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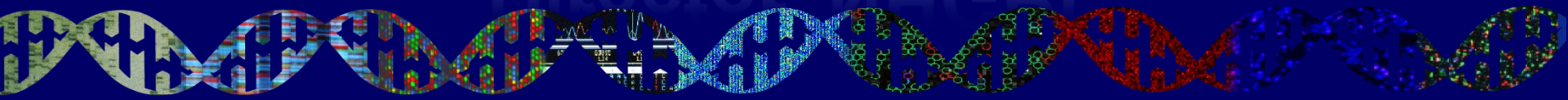
National Human Genome Research Institute
National Institutes of Health

DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

September 2011

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



Remembering 9-11-01

genome.gov/DirectorsReport

Document #

I. General NHGRI Updates

II. General NIH Updates

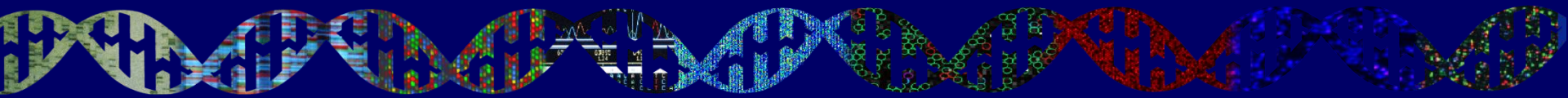
III. Genomics Updates

IV. NHGRI Extramural Program

V. NIH Common Fund Programs

VI. NHGRI Office of the Director

VII. NHGRI Intramural Program



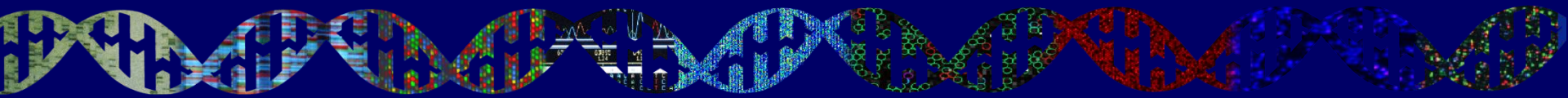
Open Session Presentations

NHGRI Office of Policy, Communications, and Education

- **Laura Lyman Rodriguez**

Chicago Genomic Medicine Meeting

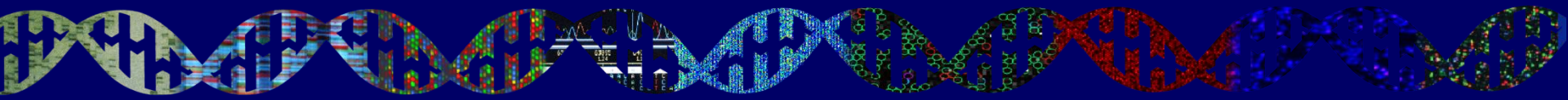
- **Teri Manolio & Geoff Ginsburg**



Open Session Presentations

Program Updates:

- H3Africa
  - Jane Peterson
- GTEx
  - Jeff Struewing
- Large-Scale Genome Sequencing Program
  - Adam Felsenfeld



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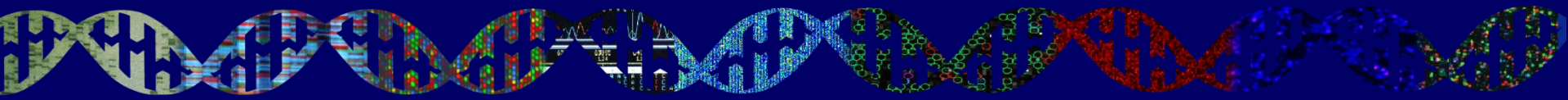
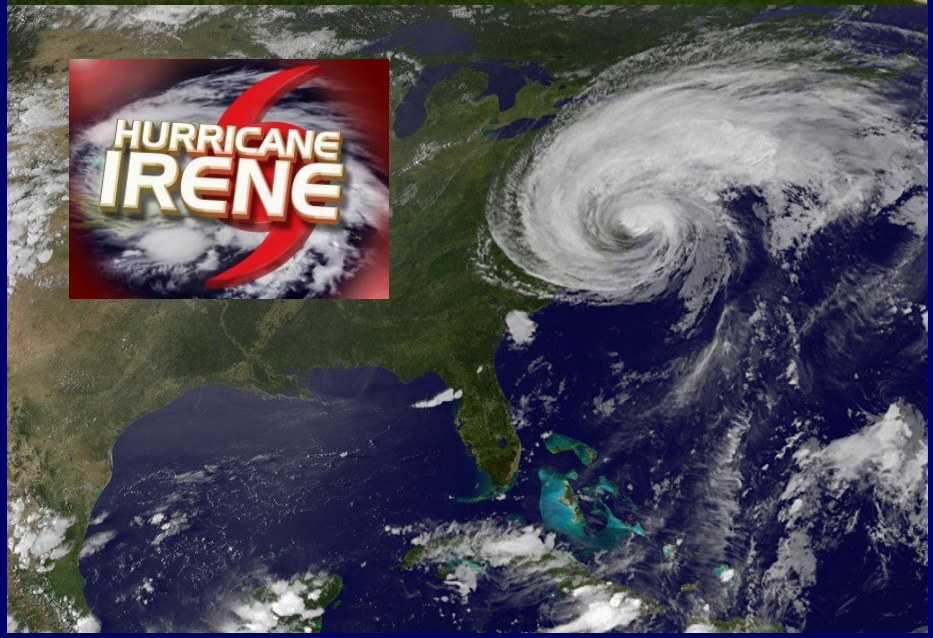


More Episodes of Nature's Wrath



New NHGRI Deputy Director

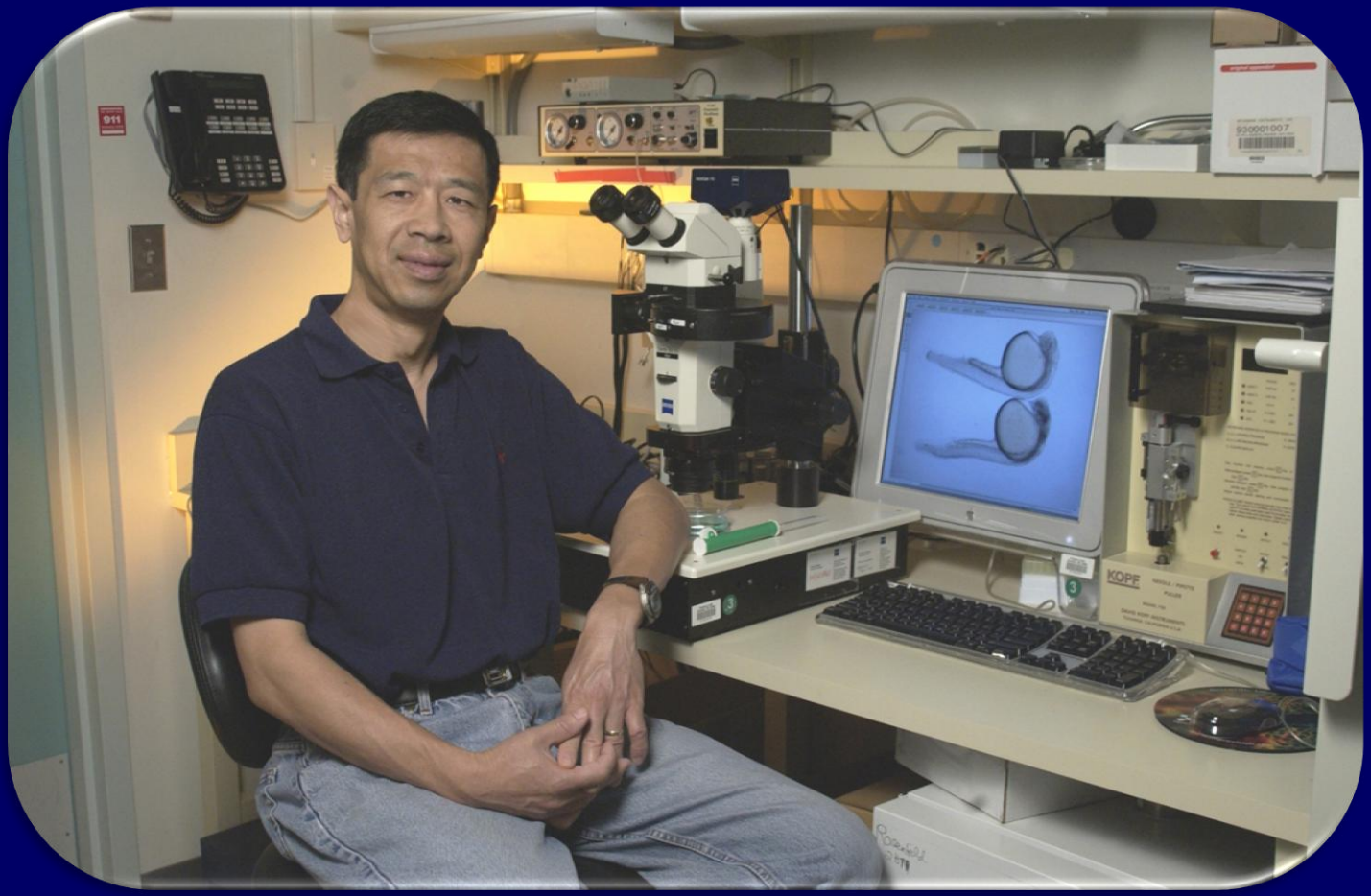
Mark Guyer, Ph.D.

Document 1

New NHGRI Policy and Program Analysis Branch Chief

Derek Scholes, Ph.D.

New NHGRI Deputy Scientific Director



P. Paul Liu, M.D., Ph.D.

Document 2

New NIH Intramural Sequencing Center (NISC) Director

Jim Mullikin, Ph.D.

Elected Fellow by the American Psychological Association

Vivian Ota Wang, Ph.D.

Special Advisors to NHGRI Director

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

Marc Williams, M.D.

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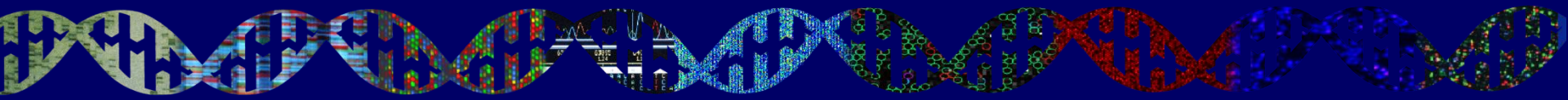
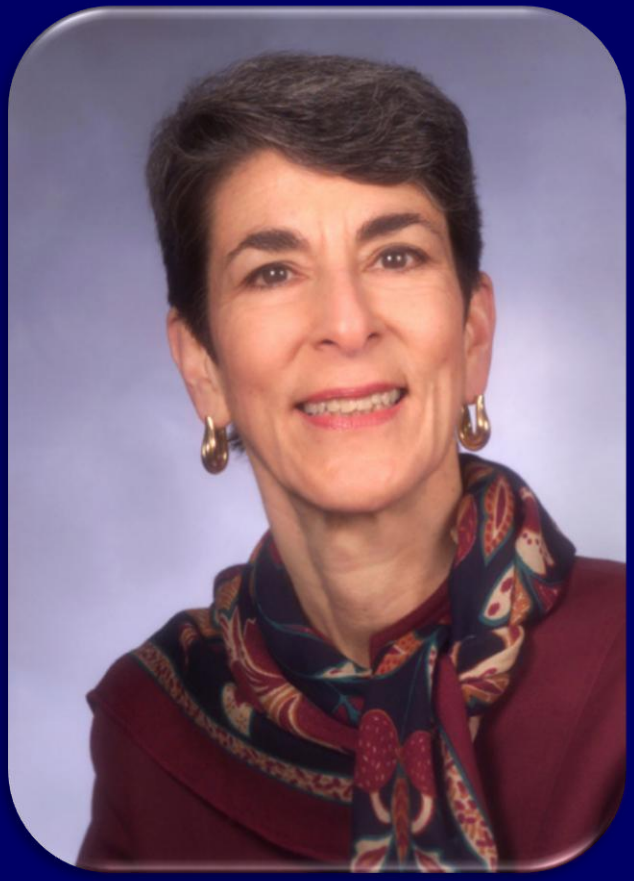


New Director of the National Institute of Dental and Craniofacial Research

Martha Somerman, D.D.S., Ph.D.

