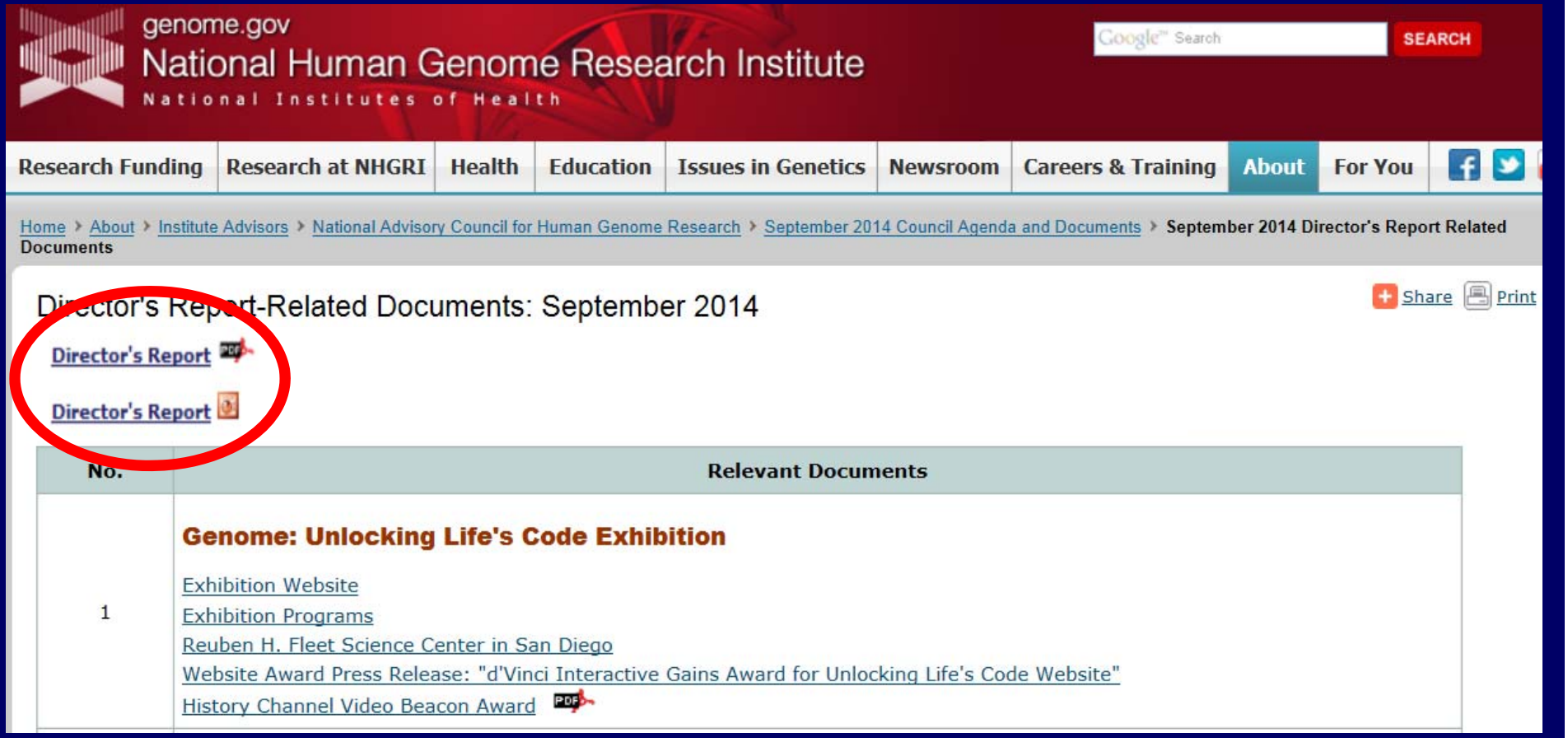
DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

September 2014

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

NIH National Human Genome Research Institute



genome.gov/DirectorsReport

Document #

Open Session Presentations

- Presentation from NIH Associate Director for Data Science

  Phil Bourne

- Genome Sequencing Workshop Report

  Adam Felsenfeld

- Concept Clearances: Genome Sequencing Program

  Adam Felsenfeld

  Lu Wang

Open Session Presentations

- **Concept Clearance: Center for Inherited Disease Research Contract Renewal**

  Lawrence Brody

- **NHGRI Division of Genomics and Society & the ELSI Research Program**

  Lawrence Brody

- **NACHGR Genomics and Society Working Group Update**

  Pamela Sankar

- **ENCODE Project Update**

  Elise Feingold

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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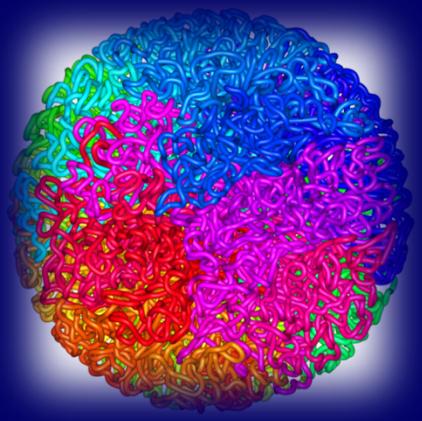
End of an NHGRI Era: Mark Guyer Retires

Mark Guyer, Ph.D.

Genome: Unlocking Life's Code Exhibition

GENOME
UNLOCKING
LIFE'S
CODE



- >3.3 million visitors since June 2013 opening
- Hosted 15 programs in conjunction with Smithsonian
- Website and videos received national recognition
- Closing Symposium on September 30
- 1st stop on 4-5 year traveling tour: San Diego

Document 1

James Watson Tours Genome: Unlocking Life's Code

New ASHG-NHGRI Fellows

Kate Blizinsky, Ph.D.

Genetics and Public Policy Fellow

Elizabeth Tuck

Genetics and Education Fellow

Document 2

NHGRI Historical Archiving Initiative

Document 3

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New DHHS Secretary: Sylvia Mathews Burwell

Document 4

Retirement of Story Landis

Story Landis, Ph.D.

Walter Koroshetz, M.D.

NIH National Institute of Neurological Disorders and Stroke

Document 5

New NIH Program on Biosecurity and Biosafety Policy

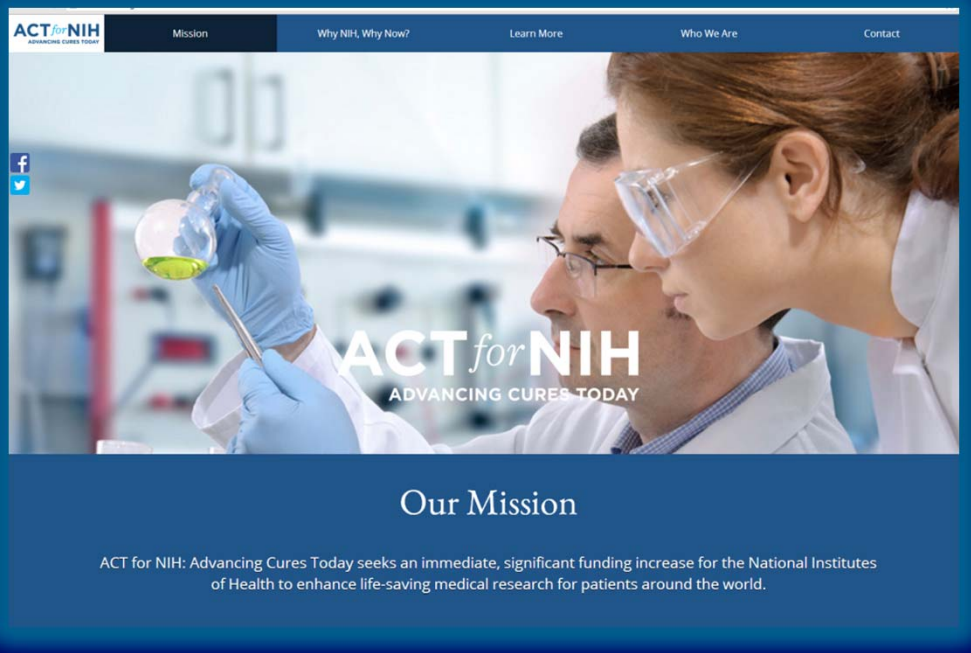
Amy Patterson, M.D.

David Shurtleff, Ph.D.

