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National Human Genome Research Institute
National Institutes of Health

DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

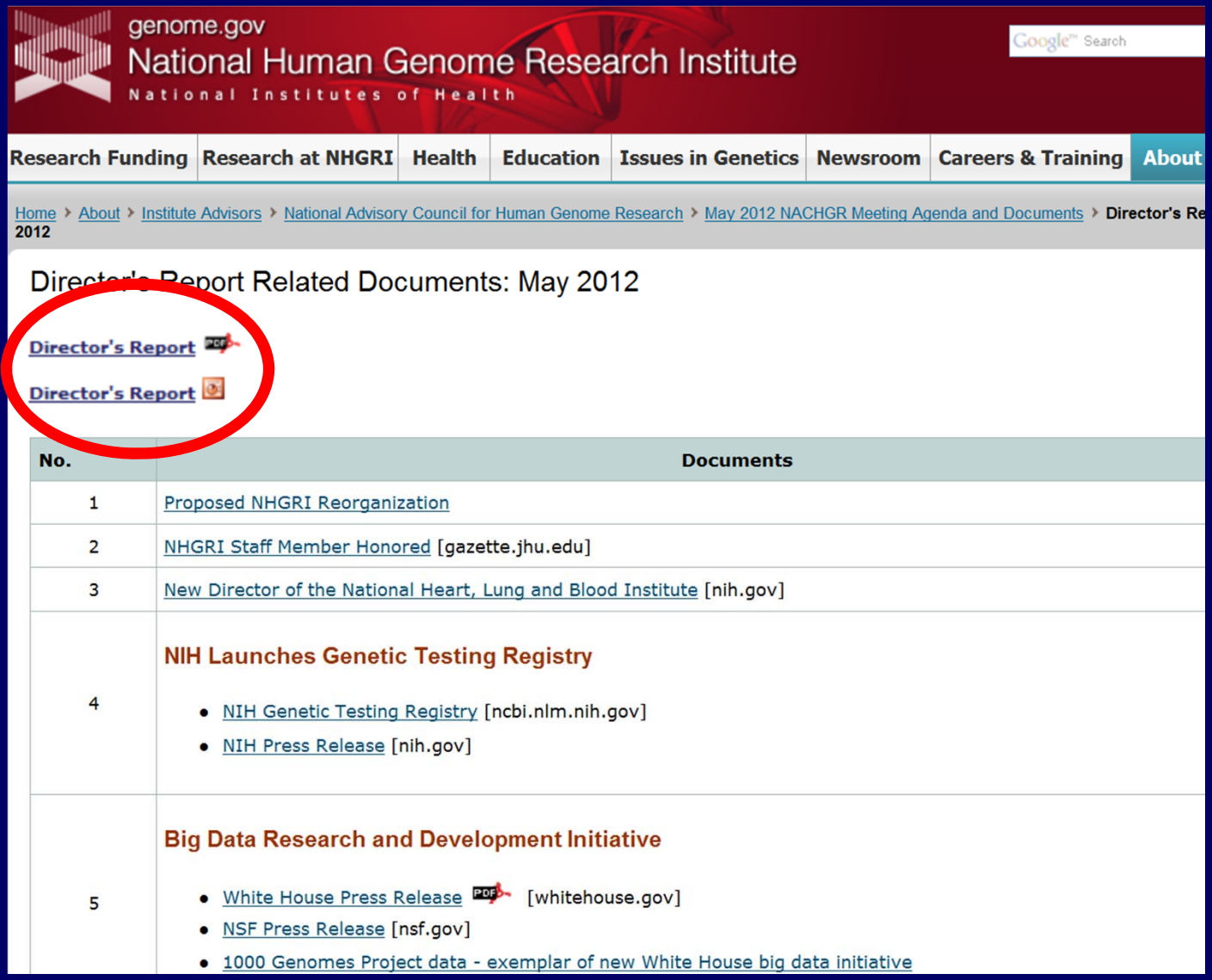
May 2012

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

National Advisory Council for Human Genome Research

Rockville, Maryland

February 13, 2012

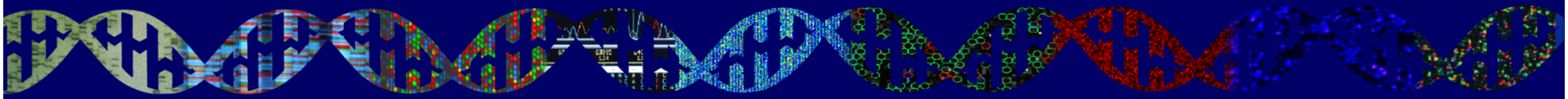


genome.gov/DirectorsReport

Document #

Open Session Presentations

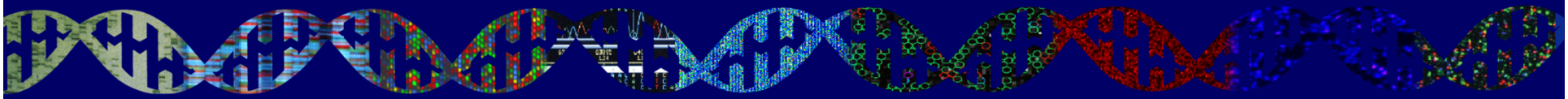
- **NHGRI Training Portfolio**
  - **Bettie Graham**
- **NIH Policy on Applicants with More than $1.5M in Grant Support**
  - **Bettie Graham**
- **Update on the X Chromosome Program Notice**
  - **Anastasia Wise**



Open Session Presentations

Concept Clearances:

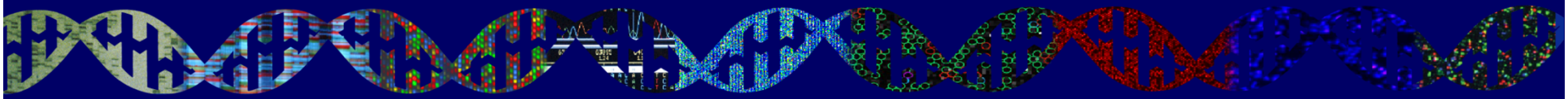
- **Genomic Sequencing and Newborn Screening Disorders**
  - **Anastasia Wise**
- **Clinically Relevant Variants Resource**
  - **Erin Ramos**



Open Session Presentations

Program Updates:

- **1000 Genomes Project**
  - **Lisa Brooks**



I. General NHGRI Updates

II. General NIH Updates

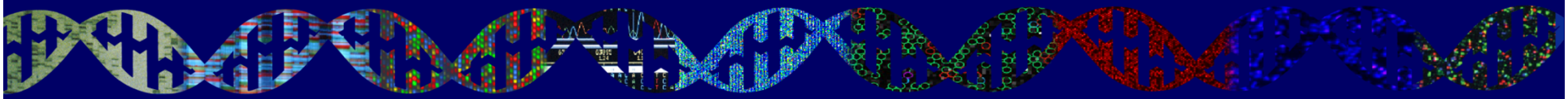
III. Genomics Updates

IV. NHGRI Extramural Program

V. NIH Common Fund Programs

VI. NHGRI Office of the Director

VII. NHGRI Intramural Program



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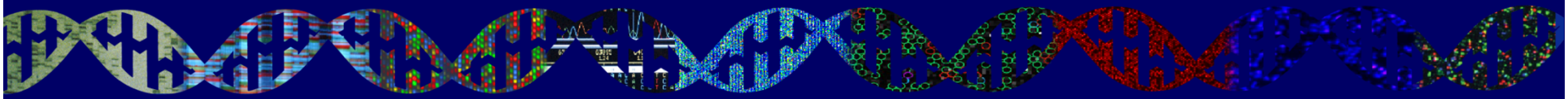

Proposed NHGRI Reorganization

Proposed NHGRI Reorganization

Times change and so, too, should institutions. For the National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH), a natural time for change has arrived, and the Institute is proposing an internal reorganization to reflect our current and future genomics research portfolio and associated activities more appropriately.

In 1988, NIH created an office that eventually became NHGRI; at the time, the single charge to that office was to oversee NIH's contributions to the Human Genome Project. As such, the office started with a simple organization — a director's office and a team managing grants. Today, NHGRI manages dozens of named scientific projects and a research portfolio that is multifaceted and highly diverse. In aggregate, NHGRI's current suite of responsibilities requires a more sophisticated management structure.

Moreover, with the completion of the Human Genome Project in 2003, NHGRI has worked with the international community of genomics researchers to develop strategic plans to guide the field as a whole. NHGRI published its most recent plan in the journal *Nature* in February 2011 (*Charting a course for genomic medicine from base pairs to bedside*). This new strategic vision is organized around five domains of research activities that together chart a progression from basic research elucidating the structure and biology of genomes to understanding the biology of disease and advancing the science of medicine. The ultimate goal, of course, is to improve the effectiveness of healthcare and advance human health.

Document 1

Departure of Greg Feero

Genomic Healthcare Branch

G2C2
GENETICS/GENOMICS COMPETENCY CENTER FOR EDUCATION

G3C
INTERACTIVE UNFOLDING CASE STUDIES

NEJM GENOMICS IN MEDICINE

NIH STATE-OF-THE-SCIENCE CONFERENCE
FAMILY HISTORY and Improving Health

NHGRI Staff Member Honored (x2)

Teri Manolio, M.D., Ph.D.