Document 3

Acting Director of the National Institute of General Medical Sciences



Judith Greenberg, Ph.D.

Departing Director of the National Center for Research Resources (NCRR)

Barbara Alving, M.D.

Document 5

Incoming Acting Director of the National Center for Research Resources (NCRR)

Louise Ramm, Ph.D.

Document 5

Departing Director of the NIH Center for Scientific Review (CSR)

Antonio Scarpa, M.D., Ph.D.

Document 6

Incoming Acting Director of the NIH Center for Scientific Review (CSR)

Richard Nakamura, Ph.D.

New Director of the NIH Center for Information Technology (CIT)

Andrea Norris, M.B.A.

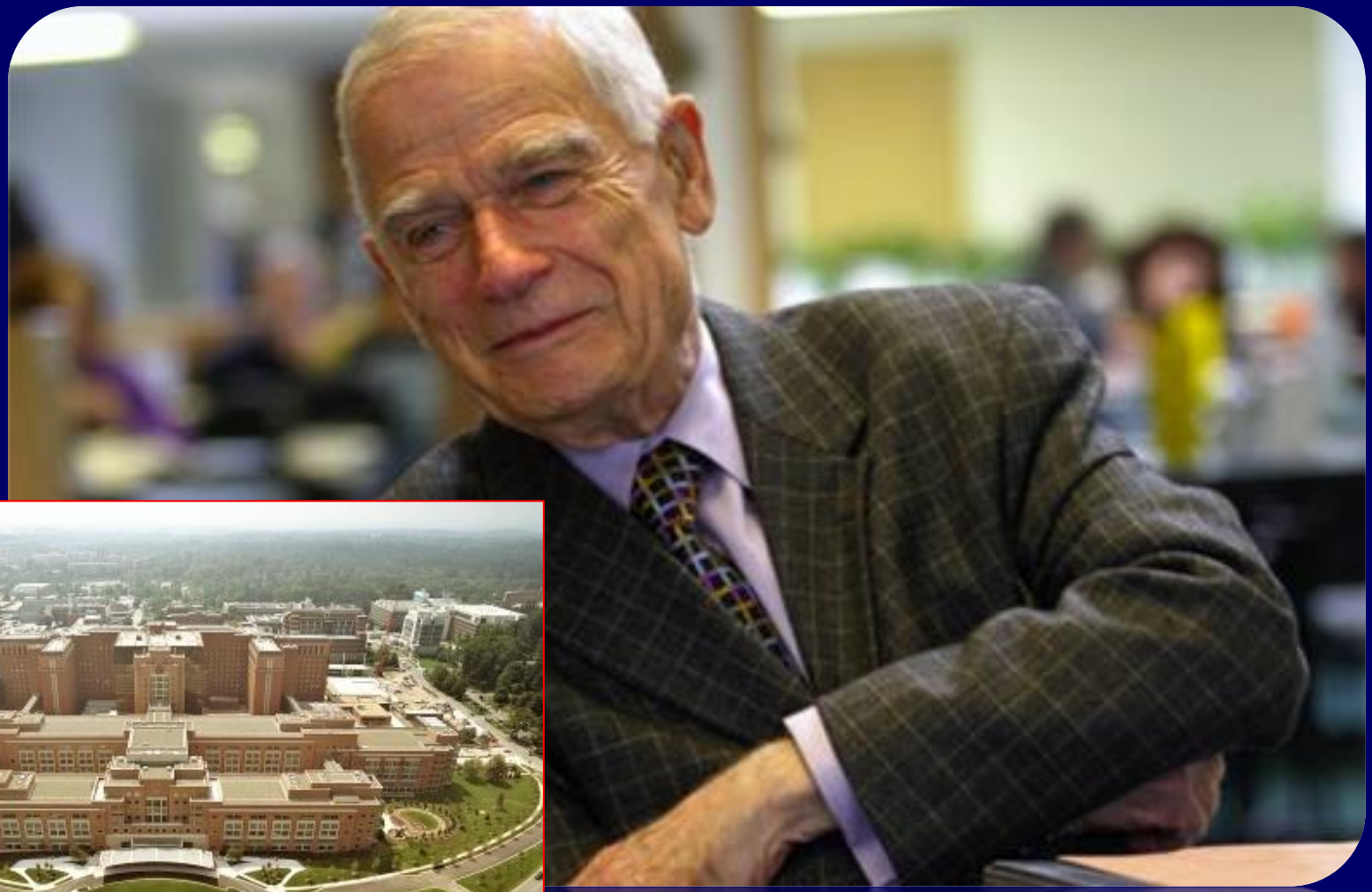
Interim Director for NCI Center for Cancer Genomics

Barbara Wold, Ph.D.

Mourning the Loss of Bernadine Healey

Document 7

Mourning the Loss of Senator Mark Hatfield



Document 8

Science Articles on Race & Ethnicity Influence on NIH Awards

Race, Ethnicity, and NIH Research Awards

Donna K. Ginther,[1]* Walter T. Schaffer,[2] Joshua Schnell,[3] Beth Masimore,[3] Faye Liu,[3] Laurel L. Haak,[3] Raynard Kington[2]†

We investigated the association between a U.S. National Institutes of Health (NIH) R01 applicant's self-identified race or ethnicity and the probability of receiving an award by using data from the NIH IMPAC II grant database, the Thoms
Although proposals with strong priority score
we find that Asians are 4 percentage points
percentage points less likely to receive NIH
whites. After controlling for the applicant's
previous research awards, publication record
applicants remain 10 percentage points less
Our results suggest some leverage points fo

POLICYFORUM

SOCIOLOGY

Weaving a Richer Tapestry in Biomedical Science

Lawrence A. Tabak* and Francis S. Collins*

Document 9

Revised NIH Regulations on Financial Conflict of Interest (FCOI)

- **Major Changes to Previous Policy:**
  - **Significant Financial Interest (SFI) Definition**
  - **Investigator Disclosure**
  - **Reporting to NIH**
  - **Public Accessibility to Information**
  - **Investigator Training**

>>>Implementation by August 24, 2012

PI **Institution** **NIH**

NIH's Proposed National Center for Advancing Translational Sciences (NCATS)

To advance the discipline of translational science and catalyze the development and testing of novel diagnostics and therapeutics across a wide range of human diseases and conditions

Commentary from the NIH Director about the Proposed NCATS

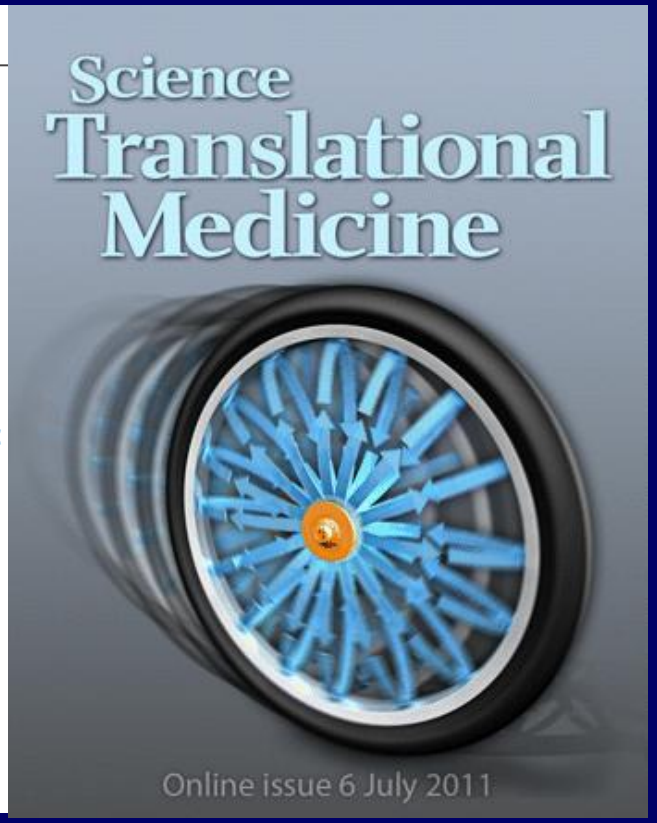
COMMENTARY

POLICY

Reengineering Translational Science: The Time Is Right

Francis S. Collins

Despite dramatic advances in the molecular pathogenesis of disease, translation of basic biomedical research into safe and effective clinical applications remains a slow, expensive, and failure-prone endeavor. To pursue opportunities for disruptive translational innovation, the U.S. National Institutes of Health (NIH) intends to establish a new entity, the National Center for Advancing Translational Sciences (NCATS). The mission of NCATS is to catalyze the generation of innovative methods and technologies that will enhance the development, testing, and implementation of diagnostics and therapeutics across a wide range of diseases and conditions. The new center's activities will complement, and not compete with, translational research being carried out at NIH and elsewhere in the public and private sectors.



Document 11

Fiscal Year 2012 Appropriations Update

FY2012 President's Request:

NIH: $32B (+2.4%)
NHGRI: $525 M (+1.7%)

- **House: Passed 6 of 12, 3 out of committee, and 3 (including Labor/HHS) without action**
- **Senate: Passed 1 of 12, 3 out of committee, and 8 without action**
- **Continuing Resolution likely**

Document 12

The Debt Deal … Waiting for the Cuts

- Caps established for appropriations over the next decade

- Super Committee must propose deficit reduction through ≥$1.2 trillion of more cuts by Nov. 23

- Substantial reductions possible for FY2013

Document 13

Senator Ben Cardin Visits NIH

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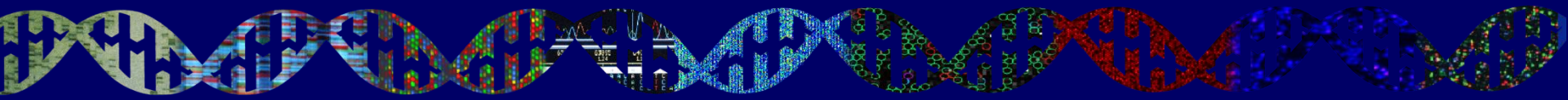


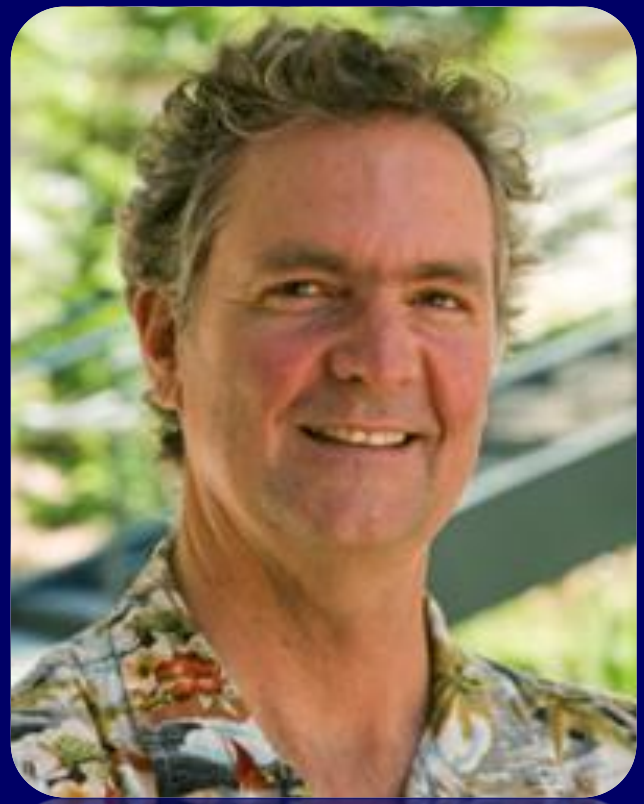
2011 Gruber Genetics Prize
The Peter and Patricia Gruber Foundation

GRUBER
FOUNDATION

Ron Davis, Ph.D.

