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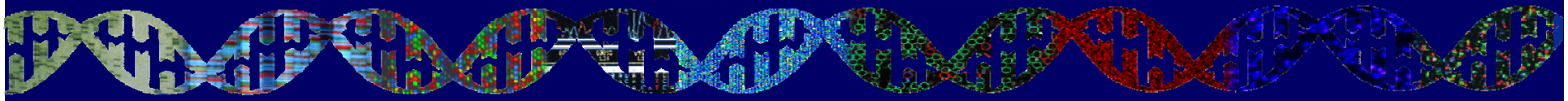
National Human Genome Research Institute
National Institutes of Health

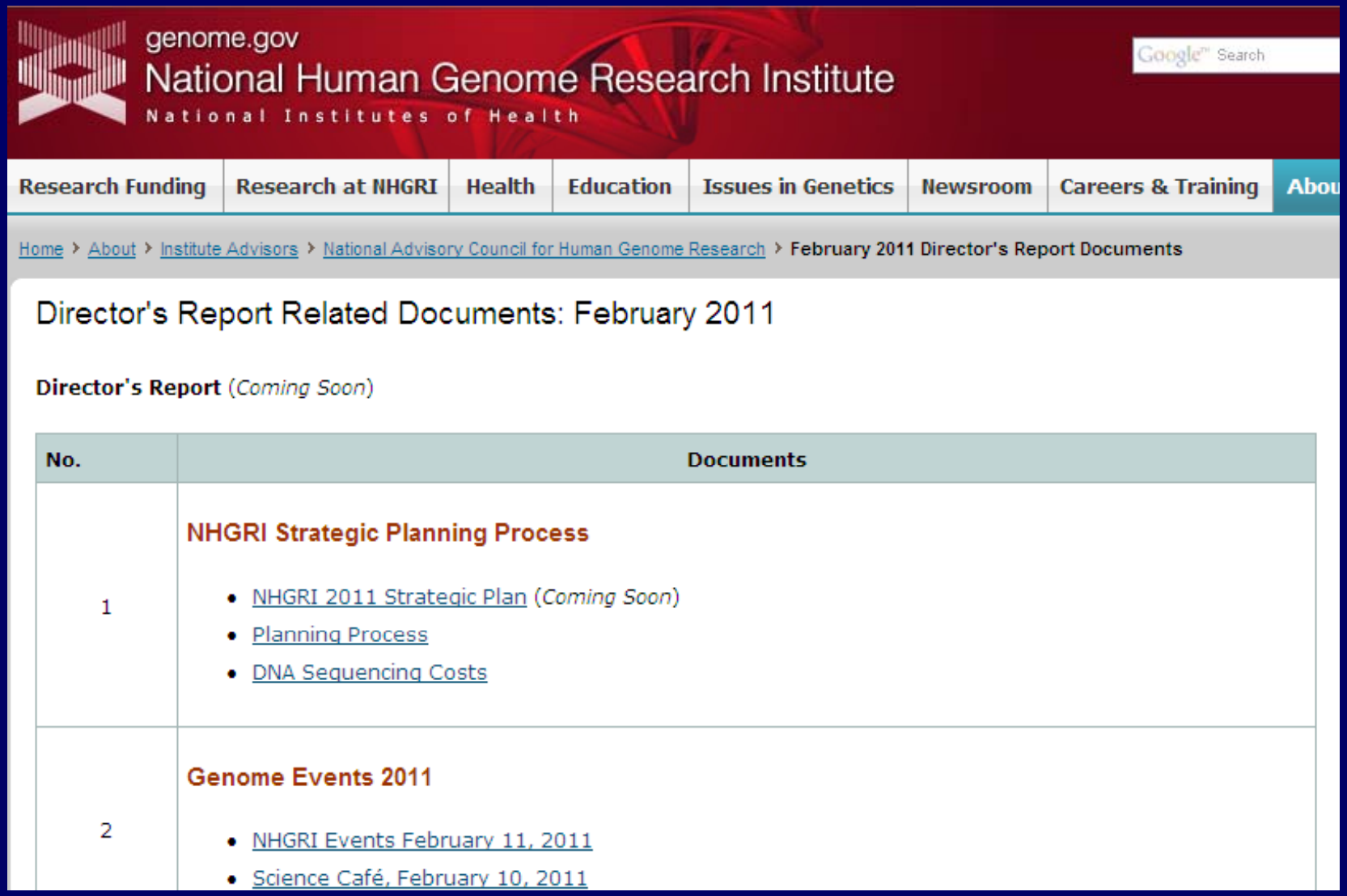
DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

February 2011

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





genome.gov/DirectorsReport

Document #

I. General NHGRI Updates

II. General NIH Updates

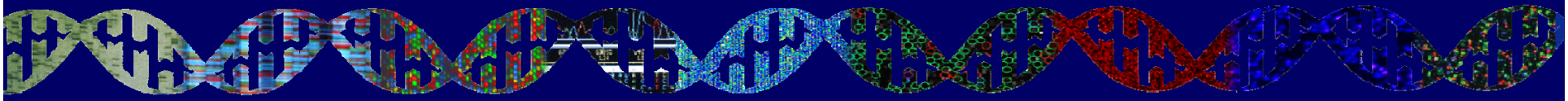
III. Genomics Updates

IV. NHGRI Extramural Program

V. NIH Common Fund Programs

VI. NHGRI Office of the Director

VII. NHGRI Intramural Program



Open Session Presentations

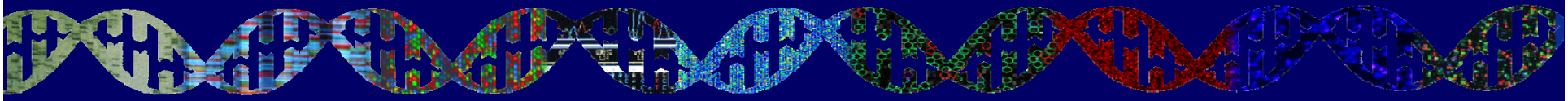
- The NIH Common Fund: James Anderson
- ELSI Grants and CSR: Rudy Pozzatti
- Scientific Presentation: Emerging Ethical Issues in Genome Research
- Concept Clearance:

  ENCODE – Mike Pazin
- Program Updates:

  TCGA – Brad Ozenberger

  LINCS – Ajay Pillai

  Molecular Libraries – Carson Loomis



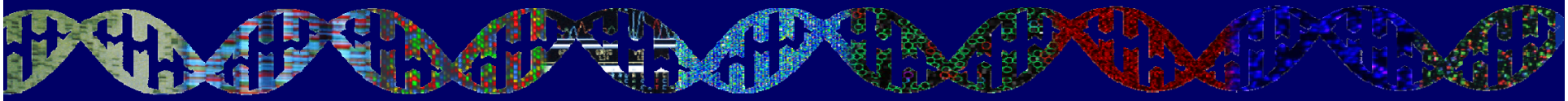
Open Session Presentations

- **Meeting Reports:**

  Protein Capture – Adam Felsenfeld

  Newborn Screening in the Genomics Era – David Valle

- **Population Tracking: Anna Rossoshek**



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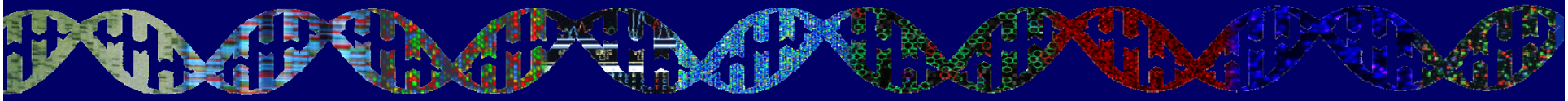


NHGRI Publishes 2011 Strategic Plan

PERSPECTIVE

doi:10.1038/nature09764

Charting a course for genomic medicine from base pairs to bedside

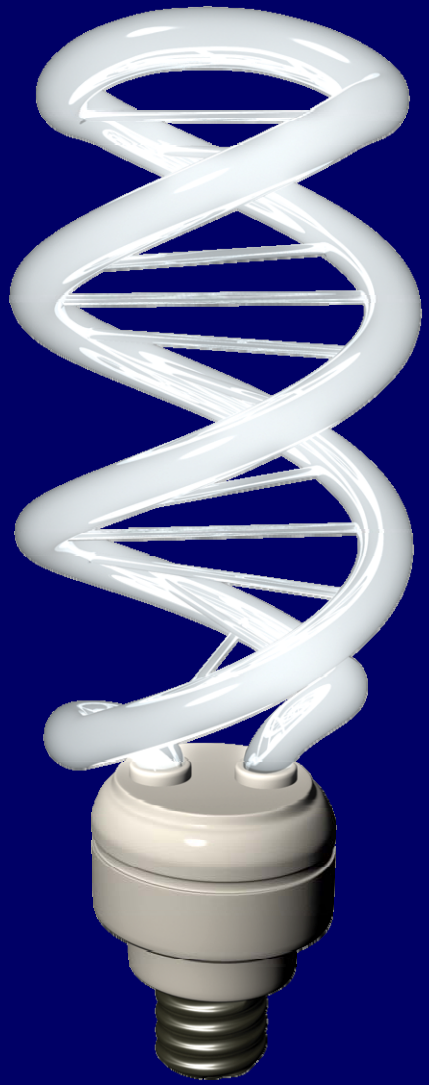
Eric D. Green[1], Mark S. Guyer[1] & National Human Genome Research Institute*

There has been much progress in genomics in the ten years since a draft sequence of the human genome was published. Opportunities for understanding health and disease are now unprecedented, as advances in genomics are harnessed to obtain robust foundational knowledge about the structure and function of the human genome and about the genetic contributions to human health and disease. Here we articulate a 2011 vision for the future of genomics research and describe the path towards an era of genomic medicine.

genome.gov/SP2011

Document 1

Illuminating the Path to Genomic Medicine



nature

THE INTERNATIONAL WEEKLY JOURNAL OF SCIENCE

THE FUTURE IS BRIGHT

Reflections on the first ten years of the human genomics age

GENOMICS

THE END OF THE BEGINNING

Eric Lander on the impact of the human genome sequence

PAGE 187

METHODS

MORE BASES PER DOLLAR

Elaine Mardis on the march of sequencing technology

PAGE 198

HEALTH

FROM LAB TO CLINIC

A road map to genomic medicine

PAGE 204

NATUREASIA.COM

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Vol. 470, No. 7333

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doi:10.1038/nature09764

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Eric D. Green[1], Mark S. Guyer[1] & Natio

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Opportunities for understanding h
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contributions to human health an
describe the path towards an era o

Since the end of the Human Genome P
publication of a reference human geno
become a mainstay of biomedical rese
nity's foresight in launching this ambitious pr
range of scientific advances that the HGP ha
(see rollfold). Optimism about the potential c
improving human health has been fuelled by
the molecular basis of inherited diseases (h
omim.and http://www.genome.gov/GWAStu
variation in disease[8], some of which have aln
Other advances have already changed medical
arrays are now used for clinical detection o
pharmacogenomic testing is routinely perfo
of certain medications[15]). Together, these achi
paper[16]) document that genomics is contribut
of human biology and to improving human

As it did eight years ago[17], the National
Institute (NHGRI) has engaged the scientif
genome.gov/Planning) to reflect on the key a
and explore future directions and challenge
sions have led to an updated vision that focu
biology and the diagnosis, prevention and t
including consideration of the implications
(but these discussions, intentionally did not
in agriculture, energy and other areas). Liketh
is broader than what any single organizatio
realizing the full benefits of genomics will be

This 2011 vision for genomics is organized
ing from basic research to health application
that, over time, the most effective way to i
understand normal biology (in this case, ge
understanding disease biology, which then bec
health. At the same time, there are other conse
Genomics offers opportunities for improvin
understanding of disease (for example, canc
based on genomic profiles that identify tumo
discoveries can lead back to understanding d

The past decade has seen genomics contrib
about biology and its perturbation in dise
understanding will accelerate the transition t
care based on genomic information). But sig

[1]National Human Genome Research Institute, National Insti
*Lists of participants and their affiliations appear at the end

REVIEW

doi:10.1038/nature09792

Initial impact of the sequencing of the human genom

Eric S. Lander[1]

The sequence of the human genome has dr
decade since its publication, on our unders
basis of inherited diseases and cancer, and d
in fulfilling the promise of genomics for m

On 15 February 2001, a decade ago this week,
62-page paper entitled 'Initial sequencing a
human genome', reporting a first global look
the human genetic code. The paper[1] marked a mile
national Human Genome Project (HGP), a discovery
ceived in the mid-1980s and launched in 1990. The s
published a paper[2] from the company Celera Genc
draft human sequence based on their own prodigio
data from the public HGP.

The human genome has had a certain tendency to
excess: from early jeremiads that the HGP would st
consuming the NIH budget (it never rose to more thar
coverage of a late-breaking genome race between p
protagonists; to a White House announcement of
sequence in June 2000, 8 months before scientific p
been written, peer-reviewed and published; to breathl
Wall Street and the press about the imminence of ge
and genome-based panaceas; to a front-page news
anniversary of the announcement that chided genom
yet having cured most diseases.

The goal of this review is to step back and assess the
from a scientific standpoint, addressing three questi
learned about the human genome itself over the past
the human sequence propelled our understanding
medicine, evolution and history? What is the road al

The past decade has shown the power of genomic m
for biomedical research. By providing a comprehe
human sequence has made it possible for scientists
fragmentary information into landscapes of biolog
function: maps of evolutionary conservation, gene tr
matin structure, methylation patterns, genetic variatio
distance, linkage disequilibrium, association to inherit
alterations in cancer, selective sweeps during human
dimensional organization in the nucleus. By providi
cross-reference information across species, it has con
of model systems to the physiology of the human. Fu
viding comprehensive catalogues of genomic informa
genes and proteins to be recognized based on unique 't
example, RNA transcripts to be assayed with arrays
probes and proteins by detection of short peptide fr
spectrometer. In turn, these measurements have been
'cellular signatures' characteristic of specific cell types, st
and catalogues of the contents of organelles such as

[1]Broad Institute of MIT and Harvard, 7 Cambridge Center, Cambridge, M

PERSPECTIVE

doi:10.1038/nature09796

A decade's perspective on DNA sequencing technology

Elaine R. Mardis[1]

The decade since the Human Genome Project ended has witnessed a remarkable sequencing technology explosion that has permitted a multitude of questions about the genome to be asked and answered, at unprecedented speed and resolution. Here I present examples of how the resulting information has both enhanced our knowledge and expanded the impact of the genome on biomedical research. New sequencing technologies also have introduced exciting new areas of biological endeavour. The continuing upward trajectory of sequencing technology development is enabling clinical applications that are aimed at improving medical diagnosis and treatment.

The sequencing of the Human Reference Genome, announced ten years ago, provided a roadmap that is the foundation for modern biomedical research. This monumental accomplishment was enabled by developments in DNA sequencing technology that allowed data production to far exceed the original description of Sanger sequencing[1]. Moving forward in the genomic era in which we now find ourselves, new (or 'next generation') DNA sequencing technology is enabling revolutionary advances in our understanding of health and disease. In essence, sequencing technology is the engine that powers the car that allows us to navigate the human genome roadmap. As that engine becomes ever more powerful, so will the questions we can ask and answer about the geography of our genetic landscape.

Of course, a car with only an engine is unworkable; as such, DNA sequencing technology provides an integral part of a larger system, one with multiple components that must be properly matched in order to achieve high throughput and efficiency. It has essentially never been as 'easy' as simply buying sequencing instruments, plugging them in, and generating data. We need the raw materials, such as fuel (DNA), sparks to ignite the fuel (reagents), mechanical parts to translate fuel and ignition into movement (robotics) and direction (bioinformatics), all working in a carefully engineered balance, and a driver (genome centre) to steer the automobile quickly and efficiently to the desired destination (biological understanding). By inference, as this 'engine' has achieved ever increasing horsepower, the supporting components have evolved to match its output with corresponding levels of performance, and new or completely revised components have been added as required.

In 2001, the technology that sequenced the human genome was based on capillary electrophoresis of individual fluorescent-labelled Sanger sequencing reaction products. Each instrument could detect 500–600 bases from each of 96 reactions in around ten hours, with 24-hour unattended operation producing 115 kbp (thousand base pairs) per day. Because of the increased scale required for the Human Genome Project, genome centres had developed a robust, highly automated and inexpensive preparatory process to feed their capillary sequencers. Once the data were produced, mature analysis software was applied to analyse the sequencing reads (each a ~500-bp sequence of A, C, G, T), then to assemble reads that shared sequence identity, reproducing that region of the genome. After assembly, each genomic region was further analysed to identify genes, repeat elements and other features. As the 'drivers' of these sequencing pipelines, genome centres could dial up capacity by increasing the amount of hardware used in the preparatory and sequencing processes, because sequence production, not sequence analysis, was rate limiting.

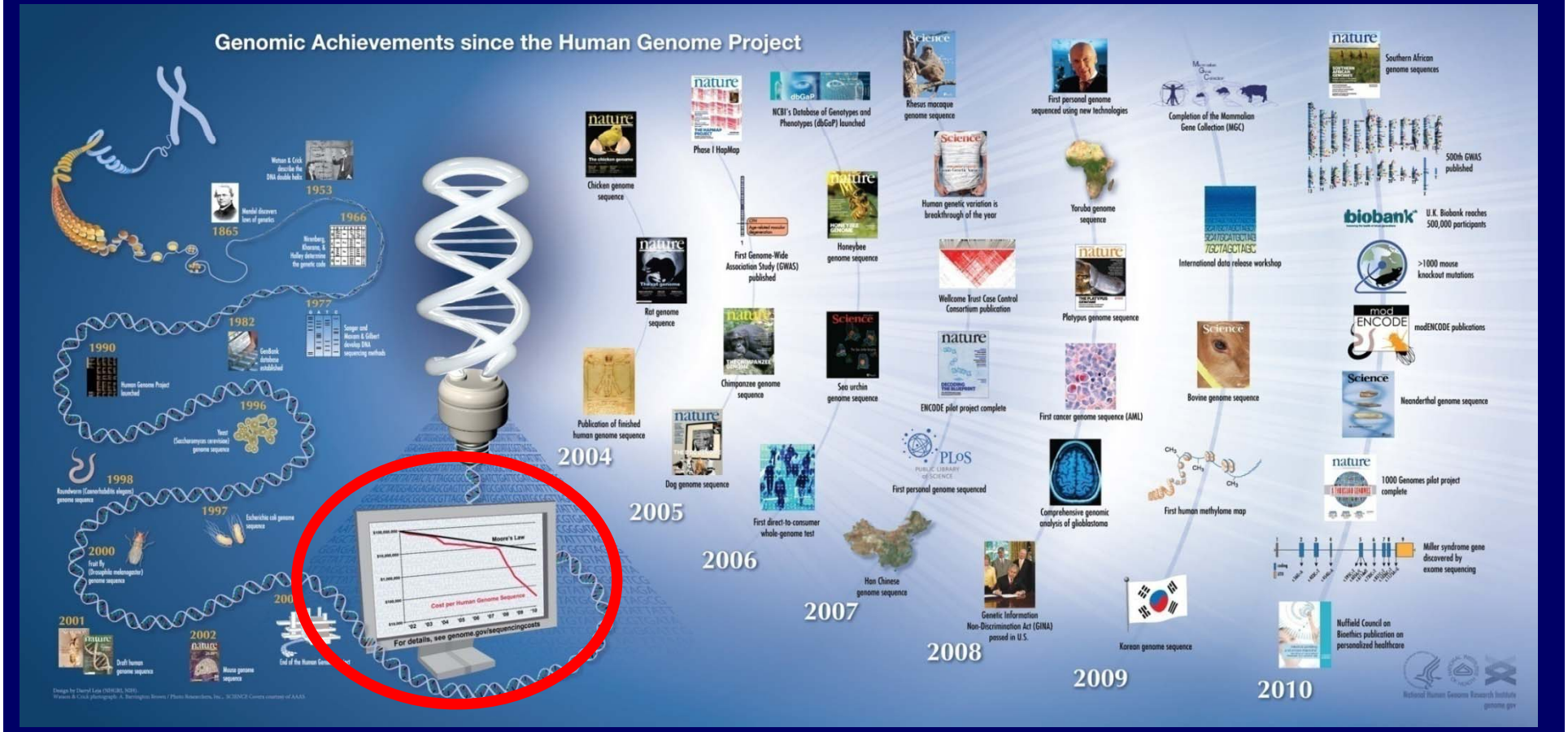
As I will describe, the ensuing 10 years has been marked by dramatic improvements in sequencing technology that have catapulted sequencing to the forefront of biological experimentation and have revolutionized the way that we approach genome-wide questions. One consequence of this revolution has been the coincident revitalization of bioinformatics, predominantly in development efforts aimed at data analysis and interpretation. Taken together, these unprecedented sequencing and analysis capabilities have inspired new areas of enquiry, have solved major questions about the regulation, variability and diaspora of the human genome, and have introduced a genomic era in medical enquiry and (ultimately) practice that will bring about the originally envisioned impact of the Human Genome Project.

