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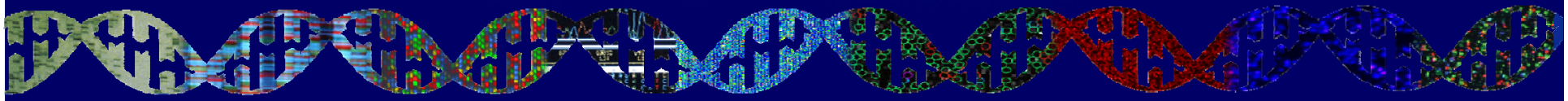
National Human Genome Research Institute
National Institutes of Health

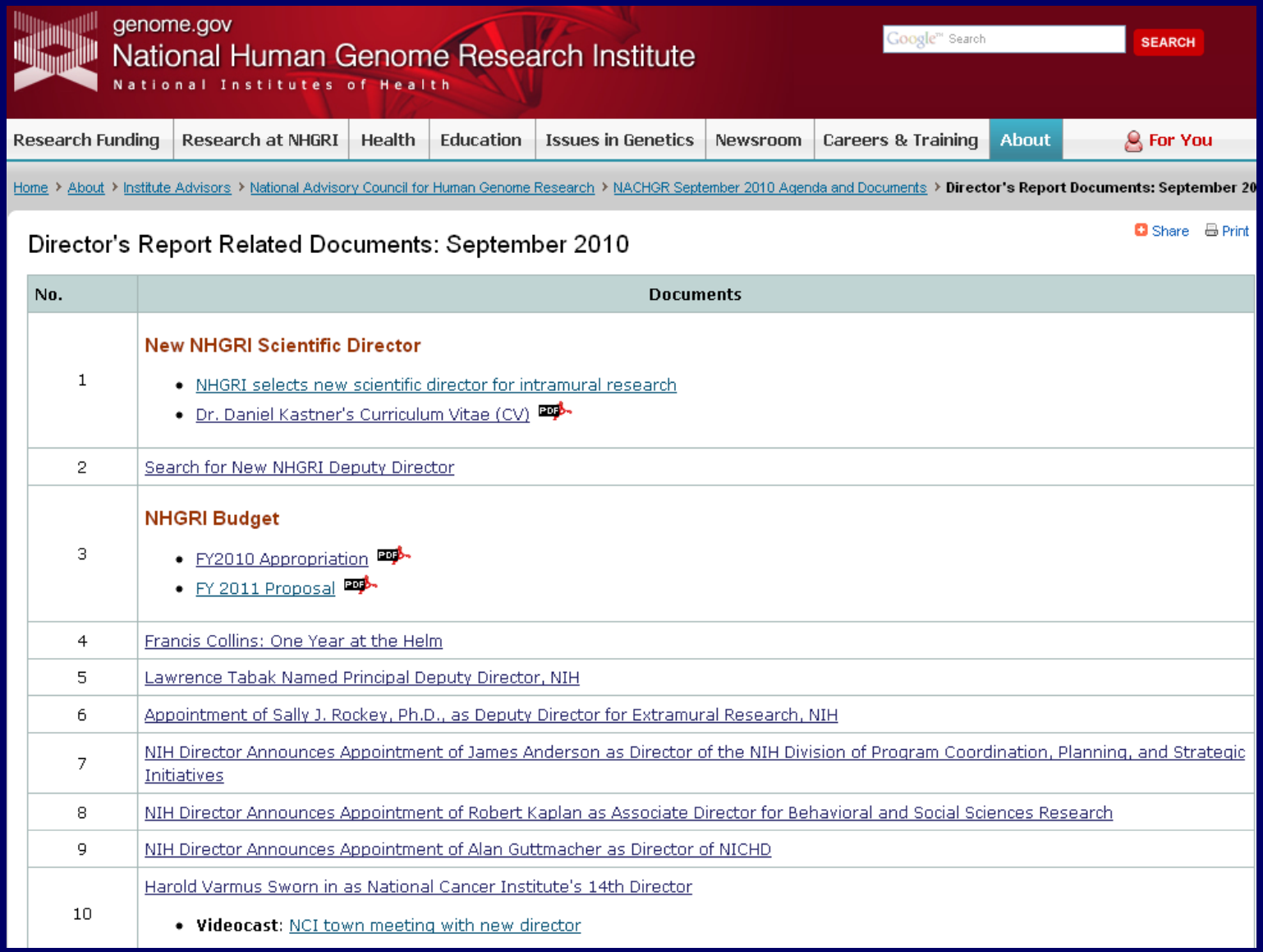
DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

September 2010

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





genome.gov/DirectorsReport

Document #

I. General NHGRI Updates

II. General NIH Updates

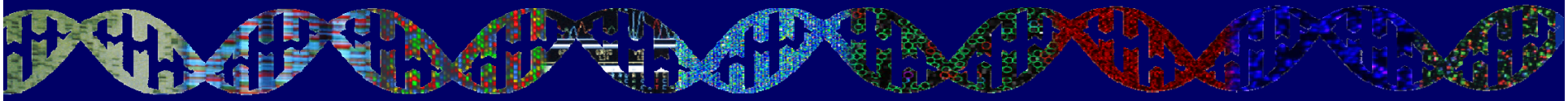
III. Genomics Updates

IV. NHGRI Extramural Program

V. NIH Common Fund Programs

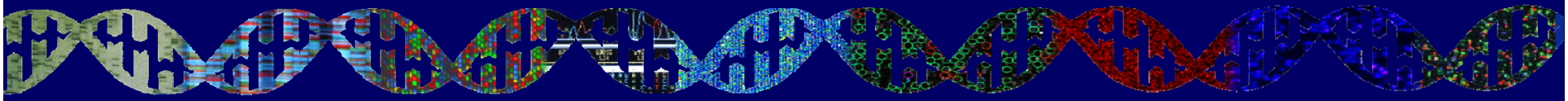
VI. NHGRI Office of the Director

VII. NHGRI Intramural Program



Open Session Presentations

- **Concept Clearances:**
  - **Jean McEwen (ELSI)**
  - **Susan Old (TRND)**
  - **Joan Bailey-Wilson (Intramural)**
- **Francis Collins: NIH Update**
- **Jane Peterson: Human Microbiome Project**
- **Bettie Graham, Kenneth Lange, & Janet Sinsheimer: NHGRI Training Programs**



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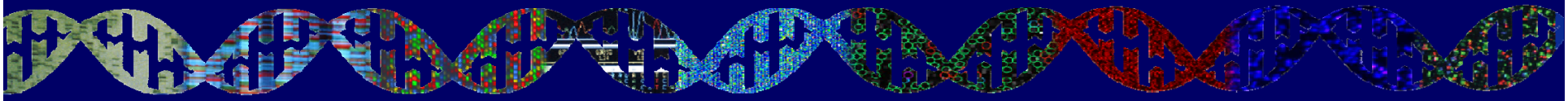


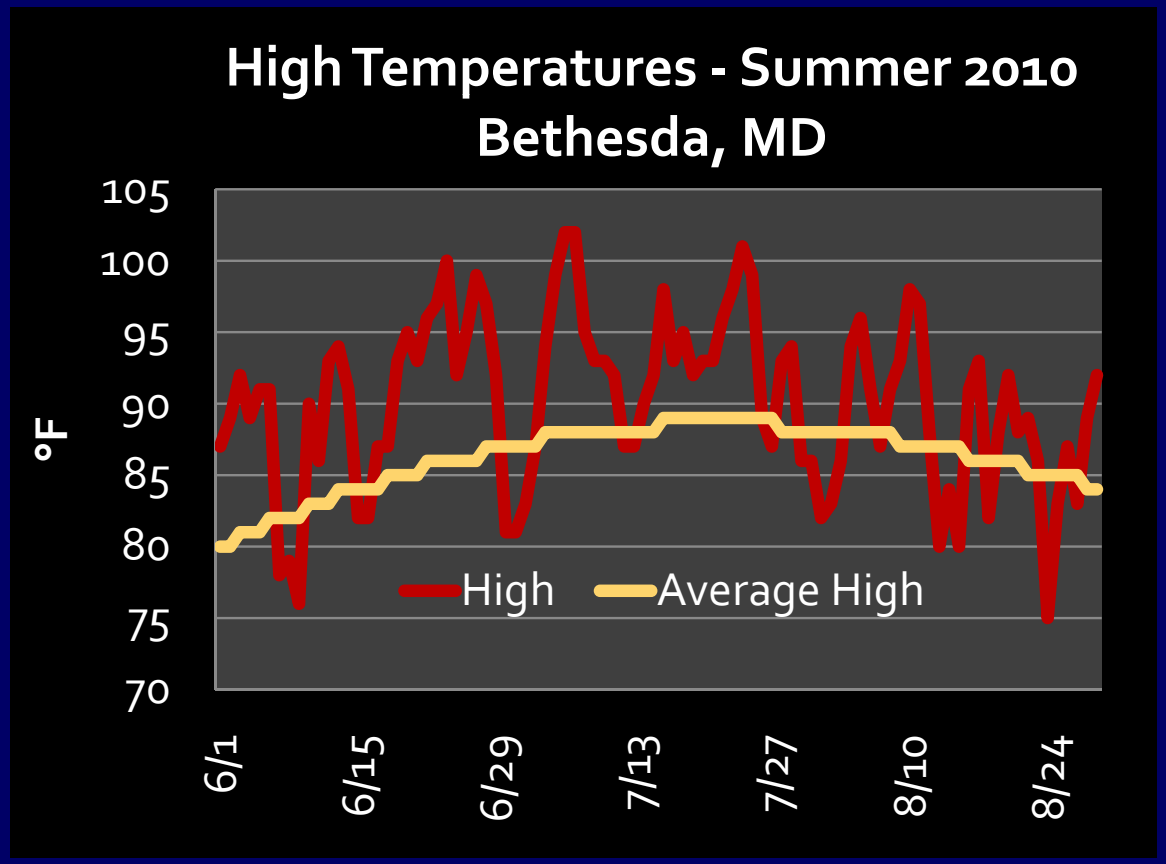
February 2010: WATER

Snowmageddon

Cancels February In-Person Council Meeting

Summer 2010: FIRE

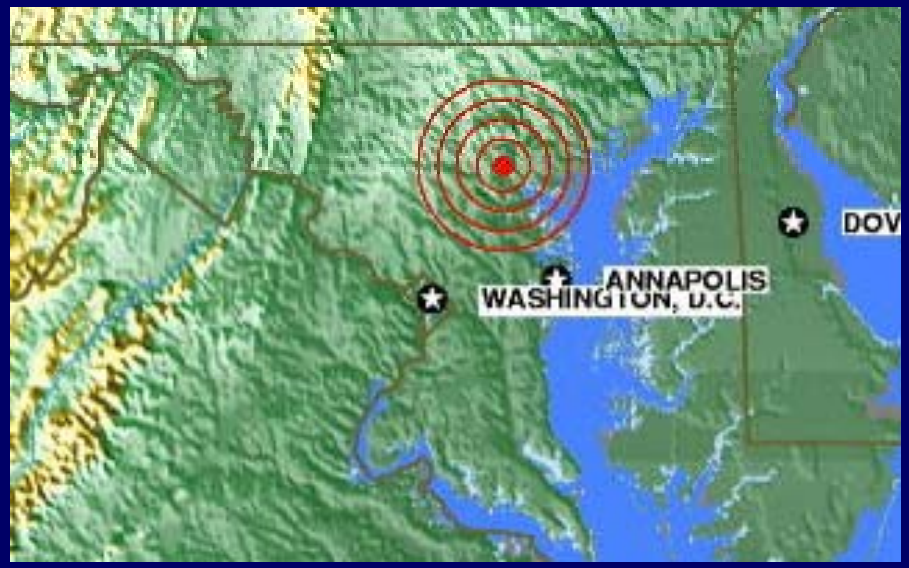
Forecast: Hot, with a chance of sweltering and sizzling