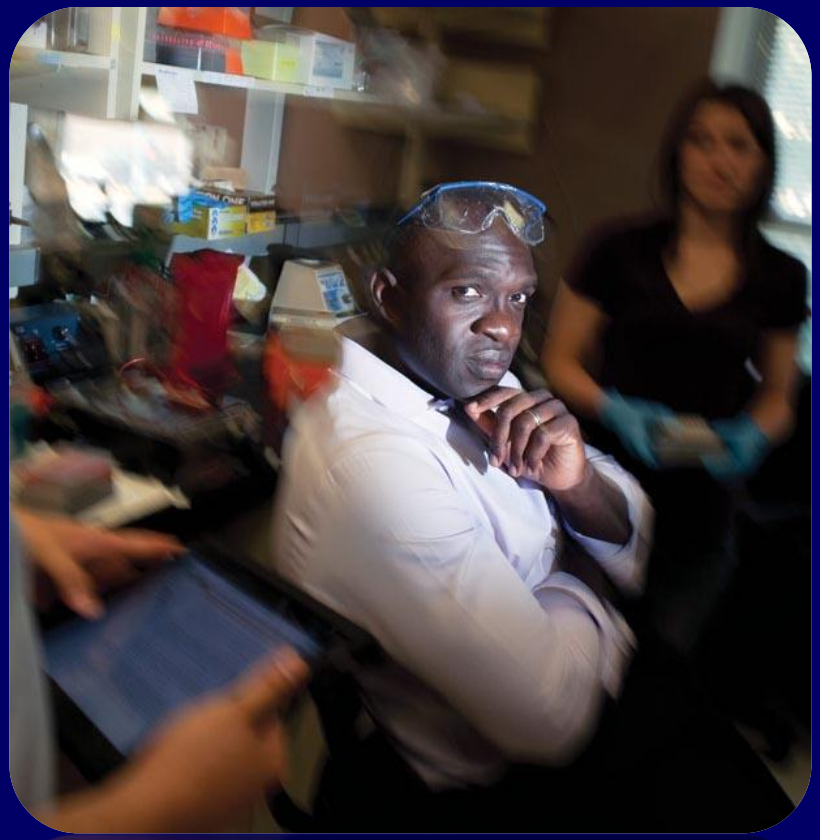
Document 14

2011 Weldon Memorial Prize
University of Oxford



David Haussler, Ph.D.

Technology Review's Annual List of 35 Innovators Under 35



tr35

Yemi Adesokan, Ph.D.

Document 16


The World
ARCTIC
ARCTIC OCEAN
EUROPE
ASIA
NORTH AMERICA
PACIFIC OCEAN
ATLANTIC OCEAN
CARIBBEAN
AFRICA
Equator
SOUTH AMERICA
OCEANIA
INDIAN OCEAN
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PACIFIC OCEAN
SOUTHERN OCEAN
ANTARCTICA
0 2,000 Miles
0 2,000 Kilometers
© MAPQUEST.


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AFGHANISTAN
PAKISTAN
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NEPAL
BHUTAN
BANGLADESH
MYANMAR
SRI LANKA
JAMMU & KASHMIR
HIMACHAL PRADESH
PUNJAB
UTTARANCHAL
HARYANA
DELHI
RAJASTHAN
UTTAR PRADESH
SIKKIM
ARUNACHAL PRADESH
ASSAM
NAGALAND
MEGHALAYA
MANIPUR
TRIPURA
MIZORAM
BIHAR
JHARKHAND
WEST BENGAL
GUJARAT
MADHYA PRADESH
CHHATTISGARH
ORISSA
MAHARASHTRA
ANDHRA PRADESH
KARNATAKA
GOA
TAMIL NADU
KERALA
I N D I A
BAY OF BENGAL
ARABIAN SEA
ANDAMAN SEA
INDIAN OCEAN
LEGEND
International Boundary
State Boundary
Country Capital
State/U.T. Capital
Tourist Places
International Airport
National Parks

Purpose of Trip

- **Invited to be Keynote Speaker at Founders Day Celebration of National Institute for Biomedical Genomics**

  New Institute outside Kolkata

  Advisory role in 2008

  Significant development for genomics in India

- **Used opportunity to tour other genomics research facilities in India**

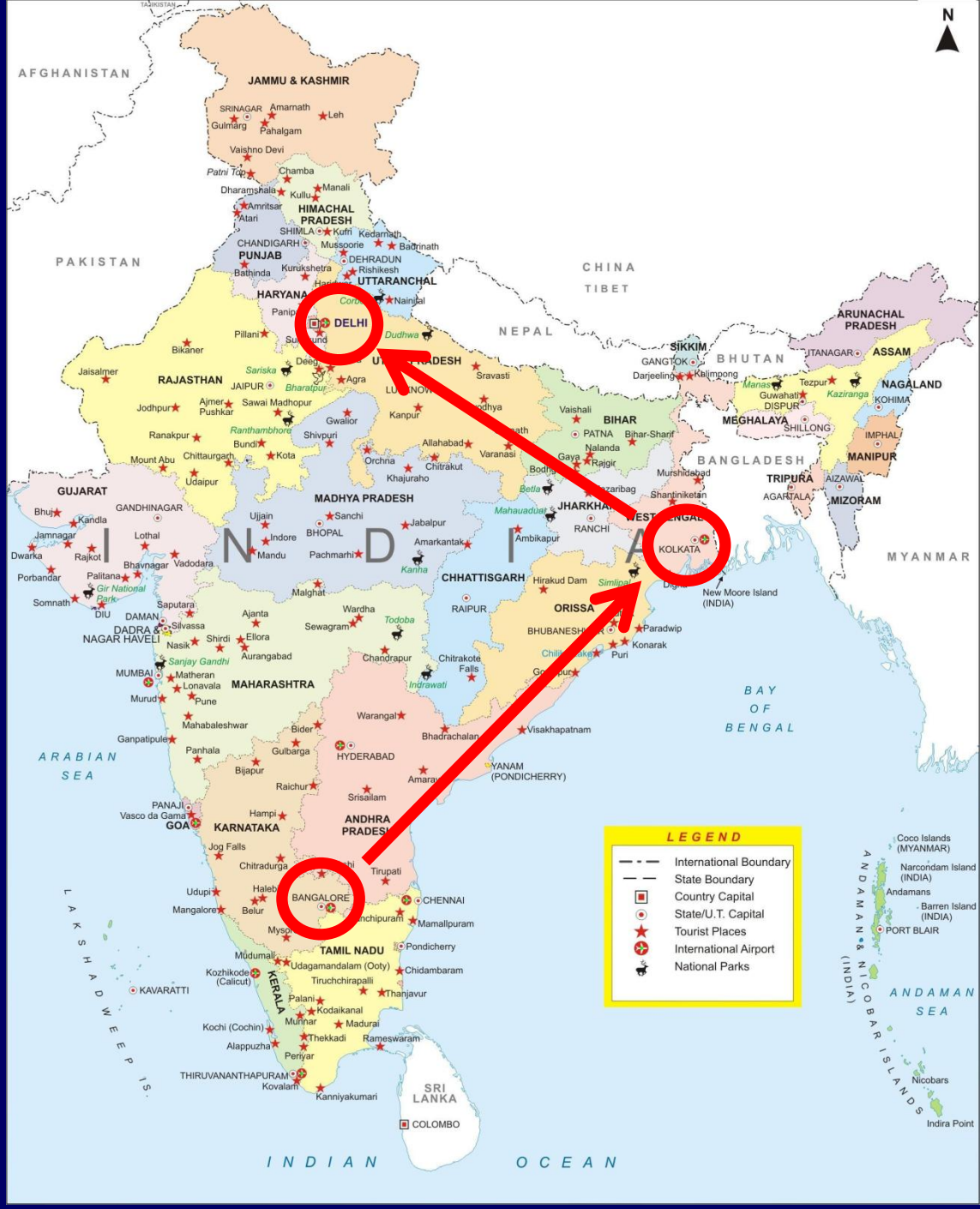
Bangalore

↓

Kolkata

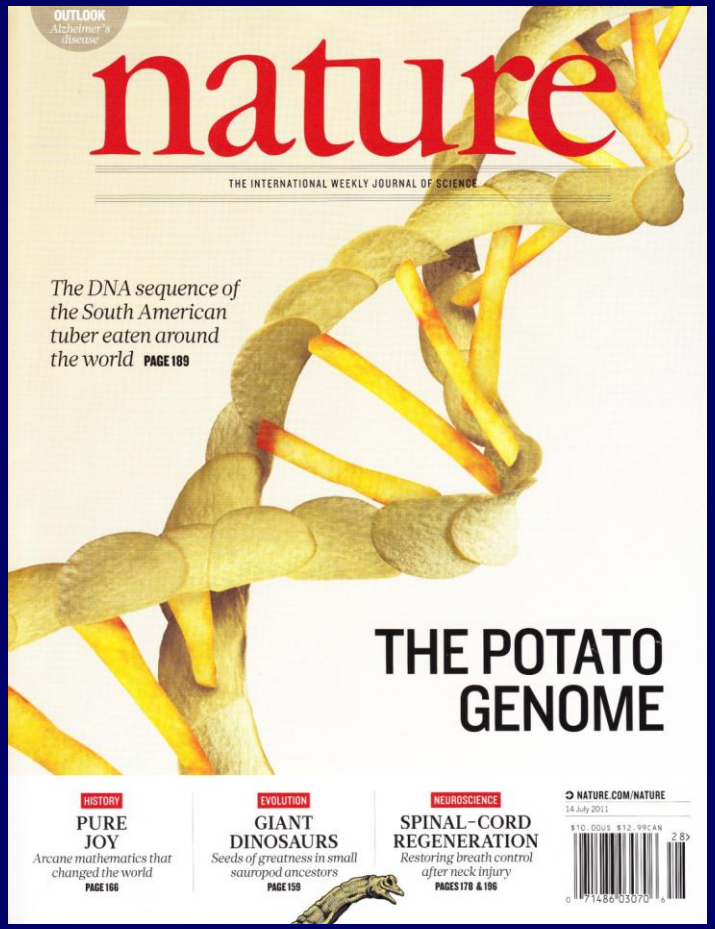
↓

Delhi




NBMG
NATIONAL INSTITUTE OF BIOMEDICAL GENOMICS

Genomics In The News…



Document 17

Genomics In The News…

Document 17

Genomics In The News…

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Genomics In The News…

CNN.com

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DNA tests provide the poop on bad dog owners

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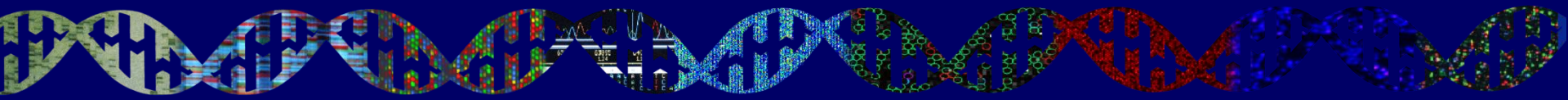
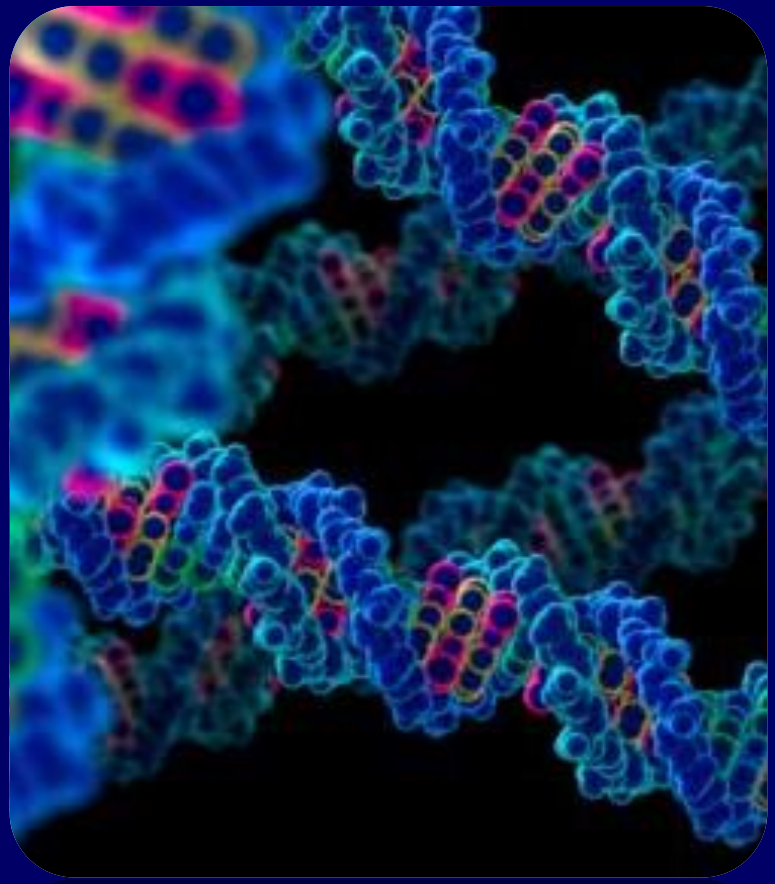