Massively parallel sequencing

The first five years following the Human Genome Project provided further definition and annotation of the human genome sequence by comparative genomics; the sequencing of several model organism genomes—such as mouse[2], rat[3], chicken[4], dog[5], chimpanzee[6], rhesus macaque[7], duckbill platypus[8] and cow[9]—provided information about highly conserved genomic elements that are likely to be functional owing to their conservation. These genomes were largely produced by conventional methods, including Sanger-based capillary sequencing. Starting in 2005, a variety of new 'engines' for DNA sequencing that were radically different from the capillary sequencers used to sequence the human and model organism genomes became available from several different manufacturers (Fig. 1). These new engines were 'turbo-charged' by several orders of magnitude compared to their predecessors, because the basic mechanisms for data generation had changed radically, producing far more sequence reads per instrument run and at a significantly lower expense. The availability of multiple commercially available instruments alone represented a paradigm shift from the previous decade, where a single capillary instrument produced by Applied Biosystems dominated the market. Many of these innovative approaches were initially developed with National Institutes of Health (NIH) funding through the 'Technology development for the $1,000 genome' program (http://www.genome.gov/11008124#al-4) introduced during Francis Collins' directorship at the National Human Genome Research Institute (NHGRI).

Since the introduction of these platforms, the past five years have been marked by fierce competition between their manufacturers to greatly

[1]The Genome Center at Washington University School of Medicine, Department of Genetics, Washington University School of Medicine, St Louis, Missouri 63108, USA


Genomic Achievements since the Human Genome Project
1865
1953
1966
1977
1982
1990
1996
1998
1997
2000
2001
2002
2004
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2006
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2008
2009
2010
Chicken genome sequence
Phase I HapMap
NCBI's Database of Genotypes and Phenotypes (dbGaP) launched
Rhesus macaque genome sequence
First personal genome sequenced using new technologies
Completion of the Mammalian Gene Collection (MGC)
Southern African genome sequences
500th GWAS published
Rat genome sequence
First Genome-Wide Association Study (GWAS) published
Honeybee genome sequence
Human genetic variation is breakthrough of the year
Yoruba genome sequence
International data release workshop
U.K. Biobank reaches 500,000 participants
>1000 mouse knockout mutations
Wellcome Trust Case Control Consortium publication
Platypus genome sequence
modENCODE publications
Chimpanzee genome sequence
Sea urchin genome sequence
ENCODE pilot project complete
First cancer genome sequence (AML)
Bovine genome sequence
Neanderthal genome sequence
Publication of finished human genome sequence
Dog genome sequence
First direct-to-consumer whole-genome test
First personal genome sequenced
Comprehensive genomic analysis of glioblastoma
First human methylome map
1000 Genomes pilot project complete
Han Chinese genome sequence
Genetic Information Non-Discrimination Act (GINA) passed in U.S.
Korean genome sequence
Miller syndrome gene discovered by exome sequencing
Nuffield Council on Bioethics publication on personalized healthcare
Draft human genome sequence
Mouse genome sequence
Moore's Law
Cost per Human Genome Sequence
For details, see genome.gov/sequencingcosts

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National Human Genome Research Institute
National Institutes of Health

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

Home › Research Funding › DER Funded Programs › The Large-Scale Genome Sequencing Program › DNA Sequencing Costs

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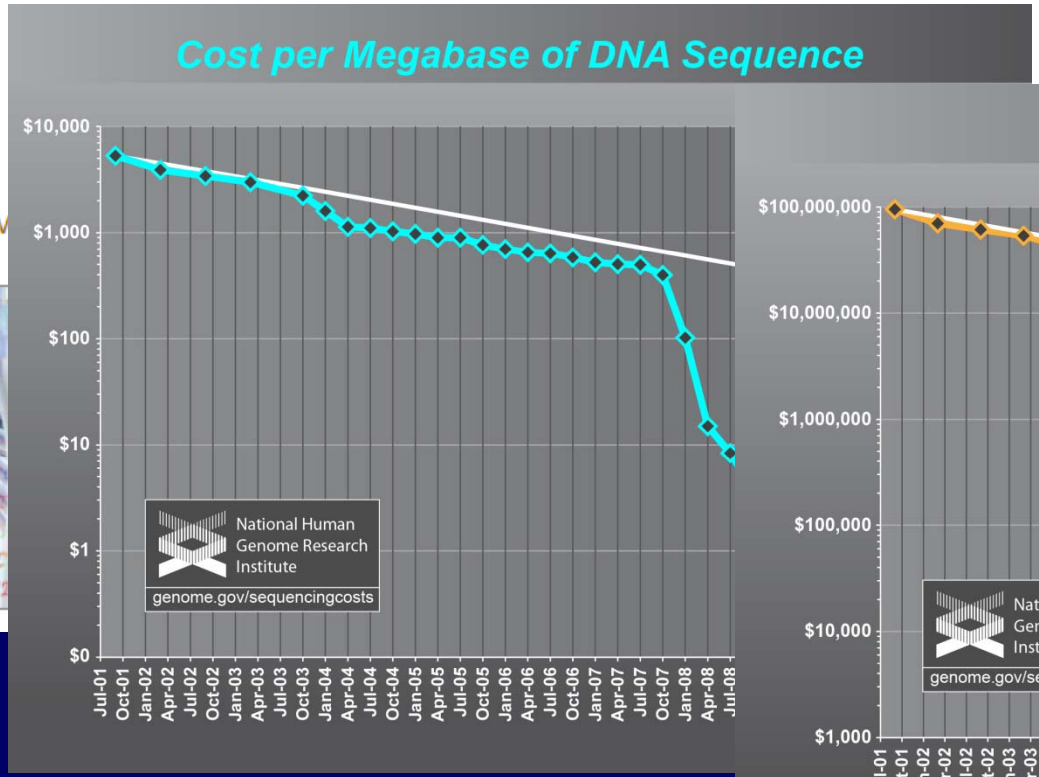
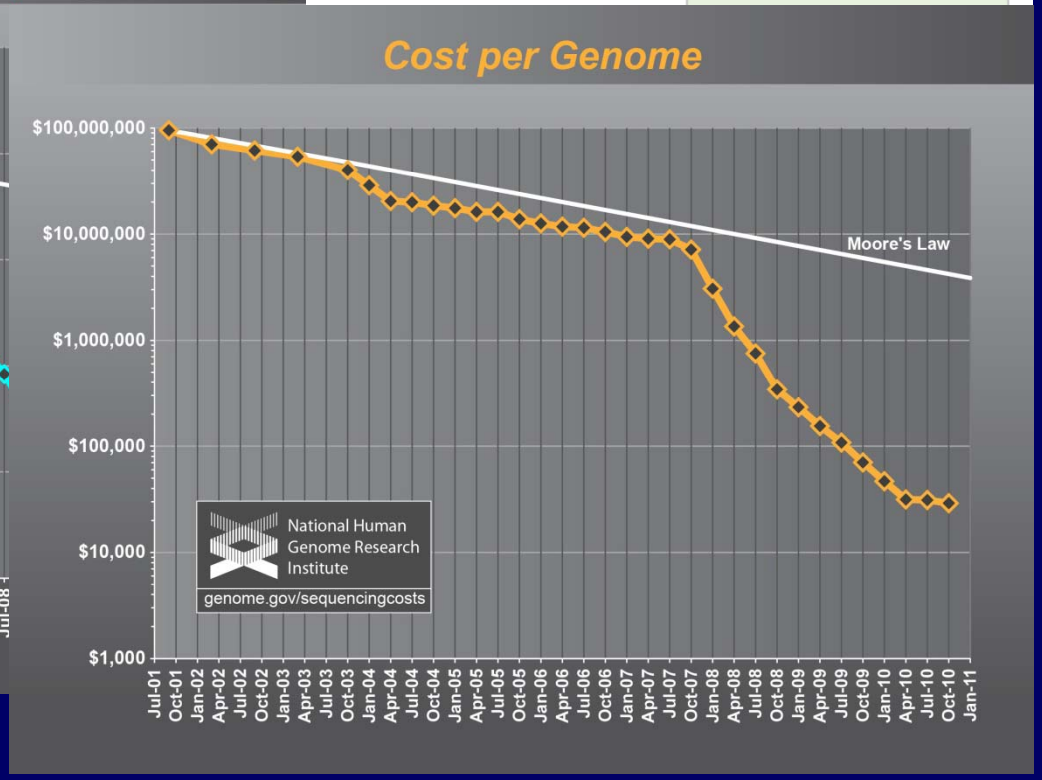
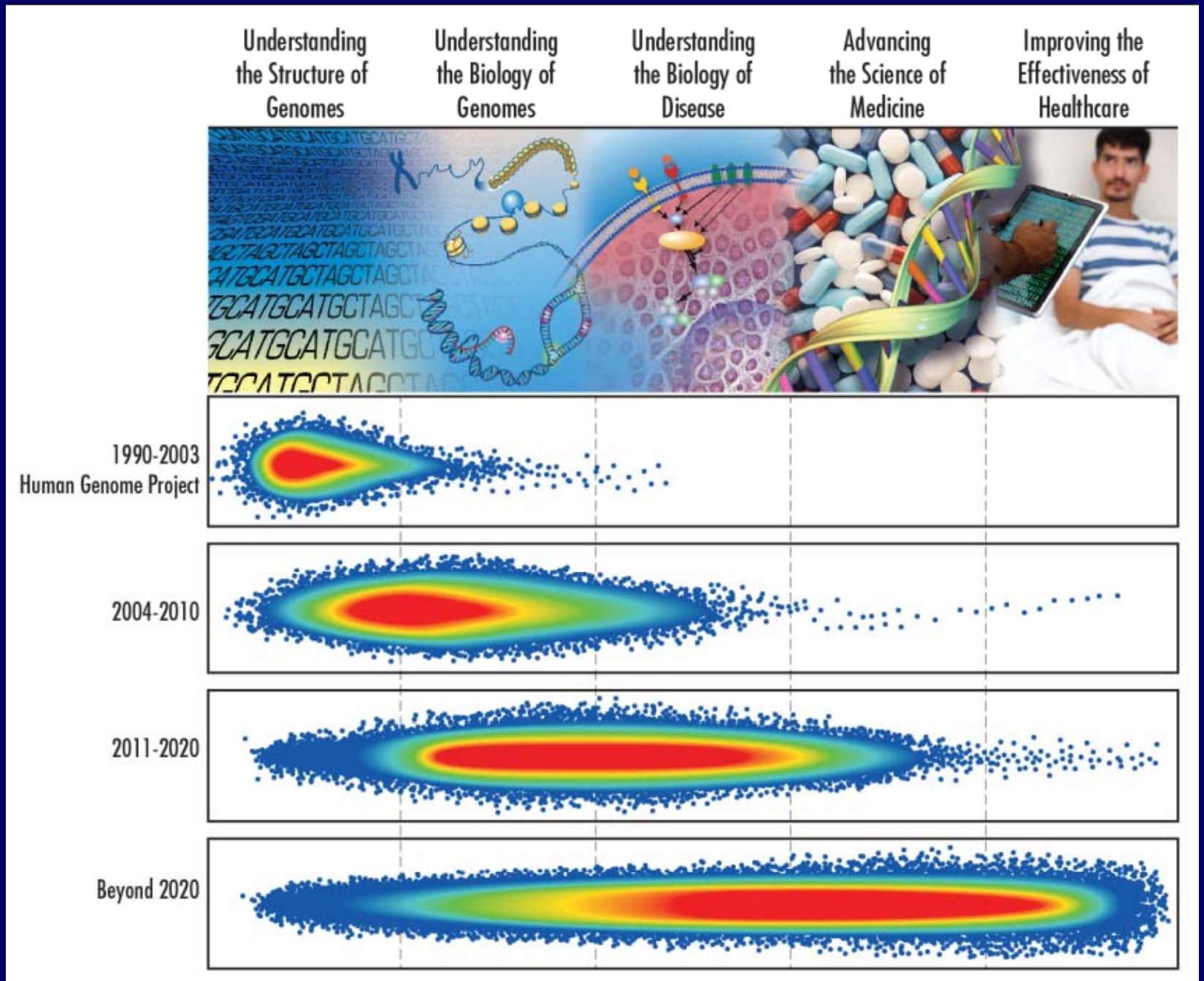
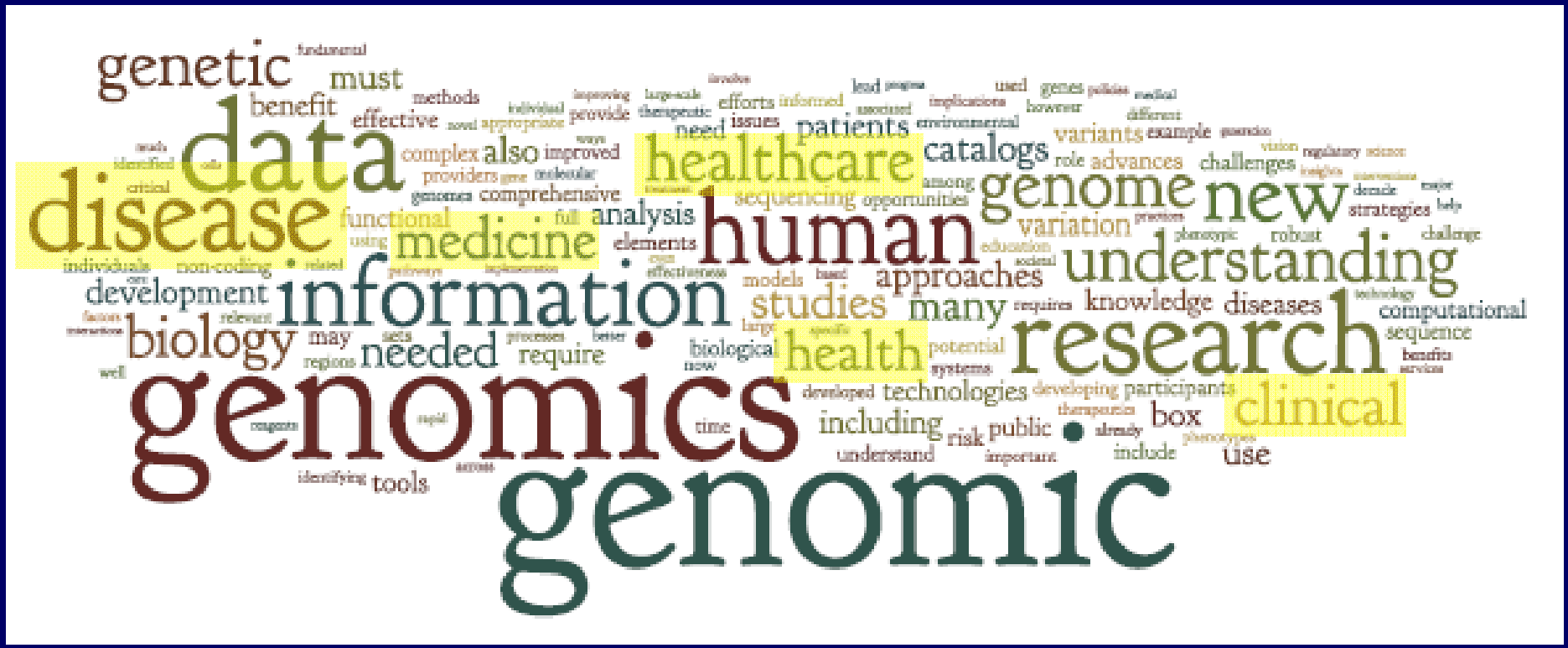




genome.gov/sequencingcosts

Document 1


Understanding the Structure of Genomes
Understanding the Biology of Genomes
Understanding the Biology of Disease
Advancing the Science of Medicine
Improving the Effectiveness of Healthcare
1990-2003 Human Genome Project
2004-2010
2011-2020
Beyond 2020