Document 2

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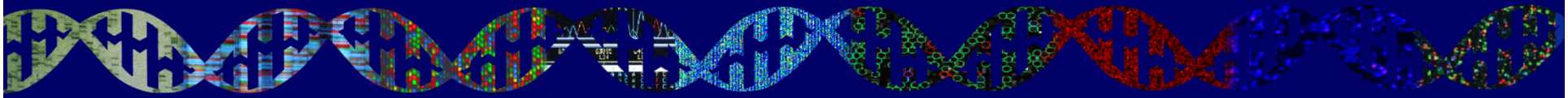


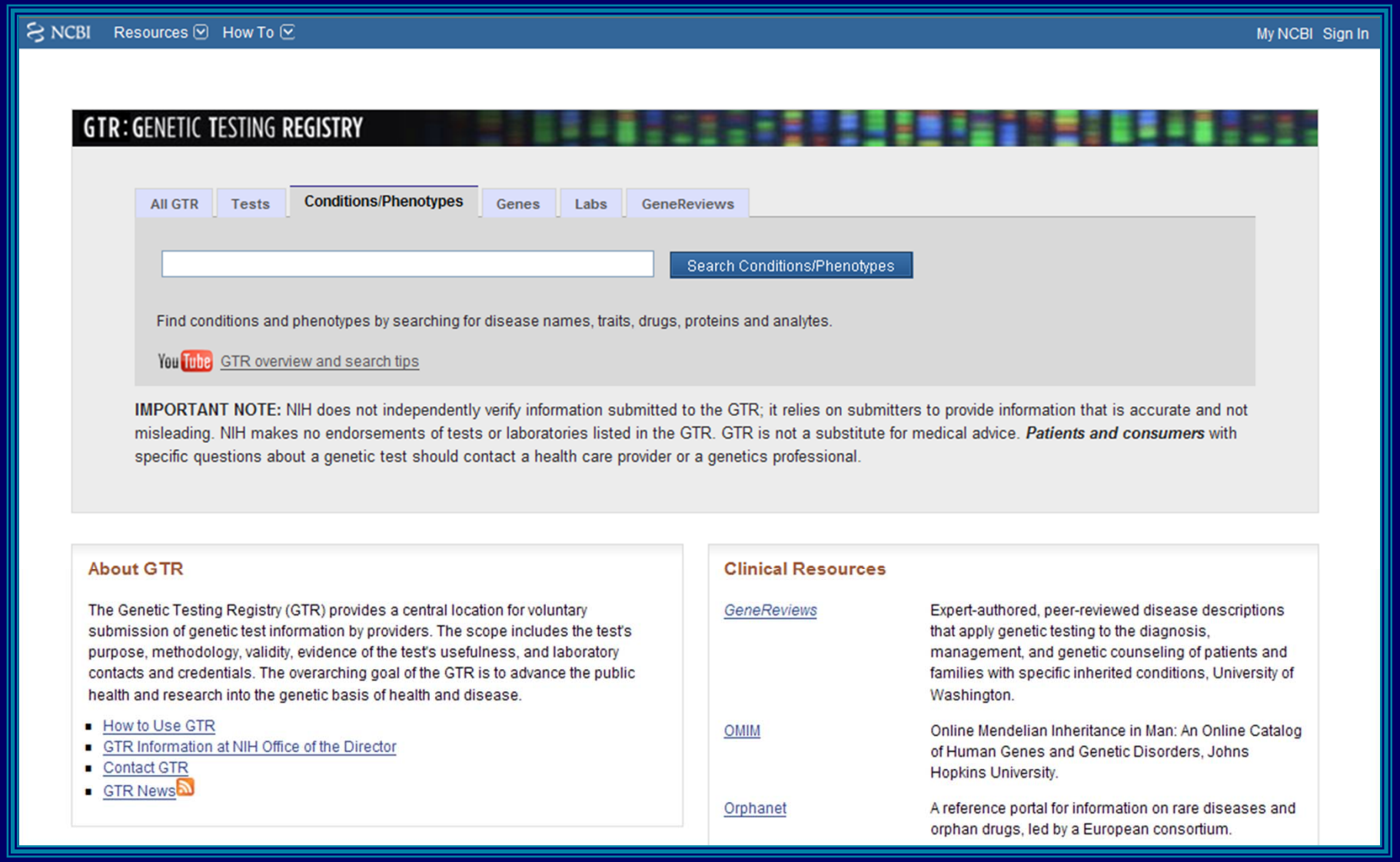
New Director: National Heart, Lung, and Blood Institute

Gary Gibbons, M.D.

Document 3

NIGMS Leadership Changes

NIH Launches Genetic Testing Registry



Document 4

‘Big Data’ Research and Development Initiative

Document 5

‘Big Data’ Planning at NIH

- **Data and Informatics Working Group**
  - **Co-Chairs: Larry Tabak and David DeMets**
  - **Report to NIH Director in June 2012**
- **Trans-NIH Subgroup on Molecular Data**
  - **Co-Leads: NHGRI and NIGMS**
- **Potential Common Fund initiative in FY2014**

Advisory Committee to the Director
NCBI Working Group

- David Ginsburg (Chair)
- Robert Gentlemen
- Richard Gibbs
- Howard Jacob
- Jill Mesirov
- Deborah Nickerson
- Christine Seidman
- Paul Sternberg
- Marc Vidal

Basic Behavioral and Social Science Opportunity Network (OppNet)

- Mission: Pursue opportunities for strengthening basic behavioral and social science research at NIH
- Collectively funded and managed

  [NHGRI FY2012 contribution: $349K]
- OppNet grants may come to September Council meeting

Document 6

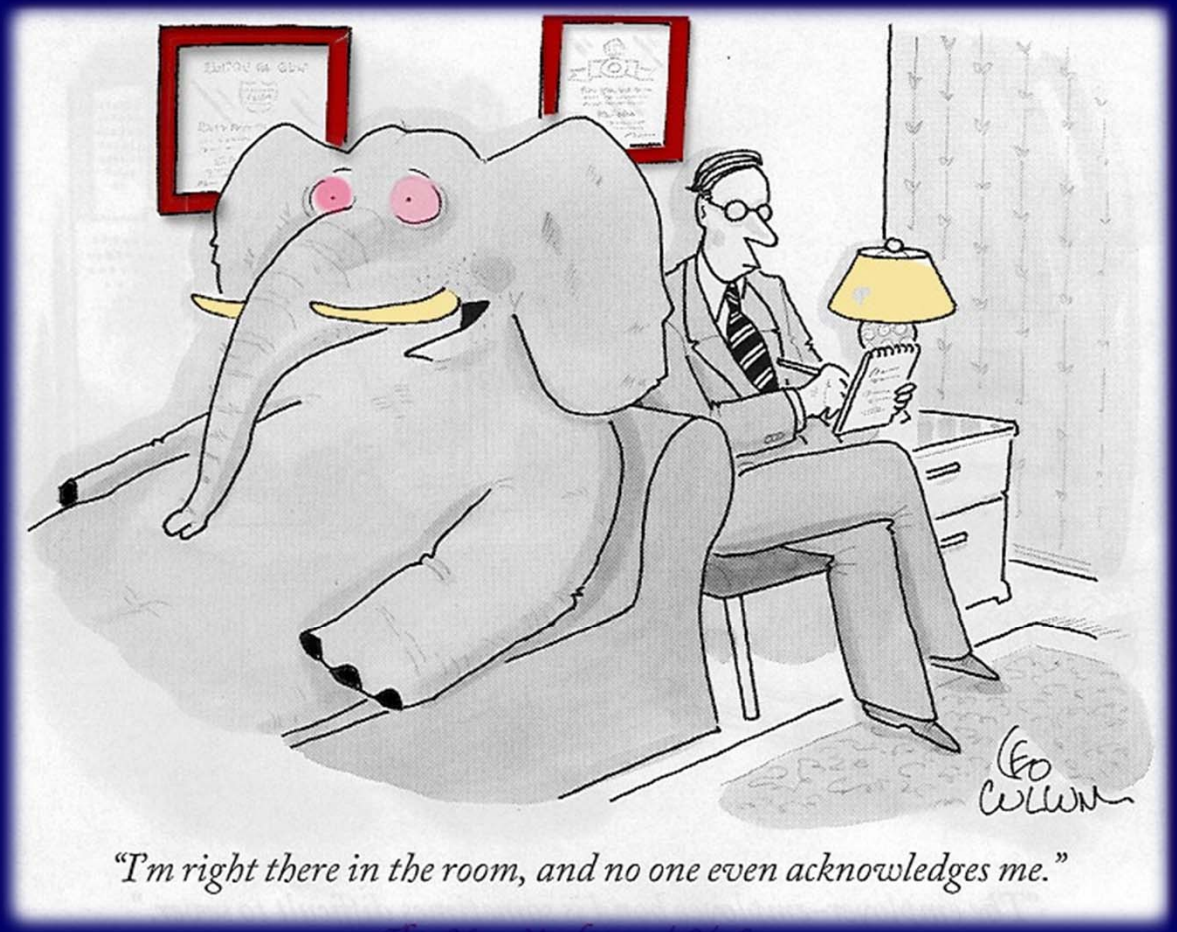
FY2013 NIH Appropriation: Overview

March 20: House NIH Hearing

March 28: Senate NIH Hearing

Document 7

FY2013 NIH Appropriation: Sequestration?



