NIH National Human Genome Research Institute

DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

September 2013

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


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National Human Genome Research Institute
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Home › About › Institute Advisors › National Advisory Council for Human Genome Research › September 2013 NACHGR Agenda and Documents › September 2013 Documents
Director's Report Related Documents: September 2013
Director's Report
Director's Report
No.
Documents
1
NHGRI-Smithsonian Exhibition
Genome: Unlocking Life's Code Exhibition Website [unlockinglifescode.org]
VOA Video
GenomeWeb [genomeweb.com]
Wall Street Journal [online.wsj.com]
Washington Post [washingtonpost.com]
New York Times [nytimes.com]
2
APA's Society for the Psychological Study of Ethnic Minority Issues Charles and Shirley Thomas Award [apa.org]
3
Arrival of New Director, National Institute of General Medical Sciences [nigms.nih.gov]
genome.gov/DirectorsReport
Document #

Open Session Presentations

Major Presentations:

- **Assoc. for Molecular Pathology et al. v Myriad Genetics Inc. et al.**

  Arti Rai

- **Data Sharing and the HeLa Genome Sequence**

  Kathy Hudson

Recent NHGRI Meetings:

- **Report of the Genomic Medicine Working Group**

  Teri Manolio

Open Session Presentations

Project Updates:

- **The NHGRI-Smithsonian Genome Exhibition**
  Vence Bonham
- **The NHGRI Bioinformatics Research Portfolio**
  Vivien Bonazzi
- **NHGRI Training & Career Development Program**
  Heather Junkins

Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

IV. NHGRI Extramural Research Program

V. NIH Common Fund/Trans-NIH

VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

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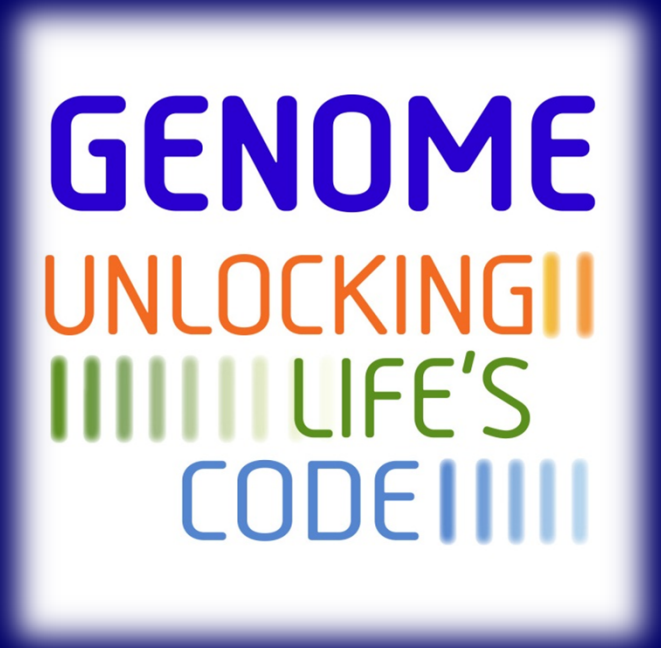
NHGRI-Smithsonian Genome Exhibition

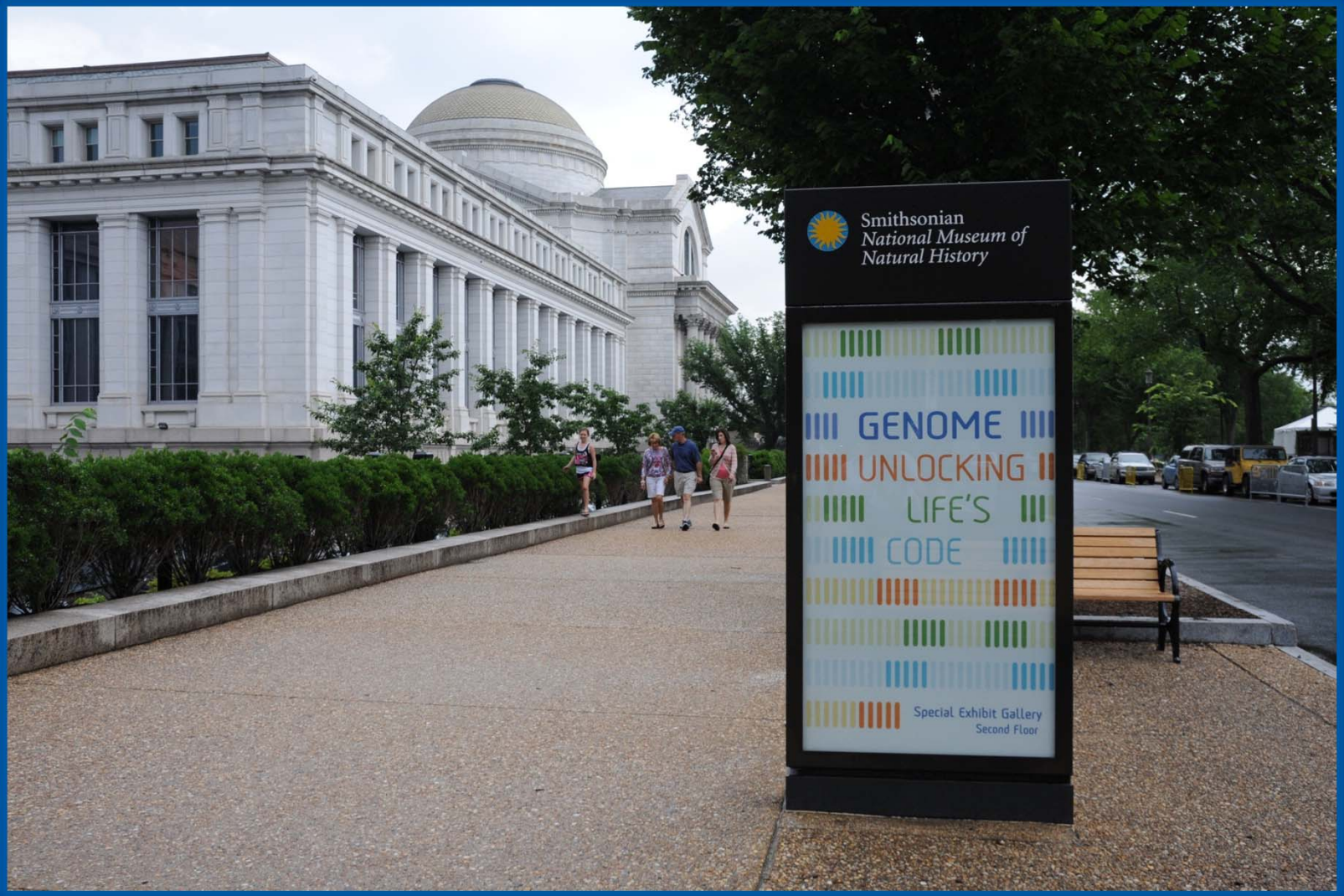


NIH National Human Genome Research Institute

Smithsonian *National Museum of Natural History*

Document 1

Exhibition Opening: June 14, 2013



Document 1

Genome Exhibition Visits

Genome Exhibition Visits

New ASHG/NHGRI Policy Fellow

Kate Donigan, Ph.D.

Extramural Staff Departure

Brad Ozenberger, Ph.D.

Ongoing NHGRI Recruitments

Director, Division of Genomics and Society

ELSI Program Director

Bioinformatics Program Directors

Chief, Genomics Healthcare Branch

APA's Society for the Psychological Study of Ethnic Minority Issues Charles and Shirley Thomas Award

AMERICAN PSYCHOLOGICAL ASSOCIATION

Vivian Ota Wang, Ph.D.

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Arrival of New Director, National Institute of General Medical Sciences

NIGMS

Jon Lorsch, Ph.D.

Document 3

Secretary Sebelius Visits NIH

Visits of Congressional Members to NIH

Sen. Harry Reid (D-NV)
Senate Majority Leader

Rep. Michael Burgess (R-TX)
House – Energy and Commerce

Rep. Scott Peters (D-CA)
House – Sci. and Tech.

"I toured the clinic where the best medical researchers in the country are trying to solve the world's most elusive medical mysteries – diseases that have yet to be identified let alone cured."

– Sen. Harry Reid *(Senate Floor, June 20 2013)*

Document 4

Congressional Briefing on Genomics

DISCOVERING KNOWLEDGE CONQUERING DISEASE

The Ad Hoc Group for Medical Research
Luncheon Briefing

THE 10TH ANNIVERSARY OF THE HUMAN GENOME PROJECT:

A DECADE OF TRANSFORMATIVE RESEARCH

Surgeon General Steps Down

Regina Benjamin, M.D., M.B.A.

Document 5

National Science Foundation
Director Nomination

France Córdova, Ph.D.

Document 6

Office of Science and Technology Policy Associate Director Nomination

Jo Handelsman, Ph.D.