genetic
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Green
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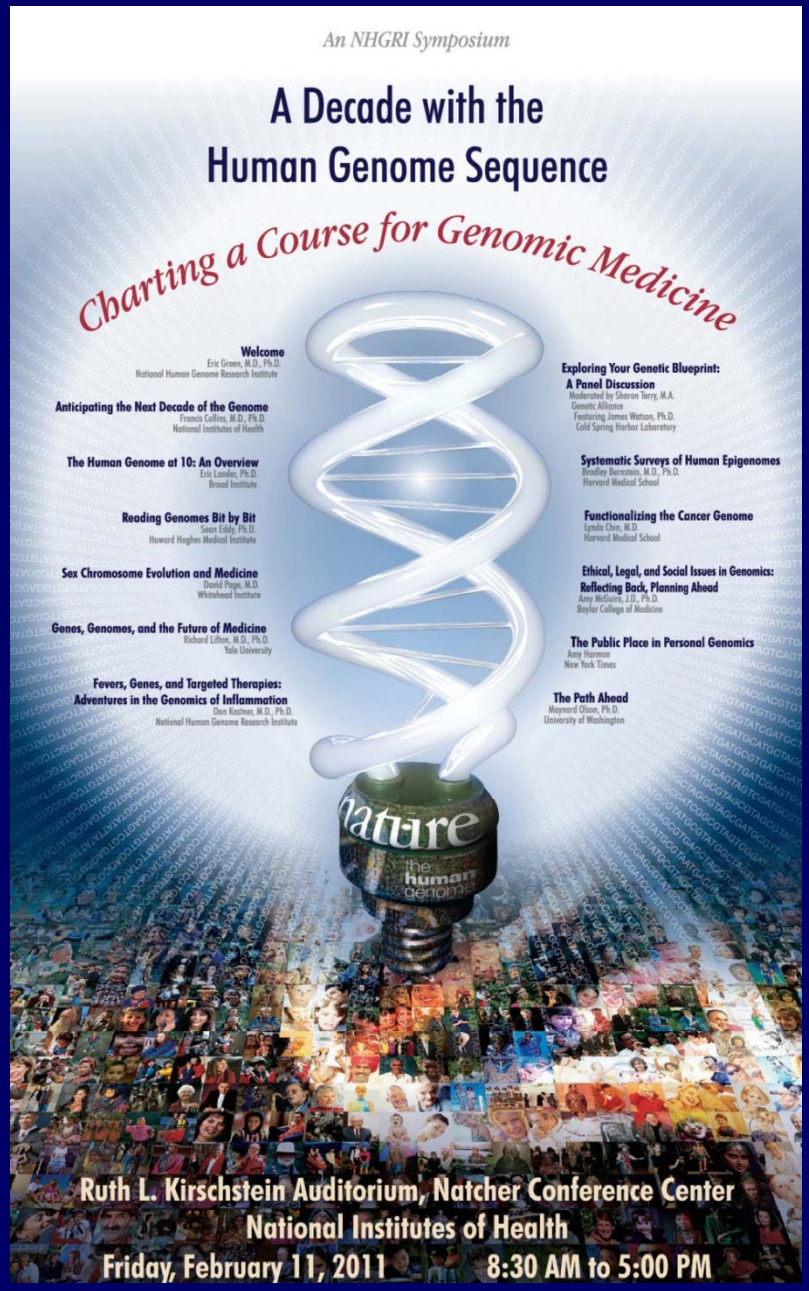
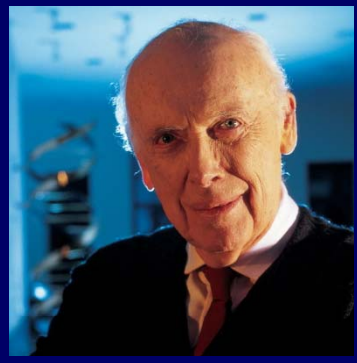
February 11, 2011 Symposium





genome.gov/Symposium2011

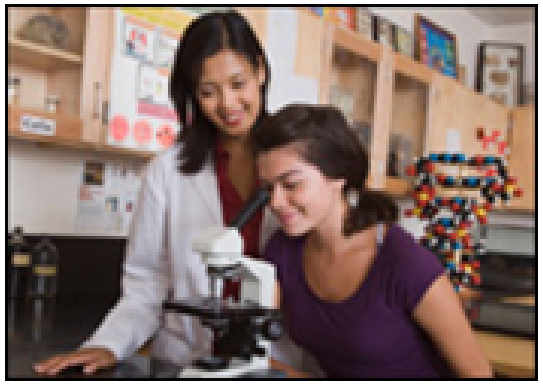
Document 2

Koshland Museum Science Café

EVENTS

Genomics and Society: Ten Years After Sequencing the Human Genome

Date: Thursday, **February 10, 2011**
Location: Koshland Science Museum
Time: 5:30 PM to 7:00 PM
Cost: Free Admission
Max Attendees: 80
Age Range: Educators



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Document 2

2011 NHGRI Strategic Plan for Genomics

>>>

NHGRI

Developing an Implementation Plan: Extramural Retreat (Jan. 25 2011)

Science Special Series on Human Genome 10th Anniversary (February 4, 2011 Issue)

Special Series: Human Genome 10th Anniversary

In February 2001, *Science* and *Nature* published two papers that provided the first detailed look at the nearly complete sequence of the human genome. *Science* is pleased to present a special month-long series celebrating the 10th anniversary of that momentous achievement, including News features and brief essays that explore the impacts of the genomics revolution on science and society.

See the historic 16 February 2001 issue of *Science* reporting the sequencing of the human genome>>

4 FEBRUARY 2011

EDITORIAL

Lessons from Genomics
B. Alberts

NEWS FOCUS

Waiting for the Revolution
E. Marshall

Human Genetics in the Clinic, One Click Away
E. Marshall

The Human Genome (Patent) Project
S. Kean

Science Podcast
The 4 February show includes several genome-related segments, including highlights from this series and a discussion about the water flea genome.

ESSAYS

Introduction: A Celebration of the Genome, Part I
B. R. Jasny and L. M. Zahn

Faces of the Genome
F. S. Collins

The Human Genome at 10: Successes and Challenges
J. C. Venter

The Golden Age of Human Population Genetics
M. Przeworski

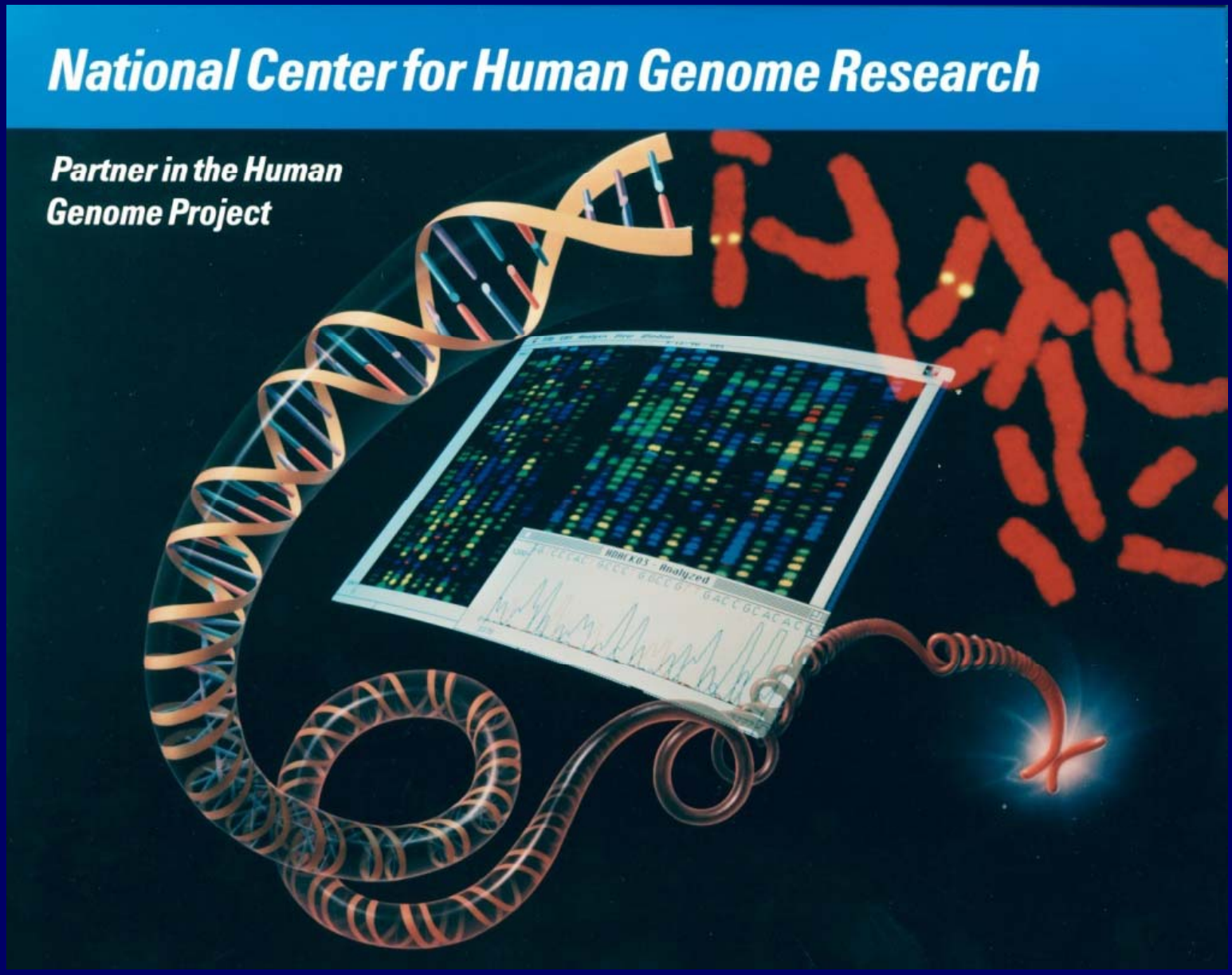
Genomics and Clinical Relevance
T. Hudson

What Defines Us?
R. Cole-Turner

Painting the Genome for the Public
X. Cortada

Bringing Genomics and Genetics Back Together
R. A. Gibbs

Happy 14th Birthday, NHGRI



Fiscal Year 2011 Appropriations Update

- Continuing Resolution until March 2011
- Operating at FY2010 levels
- Unclear what final outcome will be (for this year or next or next…)

NHGRI Deputy Director Search

Deputy Director

National Human Genome Research Institute

The National Human Genome Research Institute (NHGRI), a major research component of the National Institutes of Health (NIH) and the Department of Health and Human Services (DHHS), seeks to identify an outstanding Deputy Director.

The NHGRI Deputy Director will assist the Director in providing overall leadership of the Institute, sharing responsibilities in all phases of leading the preeminent organization dedicated to advancing genomic and genetic research, including its clinical applications. As a member of the NHGRI senior leadership, the Deputy Director will work with the Director in shaping and executing a strategic vision for the Institute as well as communicating that vision to the Institute staff and the broader scientific community. In working closely with the Director, the Deputy Director helps to develop Institute goals, priorities, policies, and program activities; this requires staying abreast of developments and needs of the Institute and the field.

Applicants must have an M.D. and/or Ph.D or equivalent degree in the biomedical sciences, as well as a broad knowledge of the field of human genetics and genomics. They must further have a compelling vision for the future of the field and the role for NHGRI within the field. Also required are senior-level research and/or clinical experience and knowledge of the major scientific areas related to genetics and genomics, in addition to well-honed administrative and interpersonal skills to meet the demands of helping to lead a complex organization. Applicants should have demonstrated leadership in dealing with different stakeholder groups within the research community, planning and assessing programs, developing plans to resolve operational problems and issues, and managing financial and human resources. Applicants should be known and respected within their profession, both nationally and internationally as individuals of outstanding scientific competence.

Salary is competitive and will be commensurate with the candidate's experience. A full Federal benefit package is available, including retirement, health and life insurance, long-term care insurance, annual and sick leave, and the Thrift Savings Plan (401K equivalent).

Interested applicants should submit a cover letter that includes a brief description of research, clinical, and/or administrative experience, a current curriculum vitae and bibliography, names and contact information of five references, and a brief written vision for becoming the NHGRI Deputy Director. Questions about the position and applications themselves should be sent to Ms. Ellen Rolfes via email at ellenr@exchange.nih.gov. All information provided by the candidates will remain confidential and will not be released outside the NHGRI search process without a signed release from the candidate.

Applications will be reviewed starting November 1, 2010, and will be accepted until the position is filled.

DHHS and NIH are Equal Opportunity Employers and encourage applications from women and minorities.

NATIONAL HUMAN GENOME RESEARCH INSTITUTE

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES | NATIONAL INSTITUTES OF HEALTH | genome.gov

Document 3

Special NHGRI Visitor: 2011-2012

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

Document 4

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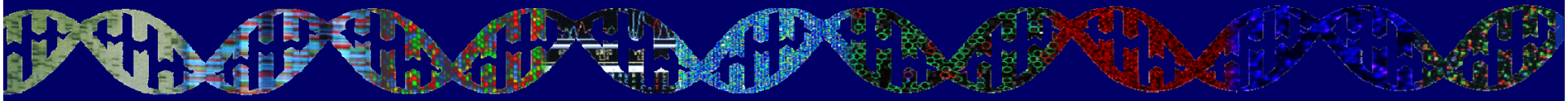


New Deputy Director for Science, Outreach, and Policy, NIH

Kathy Hudson, Ph.D.

Document 5

New Deputy Director, Office of Extramural Research, NIH

Della Hann, Ph.D.

Document 6

New Deputy Director, National Institute of Environmental Health Sciences, NIH

Rick Woychik, Ph.D.

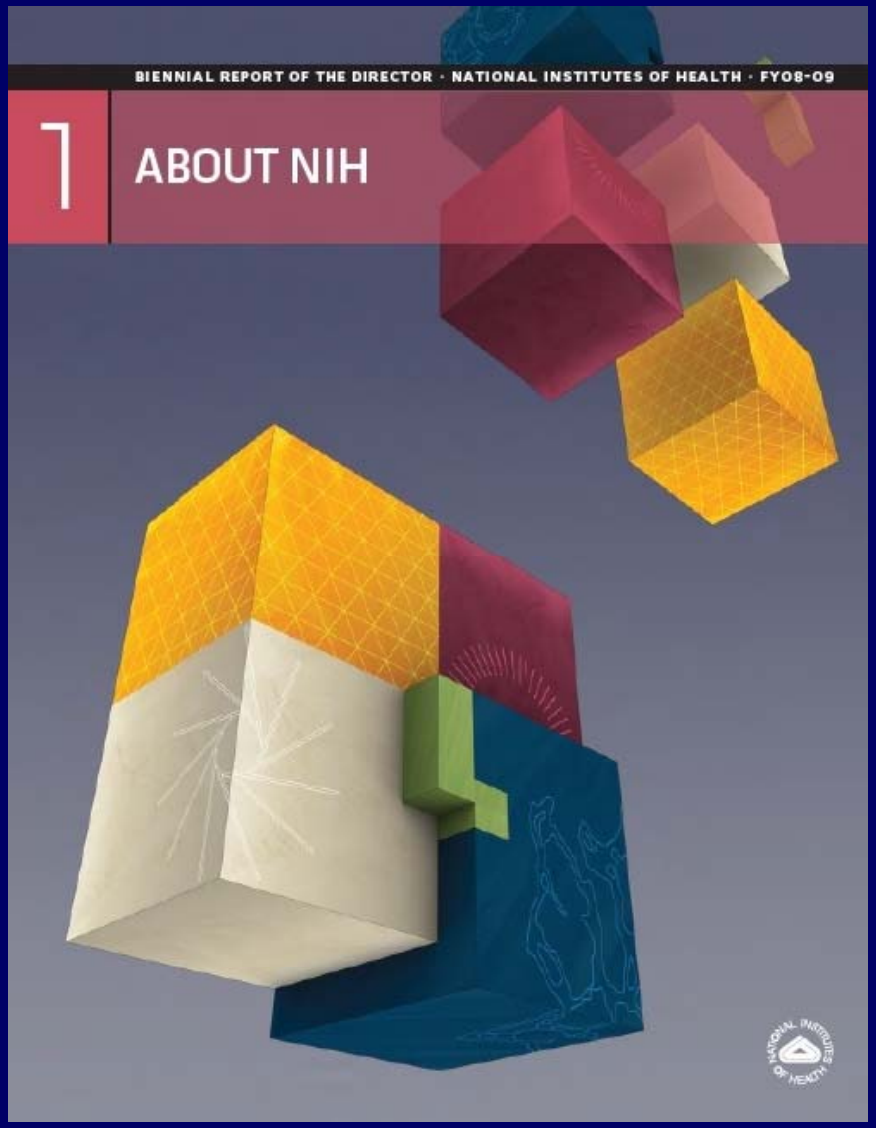
Document 7

Departing Director, National Institute of General Medical Sciences

Jeremy Berg, Ph.D.

Document 8

Biennial Report of the NIH Director 2008-2009



- **Reports the following was spent on "Human Genome" research:**

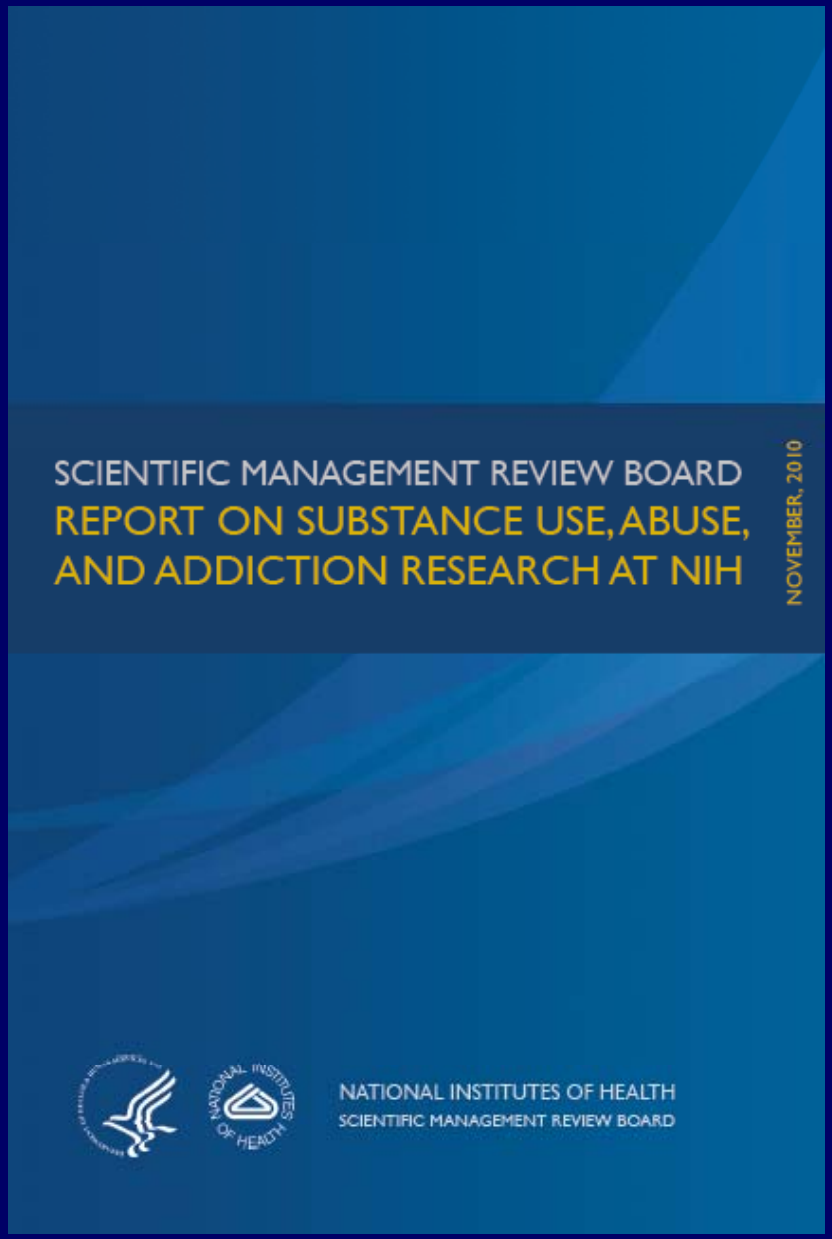
FY 2008--
$1,259 billion

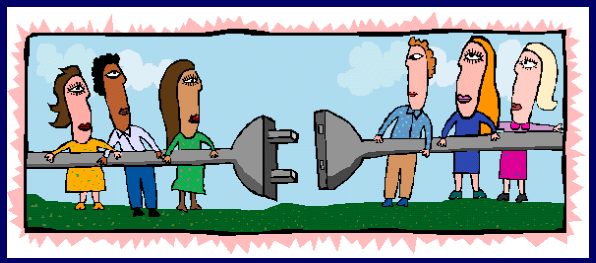
FY 2009 Non-ARRA--
$1,775 billion

FY 2009 ARRA--
$0.566 billion

Document 9

Planned Merger of NIAAA & NIDA





Document 10

Therapeutics and Translational Sciences

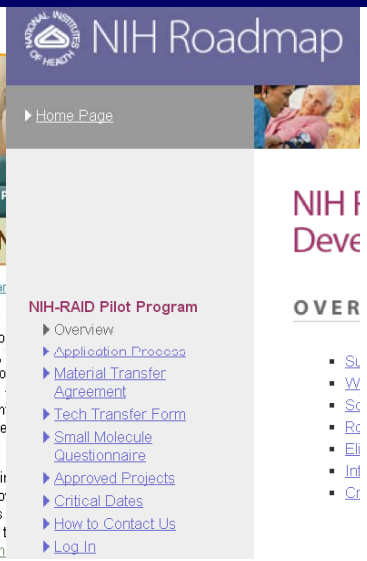
- **Evolution and Consolidation:**

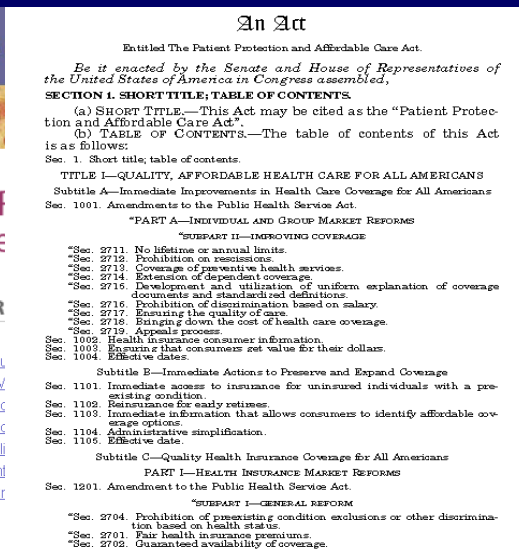
  NIH Chemical Genomics Center (NCGC)

  Therapeutics for Rare and Neglected Diseases (TRND) Program

  Rapid Access to Interventional Development (RAID) Program

  Cures Acceleration Network (CAN)





An Act

Entitled The Patient Protection and Affordable Care Act.