NIH National Institutes of Health

Departure of Francis ‘Pat’ White

ACT for NIH
ADVANCING CURES TODAY



Document 6

Changes to the Biosketch for NIH Grant Applications

- Length extended from 4 to 5 pages
- Section A – Personal Statement
- Section B – Positions and Honors
- Section C – Contributions to Science (NEW)
- Section D – Research Support
- Now in Round 2 of piloting and evaluation
- Roll-out for submissions in January 2015

BIOGRAPHICAL SKETCH—*Pilot Format (To Be Used for Specific FOAs only)*

Provide the following information for the Senior/key personnel and other significant contributors.
Follow this format for each person. **DO NOT EXCEED FIVE PAGES.**

Please refer to the Biographical Sketch—Pilot Format sample in order to complete sections A, B, C, and D of the Biographical Sketch.

NIH Genomic Data Sharing Policy

[Billing Code 4140-01-P]

DEPARTMENT OF HEALTH AND HUMAN SERVICES

National Institutes of Health

Final NIH Genomic Data Sharing Policy

SUMMARY: The National Institutes of Health (NIH) announces the final Genomic Data Sharing (GDS) Policy that promotes sharing, for research purposes, of large-scale human and non-human genomic[1] data generated from NIH-funded research. A summary of public comments on the draft GDS Policy and the NIH responses are also provided.

Document 7

“A Path to 21st Century Cures”

Document 8

Fiscal Year 2015 Funding

- **Regular appropriations process not completed**

	FY 2014 Actual	*President's Budget FY 2015*	*House FY 2015*	*Senate FY 2015 (draft)*
NIH	**$30.2B**	**$30.4B**	-	**$30.5B**
NHGRI	**$497M**	**$498M**	- -	**$504M**

- **Continuing Resolution (CR) needed**

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Richard Gibbs Awarded Companion of the Order of Australia

Richard Gibbs, Ph.D.

Document 10

Jay Shendure Awarded HudsonAlpha Life Sciences Prize

HUDSONALPHA
INSTITUTE FOR BIOTECHNOLOGY

Jay Shendure, M.D., Ph.D.

On right, with Rick Myers (HudsonAlpha)

Document 11

NHGRI Genome Advance of the Month

The Y chromosome: beyond gender determination

Share Print

By Roseanne F. Zhao, Ph.D.
NIH M.D./Ph.D. Partnership Training Program Scholar

Researchers probe inner workings of ancient human genomes, compare them to humans

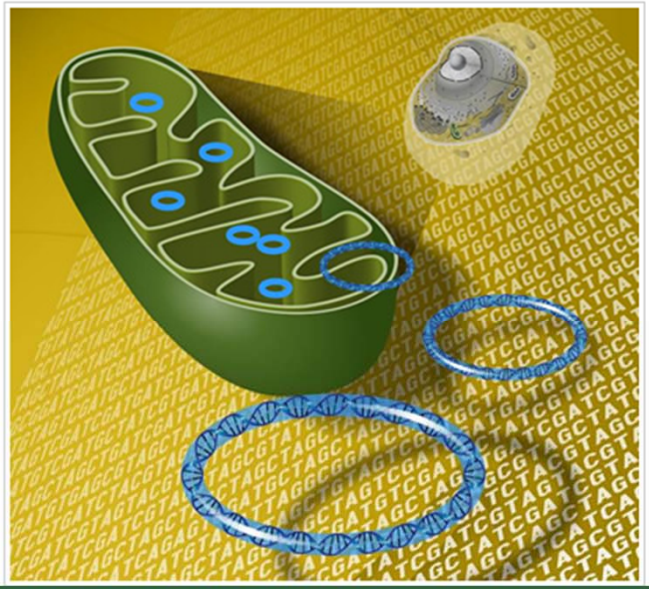
By Kyle Davis
ScM Candidate, Genetic Counseling, JHU/NHGRI

Improving the detection of heart transplant rejection with DNA sequencing

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

Researchers examine disease-causing mutations in mitochondrial genomes

By Kyle Davis
ScM Candidate, Genetic Counseling, JHU/NHGRI



The study of genetic disease has often centered on the human nuclear genome - the sprawling linear code of about 3.2 billion nucleotides and more than 24,000 genes spread across our 46 chromosomes. In contrast, the other genome that resides within us, the mitochondrial genome, has received less attention. Though our genome dwarfs it in size - a short, circular genome with just over 16,000 nucleotides and exactly 37 genes - the small (but mighty) mitochondria genome is arguably just as important.

Unlike the nuclear genome - where each cell has only one complete copy - there can be tens, hundreds or thousands of mitochondria per cell. Since each mitochondrion has on average five full mitochondrial genomes, there can, theoretically, be tens, hundreds or thousands of mitochondrial genomes per cell. New studies that sequence entire genomes have also sequenced our mitochondrial genomes, giving scientists a more detailed glimpse into the nature of diseases associated with mitochondrial mutations.

Mitochondria - often referred to as the "power plant of the cell" - are complex. They not only create energy in the form of the ever-present molecule adenosine triphosphate (ATP) used for many of the cell's actions, but they also house genes for the ribosome, which build proteins as well as the transfer RNA (tRNA) genes, which provide a sort of lock-and-key system that helps decipher the genetic code into the amino acid protein code. In this way, mitochondria keep each cell

Document 12

Genomics In The News…

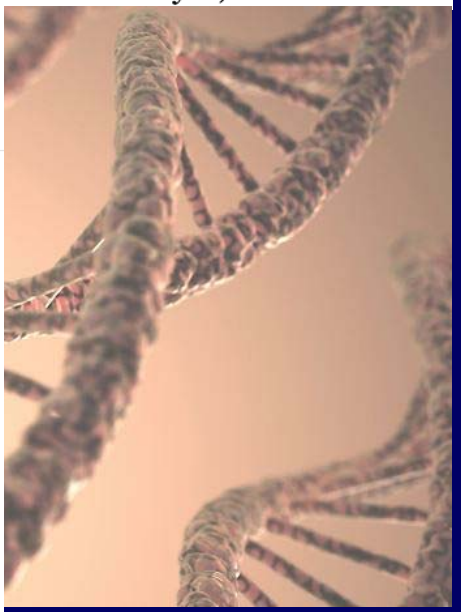
The Washington Post

Health & Science

Drugmakers find breakthroughs in medicine tailored to ind makeups

Pittsburgh Post-Gazette®

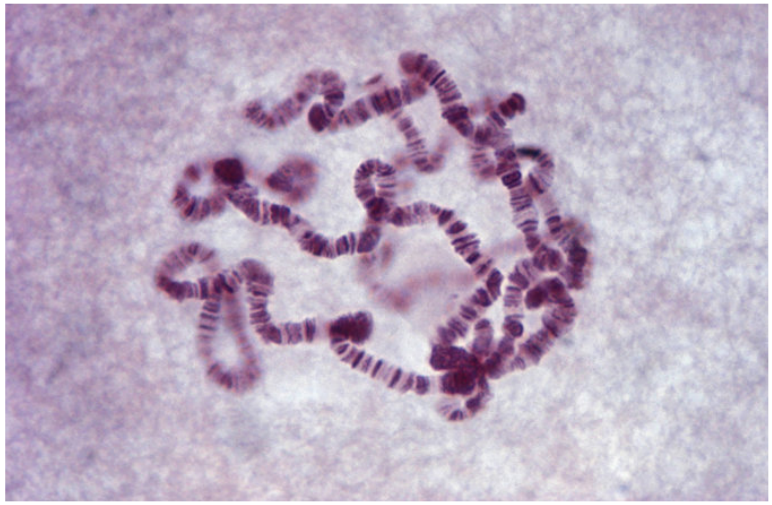
Genome not destiny, doctors stress tailor lifestyle, treatment



The New York Times

Tiny, Vast Windows Into Human DNA

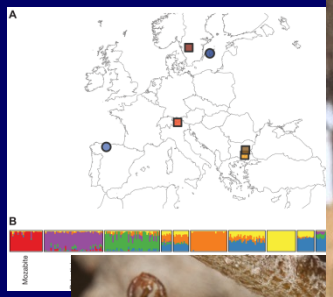
By CARL ZIMMER SEPT. 1, 2014

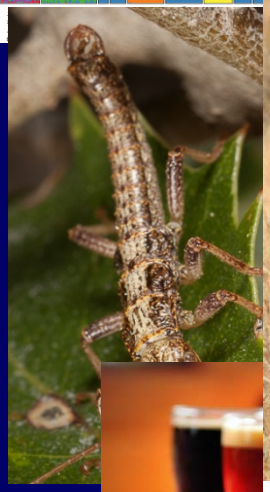


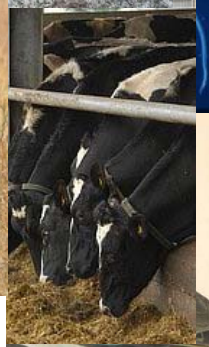
Genes found on chromosomes of the fruit fly are regulated much like those of humans though they are but distant relatives. Ed Reschke/Getty Images