Large-Scale Genome Sequencing Program Renewal

1. Genome Sequencing & Analysis Centers
2. Mendelian Disorders Genome Centers
3. Clinical Sequencing Exploratory Research
4. Informatics Tools for High-Throughput Sequence Data Analysis



Document 18

Large-Scale Genome Sequencing Program: Comparative Genomics

- Characterization of SNP variation in Rhesus Macaques (*Macaca mulatta)*

Document 19

Large-Scale Genome Sequencing Program: Comparative Genomics

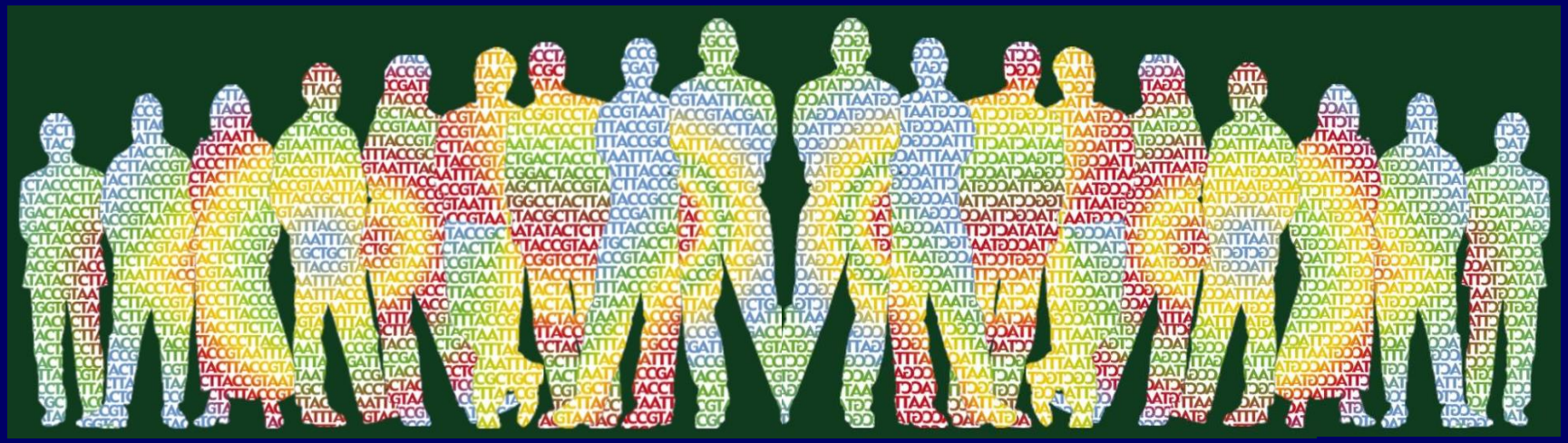
- Genome sequence of the first lizard, the North American green anole (*Anolis carolinensis*)

Document 19

Large-Scale Genome Sequencing Program: Medical Sequencing

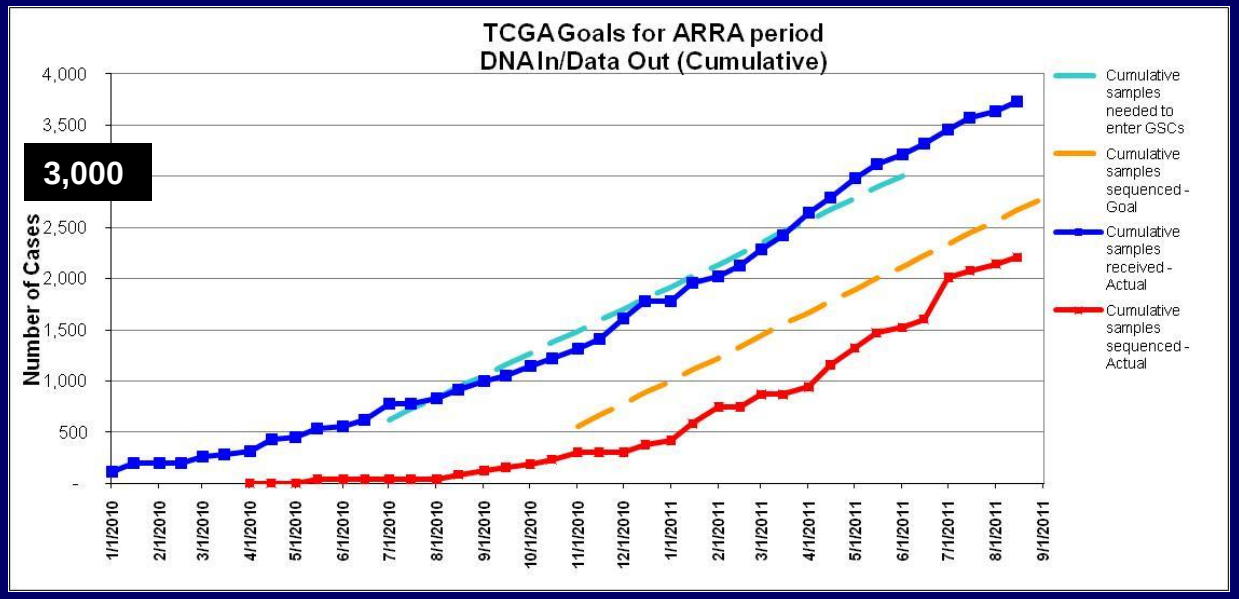
Major analyses underway for projects studying:

- **Diabetes/metabolic syndrome (n=13,000)**
- **Autism (n=2000)**
- **Lipid levels (n=1000)**
- **Tumor sequencing (n=1000 T/N pairs)**



The Cancer Genome Atlas

- **22 tumor projects in progress**
- **TCGA achieves goal of accruing and characterizing 3,000 tumor/ normal pairs in 2 years**



- Glioblastoma
- Ovarian
- Acute Myeloid Leukemia
- Colon/rectum carcinoma
- Breast
- Lung Squamous Cell
- Lung Adenocarcinoma
- Renal Clear Cell Carcin.
- Renal papillary
- Uterine (endometrial)
- Low grade glioma
- Gastric Carcinoma
- Prostate
- Bladder
- Cervical
- Head and neck
- Liver
- Melanoma
- Sarcoma
- Thyroid
- Lymphoma
- Pancreas

ARTICLE

Nature 474: 609–615, 2011

doi:10.1038/nature10166

Integrated genomic analyses of ovarian carcinoma

- **Most extensive and comprehensive cancer genome study to date**
- **#5 most highly ranked article in molecular biology by *The Scientist***
- **Several upcoming manuscripts-- colorectal carcinoma, acute myeloid leukemia, and breast carcinoma**

DOUBLE ISSUE JUNE 13, 2011

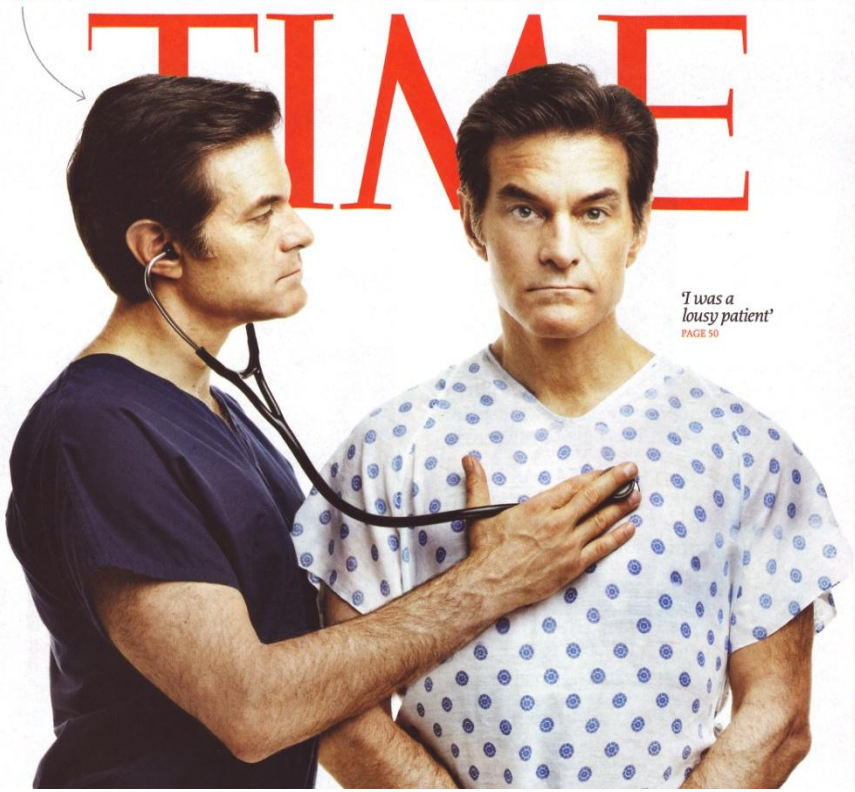
Health Special Report

THE NEXT WAVE OF CANCER TREATMENT

Dr. Oz: Lessons from my cancer scare + Is your cell phone safe?

PLUS
Fareed Zakaria: The future of innovation
Joe Klein: How to misread a mandate
Airlines: The season of sky-high add-ons

TIME



'I was a lousy patient'
PAGE 50

Cracking Cancer's Code

Tumor DNA holds the key to beating the disease

BY ALICE PARK

identifying chronic myeloid leukemia. The problem with analyzing a tumor's DNA, however, has always been one of resolution. If the physical structure of a chromosome looks different in tumor cells, then there certainly must be changes in the genes packed
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Those things can range from an overactive gene to a complete swapping of DNA from one chromosome to another. "The cardinal feature of a cancer cell is that it's lost the identity it was born with," says Dr. Ernest Hawk, head of the division of cancer prevention and population sciences at MD Anderson Cancer Center in Houston. "It simply doesn't live a normal life and then die as normal cells do."

Researchers already had some limited experience in isolating and examining the workings of cancer genes. The specific mutations linked to breast cancer in the BRCA 1 and BRCA 2 genes as well as alterations in a gene called APC, which normally suppresses tumor growth and is linked to colon cancer, are behind anywhere from 5% to a third of these diseases. But these are inherited aberrations, and cancers are triggered not only by the genes we get from our parents but also by corruptions to our genome that we acquire in our daily lives—from smoking, sun and diet as well as simple aging. "What has happened in cancer care over the past 20 years has been very piecemeal and ad hoc," says Dr. Todd Golub, director of the cancer program at the Broad Institute. "We discovered some cancer-causing genes here and there, often by stumbling across them. But the notion of being able to say that we are going to systematically and comprehensively inter-

'We expect that 10 years from now, each cancer patient is going to want to get a genomic analysis of their cancer.'
—BRAD OZENBERGER, DIRECTOR OF THE CANCER GENOME ATLAS

of other existing drugs to treat new cancers. The multiple myeloma genome also highlighted several genes that no scientist had ever even described before in the literature, which could become targets for entirely new classes of drugs.

