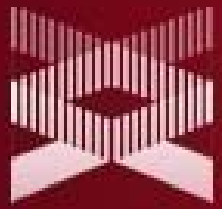


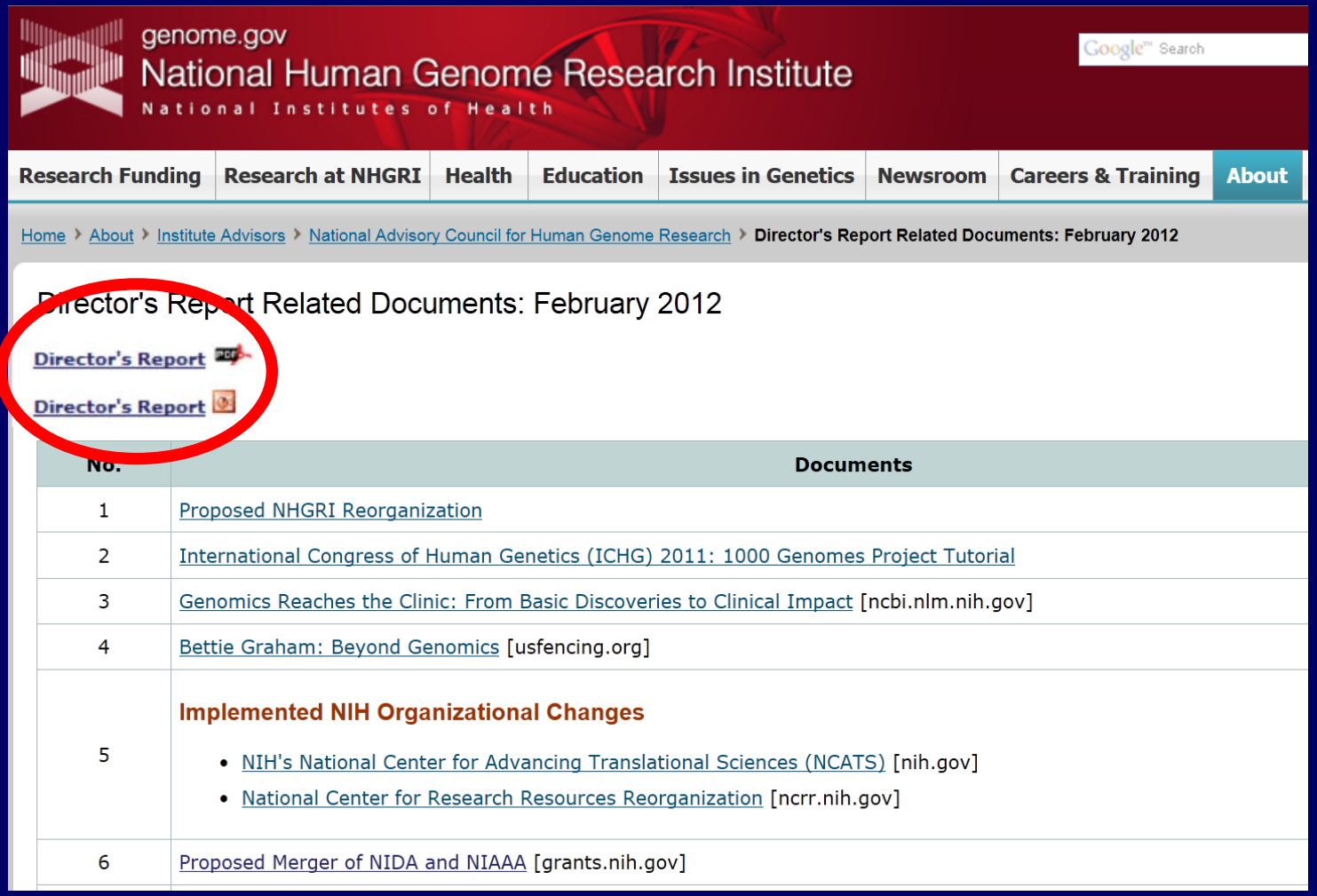
genome.gov
National Human Genome Research Institute
National Institutes of Health

DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

February 2012

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

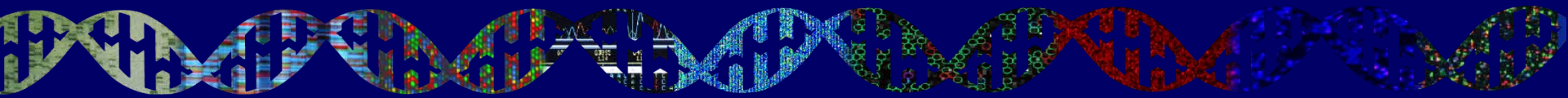


genome.gov/DirectorsReport

Document #

Open Session Presentations

- DNA Sequencing Technology 2012
  - Jeff Schloss
- Genomics and Society: The ELSI Research Program and Beyond
  - Karen Rothenberg
- NHGRI Extramural Portfolio Review
  - Mark Guyer



Open Session Presentations

Concept Clearances:

- PAGE Renewal
  - Lucia Hindorff
- Clinical Exploratory Sequencing Coordinating Center
  - Lucia Hindorff
- Genomic Medicine Pilot Projects
  - Teri Manolio

Open Session Presentations

Concept Clearances:

- ClinAction
  - Teri Manolio
- Centers of Excellence in ELSI Research
  - Joy Boyer
- High-Throughput Genomic Analysis in Children with Newborn Screening Disorders
  - Anastasia Wise

Open Session Presentations

Meeting Report:

- **Genomic Literacy**
  - **Vence Bonham**

I. General NHGRI Updates

II. General NIH Updates

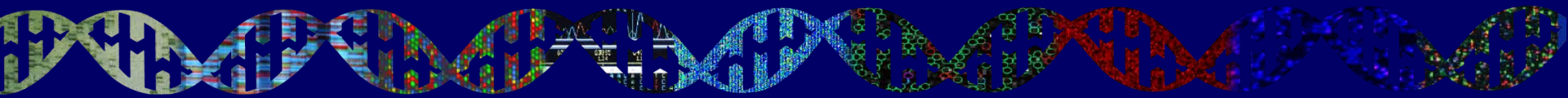
III. Genomics Updates

IV. NHGRI Extramural Program

V. NIH Common Fund Programs

VI. NHGRI Office of the Director

VII. NHGRI Intramural Program



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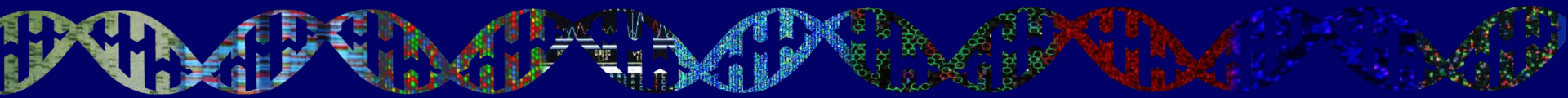


Proposed NHGRI Reorganization

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* PDF). 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.

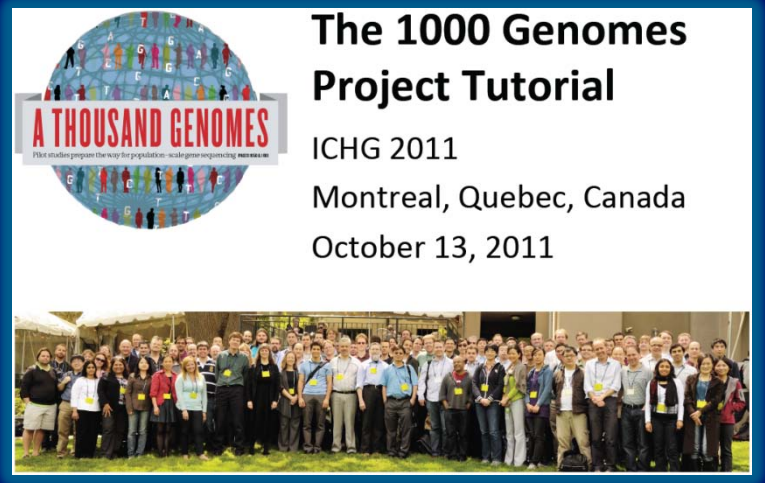
genome.gov/reorg

Document 1

NHGRI @ ICHG/ASHG

- **ELSI Concurrent Invited Session: "Emerging Ethical Issues in Large-Scale International Genomics Research Collaborations"**

- **1000 Genomes Meeting & Tutorial**



Document 2

Cell Commentary on Clinical Genomics

Cell

Leading Edge

Commentary

Genomics Reaches the Clinic: From Basic Discoveries to Clinical Impact

Teri A. Manolio[1] and Eric D. Green[1,*]
[1]National Human Genome Research Institute, National Institutes of Health, Bethesda, MD 20892, USA
*Correspondence: egreen@nhgri.nih.gov
DOI 10.1016/j.cell.2011.09.012

Today, more than ever, basic science research provides significant opportunities to advance our understanding about the genetic basis of human disease. Close interactions among laboratory, computational, and clinical research communities will be crucial to ensure that genomic discoveries advance medical science and, ultimately, improve human health.

Document 3

Bettie Graham: Beyond Genomics

Document 4

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II. General NIH Updates

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V. NIH Common Fund Programs

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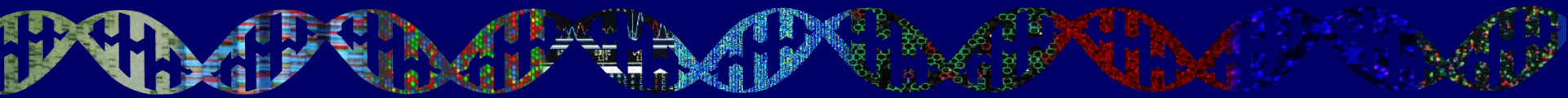
VII. NHGRI Intramural Program



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

NCATS

National Center for Advancing Translational Sciences

NCATS News

Frequently Asked Questions

Related Links

NIH Launches National Center for Advancing Translational Sciences

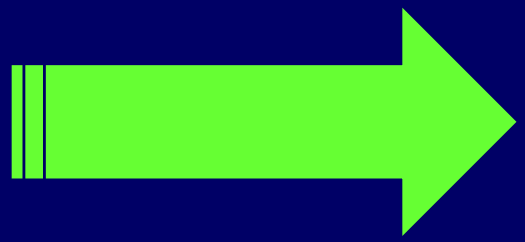
December 23, 2011

President Barack Obama has signed the Fiscal Year 2012 spending bill enabling the National Institutes of Health to establish the new National Center for Advancing Translational Sciences (NCATS). This action marks a major milestone in efforts to revolutionize the science of translation. NCATS provides our nation with an opportunity to forge a new paradigm for translational research that involves government, academia, industry, philanthropy, and patient advocacy groups. Through partnerships that capitalize upon our respective strengths, I believe we can work together to achieve our common goal: speeding the movement of scientific discoveries from the lab to patients.

Francis S. Collins, M.D., Ph.D.
Director, National Institutes of Health

Document 5

Relocation of Programs from the National Center for Research Resources



NCATS
DPCPSI
NIBIB
NIGMS
NIMHD
NIDDK
NHLBI

Document 5

Proposed Merger of Institutes

Request for Information (RFI): Input into the Scientific Strategic Plan for the proposed National Institute of Substance Use and Addiction Disorders

Notice Number: NOT-OD-12-045

Key Dates

Release Date: February 8, 2012
Response Date: May 11, 2012

Issued by

National Institutes of Health (NIH)

Purpose

This Notice is a time-sensitive Request for Information (RFI) soliciting input into the Scientific Strategic Plan for the proposed new Institute with the working name of the National Institute of Substance Use and Addiction Disorders. This new Institute would result from the proposed reorganization of substance use, abuse, and addiction-related research at the NIH.

