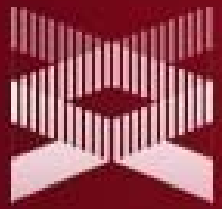


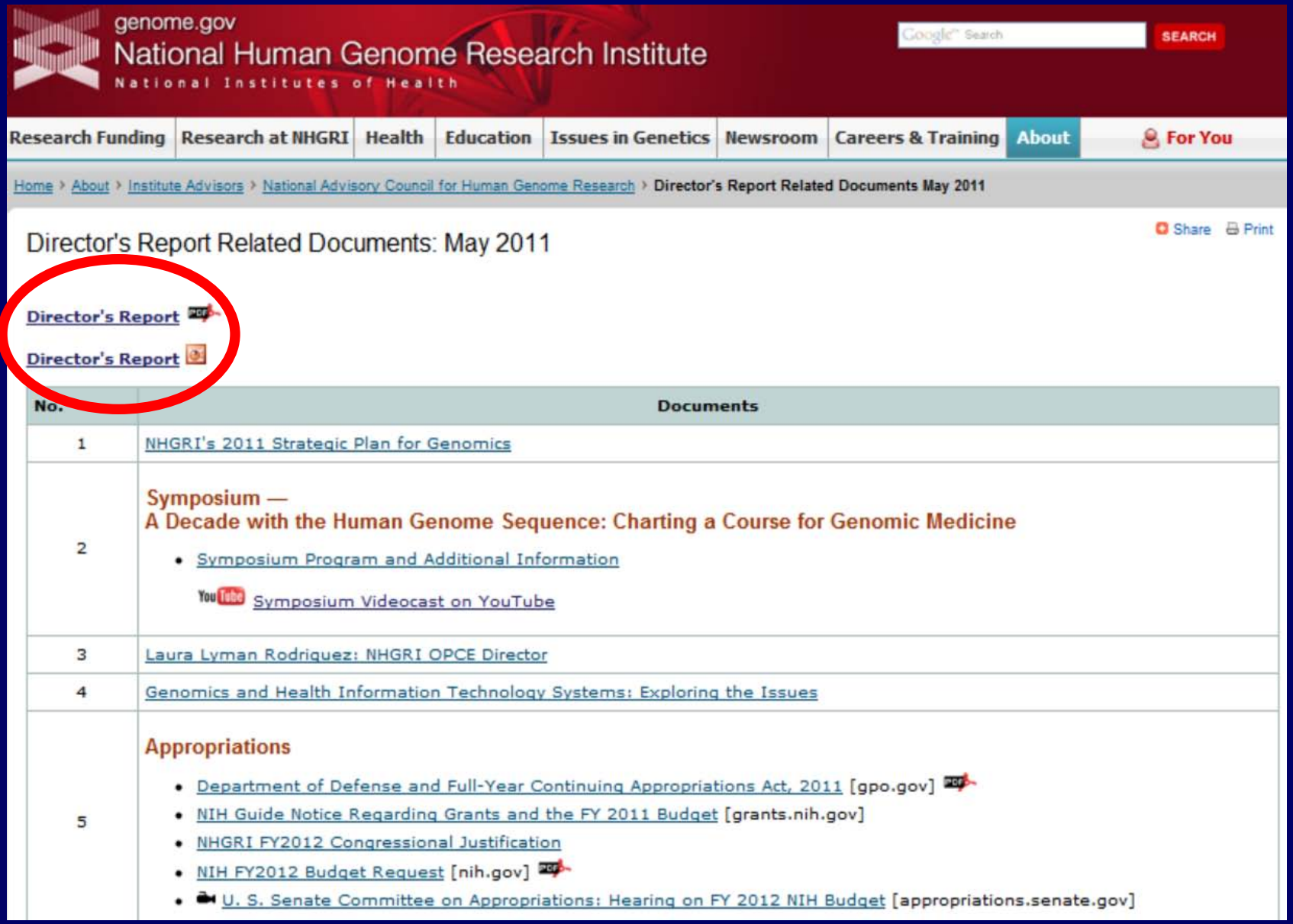
genome.gov
National Human Genome Research Institute
National Institutes of Health

DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

May 2011

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



genome.gov/DirectorsReport

Document #

I. General NHGRI Updates

II. General NIH Updates

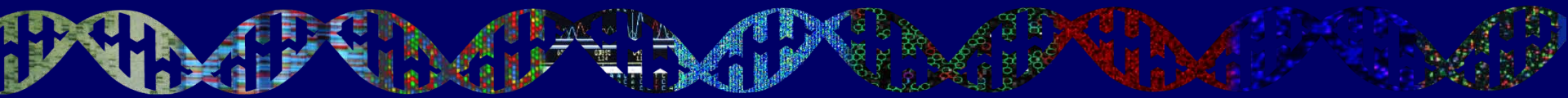
III. Genomics Updates

IV. NHGRI Extramural Program

V. NIH Common Fund Programs

VI. NHGRI Office of the Director

VII. NHGRI Intramural Program



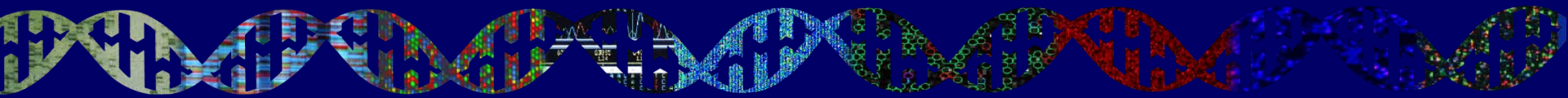
Open Session Presentations

Topic of Interest:

- PheGenI
  (Lucia Hindorff)

Program Updates:

- ENCODE Program
  (Elise Feingold)
- Microbiome Research
  (Lita Proctor)



Open Session Presentations

Meeting Reports:

- Collection, Storage, Management, and Distribution of Next-Generation Sequence Data (Peter Good)
- Genomics and Health Information Technology Systems: Exploring the Issues (Jeff Struewing)

Open Session Presentations

Concept Clearances:

- Future of ENCODE
  (Elise Feingold & Peter Good)
- Genomics of Gene Regulation
  (Peter Good & Elise Feingold)
- Analyzing the Whole Chip for GWAS
  (Teri Manolio)

I. **General NHGRI Updates**

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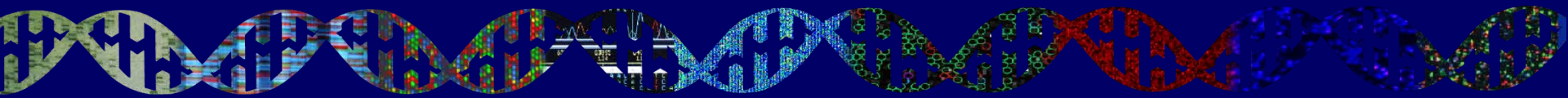
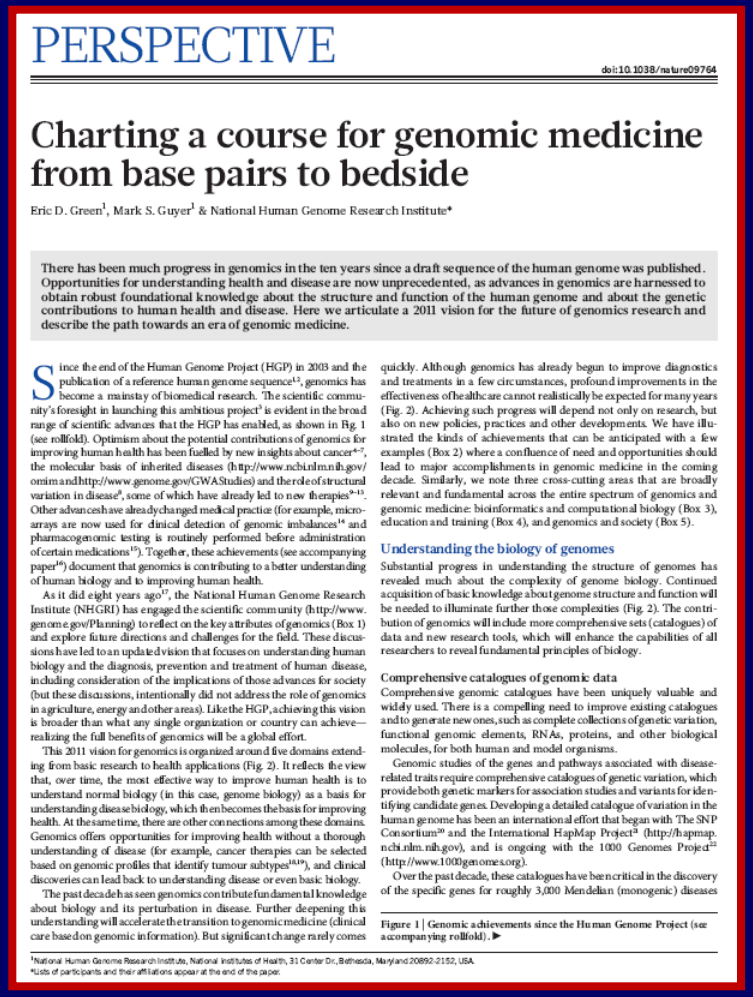


February 2011



PERSPECTIVE

doi:10.1038/nature09764

Charting a course for genomic medicine from base pairs to bedside

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

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

Since the end of the Human Genome Project (HGP) in 2003 and the publication of a reference human genome sequence[1,2], genomics has become a mainstay of biomedical research. The scientific community's foresight in launching this ambitious project[3] is evident in the broad range of scientific advances that the HGP has enabled, as shown in Fig. 1 (see rollfold). Optimism about the potential contributions of genomics for improving human health has been fuelled by new insights about cancer[4–7], the molecular basis of inherited diseases (http://www.ncbi.nlm.nih.gov/omim and http://www.genome.gov/GWAStudies) and the role of structural variation in disease[8], some of which have already led to new therapies[9–11]. Other advances have already changed medical practice (for example, microarrays are now used for clinical detection of genomic imbalances[14] and pharmacogenomic testing is routinely performed before administration of certain medications[15]). Together, these achievements (see accompanying paper[16]) document that genomics is contributing to a better understanding of human biology and to improving human health.

As it did eight years ago[17], the National Human Genome Research Institute (NHGRI) has engaged the scientific community (http://www.genome.gov/Planning) to reflect on the key attributes of genomics (Box 1) and explore future directions and challenges for the field. These discussions have led to an updated vision that focuses on understanding human biology and the diagnosis, prevention and treatment of human disease, including consideration of the implications of those advances for society (but these discussions, intentionally did not address the role of genomics in agriculture, energy and other areas). Like the HGP, achieving this vision is broader than what any single organization or country can achieve—realizing the full benefits of genomics will be a global effort.

This 2011 vision for genomics is organized around five domains extending from basic research to health applications (Fig. 2). It reflects the view that, over time, the most effective way to improve human health is to understand normal biology (in this case, genome biology) as a basis for understanding disease biology, which then becomes the basis for improving health. At the same time, there are other connections among these domains. Genomics offers opportunities for improving health without a thorough understanding of disease (for example, cancer therapies can be selected based on genomic profiles that identify tumour subtypes[18,19]), and clinical discoveries can lead back to understanding disease or even basic biology.

The past decade has seen genomics contribute fundamental knowledge about biology and its perturbation in disease. Further deepening this understanding will accelerate the transition to genomic medicine (clinical care based on genomic information). But significant change rarely comes quickly. Although genomics has already begun to improve diagnostics and treatments in a few circumstances, profound improvements in the effectiveness of healthcare cannot realistically be expected for many years (Fig. 2). Achieving such progress will depend not only on research, but also on new policies, practices and other developments. We have illustrated the kinds of achievements that can be anticipated with a few examples (Box 2) where a confluence of need and opportunities should lead to major accomplishments in genomic medicine in the coming decade. Similarly, we note three cross-cutting areas that are broadly relevant and fundamental across the entire spectrum of genomics and genomic medicine: bioinformatics and computational biology (Box 3), education and training (Box 4), and genomics and society (Box 5).

