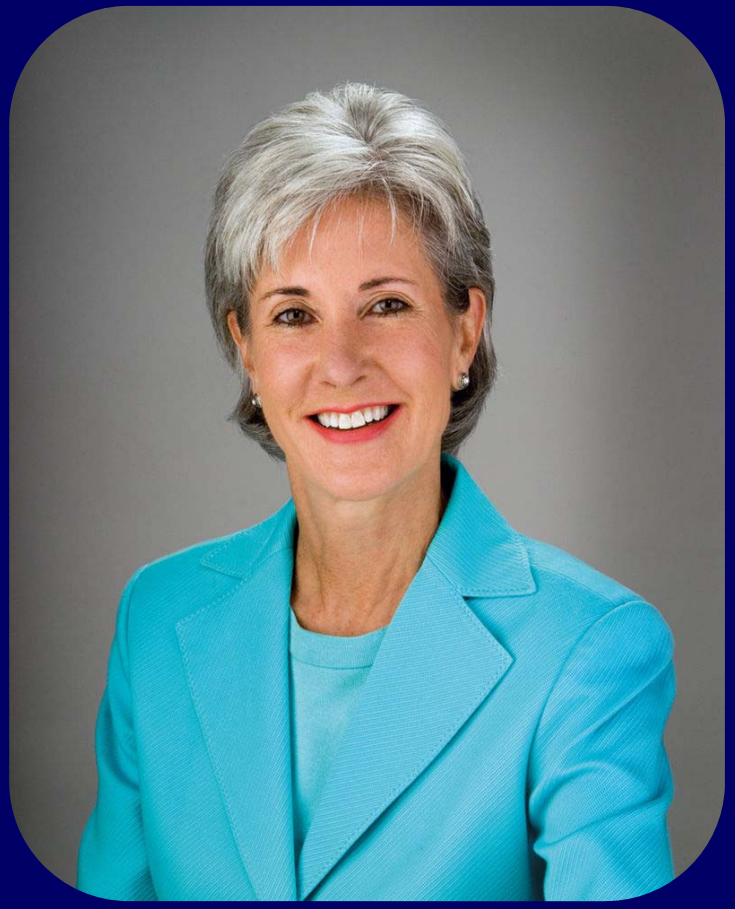
MYRIAD



Document 26

Courts Give NIH hESC Funding the All-Clear

Drs. Sherley and Deisher



Secretary Sebelius

Document 27

NHGRI State Policy Database

Welcome to the Genome Statute and Legislation Database

Share

The Genome Statute and Legislation Database is comprised of state statutes and bills introduced during the 2007-2012 U.S. state legislative sessions. Searchable topics in the database include direct-to-consumer genetic testing, employment and insurance nondiscrimination, health insurance coverage, privacy, research and the use of residual newborn screening specimens.

NHGRI's Table of State Statutes Related to Genomics new provides the total number of states that have enacted legislation on the topics in the database, together with a description of each topic. (In some cases the state legislature has enacted multiple bills pertaining to one topic area. As a result, a database search may yield several statute entries for one state on a particular topic.)

See Also:

Glossary of Statutory, Legislative and Regulatory Terms

On Other Sites:

THOMAS Legislative Information on the Internet

National Conference of State Legislatures

Cornell Legal Information Institute

Note that the database does not contain information about state *regulations*. Some states have developed regulations concerning genetic nondiscrimination in employment and insurance, health insurance coverage, genetic privacy, research and the use of residual newborn screening specimens. To search *regulations* in a particular state, visit the Cornell Legal Information Institute, click on the name of the state, and scroll down to the regulations link.

Definitions of terms such as **"bill"**, **"statute"**, and **"regulation"** are available through the Glossary of Terms.

The database does not include state genetic counselor licensing laws. For information about this topic, visit the National Society of Genetic Counselors.

Search the database:

Content Type:

- State Statute
- 2012 State Bills
- 2011 State Bills
- 2010 State Bills
- 2009 State Bills
- 2008 State Bills
- 2007 State Bills

And/Or Topic:

- Direct-to-Consumer Genetic Testing
- Employment Nondiscrimination
- Health Insurance Coverage
- Health Insurance Nondiscrimination
- Other Lines of Insurance Nondiscrimination
- Privacy
- Research
- Use of Residual Newborn Screening Specimens

And/Or State:

Alabama

Search Tips

- You may select one or more from each box; hold down your "Ctrl" key and click to select multiple.
- Selecting all the choices inside any one box will, by default, return every record in the database.
- The keyword search, by default, combines with all other selections and may result in too narrow a filter. If searching by keywords, it is best not to select from any other boxes at first, then progressively narrow down your result if

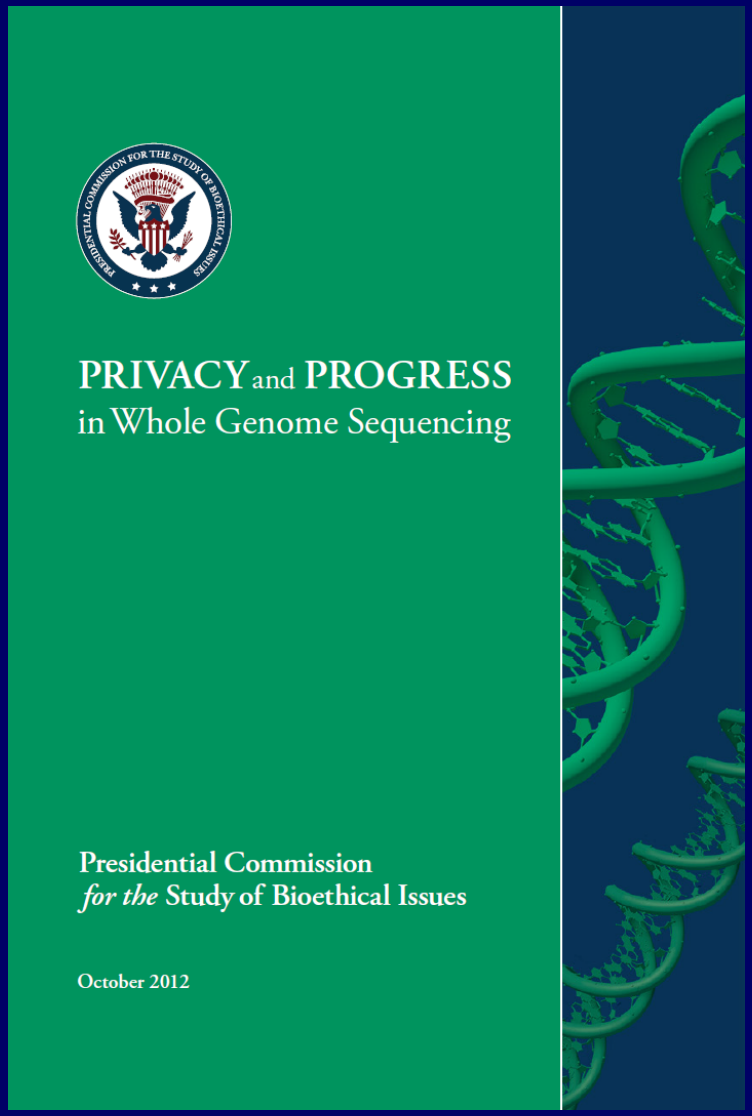
Document 28

GINA Final Rule Published

- Amends HIPAA Privacy Rule to clarify that genetic information is health information
- Prohibits use or disclosure of genetic information for underwriting purposes
- Covers both private and public health insurance