July 16, 2010: EARTH

3.7-magnitude earthquake ‘wakes’ Maryland residents

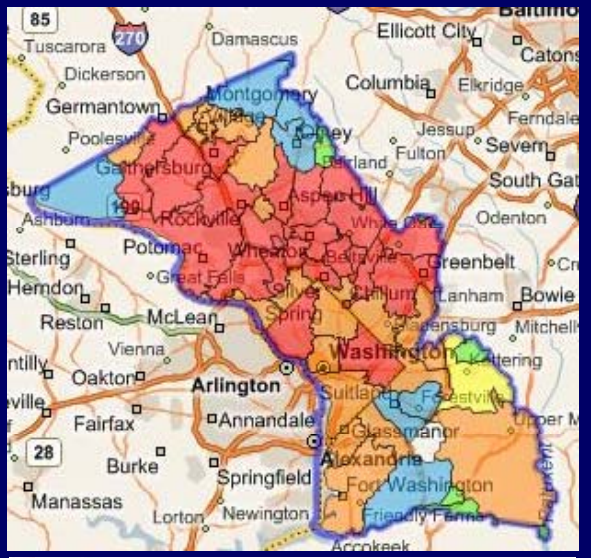
Quake centered near Gaithersburg; felt by as many as 3 million people in Mid-Atlantic region



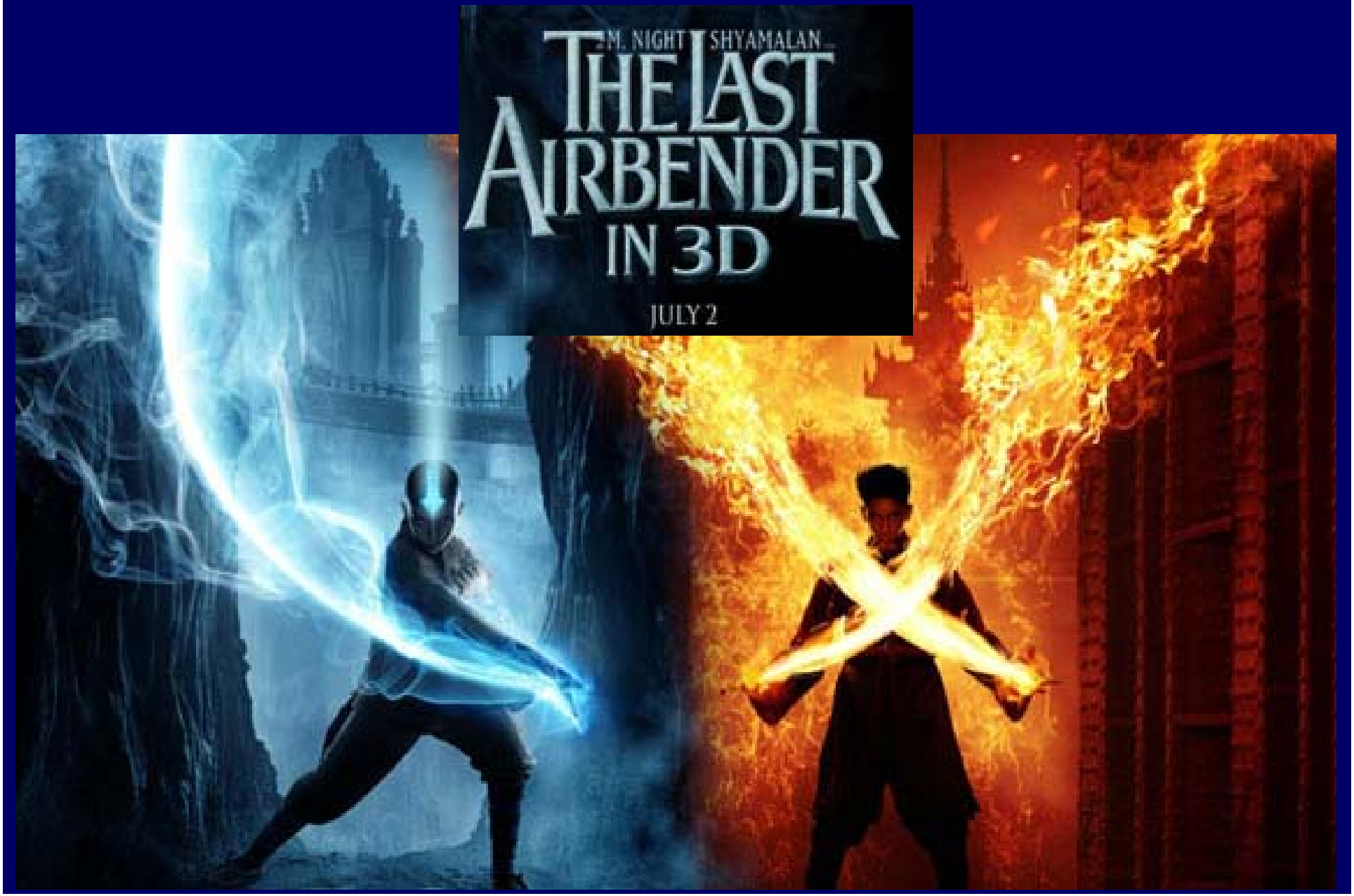
July 25, 2010: AIR

Montgomery County hit by storms with winds >75 mph

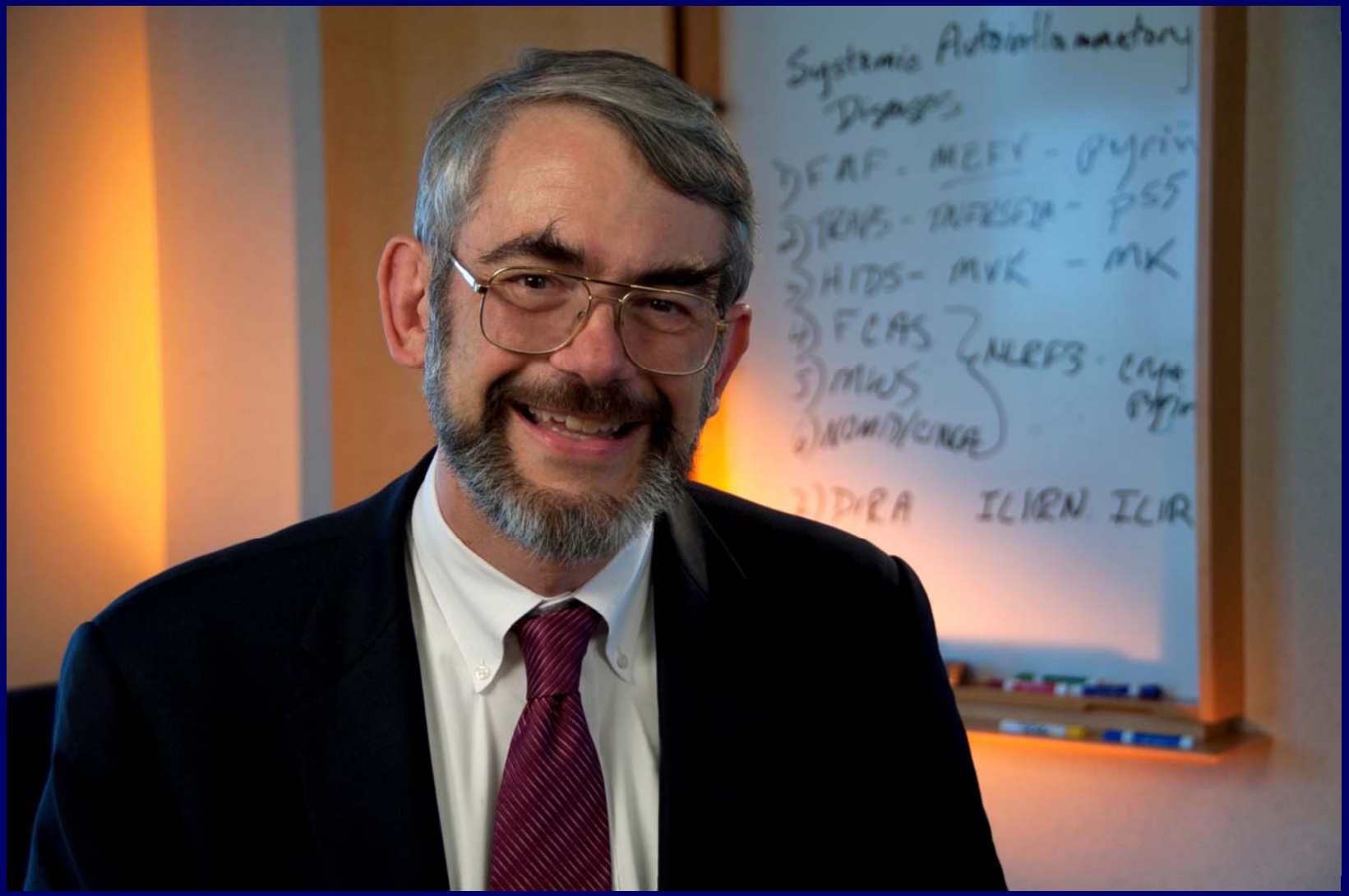
>300,000 customers lost power



NHGRI Leadership: Job Requirements



New NHGRI Scientific Director



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

Document 1

NHGRI Leadership Update

Scientific Director

Deputy Director

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 2

NHGRI Strategic Planning Process
Airlie 'Finale Meeting': July 6-8, 2010

NHGRI Strategic Planning Process
Airlie 'Finale Meeting': July 6-8, 2010

Airlie Meeting Remote Participants

- Videocasting of main sessions
- Remote participants included an elite group of ~100 genomics/genetics trainees
- Microblog

Airlie Meeting Microblog


srjefferys Group at http://groups.google.com/group/futureofgenomics
about a month ago from web at 36°0'48"N 78°54'42"W in context


orbitsoldier It's important to differentiate between medical science and genomic medicine, but I think the synergies between them should also be addressed in the draft.. eg collecting WGS in the clinic and making it available for research
about a month ago from web


srjefferys Just created the google group futureofgenomics. Please join! I'll be glad to transfer admin if desired.
about a month ago from web at 36°0'48"N 78°54'42"W


anne @cashspencer Yes, thanks. Sent another comment to @chriswellington re having a more complete transcript available at some point, too, if possible. Appreciate all you have done to help us be involved – even at a distance.
about a month ago from web at 41°38'52"N 91°36'16"W in context


ken @anne Anne, et al., although not all the notes, I have 5 pages of notes and url addresses. I would be happy to send to whoever, just let me know in direct email: kenfnp@ready2host.net
about a month ago from web in context


robertw @cashspencer I wouldn't say GWAS didn't work. I'm actually surprised they worked as well as they did (I was not one of the people hyping them up). But I do think they were subject to a great degree of irrational exuberance. The same, I think, is happening to whole genome sequencing where the statistical issues will dwarf those in GWAS.
about a month ago from web at 39°17'31"N 76°35'14"W in context


cross @orbitsoldier Sadly I don't off the top of my head but let me think about it. I am not a great off the top of my head person I am a plodder. So I'll get back to you.
about a month ago from web at 44°57'59"N 89°36'53"W

for example, the meta-discussion in the first 2 paragraphs of the "societal implications" textbox aren't about societal implications.