Warnings: Sequestration Impact on NIH

United for Medical Research

“33,000 fewer jobs across the United States”

“11.1% ($2.8 billion) reduction of the extramural budget”

Document 8

FY2013 NIH Appropriation: Survival Tips

Just Released: *Leadership in Decline*

Document 8

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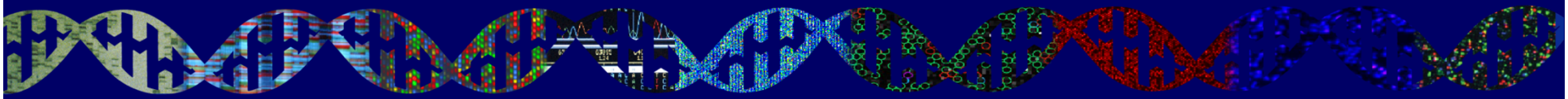
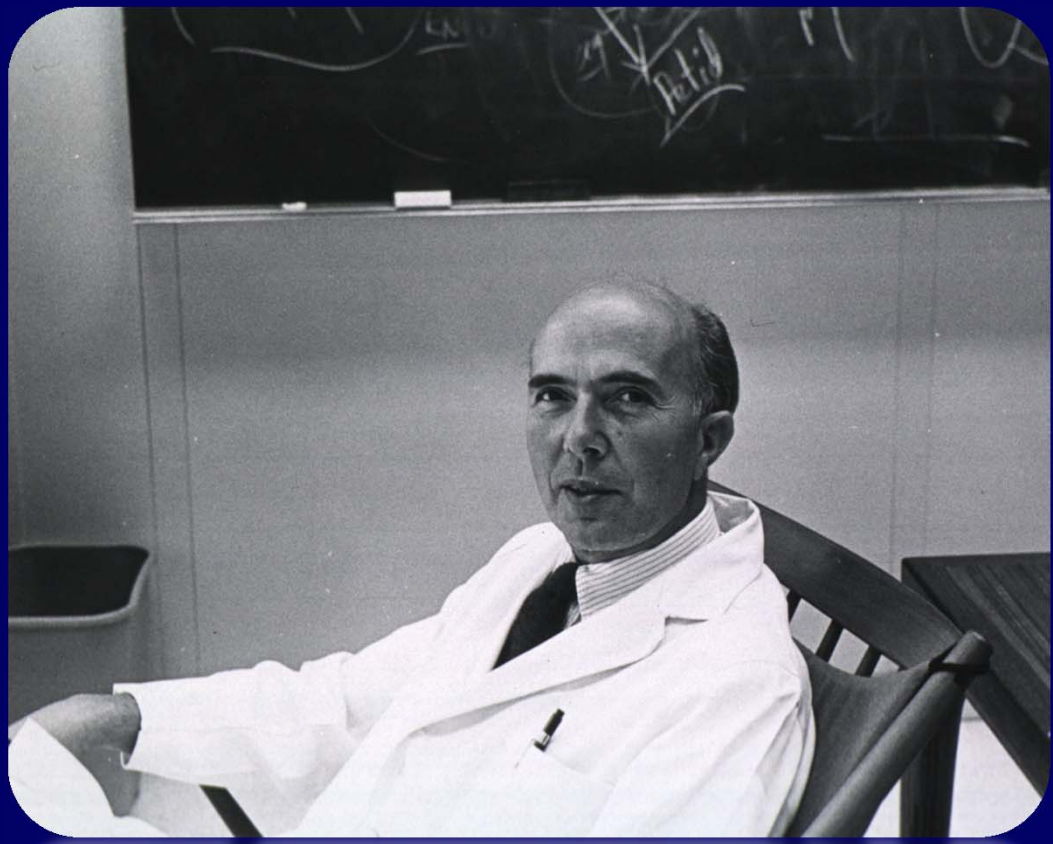


Mourning the Loss of Renato Dulbecco



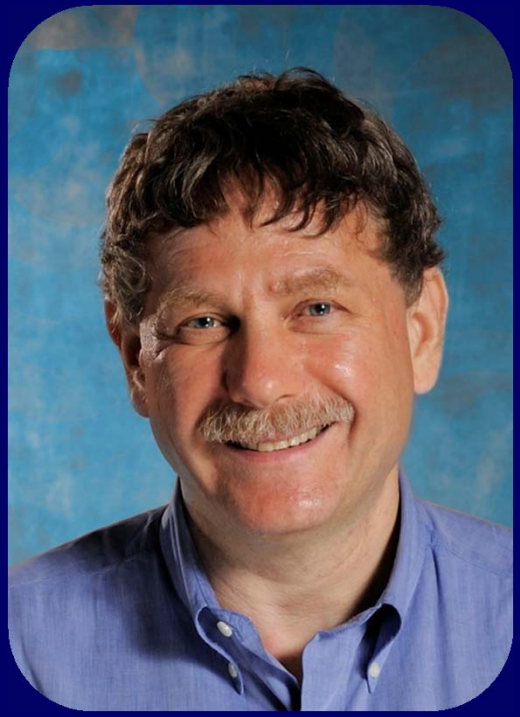
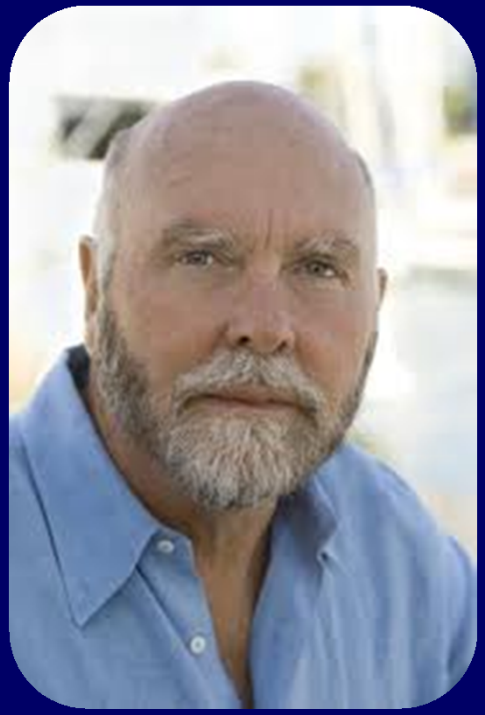
Document 9

2012 Japan Prize

Janet Rowley, M.D.

Document 10

2012 Dan David Prize

David Botstein, Ph.D.
Eric Lander, Ph.D.
Craig Venter, Ph.D.

DAN DAVID PRIZE

Document 11

Outstanding St. Louis Scientists Award

Academy of Science St. Louis

CONNECTING SCIENCE & THE COMMUNITY SINCE 1856™

Tim Ley M.D.

Elaine Mardis Ph.D.

Rick Wilson Ph.D.

Document 12

Newly Elected: National Academy of Sciences

- Andy Clark
- Ron DePinho
- Evan Eichler
- Greg Hannon
- Harris Lewin
- Rick Young

- Subra Suresh

Document 13

Bruce Korf: President of the ACMG Foundation for Genetic and Genomic Medicine

American College of Medical Genetics Foundation

Better Health Through Genetics™

Document 14

Presidential Commission for the Study of Bioethical Issues

DEPARTMENT OF HEALTH AND HUMAN SERVICES

Request for Comments on Issues of Privacy and Access With Regard to Human Genome Sequence Data

AGENCY: The Presidential Commission for the Study of Bioethical Issues, Office of the Secretary, Department of Health and Human Services.

ACTION: Notice.

SUMMARY: The Presidential Commission for the Study of Bioethical Issues is requesting public comment on the ethical issues raised by the ready availability of large-scale human genome sequence data, with regard to privacy and data access and the balancing of individual and societal interests.

DATES: To assure consideration, comments must be received by May 25, 2012. Comments received after this date will be considered only as time permits.

info@bioethics.gov

Document 15

DeciBio Report on Research Tools

DeciBio
Decisive Bio-Insights

- **Genomics sector:**

  From $6.8B in 2011 to $8.9B in 2016

- **‘Next-Gen’ DNA sequencing market:**

  From $1.05B in 2011 to $2.25B in 2016

Document 16

National Bioeconomy Blueprint

NATIONAL BIOECONOMY BLUEPRINT

April 2012

THE WHITE HOUSE
WASHINGTON

Foundational Technologies: Today and for the Bioeconomy of the Future

Decades of biological research have revealed detailed information about the components of complex systems that characterize life—genes, cells, organisms, ecosystems—and how they interact. As mentioned earlier, genetic engineering, DNA sequencing, and high-throughput technologies have transformed the practice and potential of biological research. Yet, there is substantial room for advancement and discovery; additional scientific and technological revolutions are needed to fundamentally improve the approaches needed to confront the complex societal challenges of the future.

Emerging technologies such as synthetic biology, proteomics, and information technologies, including bioinformatics and computational biology, have the potential to create a vibrant bioeconomy.

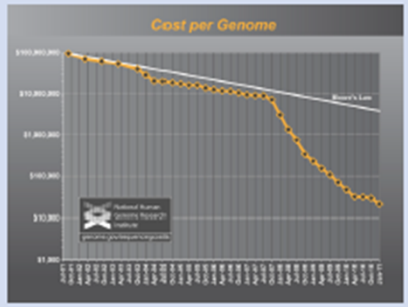


Figure 2. "Cost per Genome" – the cost of sequencing a human-sized genome. Data from 2001 through October 2007 represent the costs of generating DNA sequence using first generation sequencing technology. Beginning in January 2008, the data represent the costs of generating DNA sequence using 'second-generation' (or 'next-generation') sequencing platforms. The change in instruments represents the rapid evolution of DNA sequencing technologies that has occurred in recent years.[38]

Synthetic Biology: The ability to quickly and cheaply read and synthesize DNA sequences has transformed biological research. As shown in Figure 2, the costs of sequencing a genome decreased dramatically from 2004 to 2011. Expansive genetic libraries, with billions of genome variants created daily, are now available due to huge strides in "reading and writing" DNA. While the sequencing of the first human genome took 13 years and cost $2.7 billion, researchers can now sequence a human genome for a fraction of that cost (~$7,700) and within two weeks' time. Synthetic biology, the design and wholesale construction of new biological parts and systems, and the re-design of existing, natural biological systems for tailored purposes, integrates engineering and computer-assisted design approaches with biological research. Since natural biological systems are so complicated, a primary focus of synthetic biologists is developing technologies that make the engineering of biology easier, faster, and more predictable. This ability to quickly engineer organisms in laboratories holds vast potential for the bioeconomy, as engineered organisms could dramatically transform modern practices in high-impact fields such as agriculture, manufacturing, energy generation, and medicine. Much work lies ahead, including identifying and standardizing biological and molecular components, but this powerful new area of

38. Wetterstrand KA.DNA Sequencing Costs: Data from the NHGRI Large-Scale Genome Sequencing Program Available at: www.genome.gov/sequencingcosts.