Ellis and his team have sketched out a similar map of one type of breast cancer—tumors that are positive for receptors of the hormone estrogen. Like Golub and researchers at TCGA, they are beginning to see patterns in the genetic triggers of cancer. They suspect, for example, that cancer is not a disease of blockbuster mutations, in which a majority of patients with a particular cancer share aberrations in one or even a few genes. Instead, it's likely that each type of cancer may have a few "driver" mutations and a host of "passenger" changes that appear at a very low frequency. The good news is

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Cancer experts aren't naive enough to believe that sequencing a tumor just once will reveal all they need to know. Cancer is constantly changing its offensive and defensive plans in response to whatever treatments doctors are using against it. The idea is to rebiopsy patients periodically and allow the dynamic genetic changes in the tumors to educate doctors about how aggressive the cancer is, whether it has developed resistance to drugs and even whether it has spread. "The concept is to let the tumor teach us how to treat patients," says Dr. Waun Ki Hong, head of cancer medicine at MD Anderson.

It's all part of the leap toward personalized cancer care, the therapeutic beacon toward which researchers and doctors have been navigating for a long time. "We fully expect that 10 years from now, each cancer patient is going to want to get a genomic analysis of their cancer and will expect customized therapy based on that information," says Brad Ozenberger, TCGA's program director. Only with more individualized therapies that match the right treatment with the right patient at the right time will the battle ultimately be won. ■

'We expect that 10 years from now, each cancer patient is going to want to get a genomic analysis of their cancer.'
—BRAD OZENBERGER, DIRECTOR OF THE CANCER GENOME ATLAS

Document 21

1000 Genomes

Phase 1 sequence data:

- Low-coverage: 1094 samples
- Exome: 1128 samples
- 39 million SNPs, 100,000 indels, 84,000 SVs
- Integrated data set to be released in October

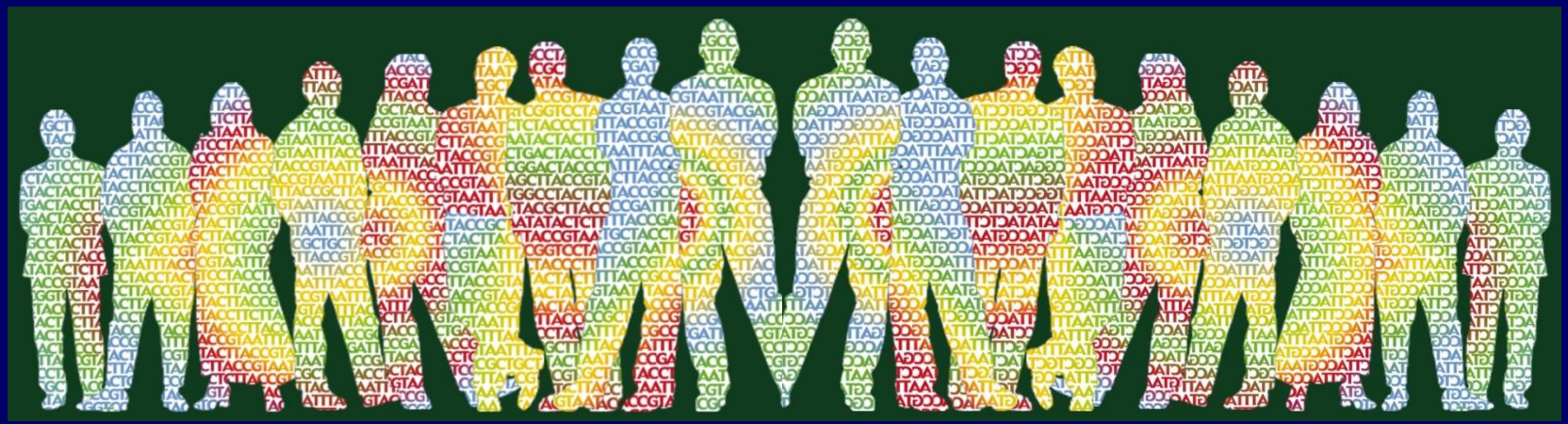
Phase 2 sequence data by Fall:

- Low-coverage and exome: 1721 samples

Document 22

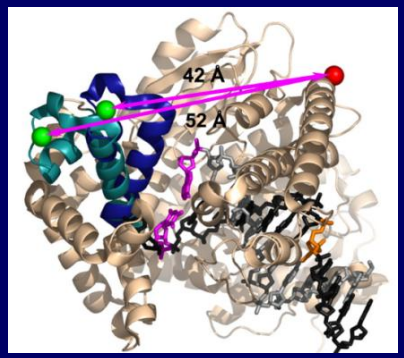
Possible/Likely NIH-Wide Inventory of Genome Sequencing Projects

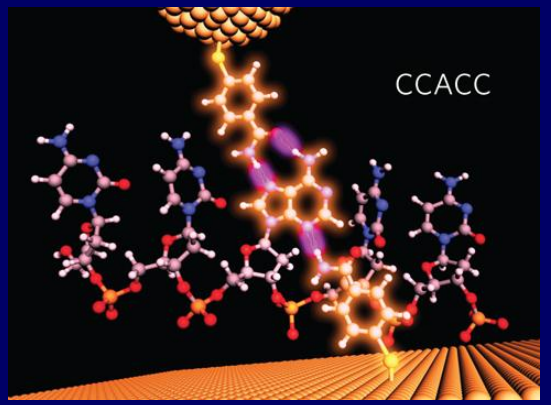
- Early discussions about desire to develop a trans-NIH inventory of ongoing genome-sequencing projects
- Recent fact-finding effort led by Teri Manolio on behalf of the NIH Director
- Momentum gathering quickly— likely will discuss at next Council meeting

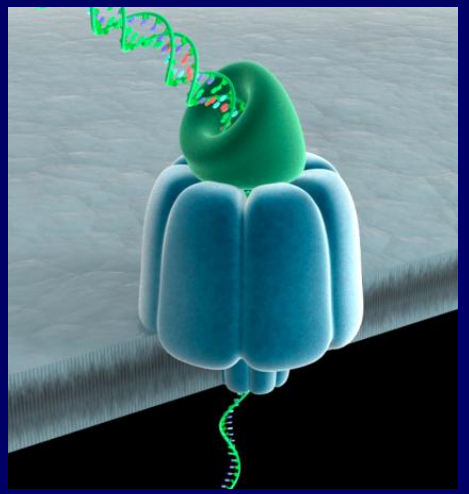


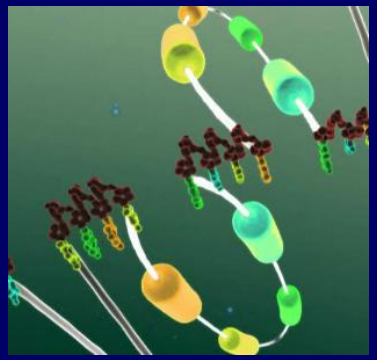
DNA Sequencing Technology Development

- 9 awards
- $14.4M committed
- Approaches:
  - polymerase conformation
  - protein and solid state nanopores, conductance and tunneling
  - electron microscopy
  - nucleotide time-of-flight
  - template expansion
  - libraries for contiguity









>>>New applications due 10/18/11

Lab to Marketplace: Tools for Biomedical and Behavioral Research

- Re-issued Program Announcement (PA-11-335) for Small Business grant mechanisms
- NIH-wide program
- Translation of technologies for biomedical or behavioral research into the marketplace

>>>Submission by November 5, 2011

Document 23

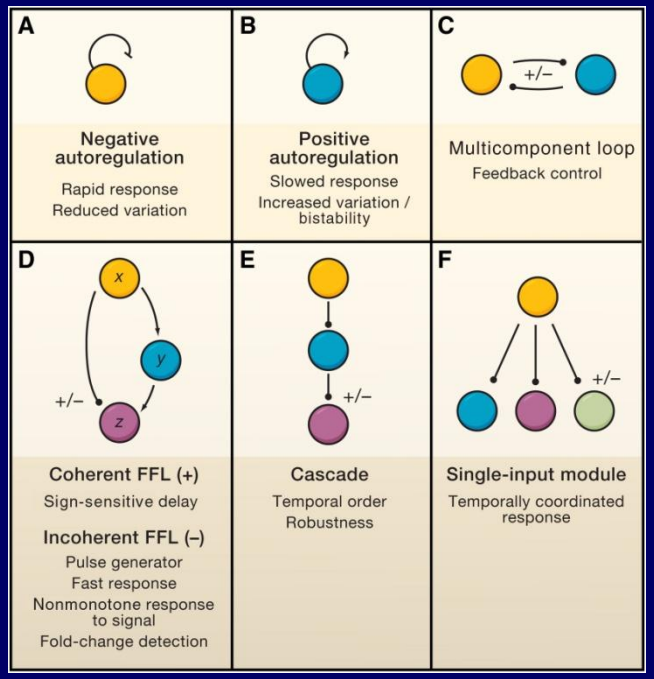
ENCODE & modENCODE

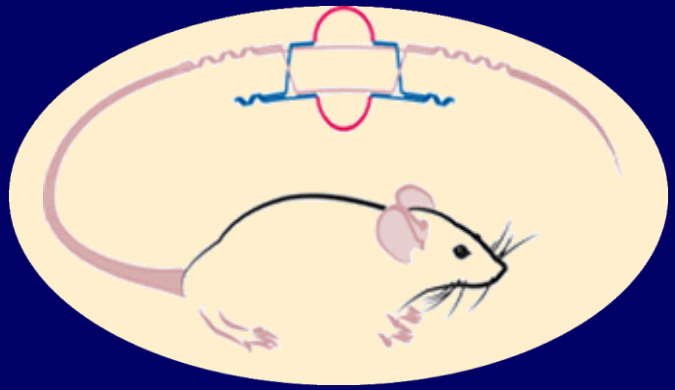
- modENCODE Symposium: June 20-21, 2012
- Integrated analysis papers
  - ENCODE
  - modENCODE – comparison of fly and worm (and human)
- Technology development FOA applications received (to be discussed at February 2012 Council)

Centers of Excellence in Genomic Science (CEGS)

- One new award in Fiscal Year 2011
- ‘Center for Cell Circuits’

P.I.: Aviv Regev
Broad Institute





KOMP/KOMP2

KOMP

- Ends September
- 8700 knockouts as of Aug 1
- 8500 is the goal

KOMP2

- Starts this year
- $111M over 5 years
- 2500 is the goal

KOMP Finale, KOMP2 Kickoff, and IMPC Launch Meeting: September 28-29, 2011

New ELSI Program Announcements

- **Three New Announcements: Regular Research Grant (R01); Small Research Grant (R03); & Exploratory/Developmental Research Grant (R21)**
- **Based on new Strategic Plan's *Genomics & Society* Priority Areas:**
  - **Genomic Research**
  - **Genomic Health Care**
  - **Broader Societal Issues**
  - **Legal, Regulatory, & Policy Issues**

Document 27

Challenges in Research Ethics and Policy: Perspectives on Data Sharing

July 2011

- Organized by the eMERGE Consent & Community Consultation Work Group
- Focused on the impact of data sharing policies on researchers, research participants, and community partners
- Explored how data on these issues can be used to inform the development of research policy
- Speakers included genomics and ELSI researchers, bioinformaticians, policy experts, and community advisors

Upcoming Extramural Meetings

- Centers of Excellence in ELSI Research (CEERs) Program Meeting: October 3-5, 2011

- ELSI Session at the International Congress of Human Genetics (ICHG): October 13, 2011

- Centers of Excellence in Genomic Science (CEGS) & Diversity Action Plan (DAP) Program Meeting: October 18-21, 2011

- Approaches for Characterizing Genetic Variants for Clinical Use: December 1-2, 2011

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III. Genomics Updates

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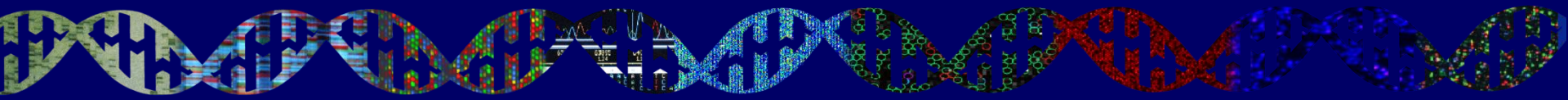
VI. NHGRI Office of the Director