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

SECTION 1. SHORT TITLE; TABLE OF CONTENTS.

(a) SHORT TITLE.—This Act may be cited as the "Patient Protection and Affordable Care Act".

(b) TABLE OF CONTENTS.—The table of contents of this Act is as follows:

Sec. 1. Short title; table of contents.

TITLE I—QUALITY, AFFORDABLE HEALTH CARE FOR ALL AMERICANS

Subtitle A—Immediate Improvements in Health Care Coverage for All Americans

Sec. 1001. Amendments to the Public Health Service Act.

"PART A—INDIVIDUAL AND GROUP MARKET REFORMS

"SUBPART II—IMPROVING COVERAGE

"Sec. 2711. No lifetime or annual limits.
"Sec. 2712. Prohibition on rescissions.
"Sec. 2713. Coverage of preventive health services.
"Sec. 2714. Extension of dependent coverage.
"Sec. 2715. Development and utilization of uniform explanation of coverage documents and standardized definitions.
"Sec. 2716. Prohibition of discrimination based on salary.
"Sec. 2717. Ensuring the quality of care.
"Sec. 2718. Bringing down the cost of health care coverage.
"Sec. 2719. Appeals process.
Sec. 1002. Health insurance consumer information.
Sec. 1003. Ensuring that consumers get value for their dollars.
Sec. 1004. Effective dates.

Subtitle B—Immediate Actions to Preserve and Expand Coverage

Sec. 1101. Immediate access to insurance for uninsured individuals with a pre-existing condition.
Sec. 1102. Reinsurance for early retirees.
Sec. 1103. Immediate information that allows consumers to identify affordable coverage options.
Sec. 1104. Administrative simplification.
Sec. 1105. Effective date.

Subtitle C—Quality Health Insurance Coverage for All Americans

PART I—HEALTH INSURANCE MARKET REFORMS

Sec. 1201. Amendment to the Public Health Service Act.

"SUBPART I—GENERAL REFORM

"Sec. 2704. Prohibition of preexisting condition exclusions or other discrimination based on health status.
"Sec. 2701. Fair health insurance premiums.
"Sec. 2702. Guaranteed availability of coverage.

Therapeutics and Translational Sciences

- **Evolution and Consolidation:**

  NIH Chemical Genomics Center (NCGC)

  Therapeutics for Rare and Neglected Diseases (TRND) Program

  Rapid Access to Interventional Development (RAID) Program

  Cures Acceleration Network (CAN)

- **Consolidating above within NIH Center for Translational Therapeutics (NCTT)**

  Distinct from Intramural and Extramural Divisions

  NHGRI as 'Interim Home' for NCTT

TRND Updates

- AesRX to collaborate with TRND (and NHLBI) to advance potential therapy (Aes-103) for sickle cell disease
- Other Pilot Projects:
  - Hereditary Inclusion Body Myopathy
  - Schistosomiasis/Hookworm
  - Chronic Lymphocytic Leukemia
  - Niemann-Pick Type C

NCRR Reorganization

National Center for Research Resources

feedback.nih.gov

NCRR Task Force Straw Model — January 2011 Draft

NCATS	NIGMS	NIBIB	NIMHD	Interim Infrastructure Unit	Not Yet Assigned
CTSAs					
				National Primate Research Centers	
				Chimpanzee Resource Centers	
				Other Primate Model Resources	
	Other Disease Model Resources				
	Beam Line and Mass Spect P41s	Imaging P41s		Remaining P41s	
	Shared and High-End Instrumentation				
				Biomedical Tech Other (R01, R21, BIRN, etc.)	
			RCMI		
				IDeA	
				SEPA	
				Extramural Construction	
					NCRR OD Not Yet Assigned

Document 12

SMRB Report on Translational Medicine and Therapeutics

Document 13

Proposed National Center for Advancing Translational Sciences (NCATS)

Component	FY10 budget
Clinical and Translational Science Awards (CTSAs)	~$450M
Molecular Libraries and Imaging program	$107M
Therapeutics for Rare and Neglected Diseases (TRND) program	$24M
Rapid Access to Interventional Development (RAID) program	$8M
NIH-FDA Regulatory Science Initiative	~$3M
Cures Acceleration Network	N/A

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Take Control

Federal Research Center Will Help Develop Medicines

By GARDINER HARRIS
Published: January 22, 2011

The Obama administration has become so concerned about the slowing pace of new drugs coming out of the pharmaceutical industry that officials have decided to start a billion-dollar government drug development center to help create medicines.

Enlarge This Image

Jennifer S. Altman for The New York Times

Creating a drug development center is a signature effort of Dr. Francis S. Collins, director of the National Institutes of Health.

Fewer New Drugs

Large drug makers have begun to reduce spending on research and

The new effort comes as many large drug makers, unable to find enough new drugs, are paring back research. Promising discoveries in illnesses like depression and Parkinson's that once would have led to clinical trials are instead going unexplored because companies have neither the will nor the resources to undertake the effort.

The initial financing of the government relatively small compared with the $45 industry estimates it invested in resear

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COMMENT ON...

- Proposed Institute for Substance Use, Abuse and Addiction
- Proposed National Center for Advancing Translational Sciences

IMPORTANT INFORMATION

- Documentation About the Proposed Institute for Substance Use, Abuse, and Addiction
- Documentation About the Proposed National Center for Advancing Translational Sciences
- FAQ: Proposed Institute for Substance Use, Abuse, and Addiction
- FAQ: Proposed National

Older Entries »

SEPARATING FACT & FICTION: NEWS ABOUT THE PROPOSED NATIONAL CENTER FOR ADVANCING TRANSLATIONAL SCIENCES

POSTED ON JANUARY 24TH, 2011 BY FRANCIS S. COLLINS, MD, PHD; JOSEPHINE BRIGGS, MD; ANTHONY FAUCI, MD; ERIC GREEN, MD, PHD; ALAN GUTTMACHER, MD; THOMAS INSEL, MD; STORY LANDIS, PHD; GRIFFIN RODGERS, MD, MACP; HAROLD VARMUS, MD; KATHY HUDSON, PHD; AND LAWRENCE A. TABAK, DDS, PHD

By now, many of you have read the recent *New York Times* article or related news coverage, about NIH's plan to establish the National Center for Advancing Translational Sciences (NCATS).

While we are pleased that the news media have recognized NIH's efforts as a significant development for translational research, the *Times* article contains some misleading statements that we would like to clarify. Those statements suggest that a much larger shakeup of NIH is underway than is actually contemplated.

So, to set the record straight, we want to share with you what we know at this point in time:

- The proposal for NCATS is that it will be assembled primarily from existing programs within the National Center for Research Resources (NCRR), the NIH Common Fund, and the National Human Genome Research Institute (NHGRI).
- NCATS is not intended to be a drug company. It is a facilitator of translational research across the NIH and complementary to translational research already being conducted and supported on a large scale in the individual NIH Institutes and Centers. NCATS will seek ways to leverage science to bring new ideas and materials to the attention of industry by demonstrating their value.

POST ARCHIVE

January 2011

S	M	T	W	T	F	S
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16	17	18	19	20	21	22
23	24	25	26	27	28	29
30	31					

« Dec

If you have comments or questions not related to the current discussions, please direct them to NIH-Listens@mail.nih.gov

If you are looking for general information about the National Institutes of Health, or the 27 Institutes and Centers, please visit http://www.nih.gov/

Document 13

NCMHD → NIMHD

Document 14

NIH Early Independence Award Program

WORLD VIEW *A personal take on events*

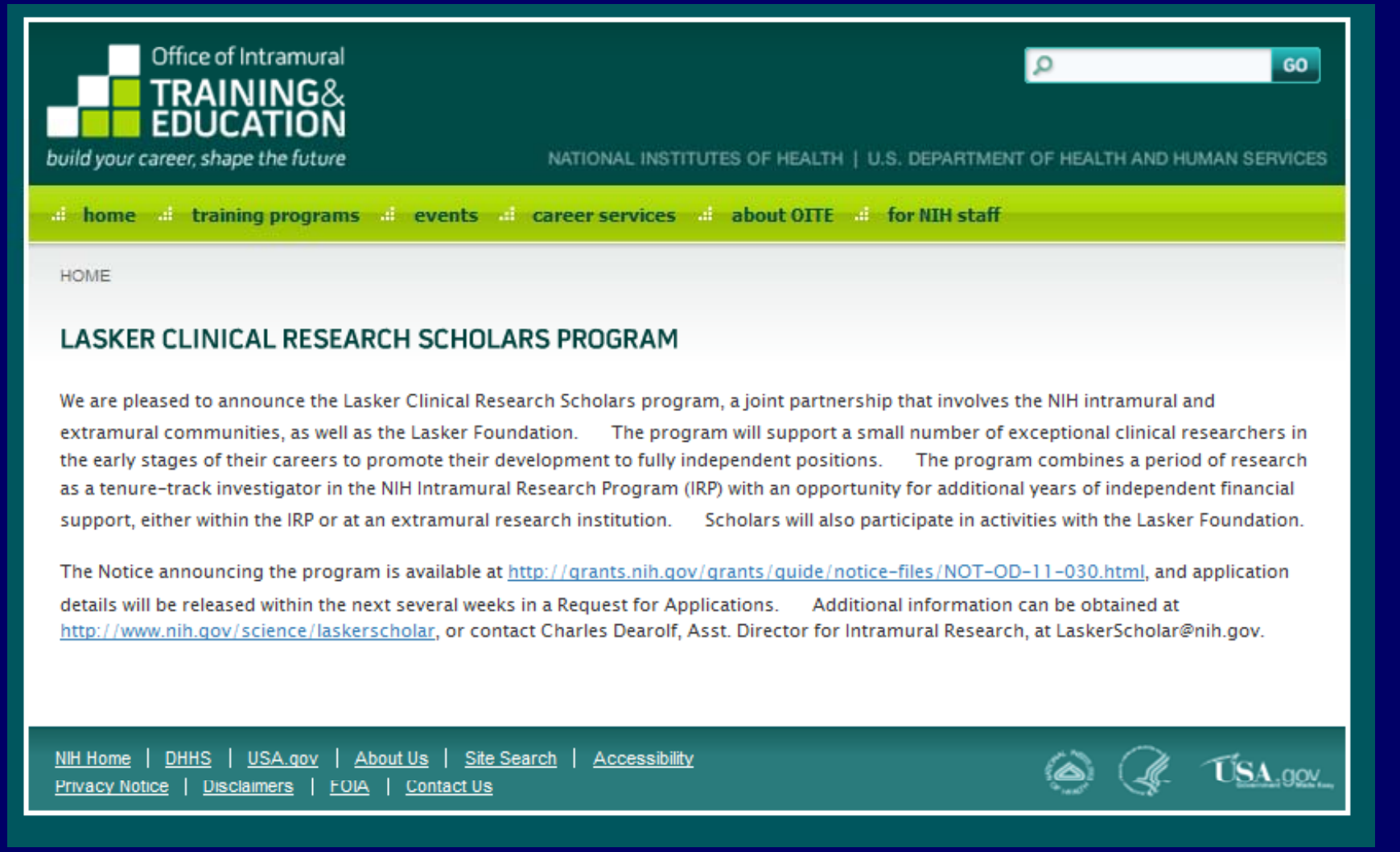
Scientists need a shorter path to research freedom

Francis Collins *explains why the NIH is launching a bid to help some doctoral students dramatically reduce the time required to start an independent career.*

WE MUST
LIBERATE
OUR BRIGHTEST MINDS
TO PURSUE HIGH-RISK,
HIGH-REWARD
IDEAS.

Document 15

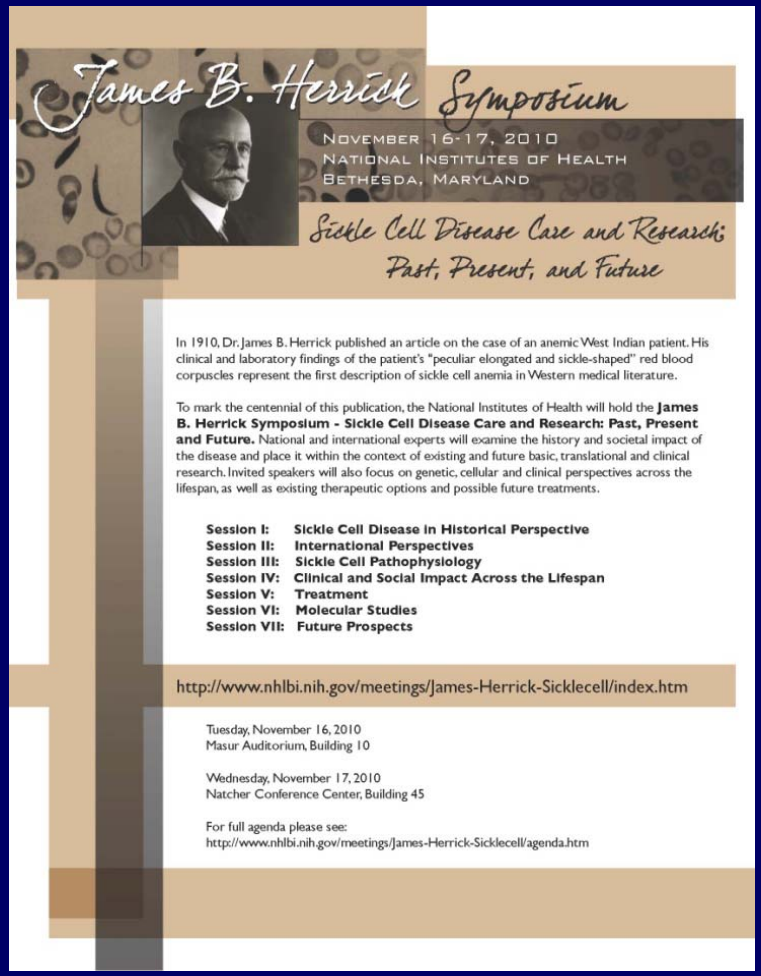
NIH Lasker Clinical Research Scholars Program



Document 16

James B. Herrick Symposium
Sickle Cell Disease Care and Research: Past, Present, and Future

- November 2010
- 39 Speakers from
  6 countries including
  Sir David Weatherall



James B. Herrick Symposium

November 16-17, 2010
National Institutes of Health
Bethesda, Maryland

Sickle Cell Disease Care and Research: Past, Present, and Future

In 1910, Dr. James B. Herrick published an article on the case of an anemic West Indian patient. His clinical and laboratory findings of the patient's "peculiar elongated and sickle-shaped" red blood corpuscles represent the first description of sickle cell anemia in Western medical literature.

To mark the centennial of this publication, the National Institutes of Health will hold the **James B. Herrick Symposium - Sickle Cell Disease Care and Research: Past, Present and Future.** National and international experts will examine the history and societal impact of the disease and place it within the context of existing and future basic, translational and clinical research. Invited speakers will also focus on genetic, cellular and clinical perspectives across the lifespan, as well as existing therapeutic options and possible future treatments.