Document 7

NIH Fiscal Year 2014 Appropriations

- Regular appropriations process will not be completed
- Continuing Resolution (CR) anticipated

	FY 2013 Actual (after sequester)	President FY 2014	House FY 2014	Senate FY 2014
NIH	$29.1B	$31.3B	-	$30.955B
NHGRI	$483M	$517M	-	$514M

Document 8

NHGRI Consequences of the Sequester

Bioinformatics Resources and Analysis Research Portfolio
REDUCED BY $5M

ENCODE RFA
REDUCED BY 16%

Genomics of Gene Regulation RFA
DELAYED FROM FISCAL YEAR 2012 TO 2014

Genomic Sequencing & Newborn Screening Disorders RFA
REDUCED BY 50%

Genomic Medicine Pilot Demonstration Projects RFA
REDUCED BY 20%

Intramural Research Program
NUMEROUS EXAMPLES OF DETRIMENTAL CUTS

Other Consequences of the Sequester

BD2K Data Catalog
REDUCED IN FISCAL YEAR 2014

BD2K Framework for Community-based Development of Data and Metadata Standards
REDUCED IN FISCAL YEAR 2014

BD2K Training
DEFERRED UNTIL FISCAL YEAR 2015

BD2K Software RFA
DEFERRED UNTIL FISCAL YEAR 2015

BD2K NIH-Initiated Centers RFA
DEFERRED UNTIL FISCAL YEAR 2015

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HeLa Whole-Genome Data Access

- NIH announced new policy for access to HeLa cell whole-genome data in August
- Presentation by Kathy Hudson later in the Open Session

Document 9

White House Open Science 'Champions' Highlight Genomic Data Pioneers

Atul Butte, M.D., Ph.D.
Atul Butte is a pediatrician, geneticist, computer scientist, and entrepreneur at Stanford University and the Lucile Packard Children's Hospital. Atul's lab at Stanford builds and uses...

David Altshuler, M.D., Ph.D.
Endocrinologist and human geneticist David Altshuler is one of four founding members of the Broad Institute of Harvard and MIT and serves as the Institute's Deputy Director and Chief Academic...

David J. Lipman, M.D.
In his 24 years as the founding director of the National Center for Biotechnology Information (NCBI) at the National Institutes of Health's National Library of Medicine (NLM), Dr. Lipman has had a...

Stephen Friend, M.D., Ph.D.
Dr. Stephen Friend is a world leader in efforts to make large scale, data-intensive biology more openly accessible to citizens and the entire research community in order to accelerate scientific...

John Quackenbush, Ph.D.
John Quackenbush is Professor of Biostatistics and Computational Biology at the Dana-Farber Cancer Institute and the Harvard School of Public Health. Since the Human Genome Project began in the...

Document 10

Warren Alpert Foundation Prize

David Botstein Ph.D.

Ronald Davis Ph.D.

David Hogness Ph.D.

WARREN ALPERT FOUNDATION

Document 11

Knighthood for Services to Medical Science

Michael Stratton, Ph.D.

Document 12

2013 Abbott Award in Clinical and Diagnostic Immunology

AMERICAN SOCIETY FOR MICROBIOLOGY

Jennifer Puck, M.D.

Document 13

Founding Director, Jackson Laboratory for Genomic Medicine

The Jackson Laboratory
Leading the search for tomorrow's cures

Charles Lee, Ph.D.

Document 14

‘Global Alliance’ to Enable Responsible Sharing of Genomic and Clinical Data

International partners describe global alliance to enable secure sharing of genomic and clinical data

By Broad Communications, June 4th, 2013

Over 70 leading health care, research, and disease ac
colleagues in over 40 countries have taken the first ste
to enabling secure sharing of genomic and clinical data
one-million fold, and more and more people are choos
available for research, clinical, and personal use. How
evidence base for biomedicine that is larger than any
to the highest standards of ethics and privacy. These
will be best served if we work together to develop and
regulatory) that make it possible to share and interpret
both effective and responsible.

wellcome trust sanger institute

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5 June 2013

Alliance will build data-sharing future

World's health researchers join together to share and use 'big data'

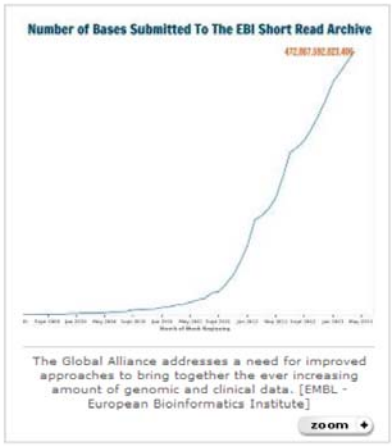
More than 60 leading health care, research and disease advocacy organisations from across the world are joining together to form an international alliance dedicated to enabling secure sharing of genomic and clinical data.

Each of these organisations has signed a 'Letter of Intent', pledging to work together to create a not-for-profit, inclusive, public-private, international, non-governmental organisation (modelled on the World Wide Web Consortium, W3C) that will develop a common framework.

The cost of genome sequencing has fallen one-million fold, and ever increasing numbers of people are making their genetic and clinical data available for research and clinical use. However, interpreting people's genetic data requires a standardised biomedical evidence base that is larger than any one party alone can develop, and that adheres to the highest ethical and privacy standards.

> *" In recent years, many groups around the world have recognized the need for improved approaches to bring together genomic and clinical data, and some have made progress addressing this. "*
>
> **Professor Mike Stratton**

"In recent years, many groups around the world have recognised the need for improved approaches to bring together genomic and clinical data, and some have made progress addressing this," said Professor Mike Stratton, Director of the Wellcome Trust Sanger Institute. *"But in coming together, and studying the challenges, we recognised that something was missing: an international body that spans diseases and institutions, committed to furthering progress in an innovative and responsible fashion."*



The Global Alliance addresses a need for improved approaches to bring together the ever increasing amount of genomic and clinical data. [EMBL - European Bioinformatics Institute]

Document 15

Genomics England Launched

Jeremy Hunt launches genomics body to oversee healthcare revolution

Genomics England will aim to attract private investment in life sciences, but campaigners raise patient privacy concerns

Randeep Ramesh, social affairs editor
The Guardian, Thursday 4 July 2013

Jeremy Hunt, the health secretary. Photograph: Jonathan Brady/PA

Genomics england

GOV.UK
Menu

Tell us what you think of GOV.UK

Document 16

Genomics and the Economy

The Impact of Genomics on the U.S. Economy

Federal research investment has contributed to medical science, improved public health, created American jobs and helped generate nearly $1 trillion in economic impacts to date

Prepared by Battelle Technology Partnership Practice
for United for Medical Research (UMR)
June 2013

Document 17

Genomics and the Economy

McKinsey Global Institute




May 2013

Disruptive technologies: Advances that will transform life, business, and the global economy

Document 17

An End to Gene Patents?

Document 18

First GINA Cases Filed

GINA
GENETIC INFORMATION NONDISCRIMINATION ACT

1. Settled

v.

FABRICUT

2. Pending

v.