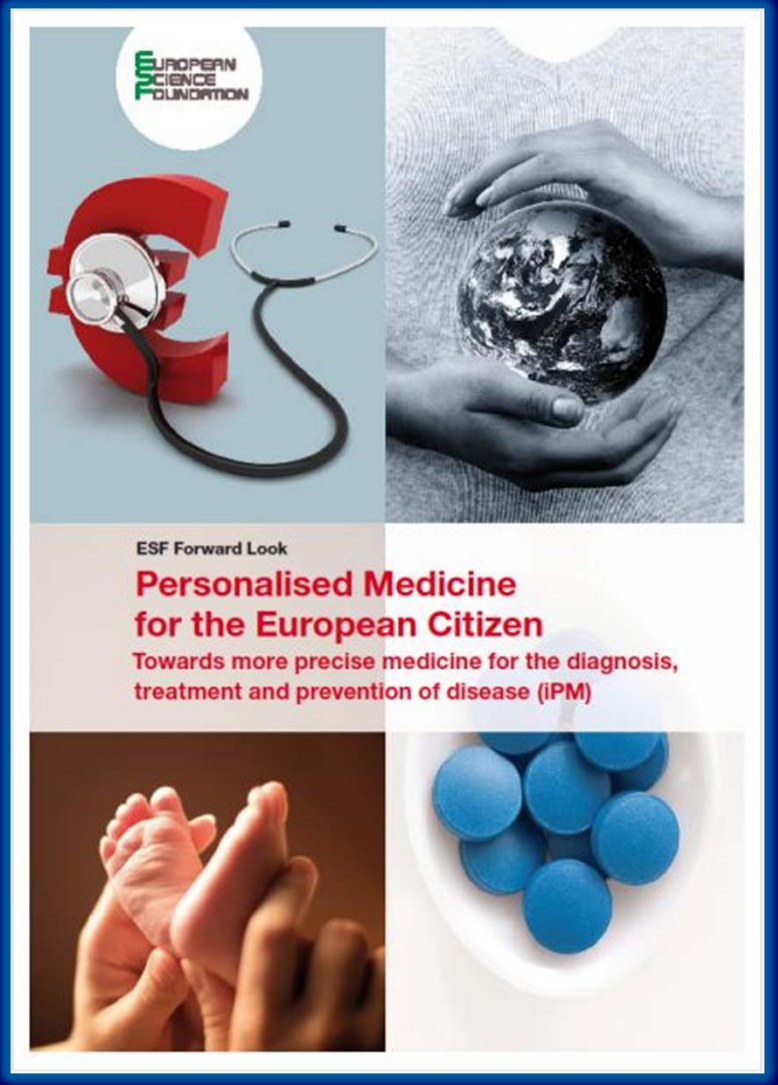
Document 29

Presidential Commission for the Study of Bioethical Issues



Document 30

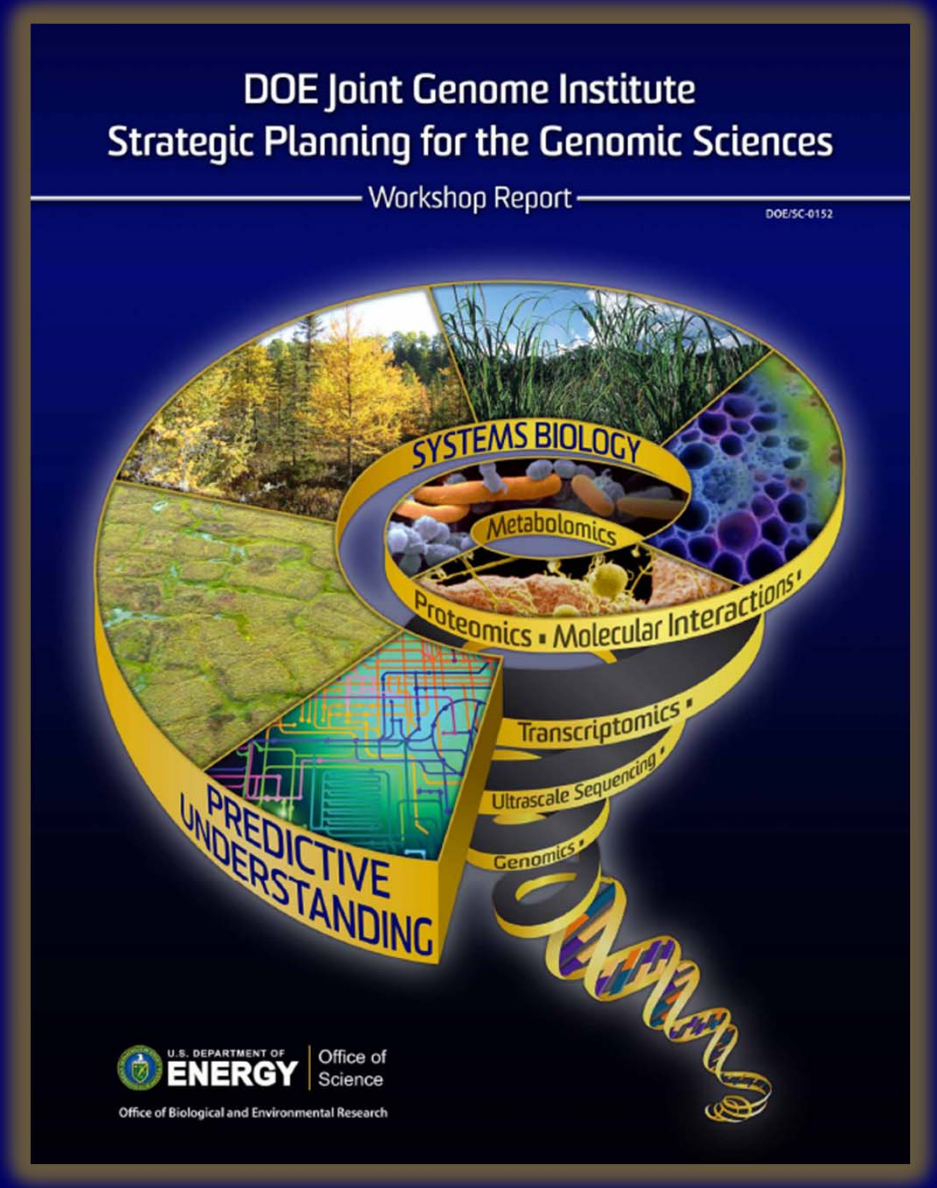
European Science Foundation Report



"…we must gain a clear understanding of what is required to achieve [the promise of personalized medicine] and begin to lay the foundations now that will allow us to benefit in the future."