Session I: Sickle Cell Disease in Historical Perspective
Session II: International Perspectives
Session III: Sickle Cell Pathophysiology
Session IV: Clinical and Social Impact Across the Lifespan
Session V: Treatment
Session VI: Molecular Studies
Session VII: Future Prospects

http://www.nhlbi.nih.gov/meetings/James-Herrick-Sicklecell/index.htm

Tuesday, November 16, 2010
Masur Auditorium, Building 10

Wednesday, November 17, 2010
Natcher Conference Center, Building 45

For full agenda please see:
http://www.nhlbi.nih.gov/meetings/James-Herrick-Sicklecell/agenda.htm

Document 17

I. General NHGRI Updates

II. General NIH Updates

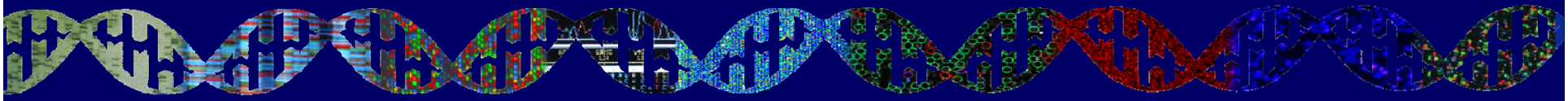
III. Genomics Updates

IV. NHGRI Extramural Program

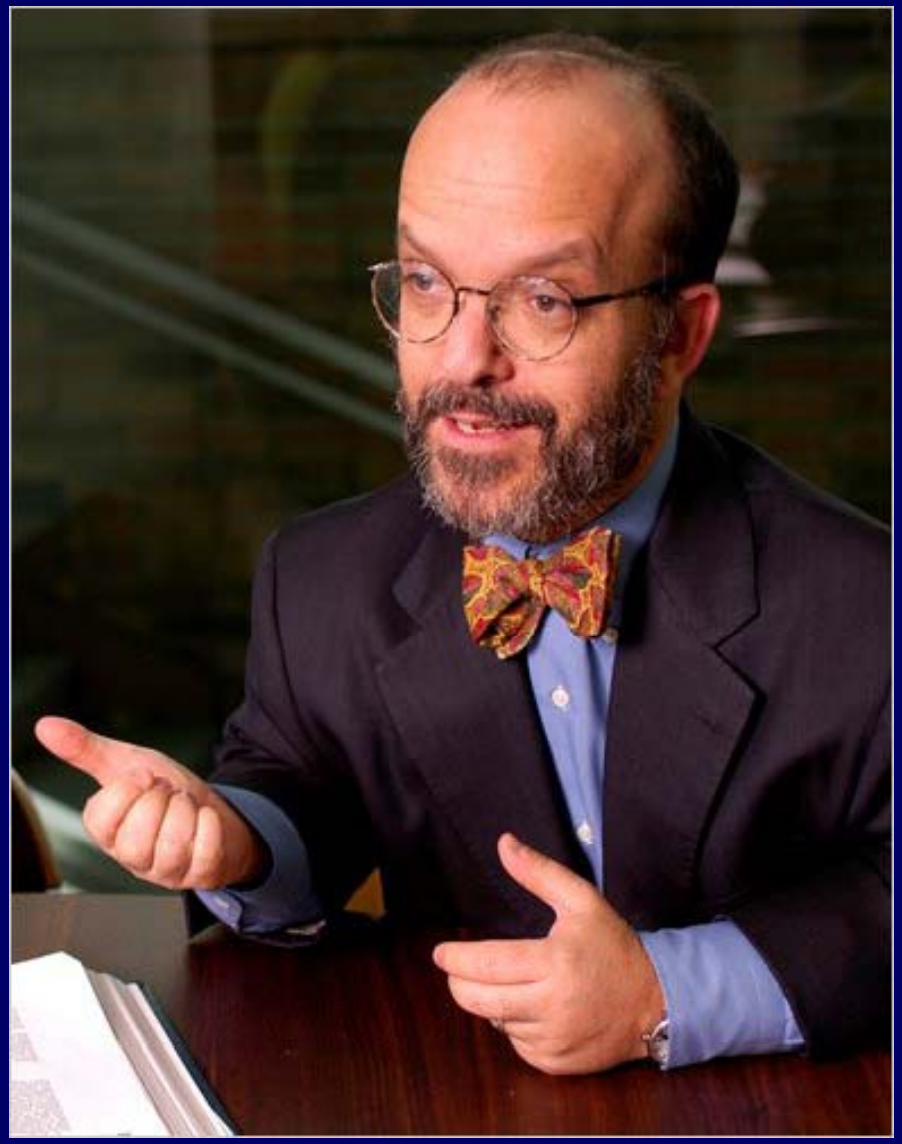
V. NIH Common Fund Programs

VI. NHGRI Office of the Director

VII. NHGRI Intramural Program



Mourning the Loss of Paul Miller



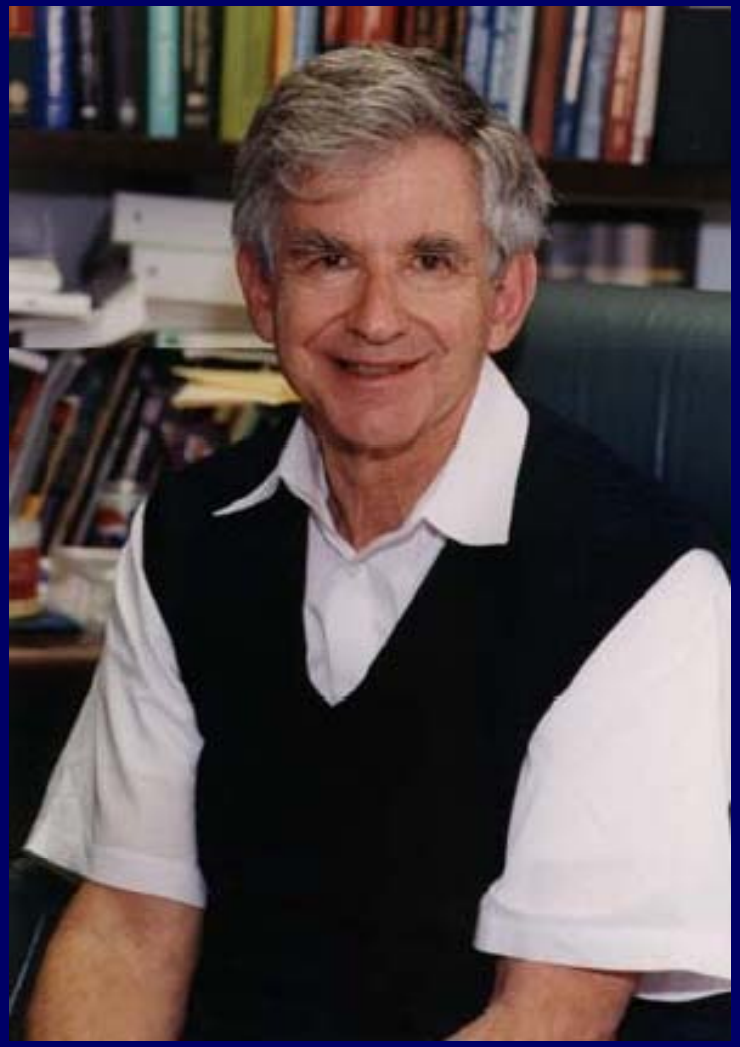
Document 18

ASHG 2010 William Allan Award

Jurg Ott, Ph.D.

Document 19

ASHG 2010 McKusick Leadership Award



Charley Epstein, M.D.

Document 20

2010 MacArthur Fellow

Carlos Bustamante, Ph.D.

Document 21

2010 Pearl Meister Greengard Prize

Janet Rowley, Ph.D. & Mary-Claire King, Ph.D.

Document 22

2011 Bower Award and Prize for Achievement in Science

George Church, Ph.D.

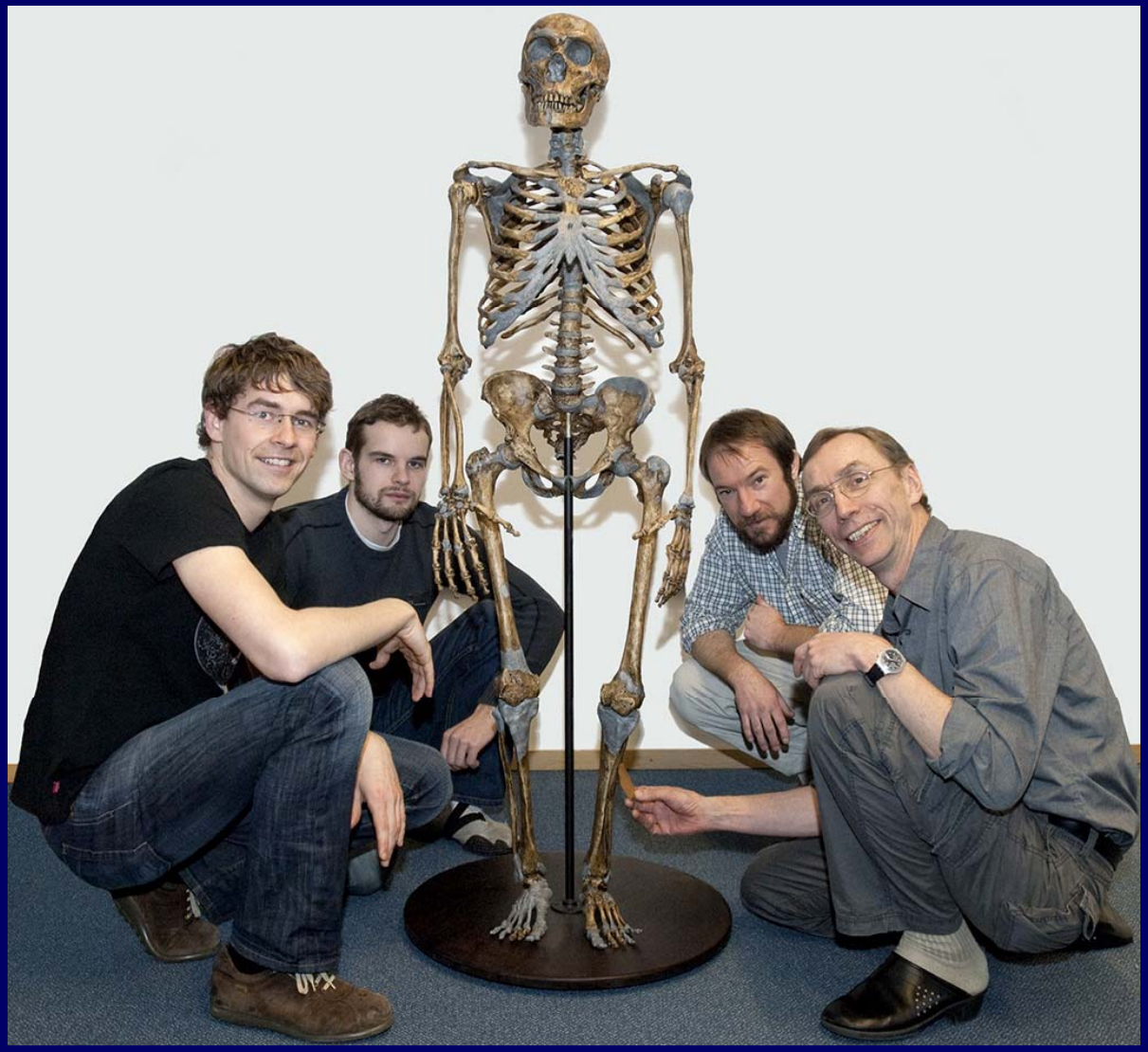
Document 23

Elected to the Institute of Medicine 2010

- Jeremy Berg (Director, NIGMS)
- Linda Birnbaum (Director, NIEHS)
- David Altshuler (Harvard & Broad)
- Sydney Brenner (Salk Institute)
- Charis Eng (Case Western Reserve University)
- Carol Greider (Johns Hopkins University)
- Caryn Lerman (University of Pennsylvania)
- Neil Risch (UCSF)

Document 24

AAAS Newcomb Cleveland Prize: Neandertal Genome Study



2011 ASHG Leadership Election

President Elect:
Mary-Claire King

Secretary:
Brendan Lee

Board of Directors:

Leslie Biesecker

Stylianos Antonarakis

Kay Davies

New Editor: American Journal of Human Genetics

David Nelson, Ph.D.

New Tufts University President

Anthony Monaco, M.D., Ph.D.

Document 25

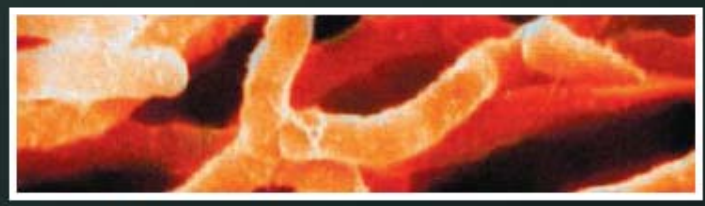
New Executive Director of NCHPEG

Joan Scott, M.S., C.G.C.

Document 26

Science's Insights of the Decade

The Dark Genome



The Microbiome

Ancient DNA

Document 27

Science's Breakthrough of the Year (2010)

Runner Ups:

Neandertal Genome

Exome Sequencing/ Rare Disease Genes

Next-Generation Genomics

Document 27

Nature's Predictions for 2011

NEWS IN FOCUS

PROSPECTS

New year, new science

Nature *looks at key findings and events that could emerge from the research world in 2011.*

- **Genome-Sequencing Explosion**
- **GWASs Prove Their Worth**

Document 28

Nature Precedings Marker Papers

nature**precedings**

Pre-publication research and preliminary findings

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Human Microbiome Project (HMP)

At the end of 2007 the NIH roadmap or common fund launched the Human Microbiome Project (HMP). The broad goal of this five year effort is to catalog and characterize the microbes living in and on the human body (the microbiome). More specifically, the project is addressing the issue of whether there is a shared core microbiome among different people and in and on different body sites. The HMP Demonstration Projects, whose "marker papers" are the first papers in this collection, are addressing another important question. Specifically, they are exploring if there is a relationship between disease states and changes in the human microbiome. In addition, HMP is working to develop new technological and bioinformatic tools needed to study these questions. Finally the project is investigating the ethical, legal, and social implications of this research. More detailed information about the HMP can be found at http://nihroadmap.nih.gov/hmp/ and http://www.hmpdacc.org/.

The NIH has been working with Nature Precedings on a pilot effort to make marker papers available electronically with the cooperation of the investigators from the Human Microbiome Project (HMP) Demonstration Projects as part of an on-going effort. These marker papers and similar papers from other community resource projects ought to provide information describing the project's purpose, experimental design and scope, data quality policies, anticipated data analyses to be included in future publications, the data release plan (including publication moratoria and any other use restrictions), and contact information. The current HMP marker papers provide the desired information, although work to facilitate obtaining information regarding the moratorium status of data is on-going. It is expected that these papers will be updated as new data sets are deposited or new policies are agreed to.

Order by: Date Title

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The Human Virome in Children and its Relationship to Febrile Illness

Gregory A. Storch et al.

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Frequently Asked Questions

Documents on Nature Precedings are not peer-reviewed.

What is Nature Precedings?
Nature Precedings is a permanent, citable archive for pre-publication research and preliminary findings.

What is voting and who can vote?
Voting is intended to be an informal way of showing support for a researcher's work.

Document 29

NHGRI @ ASHG

- NHGRI Director Met with ASHG Board of Directors and ACMG Board
- Good Press Interactions
- NIH Town Hall on Genetic Testing

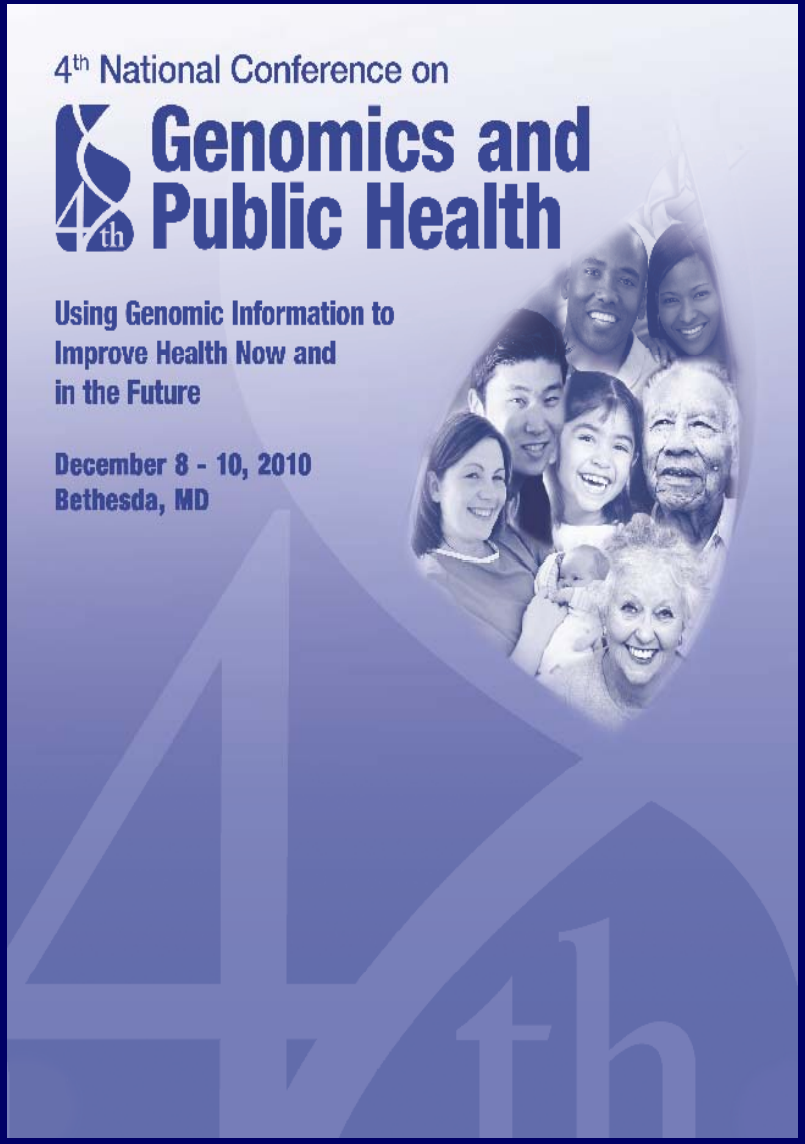
Document 30

NHGRI @ ASHG: 1000 Genomes

1000 Genomes Data Tutorial

- ~500 people in the room
- Video posted at genome.gov (with ~10K visits to date)

4th National Conference on Genomics and Public Health



Document 31

2011 Advances in Genome Biology and Technology Meeting

AGBT | Advances in Genome Biology and Technology

Document 32

NHGRI Genomic Advance of the Month

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

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Home › Newsroom › Advance of the Month: The Biology of Living Longer

Newsroom
- Advance of the Month: The Biology of Living Longer
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Genomic Advance of the Month: The Biology of Living Longer

January 2011

Editor's Note: *Genomics has become a fast-moving field with cool findings pouring out of labs all over the world. Each month, the National Human Genome Research Institute will highlight what it considers the coolest genomic advances, broadly defined, of the previous month. This process may be somewhat arbitrary and NHGRI's decisions debatable, but this is intend to be fun and your comments are definitely welcome. Here's the first installment:*

Ronald DePinho talks about his research in reversing the aging process on The Colbert Report.

By Jonathan Gitlin, Ph.D.
NHGRI Staff Writer

The ability to reverse or halt the aging process has long held allure, from early human mythology to Oscar Wilde (*The Picture of Dorian Gray* [wikipedia.org]) through to Indiana Jones (*Indiana Jones and the Last Crusade* [wikipedia.org]). It's also been the subject of considerable scientific study.

In January 2011, a paper published in the journal *Nature* has shown, for the first time, a possible biological mechanism where halting the aging process might be possible. A team of researchers at Harvard, led by Ronald DePinho, has found a way to reverse aging in a mouse by manipulating telomeres.

Document 33

Flavors of the Genome

The Ozzy Story Continues…

ST. LOUIS BUSINESS JOURNAL

Ozzy has both warrior, worrier genes, Cofactor finds

St. Louis Business Journal - by Kelsey Volkmann
Date: Monday, October 25, 2010, 9:09am CDT - Last Modified: Monday, October 25, 2010, 9:23am CDT

Related: Media & Marketing, Health Care, Technology

Sixty-one-year-old **Ozzy Osbourne** has been able to survive years of the rock 'n roll lifestyle because he has a rare combination of both warrior and worrier genes, St. Louis start-up CofactorGenomics found when it sequenced the musician's genes.

"Ozzy is a phenomenon. All these years of booze, drugs and rock 'n roll, and how is this guy still alive? What is it that he has inside of him that might be different from other people?" asked **Richard Kellner**, director of business development at CofactorGenomics.