Document 19

U.S. Supreme Court Upholds 'DNA Swabbing' of People Under Arrest

SUPREME COURT OF THE UNITED STATES

Syllabus

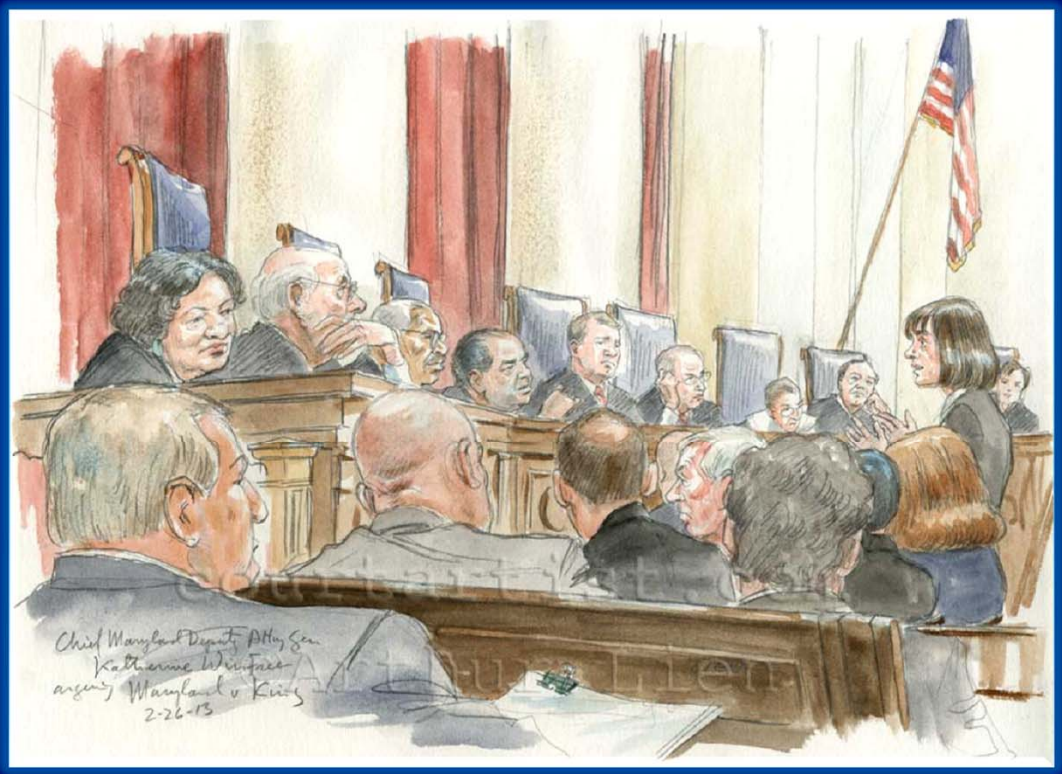
MARYLAND *v.* KING

CERTIORARI TO THE COURT OF APPEALS OF MARYLAND

No. 12–207. Argued February 26, 2013—Decided June 3, 2013

After his 2009 arrest on first- and second-degree assault charges, respondent King was processed through a Wicomico County, Maryland, facility, where booking personnel used a cheek swab to take a DNA sample pursuant to the Maryland DNA Collection Act (Act). The swab was matched to an unsolved 2003 rape, and King was charged with that crime. He moved to suppress the DNA match, arguing that the Act violated the Fourth Amendment, but the Circuit Court Judge found the law constitutional. King was convicted of rape. The Maryland Court of Appeals set aside the conviction, finding unconstitutional the portions of the Act authorizing DNA collection from felony arrestees.

Held: When officers make an arrest supported by probable cause to hold for a serious offense and bring the suspect to the station to be detained in custody, taking and analyzing a cheek swab of the arrestee's DNA is, like fingerprinting and photographing, a legitimate police booking procedure that is reasonable under the Fourth Amendment. Pp. 3–28.



Document 20

NCHPEG Ceases Operations

NCHPEG
National Coalition for Health Professional Education in Genetics

- Promoted health professional education for two decades
- NCHPEG founded in 1996 through partnership of the American Medical Association, American Nurses Association, and NHGRI
- Ceased operations on August 31
- Resources will be maintained at the American Society of Human Genetics website

Document 21

NHGRI Genome Advance of the Month

Digging into the past to uncover the cause of our cavities

Child abuse leaves epigenetic marks

By Roseanne Zhao, Ph.D.
NIH Medical Scientist Training Program Track 3 Scholar

Close encounters of the microbial kind

By Kathie Y. Sun
NHGRI Scientific Program Analyst

Alzheimer's disease: Putting the pieces together with integrative genomics

By Elizabeth Burke, Ph.D.
Intramural Postdoctoral Fellow, NHGRI

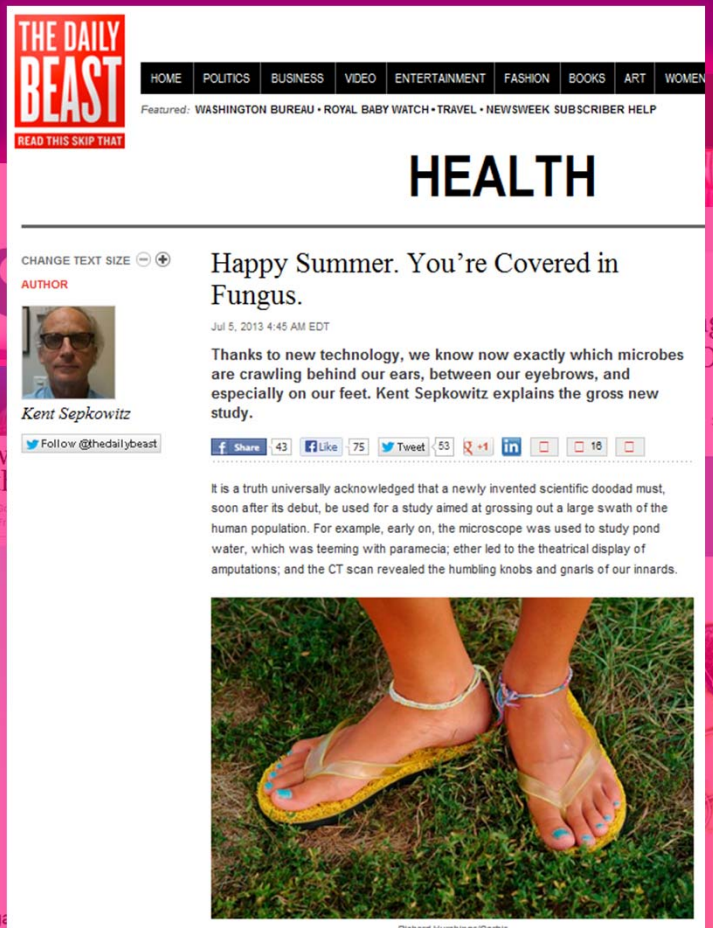
Alzheimer's disease - a neurological disorder causing progressive dementia, disorientation and behavioral changes - will affect more than 5 million Americans this year. While five percent of those with Alzheimer's disease develop it between the ages of 30-65 as a result of any one of several rare, inherited, single-gene mutations, the large majority of affected individuals develop a non-familial form after the age of 65, called late-onset Alzheimer's disease (LOAD).

In comparison to the early-onset form, the underlying cause of LOAD is much more complex; it is thought to be caused by a combination of several genetic and non-genetic risk factors. Genetic risk factors refer to common human genetic variations, or alleles, that increase a person's chance of developing a disease without directly causing it. While each risk factor is not sufficient to cause the disease on its own, multiple risk factors can combine their modest individual effects to develop LOAD.

Though advancing age is currently the strongest known risk factor for LOAD, the most influential genetic factor to be identified is one of three common alleles for the gene apolipoprotein E (*APOE*) that is referred to as *APOE4*. A person who inherits the *APOE4* allele from only one parent has a three-fold increase in LOAD risk, whereas a person that inherits *APOE4* from both parents is ten times more likely to develop LOAD. Despite this strong association, it has remained unclear how *APOE4* contributes to the disease. July's Genome Advance of the Month describes a study, published in the August 1, 2013, issue of *Nature,* which combined several genomic methods to identify important regulatory processes that link the common genetic variation *APOE4* to the development of LOAD.

Document 22

Genomics In The News…



"In the past we tended to think that the absence of microbes made people healthy, but as we're learning, bacteria are not just a source of disease—they're a source of health." Lita Proctor, PhD