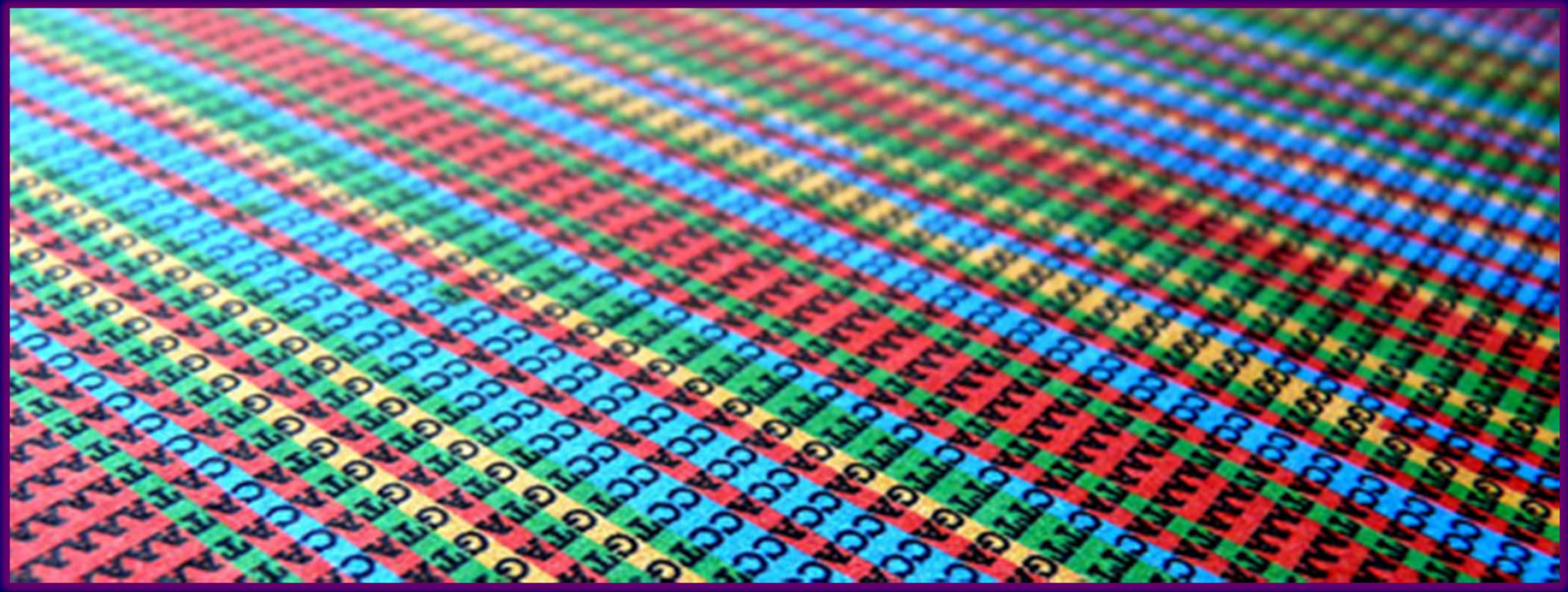
Document 31

Joint Genome Institute Workshop Report



Document 32

100,000 British Genome Sequences



DNA tests to revolutionise fight against cancer and help 100,000 NHS patients

Document 33

NHGRI Genome Advance of the Month

Whole genome sequencing used to track infection's transmission path

By Joy Yang
Post-baccalaureate Fellow

ENCODE: Deciphering Function in the Human Genome

Roseanne F. Zhao, Ph.D.
NIH Medical Scientist Training Program Track 3 Scholar

Do genes make us keep our fat jeans?

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

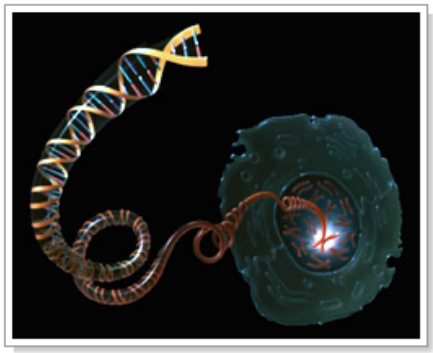
Food for thought

By Joy Yang
Post-baccalaureate Fellow

The Power of Sequencing Single Cell Genomes

Comments Share Print

Roseanne F. Zhao, Ph.D.
NIH Medical Scientist Training Program Track 3 Scholar



As the fundamental unit of life, each cell contains a complete copy of an organism's genome, which can undergo dynamic DNA mutations as the cell grows and divides. Studying the genomes of single cells is important for tracking global patterns of change across hundreds or thousands of individual cells, and will help to elucidate changes that occur in DNA over time. Among other things, this will allow scientists to gain insight into the development of mutations and diversity in the genome, as well as to track genetic changes associated with the origin and progression of different diseases.

In this issue of Genome Advance, we focus on a novel technique that allows researchers to accurately sequence a single cell.

Conventional whole genome sequencing technologies use DNA extracted from large numbers of cells to acquire enough starting material for sequencing. The normal consensus sequence is obtained by aligning, or

Document 34

Genomics In The News…

Science Top 10 Breakthroughs in 2012

GENOMICS BEYOND GENES

A decadelong, $288 million study reported this year in more than 30 papers showed the human genome to be quite a bustling place, biochemically speaking. The work—called the Encyclopedia of DNA Elements (ENCODE)—builds on the Human Genome Project, which deciphered the order of the bases that are our DNA's building blocks and found that less than 2% of those bases defined genes.

ENCODE researchers took an intensive look not just at genes but at all of the DNA in between. Their results drive home that much of the genome that at one time was dismissed as "junk DNA" actually seems to play an essential role, often by helping to turn genes on or off. They pinpointed hundreds of thousands of landing spots for proteins that influence gene activity, many thousands of stretches of DNA that code for different types of RNA, and lots of places where chemical modifications serve to silence stretches of our chromosomes, concluding that 80% of the genome was biochemically active. These details provide a much better road map for investigators trying to understand how genes are controlled. Some researchers have already used these insights to clarify genetic risk factors for a variety of diseases, including multiple sclerosis and Crohn's disease.

RUNNER-UP

When these papers were published in September, the media went wild. ENCODE was hailed in *The New York Times* as a "stunning resource" and "a major medical and scientific breakthrough" with enormous and immediate implications for human health. *The Guardian* called it "the most significant shift in scientists' understanding of the way our DNA operates since the sequencing of the human genome."

But several scientists in the blogosphere called the coverage overhyped and blamed the journals and ENCODE leaders for overplaying the significance of the results. For example, ENCODE reported that 76% of DNA is transcribed to RNA, most of which does not go on to help make proteins. Various RNAs home in on different cell compartments, as if they have fixed addresses where they operate, suggesting that they play a role in the cell. Critics argue, however, that it was already known that a lot of RNA was made, and that many of these RNAs may be spurious genome products that serve no purpose. Likewise, one ENCODE researcher found 3.9 million regions across 349 types of cells where proteins called transcription factors bind to the genome—but again, it's unclear how much of that binding is functional.

Nonetheless, ENCODE stands out as an important achievement that should ease the way for more insights into the genome. By combining these data with sampling from another data-intensive effort, the 1000 Genomes Project, researchers discovered that 8% of our DNA appears with little variation throughout the human population—a strong sign that it was important for our evolution. Overall, ENCODE's newly discovered functional regions overlap with 12% of the specific DNA bases linked to higher or lower risks of various diseases, suggesting that the regulation of genes—not just the makeup of the genes themselves—might be at the heart of these risks. Scientists have used this information to home in on relevant genes and cell types in several disorders. Experiments can now unearth the molecular basis of these connections and, from there, identify potential treatments. If that potential is realized, then ENCODE will have earned its accolades as a "stunning resource."

ENCODE BY THE NUMBERS

147	cell types studied
80%	functional portion of human genome
20,687	protein-coding genes
18,400	RNA genes
1640	data sets
>30	simultaneous papers
442	researchers
$288 MILLION	funding for pilot, technology, model organism, and current project

ENCODE

Science

AAAS

Document 35

Genomics In The News…

THE WALL STREET JOURNAL

The Future of Medicine Is Now

From cancer treatments to new devices to gene therapy, a look at six medical innovations that are poised to transform the way we fight disease

#2: Genome Sequencing for Routine Checkups

#3: Matching a Tumor to a Drug

#6: Rejigging Your Genes

Document 35

Time: Top 10 Medical Breakthroughs in 2012

Top 10 Medical Breakthroughs

8. Decoding Childhood Tumors

Top 10 Medical Breakthroughs

7. Speeding DNA-Based Diagnosis for Newborns

Top 10 Medical Breakthroughs

6. Breaking Down Breast Cancer

Top 10 Medical Breakthroughs

2. What Are Bugs For?

Top 10 Medical Breakthroughs

1. Junk No More

By Alice Park | Dec. 04, 2012 | Add a Comment

After being ignored as useless genetic garbage, the vast 98% of the human genome that does not code for genes finally has a purpose. It turns out that these previously insignificant portions of DNA are the true genetic masterminds, or metabolic switches that regulate how and when genes function as well as how prolifically genes churn out their respective proteins. Without them, scientists say, genes would be like a jumbled mess of words that have no meaning. Scientists are already exploiting the newly discovered trove of biological information and pursuing new ways of controlling, and possibly even curing, diseases with the flick of a genetic switch.