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

Document 6

Target Validation Workshop

Joint NIH-Industry Target Validation Workshop

National Institutes of Health
Building 31, C Wing, Conference Room 10
November 3–4, 2011

Executive Summary

genomeweb

NIH Plots Target Validation Initiative with Industry, Academia

October 20, 2011

NIH Plots Target Validation Initiative with Industry, Academia

By a GenomeWeb staff reporter

NEW YORK (GenomeWeb News) – The National Institutes of Health is planning to work with industry, government, and academia to create a collaborative precompetitive consortium focused on validating potential therapeutic targets using genomics, bioinformatics, and functional validation, according to NIH.

To kick off the initiative, NIH will hold a joint workshop on Nov. 3 and 4 with multiple partners from these sectors to explore the potential opportunities and challenges facing this type of initiative. The larger aim of the project is to develop faster and more accurate ways to realize the potential of translating new discoveries, identify promising targets, and predict which targets will be "biologically relevant and tractable," according to NIH.

Document 7

Appropriations Update

Fiscal Year 2012 Appropriations Update

	Fiscal Year 2011	Fiscal Year 2012 President	Fiscal Year 2012 House	Fiscal Year 2012 Senate	Fiscal Year 2012 Enacted
NIH	$30.7B	$31.7B	$31.7B	$30.5B	**$30. B**
NHGRI	$511M	$525M	$525M	$506M	**$513M (0.27%)**

Document 8

Fiscal Year 2013 Appropriations Update

Panel Fails to Reach Deal on Plan for Deficit Reduction

Brendan Hoffman for The New York Times

Senator Rob Portman of Ohio, a Republican member of the deficit committee, speaking with reporters on Monday after panel members met on Capitol Hill.

Alzheimer's Research Initiative

News Release

FOR IMMEDIATE RELEASE
February 7, 2012

Contact: HHS Press Office
(202) 690-6343

We can't wait: Administration announces new steps to fight Alzheimer's disease

The Obama Administration today announced new efforts to fight Alzheimer's disease, including immediately making an additional $50 million available fo
budget will boost fund
caregiver support, pro

In January 2011, Presi
national Alzheimer's di
brings together some
preliminary framework
disease by 2025. As

"Investments will include research to identify genes that increase the risk of Alzheimer's disease…"

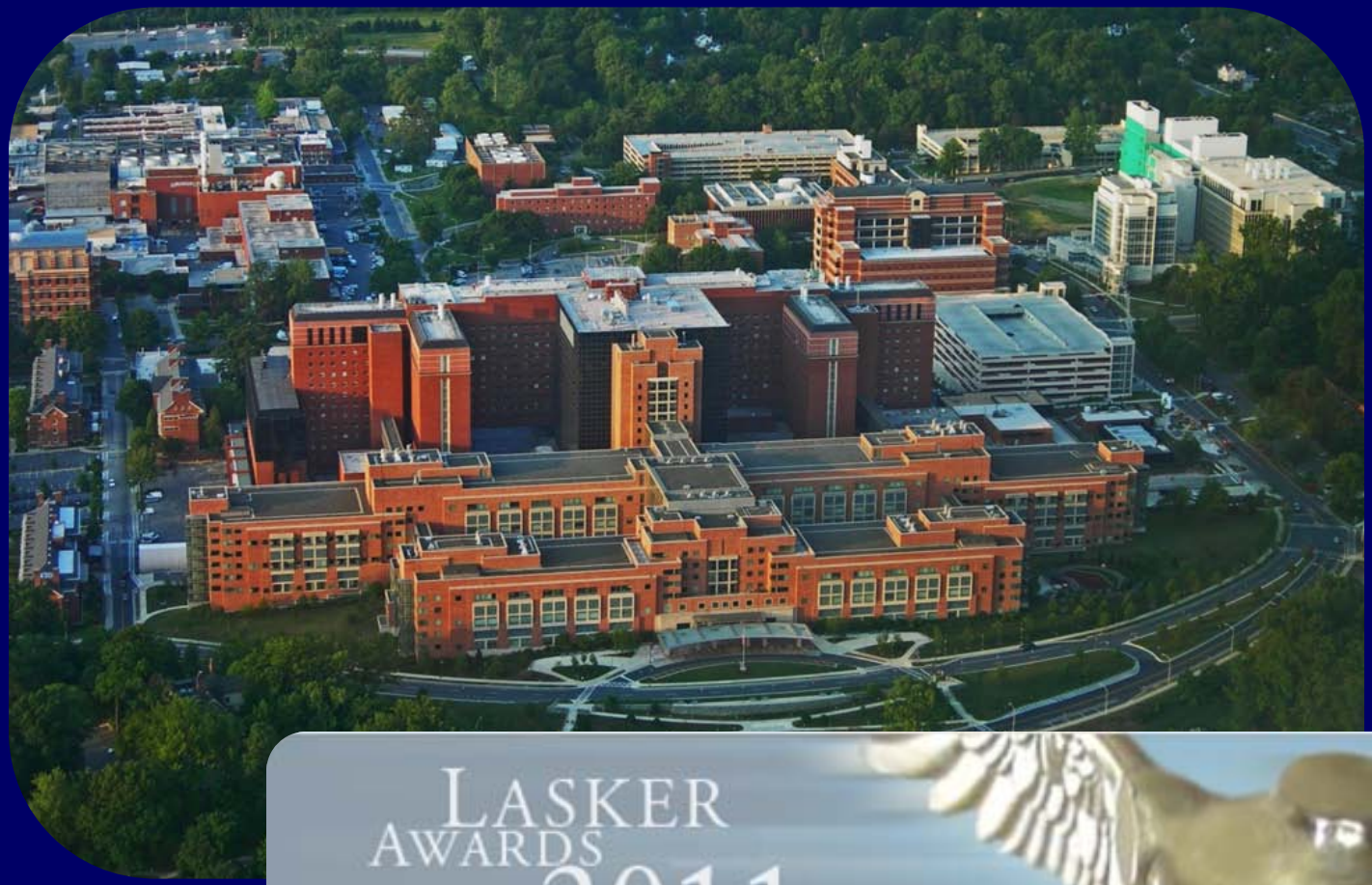
The Washington Post

Back to previous page

Obama administration proposes raise for Alzheimer's research, some now and some next year

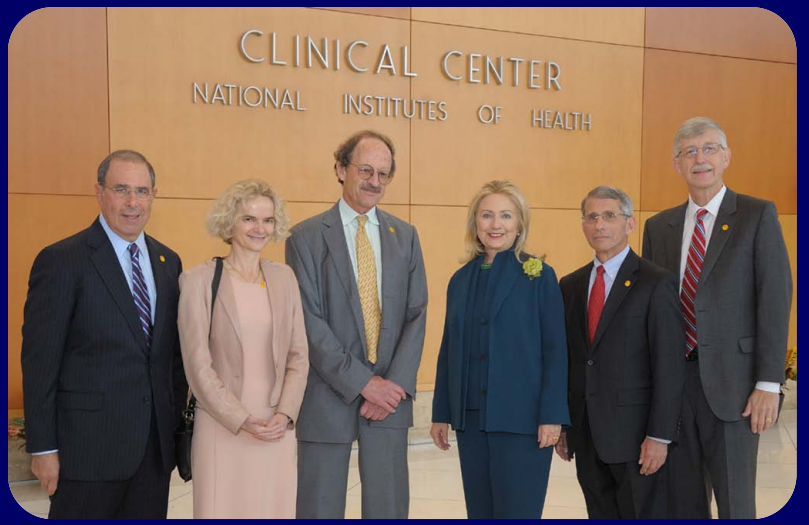
Document 9

NIH Clinical Center Receives Lasker-Bloomberg Public Service Award



Document 10

Secretary of State Hillary Clinton Visits NIH



Document 11

Advisory Committee to the NIH Director Working Group on Data and Informatics

- Working group to investigate the management, integration, and analysis of large biomedical datasets
- Called the NIH Data and Informatics Working Group
- Co-chaired by David DeMets and Larry Tabak

Document 12

Request for Information: NIH Data and Informatics Working Group

- Scope of the issues
- Standards development
- Secondary use of data
- Data accessibility
- Incentives for data sharing
- Support needs

Submit comments by March 12, 2012

Document 12

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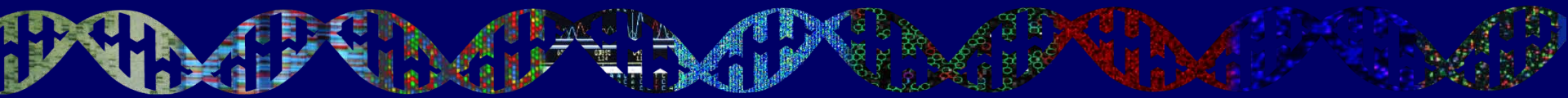
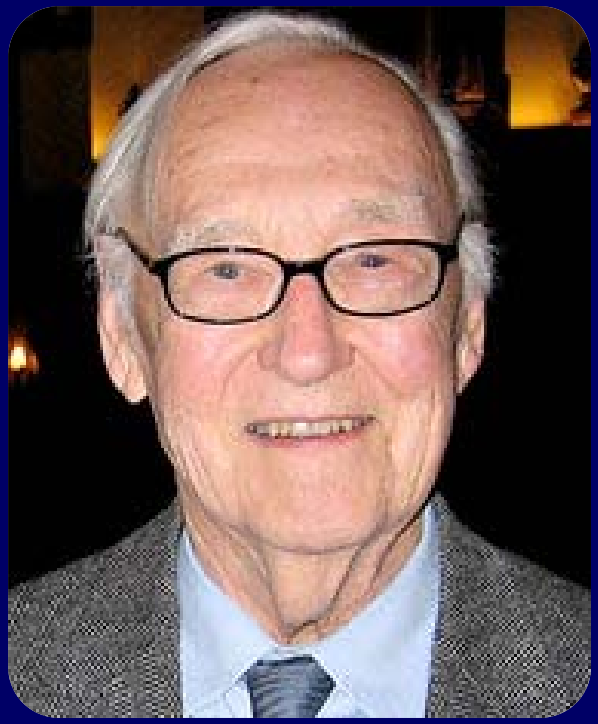
VII. NHGRI Intramural Program