Document 23

Genomics In The News…

Richard K. Wilson
Washington University, St. Louis

Field: Genomics
# of Hot Papers: 15

Eric S. Lander
Broad Inst. of MIT at Harvard

Field: Genomics
# of Hot Papers: 13

Jun Wang
BGI

Field: Genomics
# of Hot Papers: 14

SCIENCEWATCH

THE HOTTEST SCIENTIFIC RESEARCHERS AND RESEARCH

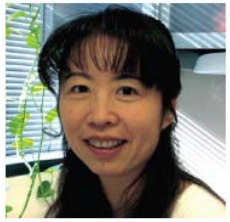
Kari Stefansson
deCODE Genetics

Field: Genomics
# of Hot Papers: 13

Elaine Mardis
Washington University, St. Louis

Field: Genomics
# of Hot Papers: 12

THOMSON REUTERS



Li Ding
Washington University, St. Louis

Field: Genomics
# of Hot Papers: 12

Goncalo Abecasis
University of Michigan

Field: Biostatistics
# of Hot Papers: 12

Robert Fulton
Washington University, St. Louis

Field: Genomics
# of Hot Papers: 11

Document 23

Genomes In The News…

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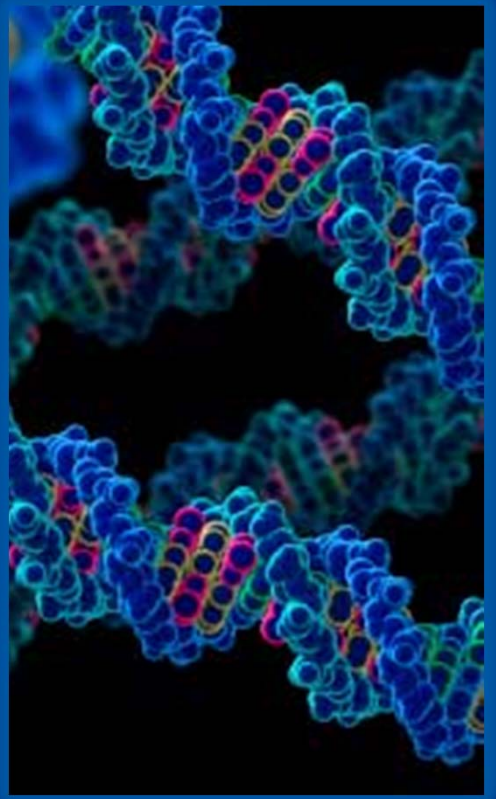
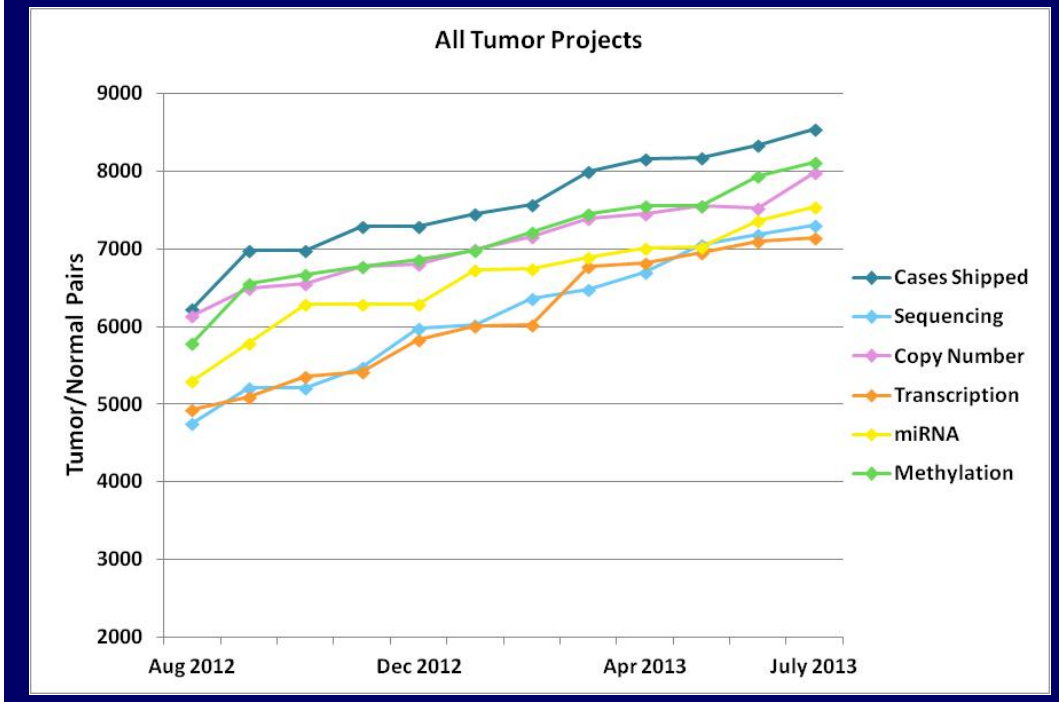
Large-Scale Genome Sequencing and Analysis Centers

- 15 publications this quarter
- Current major projects:
  - TCGA
  - Alzheimer's disease
  - Other complex genetic disorders



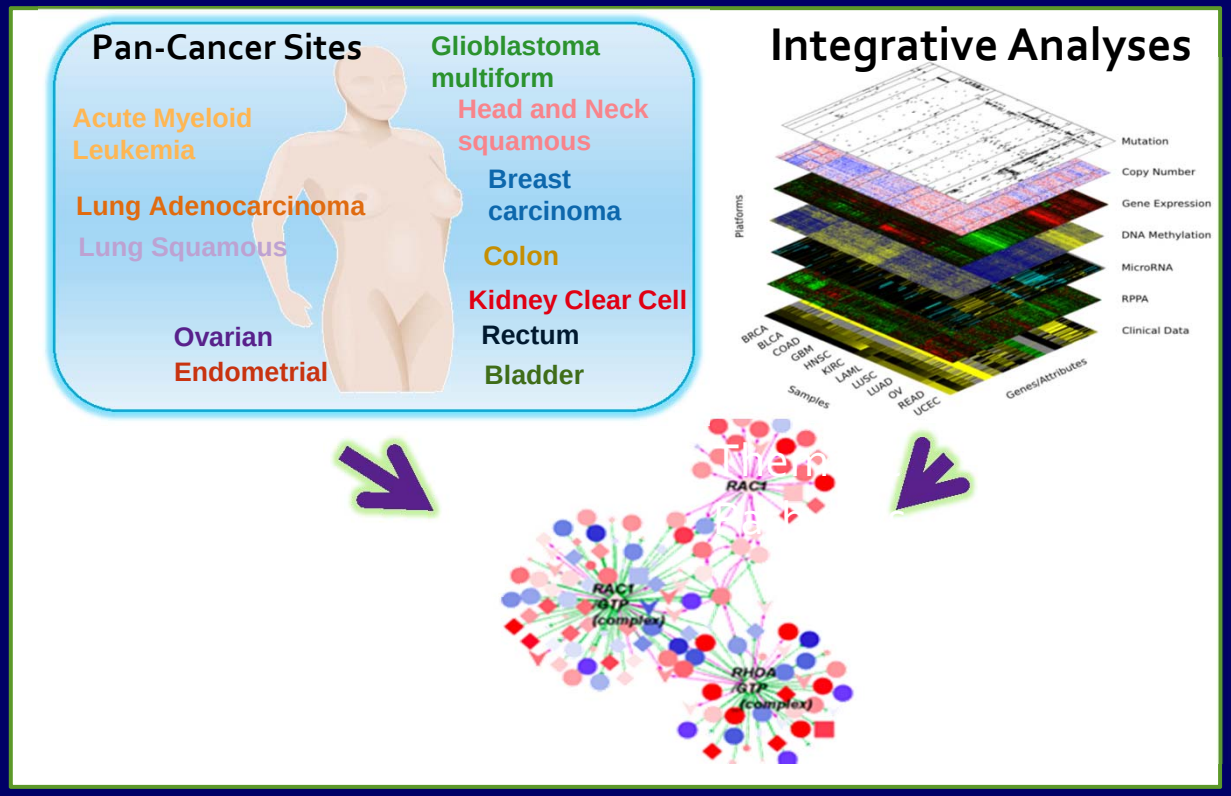
Document 25

- **Changes in TCGA leadership:**
  - **TCGA Program Directors Brad Ozenberger and Kenna Shaw left NIH**
  - **Lou Staudt named Director of the NCI Center for Cancer Genomics**



- **On target to meet goals**
- **Data available for 8,500 tumor specimens (from 26 tumor types)**
- **On course to reach ~11,000 specimens by project's end**

- **‘Pan-cancer’ analyses reveal additional features of disease**
- **~30 papers to be published in the next several weeks**



Centers for Mendelian Genomics

Finding the genes underlying human Mendelian conditions

Disease Gene Discovery:

- **>9600 whole-exome sequences for >617 diseases**
- **Discovery of 199 disease genes underlying 114 diseases**
- **Publications (31)**

  Discoveries of diseases genes (22)
  Methods and resources (6)
  Practices for data sharing (3)

Network and Outreach:

- **Collaborative Network: 384 investigators, 189 institutions, and 30 countries**
- **IRDiRC committees and working groups**

Document 27

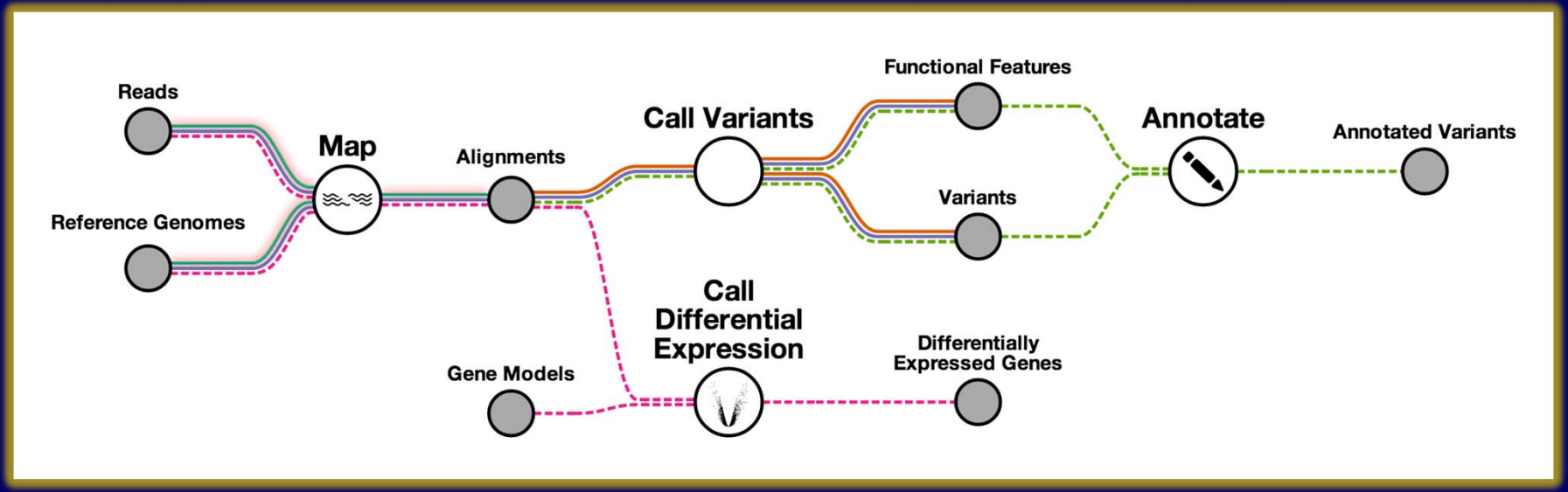
Clinical Sequencing Exploratory Research (CSER) Program