Understanding the biology of genomes

Substantial progress in understanding the structure of genomes has revealed much about the complexity of genome biology. Continued acquisition of basic knowledge about genome structure and function will be needed to illuminate further those complexities (Fig. 2). The contribution of genomics will include more comprehensive sets (catalogues) of data and new research tools, which will enhance the capabilities of all researchers to reveal fundamental principles of biology.

Comprehensive catalogues of genomic data

Comprehensive genomic catalogues have been uniquely valuable and widely used. There is a compelling need to improve existing catalogues and to generate new ones, such as complete collections of genetic variation, functional genomic elements, RNAs, proteins, and other biological molecules, for both human and model organisms.

Genomic studies of the genes and pathways associated with disease-related traits require comprehensive catalogues of genetic variation, which provide both genetic markers for association studies and variants for identifying candidate genes. Developing a detailed catalogue of variation in the human genome has been an international effort that began with The SNP Consortium[20] and the International HapMap Project[21] (http://hapmap.ncbi.nlm.nih.gov), and is ongoing with the 1000 Genomes Project[22] (http://www.1000genomes.org).

Over the past decade, these catalogues have been critical in the discovery of the specific genes for roughly 3,000 Mendelian (monogenic) diseases

Figure 1 | Genomic achievements since the Human Genome Project (see accompanying rollfold). ▶

[1]National Human Genome Research Institute, National Institutes of Health, 31 Center Dr., Bethesda, Maryland 20892-2152, USA.
*Lists of participants and their affiliations appear at the end of the paper.

NHGRI's New Strategic Plan for Genomics

Document 1

Press Coverage of New Strategic Plan

NEWS&ANALYSIS

nature International weekly journal of science

Home | News & Comment | Research | Careers & Jobs | Current Issue | Archive | Audio

Archive > Volume 470 > Issue 7333 > Editorials > Article

NATURE | EDITORIAL

previous article

Best is yet to come

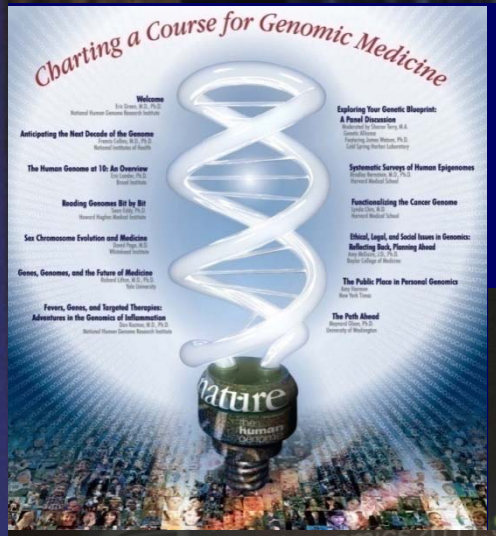
Nature **470**, 140 (10 February 2011) | doi:10.1038/470140a
Published online 09 February 2011

Companies such as Illumina would still have reimbursement issues and other problems to solve before installing sequencers in every hospital.

to the genomes of microbes in the human

really, really hit it hard.

enthusiasm about programs that are being



A Decade with the Human Genome Sequence

Document 2

A Decade with the Human Genome Sequence

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<Embed>

About this playlist

1,263 views
16 videos
Total length: 7 hours

Description: Charting a
Medicine. NHGRI hoste
symposium on Friday,
5:00 p.m. at the Ruth L
Natcher Conference Ce
of Health (NIH) campus
was to offer the NIH co
into contemporary genc
how genomics can be u
discoveries and how ge
communities and socie

ce: by GenomeTV

Director of NIH

M.D., Ph.D.

D. Ph.D.

How to extract DNA from strawberries

GenomeTV Subscribe

0:13 / 9:46 360p

Like Flag

7,831 views

Uploaded by GenomeTV on Oct 21, 2010

Drs. Eric Green and Carla Easter from the National Human Genome Research Institute (NHGRI) of the National

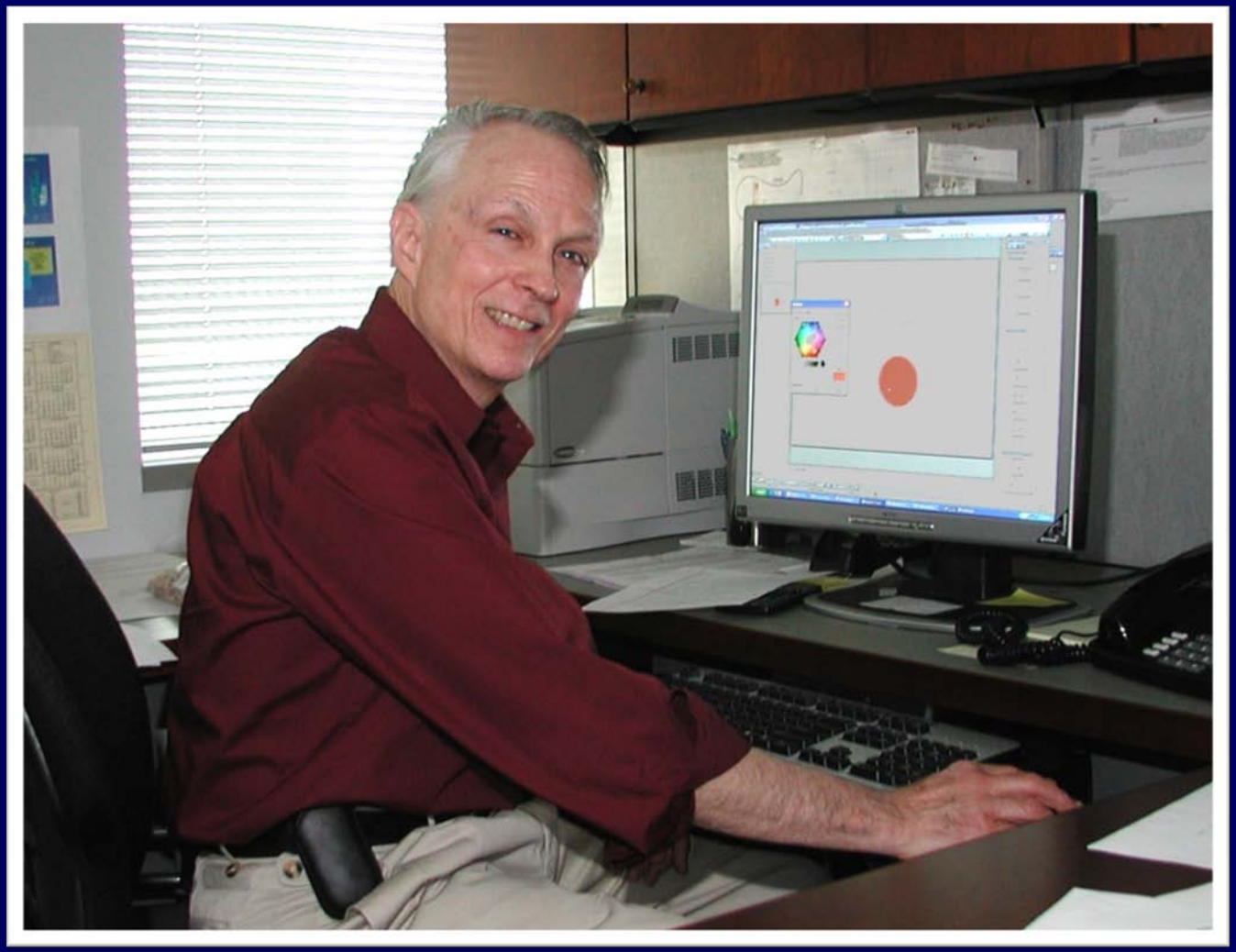
Document 2

Appointment of NHGRI OPCE Director

Laura Lyman Rodriguez, Ph.D.

Document 3

Extramural Program Director Retires



Gary Temple, M.D., Ph.D.

Special Advisor to NHGRI Director

Marc Williams, M.D.

U. of Maryland Law School Workshop: Regulation of Genomic Research

Genomics and Health Information Technology Systems: Exploring the Issues

- A direct outgrowth of the strategic planning process
- Meeting held in April 2011 (~90 participants)
- Explored the broad spectrum of issues facing the intersection of clinical informatics systems and genomics
- Upcoming presentation by Jeff Struewing

Document 4

The Collection, Storage, Management, and Distribution of Next-Generation Sequence Data

- Meeting held May 2011 (~80 participants)
- Aimed to identify the issues related to the increase in next-generation sequencing data
- Also aimed to develop a plan for how the scientific community can share large datasets in a cost-efficient and scientifically rational fashion
- Upcoming presentation by Peter Good

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FY2011 Appropriations Update

- **NIH: $30.7B**
  (0.8% less than FY2010)
- **NHGRI's final number pending**

Document 5

FY2011 Extramural Award Guidelines

- **Non-Competing Research Awards**

  Awarded at 1% below FY2010 levels
  (except for NCI with a 3% reduction)

- **Competing Research Awards**

  Individual Institutes to determine

- **New Investigators**

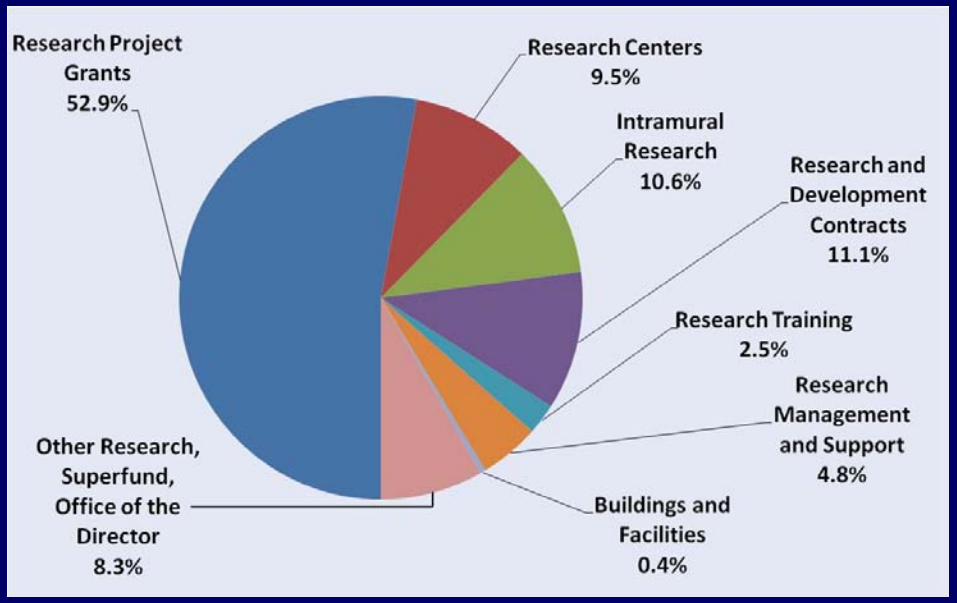
  Maintain at success rates of established investigators

- **NRSAs**

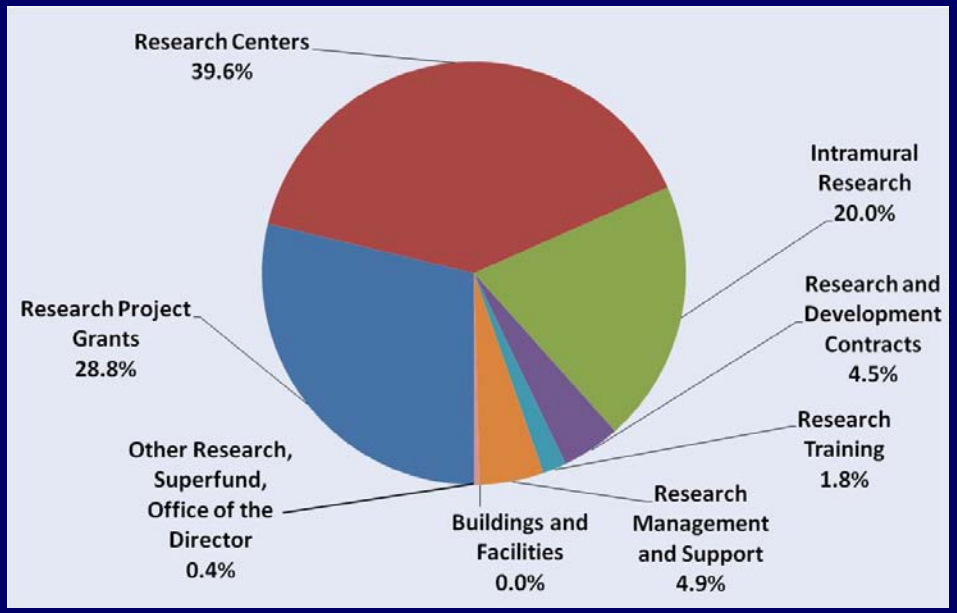
  2% increase at all stipend levels

Document 5

President's FY2012 Budget



NIH



NHGRI

President's FY2012 Budget

- **NIH: $32B**
  (2.4% increase)
- **NHGRI: $525M**
  (1.7% increase)

Document 5

House FY2012 Budget Resolution

Senate Appropriations Hearing

Senate Hearings on NIH

Genomics research was a central feature of the NIH message

Document 5

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

Document 6

NCATS will:

- **Facilitate – not duplicate – other translational research activities supported by NIH**
- **Complement – not compete with – the private sector**
- **Reinforce – not reduce – NIH's commitment to basic research**

NCATS: Research Programs

- Components of Molecular Libraries Program
- Therapeutics for Rare and Neglected Diseases
- Rapid Access to Interventional Development
- Clinical and Translational Science Awards
- Office of Rare Diseases Research
- FDA-NIH Regulatory Science
- Cures Acceleration Network

Associate Director for Science Policy, NIH

Amy Patterson, M.D.