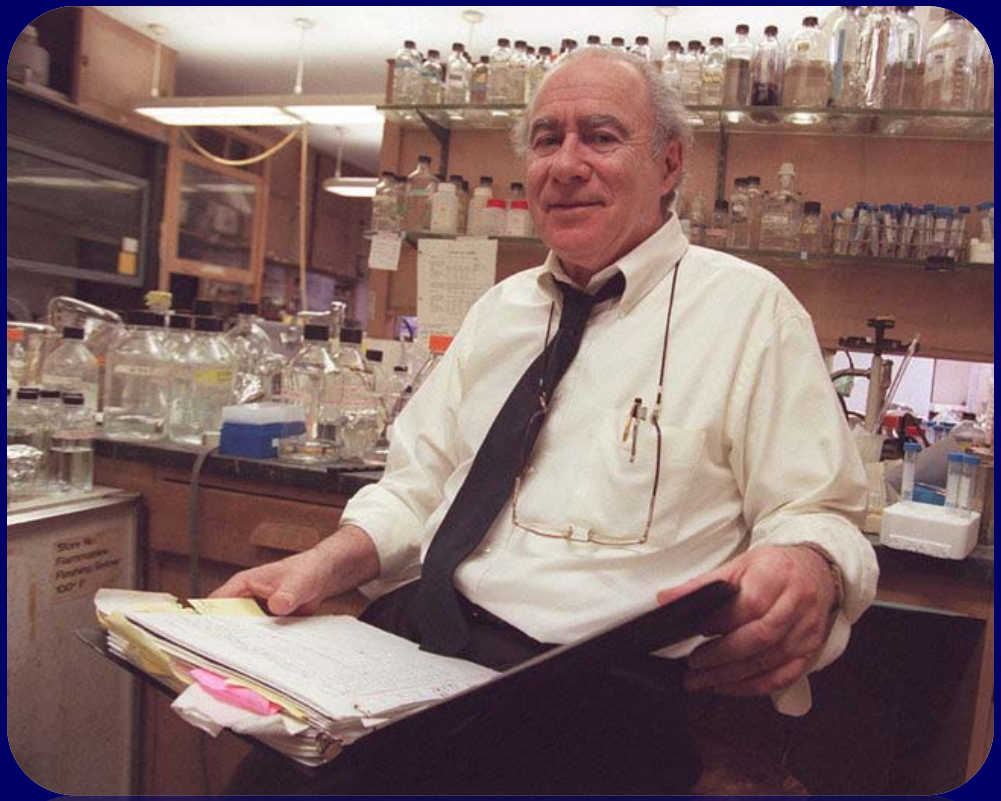


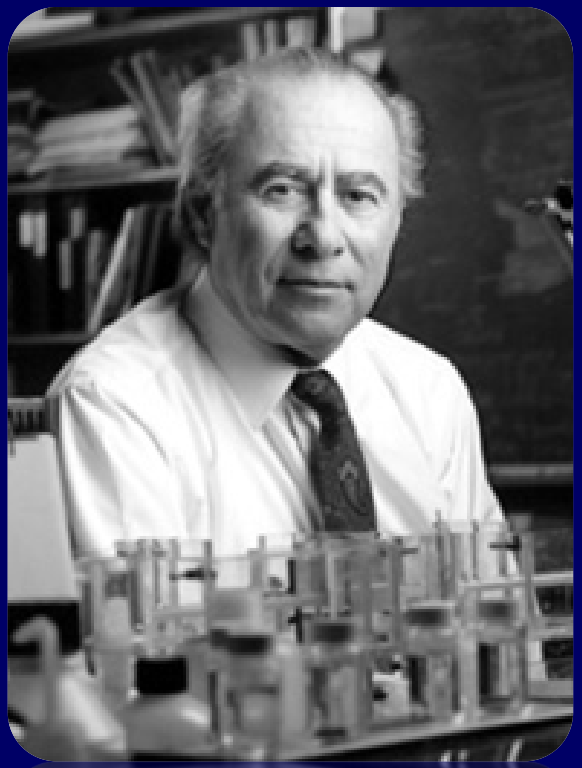
Mourning the Loss of James Crow



Document 13

Mourning the Loss of Norton Zinder





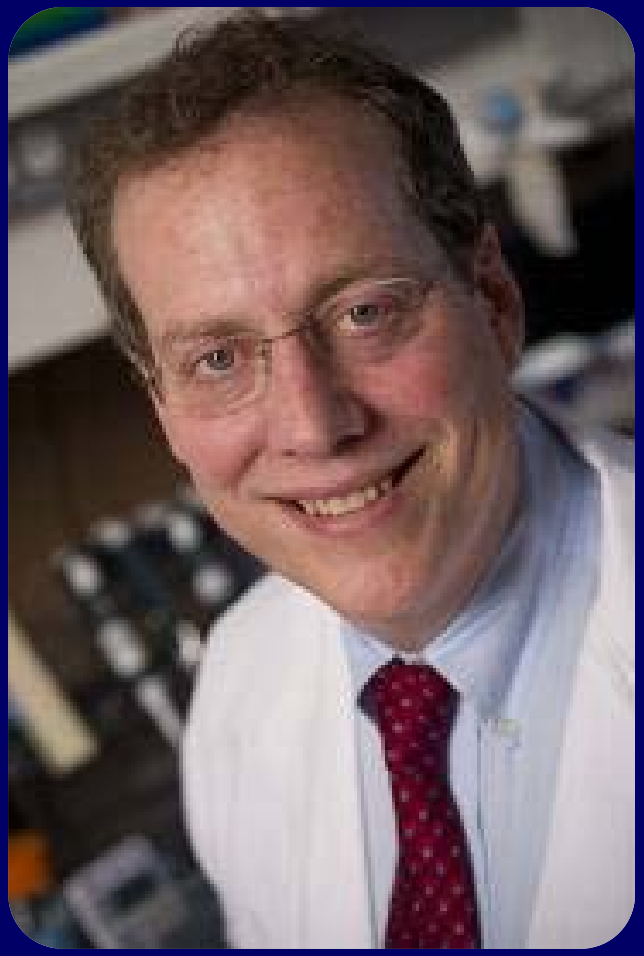
Document 13

2011 ASHG Curt Stern Award

David Altshuler, M.D., Ph.D.

Document 14

2011 NIH Director's Pioneer Award



NIH DIRECTOR'S PIONEER A·W·A·R·D

Andrew Feinberg, M.D., M.P.H.

Best Graduate Student Presenter for Genetics at SACNAS National Conference

SACNAS

Devoted to Advancing Hispanics, Chicanos & Native Americans in Science

Keolu Fox, Ph.D.

Document 16

2011 "Niki" Award Recipient

ATHENS INFORMATION TECHNOLOGY

CENTER OF EXCELLENCE FOR RESEARCH AND GRADUATE EDUCATION

Manolis Kellis, Ph.D.

National Academy of Science Public Welfare Medal Recipient

Harold Shapiro, Ph.D.

Newly Elected ASHG Leadership

President Elect:
Jeff Murray

Treasurer:
Geoff Duyk

Board of Directors:

Vivian Cheung

Evan Eichler

Richard Gibbs

Elected to the Institute of Medicine 2011

- Martin Blaser
- Vivian Cheung
- Claire Fraser-Liggett
- Richard Gibbs
- David Relman

Elected to AAAS

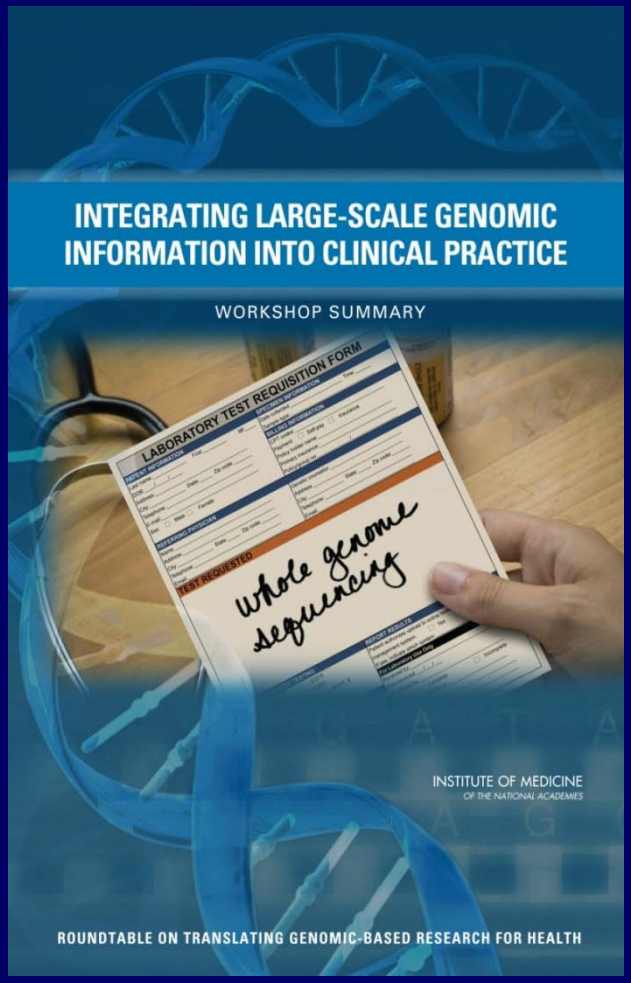
- Andrew Feinberg
- Edward Marcotte
- Richard McCombie
- Richard Myers

USPTO Study on Genetic Diagnostic Testing

Presidential Commission for the Study of Bioethical Issues Meeting:

Whole-Genome Sequencing

Institute of Medicine Report

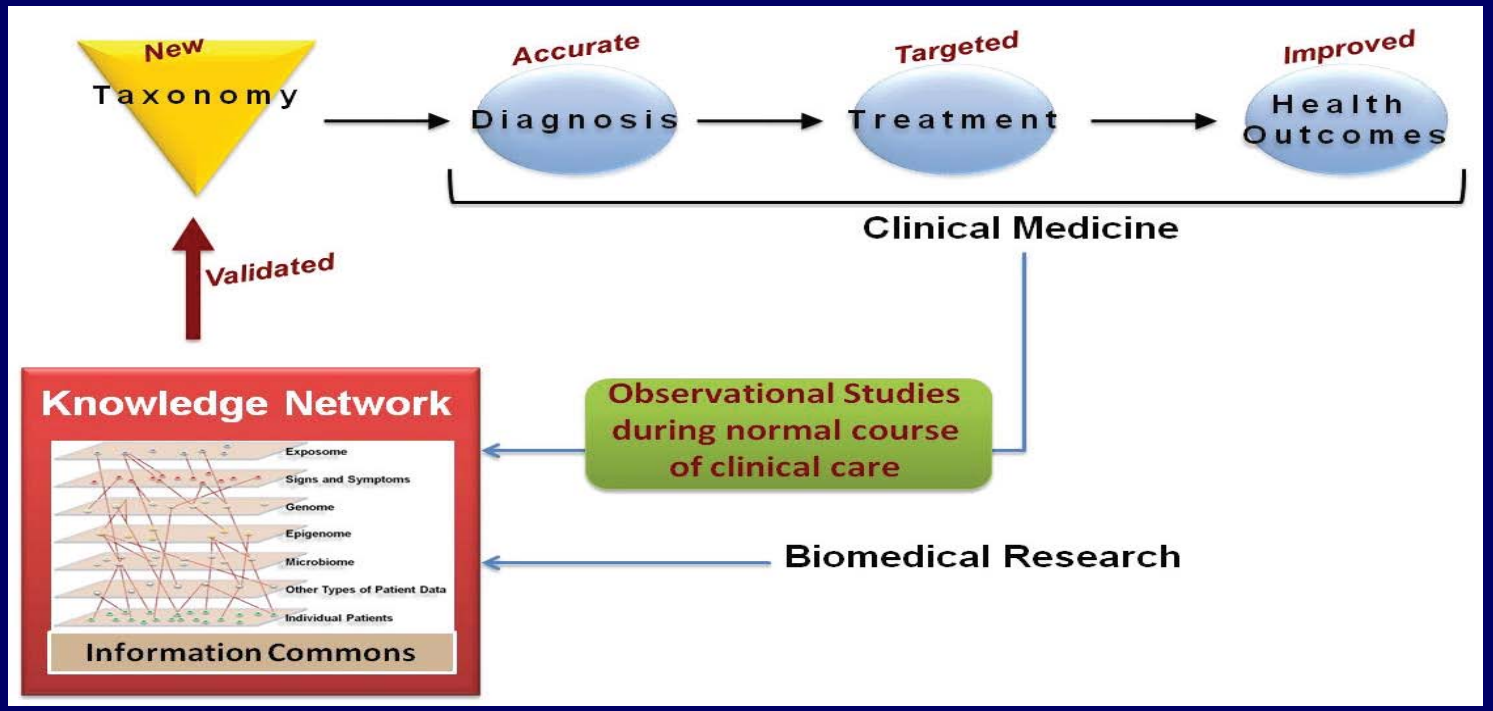


INSTITUTE OF MEDICINE
OF THE NATIONAL ACADEMIES

Document 22

National Academies Report

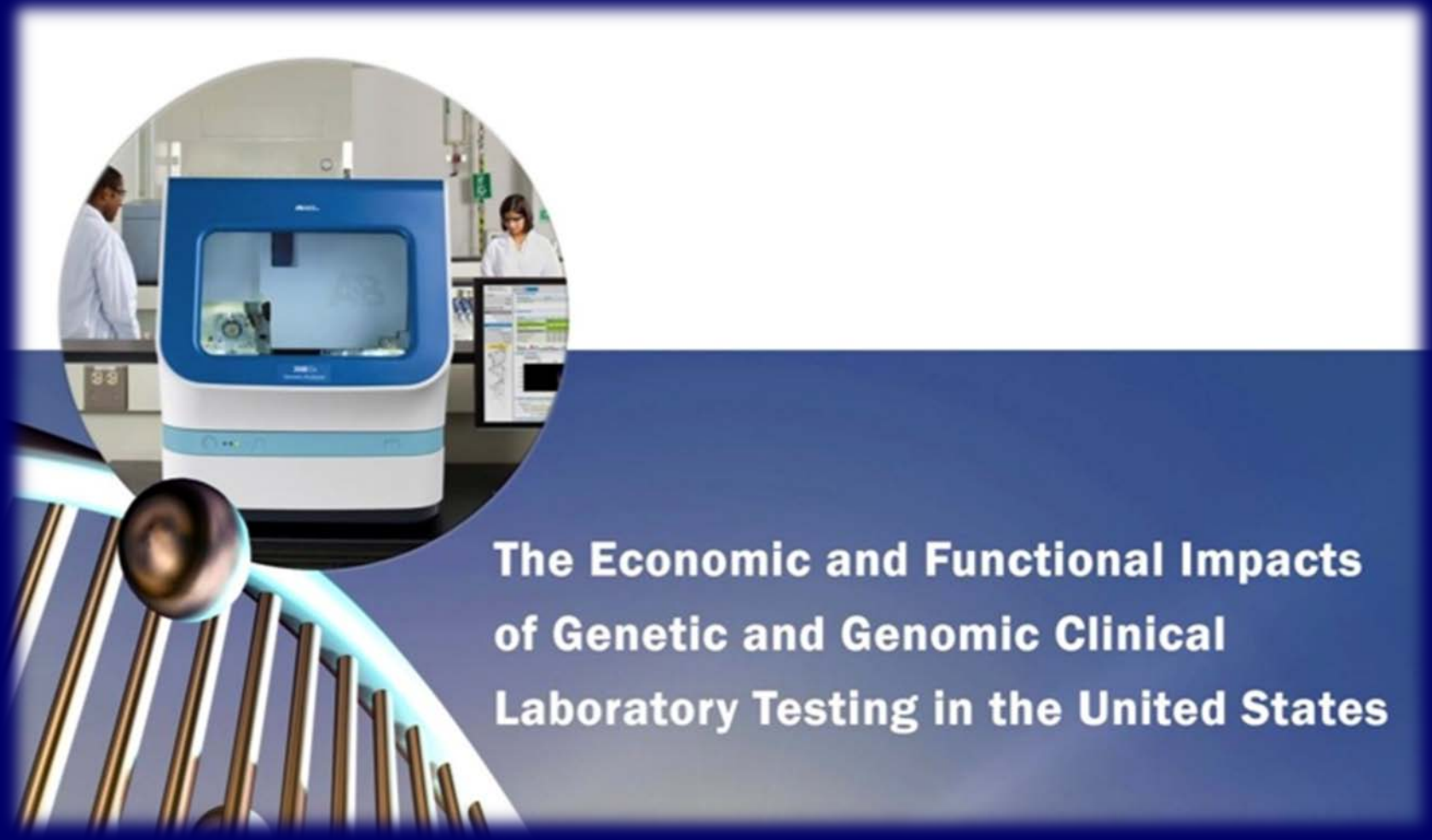
"Toward Precision Medicine: Building a Knowledge Network for Biomedical Research and a New Taxonomy of Disease"



THE NATIONAL ACADEMIES PRESS

Document 23

Battelle Report on Genomic Clinical Testing Industry



Document 24

American College of Medical Genetics Changes Name to American College of Medical Genetics and Genomics

Effective March 2012
at the ACMG Annual
Clinical Genetics Meeting

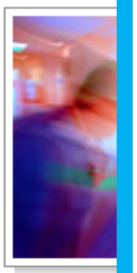
Document 25

NHGRI Genome Advance of the Month

Trauma regulates genes that predict survival

Septemb

By Jonat
Science



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Using the
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Dissecting the cause of the Black Death

October

By Jonath
Science

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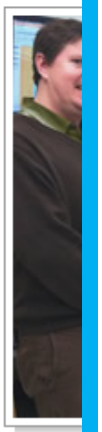
Massively parallel sequencing:
Taking an all-at-once approach to genetic testing in cancer

Comments

By Jor
Scienc



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Population genomics: Answering questions from the microscopic to the geographic

Jonathan Max Gitlin
Science Policy Analyst

The final Genome Advance of the Month for 2011 is actually a twofer, highlighting how population genomics can be used to answer questions in the diverse fields of vascular biology and anthropology. First up, from a multinational team of researchers lead by Dr. Christian Gieger in Germany, and Professor Willem Ouwehand and Dr. Nicole Soranzo in the United Kingdom, is a paper published in the journal *Nature*. The study takes advantage of genome-wide association study (GWAS) data previously collected in the course of other studies to uncover new genetic pathways that regulate how our bodies make platelets (the second most common blood cell after red blood cells).

Dr. Gieger and his colleagues began by using data gathered in 23 studies (from 48,666 participants of European descent) that had looked for genomic associations related to the number of platelets found in a patient's blood (their platelet count). They also looked at a smaller subset of studies (13 cohorts, 18,600 participants) that included mean platelet volume as well as platelet count. This approach identified 68 areas (loci) of the human genome that were associated with platelet size and number, of which 52 had not previously been linked.

The team followed up this meta-analysis by examining the functions of these genomic loci in fruit flies and zebrafish, two model organisms widely used in genomics. This revealed a number of genes already known to be involved in blood cell regulation and formation, as well as 11 novel genes. Mutations in some of these genes had previously been implicated in Mendelian (rare) diseases and human cancers.

Document 26

Genomics In The News…

First Australian Aboriginal Genome Sequenced

Genome Of World's Oldest Woman Sequenced

Document 27

Genomics In The News…

Elaine Mardis, Ph.D.

TheScientist
MAGAZINE OF THE LIFE SCIENCES

“High-Tech Choir Master”

Document 27

Genomics In The News…

Eric Lander, Ph.D.

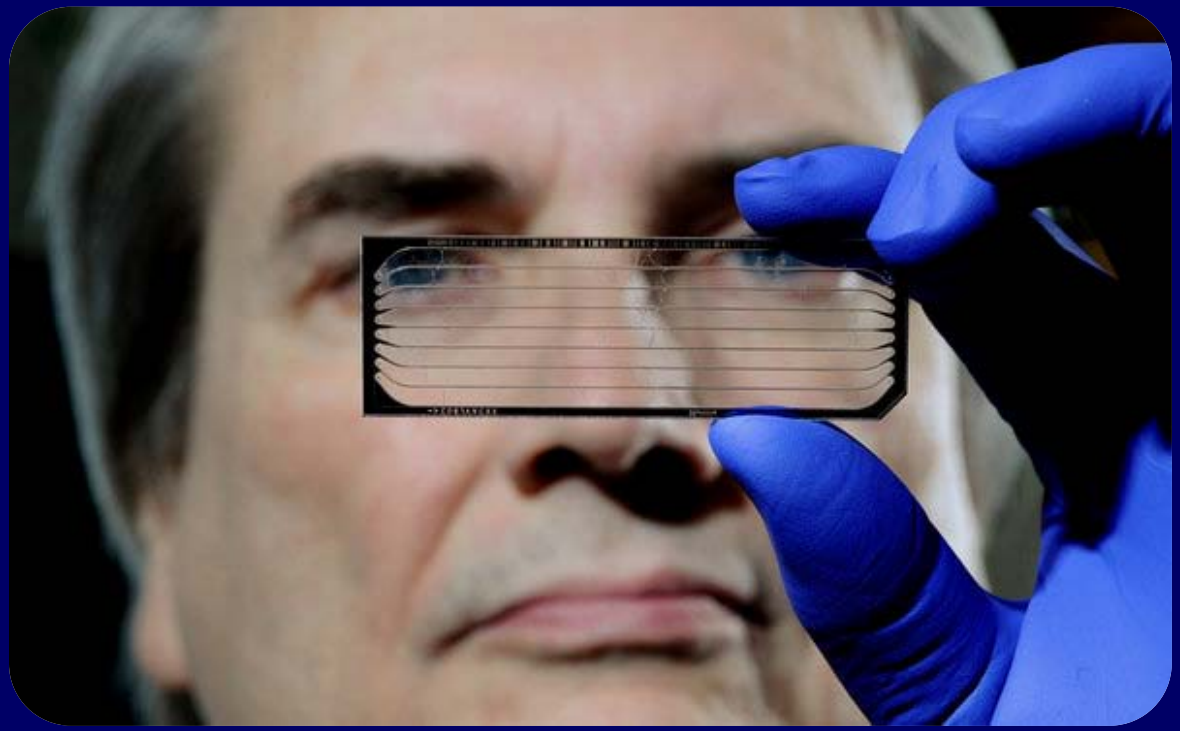
The New York Times

“Power in Numbers”

Document 27

Genomics In The News…

"DNA Sequencing Caught in Deluge of Data"



The New York Times

Dick McCombie, Ph.D.

Genomics In The News…

NEWS IN FOCUS

PROSPECTS

nature International weekly journal of science

New year, new science

Nature looks ahead to the key findings and events that may emerge in 2012.

LET'S TALK ABOUT EARTH

In June, scientists, politicians and campaigners of all stripes will flock to Rio de Janeiro, Brazil, for the United Nations' fourth Earth summit, devoted to sustainable development and the green economy. The conference — undoubtedly the major environmental meeting of 2012 — comes 20 years after the UN Framework Convention on Climate Change was signed at the first UN Earth summit, also in Rio.

THE SOURCE OF MARTIAN METHANE

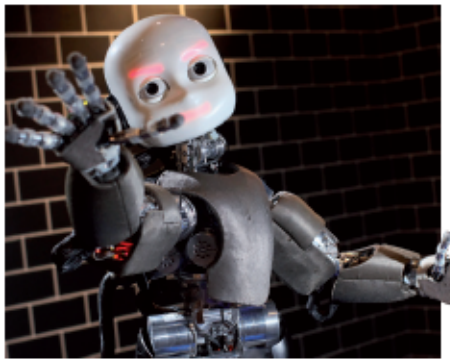
NASA's car-sized rover, Curiosity, is set to arrive on Mars in August. The US$2.5-billion craft will be lowered by an innovative landing system — the 'sky crane' — into Gale crater, where it will study rock strata in a bid to unpick the red planet's watery past. It will also sniff for methane in Mars's atmosphere, and could reveal whether the gas is being produced by geological processes — or by microbial martian life. Farther afield, NASA's Kepler mission surely ought to find a true extrasolar twin for Earth, with just the right size and orbit around a Sun-like star to be habitable.

ROBOTS, BRAINS OR GRAPHENE?

Six visionary research proposals will vie for huge grants from the European Commission's Future and Emerging Technologies Flagship scheme. The two winning projects, to be announced in the latter half of the year, will each receive €1 billion (US$1.3 billion) over the next decade. In the running are projects on graphene, hypothesized to be massless, chargeless entities able to serve as their own antiparticles, which could be useful for forming stable bits in quantum computing. Experiments have suggested that in materials known as topological insulators, the collective motions of electrons create a quasiparticle that behaves like a Majorana.

DNA ENCYCLOPEDIA

Biologists know that much of what was once termed 'junk' DNA actually has a role. But which sequences are functional — and what do they do? The best answer so far will come with a major update from the US National Institutes of Health's ENCODE (Encyclopedia of DNA Elements) project, which aims to identify all the functional elements in the human genome.



D. SCARFE GETTY IMAGES

Will iCub and his robot friends win a €1-billion grant this year?

patent protection, including the anticlotting Plavix (clopidogrel) and the antipsychotic Seroquel (quetiapine).

RAIDERS OF THE LOST LAKE

Within weeks, Russian researchers hope to finish drilling through Antarctica's ice sheet to reach Lake Vostok, a huge freshwater lake roughly 3,750 metres beneath the surface. It's a race against time: 10–50 metres of ice separate the team from its goal, which it must reach before the last aircraft of the season leaves in February. There'll be more drilling research in April, when Japan's *Chikyu* ship sets sail to bore into the underwater fault that caused the magnitude-9.0 Tohoku earthquake last year.