- **Three new CSER sites:**
  - **HudsonAlpha Institute**
  - **Kaiser Foundation Research Inst.**
  - **University of Michigan**
- **Coordinating Center:**
  - **University of Washington**
- **Enhanced integration with ELSI studies**

Genome Sequencing Informatics Tools

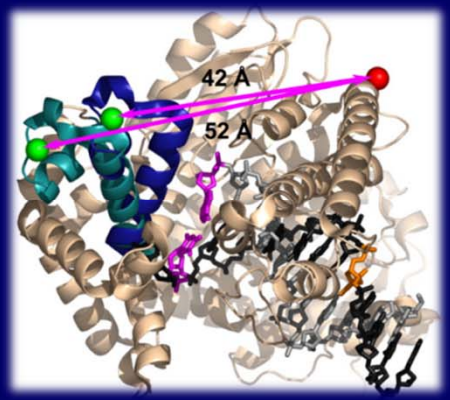
- iSeqTools Portal provides a ‘subway map’ for genome analysis

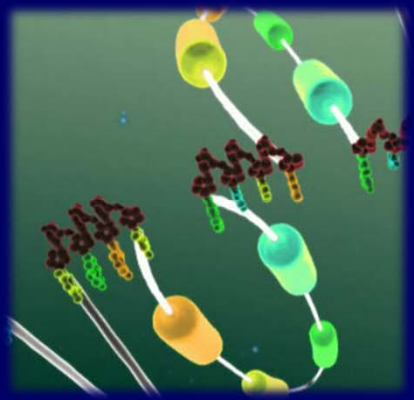
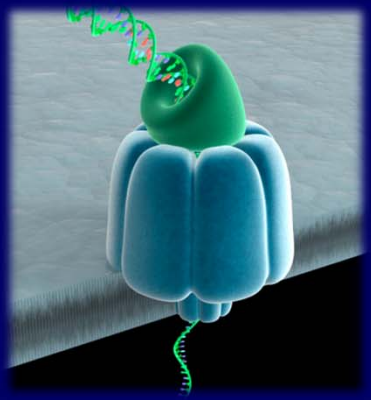
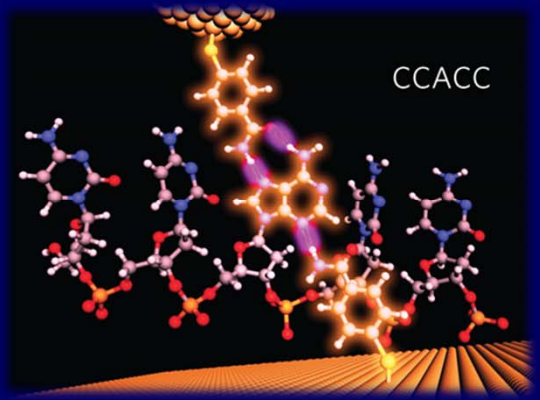
- User can drill down to details and individual tools with a ‘semantic zoom’ web feature



DNA Sequencing Technology Development

- 8 awards made this year
- RFAs posted in June with application receipt date of October 17, 2013



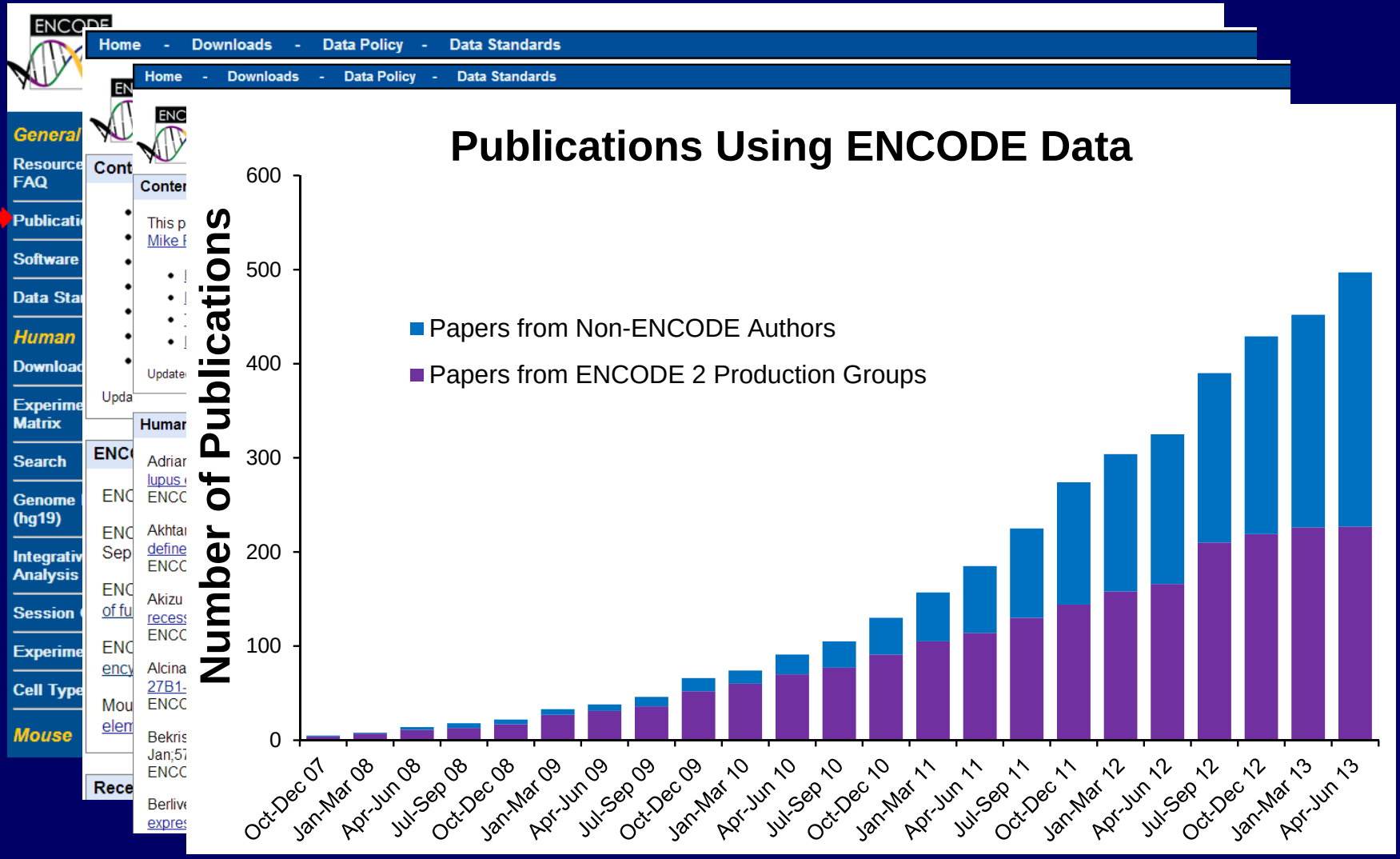




ENCODE

- **ENCODE Outreach Activities:**
  - ENCODE Tutorial at ASHG Meeting
  - ENCODE Tutorial at CHARGE Consortium Meeting

- **Cross-Species Comparison Papers Under Review:**
  - modENCODE (fly/worm/human)
  - mouse ENCODE (mouse/human)

Publications Using ENCODE Data



Genomics of Gene Regulation RFA

- Concept Clearance approved in May 2011
- RFA-HG-13-012: Genomics of Gene Regulation (U01)
- Exploring genomic approaches to understand the role of DNA sequence in gene regulatory networks
- Applications due November 15, 2013

Part 1. Overview Information

Participating Organization(s)	National Institutes of Health (NIH)
Components of Participating Organizations	National Human Genome Research Institute (NHGRI)
Funding Opportunity Title	Genomics of Gene Regulation (U01)
Activity Code	U01 Research Project – Cooperative Agreements
Announcement Type	New
Related Notices	None
Funding Opportunity Announcement (FOA) Number	RFA-HG-13-012
Companion Funding Opportunity	None
Number of Applications	See Section III. 3. Additional Information on Eligibility.
Catalog of Federal Domestic Assistance (CFDA) Number(s)	93.172
Funding Opportunity Purpose	This Funding Opportunity Announcement (FOA) solicits applications to explore genomic approaches to understanding the role of genon genomics is to decipher the rules by which gene networks are regu and disease. The GGR initiative will address the genome-proxima

Document 32

Centers of Excellence in Genomic Science (CEGS) Program

- Annual meeting at the U. of Wisconsin in October

WISCONSIN
UNIVERSITY OF WISCONSIN–MADISON

- New CEGS applications to be reviewed this fall for discussion at February 2014 Council meeting

Document 33

Genomic Medicine Working Group

- 5th Genomic Medicine Meeting (May 2013): Federal-wide strategies
- 6th Genomic Medicine Meeting (January 2014): International approaches
- Progress of the Genomic Medicine Working Group will be discussed later in the Open Session