NCI Interim Director for Center for Cancer Genomics

Barbara Wold, Ph.D.

New Look for NIH Website

Document 7

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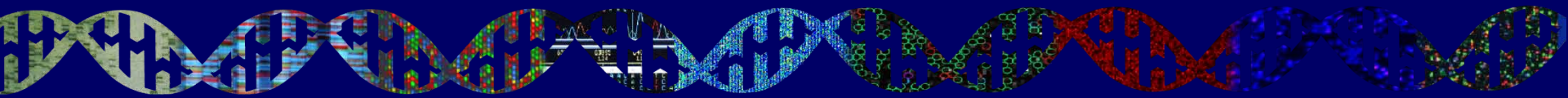
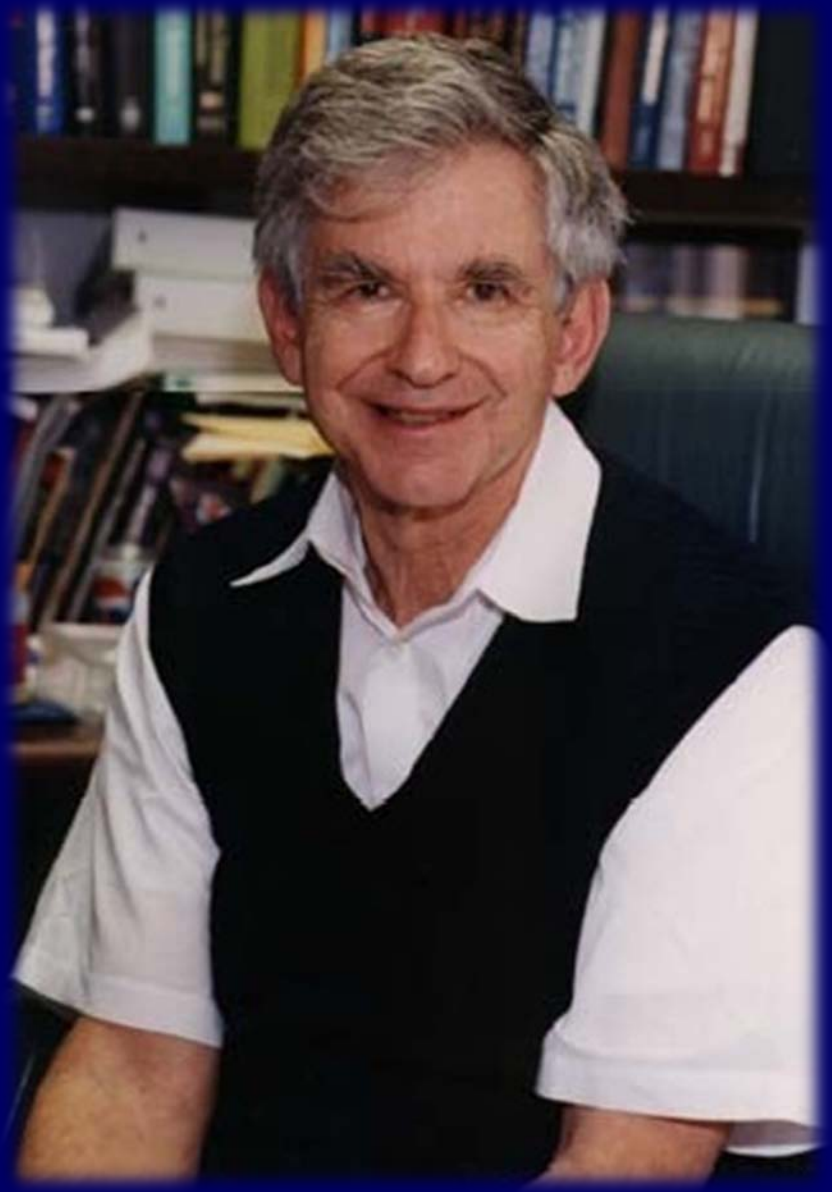


Mourning the Loss of Charley Epstein



Document 8

Presidential Award for Excellence in Science, Mathematics, and Engineering Mentoring

Jo Handelsman, Ph.D.

Document 9

American Society of Microbiology: 2011 Promega Biotechnology Research Award

AMERICAN SOCIETY FOR MICROBIOLOGY

Stephen Quake, Ph.D.

Document 10

2011 March of Dimes Prize in Developmental Biology

David Page, Ph.D.

Document 11

American Academy of Arts and Sciences

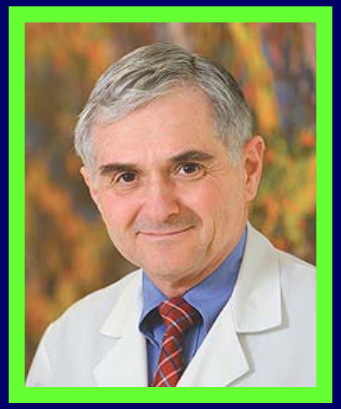
David Page, M.D.

Robert Kingston, Ph.D.

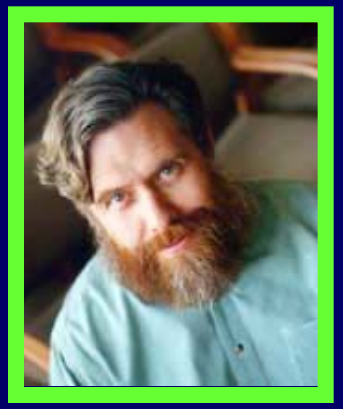
Document 12

National Academy of Science 2011 Electees

David Bartel

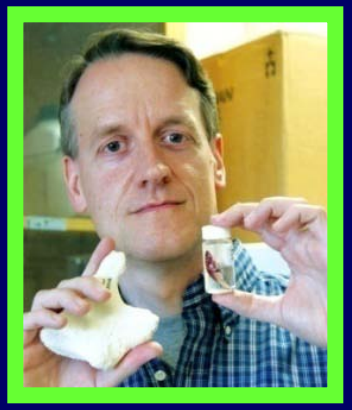


Art Beaudet



George Church

Hal Dietz



David Kingsley

Sunney Xie

Steve Warren

NATIONAL ACADEMY OF SCIENCES

Document 13

The Hottest Research of 2010: Reuters' *Science Watch*




				No. 'Hot Papers' ⬇
1	Eric Lander	Broad	Genetics	10
2	Augustine Kong	deCODE	Statistics/Genetics	9
3	Kári Stefánsson	deCODE	Genetics	9
4	Francis Collins	NIH	Genetics	9
5	Richard Durbin	Sanger	Bioinformatics	9
8	Unnur Thorsteinsdottir	deCODE	Genetics	8
9	Richard Wilson	WashU	Genetics	8
10	Mark Daly	Broad, MGH	Genetics	8

Document 14

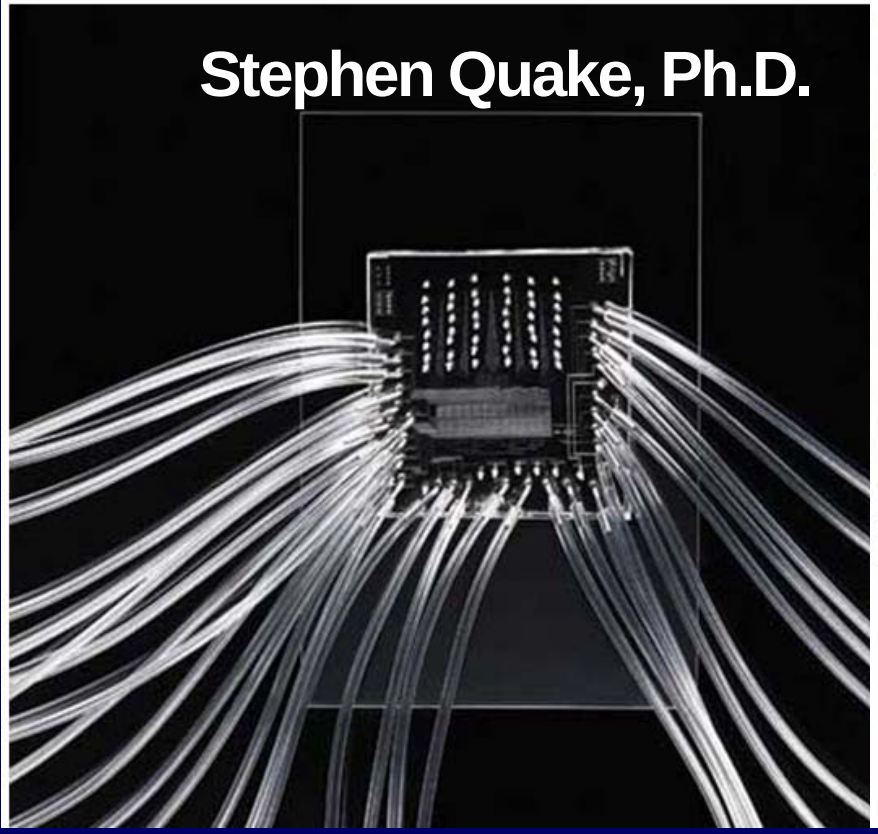
Technology Review:
10 Emerging Technologies 2011

Separating Chromosomes

A more precise way to read DNA will change how we treat disease

MAY/JUNE 2011 | BY INGFEI CHEN

Stephen Quake, Ph.D.



Cancer Genomics

Deciphering the genetics behind the disease

MAY/JUNE 2011 | BY EMILY SINGER

Elaine Mardis, Ph.D.