The St. Louis firm, which is led by President and Chief Technology Officer **Jarret Glasscock** and has 10 employees and $3

Enlarge Image

Ozzy Osbourne

THEY SAID IT

"Given the swimming pools of booze I've guzzled over the years—not to mention all of the cocaine, morphine, sleeping pills, cough syrup, LSD, Rohypnol… you name it—there's really no plausible medical reason why I should still be alive. Maybe my DNA could say why."

—Rock star Ozzy Osbourne, explaining in a column in *The Sunday Times* why he let a company sequence his genome.

Science
Nov. 19, 2010

I. General NHGRI Updates

II. General NIH Updates

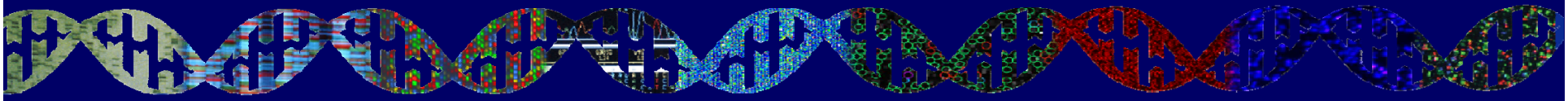
III. Genomics Updates

IV. NHGRI Extramural Program

V. NIH Common Fund Programs

VI. NHGRI Office of the Director

VII. NHGRI Intramural Program



Large-Scale Sequencing Program: RFAs Issued

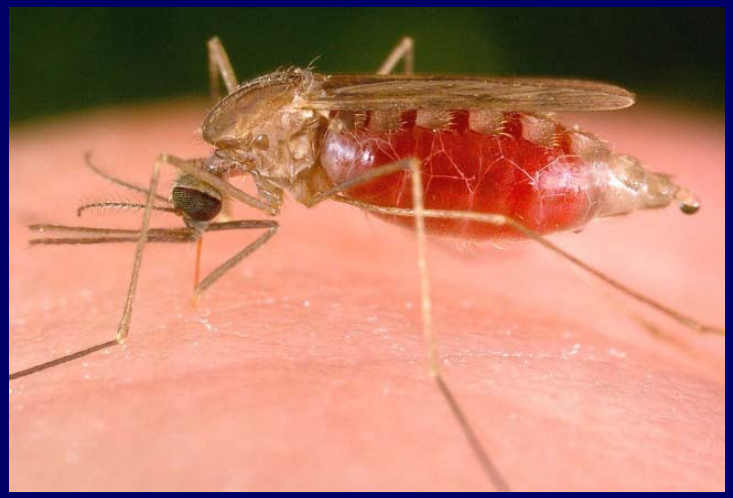
- Genome Sequencing & Analysis Centers (U54)
- Mendelian Disorders Genome Centers (U54)
- Clinical Sequencing Exploratory Research (U01)
- Informatics Tools for High-Throughput Sequence Data Analysis (U01 and R43/R44)

Letters of Intent Due : February 3, 2011

Application Due: March 3, 2011

Document 35

Large-Scale Sequencing Program: Recently Sequenced Genomes



- *Anopheles gamibae*

Widespread Divergence Between Incipient *Anopheles gambiae* Species Revealed by Whole Genome Sequences

M. K. N. Lawniczak,[1*] S. J. Emrich,[2*] A. K. Holloway,[3] A. P. Regier,[2] M. Olson,[2] B. White,[4] S. Redmond,[1] L. Fulton,[5] E. Appelbaum,[5] J. Godfrey,[5] C. Farmer,[5] A. Chinwalla,[5] S.-P. Yang,[5] P. Minx,[5] J. Nelson,[5] K. Kyung,[5] B. P. Walenz,[6] E. Garcia-Hernandez,[6] M. Aguiar,[6] L. D. Viswanathan,[6] Y.-H. Rogers,[6] R. L. Strausberg,[6] C. A. Saski,[7] D. Lawson,[8] F. H. Collins,[4] F. C. Kafatos,[1] G. K. Christophides,[1] S. W. Clifton,[5] E. F. Kirkness,[6] N. J. Besansky[4]†

- White nose fungus

Document 36

Large-Scale Sequencing Program: Orangutan Genome Sequenced

Comparative and demographic analysis of orang-utan genomes

Devin P. Locke[1], LaDeana W. Hillier[1], Wesley C. Warren[1], Kim C. Worley[2], Lynne V. Nazareth[2], Donna M. Muzny[2], Shiaw-Pyng Yang[1], Zhengyuan Wang[1], Asif T. Chinwalla[1], Pat Minx[1], Makedonka Mitreva[1], Lisa Cook[1], Kim D. Delehaunty[1], Catrina Fronick[1], Heather Schmidt[1], Lucinda A. Fulton[1], Robert S. Fulton[1], Joanne O. Nelson[1], Vincent Magrini[1], Craig Pohl[1], Tina A. Graves[1], Chris Markovic[1], Andy Cree[2], Huyen H. Dinh[2], Jennifer Hume[2], Christie L. Kovar[2], Gerald R. Fowler[2], Gerton Lunter[3,4], Stephen Meader[3], Andreas Heger[3], Chris P. Ponting[3], Tomas Marques-Bonet[5,6], Can Alkan[5], Lin Chen[5], Ze Cheng[5], Jeffrey M. Kidd[5], Evan E. Eichler[5,7], Simon White[8], Stephen Searle[8], Albert J. Vilella[9], Yuan Chen[9], Paul Flicek[9], Jian Ma[10]†, Brian Raney[10], Bernard Suh[10], Richard Burhans[11], Javier Herrero[9], David Haussler[10], Rui Faria[6,12], Olga Fernando[6,13], Fleur Darré[6], Domènec Farré[6], Elodie Gazave[6], Meritxell Oliva[6], Arcadi Navarro[6,14], Roberta Roberto[15], Oronzo Capozzi[15], Nicoletta Archidiacono[15], Giuliano Della Valle[16], Stefania Purgato[16], Mariano Rocchi[15], Miriam K. Konkel[17], Jerilyn A. Walker[17], Brygg Ullmer[18], Mark A. Batzer[17], Arian F. A. Smit[19], Robert Hubley[19], Claudio Casola[20], Daniel R. Schrider[20], Matthew W. Hahn[20], Victor Quesada[21], Xose S. Puente[21], Gonzalo R. Ordoñez[21], Carlos López-Otín[21], Tomas Vinar[22], Brona Brejova[22], Aakrosh Ratan[11], Robert S. Harris[11], Webb Miller[11], Carolin Kosiol[23], Heather A. Lawson[24], Vikas Taliwal[25], André L. Martins[25], Adam Siepel[25], Arindam RoyChoudhury[26], Xin Ma[25], Jeremiah Degenhardt[25], Carlos D. Bustamante[27], Ryan N. Gutenkunst[28], Thomas Mailund[29], Julien Y. Dutheil[29], Asger Hobolth[29], Mikkel H. Schierup[29], Oliver A. Ryder[30], Yuko Yoshinaga[31], Pieter J. de Jong[31], George M. Weinstock[1], Jeffrey Rogers[2], Elaine R. Mardis[1], Richard A. Gibbs[2] & Richard K. Wilson[1]

Document 36

1000 Genomes Project

ARTICLE

doi:10.1038/nature09534

A map of human genome variation from population-scale sequencing

The 1000 Genomes Project Consortium*

The 1000 Genomes Project aims to provide a deep characterization of human genome sequence variation as a foundation for investigating the relationship between genotype and phenotype. Here we present results of the pilot phase of the project, designed to develop and compare different strategies for genome-wide sequencing with high-throughput platforms. We undertook three projects: low-coverage whole-genome sequencing of 179 individuals from four populations; high-coverage sequencing of two mother–father–child trios; and exon-targeted sequencing of 697 individuals from seven populations. We describe the location, allele frequency and local haplotype structure of approximately 15 million single nucleotide polymorphisms, 1 million short insertions and deletions, and 20,000 structural variants, most of which were previously undescribed. We show that, because we have catalogued the vast majority of common variation, over 95% of the currently accessible variants found in any individual are present in this data set. On average, each person is found to carry approximately 250 to 300 loss-of-function variants in annotated genes and 50 to 100 variants previously implicated in inherited disorders. We demonstrate how these results can be used to inform association and functional studies. From the two trios, we directly estimate the rate of ***de novo*** germline base substitution mutations to be approximately 10^{-8} per base pair per generation. We explore the data with regard to signatures of natural selection, and identify a marked reduction of genetic variation in the neighbourhood of genes, due to selection at linked sites. These methods and public data will support the next phase of human genetic research.

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Current 1000 Genomes Data

- Now in full-scale production
- Expect to identify >35M SNPs and >2M indels from 26 Tb of whole-genome and -exome sequence data on ~1,000 individuals
- Even before the release of variants in ~1,000 samples, seeing 3,000 data downloads per month

1000 Genomes

A Deep Catalog of Human Genetic Variation

DNA Sequencing Technology

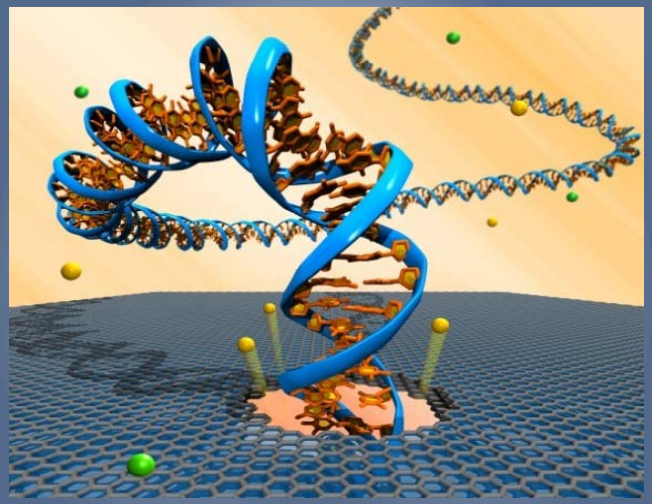
- Annual Grantee Meeting: April 4-6
- Public Meeting with Grantees: April 6-7

NHGRI Advanced Sequencing Technology Development Meeting

Wednesday, April 6 - Thursday, April 7, 2011

Catamaran Resort/Hotel
San Diego, California

Organized by:
The National Human Genome Research Institute (NHGRI)



:: Purpose
:: Agenda
:: Abstract/Poster information
:: Logistics/Hotel Information
:: Registration
:: Contact Information
:: Current Awards

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modENCODE

Integrative Analysis of the *Caenorhabditis elegans* Genome by the modENCODE Project

Mark B. Gerstein,[1,2,3*†] Zhi John Lu,[1,2*] Eric L. Van Nostrand,[4*] Chao Cheng,[1,2*] Bradley I. Arshinoff,[5,6*] Tao Liu,[7,8*] Kevin Y. Yip,[1,2*] Rebecca Robilotto,[1*] Andreas Rechtsteiner,[9*] Kohta Ikegami,[10*] Pedro Alves,[1*] Aurelien Chateigner,[11*] Marc Perry,[5*] Mitzi Morris,[12*] Raymond K. Auerbach,[1*] Xin Feng,[5,22*] Jing Leng,[1*] Anne Vielle,[13*] Wei Niu,[14,15*] Kahn Rhrissorrakrai,[12*] Ashish Agarwal,[2,3] Roger P. Alexander,[1,2] Galt Barber,[16] Cathleen M. Brdlik,[4] Jennifer Brennan,[10] Jeremy Jean Brouillet,[4] Adrian Carr,[11] Ming-Sin Cheung,[13] Hiram Clawson,[16] Sergio Contrino,[11] Luke O. Dannenberg,[17] Abby F. Dernburg,[18] Arshad Desai,[19] Lindsay Dick,[38] Andréa C. Dosé,[18] Jiang Du,[3] Thea Egelhofer,[9] Sevinc Ercan,[10] Ghia Euskirchen,[14] Brent Ewing,[20] Elise A. Feingold, Michelle Gutwein, Stefan R. Henz,[2] Judith Janette,[15] Vishal Khivansara, Isabel Latorre,[13] Rebecca F. Lowdo, Marco Mangone, David M. Miller, Taryn Phippen,[9] Joel Rozowsky,[1,] Andrea Sboner,[1] Cindie Slightam, Teruaki Takasaki, Christina M. Whi, Xingliang Zhou,[1] Kristin C. Gunsal, LaDeana W. Hill, Lincoln Stein,[5,6,]

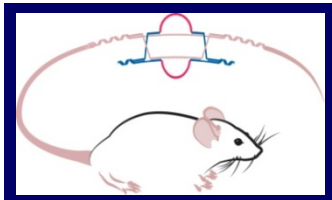
Identification of Functional Elements and Regulatory Circuits by *Drosophila* modENCODE

The modENCODE Consortium,* Sushmita Roy,[1,2†] Jason Ernst,[1,2†] Peter V. Kharchenko,[3†] Pouya Kheradpour,[1,2†] Nicolas Negre,[4†] Matthew L. Eaton,[5†] Jane M. Landolin,[6†] Christopher A. Bristow,[1,2†] Lijia Ma,[4†] Michael F. Lin,[1,2†] Stefan Washietl,[1†] Bradley I. Arshinoff,[7,18†] Ferhat Ay,[1,33†] Patrick E. Meyer,[1,30†] Nicolas Robine,[8†] Nicole L. Washington,[9†] Luisa Di Stefano,[1,31†] Eugene Berezikov,[23‡] Christopher D. Brown,[4‡] Rogerio Candeias,[1‡] Joseph W. Carlson,[6‡] Adrian Carr,[10‡] Irwin Jungreis,[1,2‡] Daniel Marbach,[1,2‡] Rachel Sealfon,[1,2‡] Michael Y. Tolstorukov,[3‡] Sebastian Will,[1‡] Artyom A. Alekseyenko,[11] Carlo Artieri,[12] Benjamin W. Booth,[6] Angela N. Brooks,[28] Qi Dai,[8] Carrie A. Davis,[13] Michael O. Duff,[14] Xin Feng,[13,18,35] Andrey A. Gorchakov,[11] Tingting Gu,[15] Jorja G. Henikoff,[o] Philipp Kapranov,[10] Renhua Li,[17] Heather K. MacAlpine,[3] John Malone,[12] Aki Minoda,[6] Jared Nordman,[22] Katsutomo Okamura,[8] Marc Perry,[18] Sara K. Powell,[5] Nicole C. Riddle,[15] Akiko Sakai,[29] Anastasia Samsonova,[19] Jeremy E. Sandler,[6] Yuri B. Schwartz,[3] Noa Sher,[22] Rebecca Spokony,[4] David Sturgill,[12] Marijke van Baren,[20] Kenneth H. Wan,[6] Li Yang,[14] Charles Yu,[6] Elise Feingold,[17] Peter Good,[17] Mark Guyer,[17] Rebecca Lowdon,[17] Kami Ahmad,[29] Justen Andrews,[21] Bonnie Berger,[1,2] Steven E. Brenner,[28,32] Michael R. Brent,[20] Lucy Cherbas,[21,24] Sarah C. R. Elgin,[15] Thomas R. Gingeras,[13,16] Robert Grossman,[4] Roger A. Hoskins,[6] Thomas C. Kaufman,[21] William Kent,[34] Mitzi I. Kuroda,[11] Terry Orr-Weaver,[22] Norbert Perrimon,[19] Vincenzo Pirrotta,[27] James W. Posakony,[26] Bing Ren,[26] Steven Russell,[10] Peter Cherbas,[21,24] Brenton R. Graveley,[14] Suzanna Lewis,[9] Gos Micklem,[10] Brian Oliver,[12] Peter J. Park,[3] Susan E. Celniker,[6§‖] Steven Henikoff,[25§‖] Gary H. Karpen,[6,28§‖] Eric C. Lai,[8§‖] David M. MacAlpine,[5§‖] Lincoln D. Stein,[18§‖] Kevin P. White,[4§‖] Manolis Kellis[1,2‖]

Document 39

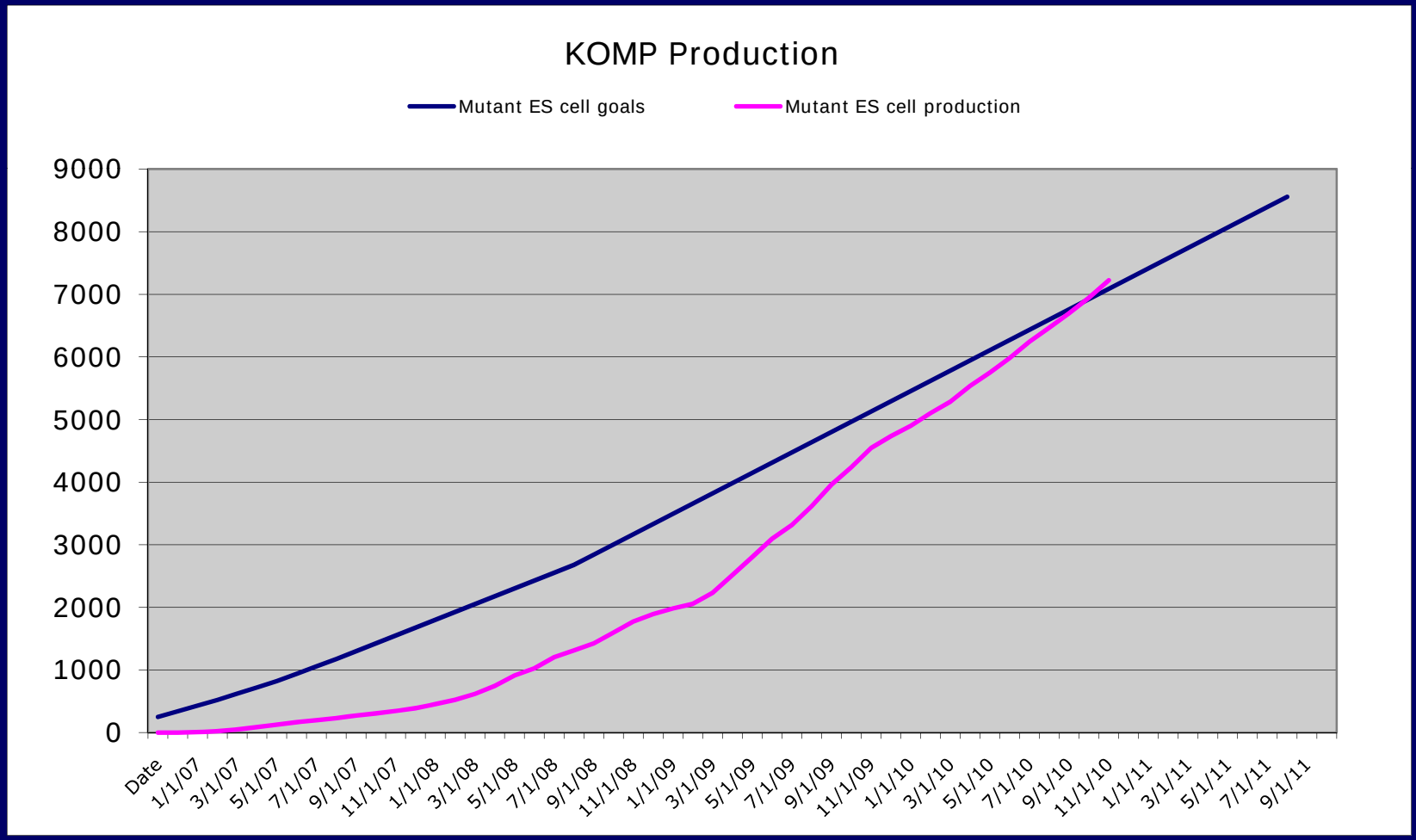
ENCODE

- ENCODE Consortium Meeting:
  November 2010
- ENCODE Analysis Workshop:
  March 2011
- Joint mod/ENCODE Consortia Meeting:
  May 2011
- ‘User’s Guide to ENCODE Data’ paper under revision