GETTY IMAGES

Document 35

Genomics In The News…

Document 35

Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

IV. NHGRI Extramural Research Program

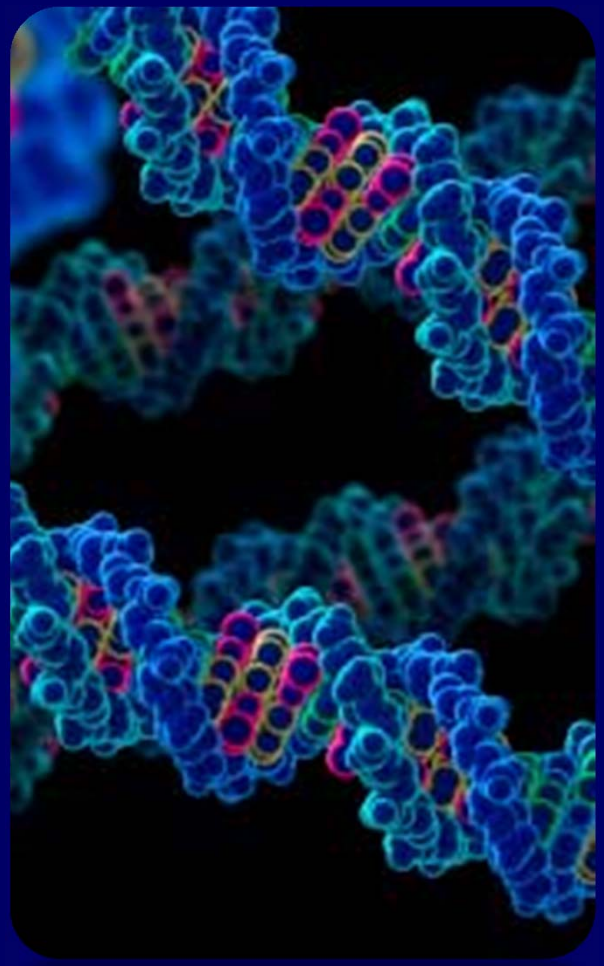
V. NIH Common Fund Programs

VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

Large-Scale Genome Sequencing and Analysis Centers

- 310 Tb generated in last grant year
- Over 120 ongoing projects: cancer, complex disease, rare diseases, and comparative sequencing
- Over 20 papers published or in press this quarter

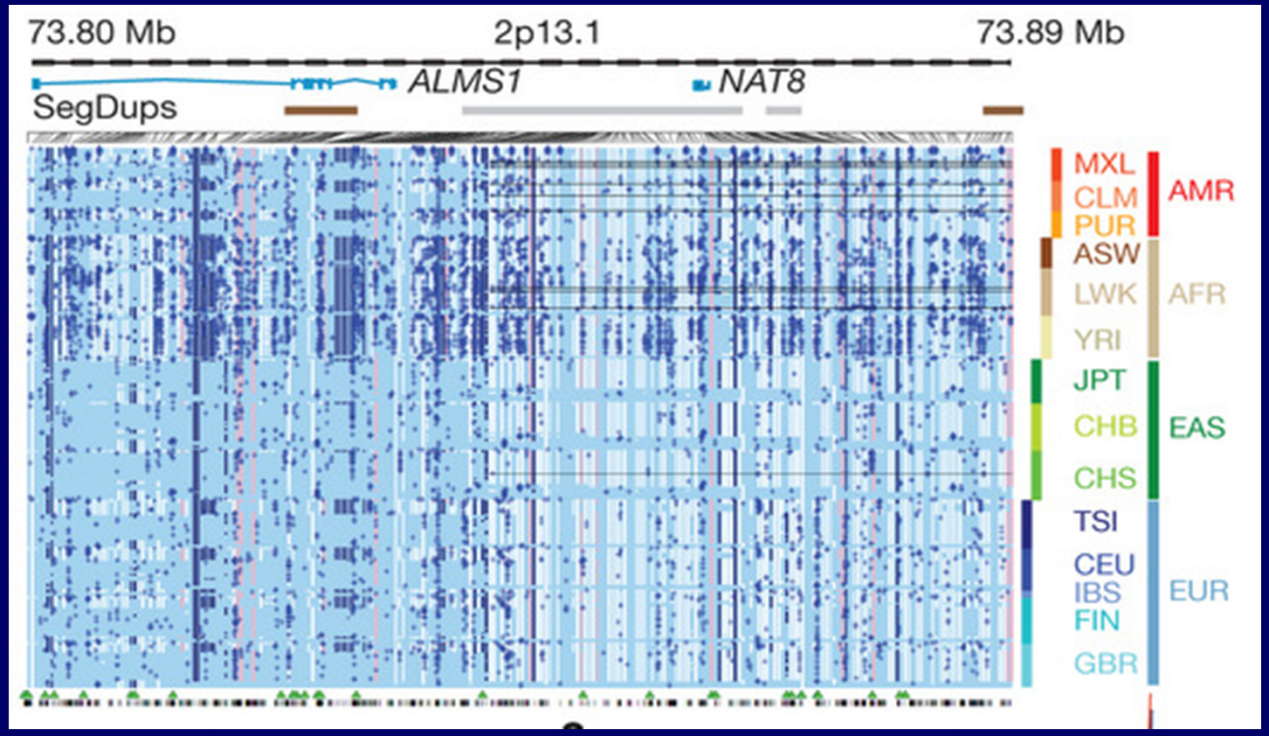


Document 36

1000 Genomes

A Deep Catalog of Human Genetic Variation

- **Phase 1 paper published in *Nature***
- **Meeting and tutorial at 2012 ASHG annual meeting**
- **Phase 3 sequencing will be complete by March**



- **2nd Annual TCGA Scientific Symposium (November 2012)**

  Two days of presentations, posters, and workshops

  >500 participants

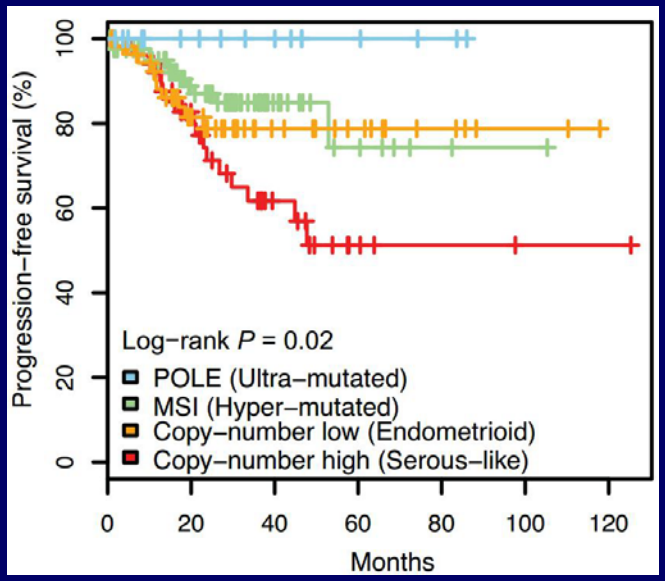
  Video at genome.gov

- **NCI-NHGRI Cancer Genomics Strategic Planning Meeting (November 2012)**

Since the last Council meeting:

- "Comprehensive molecular portraits of human breast tumours" *Nature* (2012)

- Papers under review:
  - Renal Clear Cell Carcinoma
  - Endometrial Carcinoma
  - Acute Myeloid Leukemia



No recurrence of cancer for endometrial tumors containing *POLE* mutations

Centers for Mendelian Genomics

Finding the genes underlying human Mendelian conditions

Disease Gene Discovery in Year 1

- >6000 whole-exome sequences
- >150 rare inherited diseases
- Discovery of the genes for >20 diseases
- ~12 manuscripts published/accepted

Outreach and Coordination

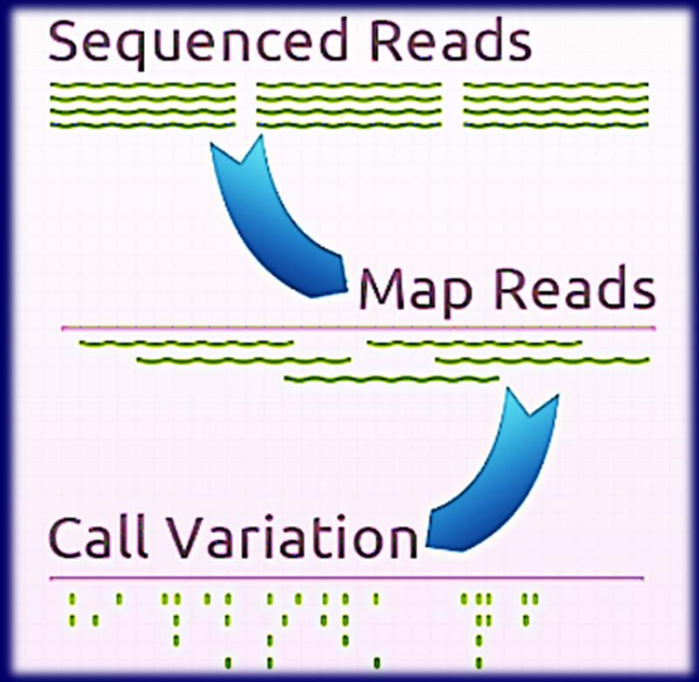
- Public phenotype lists
- Public release of genomic data
- International Rare Diseases Research Consortium
- Session at 2012 ASHG Annual Meeting

Clinical Sequencing Exploratory Research (CSER) Projects

- All CSER projects are now recruiting patients; several have begun to return results
- Manuscripts in preparation on topics such as variant actionability and sequencing standards for clinical use
- *Time* cover story feature CSER groups
- Applications currently in review for re-issued CSER RFA and new Coordinating Center RFA

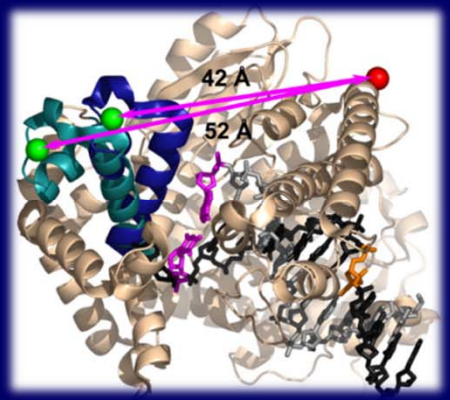
Genome Sequencing Informatics Tools

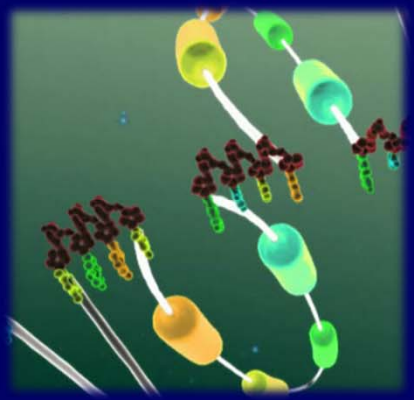
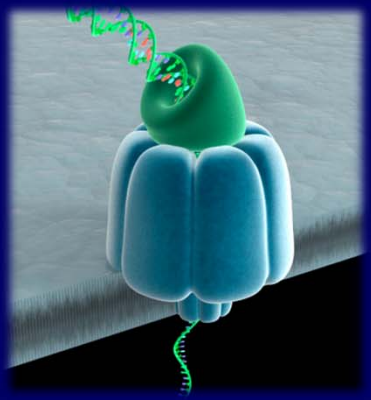
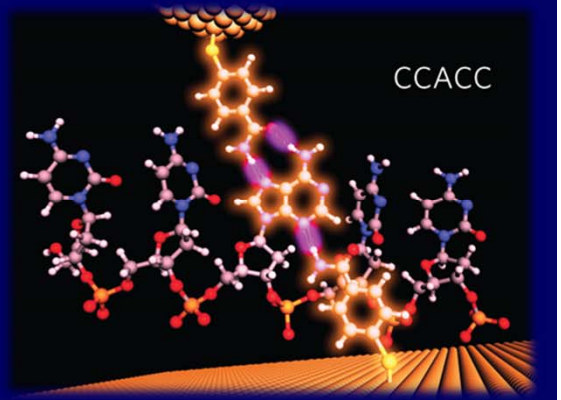
- GS-IT Program provides ‘researcher-friendly’ sequence analysis tools from six projects
- An ‘iSeqTools” Portal grant supplement has been funded to build a program-wide interface
- The Portal will use innovative visual strategies to simplify integrated analysis with GS-IT tools



DNA Sequencing Technology Development

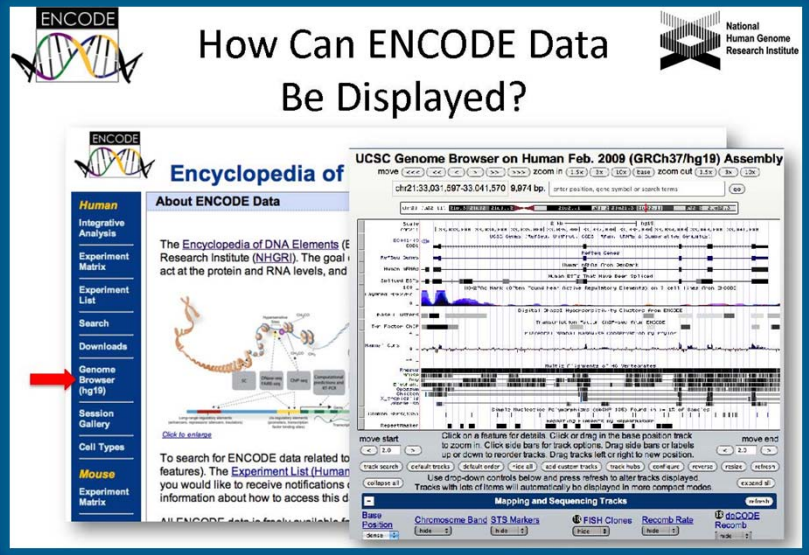
- Two instruments released ⟶ 'genome-in-a-day' later this year?
- Grant awards made in the Revolutionary Sequencing Technology Program
- Grantee meeting in the Spring, including a public meeting May 1-2, 2013