Document 17

Biotech Patents and the Courts

2012 Advances in Genome Biology and Technology Meeting

2012 Sponsors

Gold

PACIFIC BIOSCIENCES™

ronze

Caliper illumina Complete genomics

NuGEN RainDance Technologies ADVANCED ANALYTICAL

organized by:

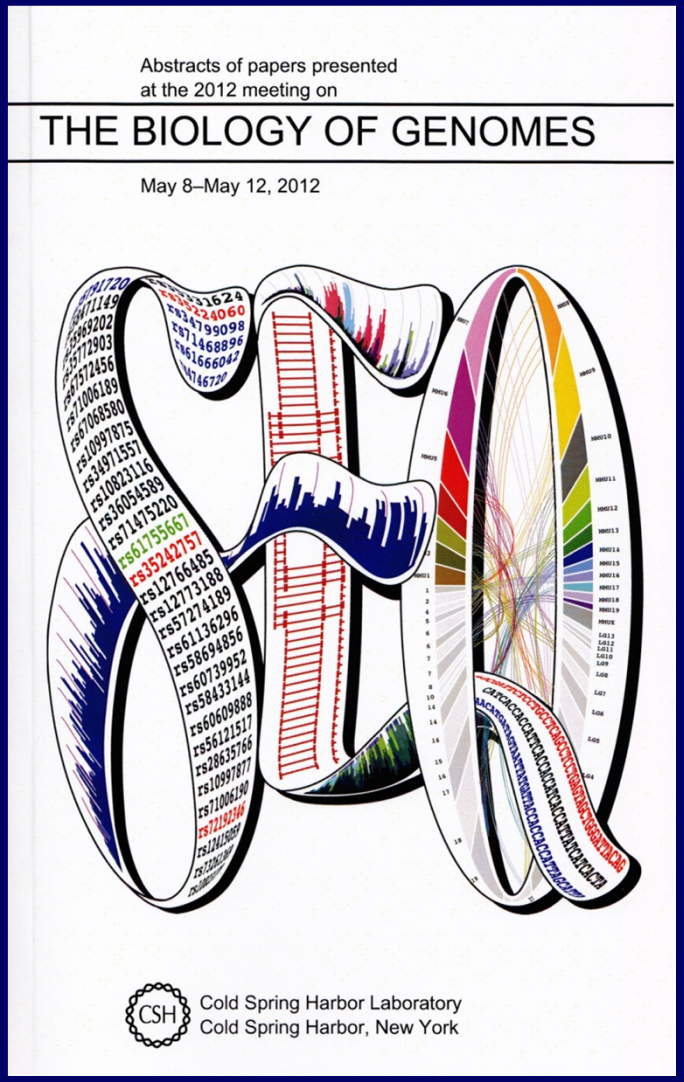
PMB 221,738 MAIN STREET, WALTHAM,MA

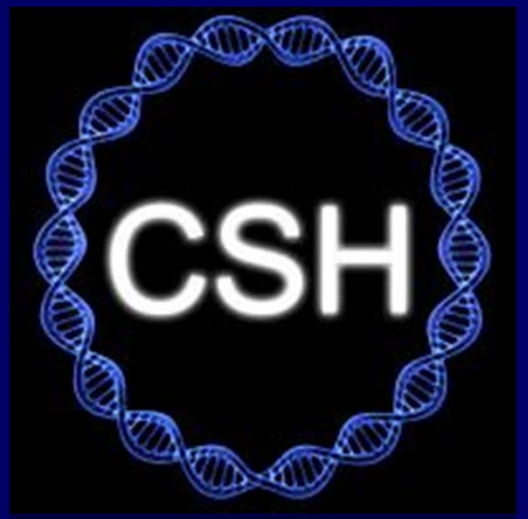
Corp

Site by the ranDimension.

Document 18

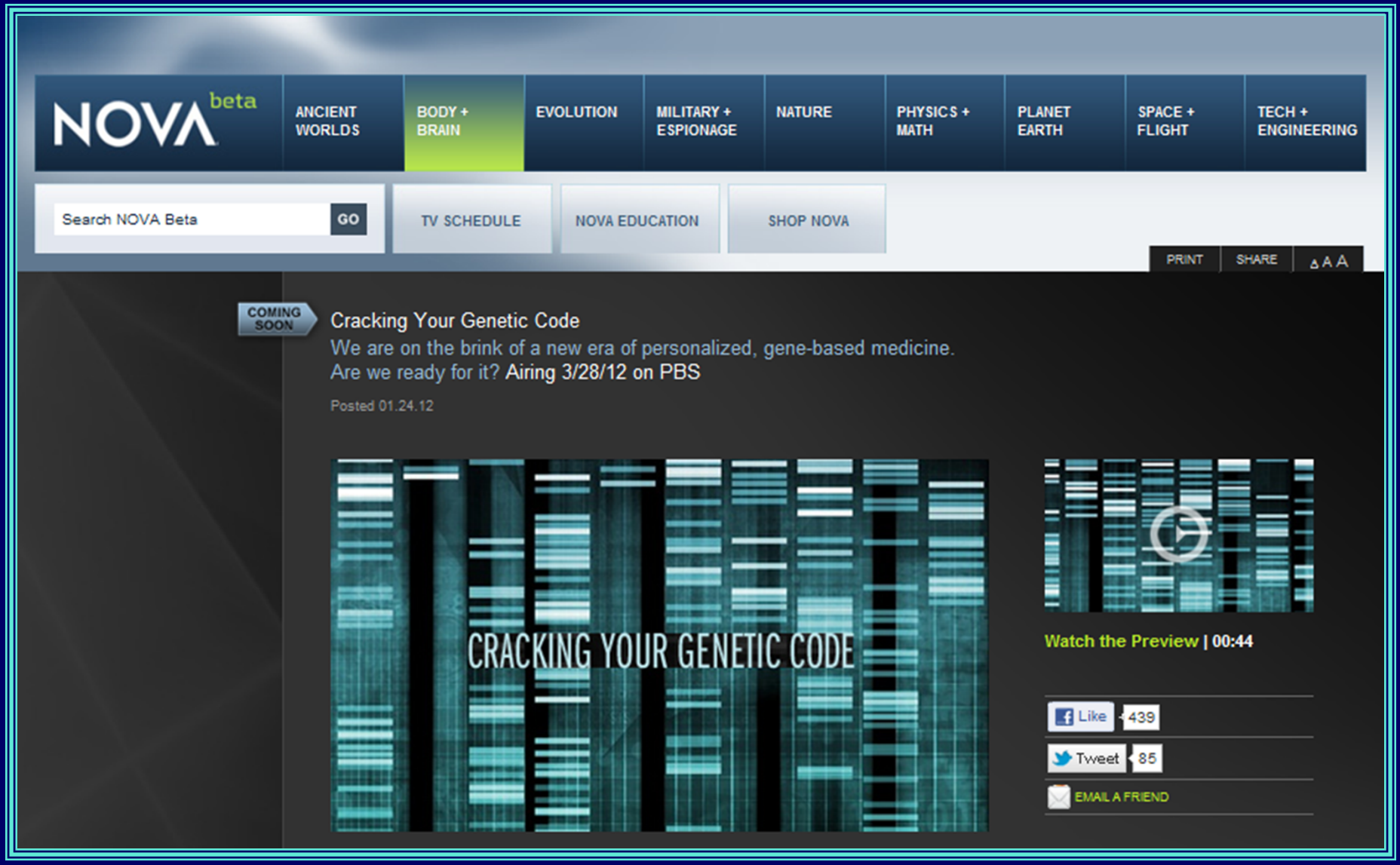
2012 Biology of Genomes Meeting
Cold Spring Harbor Laboratory





Document 19

NOVA: “Cracking Your Genetic Code”



Document 20

NHGRI Genome Advance of the Month

Harnessing the full 'omics potential of personalized medicine

By Danielle Daee, Ph.D.
Postdoctoral Fellow

Discovering the Mutants Among Us

By Joy Yang
Post-baccalaureate Fellow

Last year, the Sanger Institute boldly announced "We are all mutants" when a study was published showing that healthy individuals carry around 60 new mutations from their parents. However, not all of these mutations are meaningful, as some may fall in regions of the genome without any currently known function. The next Genome Advance of the Month focuses on a particular class of mutations, "loss-of-function (LoF) variants."

LoF is defined as a genetic variant that is predicted to cause a loss of function in protein-coding genes; in other words, some change in the genome sequence that prevents the production of a normal protein. Because LoF variants can result in debilitating diseases such as cystic fibrosis and Duchenne muscular dystrophy, they are usually thought to be rare; however, many scientists suspect that LoF variants may actually be quite common, even among healthy people. In fact, the first few whole genome sequences produced following the Human Genome Project each contained several hundred LoF variants in apparently normal individuals.