@robertw There is always the risk of giving people 'just enough knowledge to be dangerous' – if you teach people genomics but not statistics, it's easy to get artifactual results and not know how to avoid them... but the field won't ever progress if the two sides sit in their own buildings and don't start intertwining


The current speaker mentioned how grants in the early 90s from NHGRI led to important advances in cancer genomics. I think this is an example of a success that has resulted from genomics (which is a recurring theme). It strikes me that the people at this meeting are probably well qualified to come up with things like this. I think it would be good to poll them, while everyone is there, and make a list of "successes since the last document" that could be included in the current one. This will also provide "wow".


@timreddy Our lab is involved proteomics – based tagging of human genes as associated with detected peptides as part of the ENCODE project

FY 2009 PECASE Awardees

Four NHGRI-related Researchers

Chuck Venditti

Investigator
NHGRI

Brian Brooks

Investigator
NEI

Adjunct Investigator
NHGRI

Manolis Kellis

Associate Professor
MIT

Extramural Grantee
NHGRI

Bradley Malin

Assistant Professor
Vanderbilt

Extramural Grantee
NHGRI

NIH & NHGRI Appropriations Update

- **FY2010**
  - **NIH: $31B (2.3% increase)**
  - **NHGRI: $516M (2.7% increase)**

- **FY2011 (President's Budget)**
  - **NIH: $32B (3.2% increase)**
  - **NHGRI: $534M (3.5% increase)**

Document 3

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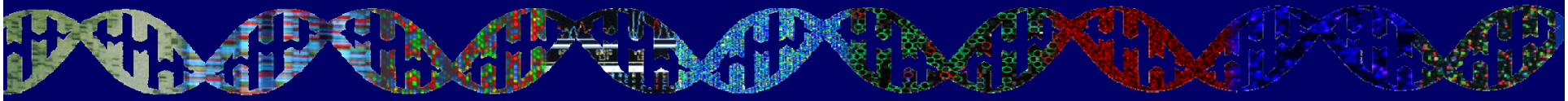


End of His Rookie Year

ONE YEAR AT THE HELM

Having taken on the biggest job in biomedicine — leading the US National Institutes of Health — Francis Collins must now help his agency over a funding cliff. **Meredith Wadman** looks at his record so far, and his plans to cushion the fall.

Document 4

New Principal Deputy Director, NIH

Larry Tabak, D.D.S, Ph.D.

Document 5

New Deputy Director for Extramural Research, NIH

Sally Rockey, Ph.D.

Document 6

New Director, NIH Division of Program Coordination, Planning, and Strategic Initiatives (DPCPSI)

James Anderson, M.D., Ph.D.

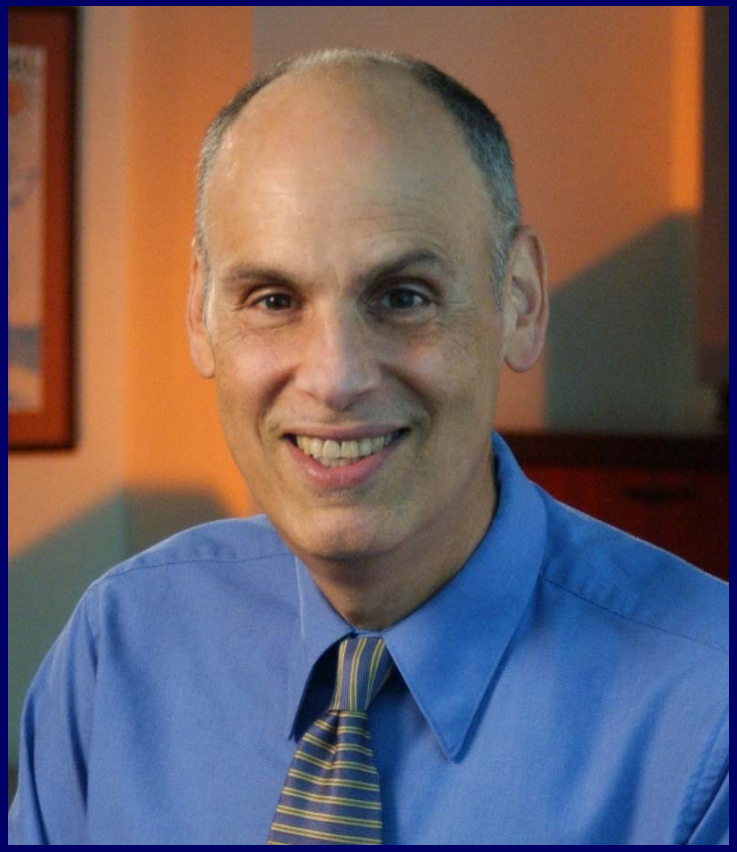
Document 7

New Director, NIH Office of Behavioral and Social Sciences Research (OBSSR)

Robert Kaplan, Ph.D.

Document 8

New Director, National Institute of Child Health and Development (NICHD)



Alan Guttmacher, M.D.

Document 9

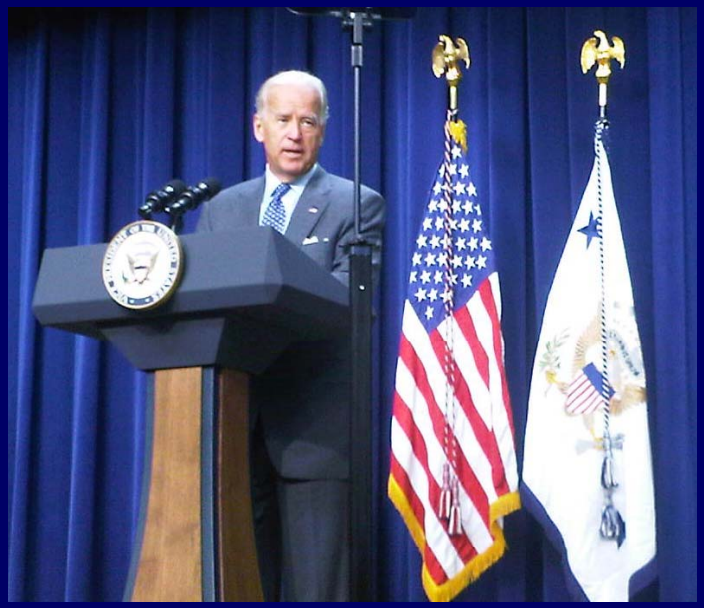
New Director, National Cancer Institute (NCI)

Harold Varmus, M.D.

New NHGRI Intramural Investigator

ARRA Report: August 24, 2010

THE RECOVERY ACT:
TRANSFORMING THE AMERICAN ECONOMY THROUGH INNOVATION



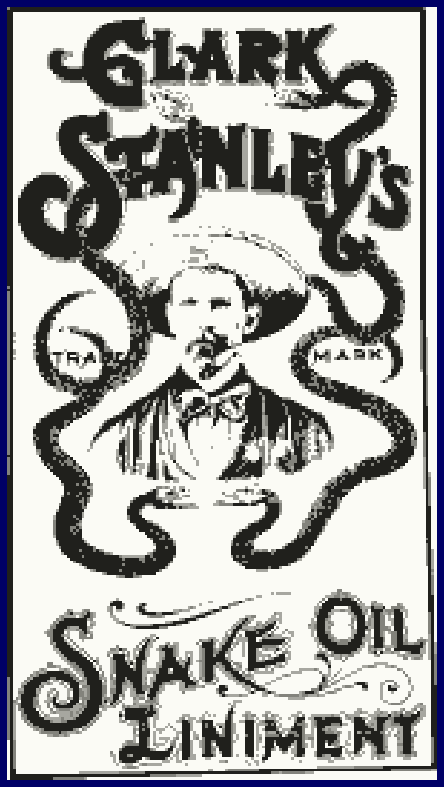
"…we stand on the verge of bringing the cost of a human genome map below $1,000 …"

"…disease prevention work is happening across the board: in human genome sequencing, cardiovascular disease, cancer, and autism."

"…NIH will be able to sequence the genes of cancers that affect 10 million Americans …"

Document 11

NIH Genetic Testing Registry



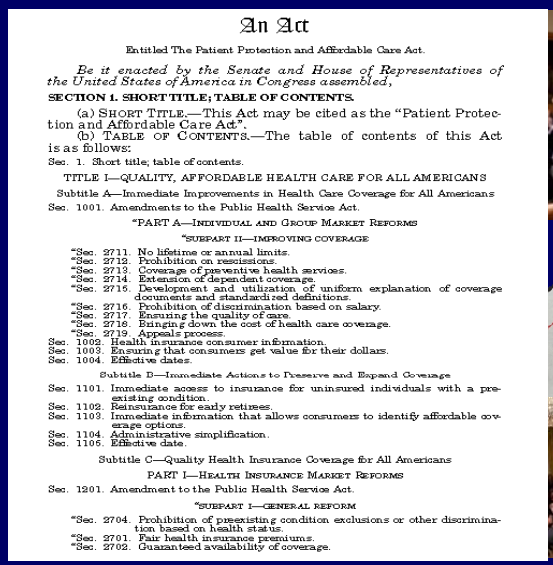
GENETIC TESTING **REGISTRY**

www.ncbi.nlm.nih.gov/gtr/

Document 12

Therapeutics @ NIH

- **Continued intense developmental activities**
  - **Therapeutics for Rare and Neglected Diseases (TRND) Program**
  - **Cures Acceleration Network (CAN)**
- **Presentation: Francis Collins**
- **Presentation: Susan Old**


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.