Document 13

Genomes In The News…







Elephants have five times more olfactory receptors than humans and the most of any animal characterized to date, according to a new study. (Courtesy of Eric Green)

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Genome Sequencing Program

Document 16

ARTICLE

Nature 2014, July 31. Vol. 511 (7511)

OPEN

ARTICLE

Nature 2014, July 23. doi.10.1038/nature13480

OPEN

Cancer Cell

Article

Cancer Cell, 2014, Aug 21. doi.org/10.1016/j.ccr.2014.07.014

CellPress

Cell 2014, Aug 14. Vol. 158

Resource

Cell

Multiplatform Analysis of 12 Cancer Types Reveals Molecular Classification within and across Tissues of Origin

Katherine A. Hoadley,[1,20] Christina Yau,[2,20] Denise M. Wolf,[3,20] Andrew D. Cherniack,[4,20] David Tamborero,[5] Sam Ng,[6] Max D.M. Leiserson,[7] Beifang Niu,[8] Michael D. McLellan,[8] Vladislav Uzunangelov,[6] Jiashan Zhang,[9] Cyriac Kandoth,[8] Rehan Akbani,[10] Hui Shen,[11,22] Larsson Omberg,[12] Andy Chu,[13] Adam A. Margolin,[12,21] Laura J. van't Veer,[3] Nuria Lopez-Bigas,[5,14] Peter W. Laird,[11,22] Benjamin J. Raphael,[7] Li Ding,[8] A. Gordon Robertson,[13] Lauren A. Byers,[10] Gordon B. Mills,[10] John N. Weinstein,[10] Carter Van Waes,[18] Zhong Chen,[19] Eric A. Collisson,[15] The Cancer Genome Atlas Research Network, Christopher C. Benz,[2,*] Charles M. Perou,[1,16,17,*] and Joshua M. Stuart[6,*]

Document 16

Centers for Mendelian Genomics

Finding the genes underlying human Mendelian conditions

- **15,790 samples / 6,421 families / 1,433 Mendelian disorders**
- **11,800 whole-exome sequences, various stages of analysis**
- **Novel discoveries about the genomic bases or clinical phenotypes of >280 Mendelian disorders**
- **Nearly 100 manuscripts published or in press**

  Phenotype-driven and genotype-driven approach

  Disease genes/pathways and disease biology

  Pleiotropy and genetic heterogeneity

Document 17

Centers for Mendelian Genomics

Finding the genes underlying human Mendelian conditions

Home Create Account About EULA Contact Us Help

GeneMatcher

GeneMatcher is a freely accessible web site designed to enable connections between clinicians and researchers from around the world who share an interest in the same gene or genes. The principle goal for making GeneMatcher available is to help solve "unsolved" exomes. This may be done with cases from research or clinical sources. No identifiable data are collected. GeneMatcher was developed with support from the Baylor-Hopkins Center for Mendelian Genomics as part of the Centers for Mendelian Genomics network.

The site allows investigators to post a gene (or genes) of interest and will connect investigators who post the same gene. The match is done automatically and there is no way to search the database. When a match occurs, the submitters will automatically receive email notification. Follow-up is at the discretion of the submitters. Aside from the site administrator, no one has access to all the information in the database. Submitters have access to their own data and may edit it or delete it at will.

As of August 1, 2014:
662 genes submitted
143 submitters
15 matches

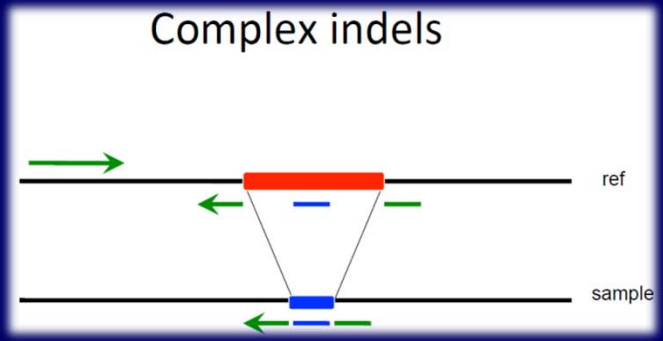
Document 17

2nd UWCMG Mendelian Data Analysis Workshop

- 17 participants
- Interaction with Center analysts, IT team, and PIs
- Concept-driven lectures on approaches to Mendelian gene discovery (e.g., how to design project)
- Hands-on data analysis using open-source software and the testing of different genetic models
- Positive post-workshop feedback from attendees

Genome Sequencing Informatics Tools

iSeqTools



- iSeqtools being used in clinical research for analysis and annotation of complex genomic variants

  Annotation of CNVs (benign vs. pathogenic)

  Analysis of complex indels

  Prediction of structural variants

- 2014 ASHG workshop “iSeqTools to demystify the cloud” sold out; “hands on” evening session added

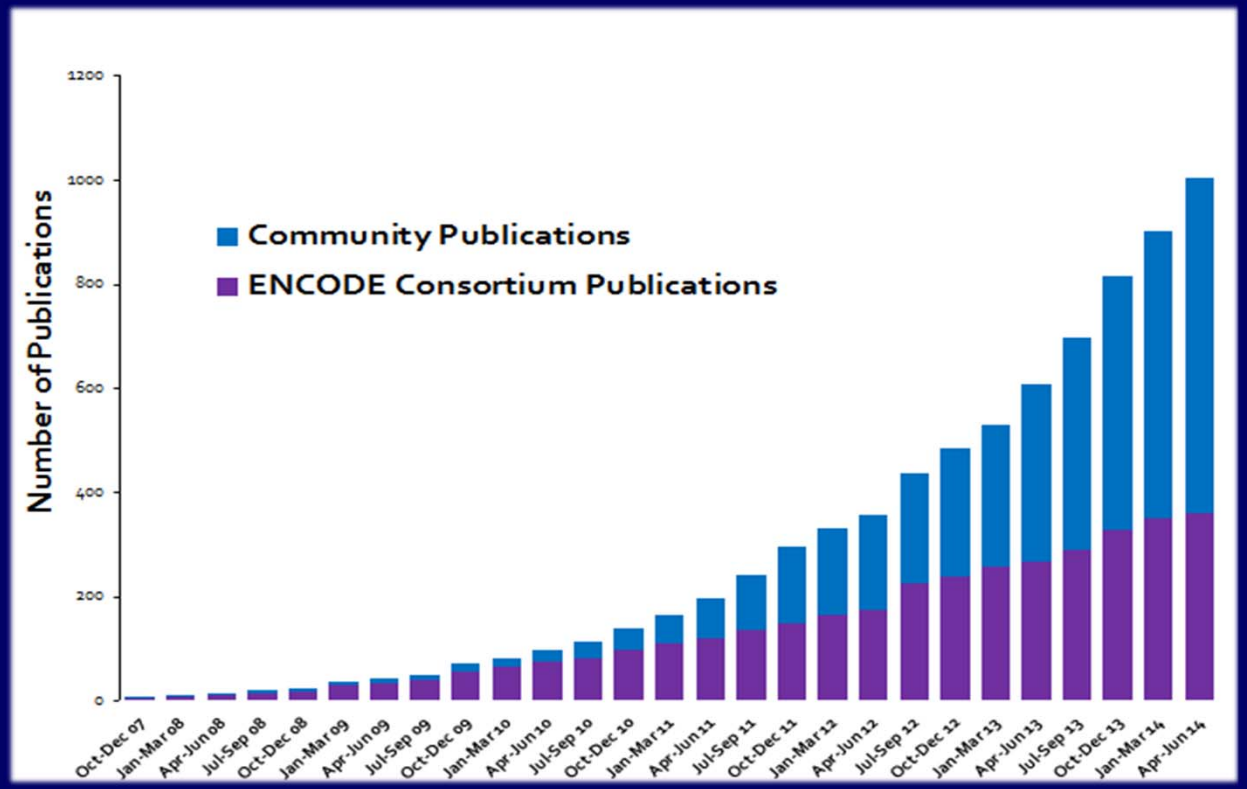
DNA Sequencing Technology Development: New Awards