Genomics In The News…

New insights on evolution from the study of sticklebacks

Insights about human evolution from the gorilla genome sequence

Document 22

Genomics In The News…

The New York Times

Research

WORLD | U.S. | N.Y. / REGION | BUSINESS | TECHNOLOGY | SCIENCE | HEALTH | SPORTS | OPINION

Scientists Link Gene Mutation to Autism Risk

Document 22

Genomics In The News…

Document 22

Genomics In The News…

N.Y. Preschool Starts DNA Testing For Admission

by NPR STAFF

Listen to the Story
All Things Considered [3 min 41 sec]

Add to Playlist
Download
Transcript

April Fools

iStockphoto.com

At the Porsafillo Preschool Academy, there are 32 spots but more than 12,000 applications.

their children.

The preschool is housed in a modern glass and steel building designed by IM Pei. It's situated in a leafy corner of the Upper West Side. On a recent afternoon, Headmaster Rebecca Unsinn showed off "Porsafillo Pre," as it's called.

"Over here, we have computer labs, C++ learning, which of course, as I'm sure you know, is a language of computers," she says. Wait, computer language? These preschoolers are learning C++?

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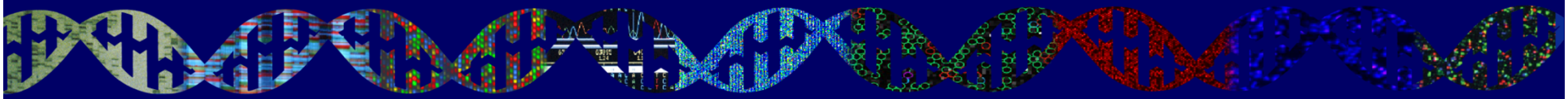


NHGRI Genome Sequencing Program

- Large-Scale Genome Sequencing Centers
- Mendelian Disorders Genome Centers
- Clinical Sequencing Exploratory Research Projects
- Informatics Tools for High-Throughput Sequence Data Analysis

- Meeting involving all components: October 2012

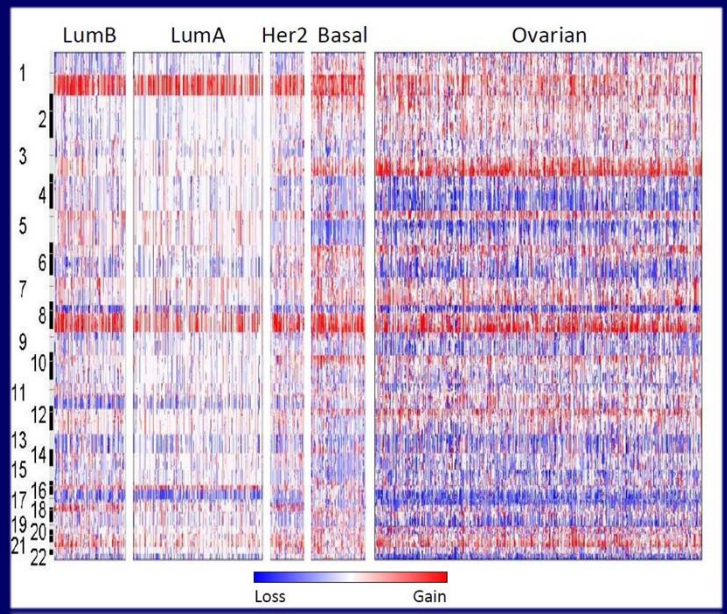
Document 23

Large-Scale Genome Sequencing Centers

- New: Alzheimer's disease sequencing project
- Recent Publications:
  - Autism Sequencing Consortium
  - Schizophrenia allele spectrum
  - Six Cancer genomics papers
  - Gorilla genome
  - Macaque Y chromosome
  - Stickleback adaptive evolution
  - Drosophila population genomics

Document 23

- **TGCA papers on three cancers (in press or under review):**
  - **Colorectal Carcinoma**
  - **Breast Carcinoma**
  - **Lung Squamous Cell Carcinoma**

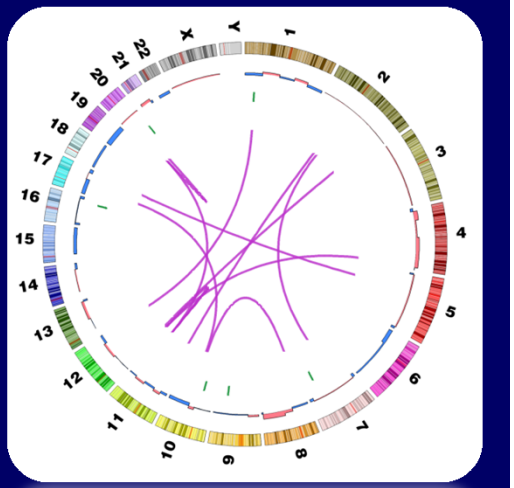


- **Each paper reports the comprehensive integrative analyses of genome sequences for 100's of tumors**

- **Several additional manuscripts are anticipated in 2012**

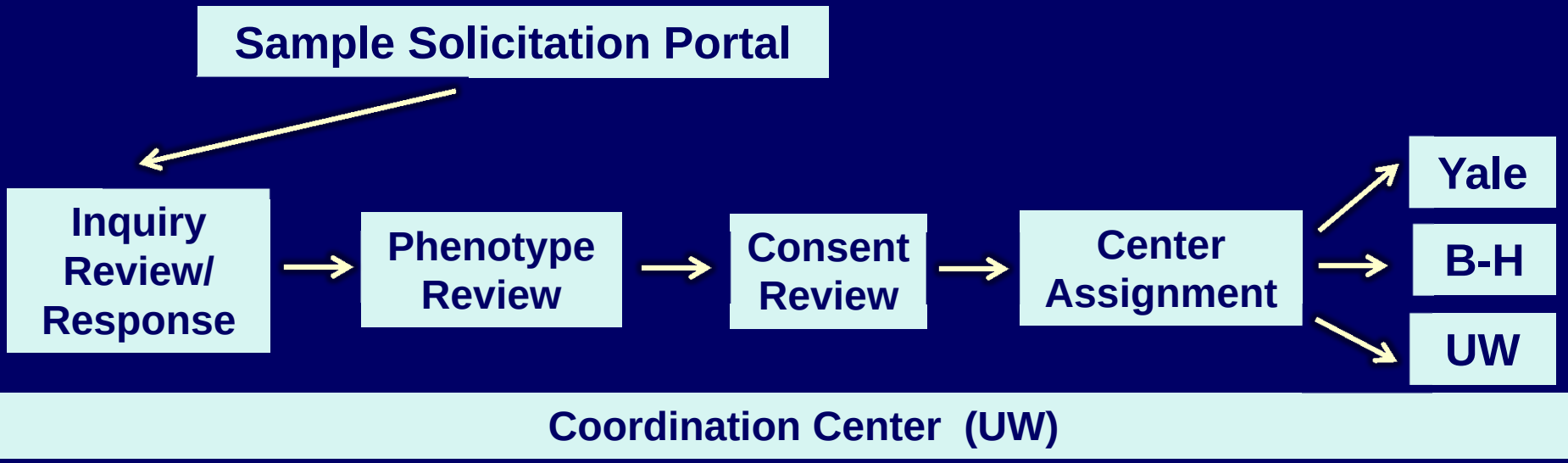
- **CGHub (TCGA sequence data repository) opens at UCSC**
  - **New innovations (e.g., BAM slicing) to be implemented soon**
  - **First 'NIH Trusted Partner' for redistribution of genome sequence data**

- **>6,000 tumors analyzed from 20 cancer types; on track to achieve program goals by 2014 target**



AGTC
Centers for Mendelian Genomics

- Implemented pipeline from the Program's sample solicitation portal through sample assignment to the Centers



- Educational program on Mendelian Genomics at the 2012 Annual ASHG Meeting
- Disease gene discovery is ongoing at various stages from sequencing to the identification of disease genes

Document 23

Clinical Sequencing Exploratory Research (CSER) Projects

- First meeting of the CSER Steering Committee and the Return of Results Consortium

- New RFAs released:
  - Reissue of original CSER solicitation
  - CSER Coordinating Center

Document 23

Informatics Tools for High-Throughput Sequence Data Analysis

- "iSeqTools" Network aims to develop robust and reliable analysis tools for researchers without specialized computational skills

- Leveraging NHGRI's investment in sequencing centers and initiatives (e.g., 1000 Genomes, Galaxy, and others)

- Building iSeqTools wiki as a knowledgebase

DNA Sequencing Technology Development

Grantee Meeting (April 2012)

Document 25

DNA Sequencing Technology Development

LETTERS

nature biotechnology

Automated forward and reverse ratcheting of DNA in a nanopore at 5-Å precision

Gerald M Cherf, Kate R Lieberman, Hytham Rashid, Christopher E Lam, Kevin Karplus & Mark Akeson

An emerging DNA sequencing technique uses protein or solid-state pores to analyze individual strands as they are driven in single-file order past a nanoscale sensor[1–3]. However, uncontrolled electrophoresis of DNA through these nanopores is too fast for accurate base reads[4]. Here, we describe forward and reverse ratcheting of DNA templates through the α-hemolysin nanopore controlled by phi29 DNA polymerase without the need for active voltage control. DNA strands were ratcheted through the pore at median rates of 2.5–40 nucleotides per second and were examined at one nucleotide spatial precision in real time. Up to 500 molecules were processed at ~130 molecules per hour through one pore. The probability of a registry error (an insertion or deletion)

through the nanopore. This event results in a brief current blockade that is influenced by DNA strand length[1] and base composition[3].