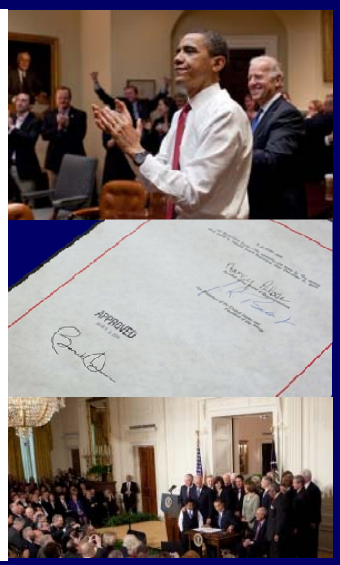
Subtitle C—Quality Health Insurance Coverage for All Americans

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.



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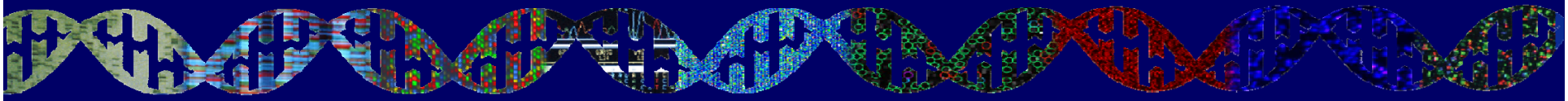

10th Anniversary: Draft Human Genome Sequence

NEWS FEATURE

NATURE Vol 464|24 June 2010

Ten Years On — The Human Genome and Medicine

The New York Times

June 12, 2010

A Decade Later, Genetic Map Yields Few New Cures

By NICHOLAS WADE

Ten years after President Bill Clinton announced that the first draft of the human genome was complete,

SPIEGEL ONLINE

07/29/2010 05:58 PM

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June 15, 2010

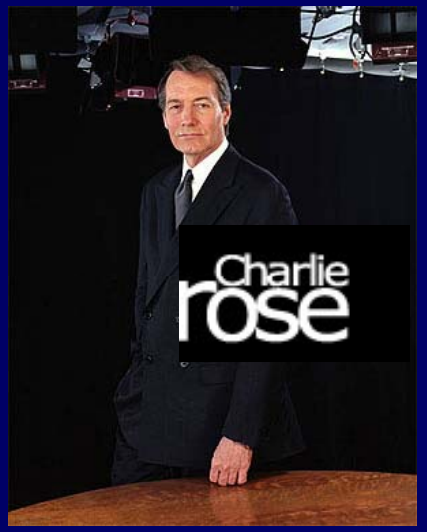
THE GENOME AT 10; Awaiting the Genome Payoff

By ANDREW POLLACK

NORTH WALES, Pa. -- At Merck's "automated biotechnology facility" here, robot arms adapted from automobile factories deftly shuttle plates containing human cells.

Assisted by the robots and other complex machinery, scientists are studying what happens to the cells as each of the roughly 22,000 human genes is turned off. They hope to find the genes involved in different diseases, the starting point for creating a drug.

It is a merger of sophisticated biology and brute force made possible only because the Human Genome Project provided the identity of all the human genes. But as with so much else that has spun off from the genome project, it is also an expensive gamble, with success far from assured.



Document 13

The Economist Series on Genomics

The Economist

A special report on the human genome

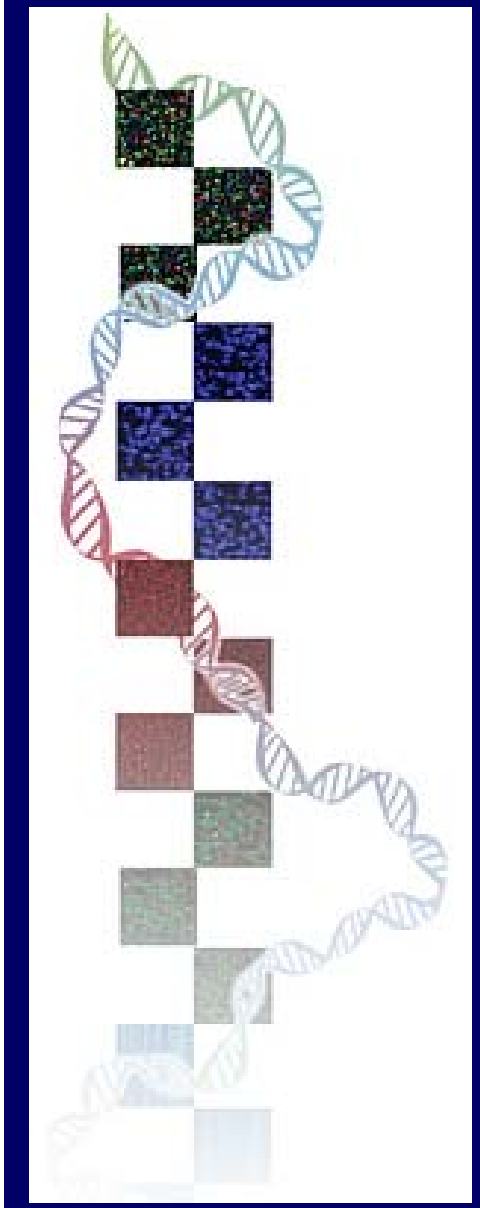
Biology 2.0

A decade after the human-genome project, writes Geoffrey Carr (interviewed here), biological science is poised on the edge of something wonderful

Jun 17th 2010

Document 14

NHGRI Science Writers Workshop



"The Human Genome: A Decade of Discovery, Creating a Healthy Future"

Goal: help science reporters write stories about the 10th anniversary of draft human genome sequence

Document 15

USA Today: July 8, 2010

USA TODAY

07/08/2010 - Updated 07:27 AM ET

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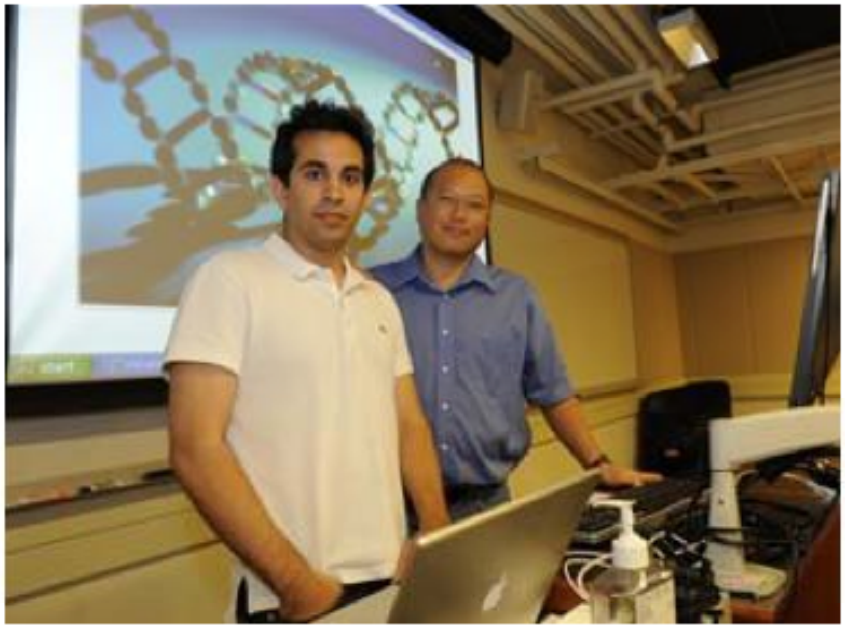
Genome gains seen as a beginning



CAPTION By Fred Mertz, for USA TODAY

Mexico schools teach survival

Schools across Mexico are teaching students to cover their heads as the violence-torn country sees more urban gunfights.

Which team will LeBron choose?

From sublime theater to manufactured drama, the LeBron James saga will end today.

LOPRESTI: Full-court press is stifling

With this chalupa, I thee wed

More couples are choosing their favorite retail spots as the backdrop for their special day. The shops range from T.J. Maxx to Taco Bell.

Document 13

Legislation

- **H.R. 5440: The Genomics and Personalized Medicine Act**
- **Francis Collins testifies at House Energy & Commerce Subcommittee on Health hearing:**

  "NIH in the 21st Century: The Director's Perspective"

Document 16

Personal Genomics Regulation

- FDA sent letters to 21 DTC genomics companies concerning lack of premarket approval for their tests
- House Committee on Energy and Commerce hearing on "Direct-To-Consumer Genetic Testing and the Consequences to the Public Health"

23andMe

Navigenics™

PATHWAY GENOMICS

Document 17

DTC Genetic Test Hearing

GAO Report on DTC Genetic Tests

GAO
Accountability · Integrity · Reliability

Highlights

Highlights of GAO-10-847T, a testimony before the Subcommittee on Oversight and Investigations, Committee on Energy and Commerce, House of Representatives

July 22, 2010

DIRECT-TO-CONSUMER GENETIC TESTS

Misleading Test Results Are Further Complicated by Deceptive Marketing and Other Questionable Practices

Why GAO Did This Study

In 2006, GAO investigated companies selling direct-to-consumer (DTC) genetic tests and testified that these companies made medically unproven disease predictions. Although new companies have since been touted as being more reputable—*Time* named one company's test 2008's "invention of the year"—experts remain concerned that the test results mislead consumers. GAO was asked to investigate DTC genetic tests currently on the

What GAO Found

GAO's fictitious consumers received test results that are misleading and of little or no practical use. For example, GAO's donors often received disease risk predictions that varied across the four companies, indicating that identical DNA samples yield contradictory results. As shown below, one donor was told that he was at below-average, average, and above-average risk for prostate cancer and hypertension.

Contradictory Risk Predictions for Prostate Cancer and Hypertension

Gender	Age	Condition	Company 1	Company 2	Company 3	Company 4
Male	48	Prostate cancer	Average	Average	Below average	Above average
		Hypertension	Average	Below average	Above average	Not tested

Source: GAO.

Document 17

Personal Genomics Regulation

- FDA held a two-day meeting to solicit public comments on how to proceed with regulation of lab developed tests (LDTs)
- 14 professional societies, 9 clinical labs, 26 companies, and 3 advocacy groups gave public comments
  - Fears of excessive regulatory burden
  - Calls not to duplicate current efforts (CLIA, CAP)
  - Need to address software analysis tools
  - Role for NIH's Genetic Test Registry?
- However, DTC genomics may not be considered LDTs according to FDA

Personal Genomes & Universities

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Office of Communication & Public Affairs

SHARE THIS VIA...

JUNE 7, 2010

Medical school to offer course that gives students option of studying their own genotype data

BY SUSAN IPAKTCHIAN

Charles Prober

It seems simple enough: Swab the inside of your cheek, mail it to a genetic testing company and a short time later you'll get a report about your risks for certain diseases. But if you then decide to share the information with your physician in hopes of getting more guidance, you may have a problem. Few doctors have the training to correctly interpret and advise their patients about the complex web of information that personal genome testing provides.

In a step toward preparing physicians and biomedical researchers for a future where genomic testing increasingly becomes part of medical care, the Stanford University School of Medicine is offering a new course in which medical and graduate students will be given the option of studying their personal genotype data. The students will learn how to analyze, evaluate and interpret the genetic data, the limitations of the existing technologies, and the legal and ethical issues raised by personal genotyping.