Document 15

Milwaukee Journal Sentinel Wins Pulitzer Prize for 'One in a Billion' Series

One In A Billion: A boy's life, a medical mystery

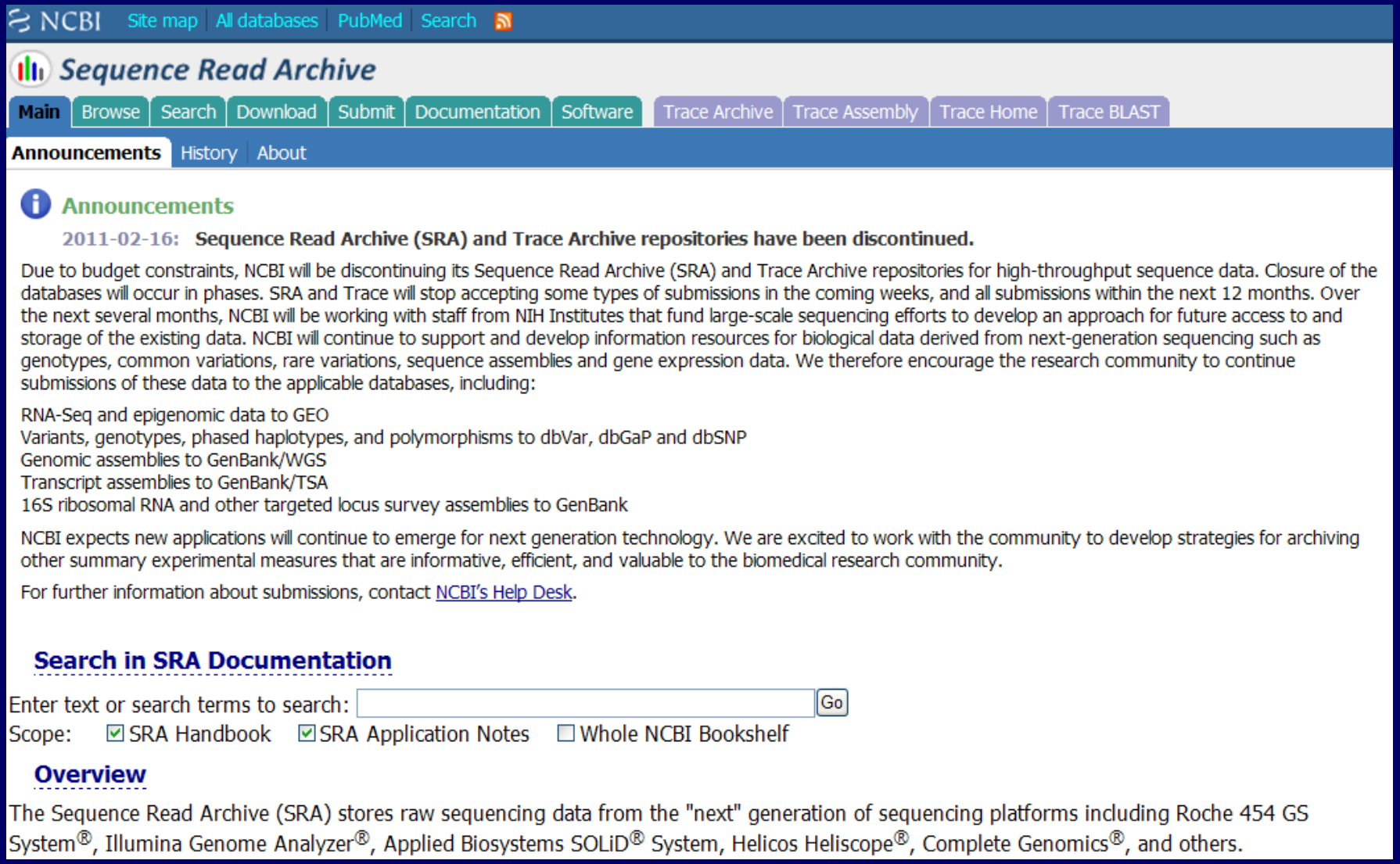
Home » Features » Health and Fitness

Part 1: *A baffling illness*
Faced with a confounding illness, doctors consider an

Nicholas Volker is a little boy with a rare, devastating disease. In a desperate bid to save his life, Wisconsin doctors must decide: Is it time to push medicine's frontier?

Document 16

NCBI's Sequence Read Archive to be Discontinued



NCBI Site map | All databases | PubMed | Search

Sequence Read Archive

Main | Browse | Search | Download | Submit | Documentation | Software | Trace Archive | Trace Assembly | Trace Home | Trace BLAST

Announcements | History | About

Announcements

2011-02-16: Sequence Read Archive (SRA) and Trace Archive repositories have been discontinued.

Due to budget constraints, NCBI will be discontinuing its Sequence Read Archive (SRA) and Trace Archive repositories for high-throughput sequence data. Closure of the databases will occur in phases. SRA and Trace will stop accepting some types of submissions in the coming weeks, and all submissions within the next 12 months. Over the next several months, NCBI will be working with staff from NIH Institutes that fund large-scale sequencing efforts to develop an approach for future access to and storage of the existing data. NCBI will continue to support and develop information resources for biological data derived from next-generation sequencing such as genotypes, common variations, rare variations, sequence assemblies and gene expression data. We therefore encourage the research community to continue submissions of these data to the applicable databases, including:

RNA-Seq and epigenomic data to GEO
Variants, genotypes, phased haplotypes, and polymorphisms to dbVar, dbGaP and dbSNP
Genomic assemblies to GenBank/WGS
Transcript assemblies to GenBank/TSA
16S ribosomal RNA and other targeted locus survey assemblies to GenBank

NCBI expects new applications will continue to emerge for next generation technology. We are excited to work with the community to develop strategies for archiving other summary experimental measures that are informative, efficient, and valuable to the biomedical research community.

For further information about submissions, contact NCBI's Help Desk.

Search in SRA Documentation

Enter text or search terms to search: Go

Scope: ☑ SRA Handbook ☑ SRA Application Notes ☐ Whole NCBI Bookshelf

Overview

The Sequence Read Archive (SRA) stores raw sequencing data from the "next" generation of sequencing platforms including Roche 454 GS System®, Illumina Genome Analyzer®, Applied Biosystems SOLiD® System, Helicos Heliscope®, Complete Genomics®, and others.

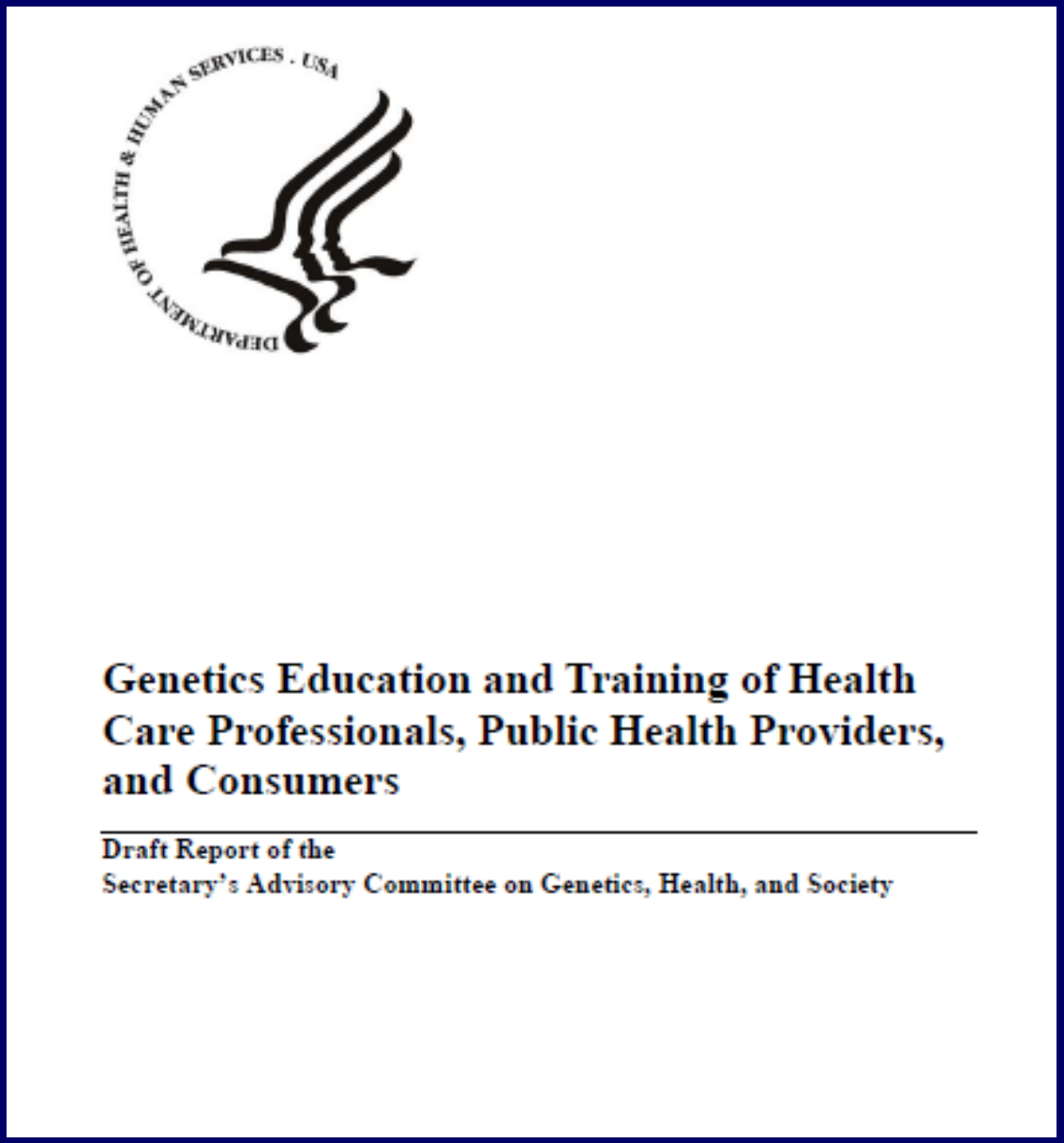
Document 17

CDC Office of Public Health Genomics

- Est. 1997, $8.9M budget in FY2010
- Funded knowledge synthesis, translational research, and population data studies
- Being downsized by >90% FY2011-12

Document 18

SACGHS's Final Report



Available for public comment until June 30, 2011

Document 19

Presidential Commission for the Study of Bioethical Issues

- Emerging technologies for diagnostic and predictive tools
  - Genetics
  - Neuroimaging
- International Human Subjects Research

Document 20

International Rare Disease Research Consortium (IRDiRC)

- Workshop April 2011
- Participants: funding agencies, patient advocacy groups, researchers, industry, and regulatory agencies
- Consortium goals: deliver by 2020 diagnostic tests for most rare diseases and 200 new therapies for rare diseases

Document 21

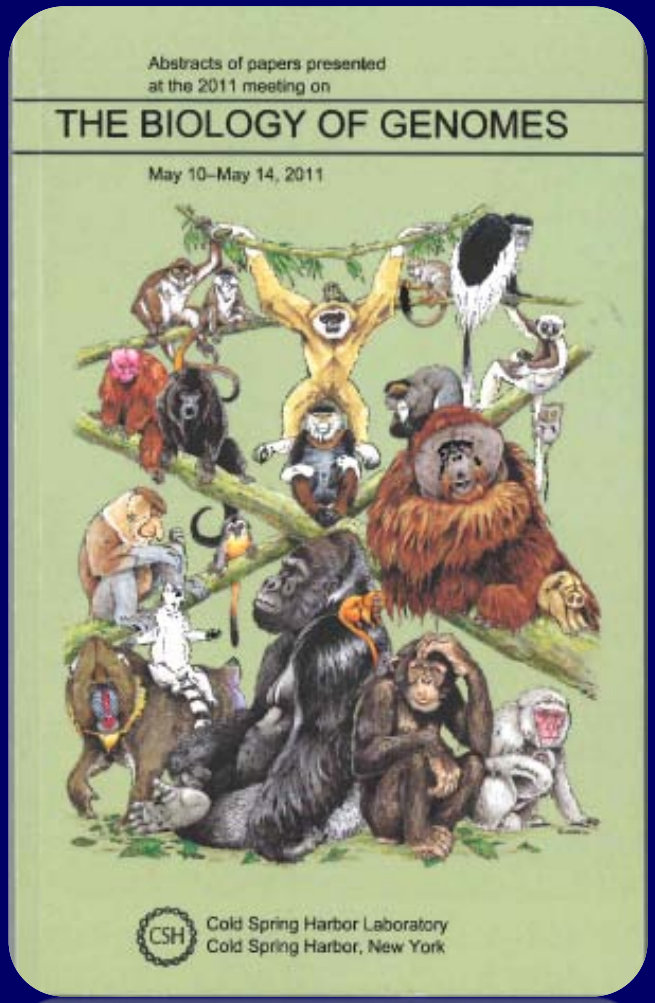
New Journal: *Gigascience*

- **BGI and Biomed Central to launch a new journal for large-scale biology called *GigaScience* in 2011**
- **Will publish and serve as a data repository for studies generating large biomedical data sets, including genomics studies**
- **Will provide DOI numbers for large datasets so that they can 'count' as publications**

2011 ELSI Congress

- April 2011; >350 attendees (8 countries)
- Plenary talks (Eric Green, David Williams, Pearl O'Rourke, Kathy Hudson)
- 19 panels, 79 individual papers, 12 focused workshops, 54 posters
- Themes: Genomic Data Sharing, Return of Clinical & Research Results, Behavioral Genetics, Health Equity, Race & Genomics, IP, HMP, Epigenomics
- Prominence of CEER trainees from diverse disciplines and population groups

2011 Biology of Genomes Meeting: Cold Spring Harbor Laboratory



NHGRI Genome Advance of the Month

Genomic Advance of the Month: The Biology of Living Longer

Share Print Comments

January 2011

Ronald DePinho talks about aging process on The Colbe

OK, it's a mouse model aging and extend lifespa of an enzyme called telo

Withhold Tamoxifen and control mice that expres tissue atrophy, especiall lacking normal telomera

Genome Advance of the Month: Undiagnosed Diseases Program (UDP) Discovers a New Disease

Share Print Comments

See Also:

February 2011

By Jonathan Gitlin, P Science Policy Analy

From left to right: Research M.D.; patients with ACDC P Benge; UDP Director Willian and Researcher Cynthia St.

advance medical knowled accepted, travel to Bethe

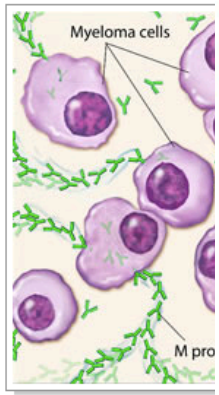
Conceived by William A. (is only possible because i Some 1,500 different stu stating the obvious, but individuals with severe h such individuals.