Document 41

ENCODE



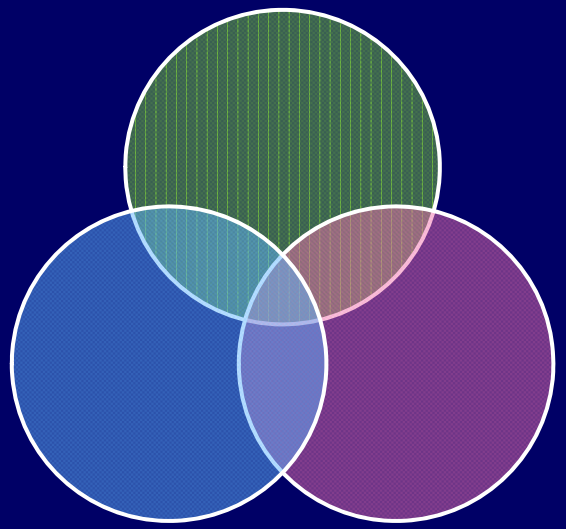
- ENCODE/Common Fund Epigenomics Workshop at 2012 ASHG Annual Meeting
- ENCODE Phase 3 grants funded: September 2012
- ENCODE Production PI Meeting: December 2012
- ENCODE Consortium Meeting: May 2013

Document 42

ENCODE Analysis

Cross-species comparison papers planned:

- **modENCODE– comparison of fly, worm, and human data**
- **mouse ENCODE– comparison of mouse and human data**



CEGS Program

- Grantee meeting at UNC in October 2012
- To discuss CEGS Program continuation today
- Applications considered in Closed Session

Document 43

Genomic Medicine I Colloquium Report

REVIEW | Genetics inMedicine

Open

Implementing genomic medicine in the clinic: the future is here

Teri A. Manolio, MD, PhD[1], Rex L. Chisholm, PhD[2], Brad Ozenberger, PhD[1], Dan M. Roden, MD[3], Marc S. Williams, MD[4,5], Richard Wilson, PhD[6], David Bick, MD[7], Erwin P. Bottinger, MD[8], Murray H. Brilliant, PhD[9], Charis Eng, MD, PhD[10], Kelly A. Frazer, PhD[11], Bruce Korf, MD, PhD[12], David H. Ledbetter, PhD[5], James R. Lupski, MD, PhD[13], Clay Marsh, MD[14], David Mrazek, MD[15], Michael F. Murray, MD[16], Peter H. O'Donnell, MD[17], Daniel J. Rader, MD[18], Mary V. Relling, PharmD[19], Alan R. Shuldiner, MD[20], David Valle, MD[21], Richard Weinshilboum, MD[22], Eric D. Green, MD, PhD[1] and Geoffrey S. Ginsburg, MD, PhD[23]

Document 44

Recent Advances in Genomic Medicine

RESEARCH ARTICLE

RESEARCH ARTICLE

RESEARCH ARTICLE

ORIGINAL ARTICLE

Aspirin Use, Tumor *PIK3CA* Mutation, and Colorectal-Cancer Survival

Xiaoyun Liao, M.D., Ph.D., Paul Lochhead, M.B., Ch.B., Reiko Nishihara, Ph.D., Teppei Morikawa, M.D., Ph.D., Aya Kuchiba, Ph.D., Mai Yamauchi, Ph.D., Yu Imamura, M.D., Ph.D., Zhi Rong Qian, M.D., Ph.D., Yoshifumi Baba, M.D., Ph.D., Kaori Shima, D.D.S., Ph.D., Ruifang Sun, M.B., Katsuhiko Nosho, M.D., Ph.D., Jeffrey A. Meyerhardt, M.D., M.P.H., Edward Giovannucci, M.D., M.P.H., Sc.D., Charles S. Fuchs, M.D., M.P.H., Andrew T. Chan, M.D., M.P.H., and Shuji Ogino, M.D., Ph.D.

isease

er Saffrey,[6]

ne Safina,[14]

Document 44

Genomic Medicine IV

- **Main goals:**
  - **Identify ongoing efforts and current needs in physician education in genomics**
  - **Understand processes for guideline development**
- **Major outcomes:**
  - **Convene a Coordinating Committee**
  - **Collect current approaches and develop and disseminate best practices**

Document 44

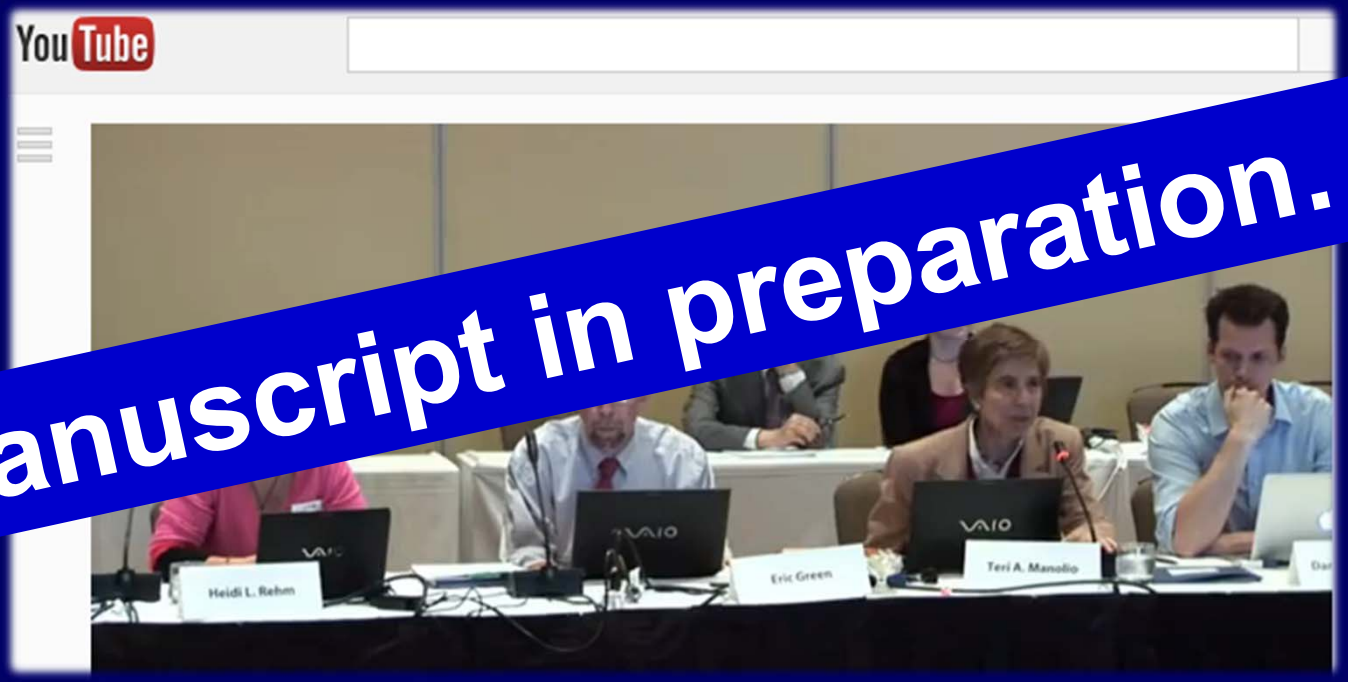
Reimbursement Models to Promote Evidence Generation and Innovation for Genomic Tests

- Meeting held in October 2012
- Attended by members of Genomic Medicine Working Group and stakeholder representatives
- Explored coupling coverage and evidence development
- Action items being pursued

Document 44

Implicating Sequence Variants in Human Disease Workshop: September 2012

Goal: To develop guidelines for assessing the evidence implicating sequence variants or genes as causal in a specific disease



Document 45

The eMERGE Network

electronic Medical Records & Genomics

A consortium of biorepositories linked to electronic medical records data for conducting genomic studies

High Density GWAS for LDL Cholesterol in African Americans Using Electronic Medical Records Reveals a Strong Protective Variant in *APOE*

Laura J. R
Jennifer A
M.D., Ph.D
Marylyn D
Kho M.D.,
M. Geoff H
M.Eng.[6],
John T. W
Zuvich Ph
, Dan M.
M.D., M.S
Christoph
Kullo M.D
M.P.H.[14],
M.P.H.[15],
, Teri A. Manolio M.D
Crawford Ph.D.[7], Re

The American Journal of Human Genetics *89*, 529–542, October 7, 2011

ARTICLE

Variants Near *FOXE1* Are Associated with Hypothyroidism and Reco

Joshua
Melissa
Peggy
Luke R
Noah
Ronglin
Eric B.