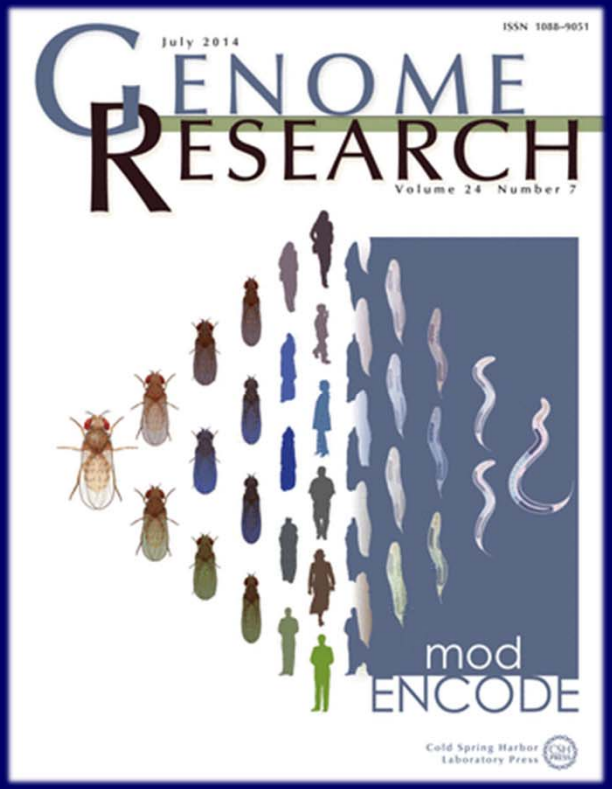
- Molecular biology to produce long read information using short-read sequencing technology
- Increased sequence accuracy with a protein nanopore
- Hybrid nanopore arrays with protein pores in solid-state or other support structures
- Solid-state graphene nanopore
- Electronic detection of polymerase motion
- Single-molecule sequencing using chemical detection of amplified reporter molecule

Document 19

ENCODE

- Updated Data Release Policy
- ENCODE Publications



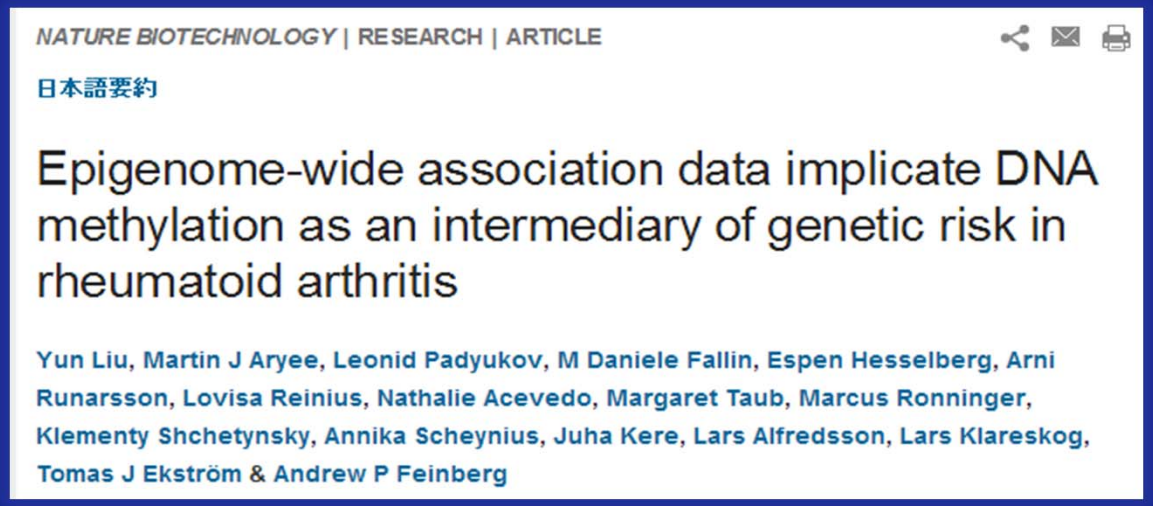


Document 20

DHHS Award for Distinguished Service Given to NHGRI ENCODE Team

Document 20

CEGS Program: High-Visibility Papers



NATURE BIOTECHNOLOGY | RESEARCH | ARTICLE

日本語要約

Epigenome-wide association data implicate DNA methylation as an intermediary of genetic risk in rheumatoid arthritis

Yun Liu, Martin J Aryee, Leonid Padyukov, M Daniele Fallin, Espen Hesselberg, Arni Runarsson, Lovisa Reinius, Nathalie Acevedo, Margaret Taub, Marcus Ronninger, Klementy Shchetynsky, Annika Scheynius, Juha Kere, Lars Alfredsson, Lars Klareskog, Tomas J Ekström & Andrew P Feinberg

"This is one of the first studies that looks for an epigenetic disease association in a really rigorous fashion."

"I am quite impressed with their level of rigor and sophistication."

Document 21

eMERGE Phase III RFAs

1. **RFA-HG-14-025 – Study Investigators (U01)**
   Genomic discovery and clinical implementation research

2. **RFA-HG-14-026 – Coordinating Center (U01)**
   Centralized support and communications

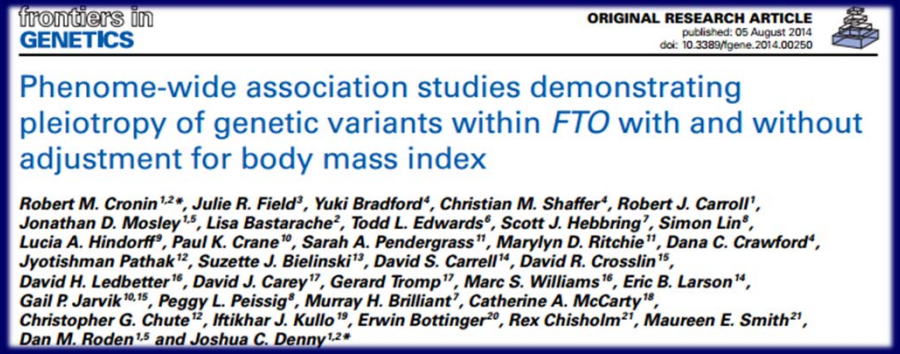
3. **RFA-HG-14-027 – Central Genome Sequencing and Genotyping Center (U01)**
   High-quality sequencing and genotyping in a CLIA environment

Deadline for submission: November 12, 2014

Document 22

eMERGE Network

- **Special Issue of**
  Frontiers in Genetics
  Genetic Research in Electronic Health Records Linked to DNA Biobanks

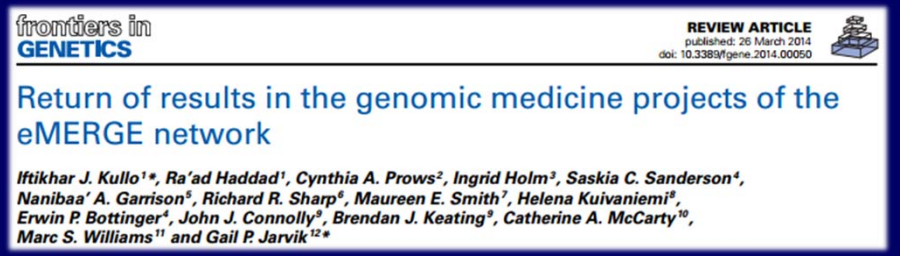


frontiers in GENETICS

ORIGINAL RESEARCH ARTICLE
published: 05 August 2014
doi: 10.3389/fgene.2014.00250

Phenome-wide association studies demonstrating pleiotropy of genetic variants within *FTO* with and without adjustment for body mass index

Robert M. Cronin[1,2], Julie R. Field[3], Yuki Bradford[4], Christian M. Shaffer[4], Robert J. Carroll[1], Jonathan D. Mosley[1,5], Lisa Bastarache[2], Todd L. Edwards[6], Scott J. Hebbring[7], Simon Lin[8], Lucia A. Hindorff[9], Paul K. Crane[10], Sarah A. Pendergrass[11], Marylyn D. Ritchie[11], Dana C. Crawford[4], Jyotishman Pathak[12], Suzette J. Bielinski[13], David S. Carrell[14], David R. Crosslin[15], David H. Ledbetter[16], David J. Carey[17], Gerard Tromp[17], Marc S. Williams[16], Eric B. Larson[14], Gail P. Jarvik[10,15], Peggy L. Peissig[8], Murray H. Brilliant[7], Catherine A. McCarty[18], Christopher G. Chute[12], Iftikhar J. Kullo[19], Erwin Bottinger[20], Rex Chisholm[21], Maureen E. Smith[21], Dan M. Roden[1,5] and Joshua C. Denny[1,2]**