Genome Advance of the Month: Sequencing Insights Into Multiple Myeloma

Share Print Comments

March 2011

By Jonathan Gitlin, P Science Policy Analy



Drawing of a Myeloma cell making M proteins. M prote created by a Myeloma cell.

The study, published in t organized by Todd R. Gol made several new discov a type of immune cell ca that number die from th

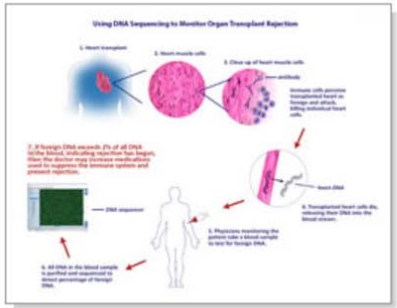
Genome Advance of the Month

Comments Share Print

Using DNA Sequencing to Monitor Organ Transplant Rejection

April 2011

By Jonathan Gitlin, Ph.D.
Science Policy Analyst



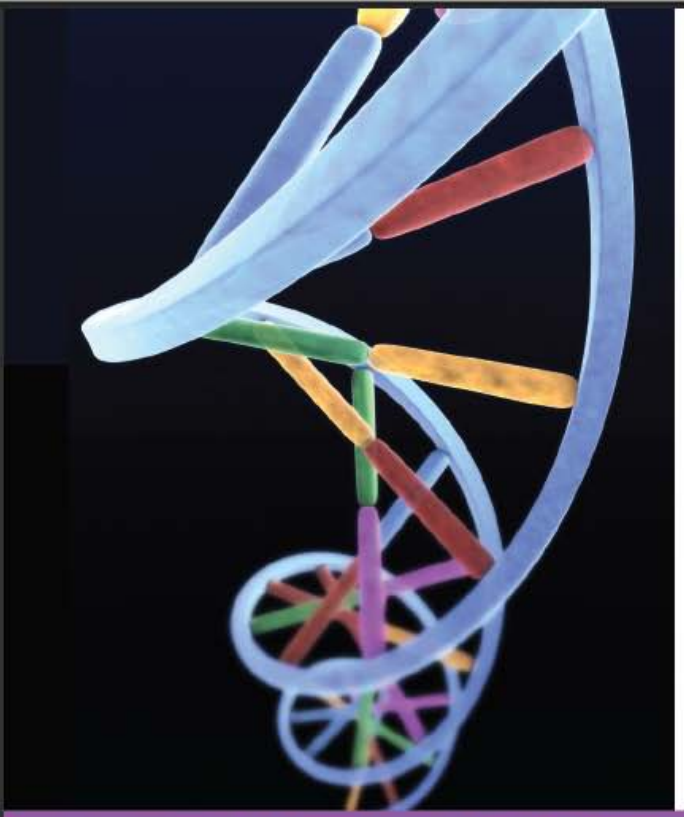
Using DNA Sequencing to Monitor Organ Transplant Rejection. Click on the image to enlarge.

For people who have received an organ transplant — a heart or a kidney, for example — detecting rejection early can significantly improve their long-term health, even survival. But until now, doctors have had a hard time monitoring the health of transplanted organs; detecting rejection has required invasive — and risky — biopsies. A novel application of genome-sequencing technology developed by a group of researchers at Stanford University may soon solve that problem.

Thomas Snyder and his colleagues demonstrated that it is possible to identify organ rejection noninvasively by detecting DNA from the transplanted organ — which is essentially foreign genetic material — released into the recipient's bloodstream when the patient's immune system attacks the transplanted organ as if it were a dangerous infection. This is yet another example of how the genome-analyzing technologies developed during Human Genome Project, and ever-decreasing costs for DNA sequencing, are beginning to impact the field of medicine.

The history of organ transplantation goes back a long way, but until the development of immunosuppressive drugs, allotransplantation (transplanting tissue from a donor with different DNA) always ended in failure. Basically, the recipient's immune system recognizes the donor tissue as foreign and mounts a massive attack to destroy the invader (the transplanted organ), resulting in rejection of the organ. Understanding the mechanism behind rejection and developing drugs such as cyclosporin to suppress it, enabled surgeons to transplant organs with much greater success. Even though transplantation patients now take these powerful immunosuppressants, the drugs do not always prevent tissue rejection.

Document 23



Economic Impact of the Human Genome Project

How a $3.8 billion investment drove $796 billion in economic impact, created 310,000 jobs and launched the genomic revolution

Prepared by Battelle Technology Partnership Practice

May 2011

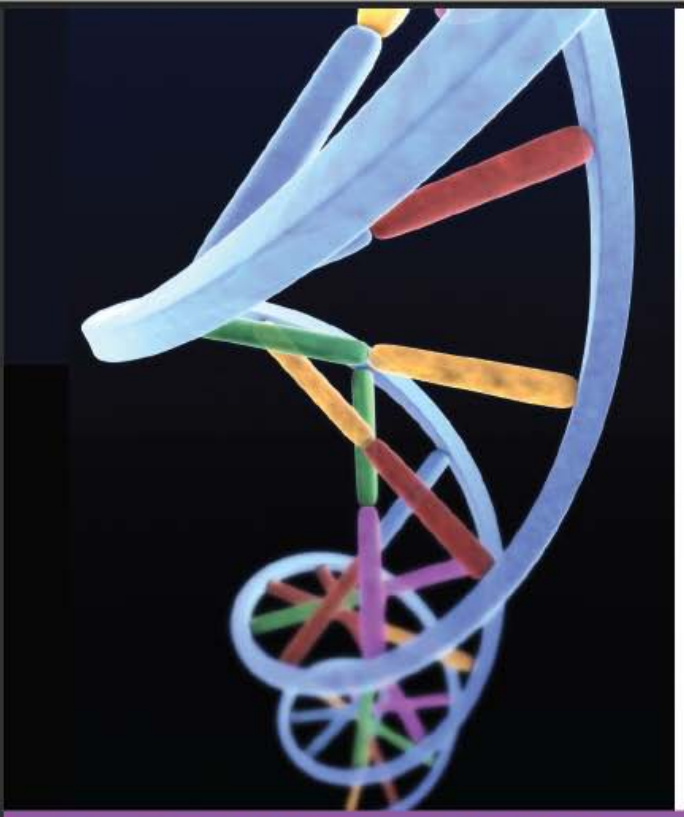
Battelle Memorial Institute Study: Key Findings

- **Between 1988 and 2010, the Federal government invested $5.6 billion (in 2010 dollars) in the Human Genome Project and generated for the American economy:**

  $796 billion in economic output
  $244 billion in personal incomes
  3.8 million job-years of employment
  $141 returned for every $1 invested

- **In 2010 alone, genomics-related projects produced:**

  $67 billion in U.S. economic output
  $20 billion in personal income for Americans
  310,000 jobs for the U.S. economy
  $3.7 billion in federal taxes
  $2.3 billion in U.S. state and local taxes



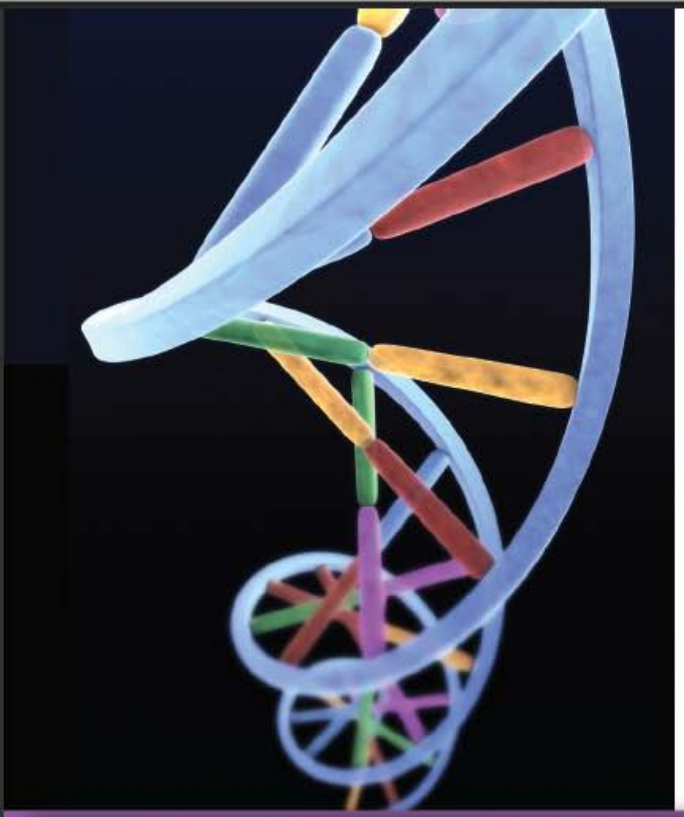
Economic Impact of the Human Genome Project

How a $3.8 billion investment drove $796 billion in economic impact, created 310,000 jobs and launched the genomic revolution

Prepared by Battelle Technology Partnership Practice

May 2011

“The HGP is arguably the single most influential investment to have been made in modern science and a foundation for progress in the biological sciences moving forward.”



Economic Impact of the Human Genome Project

How a $3.8 billion investment drove $796 billion in economic impact, created 310,000 jobs and launched the genomic revolution

Prepared by Battelle Technology Partnership Practice

May 2011

“The impacts of the human genome sequencing are just beginning— large-scale benefits in human medicine, agriculture, energy, and environment are still in their early stages. The best is truly yet to come.”