Document 18

Personal Genomes & Universities

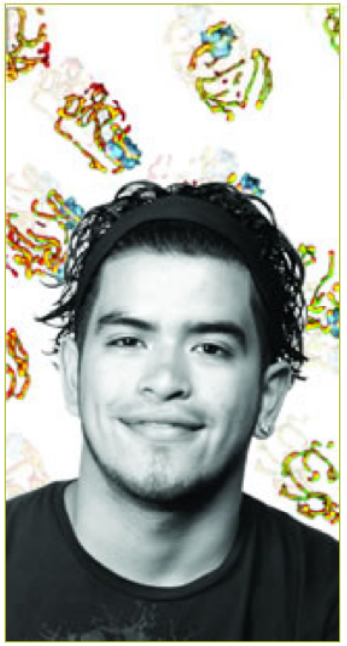
ON THE *Same Page*

BRING YOUR **GENES** TO CAL

College of Letters & Science
UC Berkeley

- CONTEST
- SEMINARS
- EVENTS
- PRESS COVERAGE
- OPEN LETTER
- FAQ
- ARCHIVE
- CONTACT US

On the Same Page gives new students in the College of Letters & Science something to talk about. This year, we'll all be on the same page exploring the theme of **Personalized Medicine**—the set of emerging technologies that promises to transform our ability to predict, diagnose, and treat human disease —with **Professor Jasper Rine** as our guide.



The program announcement has already generated a lot of discussion and questions, about everything from our reasons for tackling this topic to the safeguards we have put in place for our students. Dean Mark Schlissel provides answers in the FAQ. See also his open letter.

Participate in the Grand Experiment

All new L&S freshmen and transfers will receive a package in the mail over the summer, after they have submitted their statement of intent to register. This package will contain, among other things, a swab and a consent form. Send us a small sample of your own genetic material (along with your consent form) and Professor Rine will analyze it for variants of three genes that effect the impact of nutrition on your health. Participation is purely voluntary and completely anonymous. For more information on how the program works, see the FAQ.

Document 19

AJCP Article on Pathology Training

AJCP / Special Article

A Call to Action

Training Pathology Residents in Genomics and Personalized Medicine

Richard L. Haspel, MD, PhD,[1] Ramy Arnaout, MD, DPhil,[1] Lauren Briere, MS,[2] Sibel Kantarci, PhD,[1] Karen Marchand, MS,[2] Peter Tonellato, PhD,[1,3] James Connolly, MD,[1] Mark S. Boguski, MD, PhD,[1,3] and Jeffrey E. Saffitz, MD, PhD[1]

Document 20

Celebrity Genomes: Recent Addition

NYDailyNews.com
DAILY NEWS

News Sports Gossip Entertainment NY Events Local Opinion

Lifestyle › Scientists to map Ozzy Osbourne's genetic code to find out

Article Comments Email Print Facebook Twitter

Scientists to map Ozzy Osbourne's genetic code to find out how he survived so much substance abuse

BY NICK KLOPSIS
DAILY NEWS WRITER

Monday, June 14th 2010, 1:39 PM

stltoday.com
the #1 St. Louis website

Home News Business Sports Entertainment Health Life & Style Photos

What keeps Ozzy going? It could be in the genes

St. Louis • He is famous for many things. For his eerie scream. For his "Satan worship." For biting the head off a dove. And a bat.

Jun 29, 2010 | 11:45 pm | No Comments Posted

top news headlines

VA patients get letter of warning

Some may have been exposed to viruses from dental work performed at the St. Louis VA Medical Center.

Camp teaches kids to deal with life without limbs

WILDWOOD • The soft tap, tap, tap of prosthetic feet against pavement ...

- Steals & Deals: Wehrenberg lowers ticket prices during heat advisory
- Florissant police officer with career-ending injuries now getting a helping h...
- Boeing contract vote reflects the times
- Heads up
- Men set Lincoln County fireworks stand on fire
- Man set on fire described attackers
- Law and order
- Missouri Legislature concerned with family ties
- Corrections & clarifications
- Double shooting in St. Louis is investigated
- Local news digest

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Health | Longevity

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CHOLESTEROL
Q&A WITH DR. MANNY
FOX ON EDUCATION
INCREDIBLE HEALTH

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MEN'S HEALTH
WOMEN'S HEALTH
CHILDREN'S HEALTH
LONGEVITY

HEALTH CENTERS
ACID REFLUX / GERD
ALLERGY & ASTHMA
ALTERNATIVE MEDICINE
ARTHRITIS
BEAUTY & SKIN
CANCER

Genetics to Solve Why Ozzy Osbourne Is Still Alive

Monday, June 14, 2010

Print | ShareThis

The mystery of why rocker Ozzy Osbourne is still alive after decades of drug and alcohol abuse may finally be solved, reported The London Sunday Times.

The 61-year-old former Black Sabbath lead singer is to become one of only a few people in the world to have his full genome sequenced.

In addition to giving Osbourne information that could help prevent diseases, it is hoped the results will provide insights into the way drugs are absorbed into the body.

Reuters

Before he reformed, Osbourne survived decades of substance abuse

THE TIMES | THE SUNDAY TIMES | TIMES+

THE TIMES
THE SUNDAY TIMES

Archive Article Please enjoy this article from The Times & The Sunday Times archives. For

From The Sunday Times

June 13, 2010

Genetics to solve why Ozzy Osbourne is still alive

Document 21

Celebrity Genomes: Future Addition?

- Researchers at the U. of Copenhagen have the approval of Sitting Bull's descendents to sequence DNA from sample of his hair
- If successful, would become the first ancient, non-frozen, Native American genome sequenced

Document 21

CSHL Personal Genomes Meeting

Cold Spring Harbor Laboratory

Home | Meetings | Courses

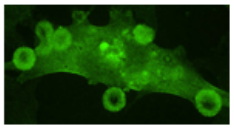
- Register
- Abstract Submission
- Abstract Status
- Media Registration
- General Information
- Travel Info
- Campus Info

PERSONAL GENOMES
September 10 - 12, 2010
Abstract Deadline: June 18, 2010

Organizers:
George Church, *Harvard University*
Paul Flicek, *European Bioinformatics Institute, UK*
Richard Gibbs, *Baylor College of Medicine, Houston*
Elaine Mardis, *Washington University School of Medicine*

We are pleased to host the third meeting on **Personal Genomes**, which will begin in the morning (9am) on Friday, September 10, 2010 and conclude in the late afternoon of Sunday September 12.

The first two conferences have spanned a significant milestone in human genetics - the appearance of individual, or personal, human genomes, and it is estimated that as many as 1,000 full human genomes will be completed by fall 2010. Ultra high throughput sequencing strategies have now been used to study more individual genomes – and yet few scientists, and even fewer physicians and clinical geneticists are familiar with the implications of this new data. This meeting will address the issues of individual genomes being part of research and routine clinical medicine within the next few years.



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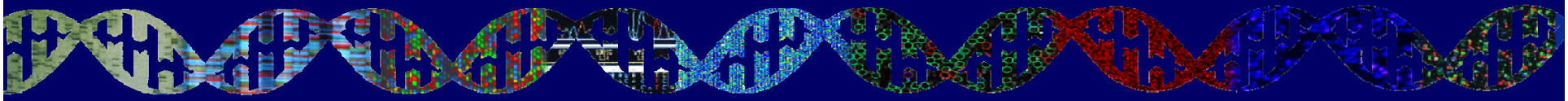


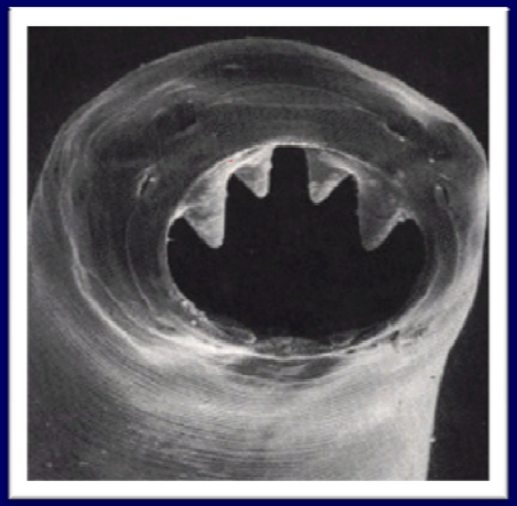
Large-Scale Sequencing Program: Organism Sequencing

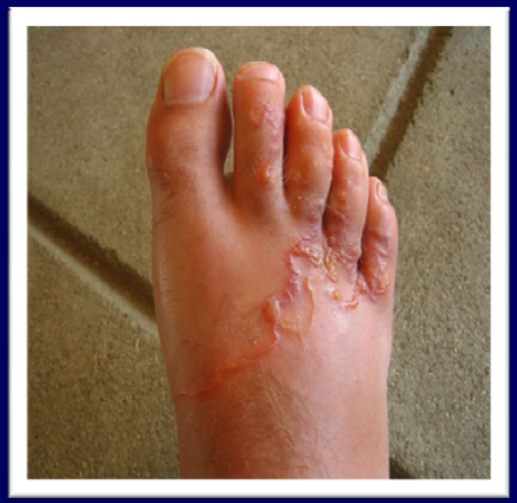
- Consortium published genome sequence of the Pea Aphid

- Washington University researchers published the genome sequence of the Western Clawed Frog

Document 22

Large-Scale Sequencing Program: Organism Sequencing

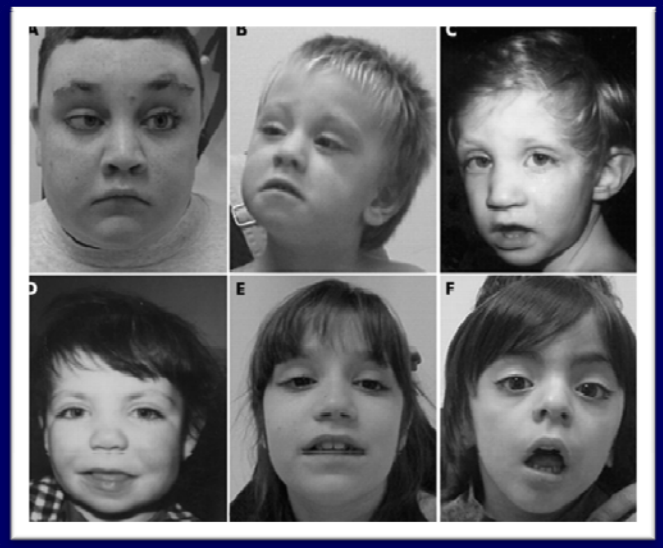




- Washington University researchers publish the transcriptome of the hookworm (*Ancylostoma caninum*)

Document 23

Large-Scale Sequencing Program: Medical Sequencing



- NIH-funded researchers identify *MLL2* mutations as a cause of Kabuki syndrome

Document 24

The Cancer Genome Atlas (TCGA)