THE BIGGEST ARRAY

South Africa and Australia will find out by March which of them might host the $2.1-billion Square Kilometre Array (SKA), which would be the world's largest radio telescope if it is built. The decision will be made by the SKA's programme development office in Manchester, UK. Meanwhile, the Atacama Large Millimeter/Submillimeter Array in Chile's Atacama Desert should be 60% complete by the end of the year.

SPACEFLIGHT ADVANCES

In February, SpaceX of Hawthorne, California, hopes to be the first commercial firm to fly an unmanned cargo craft to the Interna-

Document 27

Genomics In The News…

U.S. NEWS | JANUARY 10, 2012

Soon, $1,000 Will Map Your Genes

THE WALL STREET JOURNAL.

The goal, triggered in part by an initiative launched by the U.S. government's National Human Genome Research Institute in 2004, already has resulted in a dramatic cost reduction in sequencing all three billion units of DNA, known as base-pairs, that make up the human genetic code.

Document 27

Genomics In The News…

UC Davis police chief on leave after pepper spraying

UC Davis said early Monday in a news release that it was necessary to place police Chief Annette Spicuzza on administrative leave to restore trust and calm tensions.

By Jason Dearen, *Associated Press* / November 21, 2011

"Free speech is part of the DNA of this university, and non-violent protest has long been central to our history," UC President Mark G. Yudof said in a statement Sunday in response to the spraying of students sitting passively at UC Davis. "It is a value we must protect with vigilance."

Document 27

What's in your car's DNA?

Our exclusive **Diagnostic Needs Analysis** *can provide you the necessary information to prevent future problems down the road.*

A quick analysis today can save costly repairs tomorrow.

We'll diagnose the condition of your vital fluids and other components

- *motor oil*
- *transmission fluid*
- *brake fluid*
- *coolant*
- *belts and hoses*
- *battery*
- *fuel system*

and more!

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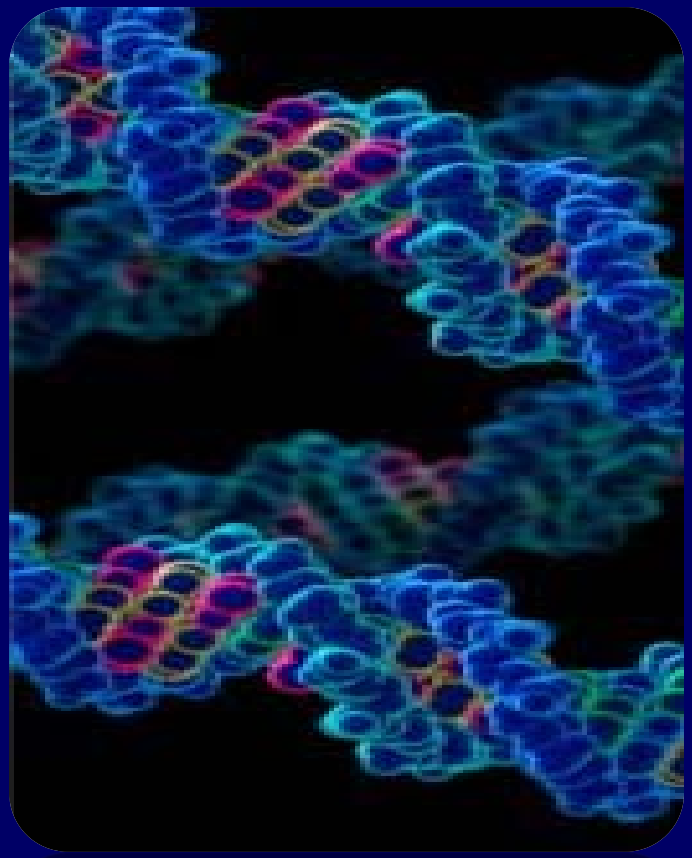
VII. NHGRI Intramural Program

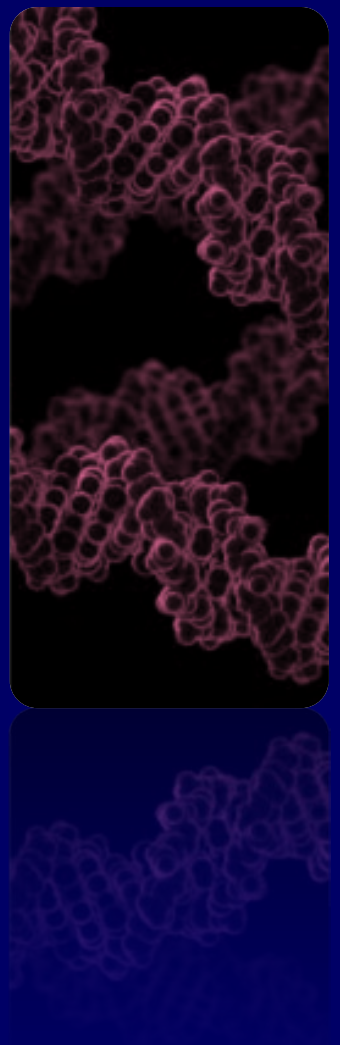
Large-Scale Genome Sequencing Centers

- **Baylor College of Medicine**
  PI: Richard Gibbs
  $21.3M in Year 1
- **Broad Institute**
  PI: Eric Lander
  $35.9M in Year 1
- **Washington University**
  PI: Richard Wilson
  $28.4M in Year 1



Mendelian Disorders Genome Centers

- University of Washington (Coordination Center)
  - PIs: Deborah Nickerson, Michael Bamshad, Mark Rieder, & Jay Shendure
  - $5.2M per year
- Yale University
  - PIs: Richard Lifton, Murat Gunel, Shrikant Mane, & Mark Gerstein
  - $2.8M per year
- Baylor College of Medicine & Johns Hopkins University
  - PIs: David Valle (Hopkins) & James Lupski (Baylor)
  - $4M per year

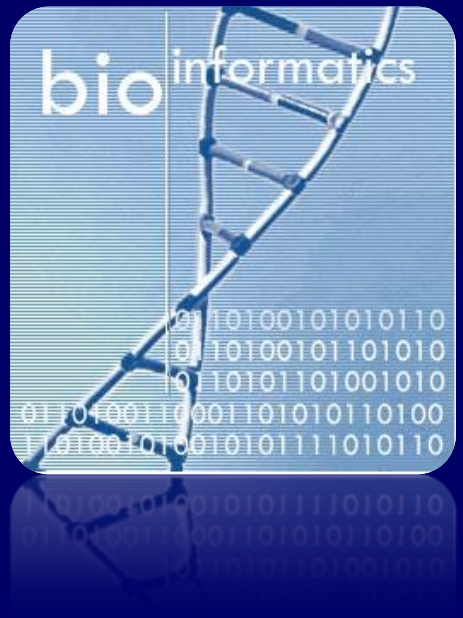


Clinical Sequencing Exploratory Research Projects

- **Baylor College of Medicine**
  PIs: Sharon Plon & Will Parsons, $1.8M per year (with NCI)
- **Brigham and Women's Hospital**
  PI: Robert Green, $2.4M per year
- **Children's Hosp. of Philadelphia**
  PIs: Ian Krantz & Nancy B. Spinner, $2.2M per year
- **University of North Carolina**
  PI: James Evans, $1.6M per year
- **University of Washington**
  PI: Gail Jarvik, $2.3M per year (with NCI)
- **Dana Farber Cancer Institute**
  PIs: Levi Garraway & Pasi Janne, $1.6M per year

Informatics Tools for High-Throughput Sequence Data Analysis Awards

- **Broad Institute ($1.0M in Year 1)**
  PI: Mark DePristo
- **Boston College & U. Michigan ($1.0M in Year 1)**
  PIs: Gabor Marth & Goncolo Abecasis
- **Washington University ($805K in Year 1)**
  PIs: Li Ding & David Dooling
- **Harvard ($448K in Year 1)**
  PI: Steven McCarroll
- **Scripps Institute ($382K in Year 1)**
  PI: Ali Torkamani
- **University of Southern California ($345K in Year 1)**
  PIs: Ting Chen, Ewa Deelman, & James Knowles



Genome Sequencing Program: Comparative Genomics

A high-resolution map of human evolutionary constraint using 29 mammals

Kerstin Lindblad-Toh[1,2], Manuel Garber[1]*, Or Zuk[1]*, Michael F. Lin[1,3]*, Brian J. Parker[4]*, Stefan Washietl[3]*, Pouya Kheradpour[1,3]*, Jason Ernst[1,3]*, Gregory Jordan[5]*, Evan Mauceli[1]*, Lucas D. Ward[1,3]*, Craig B. Lowe[6,7,8]*, Alisha K. Holloway[9]*, Michele Clamp[1,10]*, Sante Gnerre[1]*, Jessica Alföldi[1], Kathryn Beal[5], Jean Chang[1], Hiram Clawson[6], James Cuff[11], Federica Di Palma[1], Stephen Fitzgerald[5], Paul Flicek[5], Mitchell Guttman[1], Melissa J. Hubisz[12], David B. Jaffe[1], Irwin Jungreis[3], W. James Kent[9], Dennis Kostka[9], Marcia Lara[1], Andre L. Martins[12], Tim Massingham[5], Ida Moltke[4], Brian J. Raney[6], Matthew D. Rasmussen[3], Jim Robinson[1], Alexander Stark[13], Albert J. Vilella[5], Jiayu Wen[4], Xiaohui Xie[1], Michael C. Zody[1], Broad Institute Sequencing Platform and Whole Genome Assembly Team†, Kim C. Worley[14], Christie L. Kovar[14], Donna M. Muzny[14], Richard A. Gibbs[14], Baylor College of Medicine Human Genome Sequencing Center Sequencing Team†, Wesley C. Warren[15], Elaine R. Mardis[15], George M. Weinstock[14,15], Richard K. Wilson[15], Genome Institute at Washington University†, Ewan Birney[5], Elliott H. Margulies[16], Javier Herrero[5], Eric D. Green[17], David Haussler[6,8], Adam Siepel[12], Nick Goldman[5], Katherine S. Pollard[9,18], Jakob S. Pedersen[4,19], Eric S. Lander[1] & Manolis Kellis[1,3]

Nature (2011)

Document 28

1st TCGA Scientific Symposium November 17 & 18, National Harbor

- 2-day symposium describing analyses of TCGA data
- Open to all
- 470 Attendees, representing 13 countries
- Co-chairs Lynda Chin (MD Anderson) & Elaine Mardis (WashU Genome Institute)
- 30 Talks (available on YouTube / Genome TV)
- 8 Hands-on sessions
- 159 Posters

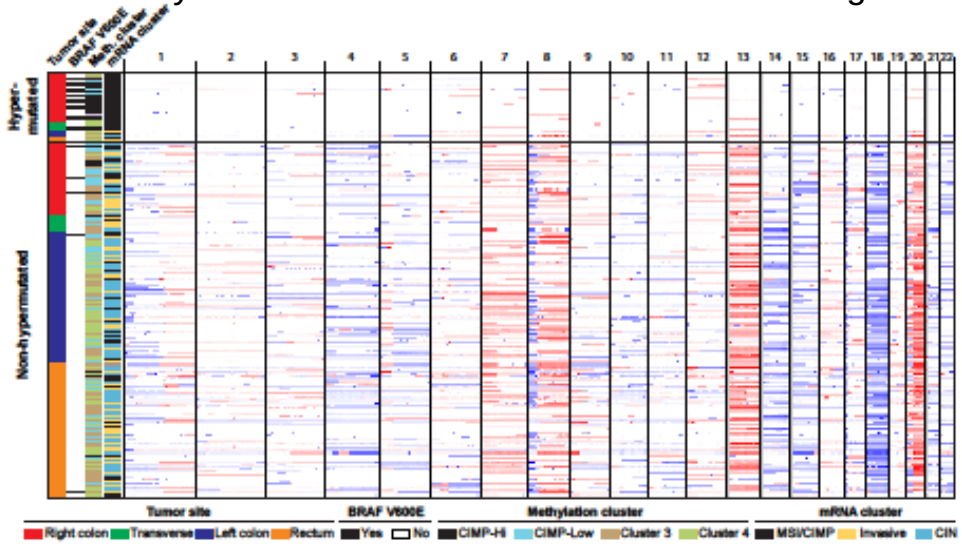
2nd Annual TCGA Symposium being planned for November 2012; American Asso. For Cancer Research (AACR) to co-sponsor

Document 29

TCGA: by the numbers

- TCGA achieves goal of 5,000 specimens analyzed by end of 2011.
- 22 tumor projects underway
- Many cancers of greatest incidence nearing completion
  - Published: Glioblastoma, Ovarian
  - Submitted: Colorectal (see figure)
  - Expected 2012 publications: Breast, AML, Lung, Kidney

Molecular analyses show colon and rectal cancers are single disease.