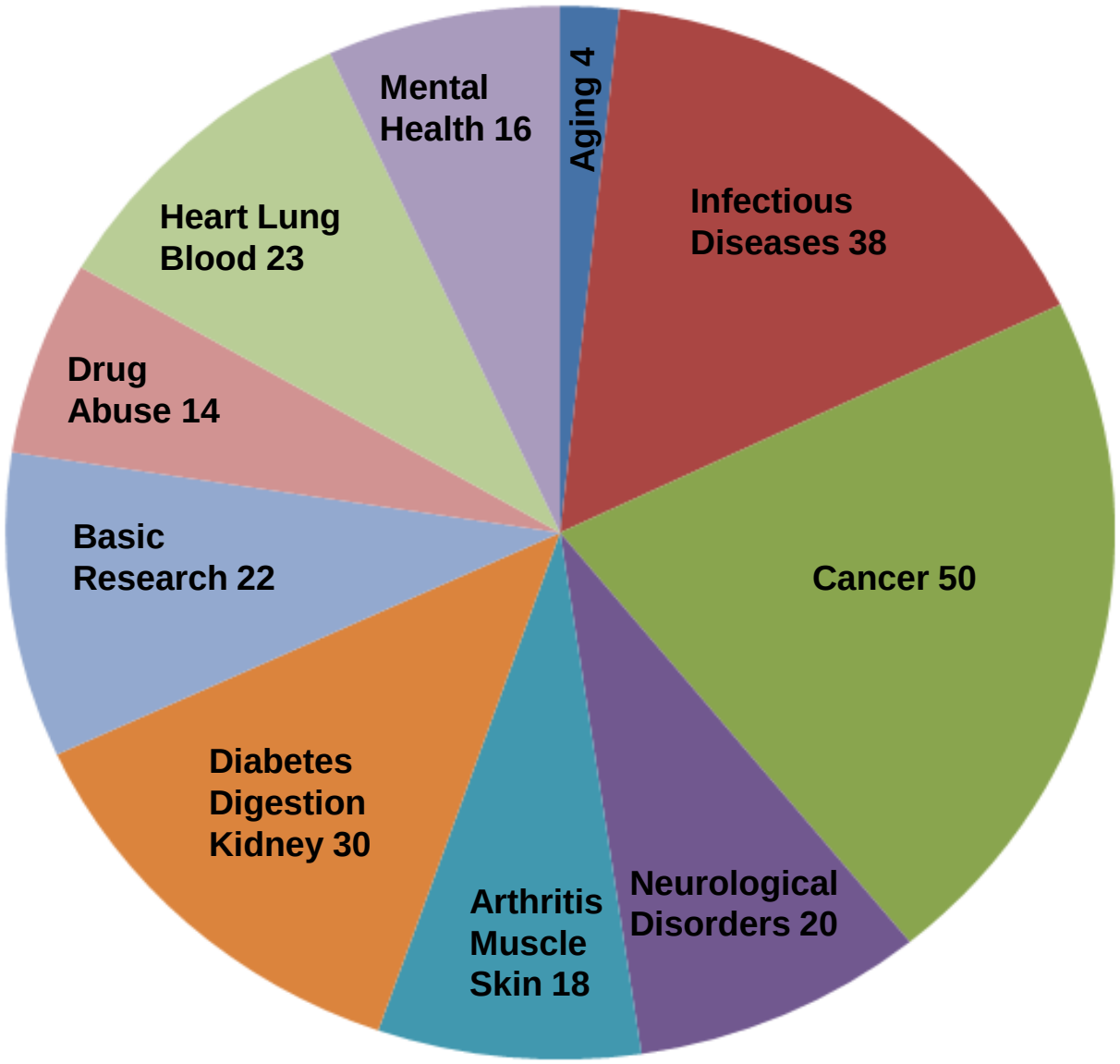
VII. NHGRI Intramural Program



Molecular Libraries Program MLP)

- MLP begins Year 4 of its Production Phase
- MLP met Year 3 quantitative milestones
- To date, the MLP network has:
  - Accepted 490 HTS assays
  - Completed >180 chemistry projects
  - Produced 229 small-molecule probes
- Future focus on qualitative milestones
  - Higher-quality probes
  - Better-characterized probes

MLP: Probes by Health Area



MLP: First IND Filing on MLP Probe

Scripps Probe ML007: Sphingosine 1-Phosphate Receptor 1 Antagonist for Multiple Sclerosis

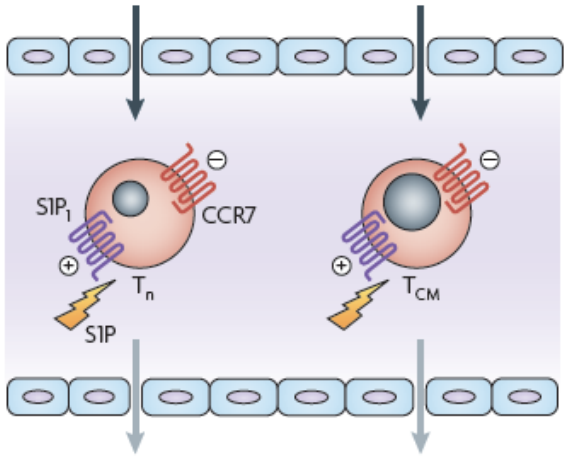
in the '902 patent were not obvious lead compounds. Moreover, at the time that the '599 patent was filed, there was a clear preference in the prior art for lipophilic groups at position 4 — indeed this often led to improved properties — so adding a hydrophilic hydroxy-isopropyl group was also not obvious.

Daiichi Sankyo versus Mylan Laboratories: http://www.cafc.uscourts.gov/images/stories/opinions-orders/09-1511.pdf

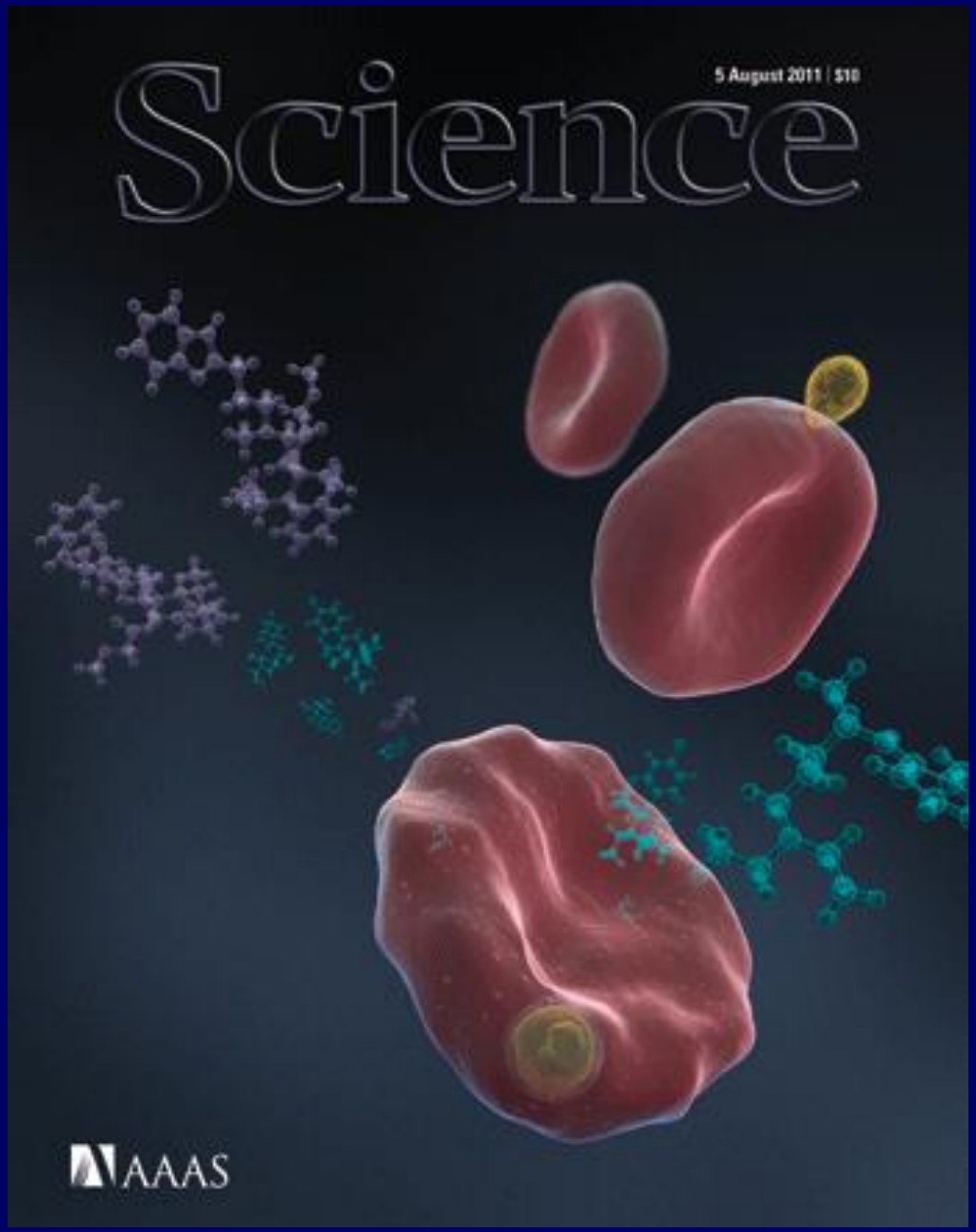
Charlotte Harrison

Sphingosine 1-phosphate

Shingosine 1-phosphate (S1P) is a lipid mediator that produces biological actions — such as on the vascular and the immune systems — through the activation of G protein-coupled S1P receptors ($S1P_1$–$S1P_5$). In their Case History on page 883, Brinkmann and colleagues describe the discovery and the development of fingolimod, a sphingosine analogue that is phosphorylated by sphingosine kinases in the cell. The drug was approved by the US Food and Drug Administration in September 2010 as a first-line treatment for relapsing forms of multiple sclerosis, thereby becoming the first oral disease-modifying therapy to be approved for multiple sclerosis in the United States. Here in TABLE 1, we summarize international patent applications published in the past year related to S1P. Data were researched using the SureChem database from Macmillan Publishers.



NIH Center for Translational Therapeutics



RESEARCH ARTICLES

Chemical Genomic Profiling for Antimalarial Therapies, Response Signatures, and Molecular Targets

Jing Yuan,[1]* Ken Chih-Chien Cheng,[2]* Ronald L. Johnson,[2]† Ruili Huang,[2] Sittiporn Pattaradilokrat,[1] Anna Liu,[1] Rajarshi Guha,[2] David A. Fidock,[3] James Inglese,[2] Thomas E. Wellems,[1] Christopher P. Austin,[2]‡ Xin-zhuan Su[1]‡

Malaria remains a devastating disease largely because of widespread drug resistance. New drugs and a better understanding of the mechanisms of drug action and resistance are essential for fulfilling the promise of eradicating malaria. Using high-throughput chemical screening and genome-wide association analysis, we identified 32 highly active compounds and genetic loci associated with differential chemical phenotypes (DCPs), defined as greater than or equal to fivefold differences in half-maximum inhibitor concentration (IC_{50}) between parasite lines. Chromosomal loci associated with 49 DCPs were confirmed by linkage analysis and tests of genetically modified parasites, including three genes that were linked to 96% of the DCPs. Drugs whose responses mapped to wild-type or mutant *pfcrt* alleles were tested in combination in vitro and in vivo, which yielded promising new leads for antimalarial treatments.

MICROBIOLOGY

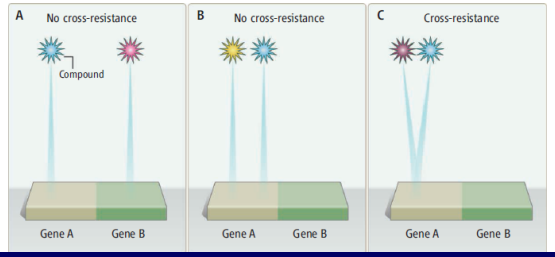
Exploiting Malaria Drug Resistance to Our Advantage

High-throughput chemical and gene analyses help identify promising pairs of antimalarial compounds that could prevent resistance.