- **Harold Varmus on board, affirms support for TCGA, and pursues close collaboration with NHGRI**
- **TCGA research network submits manuscript on ovarian cancer**
  - **489 ovarian tumors examined for genomic copy number and structural changes, and for gene expression**
  - **Complete exome sequencing of 315 cases reveals >20,000 somatic mutations**
- **Datasets made available for AML, colon, rectal, breast, and kidney cancers**

HapMap 3 Analysis Published

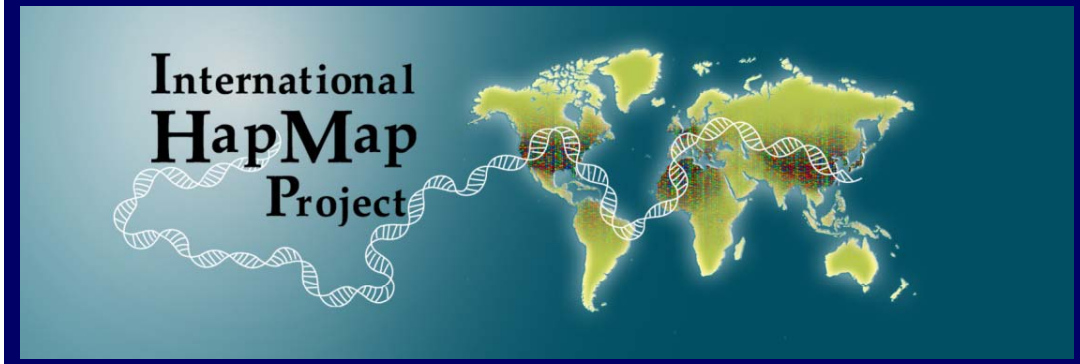
doi:10.1038/nature09298 nature

ARTICLES

Integrating common and rare genetic variation in diverse human populations

The International HapMap 3 Consortium*

Despite great progress in identifying genetic variants that influence human disease, most inherited risk remains unexplained. A more complete understanding requires genome-wide studies that fully examine less common alleles in populations with a wide range of ancestry. To inform the design and interpretation of such studies, we genotyped 1.6 million common single nucleotide polymorphisms (SNPs) in 1,184 reference individuals from 11 global populations, and sequenced ten 100-kilobase regions in 692 of these individuals. This integrated data set of common and rare alleles, called 'HapMap 3', includes both SNPs and copy number polymorphisms (CNPs). We characterized population-specific differences among low-frequency variants, measured the improvement in imputation accuracy afforded by the larger reference panel, especially in imputing SNPs with a minor allele frequency of ≤5%, and demonstrated the feasibility of imputing newly discovered CNPs and SNPs. This expanded public resource of genome variants in global populations supports deeper interrogation of genomic variation and its role in human disease, and serves as a step towards a high-resolution map of the landscape of human genetic variation.



- 3rd generation map of human genetic variation
- Added data from 7 populations

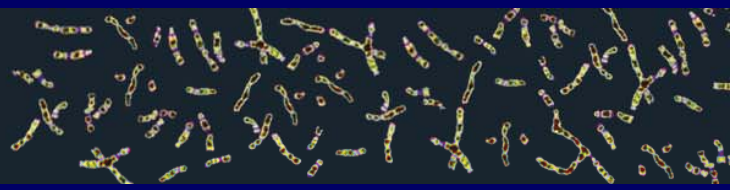
Document 25

1000 Genomes

- 1000 Genomes project releases data from pilot projects
- Pilot paper will be published in early November
- FTP site has more than 10 Gb of sequence data for 624 samples
- Variants from the first 1100 samples to be released by November

1000 Genomes

A Deep Catalog of Human Genetic Variation



Large-Scale Population Sequencing

wellcome trust
Gibbs Building, 215 Euston Road, London NW1 2BE, UK
T:+44 (0)20 7611 8888

Home > News and features > Media office > Press releases > 2010 > Wellcome Trust launches study of 10 000 human genomes in UK

Ten years on, Wellcome Trust launches study of 10 000 human genomes in UK

25 June 2010

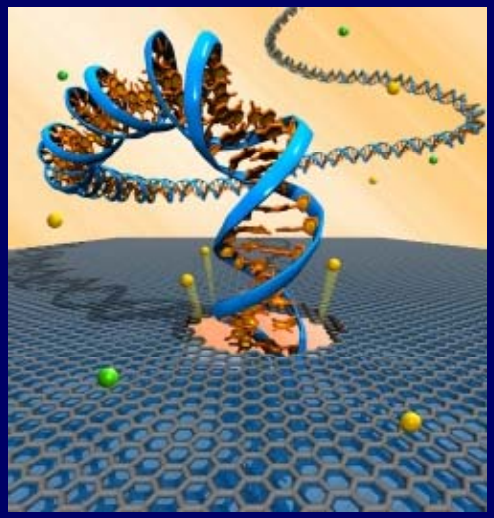
On the tenth anniversary of the completion of the first draft of the human genome - a draft that took ten years to complete - the Wellcome Trust has launched a project to decode the genomes of 10 000 people over the next three years. This will be one of the largest genome-sequencing programmes ever undertaken and will analyse the genomes of one in 6000 people in the UK.

- **Wellcome Trust and Sanger Institute announce the UK 10K project**
- **In 3 years, plan to sequence:**
  - **Complete genomes of 4000 well-phenotyped people**
  - **Exomes of 6000 people with particular disorders**

Document 27

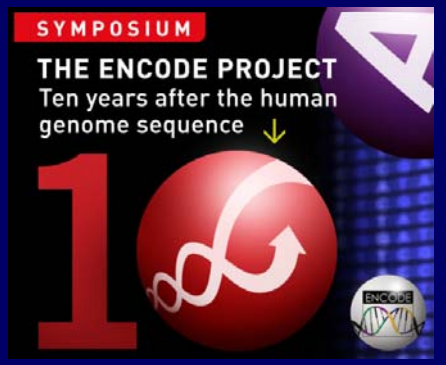
DNA Sequencing Technology

- 10 new or continuing awards for $1,000 genome sequencing technology development grants
- 50 papers published this year
- New RFAs published (Deadlines: 10/2010, 10/2011, 10/2012)



Document 28

ENCODE and modENCODE



- ENCODE Analysis Workshop in Barcelona (July 2010) focused on integrative analysis paper
- ENCODE will soon submit an ‘Users Guide’ paper for ENCODE data
- modENCODE close to submitting fly & worm integrated analysis papers
- Mouse ENCODE project (funded through ARRA) has begun submitting data to the ENCODE DCC at UCSC

Centers of Excellence in Genome Science (CEGS)

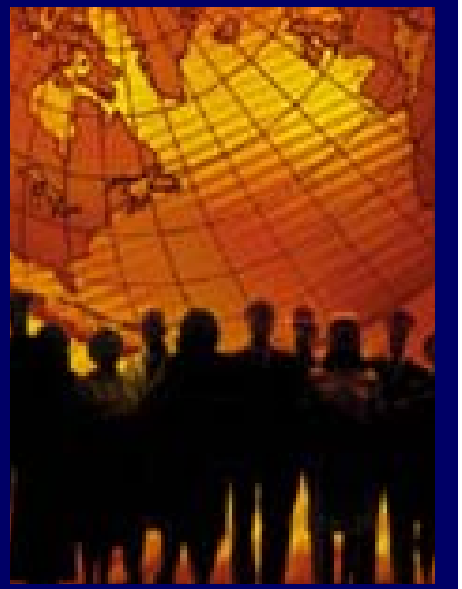
- One new award in 2010: George Church
- Revised Program Announcement last year
- Applications to be considered at February Council

CEGS Annual Meeting
HudsonAlpha
2009

ELSI Program

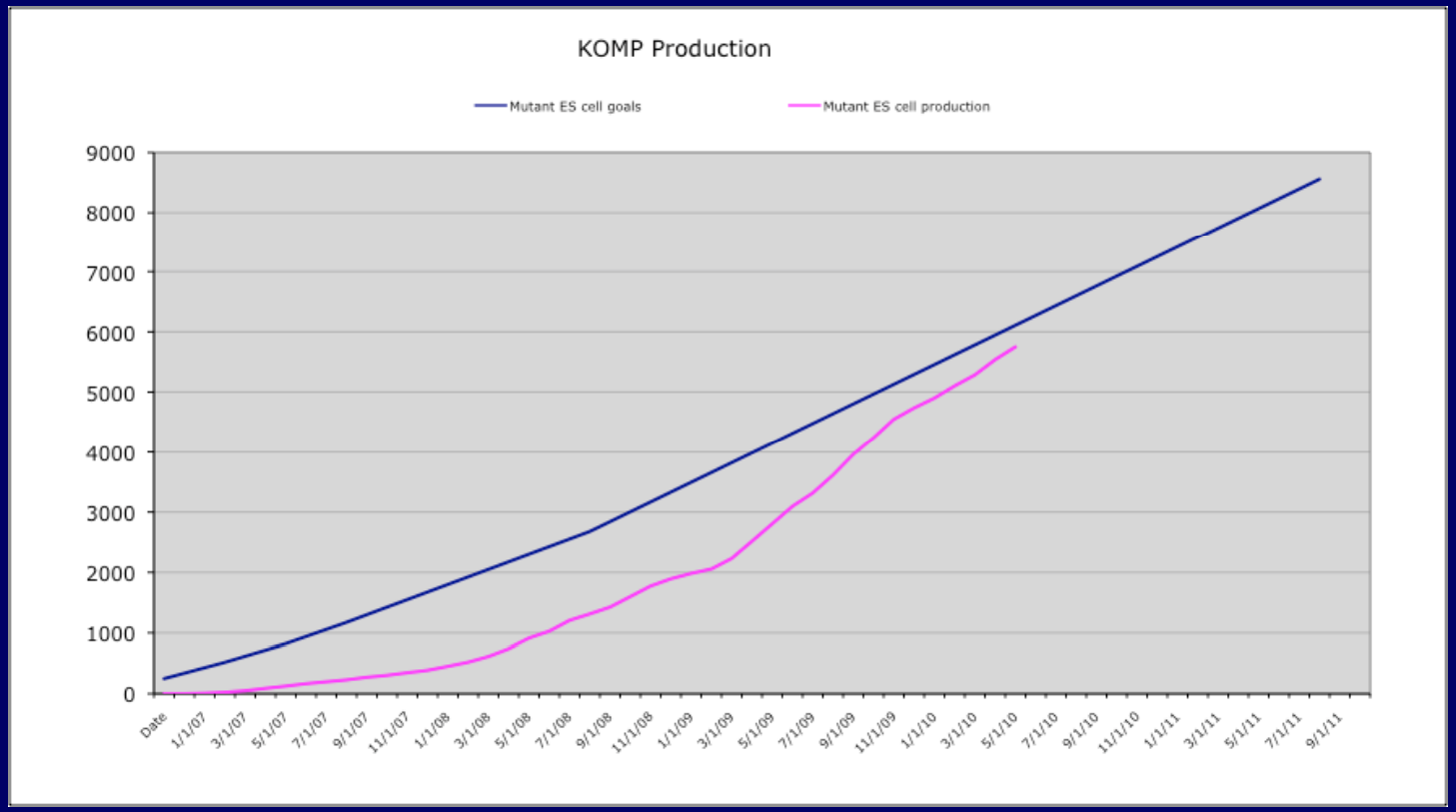
- NHGRI received $1.8M FY 2010 OD Bioethics Common Funds to support three ELSI research grants and an administrative supplement
- Presentation: Jean McEwen
- The Triennial ELSI Research Congress:

  "Exploring the ELSI Universe"
  April 12-14, 2011
  Chapel Hill, NC



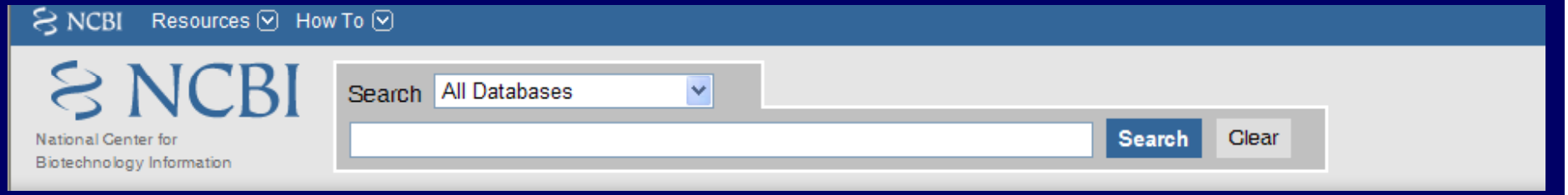
Knockout Mouse Program (KOMP)

- RFAs published for Knockout Mouse Phenotyping Project (KOMP^2)
- KOMP on track to achieve goals at end of project (Fall, 2011)



Informatics & Computational Biology

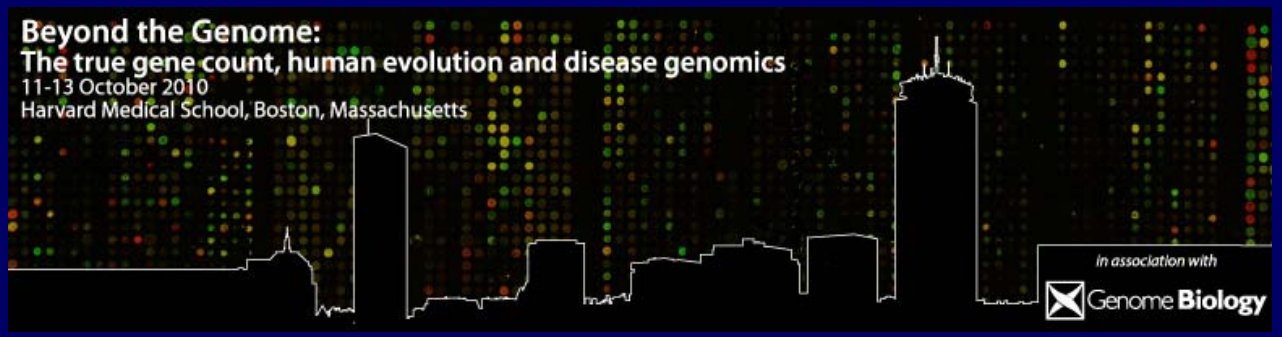
- **Quarterly meetings with NCBI**
  - **Improved communication and coordination**
  - **Tracking NHGRI projects at NCBI**
  - **Discuss project progress and resolve issues**
- **Vivien Bonazzi is official 'liaison' between NHGRI & NCBI (with help from Kris Wetterstrand)**



Informatics & Computational Biology

Beyond the Genome

(Harvard Medical School, October 2010)



Petascale Computing and Personalized Medicine

(U. of Illinois, October 2010)

2010 Workshop on Petascale Computing and Personalized Medicine

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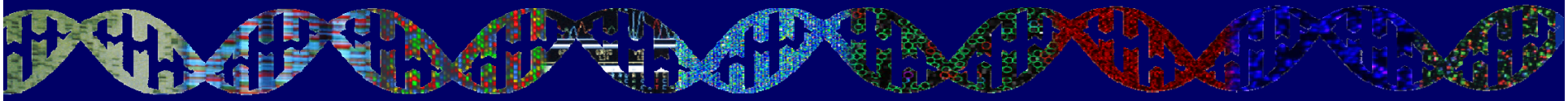

Human Microbiome Project

- Publication of first collection of human microbial genome sequences
- HMP Research Network Meeting
  - August 31-September 2, 2010
  - 450 attendees
- New awards in three areas:
  - 8 demonstration project (UH3s)
  - 5 technology development awards
  - 6 computational tools awards
- Presentation: Jane Peterson

Document 30

Genotype-Tissue Expression (GTEx)

- 2.5-year pilot with goal of collecting multiple tissues (n>30) from 160 deceased donors for eQTL analyses
- Broad Institute awarded contract for Laboratory, Data Analysis, and Coordinating Center contract
- University of Miami will serve as the Brain Bank (whole brains sent there)
- 3-4 Biospecimen Source Sites (awards soon)
- Inaugural GTEx kick-off meeting at end of September
- External Scientific Panel formed

Library of Integrated Network-based Cellular Signatures (LINCS)

- **Phase 1 Initiatives:**
  - RFA-RM-10-003: Large Scale Production of Perturbation-Induced Cellular Signatures (U54)
  - RFA-RM-10-004: New Laboratory-based Technology Development (receipt date: 02/22/2011)
  - RFA-RM-10-005: Computational Tool Development and Integrative Data Analyses (receipt date: 02/22/2011)

- **U54 Awards:**
  - Todd Golub (Broad) & Tim Mitchison (Harvard)
  - First meeting of grantees: Sept 27-28, 2010
  - Meeting goal is to map routes to collaboration & data standardization

Protein Capture Reagents

- **Ultimate Goal: a renewable community resource of high-quality affinity reagents for all human proteins**
- **Enable a wide range of applications: Immunoprecipitation (ChIP, protein-protein interactions); immunostaining; arrays, etc.**
- **Initially prioritize monoclonals; start with human transcription factors and imunoprecipitation applications**
- **Encourage development of 'near term' better alternatives**
- **Challenges: technical capabilities/range of applications; scalability (cost, throughput, quality); intellectual property**

Document 34

Human Heredity and Health in Africa (H3Africa)

- June 22, 2010: NIH and Wellcome Trust announce partnership to support population-based genome studies in Africa

- NIH pledged $25 million in Common Fund money over five years, with Wellcome Trust pledging an additional $12 million

Document 32

Human Heredity and Health in Africa (H3Africa)

- Meeting in Oxford (August 2010) to discuss scientific vision and goals for the project

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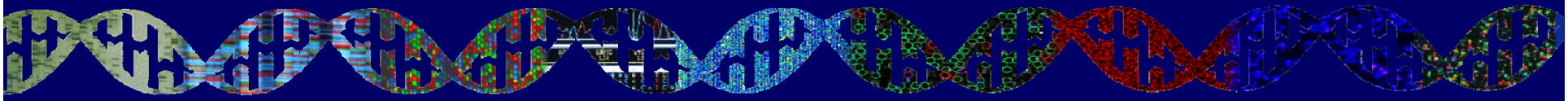
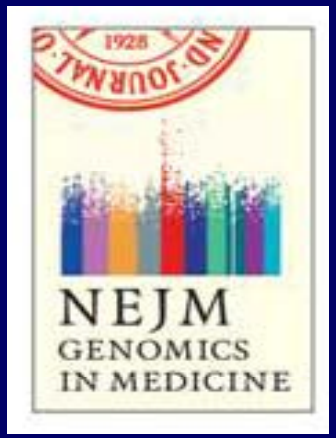


NEJM Genomic Medicine Series



The NEW ENGLAND JOURNAL of MEDICINE

REVIEW ARTICLE

The NEW ENGLAND JOURNAL of MEDICINE

REVIEW ARTICLE

The NEW ENGLAND JOURNAL of MEDICINE

REVIEW ARTICLE

GENOMIC MEDICINE

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

New Therapeutic Approaches to Mendelian Disorders

Harry C. Dietz, M.D.

PROGRESS IN THE TREATMENT OF GENETIC DISORDERS HAS DERIVED FROM insight into their causes and has focused on nutritional limitation of a substrate, the purging of a toxic metabolite, or compensatory expression of a protein whose deficiency causes disease, through gene delivery. In this review, I focus on therapeutic strategies that exploit a precise understanding of the pathogenesis of a mendelian disease, giving examples that illustrate the strengths and limitations of each approach, as well as the potential for broadening its application to the treatment of more common disorders.

Document 35

NHGRI Scientists Present Genomic Advances to Visiting Judges

June 2010: 60 judges participated in an NIH continuing education program "*Genomics, Medicine, and Discrimination*"

Document 36

Summer Workshop in Genomics August 1-6, 2010

- 35 Participants from 18 colleges/universities

- New collaboration with NIH Office of Intramural Training and Education around faculty mentoring

Document 37

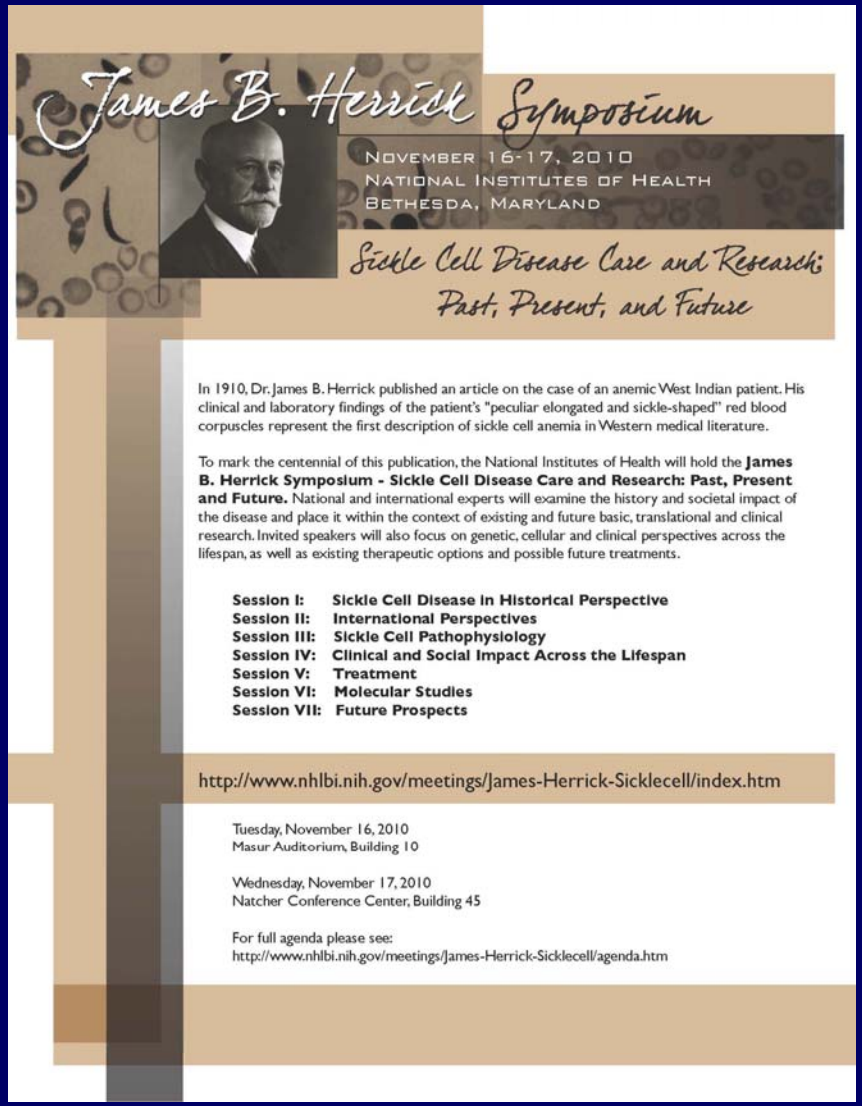
Carrier Screening Comes to College Sports