Hum Genet (2012) 131:639–652
DOI 10.1007/s00439-011-1103-9

ORIGINAL INVESTIGATION

Genetic variants associated with the white blood cell count in 13,923 sub

David R. Crosslin · A
Mariza de Andrade
Abel Kho · M. Geoff
Paul K. Crane · Kath
Chris S. Carlson · G

©American College of Medical Genetics and Genomics

COMMENTARY | Genetics inMedicine

Leveraging the electronic health record to implement genomic medicine

Iftikhar J. Kullo, MD[1], Gail P. Jarvik, MD, PhD[2,3], Teri A. Manolio, MD, PhD[4], Marc S. Williams, MD[5] and Dan M. Roden, MD[6,7]

Document 46

Genomics And Randomized Trials NETwork (GARNET)

- **GARNET randomized clinical trials:**

  SUCCESS-A Breast Cancer Trial

  Vitamin Intervention for Stroke Prevention Trial (VISP)

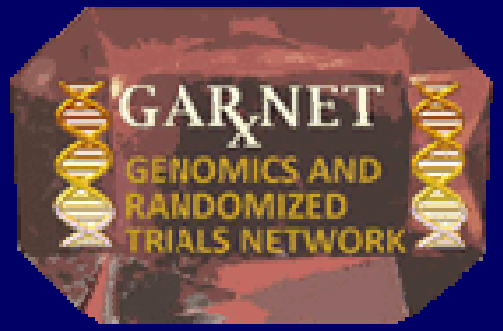
  Women's Health Initiative Hormone Therapy Trial (WHI)

- **30 manuscripts:**

  9 published

  8 accepted or submitted

  13 in preparation



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

- Genomics and Society Working Group established
- Centers of Excellence in ELSI Research (CEER) applications reviewed
- Return of Research Results Consortium to hold joint meeting with CSER in May 2013

Document 48

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

Molecular Libraries Program (MLP)

Strategies to transition MLP initiatives off of Common Fund support:

- **Continue with NIH Institute/Center support**
- **Close out completed initiatives**
- **Transition to private sector support**

MLP: Maintain Program Resources

- **Small Molecule Repository (SMR)**
  - **Partial support from NCATS**
  - **Public access to compounds at cost**
- **PubChem Database support from NCBI**
- **Assay guidance information at NCATS**
- **Probe Reports on NCBI Bookshelf**
- **BioAssay Research Database at NCATS**

Human Microbiome Project (HMP)

- **Applications received for HMP2**

  $15M, 3 years

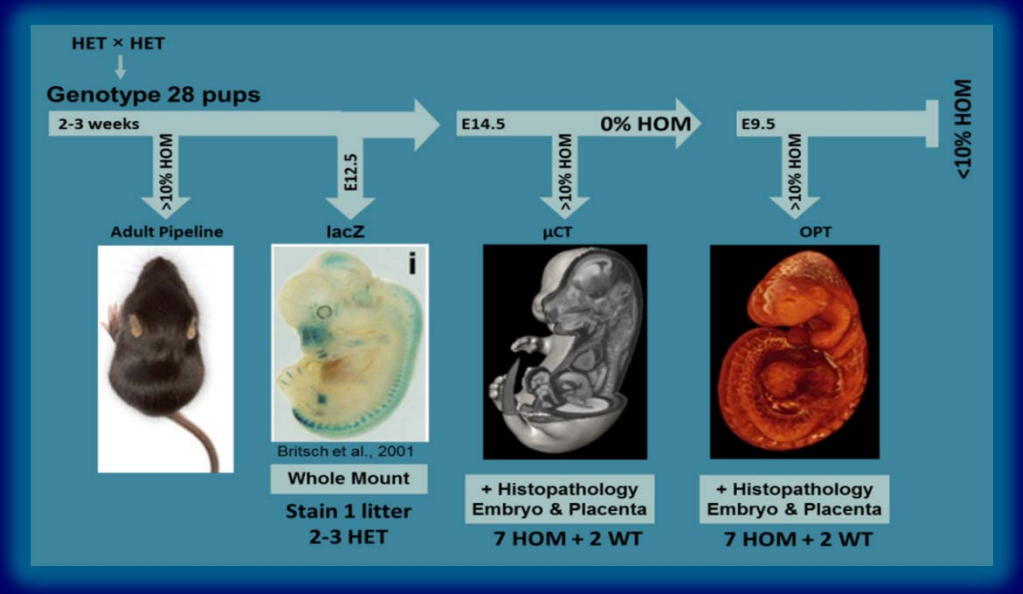
  Starting in Fiscal Year 2013

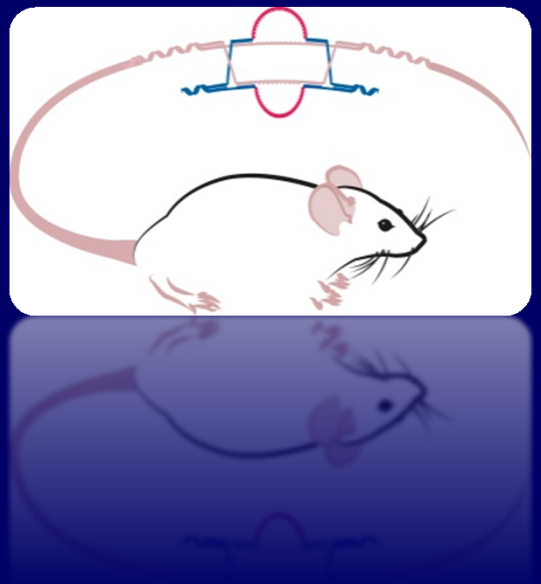
- **Trans-NIH Microbiome Working Group**

  64 members, 16 Institutes/Centers

- **2013 HMP Meeting: “NIH Human Microbiome Science: Vision for the Future”**

Knockout Mouse Phenotyping Project (KOMP^2)



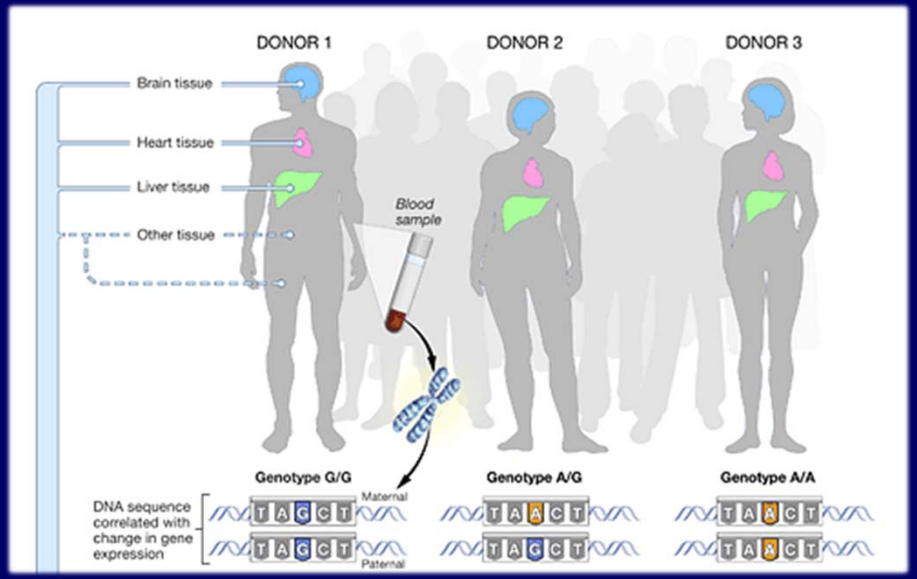


- Supplemental funds for phenotyping of embryonic lethal knockouts
- High resolution microCT and Optical Projection Tomography (OPT)
- $9.6M to be awarded over 3 years