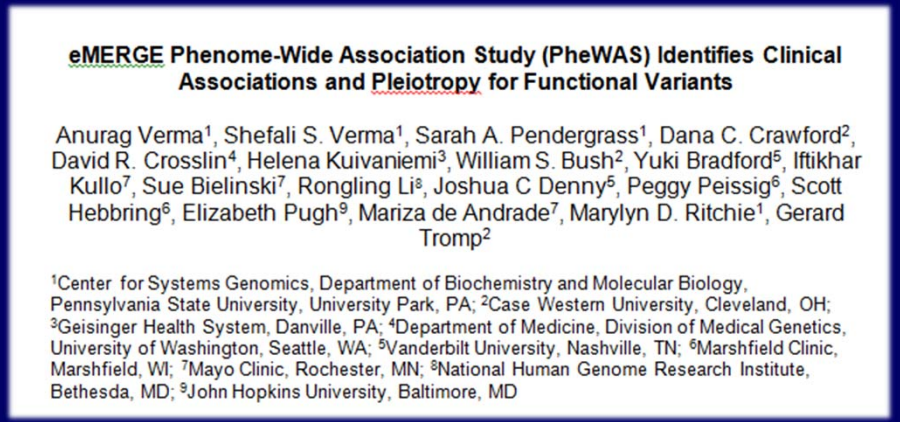
frontiers in GENETICS

REVIEW ARTICLE
published: 26 March 2014
doi: 10.3389/fgene.2014.00050

Return of results in the genomic medicine projects of the eMERGE network

Iftikhar J. Kullo[1], Ra'ad Haddad[1], Cynthia A. Prows[2], Ingrid Holm[3], Saskia C. Sanderson[4], Nanibaa' A. Garrison[5], Richard R. Sharp[6], Maureen E. Smith[7], Helena Kuivaniemi[8], Erwin P. Bottinger[4], John J. Connolly[9], Brendan J. Keating[9], Catherine A. McCarty[10], Marc S. Williams[11] and Gail P. Jarvik[12]**

- **IGES 2014**



eMERGE Phenome-Wide Association Study (PheWAS) Identifies Clinical Associations and Pleiotropy for Functional Variants

Anurag Verma[1], Shefali S. Verma[1], Sarah A. Pendergrass[1], Dana C. Crawford[2], David R. Crosslin[4], Helena Kuivaniemi[3], William S. Bush[2], Yuki Bradford[5], Iftikhar Kullo[7], Sue Bielinski[7], Rongling Li[8], Joshua C Denny[5], Peggy Peissig[6], Scott Hebbring[6], Elizabeth Pugh[9], Mariza de Andrade[7], Marylyn D. Ritchie[1], Gerard Tromp[2]

[1]Center for Systems Genomics, Department of Biochemistry and Molecular Biology, Pennsylvania State University, University Park, PA; [2]Case Western University, Cleveland, OH; [3]Geisinger Health System, Danville, PA; [4]Department of Medicine, Division of Medical Genetics, University of Washington, Seattle, WA; [5]Vanderbilt University, Nashville, TN; [6]Marshfield Clinic, Marshfield, WI; [7]Mayo Clinic, Rochester, MN; [8]National Human Genome Research Institute, Bethesda, MD; [9]John Hopkins University, Baltimore, MD

ClinGen

Clinical Genome Resource

- **Standardizing clinical assessment of variants and deposition into ClinVar**
- **Consensus processes for identifying clinically relevant variants**
- **2014 NSGC Education Conference and ASHG Annual Meeting**

PhenX Toolkit

- PhenX user community continues to grow
- Rare Conditions and Obesity are newest PhenX domains
- Adding depth to Toolkit through NIH collaborations

Contents lists available at ScienceDirect

Drug and Alcohol Dependence

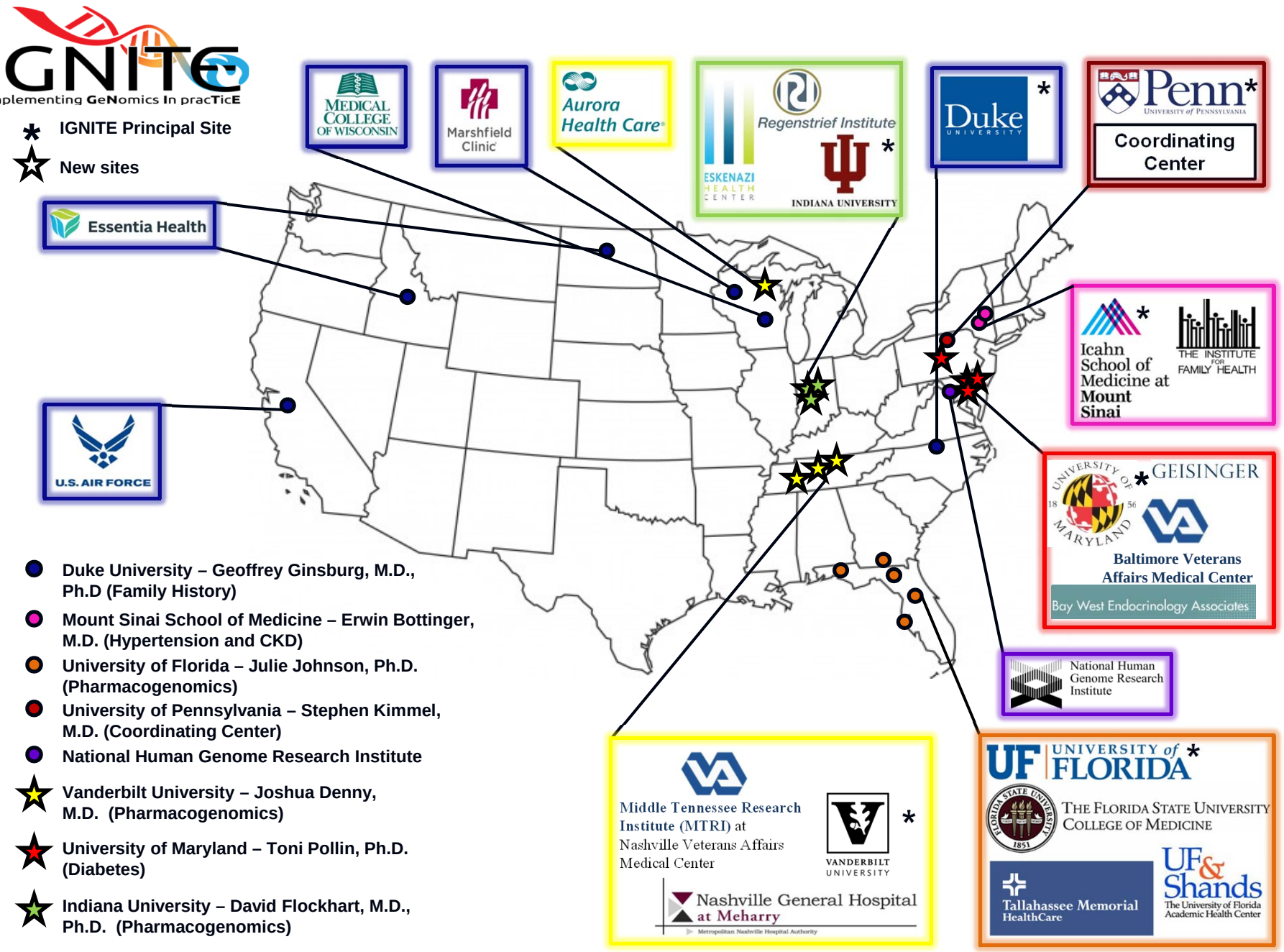
journal homepage: www.elsevier.com/locate/drugalcdep

Short communication

Data compatibility in the addiction sciences: An examination of measure commonality☆

Kevin P. Conway[a,*], Genevieve C. Vullo[a,b], Ashley P. Kennedy[c], Matthew S. Finger[a], Arpana Agrawal[d], James M. Bjork[e], Lindsay A. Farrer[f], Dana B. Hancock[g], Andrea Hussong[h], Paul Wakim[i], Wayne Huggins[g], Tabitha Hendershot[g], Destiney S. Nettles[g], Joseph Pratt[g], Deborah Maiese[g], Heather A. Junkins[j], Erin M. Ramos[j], Lisa C. Strader[g], Carol M. Hamilton[g], Kenneth J. Sher[k]