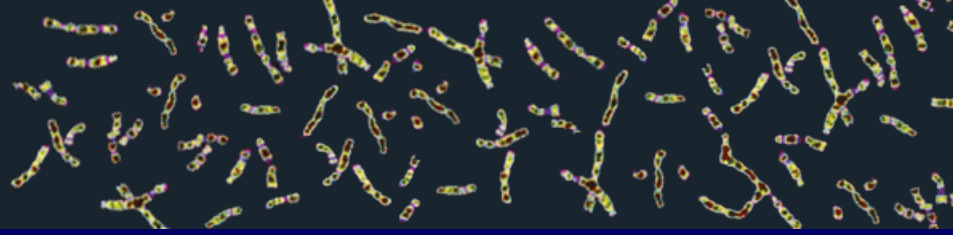


- Glioblastoma
- Ovarian
- Acute Myeloid Leukemia
- Colorectal Carcinoma
- Breast
- Lung Squamous Cell
- Lung Adenocarcinoma
- Renal Clear Cell Carcin.
- Renal papillary
- Uterine (endometrial)
- Low grade glioma
- Gastric Carcinoma
- Prostate
- Bladder
- Cervical
- Head and neck
- Liver
- Melanoma
-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

The Cancer Genome Atlas

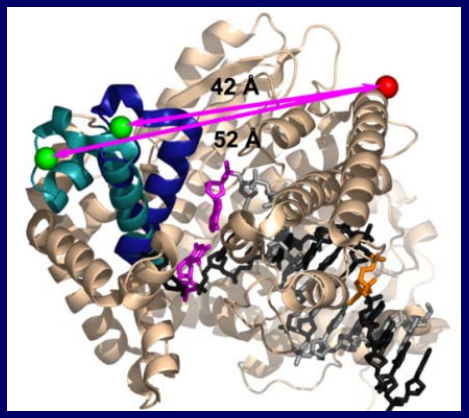
1000 Genomes

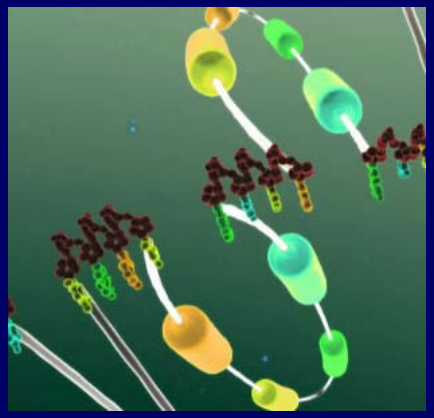
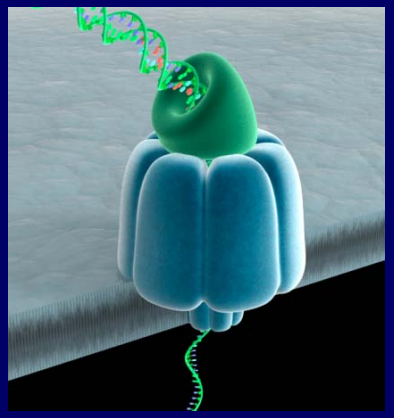
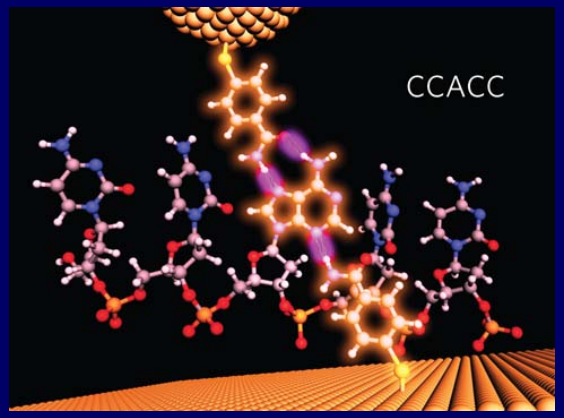
A Deep Catalog of Human Genetic Variation



- The Phase 1 paper is being written on 1094 samples from 14 populations, with 40 million SNP, indel, and deletion variants on integrated haplotypes
- The Phase 2 data set contains low-coverage and exome sequencing data on 1600 unrelated samples from 19 populations
- The Phase 3 sample collection from 7 more populations should be completed by April; all the sequencing should be completed by fall 2012, on 2500 unrelated individuals plus 161 trio kids
- Complete Genomics will deeply sequence 500 samples, including 161 trios

DNA Sequencing Technology Development



- **Annual grantee meeting**

  Grantees only: April 9-11

  Public: April 11-12

- **Presentation by Jeff Schloss later in Open Session**

Document 31

ENCODE & modENCODE

- Technology development RFA applications to be discussed in Closed Session

- Applications for the next production phase of ENCODE have been received and will be discussed at May 2012 Council meeting

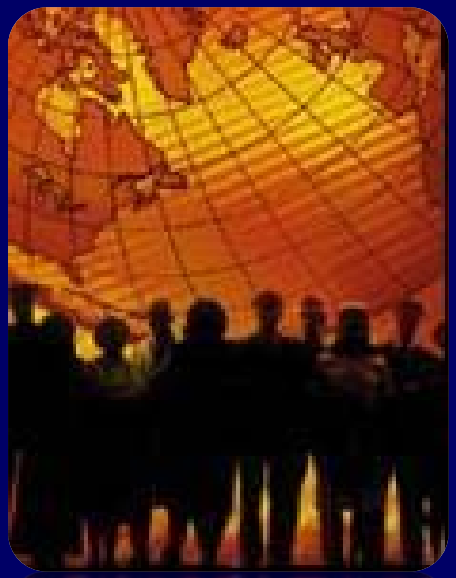
- modENCODE Symposium: June 20-21, 2012

Analyses

- ENCODE analysis session at December GENEVA Steering Committee meeting launched new collaborations
- Integrated analysis papers:
  - ENCODE – integrated manuscript under revision along with many companion papers
  - modENCODE – comparison of fly, worm, and human
  - Mouse – comparison of mouse and human

Return of Results Consortium

- **Components:**
  - **ELSI Return of Results RFAs (R01 and R21)**
  - **ELSI investigators in the Clinical Sequencing Exploratory Research Projects (U01)**
  - **Related investigator-initiated projects**
- **Goal: Identify areas of possible consensus that can form basis for policy development**
- **Plans for sharing outcome measures and instruments underway**



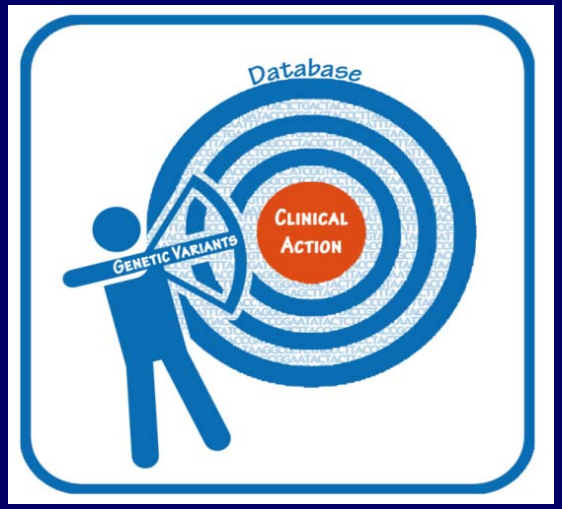
Centers of Excellence in Genomic Science (CEGS) & Diversity Action Plan (DAP)

- CEGS applications to be discussed in the Closed Session of this Council meeting
- Next receipt dates:
  CEGS: May 17, 2012
  DAP: May 25, 2012

Document 32

Characterizing and Displaying Genetic Variants for Clinical Action Workshop

- Goal: Consider processes, databases, and other resources needed to:
  - Identify clinically relevant variants
  - Decide whether they are actionable and what the action should be
  - Provide for clinical use
- Video, presentations, and recommendations posted on genome.gov
- Concept Clearance presentation later today



Genomic Medicine II Meeting

- Second in series of four planned genomic medicine meetings
- Goals:
  - Develop ideas for multicenter collaborative pilot projects in genomic medicine
  - Learn of new projects ongoing at partner sites
  - Identify infrastructure needs and solutions to speed adoption of genomic medicine
- Video, presentations, and recommendations posted on genome.gov
- Concept Clearance presentation later today

I. General NHGRI Updates

II. General NIH Updates

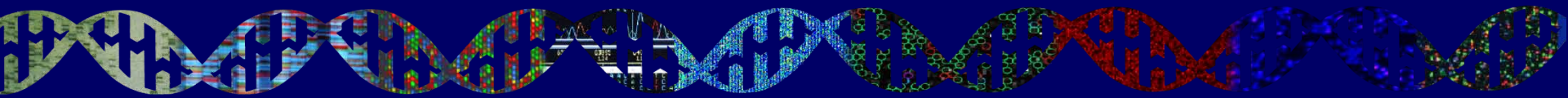
III. Genomics Updates

IV. NHGRI Extramural Program

V. NIH Common Fund Programs

VI. NHGRI Office of the Director

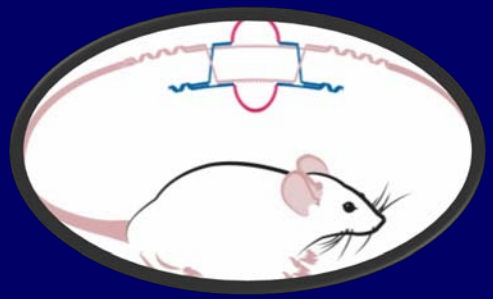
VII. NHGRI Intramural Program



Molecular Libraries Program (MLP)

- MLP Screening & Chemistry Centers' mid-year reports showed good progress toward milestones
- MLP Steering Committee meeting:
  - Presentations on center-driven research projects
  - Begin development of Molecular Libraries Biological Database

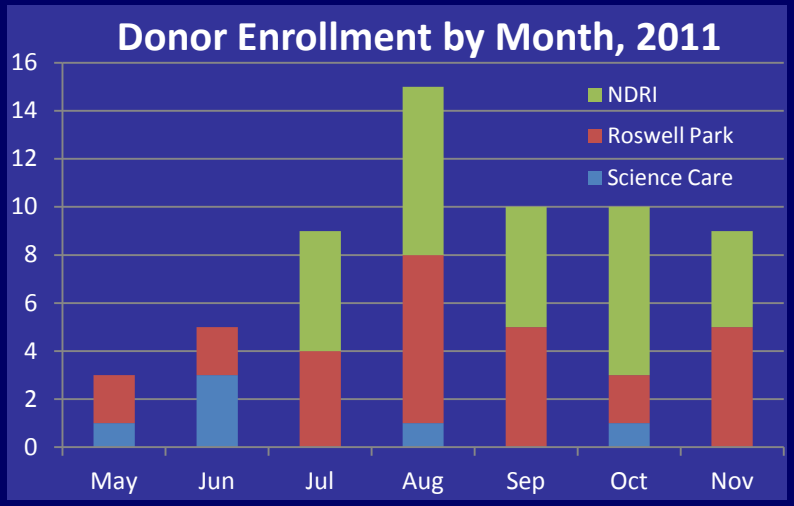
Knockout Mouse Projects



- KOMP Finale, KOMP2 Kickoff, and IMPC Launch Meeting: September 2011
- KOMP2 Awards for Knockout Mouse Production and Knockout Mouse Phenotyping
  - Baylor College of Medicine: Justice/Justice
  - The Jackson Laboratory: Donahue/Braun
  - UC Davis: Lloyd/Lloyd
- KOMP2 Award for Data Coordination Center and Database
  - European Bioinformatics Institute: Flicek