Document 34

Inter-Society Coordinating Committee

- Inter-Society Coordinating Committee for Practitioner Education in Genomics to meet in September
- 20 professional societies currently participating

AACC, AACME, AAFP, AAP, ABMG, ABMS, ACC, ACGME, ACMG, ACOG, ACP, AHA, AMP, APHMG, ASCO, ATS, CAP, CMSS, ISPG, SGIM

Population Architecture using Genomics and Epidemiology (PAGE) II

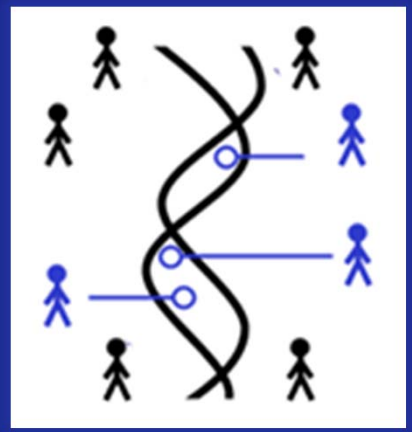
- **Study Cohorts in PAGE II:**

  Women's Health Initiative
  Fred Hutchinson Cancer Research Center

  Mount Sinai Biobank
  Mount Sinai School of Medicine

  CALiCo Consortium
  University of North Carolina

  Multiethnic Cohort Study
  University of Hawaii



- **Coordinating Center: Rutgers University**
- **Genotyping: Center for Inherited Disease Research**

Document 35

Genomic Sequencing and Newborn Screening Disorders

- Brigham and Women's Hospital
- Children's Mercy Hospital
- University of California, San Francisco
- University of North Carolina at Chapel Hill

Document 35

Genome-Wide Association Studies (GWAS) Catalog

Webinar: “Current uses of and future directions for the Genome-Wide Association Studies Catalog”

Webinar Agenda

Share Print

Current uses of and future directions for the Genome-Wide Association Studies Catalog

July 18, 2013

9 a.m. Eastern (U.S.) | 2 p.m. BST (U.K.) | 3 p.m. CET (Europe) | 11 p.m. Australia

Chair: Nancy Cox, Ph.D.

YouTube Video Playlist

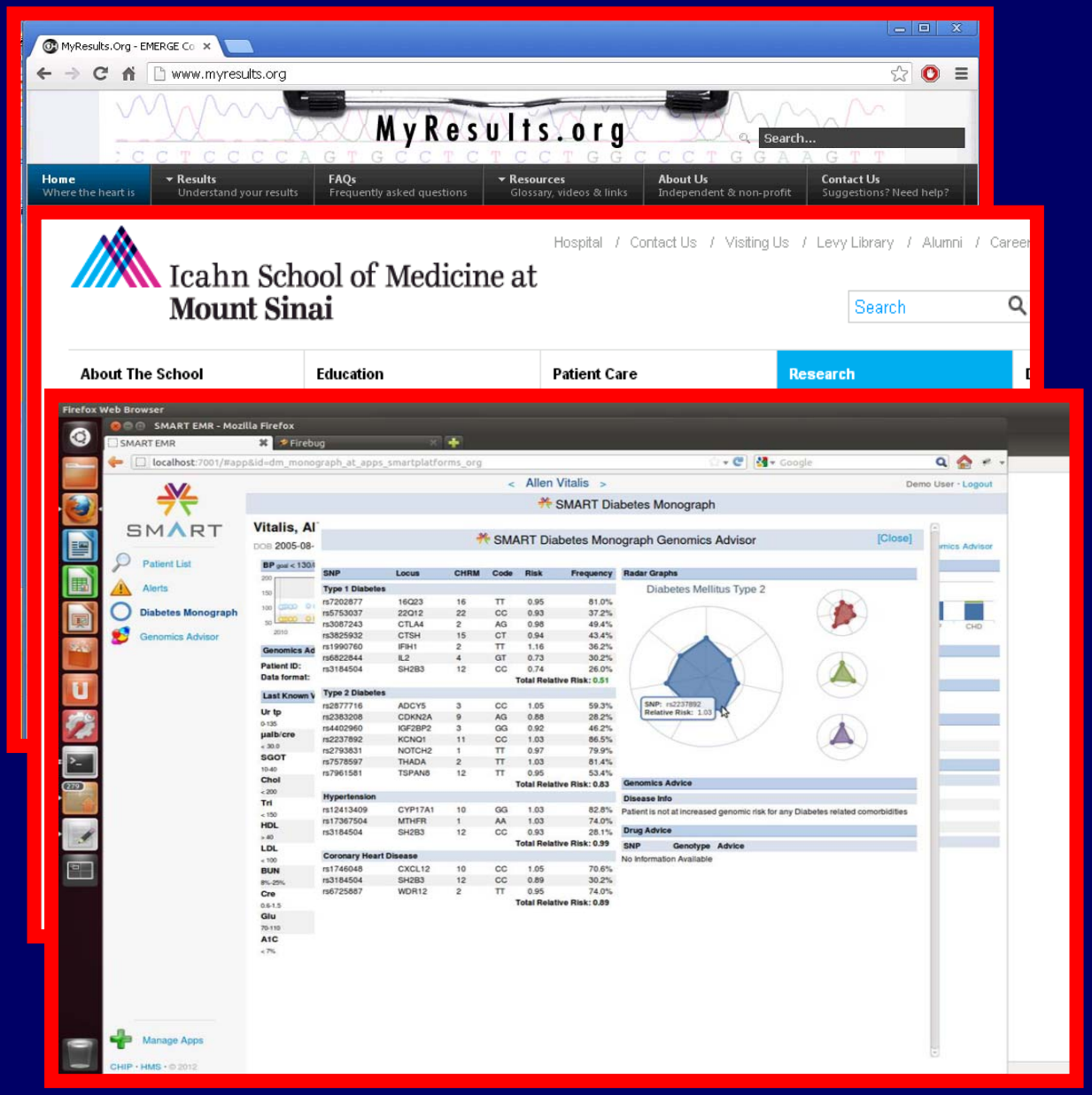
Video of each presentation has been provided by The European Bioniformatics Institute, part of the European Molecular Biology Laboratory (EMBL-EBI).

Time	Talk	Speaker
09.00	***Introduction to the GWAS Catalog*** YouTube Video | Slides PDF	**Lucia Hindorff, Ph.D.** National Human Genome Research Institute, NIH
09:40	***Uncovering hidden genes in intergenic GWAS regions with RNA capture-sequencing*** YouTube Video | Slides PDF	**Michael Clark, Ph.D.** University of Queensland, Australia
	GWAS and prior knowledge to uncover gene-gene interactions	

To begin, enroll your fingers.
You have not registered fingerprints.

Document 36

eMERGE Tools



- My Results web page
- Clinical Decision Support (CDS) tools: CLIPMERGE
- SMART Genomics Advisor will be evaluated and piloted to standardize CDS approaches across eMERGE sites

Document 37

Centers of Excellence in ELSI Research (CEER) Program

CEERs Grants Funded this Summer:

- University of North Carolina (P50)
- Columbia University (P50)

- University of Utah (P20)
- Johns Hopkins University (P20)
- Kaiser/UCSF (P20)

Document 38

Genomics & Society Working Group

- **Next Meeting: November 2013**
- **Areas to be discussed:**

  Priority Setting

  Funding Mechanisms

  Boundaries of ELSI Research

Document 39

Training and Career Development

- To align research training and career development programs with NHGRI strategic plan
- To prepare future leaders in genomic science and genomic medicine
- To harmonize our training and career investment with the rest of NIH

Document 40

Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

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V. NIH Common Fund/Trans-NIH

VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

Molecular Libraries Program (MLP)

- Final year of production phase
- Screening Centers' Goals:

  Completing projects

  Placing all data in PubChem

  Filing probe reports

Human Microbiome Project (HMP)

Document 42

HMP Data to be Hosted in Amazon Cloud

- September 2013: HMP data publically available on Amazon cloud

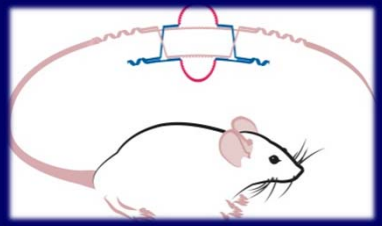
- December 2013: Collection of HMP analysis tools, tutorials, and documentation will be made available on Amazon cloud