Document 24


IGNITE
Implementing GeNomics In practicE
* IGNITE Principal Site
New sites
Medical College of Wisconsin
Marshfield Clinic
Aurora Health Care
Regenstrief Institute
Eskenazi Health Center
Indiana University
Duke University
Penn
University of Pennsylvania
Coordinating Center
Essentia Health
U.S. Air Force
Icahn School of Medicine at Mount Sinai
The Institute for Family Health
University of Maryland
Geisinger
Baltimore Veterans Affairs Medical Center
Bay West Endocrinology Associates
National Human Genome Research Institute
Middle Tennessee Research Institute (MTRI) at Nashville Veterans Affairs Medical Center
Vanderbilt University
Nashville General Hospital at Meharry
Metropolitan Nashville Hospital Authority
UF University of Florida
The Florida State University College of Medicine
Tallahassee Memorial HealthCare
UF & Shands
The University of Florida Academic Health Center
Duke University – Geoffrey Ginsburg, M.D., Ph.D (Family History)
Mount Sinai School of Medicine – Erwin Bottinger, M.D. (Hypertension and CKD)
University of Florida – Julie Johnson, Ph.D. (Pharmacogenomics)
University of Pennsylvania – Stephen Kimmel, M.D. (Coordinating Center)
National Human Genome Research Institute
Vanderbilt University – Joshua Denny, M.D. (Pharmacogenomics)
University of Maryland – Toni Pollin, Ph.D. (Diabetes)
Indiana University – David Flockhart, M.D., Ph.D. (Pharmacogenomics)

SBIR and STTR Grants

- **SBIR ($10.5M): 23 Phase I and 9 Phase II**
- **STTR ($1.5M): 2 Phase I and 1 Phase II**
- **Small business funding increasing by ~0.6M/year**
- **2014 ASHG Annual Meeting Workshop:**

"Building your genomics business with SBIR/STTR support from NHGRI and NIH"

Restructuring NHGRI Diversity Action Plan (DAP) Program

- Notices: NOT-HG-14-029, NOT-HG-14-032
- Goal: prepare students to pursue a Ph.D. or M.D./Ph.D.
- Focus on undergraduates, post-baccs, and graduate students
- Annual DAP meeting will be folded into new annual meeting for all trainees
- DAP participation optional but strongly encouraged

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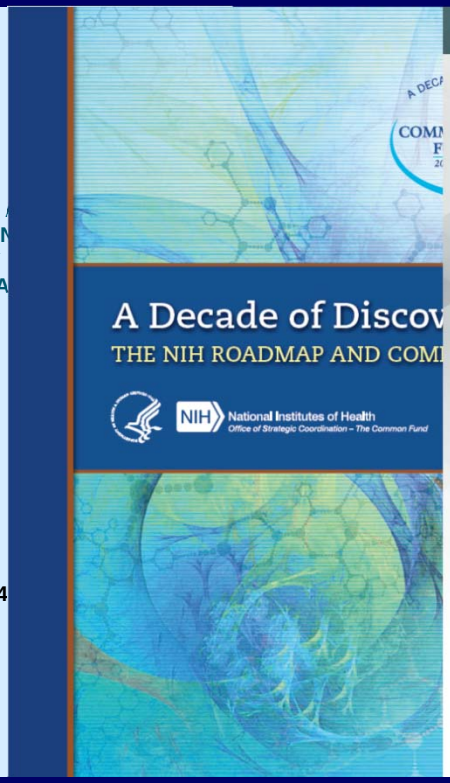
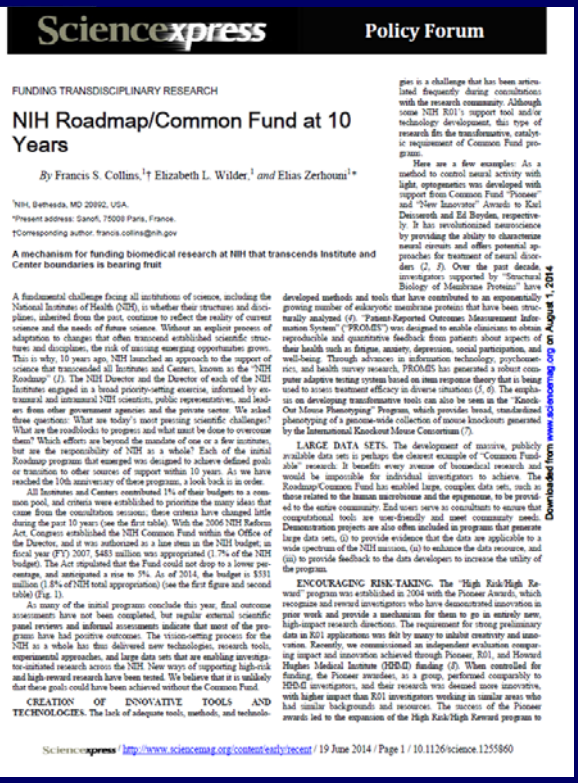
NIH Common Fund Evaluation and 10th Anniversary Commemoration

THE 2014 REVIEW OF PROCESS
OF THE NIH COMMON
REPORT BY
THE COMMON FUND EVALUA
GROUP

June 19, 2014




Sciencexpress Policy Forum

FUNDING TRANSDISCIPLINARY RESEARCH

NIH Roadmap/Common Fund at 10 Years

By Francis S. Collins,[1]† Elizabeth L. Wilder,[1] and Elias Zerhouni[1]*

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A mechanism for funding biomedical research at NIH that transcends Institute and Center boundaries is bearing fruit

A fundamental challenge facing all institutions of science, including the National Institutes of Health (NIH), is whether their structures and disciplines, inherited from the past, continue to reflect the reality of current science and the needs of future science. Without an explicit process of adaptation to changes that often transcend established scientific structures and disciplines, the risk of missing emerging opportunities grows. This is why, 10 years ago, NIH launched an approach to the support of science that transcended all Institutes and Centers, known as the "NIH Roadmap" (1). The NIH Director and the Director of each of the NIH Institutes engaged in a broad priority-setting exercise, informed by extramural and intramural NIH scientists, public representatives, and leaders from other government agencies and the private sector. We asked three questions: What are today's most pressing scientific challenges? What are the roadblocks to progress and what must be done to overcome them? Which efforts are beyond the mandate of one or a few institutes, but are the responsibility of NIH as a whole? Each of the initial Roadmap programs that emerged was designed to achieve defined goals or transition to other sources of support within 10 years. As we have reached the 10th anniversary of these programs, a look back is in order.

All Institutes and Centers contributed 1% of their budgets to a common pool, and criteria were established to prioritize the many ideas that came from the consultation sessions; these criteria have changed little during the past 10 years (see the first table). With the 2006 NIH Reform Act, Congress established the NIH Common Fund within the Office of the Director, and it was authorized as a line item in the NIH budget; in fiscal year (FY) 2007, $483 million was appropriated (1.7% of the NIH budget). The Act stipulated that the Fund could not drop to a lower percentage, and anticipated a rise to 5%. As of 2014, the budget is $531 million (1.8% of NIH total appropriation) (see the first figure and second table) (Fig. 1).

As many of the initial programs conclude this year, final outcome assessments have not been completed, but regular external scientific panel reviews and informal assessments indicate that most of the programs have had positive outcomes. The vision-setting process for the NIH as a whole has thus delivered new technologies, research tools, experimental approaches, and large data sets that are enabling investigator-initiated research across the NIH. New ways of supporting high-risk and high-reward research have been tested. We believe that it is unlikely that these goals could have been achieved without the Common Fund.

CREATION OF INNOVATIVE TOOLS AND TECHNOLOGIES. The lack of adequate tools, methods, and technologies is a challenge that has been articulated frequently during consultations with the research community. Although some NIH R01's support tool and/or technology development, this type of research fits the transformative, catalytic requirement of Common Fund programs.

Here are a few examples: As a method to control neural activity with light, optogenetics was developed with support from Common Fund "Pioneer" and "New Innovator" Awards to Karl Deisseroth and Ed Boyden, respectively. It has revolutionized neuroscience by providing the ability to characterize neural circuits and offers potential approaches for treatment of neural disorders (2, 3). Over the past decade, investigators supported by "Structural Biology of Membrane Proteins" have developed methods and tools that have contributed to an exponentially growing number of eukaryotic membrane proteins that have been structurally analyzed (4). "Patient-Reported Outcomes Measurement Information System" ("PROMIS") was designed to enable clinicians to obtain reproducible and quantitative feedback from patients about aspects of their health such as fatigue, anxiety, depression, social participation, and well-being. Through advances in information technology, psychometrics, and health survey research, PROMIS has generated a robust computer adaptive testing system based on item response theory that is being used to assess treatment efficacy in diverse situations (5, 6). The emphasis on developing transformative tools can also be seen in the "Knock-Out Mouse Phenotyping" Program, which provides broad, standardized phenotyping of a genome-wide collection of mouse knockouts generated by the International Knockout Mouse Consortium (7).