- In April 2010, NCAA approved mandatory testing for sickle cell carrier status
- NHGRI researchers Larry Brody and Vence Bonham joined George Dover (Johns Hopkins) to publish a Perspective in NEJM

Document 38

Sickle Cell Symposium

- November 16-17, 2010
- NIH Bethesda Campus
- Sponsored by NHGRI and 7 other NIH Institutes/Centers



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 38

Newborn Screening in the Genomics Era Workshop

- NICHD-NHGRI Joint Workshop
- December 13-14, 2010
- Goal is to frame a research agenda for stimulating interface between newborn screening and new genomic technologies

Electronic Health Records and Genomic Information Workshop

- Two-day event in the late Spring 2011
- Will explore issues surrounding integration of genomic information into clinical electronic health/medical records
- Stakeholders to include industry, academia, advocacy community, state and federal government

USA Science & Engineering Festival

Deepwater Horizon Disaster

Deepwater Horizon and NIH

- NIH/NIEHS commit $28M for 5-year study of clean-up workers (to follow for 10-20 years)
- Focus on clean-up workers because of likely heavy exposure
  - Few long-term studies of oil spill workers
  - Suggestions of genotoxic effects
- Plans to study other vulnerable populations, especially children and pregnant women
- Teri Manolio (NHGRI) and Harold Jaffe (CDC) leading DHHS efforts

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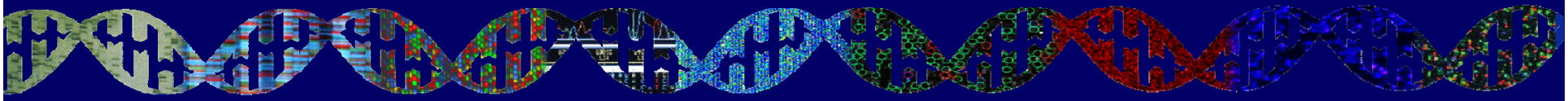
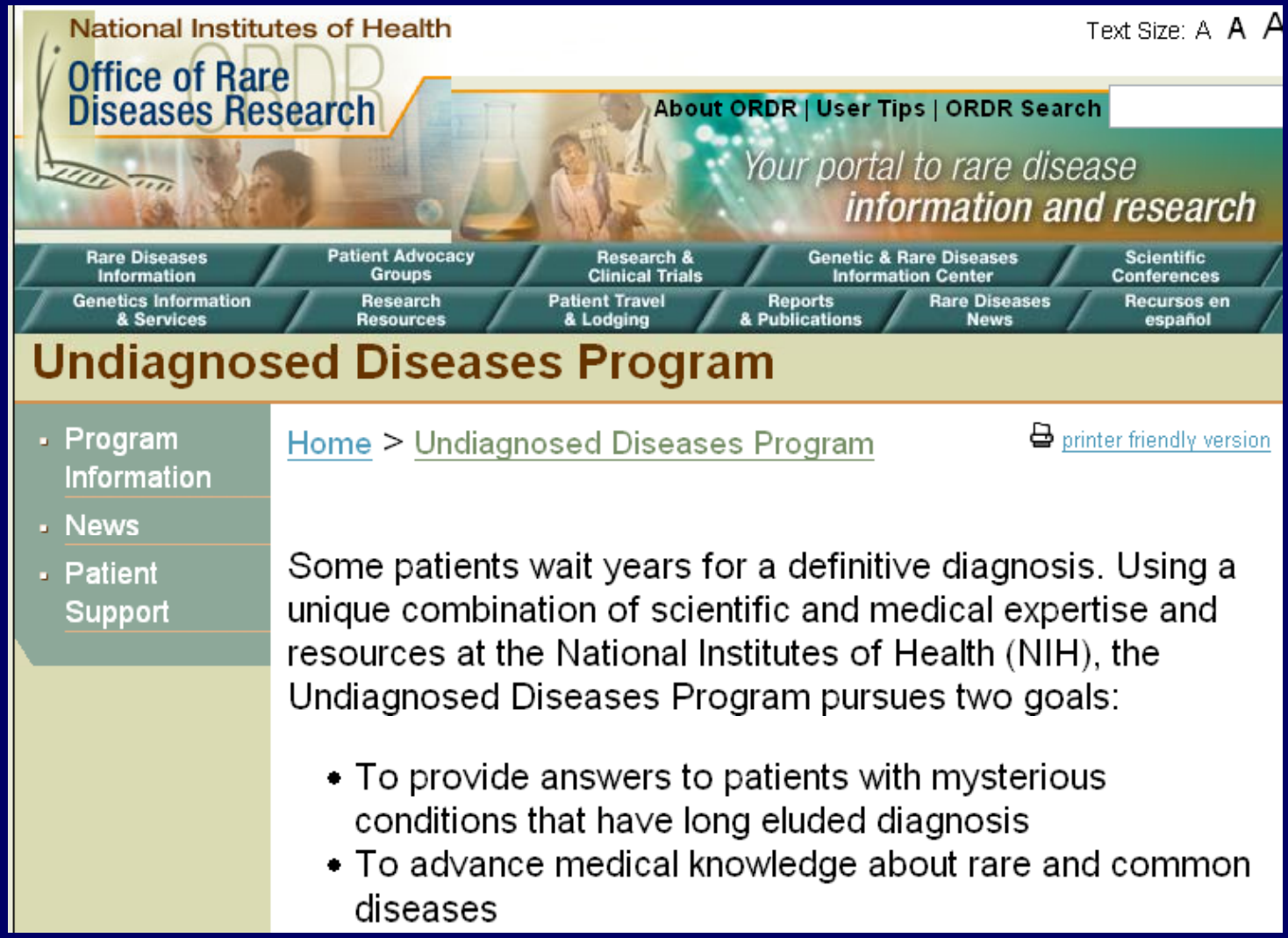
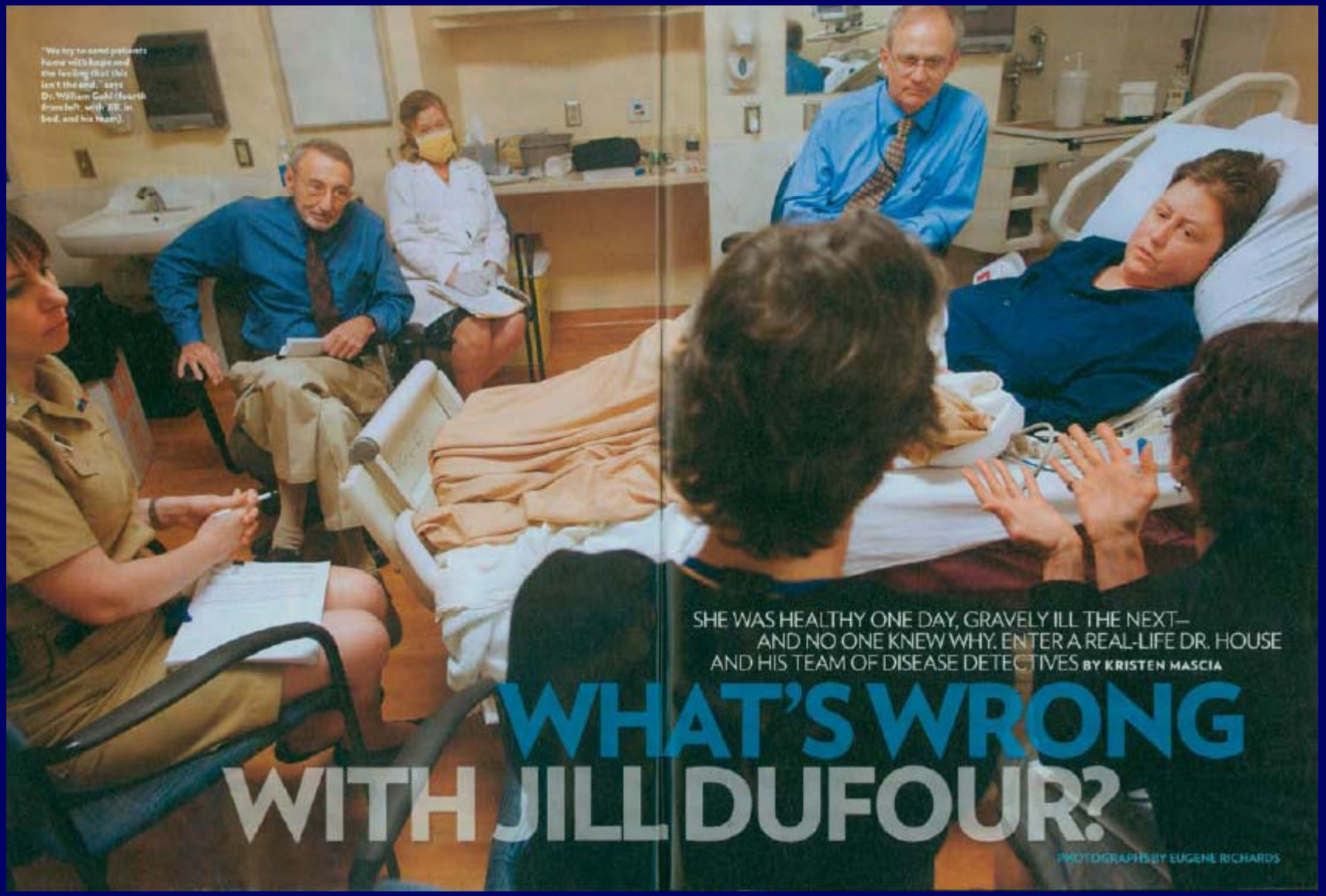


Undiagnosed Diseases Program (UDP)



rarediseases.info.nih.gov

Routine Media Coverage of UDP



***People* (June 2010)**

NHGRI Intramural Research Highlights

Colleen McBride
(Multiplex Project)

Yingzi Yang
(Cell Polarity)

Elaine Ostrander
(Dog Genetics)

Document 41

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

Special Thanks!


NHGRI