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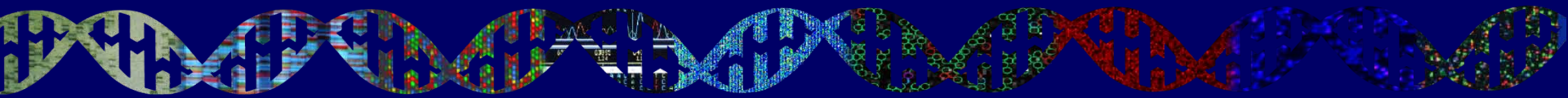
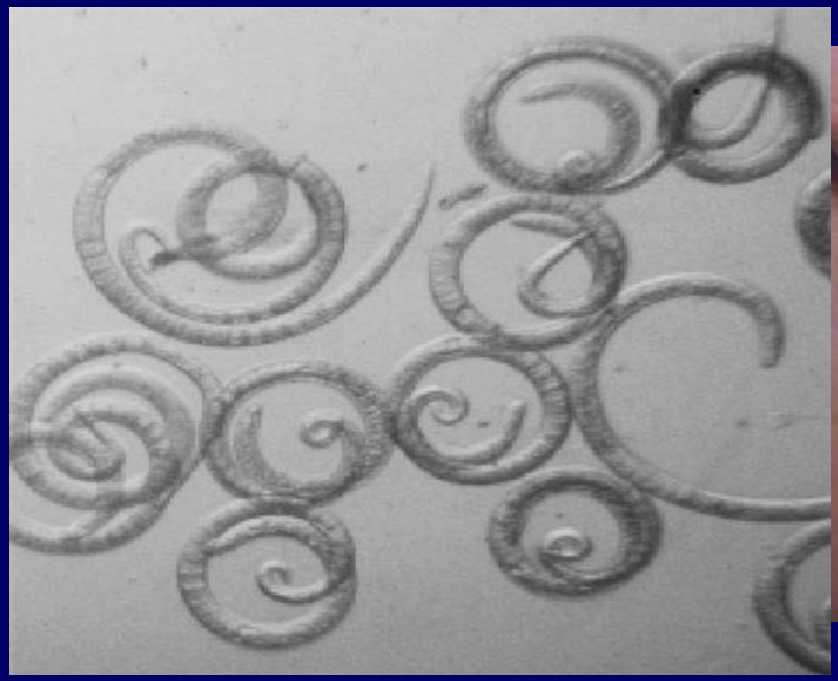
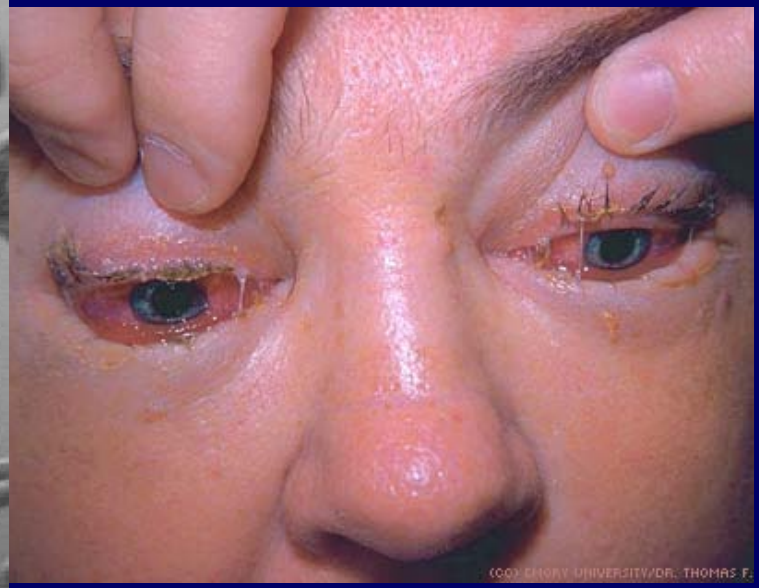


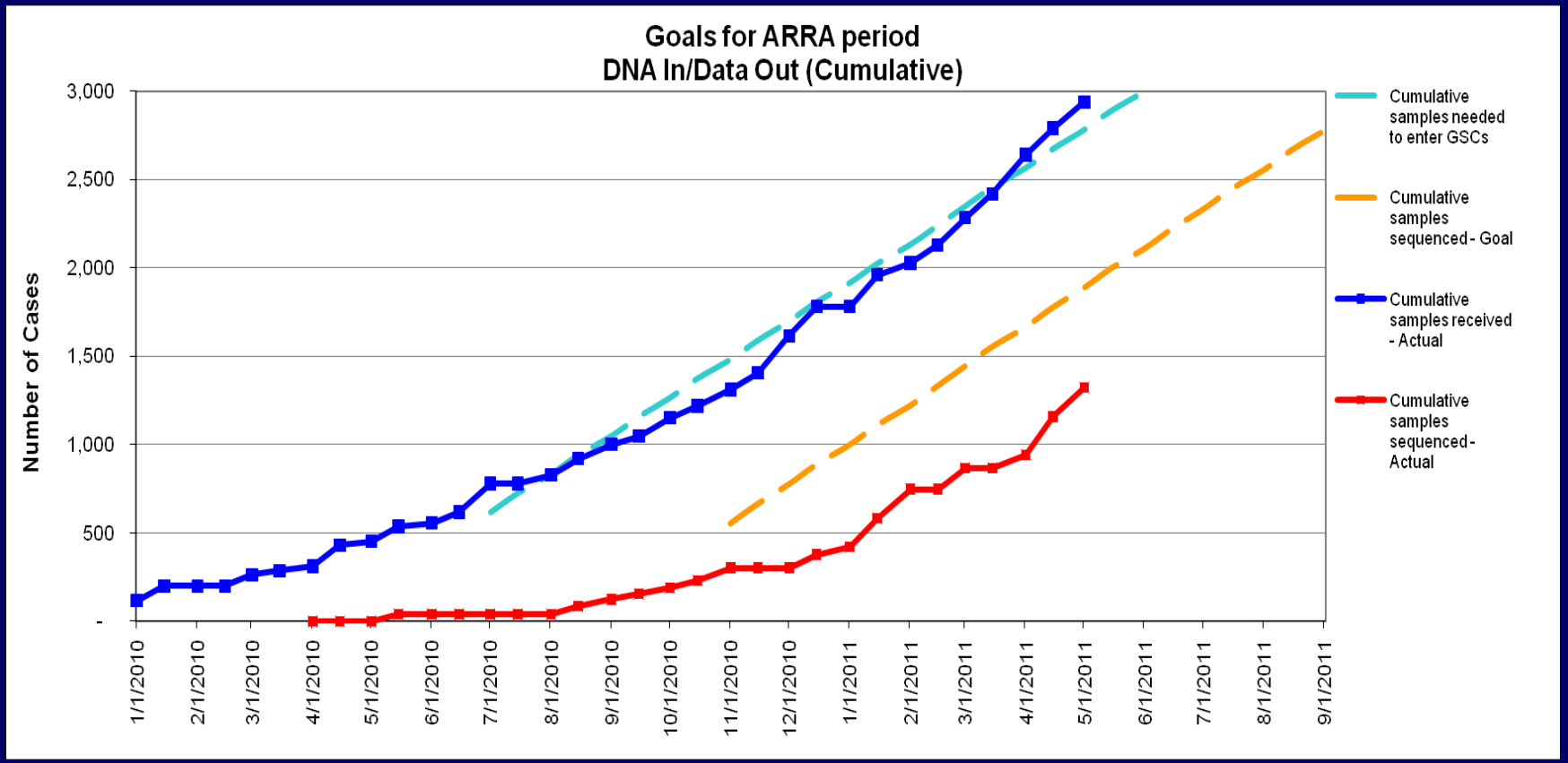
Large-Scale Genome Sequencing Program: Organisms

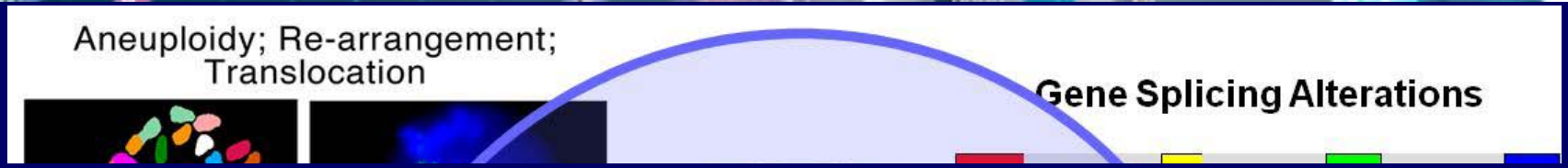
- **Draft genome sequence of parasitic nematode *Trichinella spiralis***



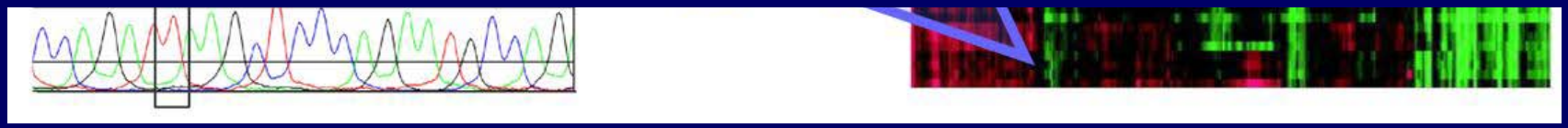


- **Post-pilot production approaching the ARRA goal of 3,000 tumor/normal pairs, representing 22 tumor projects by Sept '11**





- **Ovarian cancer manuscript *in press***
- **Major analyses underway for projects in:**
  - **AML (203 cases)**
  - **Colorectal (332 cases)**
  - **Breast (647 cases, multiple subtypes)**
  - **Kidney clear cell (461 cases)**
  - **Endometrial (298 cases)**



1000 Genomes

A Deep Catalog of Human Genetic Variation

- **Data Sets:**

  Low-coverage sequence data on 1150 samples; initial variant calls on 1094 samples

  Exome sequence data on 1000 samples; initial variant calls on 458 samples

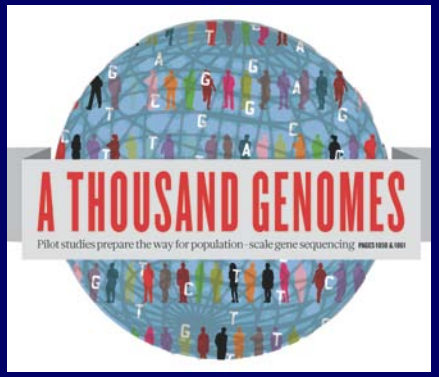
  OMNI 2.5-million SNP genotype data on 1500 samples, including trio children

- **Variants Found:**

  ~39 million SNPs

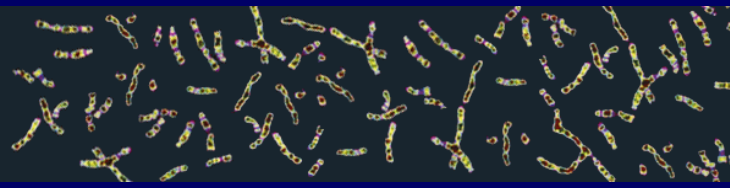
  ~100,000 indels

  ~84,000 structural variants



1000 Genomes

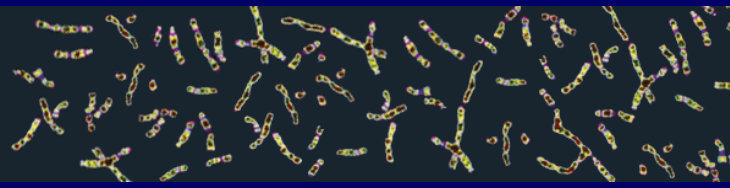
A Deep Catalog of Human Genetic Variation



- **The project is finding 95% of SNPs at 2% frequency by analyzing 1094 samples (short of goal); but expects to find 95% of SNPs at 1% frequency by analyzing all 2500 samples**
- **The haplotypes are accurate enough for imputation in disease studies**
- **The DCC has a new browser, allowing users to get 'slices' of the data (such as genomic regions of interest)**

1000 Genomes

A Deep Catalog of Human Genetic Variation



- **Project Timeline:**

  Phase 1: 1150 samples currently sequenced, manuscript by the end of 2011 on data set integrating all the variant types

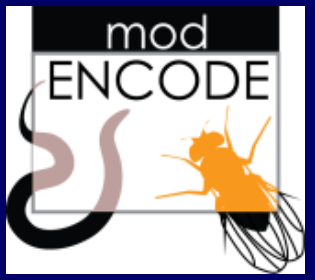
  Phase 2: 571 samples in by May 2011, sequenced by Fall 2011

  Phase 3: 779 samples in by March 2012, sequenced by Fall 2012

DNA Sequencing Technology

- 2011 Sequencing Technology Program Grantee Meeting in San Diego
- 1-day public meeting

ENCODE & modENCODE



- ENCODE and modENCODE Consortia are working on integrative analysis papers
- The annual Joint ENCODE and modENCODE Consortia meeting will be held later in May
- Upcoming presentation by Elise Feingold and Peter Good

OPEN ACCESS Freely available online

PLoS BIOLOGY

A User's Guide to the Encyclopedia of DNA Elements (ENCODE)

The ENCODE Project Consortium¶*

Abstract

The mission of the Encyclopedia of DNA Elements (ENCODE) Project is to enable the scientific and medical communities to interpret the human genome sequence and apply it to understand human biology and improve health. The ENCODE Consortium is integrating multiple technologies and approaches in a collective effort to discover and define the functional elements encoded in the human genome, including genes, transcripts, and transcriptional regulatory regions, together with their attendant chromatin states and DNA methylation patterns. In the process, standards to ensure high-quality data have been implemented, and novel algorithms have been developed to facilitate analysis. Data and derived results are made available through a freely accessible database. Here we provide an overview of the project and the resources it is generating and illustrate the application of ENCODE data to interpret the human genome.