LARGE DATA SETS. The development of massive, publicly available data sets is perhaps the clearest example of "Common Fund-able" research. It benefits every avenue of biomedical research and would be impossible for individual investigators to achieve. The Roadmap/Common Fund has enabled large, complex data sets, such as those related to the human microbiome and the epigenome, to be provided to the entire community. End users serve as consultants to ensure that computational tools are user-friendly and meet community needs. Demonstration projects are also often included in programs that generate large data sets, (i) to provide evidence that the data are applicable to a wide spectrum of the NIH mission, (ii) to enhance the data resource, and (iii) to provide feedback to the data developers to increase the utility of the program.

ENCOURAGING RISK-TAKING. The "High Risk/High Reward" program was established in 2004 with the Pioneer Awards, which recognize and reward investigators who have demonstrated innovation in prior work and provide a mechanism for them to go in entirely new, high-impact research directions. The requirement for strong preliminary data in R01 applications was felt by many to inhibit creativity and innovation. Recently, we commissioned an independent evaluation comparing impact and innovation achieved through Pioneer, R01, and Howard Hughes Medical Institute (HHMI) funding (8). When controlled for funding, the Pioneer awardees, as a group, performed comparably to HHMI investigators, and their research was deemed more innovative, with higher impact than R01 investigators working in similar areas who had similar backgrounds and resources. The success of the Pioneer awards led to the expansion of the High Risk/High Reward program to

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The NIH Common Fund

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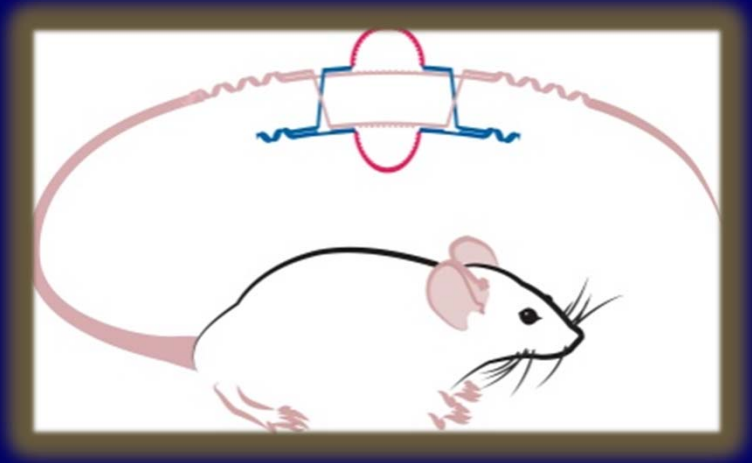
Knockout Mouse Phenotyping Project (KOMP2)



- Annual meeting held in late June
  - New version of website deployed
  - Phenotype data for 1,000 knockout strains uploaded
- CRISPR technology pilots funded
- Metabolomics pilot approved
- Annual PSC Review Meeting in November

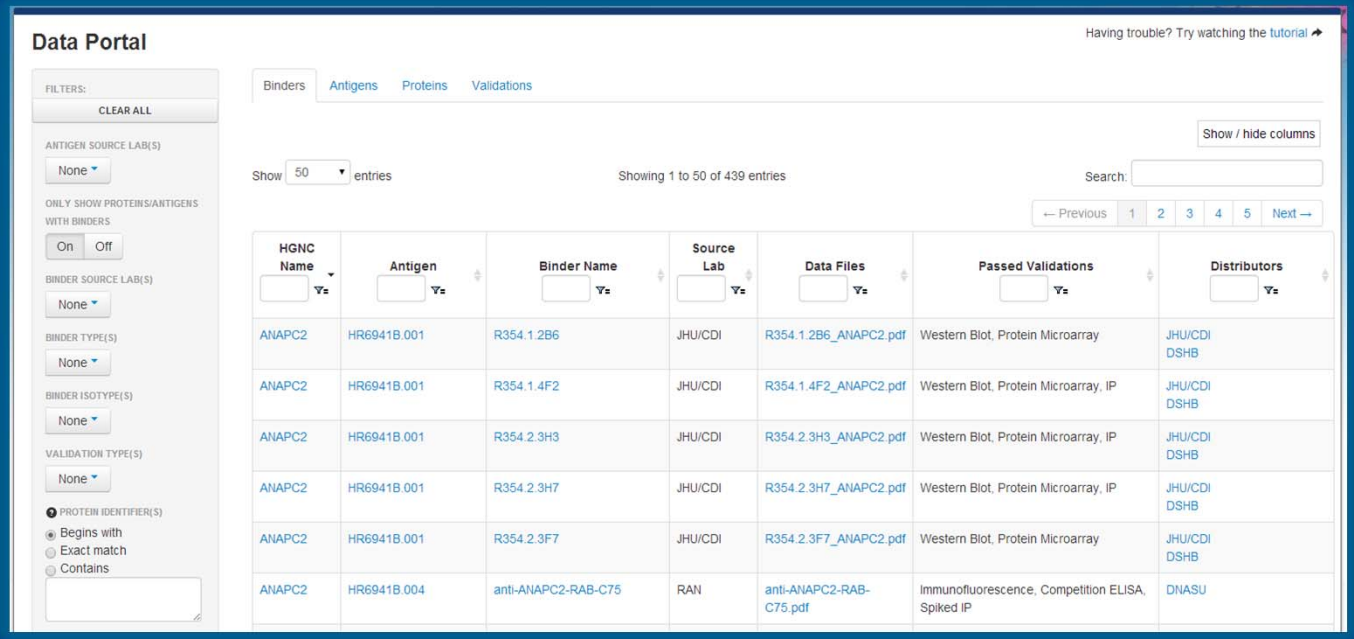
Human Microbiome Project (HMP)

- Phase 2: “integrative HMP (iHMP)”
- iHMP marker paper
  - Published in *Cell Host and Microbe*
  - Immediate open access
- iHMP Data Coordination Center
- Report on 2013 NIH meeting: “Human Microbiome Science: Vision for the Future”
  - Published in BMC *Microbiome*

- **Scale-up phase underway**
  - **700 donors enrolled /12.5K RNA-Seq studies**
  - **Whole-genome sequencing added**
  - **Portal revamped**
- **Biospecimen access mechanism in place**
- **2nd GTEx Community Scientific Meeting**

PROTEIN CAPTURE REAGENTS PROGRAM

- **>400 validated antibodies to human transcription factors available on the Data Portal**



Data Portal

Having trouble? Try watching the tutorial

FILTERS:
CLEAR ALL
ANTIGEN SOURCE LAB(S): None
ONLY SHOW PROTEINS/ANTIGENS WITH BINDERS: On Off
BINDER SOURCE LAB(S): None
BINDER TYPE(S): None
BINDER ISOTYPE(S): None
VALIDATION TYPE(S): None
PROTEIN IDENTIFIER(S): Begins with / Exact match / Contains

Binders | Antigens | Proteins | Validations

Show / hide columns

Show 50 entries — Showing 1 to 50 of 439 entries — Search:

← Previous 1 2 3 4 5 Next →

HGNC Name	Antigen	Binder Name	Source Lab	Data Files	Passed Validations	Distributors
ANAPC2	HR6941B.001	R354.1.2B6	JHU/CDI	R354.1.2B6_ANAPC2.pdf	Western Blot, Protein Microarray	JHU/CDI DSHB
ANAPC2	HR6941B.001	R354.1.4F2	JHU/CDI	R354.1.4F2_ANAPC2.pdf	Western Blot, Protein Microarray, IP	JHU/CDI DSHB
ANAPC2	HR6941B.001	R354.2.3H3	JHU/CDI	R354.2.3H3_ANAPC2.pdf	Western Blot, Protein Microarray, IP	JHU/CDI DSHB
ANAPC2	HR6941B.001	R354.2.3H7	JHU/CDI	R354.2.3H7_ANAPC2.pdf	Western Blot, Protein Microarray, IP	JHU/CDI DSHB
ANAPC2	HR6941B.001	R354.2.3F7	JHU/CDI	R354.2.3F7_ANAPC2.pdf	Western Blot, Protein Microarray	JHU/CDI DSHB
ANAPC2	HR6941B.004	anti-ANAPC2-RAB-C75	RAN	anti-ANAPC2-RAB-C75.pdf	Immunofluorescence, Competition ELISA, Spiked IP	DNASU

- **4th Annual Consortium Meeting in December**

- **LINCS Multi-Program Meeting: July 2014**
- **Funding announcement for Phase 2 investigators to be released in early September**
- **LINCS Phase 2 Consortium Meeting: October 2014**
- **Funding plan for the BD2K-LINCS Data Integration and Coordination Center approved by the BD2K Multi-Council Working Group**

H3Africa Initiative

- 4th H3Africa Consortium Meeting: May 2014

  Cardiovascular Disease, Ethics, & Genome Analysis Workshops

- H3Africa marker paper published in *Science*
- Two new H3Africa ELSI grants
- 5th H3Africa Consortium Meeting: November 2014

  Sickle Cell Disease, Grant Writing, & Custom Chip Workshops

Undiagnosed Diseases Network (UDN)

JAMA The Journal of the American Medical Association

News & Analysis

NIH's Undiagnosed Diseases Program Expands
6 New Sites Offer Potential Answers to More Patients

Bridget M. Kuehn, MSJ

Louise Benge, of Brodhead, Kentucky, enjoyed a normal, active childhood, including playing running games with friends, the 51-year-old woman explained at a National Institutes of Health (NIH) press briefing in July. Then in her teens, Benge developed crippling pain in her hands.