A consensus has emerged that the average rate of ssDNA electrophoresis through nanopores (~3 μs nt^{-1} at 120 mV for α-HL) is too fast to allow bases to be accurately identified[4]. Therefore, a functional nanopore sequencing device will require a means to systematically slow DNA template movement. One proposed strategy involves coupling an enzyme motor to the nanopore[8]. This strategy is attractive because many processive enzymes, including polymerases, ratchet along DNA strands one nucleotide at a time, up to tens of thousands of times in succession in bulk phase[9]. Systematic, enzyme-driven displacement of a captured DNA strand relative to the nanopore would be anticipated at millisec-

LETTERS

nature biotechnology

Reading DNA at single-nucleotide resolution with a mutant MspA nanopore and phi29 DNA polymerase

Elizabeth A Manrao[1], Ian M Derrington[1], Andrew H Laszlo[1], Kyle W Langford[1], Matthew K Hopper[1], Nathaniel Gillgren[1], Mikhail Pavlenok[2], Michael Niederweis[2] & Jens H Gundlach[1]

Nanopore technologies are being developed for fast and direct sequencing of single DNA molecules through detection of ionic current modulations as DNA passes through a pore's constriction[1,2]. Here we demonstrate the ability to resolve changes in current that correspond to a known DNA sequence by combining the high sensitivity of a mutated form of the protein pore *Mycobacterium smegmatis* porin A (MspA)[3] with phi29 DNA polymerase (DNAP)[4], which controls the rate of DNA translocation through the pore. As phi29 DNAP synthesizes DNA and functions like a motor to pull a single-stranded template through MspA, we observe well-resolved and reproducible ionic current levels with median durations of ~28 ms and ionic current differences of up to 40 pA. Using six different DNA sequences with readable regions 42–53 nucleotides long, we record current traces that map to the known DNA sequences. With single-nucleotide resolution and DNA translocation control, this system integrates solutions to two long-standing hurdles to nanopore sequencing[2].

nanopore sequencing because it has a short and narrow constriction ~1.2 nm wide and ~0.6 nm long[12] (**Fig. 1a**). Thus, the ionic current through MspA is affected by a smaller number of nucleotides compared with other pores[3,7,9]. Previously, we engineered mutants of MspA by replacing negative charges in the constriction with neutral residues, which enabled DNA to electrophoretically pass through the pore[13]. We also added 24 positively charged residues in the vestibule and entrance, which enhanced the rate of entry of DNA into the pore[13]. This mutant, previously called M2-NNN MspA, is used in the present study and is here designated MspA. When DNA was held statically in the constriction of a mutant MspA by a conjugated NeutrAvidin molecule[3], different homopolymer strands resulted in conductance differences of as much as ~0.23 nS, nearly ten times more separation than that observed with the widely used α-hemolysin nanopore (~0.028 nS)[7,8]. However, this was not sufficient for nanopore sequencing because freely translocating DNA acting under the force of an electric field moves through nanopores at an average rate greater than one nucleotide/μs, ~1,000 times too fast to distinguish nucleotide-specific current changes from noise[1,2,13,14]. Simple techniques for reducing the velocity of translocating DNA, such

Document 25

DNA Sequencing Technology Development

Branton, Deamer, Church

Gundlach

Akeson

Manrao

Cherf

Derrington

Lazlo

Nanopore sequencing demonstrated

ENCODE & modENCODE

- ENCODE Technology Development awards funded this spring
- modENCODE Symposium: June 20-21, 2012
- Integrative analysis papers planned:

  ENCODE – integrative manuscript submitted along with many companion papers

  modENCODE/ENCODE – comparison of fly, worm, and human

  Mouse – comparison of mouse and human

ELSI Program

- New RFAs for Centers of Excellence in ELSI Research (CEER) Program: P20 and P50
- Return of Results Consortium launched
- April 2012 issue of *Genetics in Medicine* focused on return of results and incidental findings, featuring multiple papers by ELSI-funded investigators

Document 27

Upcoming Planning Meeting

- **July 2012 Workshop:**

  Integrating Functional Data for Connecting Genotype to Phenotype

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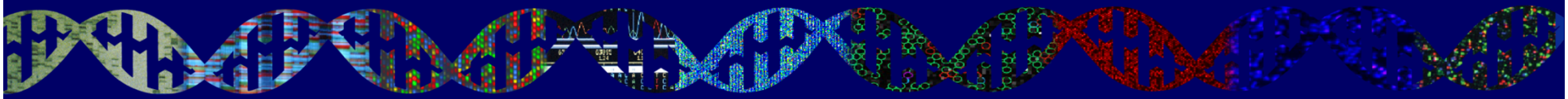


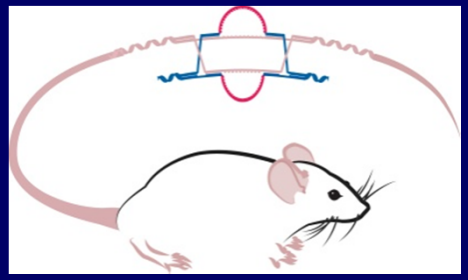
Human Microbiome Project (HMP)

- HMP funding ends in FY2012
- Two major HMP Consortium papers to be published in *Nature*; coordinated release of >20 companion papers in *PLoS* ('HMP Collection')
- HMP2 proposal submitted to Common Fund
- Report at September Council meeting

Document 28

Knockout Mouse Phenotyping Project ($KOMP^2$)

- In 5 years, make 2,500 live mouse strains from knockout ES cells
- Comprehensively phenotype the mouse strains



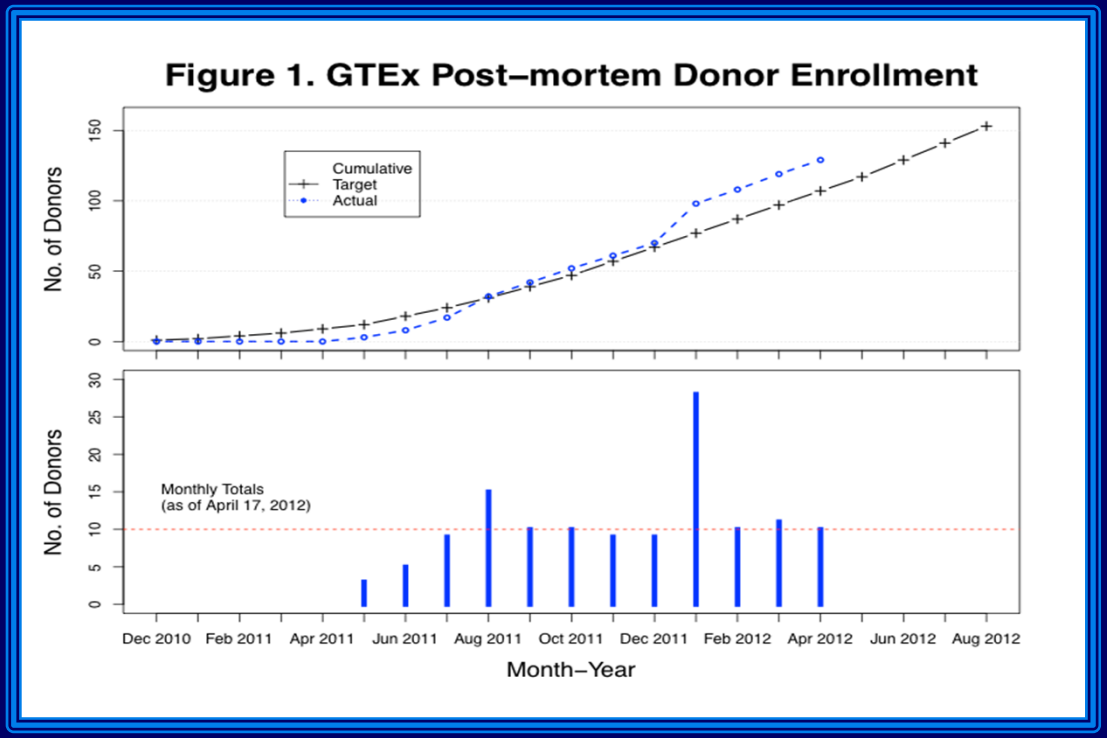
IMPC

- Make data and mice available to researchers
- Collaborate with other international projects to achieve a total of 5,000 phenotyped strains through the IMPC

Document 29

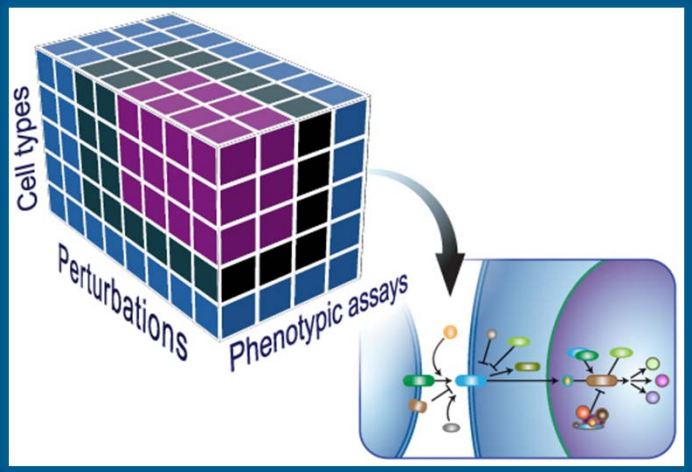
Genotype-Tissue Expression (GTEx)

- Pilot goals have been met
- Scale-up is under consideration
- First dbGaP data release later this month



Document 30

Library of Integrated Network-based Cellular Signatures (LINCS)