Nick Cammack

In the world of developing anti-infective drugs, resistance comes with the territory. It is literally in the DNA (or RNA) of the viruses and pathogens targeted for treatment. Because resistance has far-reaching consequences for human health, researchers have studied the resistance of infectious agents such as human immunodeficiency virus (HIV) type 1 with unprecedented intensity and documented in excruciating detail the genetic determinants of resistance to FDA-approved drugs (*1*). However, understanding drug resistance in a complex eukaryotic parasite, such as the *Plasmodium* parasite that causes malaria, is a very different challenge. On page 724 of this issue, Yuan *et al.* (*2*) confront the issue head-on. Using high-throughput chemical and gene analysis methods, they not only identify potential new antimalarial drugs that could be used in combination to suppress the development of drug resistance but

In the context of infectious pathogens, drug resistance is the response of the organism to a new environment. In the case of malaria, resistance reflects the parasite's ability to survive and replicate despite the presence of a drug, reducing the drug's effectiveness. Many mechanisms can confer resistance. The parasite can decrease drug accumulation, for example, by actively pumping the drug out of its cell membrane. It could also modify a target site on a specific protein, interfere with a metabolic pathway, and per-



Document 29

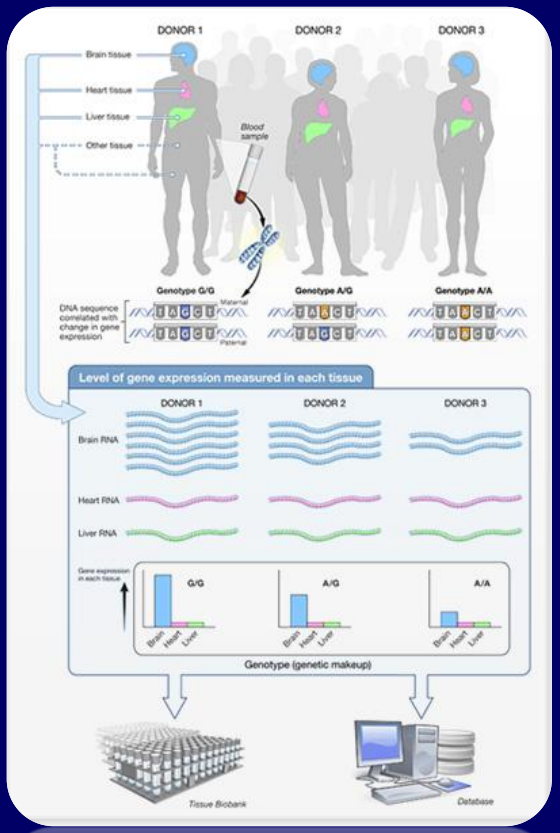
Human Microbiome Project

- 108 publications in PubMed cite HMP support
- Two major papers from the HMP Consortium near completion

- Anticipated submission in Fall 2011

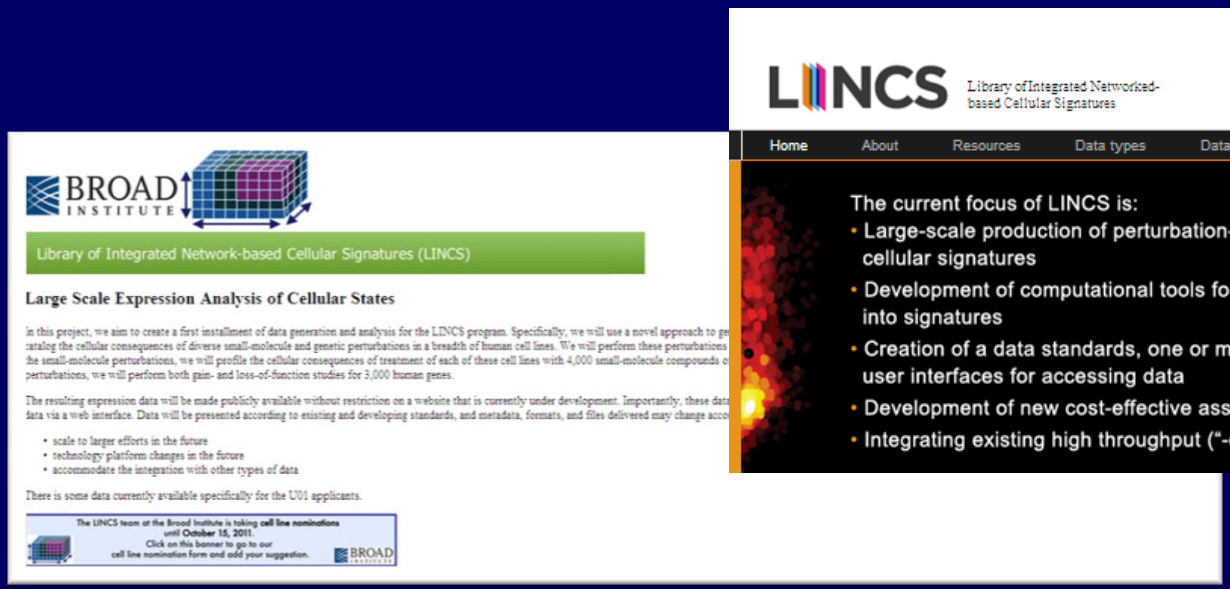
Genotype-Tissue Expression (GTEx)

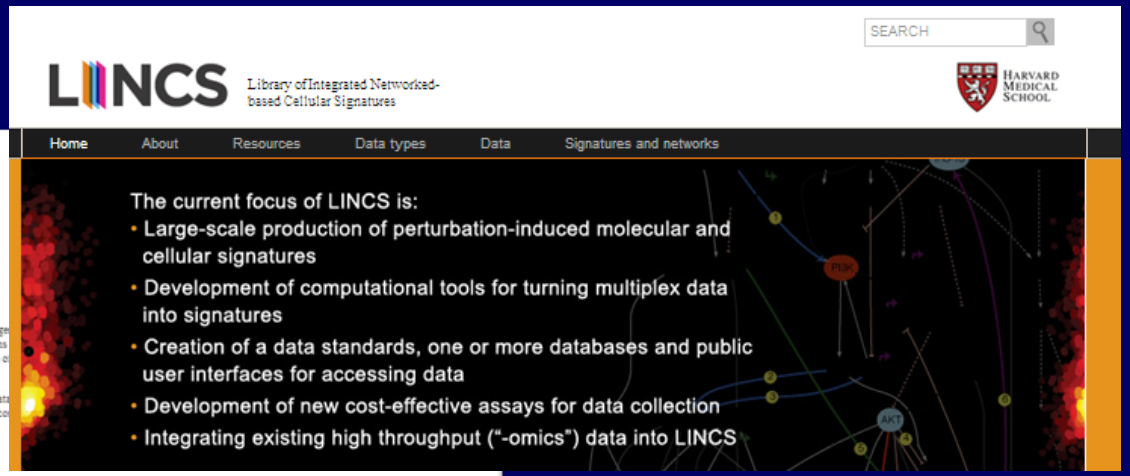
- Recruitment well underway
- Laboratory analysis ongoing
- External Scientific Panel met during June 2011 meeting
- Presentation by Jeff Struewing later in Open Session



Library of Integrated Network-based Cellular Signatures (LINCS)

- **Fall Consortia Meeting: October 2011**
- **Eight UO1 applications will be awarded in September 2011**
- **Production Center public websites now live**

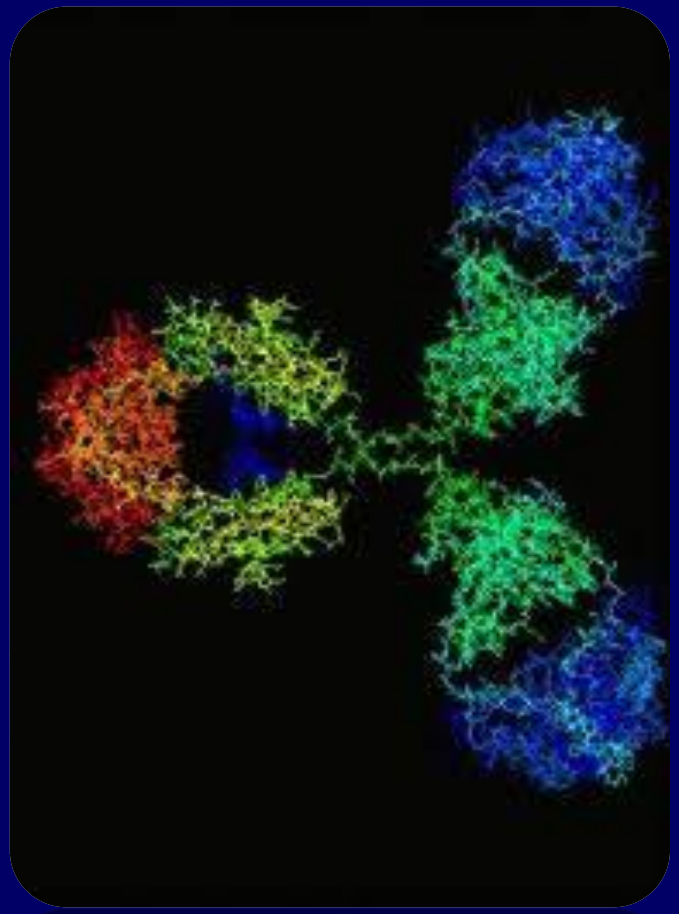




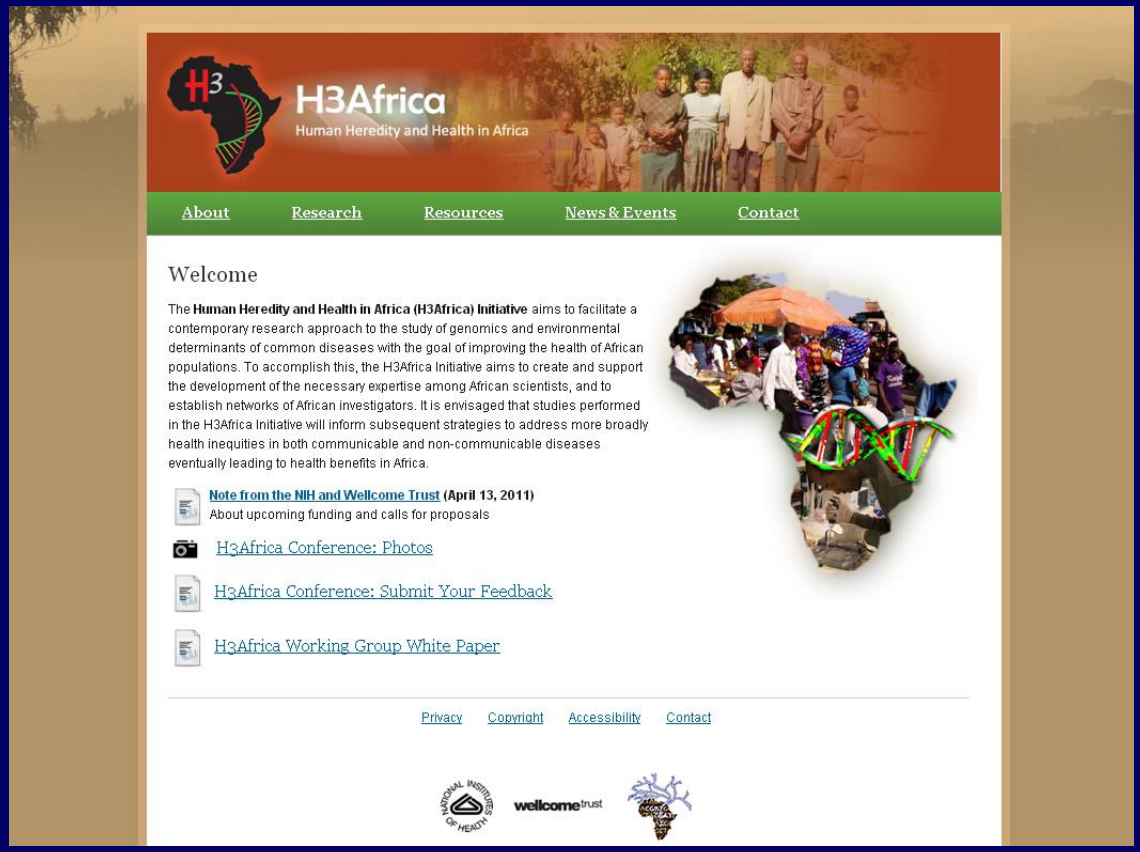
Protein Capture Reagents Program

- Production and Technology Development U54 awards to be discussed during Closed Session