Knockout Mouse Program (KOMP)

- **ES cell production on track to meet KOMP goals in Fall, 2011**



Centers of Excellence in Genome Science

Annual CEGS Meeting
Arizona State University
(October 2010)

Diversity Action Plan (DAP)

October 2010 Meeting in Arizona

- **Progress Reports**
- **IRB workshop**
- **T32 measures of program success**

ELSI Funding Opportunities

- **RFA-HG-11-003: Development of a Preliminary Evidence Base to Inform Decision-making about Returning Research Results to Participants in Genomics Studies (R01)**

- **RFA-HG-11-004: Ethical, Legal, and Social Implications of Returning Research Results to Genomic Research Participants (R21)**

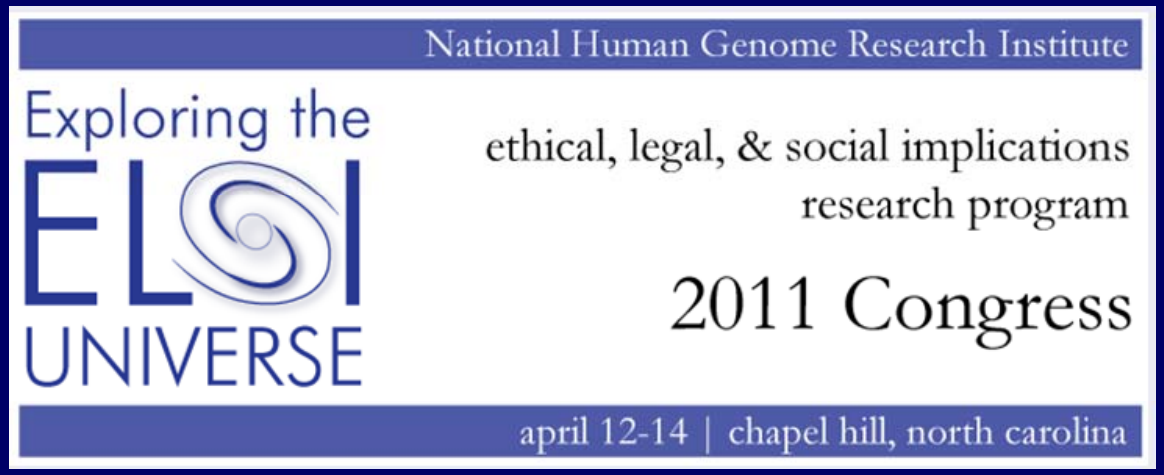
- **RFA HG-10-017: Clinical Sequencing Exploratory Research (U01) (requires ELSI research as one of three main components)**

- **Revised NIH-wide Bioethics FOA to incorporate many core ELSI and genomic issues**

ELSI Program Events

- October 2010: Centers of Excellence in ELSI Research (CEERS) Meeting

- April 2011: NHGRI ELSI Congress (Chapel Hill, NC)



- April 2011: eMERGE C&CC Policy Meeting

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I. General NHGRI Updates

II. General NIH Updates

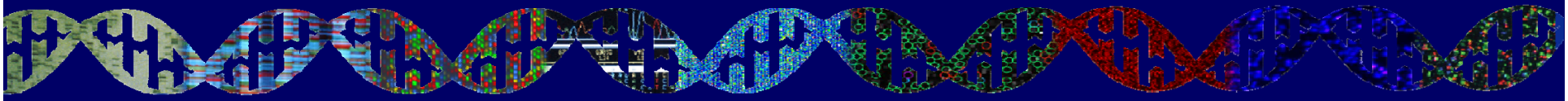
III. Genomics Updates

IV. NHGRI Extramural Program

V. NIH Common Fund Programs

VI. NHGRI Office of the Director

VII. NHGRI Intramural Program



Human Microbiome Project

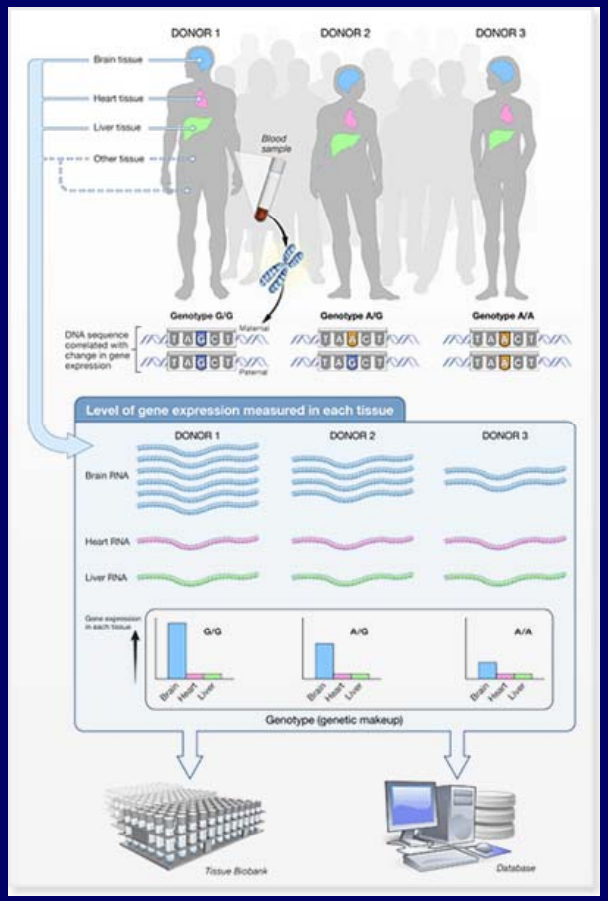
- Clinical sampling completed in October 2010

- 9 demonstration projects ramped up

- International Human Microbiome Congress in Vancouver, BC (March 2011)

Genotype-Tissue Expression (GTEx)

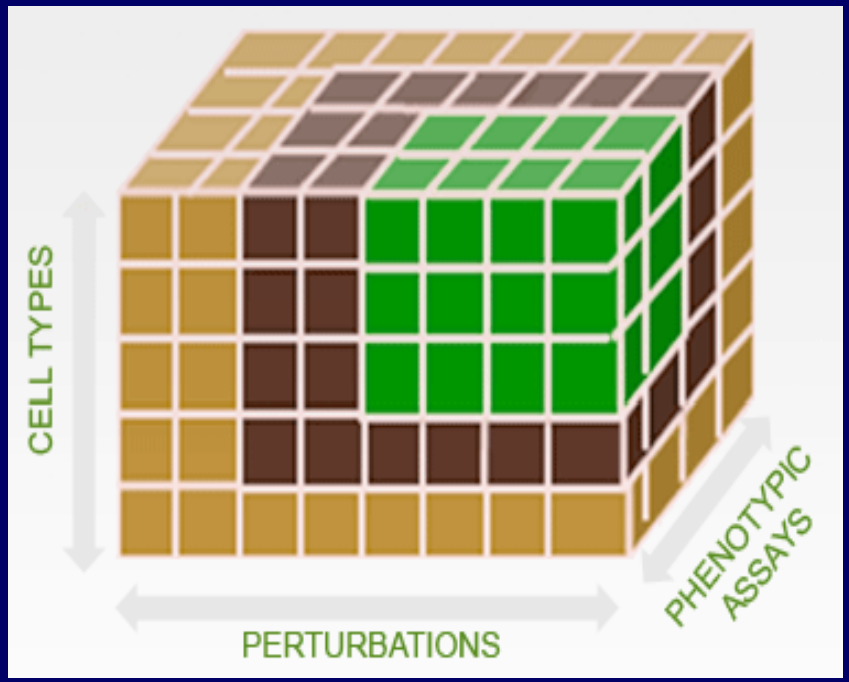
- 1 Lab/Coordinating Center and 3 Biospecimen Source Sites
- Collections by April/May 2011
- PI Meetings:
  - Dec 2010
  - June 2011
- 2nd Meeting to involve External Scientific Panel and R01 grantees and genome browser groups



Document 43

Library of Integrated Network-based Cellular Signatures (LINCS)

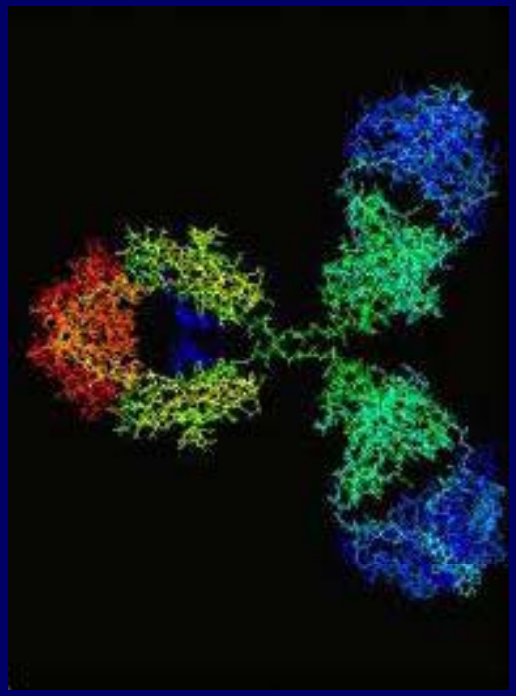
- Established data production metrics with U54 centers (for 1st year)
- Established External Scientific Panel
- Effort to engage non-cancer community to work with LINCS data



Document 44

Protein Capture Reagents

- Goal: Develop a renewable resource of protein capture reagents against human transcription factors



- 3 Components:
  - Antigen generation
  - Production of reagents
  - Methods development

- Long-Term Aim: inform possible future effort for whole human proteome

Document 45

Human Heredity and Health in Africa (H3Africa)

- White paper with recommendations for scientific scope of H3Africa now posted on the website (H3Africa.org)
- Meeting in Cape Town to discuss white paper (March 2011)

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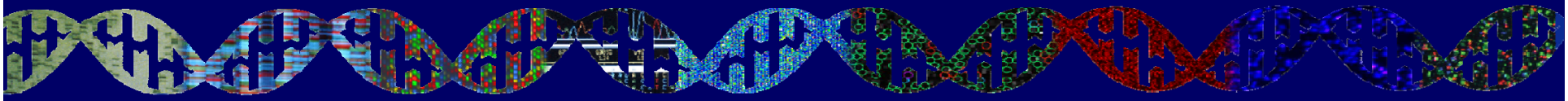
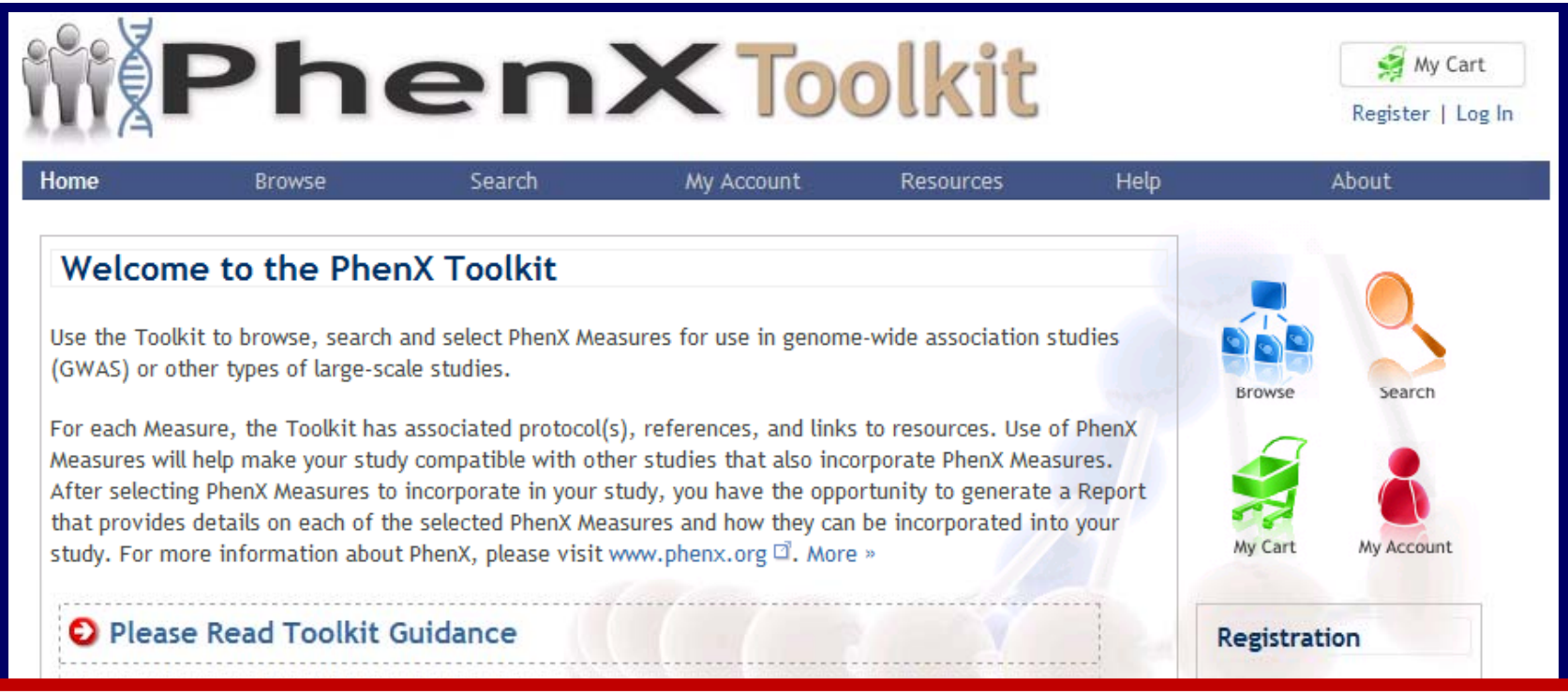


Office of Population Genomics

- GENEVA has released GWA genotyping data on 15 studies and HapMap 3 imputed genotypes on 6 studies
- Responses to eMERGE network RFAs (HG 10-009 and -010) received in November
- NHGRI GWAS catalog now includes 794 publications and 1,225 SNPs associated at $p < 5 \times 10^{-8}$

Document 47



PhenX Toolkit: published 21 domains for a total of 291 standard measures thus far

Social Environments, Speech and Hearing, Infectious Diseases, Gastrointestinal, and Psychosocial measures are recent additions.

Document 47

Teri Manolio Returns from the Deep…

GuLF STUDY

A health study for oil spill clean-up workers and volunteers

Study Summary

The Gulf Long-term Follow-up (GuLF) Study will investigate potential short- and long-term health effects associated with clean-up activities following the Deepwater Horizon disaster in the Gulf of Mexico on April 20, 2010. Crude oil, burning oil, and the dispersants used during clean-up efforts contain a range of known and suspected toxins. Over 130,000 persons have completed safety training in preparation for participation in clean-up activities related to the spill or were deployed to the Gulf as part of the Federal military and civilian response to the spill.

NEJM Genomic Medicine Series

REVIEW ARTICLE

GENOMIC MEDICINE

W. Gregory Feero, M.D., Ph.D., and Alan E. Guttmacher, M.D., *Editors*

Ancestry and Disease in the Age of Genomic Medicine

Charles N. Rotimi, Ph.D., and Lynn B. Jorde, Ph.D.

HUMAN GENETIC
pace, and whole g
tions are now pub
netic variation is facilitatir
diseases varies among ind
insights that may improve t
edge is relevant to fundame
larities. Here, we provide a
variation and how it contri
tory, group identity, and he

REVIEW ARTICLE

GENOMIC MEDICINE

W. Gregory Feero, M.D., Ph.D., and Alan E. Guttmacher, M.D., *Editors*

Genomics, Type 2 Diabetes, and Obesity

Mark I. McCarthy, M.D.

TYPE 2 DIABETES, THOUGH POORLY UNDERSTOOD, IS KNOWN TO BE A DISease characterized by an inadequate beta-cell response to the progressive insulin resistance that typically accompanies advancing age, inactivity, and weight gain.[1] The disease accounts for substantial morbidity and mortality from adverse effects on cardiovascular risk and disease-specific complications such as blindness and renal failure.[2] The increasing global prevalence of type 2 diabetes is tied to rising rates of obesity[2] — in part a consequence of social trends toward higher energy intake and reduced energy expenditure. However, the mechanisms that underlie individual differences in the predisposition to obesity remain obscure.

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ASHG & NHGRI Policy Fellowship

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

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

Home › Careers & Training › Educational Programs › Genetics and Public Policy Fellowship

Educational Programs
- **Genetics and Public Policy Fellowship**
- Lindau-Nobel Laureates Meeting Nomination
- NHGRI Summer Workshop in Genomics

Genetics and Public Policy Fellowship

Share Print

The Genetics and Public Policy Fellowship application is now available at: http://www.ashg.org/pages/education_fellowship.shtml. All completed applications must be received by April 22, 2011.

Sponsored by:
The American Society of Human Genetics
The National Human Genome Research Institute
The National Institutes of Health

Keywords: what's this?
- educational programs
- careers and training
- fellowships
- ASHG

Past ASHG fellows at a recent GINA hearing on Capitol Hill. Left to right: Jennifer Leib, Derek Scholes, Daryl Pritchard, Michael Stebbins.