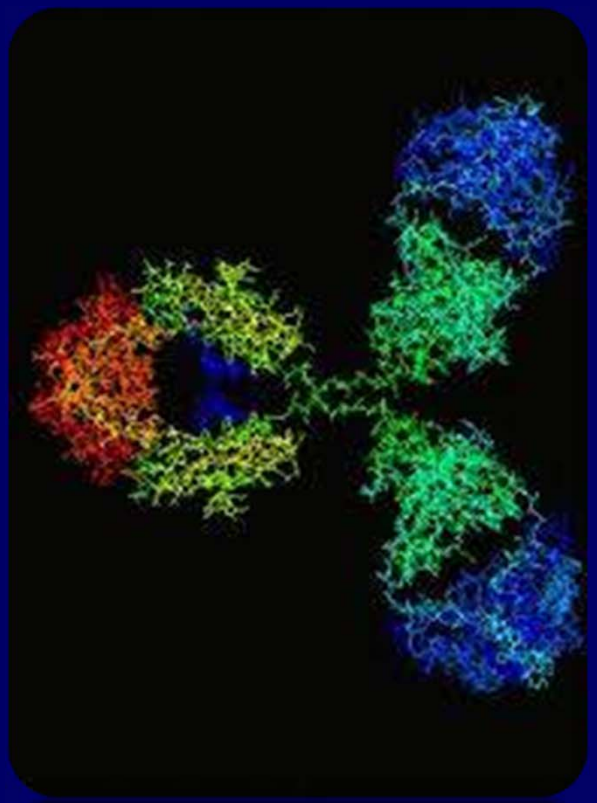


- Consortium meeting in November 2012
- Request to the Common Fund for a 1-year extension of pilot (FY2013 bridge funds)
- Quarterly public release of LINCS data and metadata (started March 2012)

Document 31

Protein Capture Reagents Program

- New working groups:
  - Data dissemination
  - Validation
  - Target list prioritization
- Soliciting community input about production of human transcription factor reagents



Human Heredity and Health in Africa (H3Africa)

- Applications reviewed in March and April
- A funding plan will be discussed in Closed Session of this Council meeting
- Inaugural H3Africa Research Network meeting will be held in Ethiopia in October 2012

Document 33

Single Cell Analysis

- **Three RFAs:**
  - **Evaluate cellular heterogeneity using transcriptional profiling**
  - **Innovative tools and technologies**
  - **Single cell technology validation in clinical setting**

- **Single Cell Analysis workshop recently held**

Document 34

I. General NHGRI Updates

II. General NIH Updates

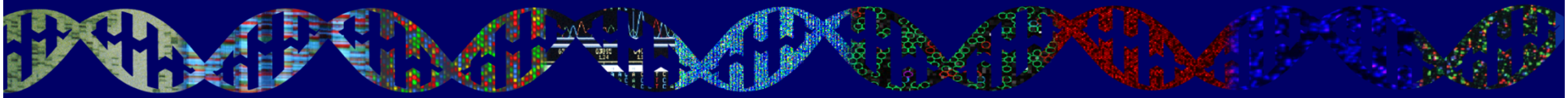
III. Genomics Updates

IV. NHGRI Extramural Program

V. NIH Common Fund Programs

VI. NHGRI Office of the Director

VII. NHGRI Intramural Program



The eMERGE Network

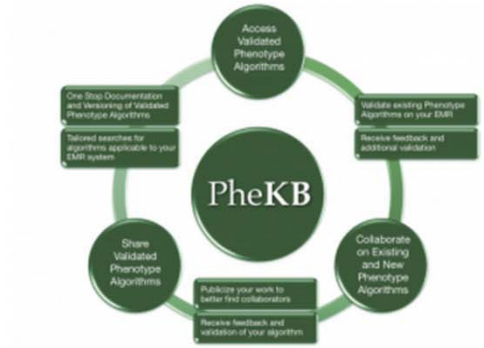
electronic Medical Records & Genomics

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

PheKB a knowledgebase for discovering phenotypes from electronic medical records

Home | Phenotypes | Implementations | Groups | Institutions

What is the Phenotype KnowledgeBase?



The reuse of data from electronic medical records (EMRs) and other clinical data systems holds tremendous promise for improving the efficiency and effectiveness of health research. Clinical data in the EMR is a potential source of rich longitudinal data for research, and the recent government efforts to promote the use of EMRs in the clinical setting may further promote the use of such systems in the US healthcare system. As the use of EMRs expands, the demand for usable data from these systems for research has also expanded.

One such effort by the Electronic Medical Records and Genomics Network (eMERGE) has investigated whether data captured through routine clinical care using EMRs can identify disease phenotypes with sufficient positive and negative predictive values for use in genome-wide association studies (GWAS). Most EMRs captured key information (diagnoses, medications, laboratory tests) used to define phenotypes in a structured format; in addition, natural language processing has also been shown to improve case identification rates.*

- **Purpose: To provide a collaborative environment for building and validating electronic phenotype algorithms**

Document 35

eMERGE Testimony to the National Committee on Vital and Health Statistics

- Data collection: community engagement models, community advisory boards, and promotion of research to the community
- Data utilization: data use agreement for sharing data in the consortium
- Data dissemination: de-identified data submitted to dbGaP for sharing data beyond the eMERGE sites

NCVHS

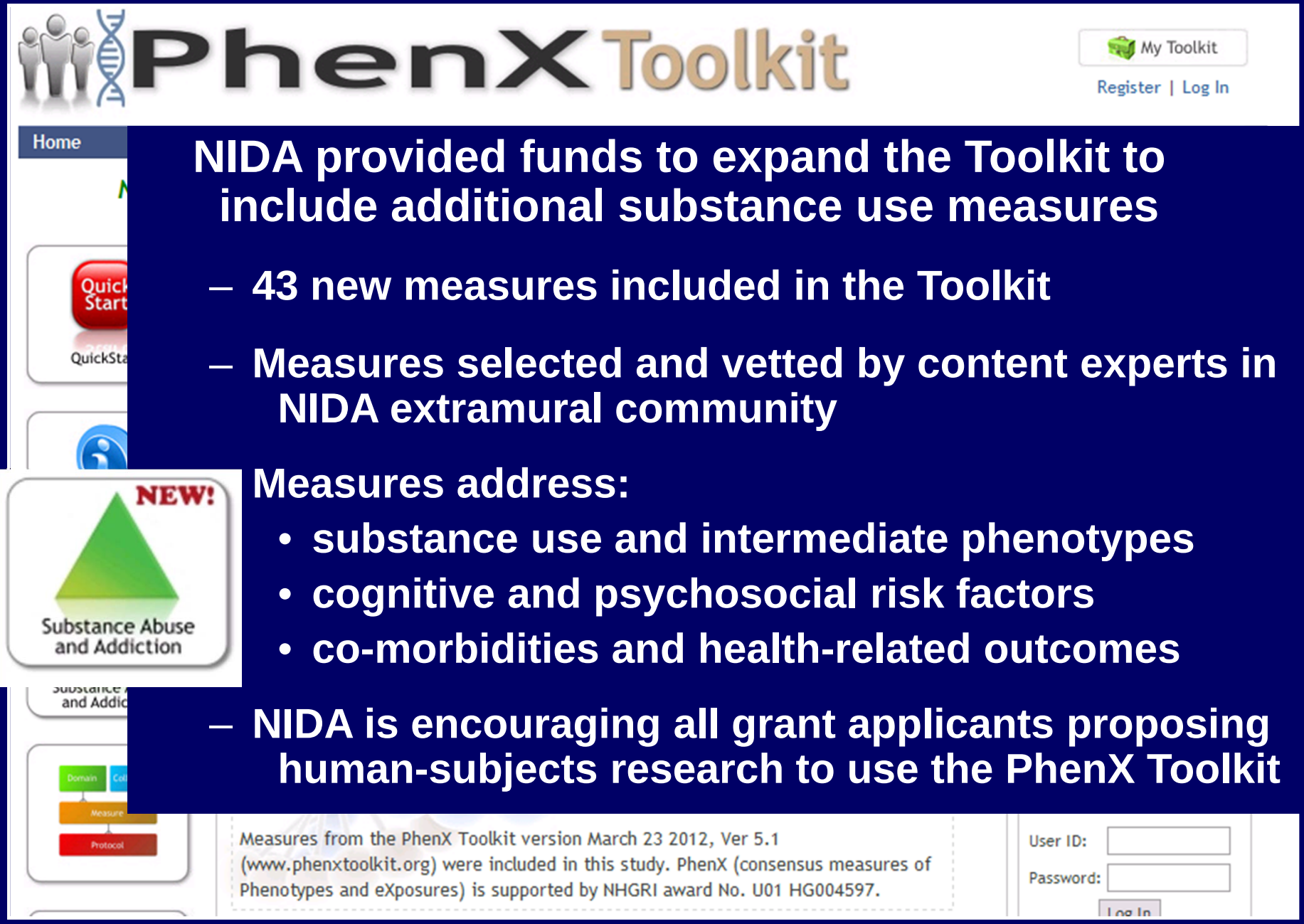
GENEVA Initiative Complete

- 20 datasets posted to dbGaP, all imputed

Addiction	Glaucoma	Oral Clefts
Blood Clotting	Heart Disease	Prematurity
Blood Pressure	Lung Cancer	Prostate Cancer
COPD	Maternal Metabolism	Stroke
Dental Carries	Melanoma	Venous Thombosis
Diabetes		

- 50+ publications
- GWASTools website developed

GENE.ENVIRONMENT ASSOCIATION.STUDIES GENEVA



NIDA provided funds to expand the Toolkit to include additional substance use measures

- **43 new measures included in the Toolkit**
- **Measures selected and vetted by content experts in NIDA extramural community**
- **Measures address:**
  - **substance use and intermediate phenotypes**
  - **cognitive and psychosocial risk factors**
  - **co-morbidities and health-related outcomes**
- **NIDA is encouraging all grant applicants proposing human-subjects research to use the PhenX Toolkit**

NEJM Genomic Medicine Series

Genomics, Intellectual Disability, and Autism

Heather C. Mefford, M.D., Ph.D., Mark L. Batshaw, M.D., and Eric P. Hoffman, Ph.D.