***PLoS Biol* (2011)**

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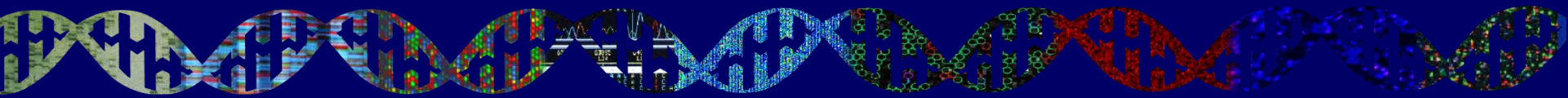


Human Microbiome Project

- Anticipated surge in paper submissions
- Pre-publication release of HMP datasets

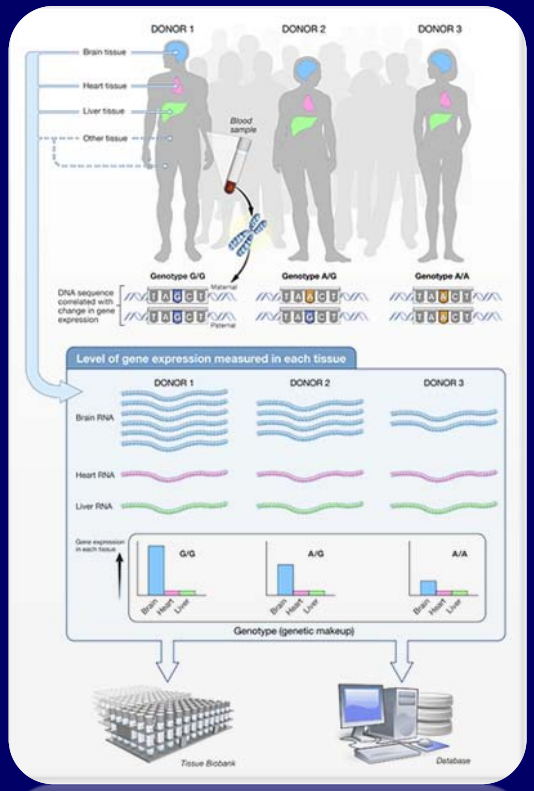
- Meetings:
  - International Human Microbiome Congress (March 2011, Vancouver)

  - Brainstorming meeting on future initiative (April 2011, Bethesda)
  - ASM session to feature HMP (May 2011, New Orleans)

- Upcoming presentation by Lita Proctor

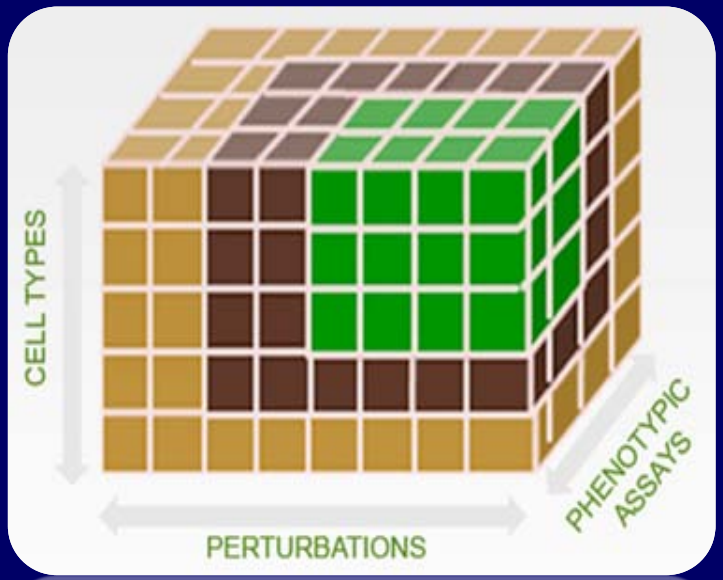
Genotype-Tissue Expression (GTEx)

- 1 Lab/Coordinating Center & 3 Biospecimen Source Sites
- Updates on donor pipeline
  - 4 post-mortem donors collected and analyzed; 6 more by the end of May
  - Early molecular data encouraging
- Upcoming meeting: June 2011
  - Involve PIs, External Scientific Panel, R01 grantees, & genome browser groups
- Development of a donor brochure & website



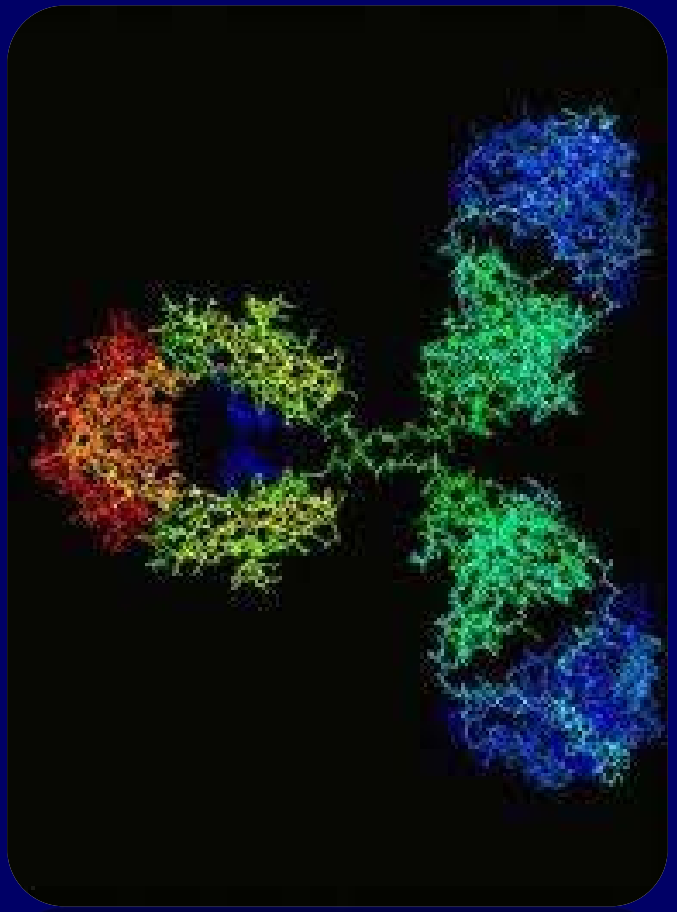
Library of Integrated Network-based Cellular Signatures (LINCS)

- LINCS Spring Meeting: March 2011
- Fall Consortia Meeting: October 2011
- RO1 outreach supplements awarded
- Review for U01 applications will occur in Summer 2011
- Paper published in May issue of *Nature Methods*



Protein Capture Reagents Program

- Applications received for Production and Technology Development U54 awards
- Review of applications will occur in Summer 2011
- NACHGR will perform the Council review for the Production RFA and funding plan in Fall 2011



Human Heredity and Health in Africa (H3Africa)

- Meeting in Cape Town to discuss white paper (March 2011)

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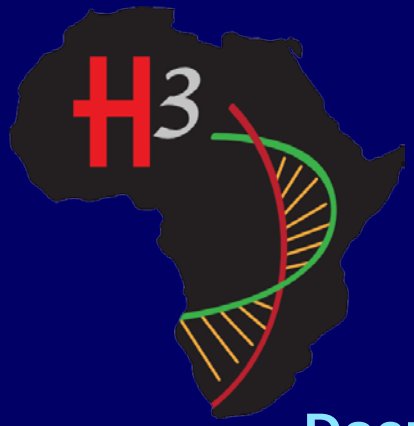
Human Heredity and Health in Africa (H3Africa)

- April: Presentation to IC Directors to try to get additional support for projects
- Released Notices of NIH's Intention to Release an FOA to solicit applications for a Bioinformatics Network and for a Repository
- June: Presentation to Council of Councils for Concept Clearance
- July: Publication of first FOAs?



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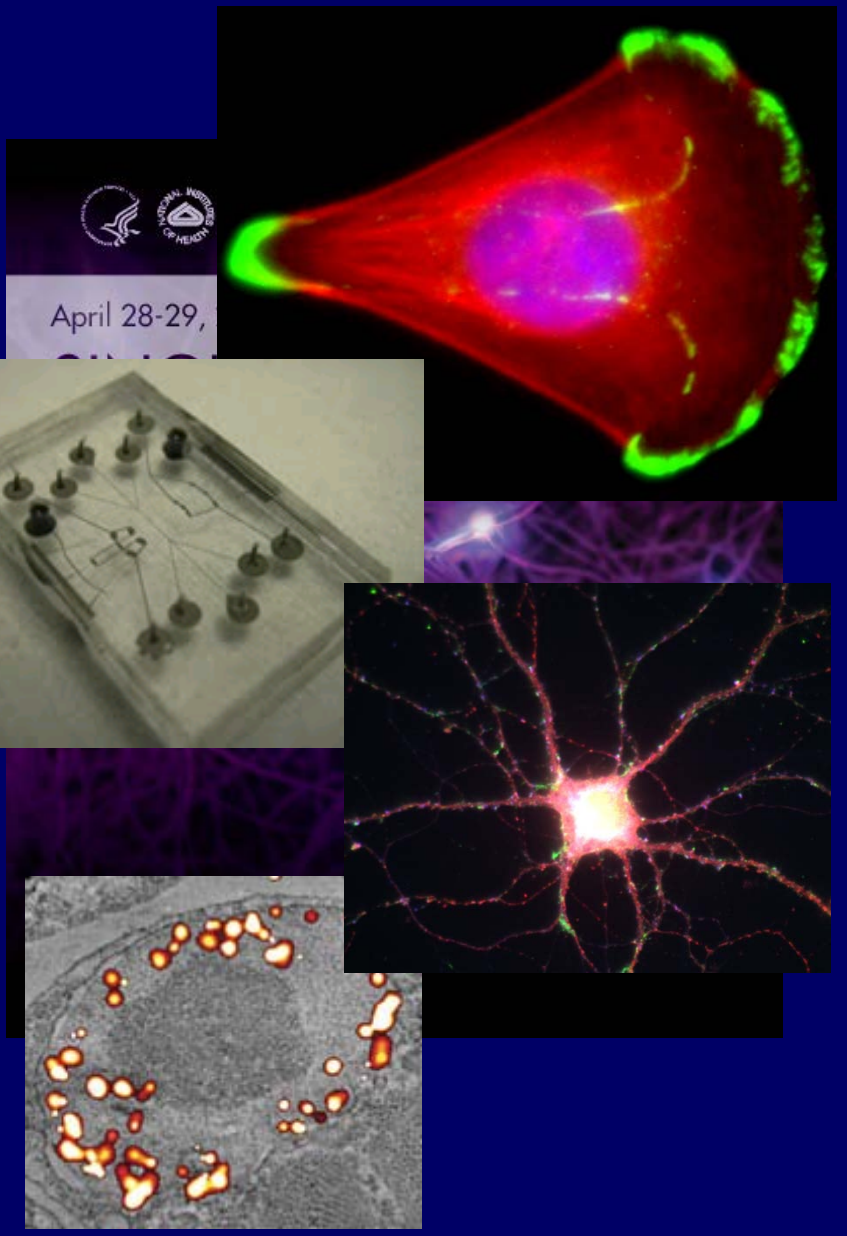
Single Cell Biology Workshop

- Concept proposed at "Big Think" Meeting
- RFI in February
- Workshop in April
  - 22 'practitioners'
  - Driving biomedical problems & limitations of current technologies/assays
- Proposal to CF in May



Innovation Brainstorm Meeting: Transforming Discovery Into Impact

- 25 junior investigators nominated by IC Directors
- Discussion focused on a set of high-impact papers selected by attendees

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eMERGE Network: Recent Publications

Genetics

Identification of Genomic Predictors of Atrioventricular Conduction

Using Electronic Medical Records as a Tool for Genome Science

Joshua C. Denny, MD, MS*; Marylyn D. Ritchie, PhD*; Dana C. Crawford, PhD; Jonathan S. Schildcrout, PhD; Andrea H. Ramirez, MD; Jill M. Pulley, MBA; Melissa A. Basford, MBA; Daniel R. Masys, MD; Jonathan L. Haines, PhD; Dan M. Roden, MD

Background—Recent genome-wide association studies in which selected community populations are used have identified genomic signals in *SCN10A* influencing PR duration. The extent to which this can be demonstrated in cohorts derived from electronic medical records is unknown.