- Background
- Program Overview
- Rotations
- Activities
- Qualifications and Skills
- Selection Process and Application
- Contact

- **Current cycle will select the10th joint fellow**
- **Previous fellows work in many different sectors of science policy**

Journal of Nursing Scholarship Special Series

Editors:

Kathleen Calzone (NCI)

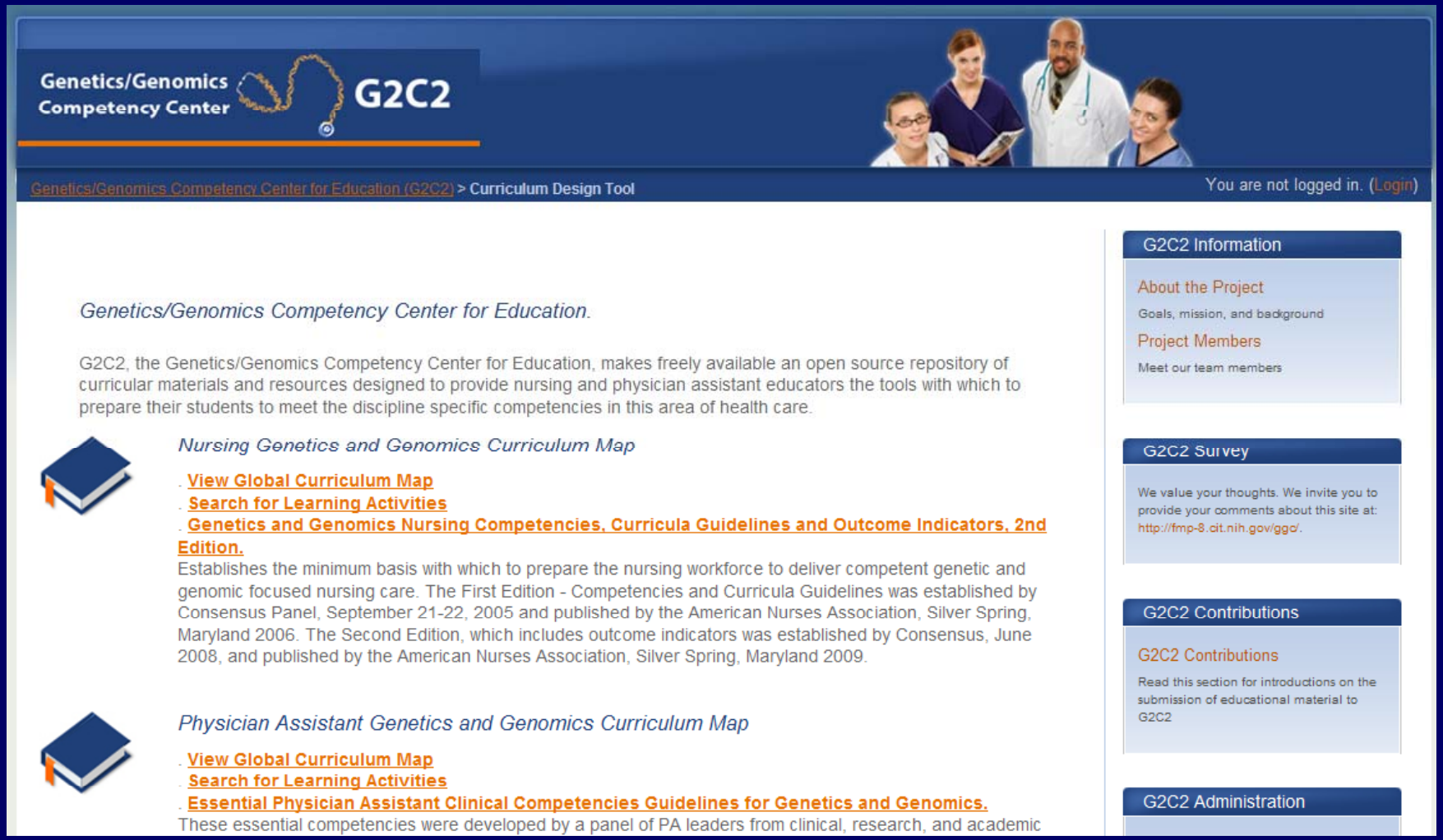
Jean Jenkins (NHGRI)

Begins: March 2011

Target: Nursing educators

Editorial and five articles

Genetics/Genomics Competency Center for Education (G2C2)



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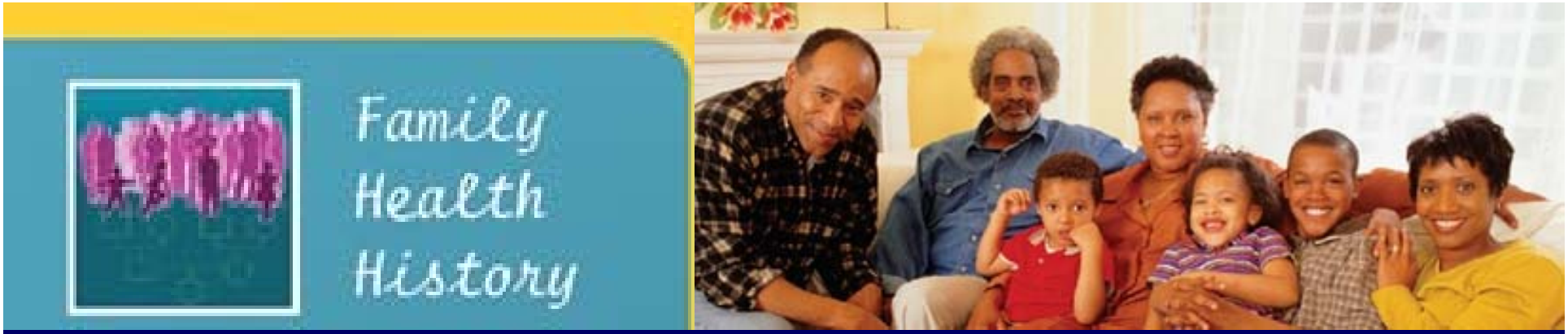
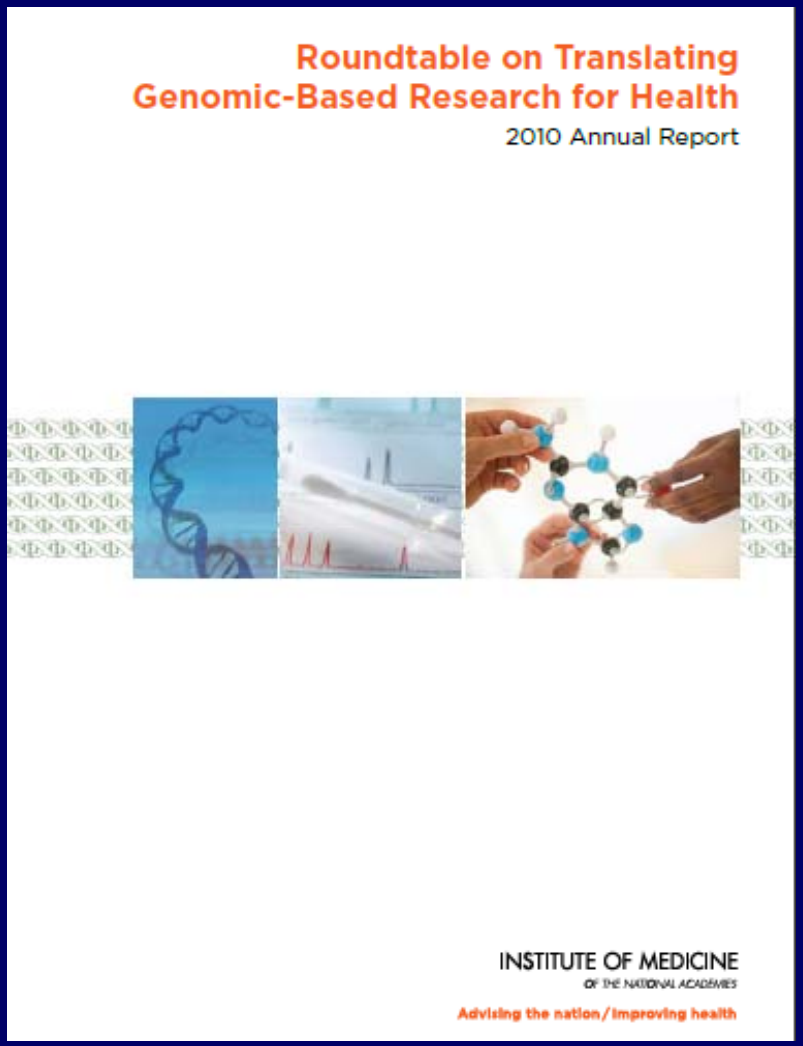


- **My Family Health Portrait (MFHP):**
  282,795 visitors in 2010
- **MFHP formally validated:**
  Facio et al., *Genetics in Medicine*, 2010
- **New trans-NIH committee established to develop a strategy for long-term governance (Greg Feero, Chair)**

Document 50

IOM Roundtable on Translating Genomic-Based Research for Health



- Three workshop reports in 2010
- Two meetings in 2011

Document 51

Faculty Champion Initiative

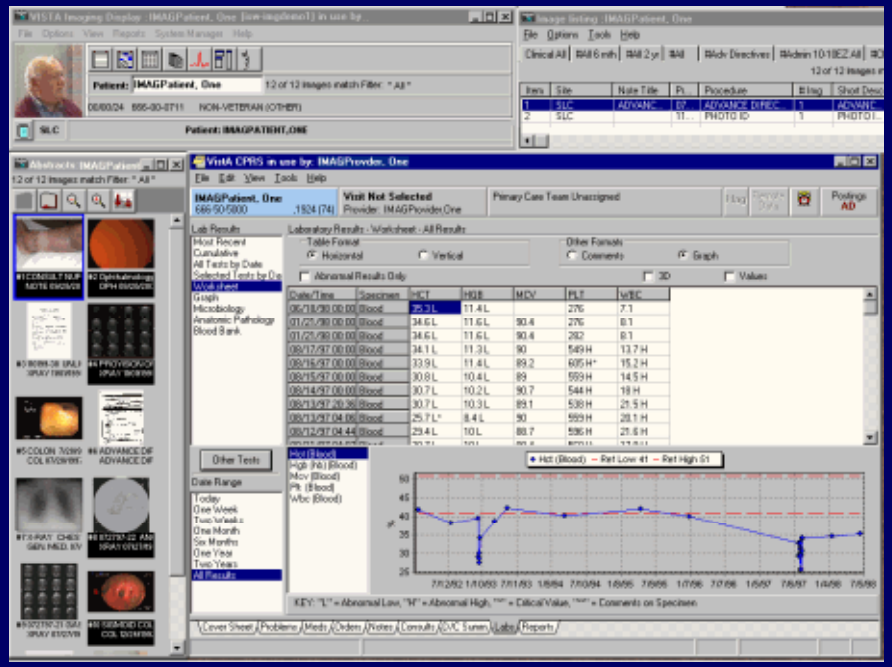
Document 52

Newborn Screening in the Genomics Era

- NHGRI, NICHD and ORDR
- Goals:
  - Generate crosstalk between the genomic technology and newborn screening communities
  - Designed to identify new research opportunities
- Report due this spring

Genomics and Health Information Technology Systems: Exploring the Issues

- Meeting: April, 2011
- Goal: Explore research and policy issues facing the integration of genomic information into health information technology systems



USA Science & Engineering Festival

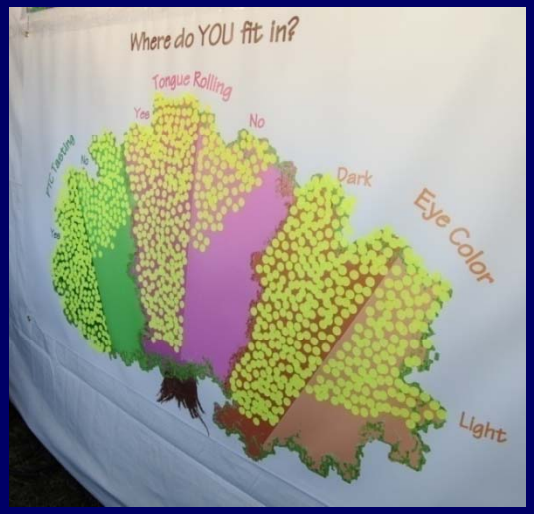
- Event : October 23-24, 2010 (Washington, DC)
- Location: National Mall and Freedom Plaza
- NIH Participants: 28 Institutes/Centers/Offices
- Visitors: ~500,000

Document 53

USA Science & Engineering Festival

NHGRI Activities:

- **Strawberry DNA Isolation**
- **A Tree of Genetic Traits**
- **DNA Bracelets**
- **Online Education Resources**
- **Game Show**



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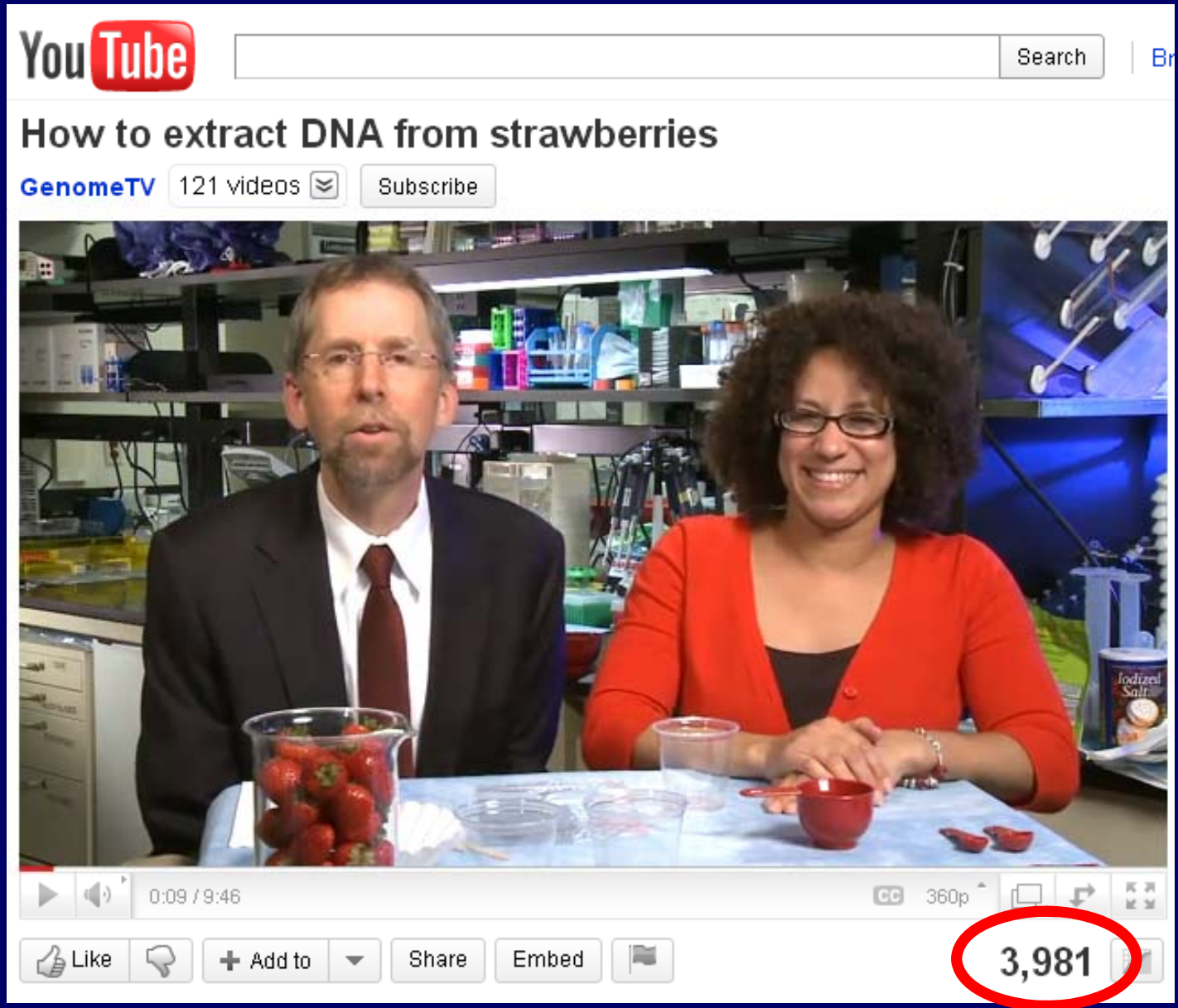
USA Science & Engineering Festival

USA Science & Engineering Festival



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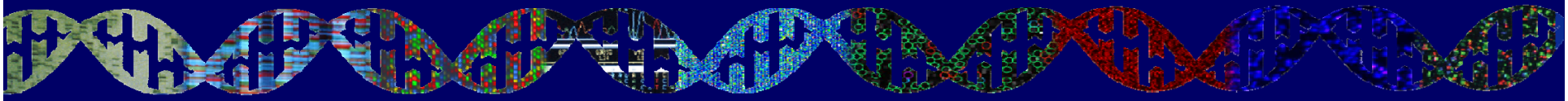
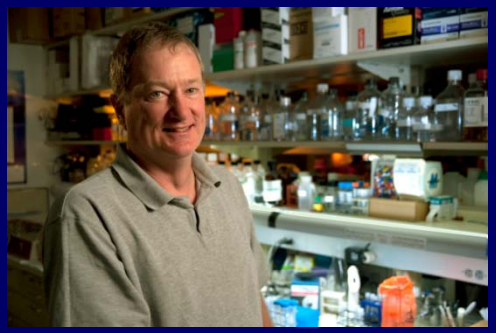


NHGRI Intramural Research Highlights

Human Gene Therapy

Adeno-associated virus serotype 8 (AAV8) Gene Transfer Rescues a Neonatal Lethal Murine Model of Propionic Acidemia

To cite this article:
Randy Joseph Chandler, Suma Chandrasekaran, Nuria Carrillo-Carrasco, Julien Simon Senac, Sean Hofherr, Michael A Barry, Charles Paul Venditti. Human Gene Therapy. -Not available-, ahead of print. doi:10.1089/hum.2010.164.



Mutation of a barrier insulator in the human ankyrin-1 gene is associated with hereditary spherocytosis

Patrick G. Gallagher,[1,2] Laurie A. Steiner,[1] Robert I. Liem,[3] Ashley N. Owen,[3] Amanda P. Cline,[3] Nancy E. Seidel,[3] Lisa J. Garrett,[3] and David M. Bodine[3]

[1]Departments of Pediatrics and [2]Genetics, Yale University School of Medicine, New Haven, Connecticut, USA.
[3]Hematopoiesis Section, Genetics and Molecular Biology Branch, National Human Genome Research Institute, NIH, Bethesda, Maryland, USA.

NT5E Mutations and Arterial Calcifications

Cynthia St. Hilaire, Ph.D., Shira G. Ziegler, B.A., Thomas C. Markello, M.D., Ph.D., Alfredo Brusco, Ph.D., Catherine Groden, M.S., Fred Gill, M.D., Hannah Carlson-Donohoe, B.A., Robert J. Lederman, M.D., Marcus Y. Chen, M.D., Dan Yang, M.D., Ph.D., Michael P. Siegenthaler, M.D., Carlo Arduino, M.D., Cecilia Mancini, M.Sc., Bernard Freudenthal, M.D., Horia C. Stanescu, M.D., Anselm A. Zdebik, M.D., Ph.D., R. Krishna Chaganti, M.D., Robert L. Nussbaum, M.D., Robert Kleta, M.D., Ph.D., William A. Gahl, M.D., Ph.D., and Manfred Boehm, M.D.

Document 54

Continued Media Coverage of UDP

The New York Times

Week in Review

HEALTH

Mysterious Maladies

By GINA KOLATA
Published: February 5, 2011

Patients who come to the Undiagnosed Disease Program at the National Institutes of Health know they're extremely sick. What they want to know is, what do they call their affliction.

Enlarge This Image

"We tell the patient we will do our best, but we have an 80 percent chance of failing," said Dr. William Gahl, the director of the program, which opened its doors in 2008.

For these patients, Dr. Gahl's program in suburban Washington is the destination of last resort. They have suffered for years, been prodded and poked by specialist after specialist, only to end up without

2011 Dr. Nathan Davis Award

- **Category: Outstanding Member of the Federal Executive Branch in Career Public Service**

National Institutes of Health / Johns Hopkins University Medical Genetics and Genomic Medicine Residency Training

- About us
- Application Process
- Contact
- Curriculum
- Diversity and Disability
- Eligibility Requirements
- Our Team
- Selectives

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

Special Thanks!


NHGRI