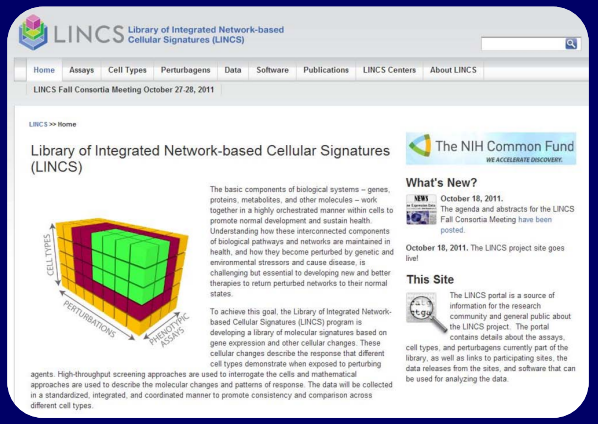
Genotype-Tissue Expression (GTEx)

- December 2011 meeting: Pilot roughly half completed



- Enrolling 10 post-mortem donors/month (on target); RNA integrity very good
- Proposal for scale up being developed

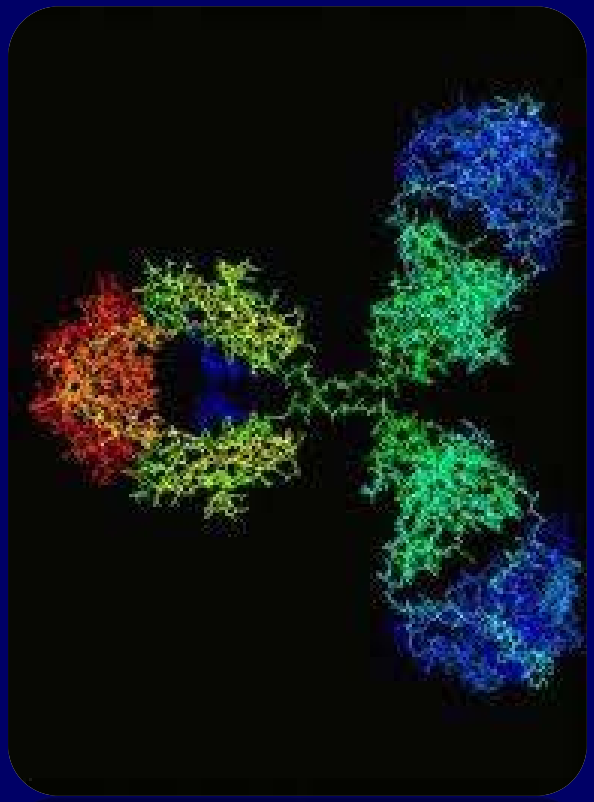
Library of Integrated Network-based Cellular Signatures (LINCS)



- Consortium meeting in October 2011
- LINCS program public web site now live
- Bridge funding proposal for Fiscal Year 2013
- Data release policy under development

Document 38

Protein Capture Reagents Program

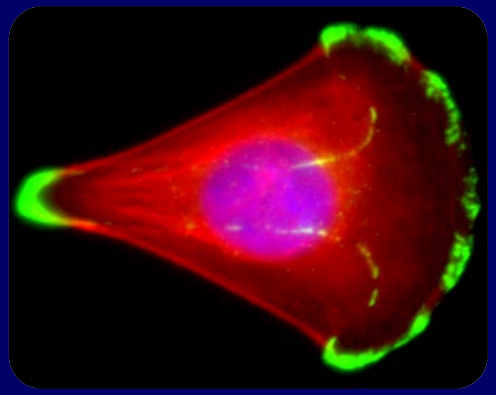


- Protein Capture Consortium meeting in December
  - Working groups formed
  - Development of public portal
- External Scientific Panel established for the program

Human Heredity and Health in Africa (H3Africa)

- Applications for four RFAs (centers, research projects, biorepositories, and bioinformatics network) received in December; excellent response
- Review of applications: March → May
- Biorepository RFA re-issued
- Wellcome Trust application process proceeding well and on similar timeline
- Ethics and Genomics Research in Africa (EAGER-Africa) Meeting in November 2011

Single Cell Analysis RFAs Issued

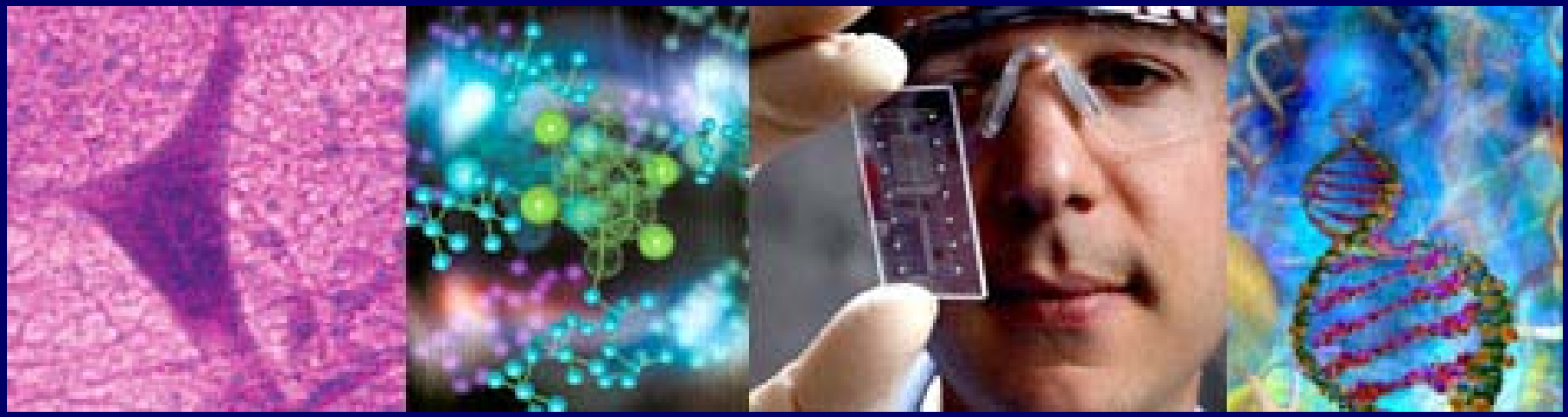


- Studies to Evaluate Cellular Heterogeneity using Transcriptional Profiling of Single Cells (U01)
- Exceptionally Innovative Tools and Technologies for Single Cell Analysis (R21)
- Accelerating the Integration and Translation of Technologies to Characterize Biological Processes at the Single Cell Level (R01)
- Applications were due in January

New Common Fund Initiative?

‘Disruptive Proteomics Technologies’

- Chosen for potential new Common Fund program starting in Fiscal Year 2013
- NIGMS and NHGRI as co-leads
- Strategic planning underway



Document 42

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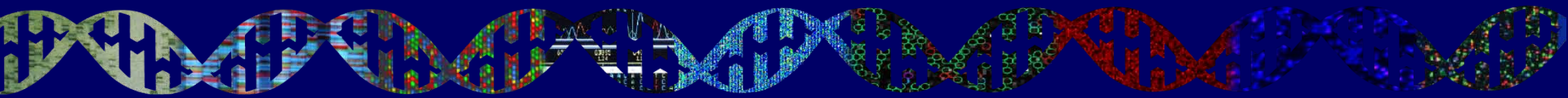
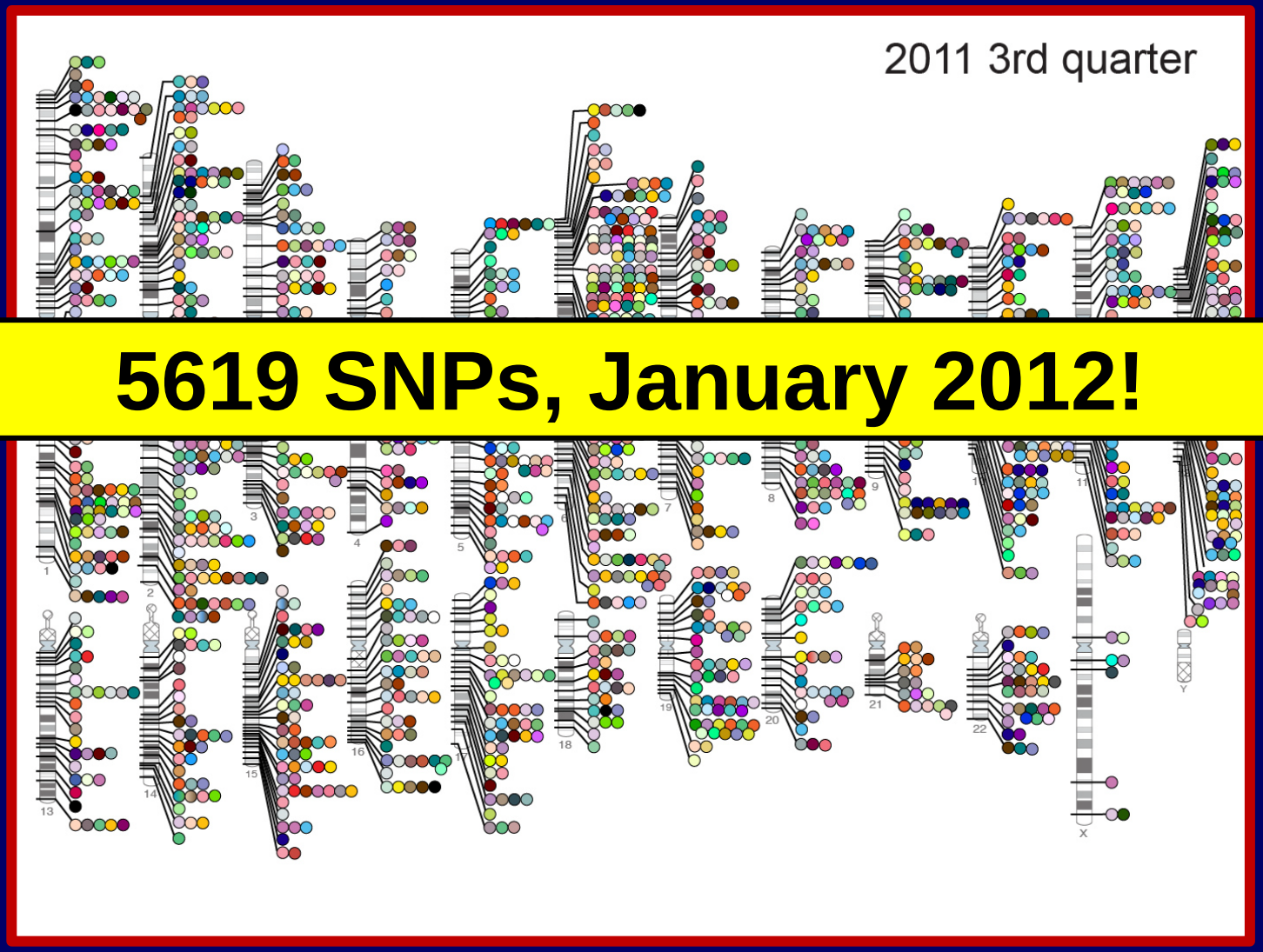
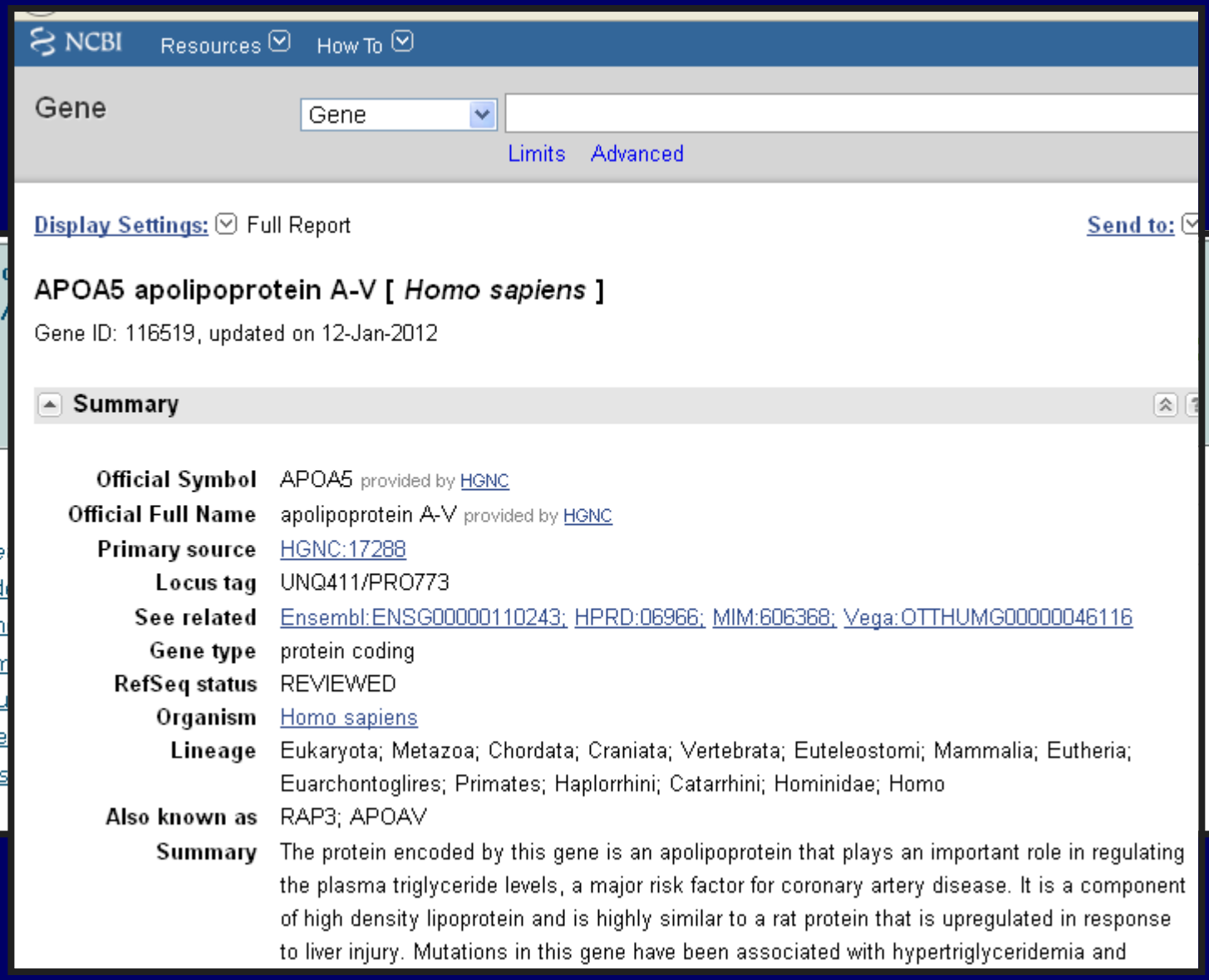