National Human Genome Research Institute

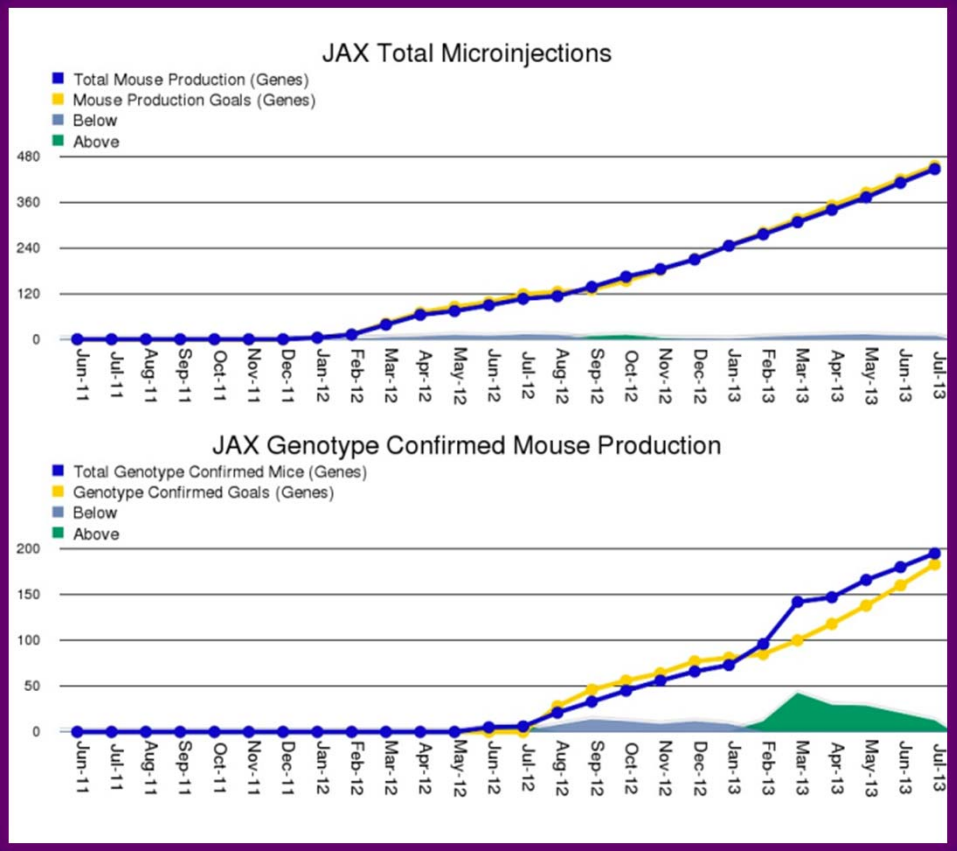
National Institute of Allergy and Infectious Diseases

amazon web services™

Knockout Mouse Phenotyping Project (KOMP2)



- ‘Perfect Host’ technology
- KOMP2 production on track



Document 43

GTEx
Genotype-Tissue Expression Project

- GTEx pilot data available in dbGaP
- Scale-up phase started
  - 900 genotyped donors
  - >25,000 RNA-Seq studies

- June 2013 GTEx Community Meeting
- Funding plan for RFA “Enhancing GTEx with molecular analyses of stored biospecimens (U01)” will be presented in Closed Session

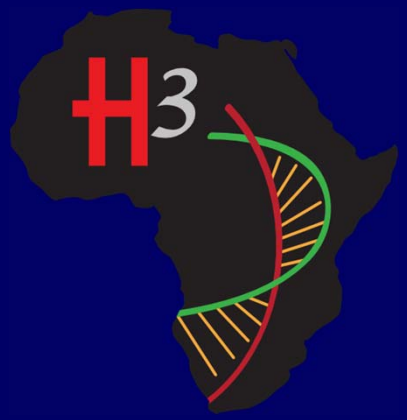
Library of Integrated Network-based Cellular Signatures (LINCS)

- New LINCS RFA (RM13-013) published
- LINCS-BD2K Coordinating Center RFA being drafted
- LINCS 2013 Community Meeting and Workshop at Broad Institute in November

H3Africa

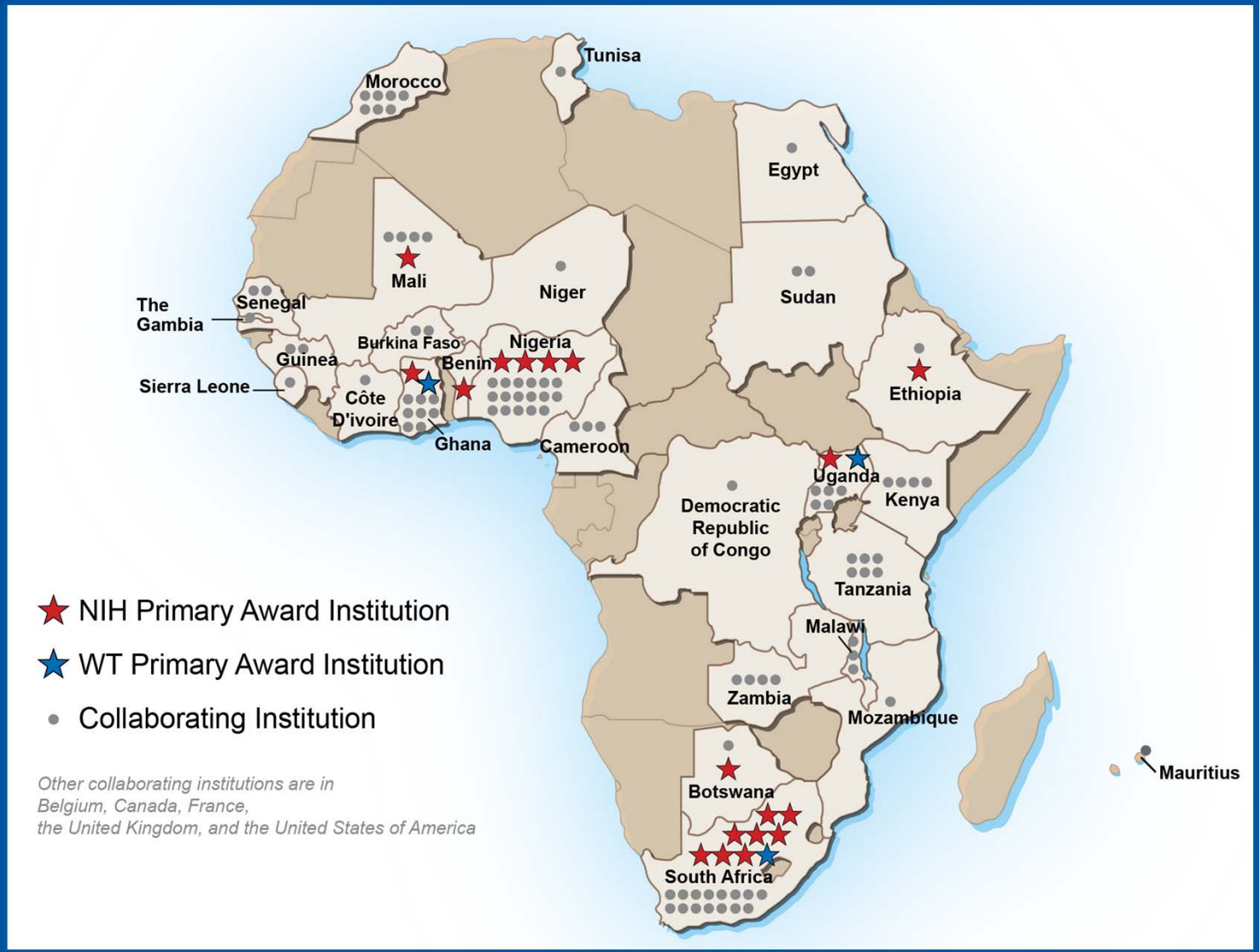
- **H3Africa Consortium has funded 21 projects**
  - **Bioinformatics network**
  - **Pilot biorepositories**
  - **Projects working on:**

Obesity	Sickle Cell Disease
HIV	Kidney disease
Stroke	Microbiome
Trypanosomes	Fevers of unknown origin
Heart disease	Diabetes
Neurological disorders	Schizophrenia