Document 51

Genotype-Tissue Expression Project (GTEx)

- GTEx moves from pilot to scale up
- eQTL Browser and Data Portal are live



- RFA: Enhancing GTEx with molecular analyses of stored biospecimens (U01)
- Full program update later today

Document 52

Library of Integrated Network-based Cellular Signatures (LINCS)

- 2nd Annual Consortium Meeting occurred in November 2012
- External Scientific Panel recommendations
- Lincsproject.org: LINCS data and tools
- LINCS Data Forum: March 2013 (Boston)

H3Africa

Inaugural Meeting of the H3Africa Consortium held in Ethiopia in October 2012

- Official press announcement of awards
- H3Africa Steering Committee convened
- 10 working groups created

Document 54

Undiagnosed Diseases Program

- **RFA-RM-12-020: Coordinating Center for an Undiagnosed Diseases Network (U01)**

  Applications received; upcoming review

- **PA-13-076: Gene Function Studies to Investigate Rare and Undiagnosed Diseases (administrative supplement)**

  Receipt date: February 26, 2013

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V. NIH Common Fund Programs

VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

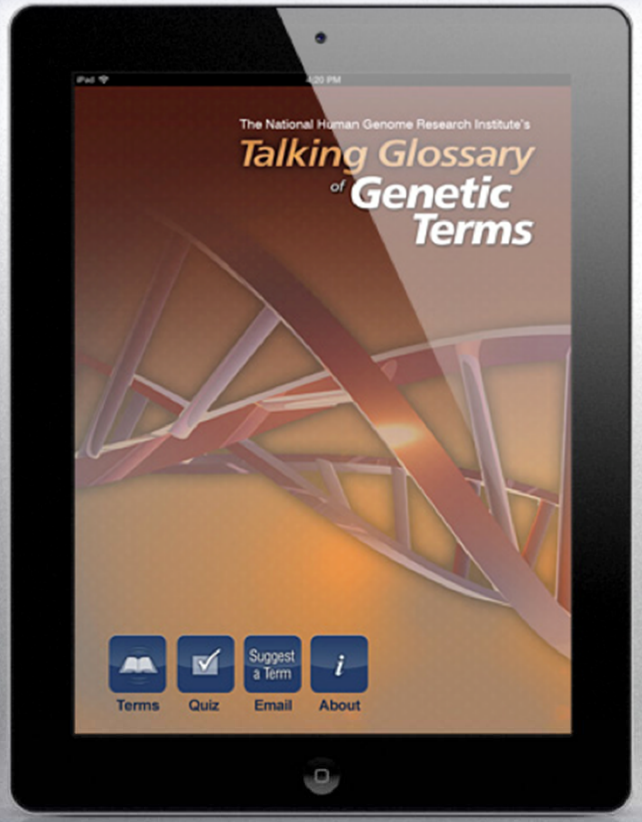
Genomics in Medicine Lecture Series

Monthly seminars on intersection of genomics and medicine held at Suburban Hospital

Daniel Kastner, M.D., Ph.D.

Document 56

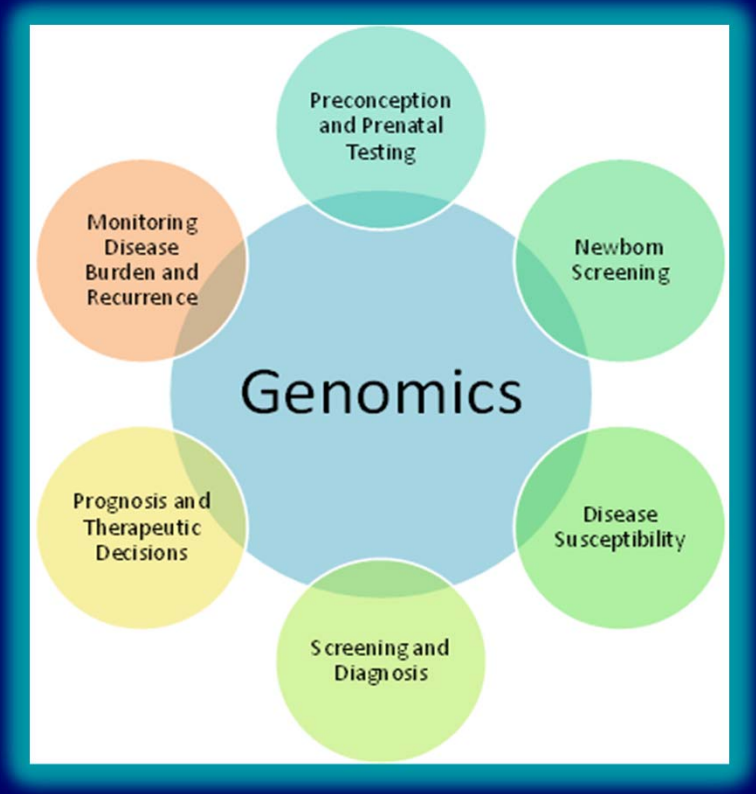
iPad App for NHGRI
Talking Glossary of Genetic Terms



Document 57

Journal of Nursing Scholarship

- 2013 Genomics Special Issue
- Articles available online
- Editorial: “Relevance of genomics to healthcare and nursing practice”
- Closing Paper: “A blueprint for genomic nursing science”
- Webinars coming soon



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Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

IV. NHGRI Extramural Research Program

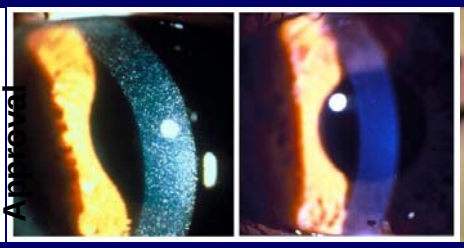
V. NIH Common Fund Programs

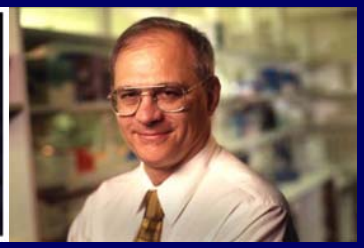
VI. NHGRI Division of Policy, Communications, and Education

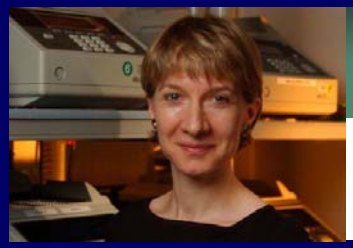
VII. NHGRI Intramural Research Program

NHGRI Intramural Research Highlights

FDA Approval





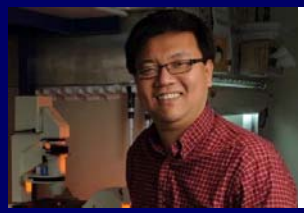


nature genetics

Exome sequencing of serous endometrial tumors identifies recurrent somatic mutations in chromatin-remodeling and ubiquitin ligase complex genes

BMC Biology IMPACT FACTOR 5.75

Genomic organization, evolution, and expression of photoprotein and opsin genes in Mnemiopsis leidyi: a new view of ctenophore photocytes



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nature genetics

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