- Protein Capture Consortia Meeting planned for Winter 2011



Human Heredity and Health in Africa (H3Africa)

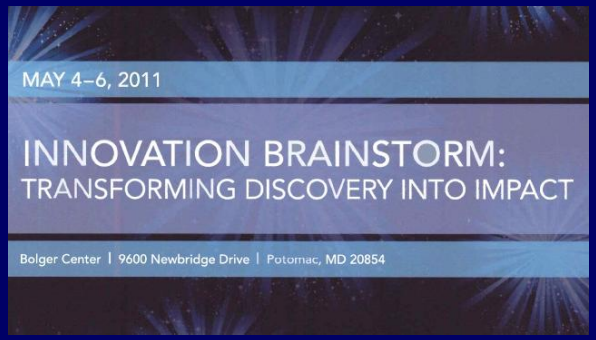


- **Presentation by Jane Peterson later in Open Session**

Document 34

New Common Fund Initiatives

- **Innovation Brainstorm Meeting**



- **NHGRI Proposals**
  1. **Disruptive Proteomics Technologies**
  2. **Molecular Phenotypes for Genome Function and Disease**

Comment Period open until September 14, 2011.

Division of Program Coordination, Planning, and Strategic Initiatives (DPCPSI) | National Institutes of Health | U.S. Department of Health and Human Services

The NIH Common Fund
WE ACCELERATE DISCOVERY.

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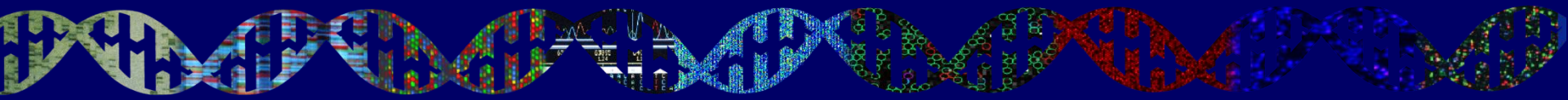
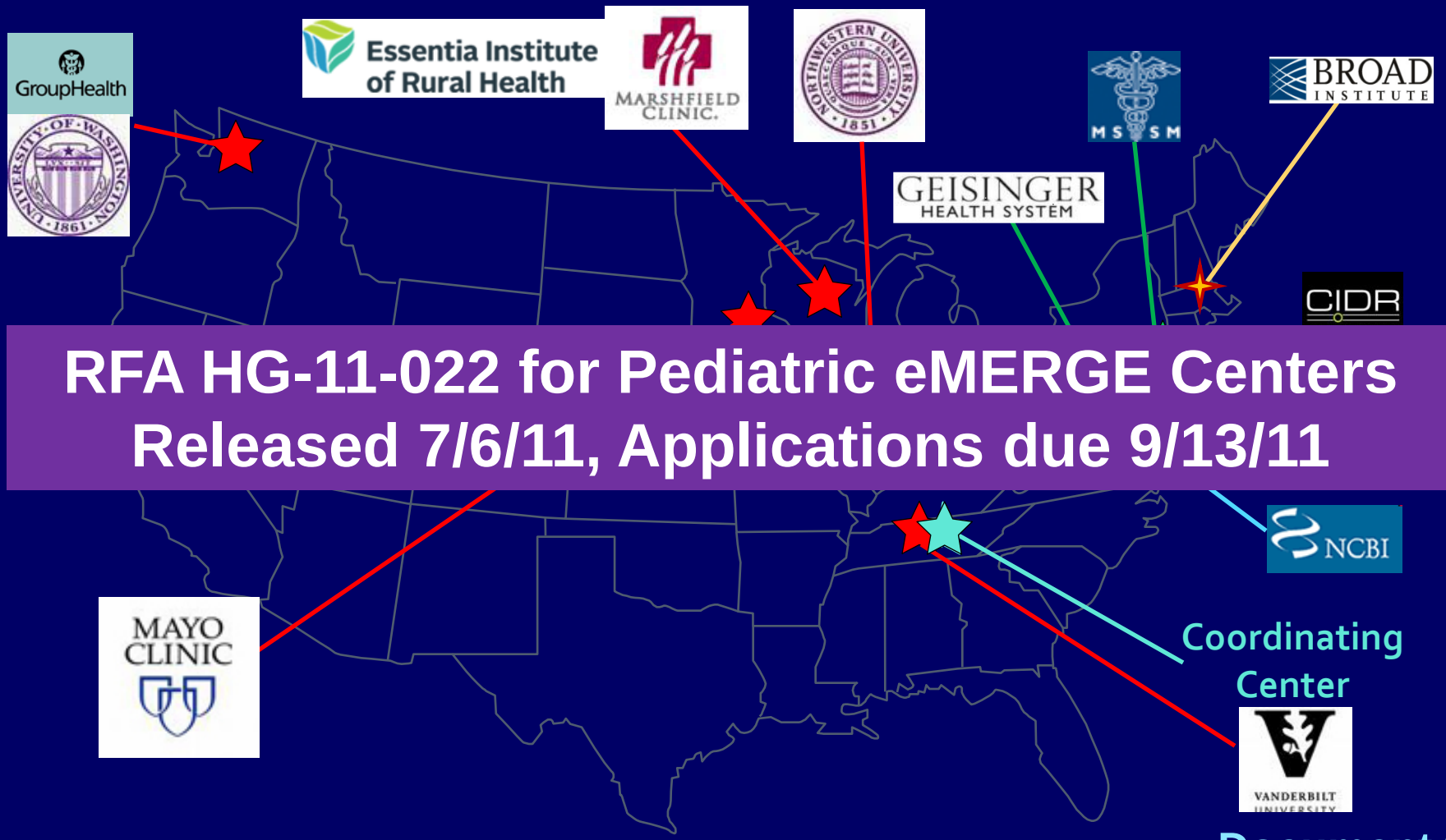


The eMERGE Network

electronic Medical Records & Genomics

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



RFA HG-11-022 for Pediatric eMERGE Centers
Released 7/6/11, Applications due 9/13/11

Coordinating Center

Document 36

NHGRI Catalog of Published Genome-Wide Association Studies (GWAS)

Document 37

PhenX Program

- **NHGRI-NIDA collaboration to expand substance use measures in Toolkit**
  - ❖ **NIDA provided $730,000 for project**
  - ❖ **Working Groups to focus on:**
    Substance Use, Intermediate Phenotypes, Risk Factors (psychosocial, Cognitive, community), Co-Morbid Conditions
- **Kick-off meeting for 7 PIs using PhenX measures**
- **Validating PhenX measures and evaluating usefulness of Toolkit**

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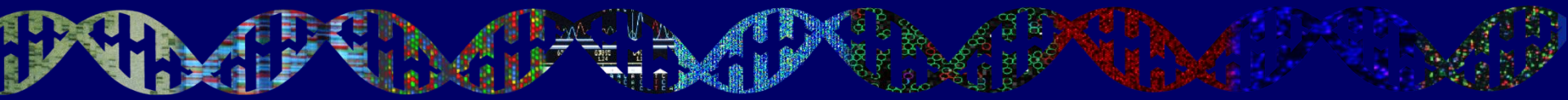


Blue Ribbon Panel Review of NHGRI Intramural Research Program

- David Page, M.D. (Chair)
- Rick Myers, Ph.D. (NACHGR)
- Bruce Korf, M.D., Ph.D. (BSC)
- Wylie Burke, M.D., Ph.D
- Nancy Cox, Ph.D
- Bob Waterston, M.D., Ph.D
- Huda Zoghbi, M.D.

NHGRI Intramural Research Highlights: Proteus Syndrome Gene Discovered

The NEW ENGLAND JOURNAL of MEDICINE

ORIGINAL ARTICLE

A Mosaic Activating Mutation in *AKT1* Associated with the Proteus Syndrome

Marjorie J. Lindhurst, Ph.D., Julie C. Sapp, Sc.M., Jamie K. Teer, Ph.D., Jennifer J. Johnston, Ph.D., Erin M. Finn, B.A., Kathryn Peters, M.S., Joyce Turner, M.S., Jennifer L. Cannons, Ph.D., David Bick, M.D., Laurel Blakemore, M.D., Catherine Blumhorst, M.S.N., Knut Brockmann, M.D., Peter Calder, M.B., B.S., Natasha Cherman, Ph.D., Matthew A. Deardorff, M.D., Ph.D., David B. Everman, M.D., Gretchen Golas, M.S., Robert M. Greenstein, M.D., B. Maya Kato, M.D., Kim M. Keppler-Noreuil, M.D., Sergei A. Kuznetsov, Ph.D., Richard T. Miyamoto, M.D., Kurt Newman, M.D., David Ng, M.D., Kevin O'Brien, M.S., Steven Rothenberg, M.D., Douglas J. Schwartzentruber, M.D., Virender Singhal, M.D., M.B.A., Roberto Tirabosco, M.D., Joseph Upton, M.D., Shlomo Wientroub, M.D., Elaine H. Zackai, M.D., Kimberly Hoag, Tracey Whitewood-Neal, Pamela G. Robey, Ph.D., Pamela L. Schwartzberg, M.D., Ph.D., Thomas N. Darling, M.D., Ph.D., Laura L. Tosi, M.D., James C. Mullikin, Ph.D., and Leslie G. Biesecker, M.D.

NHGRI Intramural Research Highlights

nature genetics

Exome sequencing identifies *ACSF3* as a cause of combined malonic and methylmalonic aciduria

Jennifer L Sloan, Jennifer J Johnston, Irini Manoli, Randy J Chandler, Caitlin Krause, *et al.*

NBEAL2 is mutated in gray platelet syndrome and is required for biogenesis of platelet α-granules

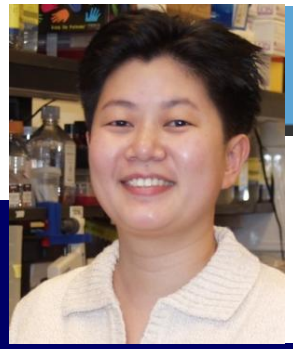
Meral Gunay-Aygun, *et al.*

Rapamycin Reverses Cellular Phenotypes and Enhances Mutant Protein Clearance in Hutchinson-Gilford Progeria Syndrome Cells

Kan Cao, *et al.*

Science Translational Medicine

AAAS



JCI The Journal of Clinical Investigation

FREE ACCESS TO ALL RESEARCH

Progerin and telomere dysfunction collaborate to trigger cellular senescence in normal human fibroblasts

Kan Cao,[1,2] Cecilia D. Blair,[1] Dina A. Faddah,[1] Julia E. Kieckhaefer,[2] Michelle Olive,[1] Michael R. Erdos,[1] Elizabeth G. Nabel,[1] and Francis S. Collins[1]

Neuron

A Transcriptomic Atlas of Mouse Neocortical Layers

T. Grant Belgard[1, 2, 3], Ana C. Marques[1], Peter L. Oliver[1], Hatice Ozel Abaan[3], Tamara M. Sirey[1, 2], Anna Hoerder-Suabedissen[2], Fernando García-Moreno[2], Zoltán Molnár[2, ✉, ✉], Elliott H. Margulies[3, 4, ✉, ✉] and Chris P. Ponting[1, ✉, ✉]

Special Thanks!


genome.gov
THE BRIGHT FUTURE OF HUMAN GENOMICS