- **Next Consortium meeting in October**

H3Africa Research Network



Document 46

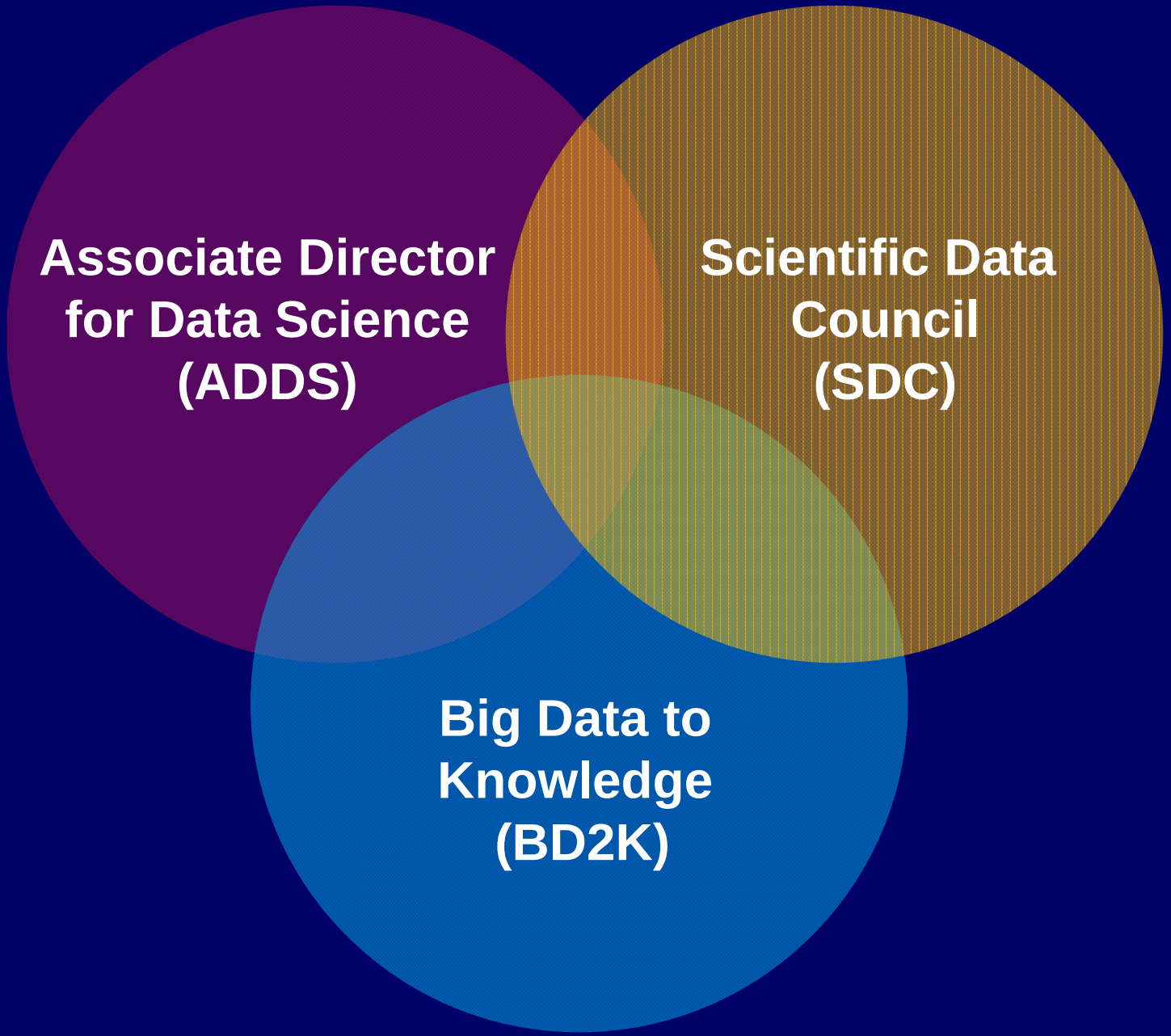
Undiagnosed Diseases Network (UDN)

- UDN aims to increase the capacity for and use of genomic data in the diagnosis and management of rare and new diseases

- RFA-RM-13-018: DNA Sequencing Core for an Undiagnosed Diseases Network (U01)

  Application Due Date: November 19, 2013

NIH Data Science Components



Scientific Data Council: Membership

Acting Chair: Eric Green (Acting ADDS & NHGRI)

Members:

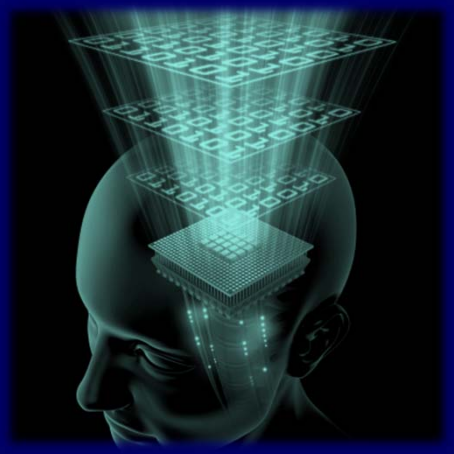
James Anderson (DPCPSI)
Sally Rockey (OER)
Michael Gottesman (OIR)
Kathy Hudson (OD)
Amy Patterson (OSP)
Andrea Norris (CIT)
Judith Greenberg (NIGMS)
Betsy Humphreys (NLM)
Douglas Lowy (NCI)
John J. McGowan (NIAID)
Alan Koretsky (NINDS)
Michael Lauer (NHLBI)
Belinda Seto (NIBIB)

Acting Executive Secretary: Allison Mandich (NHGRI)

Big Data to Knowledge (BD2K): Update

- Sequester is delaying launch of training, software development, and second round of centers components
- Three RFIs published, all with excellent responses
- Two workshops have been held, with two more coming later this month

Big Data to Knowledge (BD2K): Update



- **FOA published for Investigator-Initiated Centers of Excellence (U54)**

  Applicant Information Webinar: September 12

  Questions to BD2KCenterRFA@mail.nih.gov

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G2C2 Pharmacist Resources

- Pharmacist G2C2 Resource Review meeting was held in June

- Planned availability this Fall

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Honorary Professor
University of Cape Town

Charles Rotimi, Ph.D.

NHGRI Intramural Research Highlights

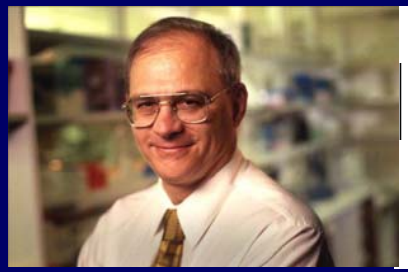
nature International weekly journal of science

Topographic diversity of fungal and bacterial communities in human skin

PNAS

Targeting proximal tubule mitochondrial dysfunction attenuates the renal disease of methylmalonic acidemia

Irini Manoli[a,1], Justin R. Sysol[a,1], Lingli Li[b], Pascal Houillier[b,c], Caterina Garone[d,e], Cindy Wang[a], Patricia M. Zerfas[f], Kristina Cusmano-Ozog[g], Sarah Young[h], Niraj S. Trivedi[i], Jun Cheng[j], Jennifer L. Sloan[a], Randy J. Chandler[a], Mones Abu-Asab[k], Maria Tsokos[k], Abdel G. Elkahloun[l], Seymour Rosen[m,n], Gregory M. Enns[g], Gerard T. Berry[o], Victoria Hoffmann[f], Salvatore DiMauro[d], Jurgen Schnermann[b], and Charles P. Venditti[a,2]

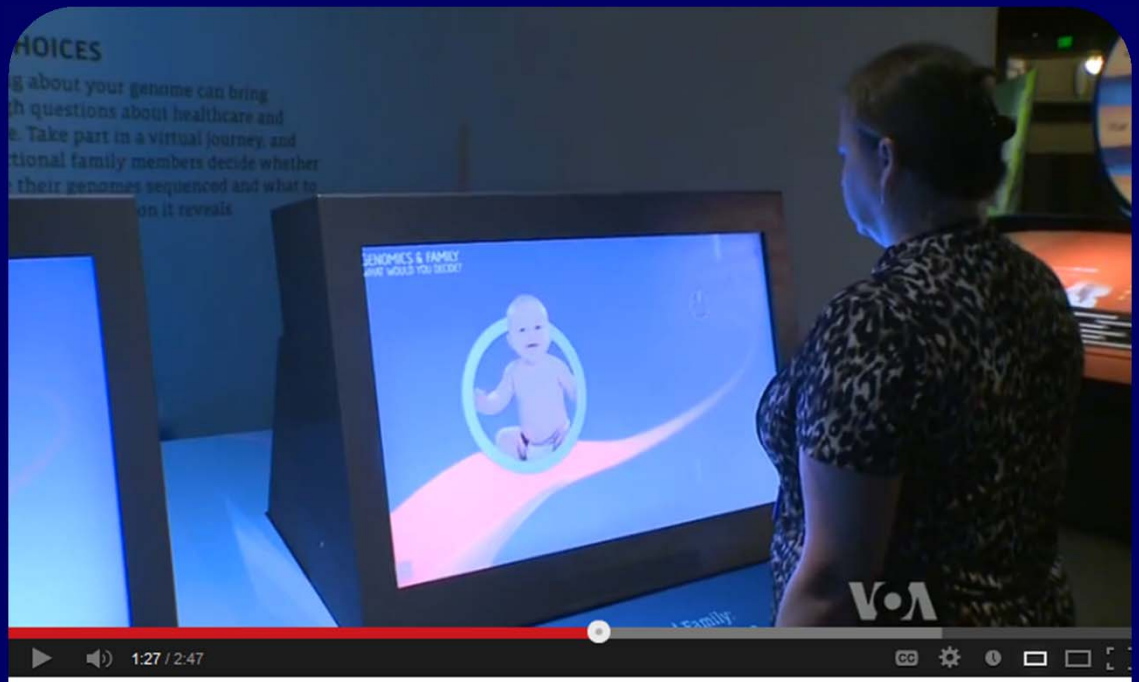


The NEW ENGLAND JOURNAL of MEDICINE

ORIGINAL ARTICLE

A Congenital Neutrophil Defect Syndrome Associated with Mutations in *VPS45*

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



Smithsonian Genome Exhibit Unlocks 21st Century Science of Life

Special Thanks!


NATIONAL HUMAN GENOME
RESEARCH INSTITUTE
Advancing human health
through genomics research