NHGRI Catalog of Published Genome-Wide Association Studies (GWAS)



Document 43

GWAS Catalog: New Features



Document 43

NEJM Genomic Medicine Series

Genomics, Health Care, and Society

Kathy L. Hudson, Ph.D.

Genomics and the Multifactorial Nature of Human Autoimmune Disease

Judy H. Cho, M.D., and Peter K. Gregersen, M.D.

Genomics of Cardiovascular Disease

Christopher J. O'Donnell, M.D., and Elizabeth G. Nabel, M.D.

CARDIOVASCULAR DISEASE IS THE LEADING CAUSE OF DEATH IN THE UNITED States. Considerable progress has been made in the past 50 years to define,

Genomics and Perinatal Care

Joann Bodurtha, M.D., M.P.H., and Jerome F. Strauss, III, M.D., Ph.D.

AMONG BOTH PROSPECTIVE PARENTS AND PROVIDERS OF MEDICAL CARE, genetic and social concerns peak during the perinatal period. Advances in genomics and assisted reproductive technology have created new opportunities to detect genetic disorders and susceptibilities at multiple times during perinatal care and thus are relevant to these concerns. Emerging therapies for single-gene disorders may reshape these discussions.

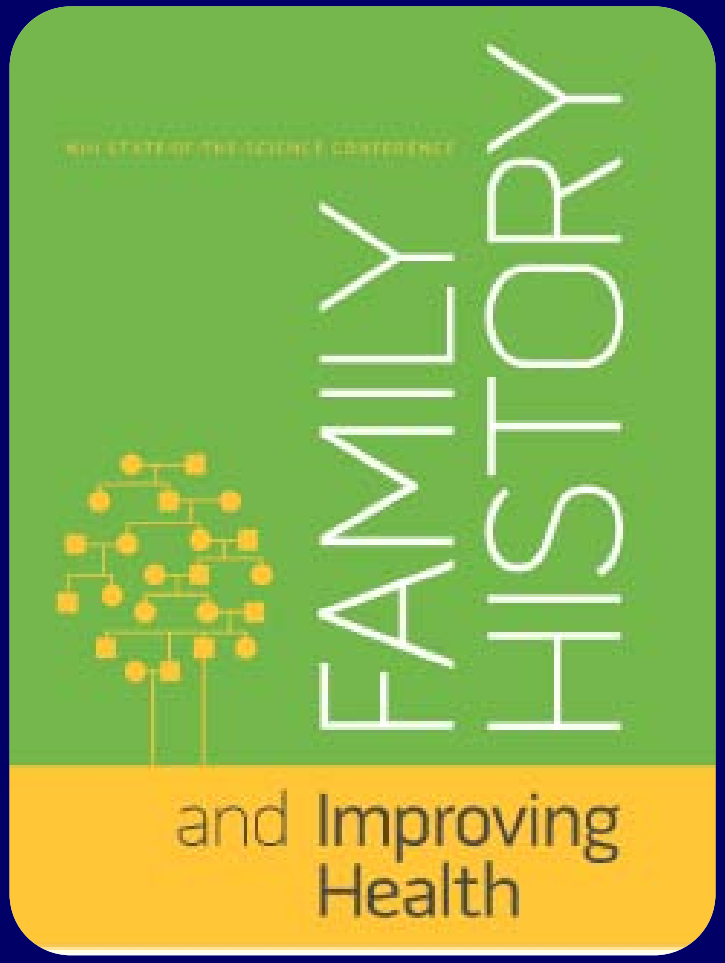
NEJM GENOMICS IN MEDICINE

Document 44

U.S. Science and Engineering Festival

Document 45

My Family Health Portrait



My Family Health Portrait

A tool from the Surgeon General

Genomic Medicine Lecture Series

- Monthly seminars being held at Suburban Hospital in Bethesda

David Valle, M.D.

Larry Brody, Ph.D.

Genomic Opportunities for Studying Sickle Cell Disease Meeting: December 2011

Co-Chairs: Michael DeBaun, Richard Gibbs, and Julie Makani

Pharmacist Education Meeting

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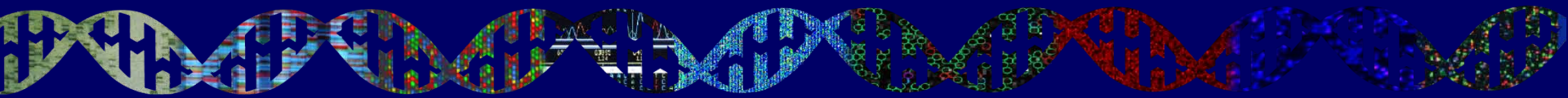
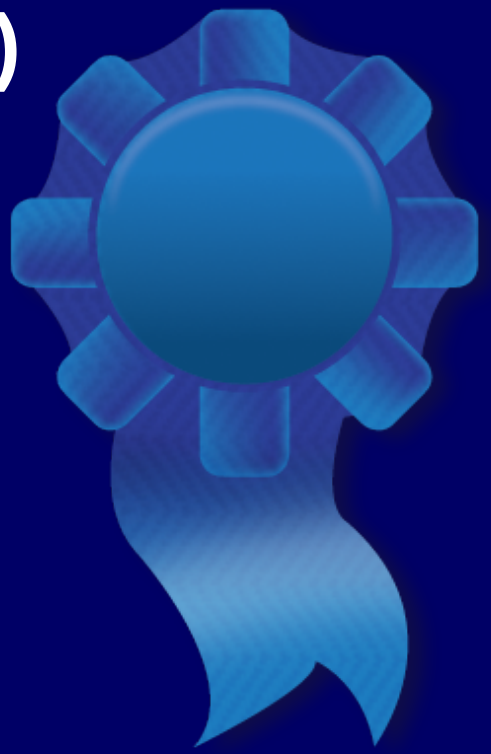


Blue Ribbon Panel Review of NHGRI Intramural Research Program

- 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

The NEW ENGLAND JOURNAL of MEDICINE

ORIGINAL ARTICLE

Cold Urticaria, Immunodeficiency, and Autoimmunity Related to *PLCG2* Deletions

Michael J. Ombrello, M.D., Elaine F. Remmers, Ph.D., Guangping Sun, M.D., Daniel L. Kastner, M.D., Ph.D., Matilda Katan, Ph.D., Hal M. Hoffman, M.D., and Joshua D. Milner, M.D.

nature genetics

NATURE GENETICS | LETTER

Exon capture analysis of G protein-coupled receptors identifies activating mutations in *GRM3* in melanoma

Todd D Prickett, Xiaomu Wei, Isabel Cardenas-Navia, Jamie K Teer, Jimmy C Lin, Vijay Walia, Jared Gartner, Jiji Jiang, Praveen F Cherukuri, Alfredo Molinolo, Michael A Davies, Jeffrey E Gershenwald, Katherine Stemke-Hale, Steven A Rosenberg, Elliott H Margulies & Yardena Samuels

AJHG

Cell PRESS

Available online 2 February 2012

The Phenotype of a Germline Mutation in *PIGA*: The Gene Somatically Mutated in Paroxysmal Nocturnal Hemoglobinuria

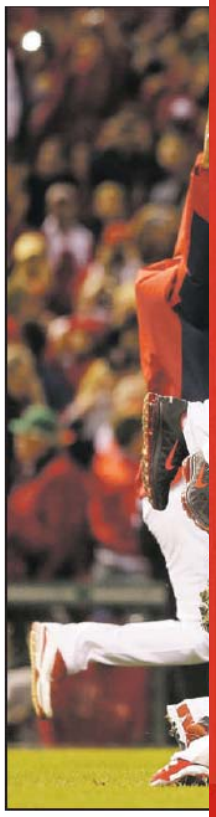
Jennifer J. Johnston[1], Andrea L. Gropman[2], Julie C. Sapp[1], Jamie K. Teer[1], Jodie M. Martin[2], Cyndi F. Liu[3], Xuan Yuan[3], Zhaohui Ye[3], Linzhao Cheng[3], Robert A. Brodsky[3], Leslie G. Biesecker[1,4]

William Gahl Honored with Prestigious Service to America Medal

Document 48

St. Louis

Laurie Skrivan | Post-Dispatch



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Congratula

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

Special Thanks!


genome.gov
THE BRIGHT FUTURE OF HUMAN GENOMICS