INTELLECTUAL DISABILITY, WHICH IS CHARACTERIZED BY SIGNIFICANT LIM-
itations in both i
the age of 18 yea
A diagnosis of intelle
less than 70, which
or early adulthood. H
early in childhood o
include motor, cogni
has long been recog
detectable by chrom
portant chromosom
been identified for a
autosomal and X-lin
of inherited syndron
male patients.

Realizing Genomic Medicine

Elizabeth G. Phimister, Ph.D., W. Gregory Feero, M.D., Ph.D., and Alan E. Guttmacher, M.D.

The current series of Genomic Medicine review articles concludes in this issue of the *Journal* with the publication of an article on cognitive impairment and autism by Mefford and colleagues.[1] The topic of this article is an appropriate capstone for the Genomic Medicine series: it highlights the clinical advances in genomics regarding the care of patients with neurologic conditions, and it shows the potential of genomic science to further accelerate the pace of discovery in the neurosciences.

solved. Their resolution will be critical to realizing the full benefit of genomic advances. Central to some of these issues is the boundary between research and clinical care, as defined in the Belmont Report produced by the National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research and the "Common Rule" that governs much of federally funded biomedical research in the United States.[2,3]

Making clinical use of the vast complexity of

Document 38

2012 DNA Day Chat Room

- April 20, 8:00 am to 5:00 pm EST
- More than 70 experts answered questions
- Received more than 900 questions and experts answered 764 of the questions
- Questions from 37 states and internationally

DNA DAY APRIL 20 '12

Document 39

USA Science and Engineering Festival

Document 40

USA Science and Engineering Festival

GENOMICS in Medicine Lecture Series

First Friday of each month, 8-9 AM

Suburban Hospital Auditorium

June 1, Barbara Biesecker, NHGRI

Genomics in Maternal Child Health

Invited Speakers: July 2012 through January 2013

Paul Sieving, NEI
Dan Kastner, NHGRI
Kenneth Fishbeck, NINDS
Ellen Sidransky, NHGRI
Max Muenke, NHGRI

JOHNS HOPKINS MEDICINE

NATIONAL INSTITUTES OF HEALTH

National Human Genome Research Institute

DEPARTMENT OF HEALTH & HUMAN SERVICES · USA

SUBURBAN HOSPITAL JOHNS HOPKINS MEDICINE

Document 41

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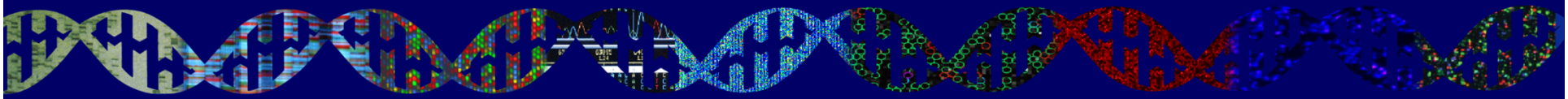
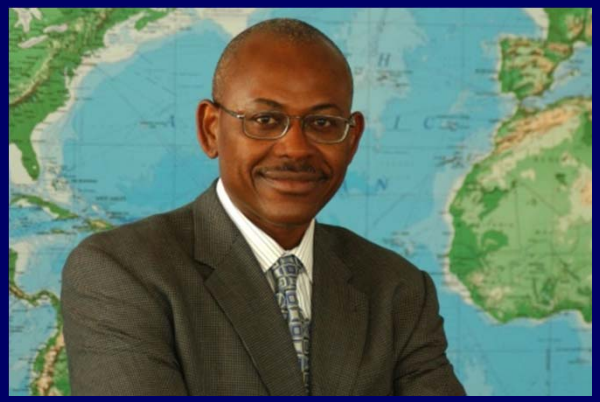


NHGRI Intramural Research Highlights



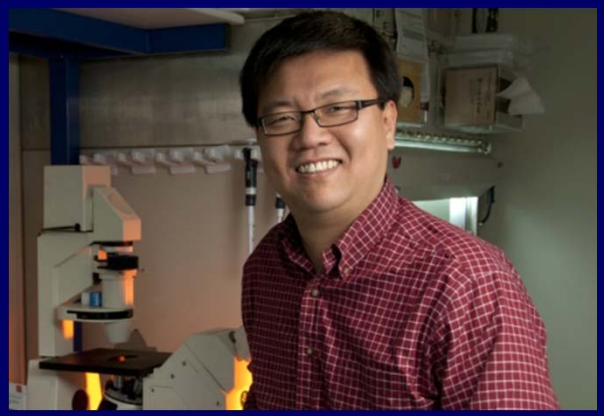
The NEW ENGLAND JOURNAL of MEDICINE

ORIGINAL ARTICLE

HLA Class II Locus and Susceptibility to Podoconiosis

Fasil Tekola Ayele, Ph.D., M.P.H., Adebowale Adeyemo, M.D., Chris Finan, Ph.D., Elena Hailu, M.Sc., Paul Sinnott, Ph.D., Natalia Diaz Burlinson, M.Sc., Abraham Aseffa, M.D., Ph.D., Charles N. Rotimi, Ph.D., M.P.H., Melanie J. Newport, M.D., Ph.D., and Gail Davey, M.D.

N Engl J Med 2012; 366:1200-1208 | March 29, 2012



PNAS

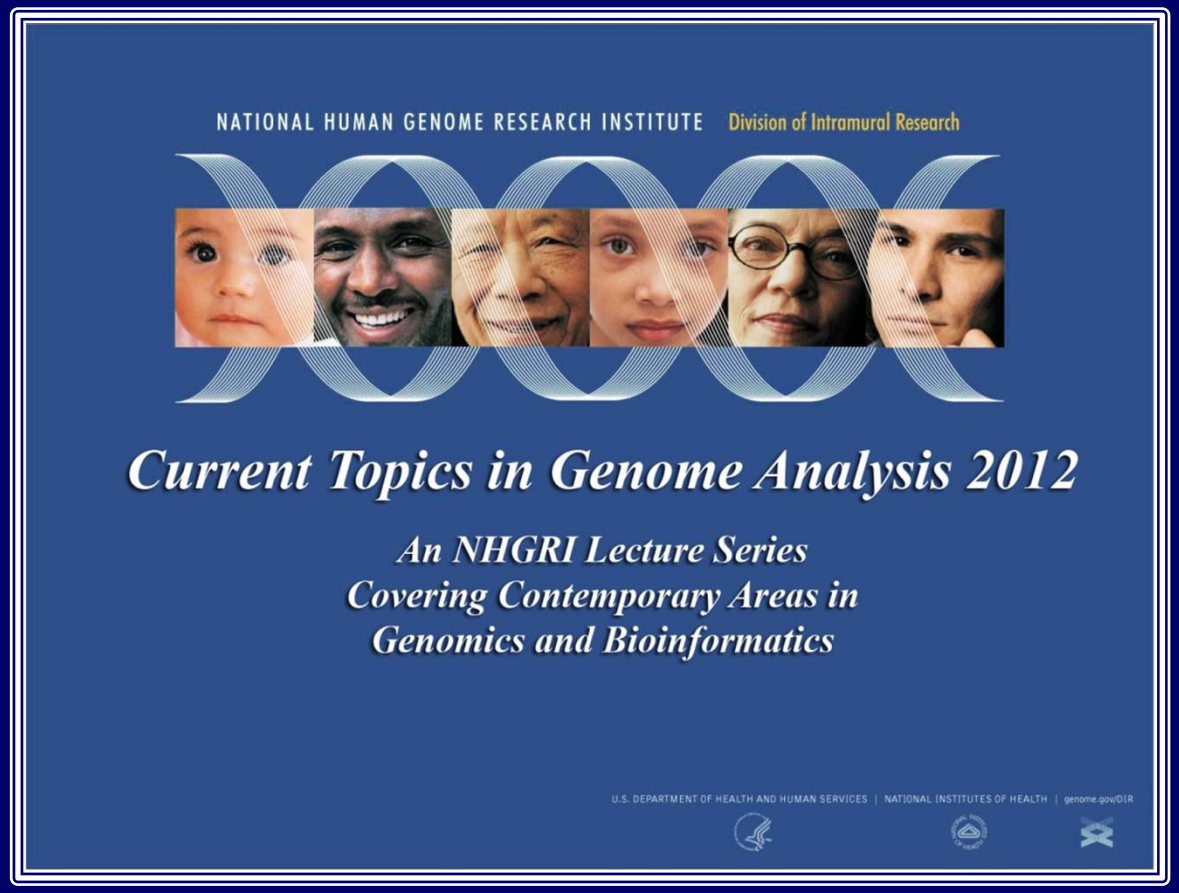
Proceedings of the National Academy of Sciences of the United States of America

High-throughput genotoxicity assay identifies antioxidants as inducers of DNA damage response and cell death

Jennifer T. Fox[a], Srilatha Sakamuru[b], Ruili Huang[b], Nedelina Teneva[a], Steven O. Simmons[c], Menghang Xia[b], Raymond R. Tice[d], Christopher P. Austin[b], and Kyungjae Myung[a,1]

Document 42

Current Topics in Genome Analysis



- **Views to date for 2012 Series: 22,852**
- **Views for 2010 Series: 202,812**

Document 43

Blue Ribbon Panel Review of NHGRI Intramural Research Program

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

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

Special Thanks!


genome.gov
THE BRIGHT FUTURE OF HUMAN GENOMICS