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rs6800541, rs6795970, rs6798015, and rs7430477 linked to *SCN10A* associated with PR interval ($p = 5.73 \times 10^{-7}$ to 1.78×10^{-6}).

Conclusions—This genome-wide association study confirms a gene heretofore not implicated in cardiac pathophysiology as a modulator of PR interval in humans. This study is one of the first replication genome-wide association studies performed with the use of an electronic medical records–derived cohort, supporting their further use for genotype-phenotype analyses. (***Circulation***. **2010;122:2016-2021.**)

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NEJM Genomic Medicine Series

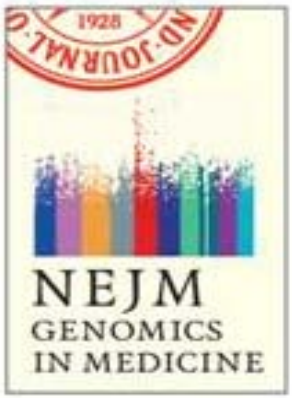
REVIEW ARTICLE

GENOMIC MEDICINE

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

Genomics and the Continuum of Cancer Care

Ultar



REVIEW ARTICLE

GENOMIC MEDICINE

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

Genomics and Drug Response

Liewei Wang, M.D., Ph.D., Howard L. McLeod, Pharm.D., and Richard M. Weinshilboum, M.D.

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GENETICS/GENOMICS COMPETENCY CENTER FOR EDUCATION

Competencies & Guidelines

Search Educator Resources:

Genetic Counselor	Nurse	Physician Assistant
		
Search	Search	Search

Share Your Resources

What you can do on this site:

1. **Search for genetics/genomics resources to use in your classroom**
   Find websites, download PDFs, locate courses–enhance your class content with genetic/genomic resources for Nurses, Physician Assistants, and Genetic Counselors.
2. **Review the Competency Guidelines and Curriculum Map**
   Need to match your classroom genomics teaching to competencies? View the guidelines and the curriculum map to find the right resources to download.
3. **Share resources, activities, and assessments to be included on this site**
   Do you have activities, resources or assessments you would like to share? Submit websites, PDFs or any other resources to include in the curricular materials.

The **Genetics/Genomics Competency Center for Education** provides links to curricular materials and resources for educators of Genetic Counselors, Nurses, and Physician Assistants.

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OPCE Staff Member Award

Rocky Rackover to receive 2011 PAragon Outstanding Physician Assistant of the Year Award

DNA DAY 11 Activities

- **2011 DNA Day Chatroom**
  - **April 15 from 8:00 am to 6:00 pm EST**
  - **45 experts on site and remotely answered questions**
  - **1031 Questions received and 782 answered**
- 

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Johns Hopkins Center for Talented Youth Family Academic Program

- NHGRI hosted 200 students and parents from the Johns Hopkins Center for Talented Youth
  - NHGRI staff discussed the Neanderthal Genome and the Human Microbiome with participants
  - Students did hands-on activities
  - Parents toured National Library of Medicine and learned about genetic counseling

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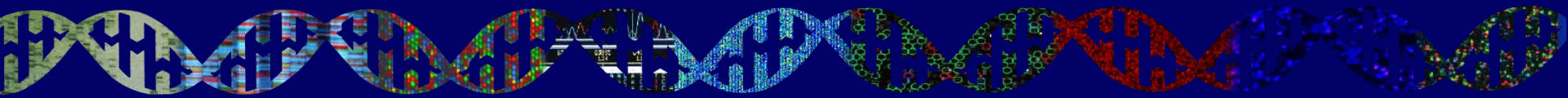

NISC Director Search

NISC Director

NIH Intramural Sequencing Center, National Human Genome Research Institute

The National Human Genome Research Institute (NHGRI), a major research component of the National Institutes of Health (NIH) and the Department of Health and Human Services (DHHS), seeks to identify an outstanding Director to lead the NIH Intramural Sequencing Center (NISC), located in Rockville, Maryland. The NISC Director leads a multi-disciplinary genomics facility that emphasizes the generation and analysis of DNA sequence. NISC brings together diverse and unique scientific expertise to perform state-of-the-art genome sequencing and sequence analysis for basic and translational research projects. The NISC Director has the responsibility for an annual budget exceeding $7 million and a staff of ~40. In addition to providing scientific and administrative leadership of this premier research enterprise, the Director is expected to be an internationally recognized, highly collaborative, and accomplished genomics researcher.

Applicants must possess a doctoral-level scientific degree. The applicant must have extensive experience in genomics research, computational biology, and large-scale DNA sequencing; this should include a productive track record of high profile publications. S/he must have proven experience in directing and managing a scientific research program, with well-honed administrative and interpersonal skills to meet the demands of both research and program direction.

Salary is competitive and will be commensurate with candidate's experience. A full Federal benefit package is available, including retirement, health and life insurance, long-term care insurance, annual and sick leave, and the thrift savings plan (401K equivalent). Appropriate support for this program will be provided and exceptional candidates may be eligible for tenure.

Interested applicants should submit a cover letter that includes a brief description of research and administrative experience, a current curriculum vitae and bibliography, names and contact information of three references, and a brief written vision for leading NISC. Questions about the position and applications themselves should be sent to Ms. Ellen Rolfes via email at ellenr@exchange.nih.gov.

Applications must be submitted by May 15, 2011.

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

NATIONAL HUMAN GENOME RESEARCH INSTITUTE

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

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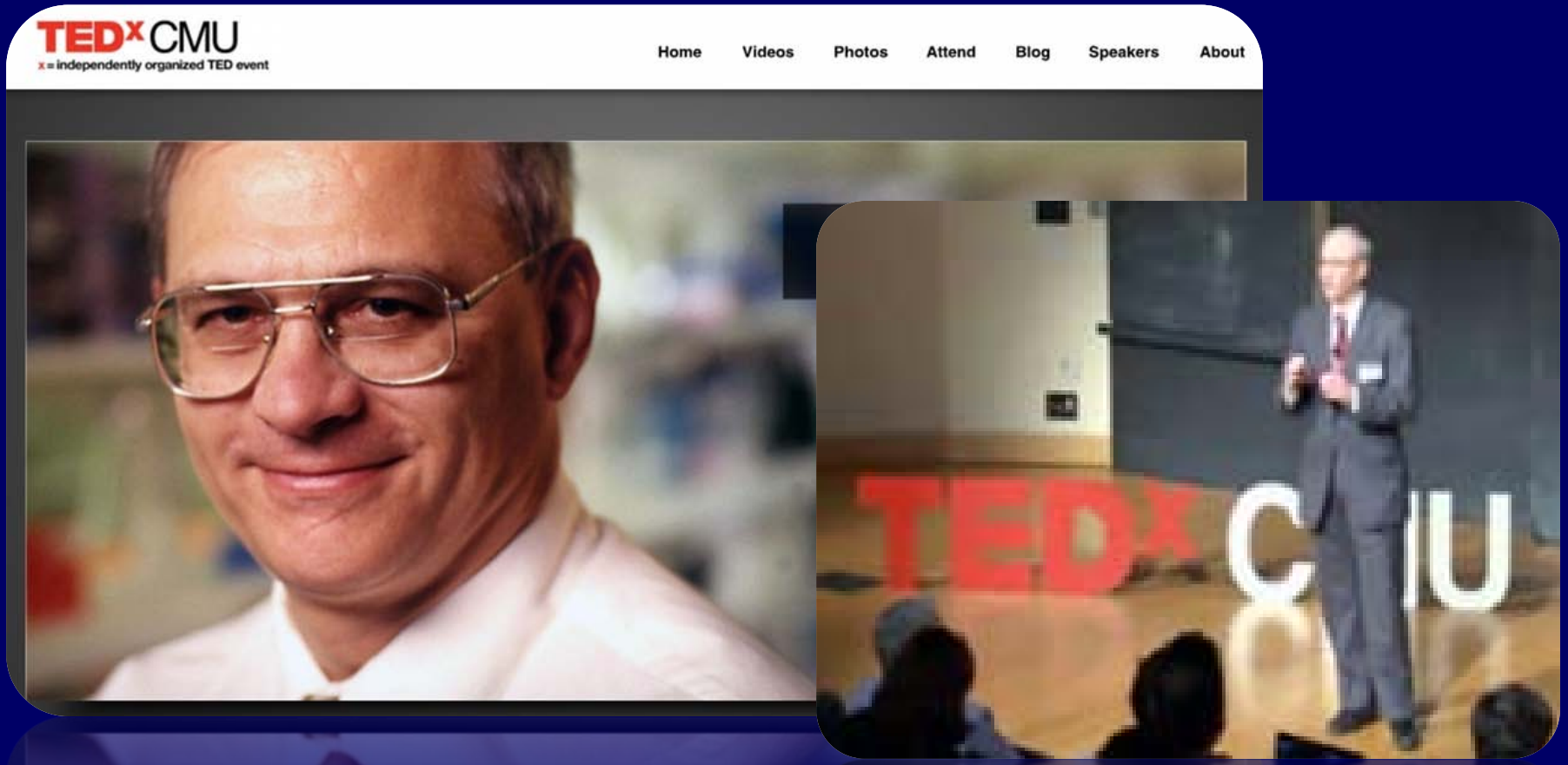
Rare Disease TED^{x} Talk



Dr. Bill Gahl opined on the need to speed up approvals of drugs for rare diseases' drugs during a recent TED^{x} talk

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Public Service Award Finalist: Bill Gahl

Bill Gahl has been named a finalist for a 2011 Samuel J. Heyman Service to America Award

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Undiagnosed Diseases Program Awarded Society for Clinical and Translational Science Team Science Award

NHGRI Intramural Research Highlights

PNAS

Periodic fever, aphthous stomatitis, pharyngitis, and adenitis (PFAPA) is a disorder of innate immunity and Th1 activation responsive to IL-1 blockade

Silvia Stojanov[a,b,1], Sivia Lapidus[a,1], Puja Chitkara[a], Henry Feder[c], Juan C. Salazar[c], Thomas A. Fleisher[d], Margaret R. Brown[d], Kathryn M. Edwards[e], Michael M. Ward[a], Robert A. Colbert[a], Hong-Wei Sun[a], Geryl M. Wood[a,f], Beverly K. Barham[a,f], Anne Jones[a,f], Ivona Aksentijevich[a,f], Raphaela Goldbach-Mansky[a], Balu Athreya[g], Karyl S. Barron[h], and Daniel L. Kastner[a,f,2]

NATURE GENETICS | LETTER

Exome sequencing identifies GRIN2A as frequently mutated in melanoma

Xiaomu Wei, Vijay Walia, Jimmy C Lin, Jamie K Teer, Todd D Prickett, Jared Gartner, Sean Davis, NISC Comparative Sequencing Program, Katherine Stemke-Hale, Michael A Davies, Jeffrey E Gershenwald, William Robinson, Steven Robinson, Steven A Rosenberg & Yardena Samuels

PEDIATRICS®

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

Parents' Attitudes Toward Pediatric Genetic Testing for Common Disease Risk

Kenneth P. Tercyak, Sharon Hensley Alford, Karen M. Emmons, Isaac M. Lipkus, Benjamin S. Wilfond and Colleen M. McBride

Pediatrics published online Apr 18, 2011;
DOI: 10.1542/peds.2010-0938

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

Special Thanks!


NHGRI