At first, her physician diagnosed her with arthritis. But tests for arthritis were negative, and when the pain spread to her legs in her 20s, another physician suggested that she undergo arterial vascular surgery repeated every few years. Louise declined the surgery. Eventually, each of her 4 siblings developed identical symptoms.

"It hurts to stand and walk, but I keep up with my family," she said.

The cause of the problem remained elusive. Finally, a physician sent Louise and her sister to the Undiagnosed Diseases Program

SCIENCEINSIDER

Breaking news and analysis from the world of science policy

NIH expands program to crack medical mysteries

Science AAAS

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By Kelly Servick | 1 July 2014 4:15 pm

Kelly is a staff writer at *Science*.

Email Kelly

An effort at the U.S. National Institutes of Health (NIH) to diagnose mysterious diseases is undergoing a major expansion. Representatives of the Undiagnosed Diseases Program (UDP), administered by NIH's National Human Genome Research Institute (NHGRI), announced today that six medical centers will join the program, forming a network of clinical sites to investigate intractable cases from patients around the country. The program aims to offer patients a long-awaited diagnosis—and sometimes treatment—while building up data for scientists studying the genetic basis of rare diseases.

Undiagnosed Diseases Network

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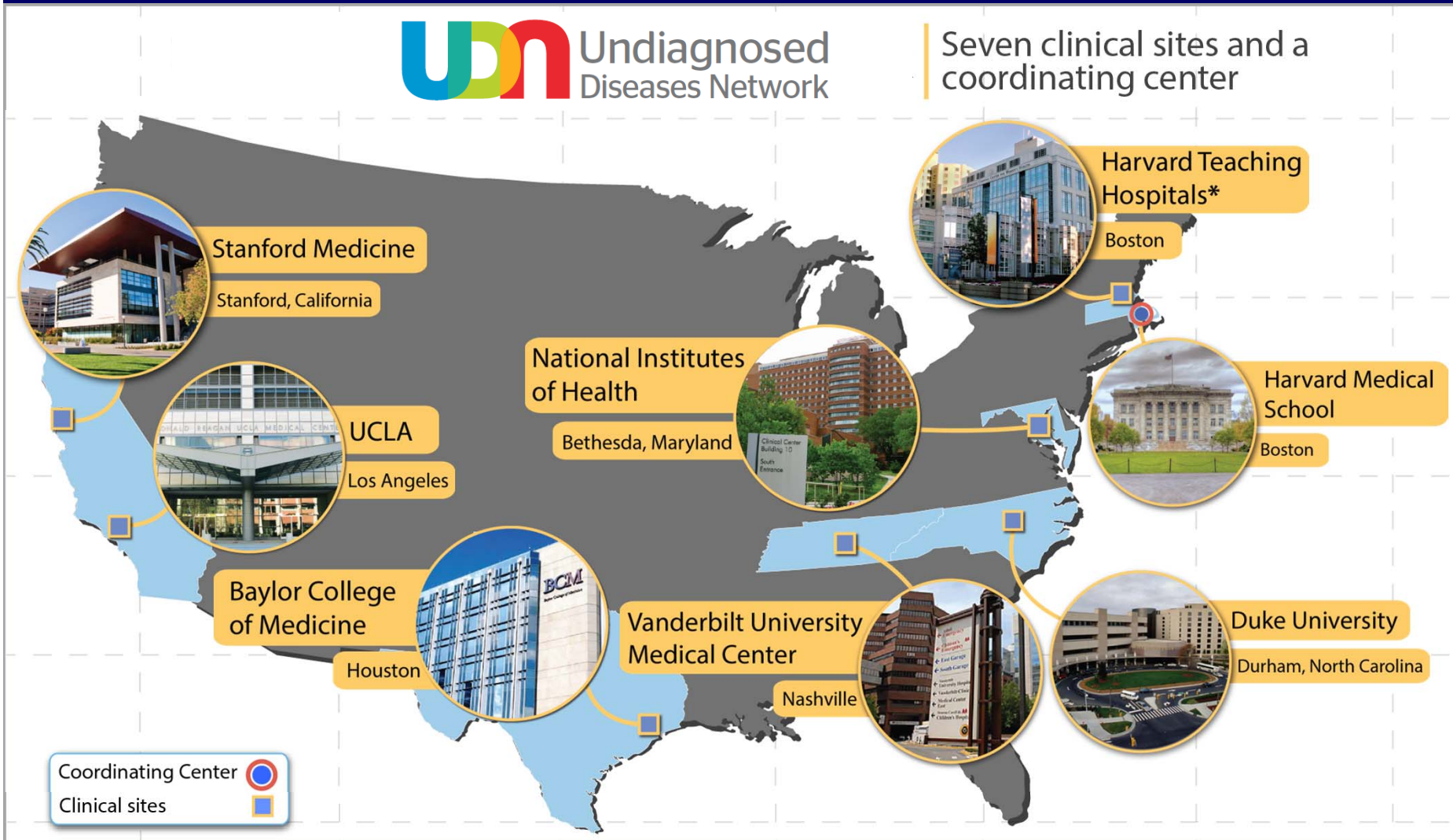
Undiagnosed Diseases Network (UDN)



The NIH site will continue to enroll about 150 patients per year; each of the clinical sites will ultimately enroll about 50 patients per year. A DNA sequencing core facility to be announced in the coming weeks.

*Boston Children's Hospital, Brigham and Women's Hospital and Massachusetts General Hospital participate jointly in the Harvard Center for Integrated Approaches to Undiagnosed Diseases

Big Data to Knowledge (BD2K)

Status of BD2K Programs

- Centers of Excellence
- LINCS-BD2K Perturbation Data Coordination and Integration Center— NHGRI and NHLBI Councils
- Data Discovery Index Coordination Consortium— NHLBI Council
- Joint Kickoff Meeting in November

Big Data to Knowledge (BD2K)

- **Training**

  K01s & R25s— through Multi-Council Working Group

  T32s (new and supplements)— FOAs issued

- **Targeted Software Development— applications received**

BD2K Multi-Council Working Group

- Formed from council representatives to perform council-level review of BD2K applications
- Has met twice
- So far has primarily reviewed applications
- In future will provide input about future projects

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Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

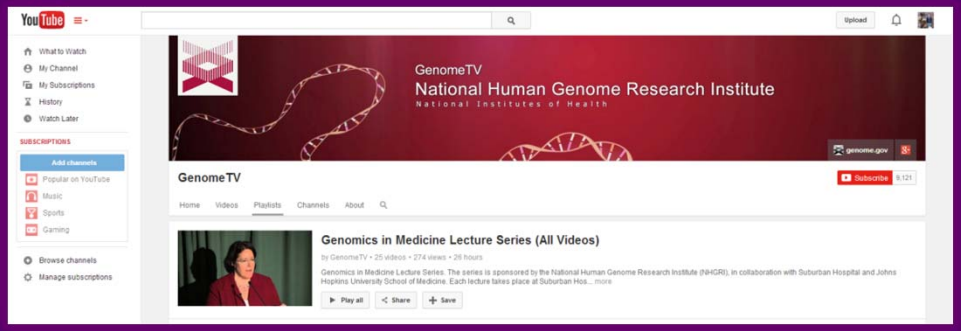
IV. NHGRI Extramural Research Program

V. NIH Common Fund/Trans-NIH

VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

Genomics in Medicine Lecture Series



- Infectious disease
- Rational therapeutics
- Maternal-child health
- Cardiovascular disease
- Autoinflammatory diseases
- Neuromuscular diseases
- Ophthalmic diseases
- Pharmacogenomics
- Oncology
- Neurology
- Psychiatry

FDA Lab-Developed Test Guidance

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FDA Companion Diagnostic Guidance

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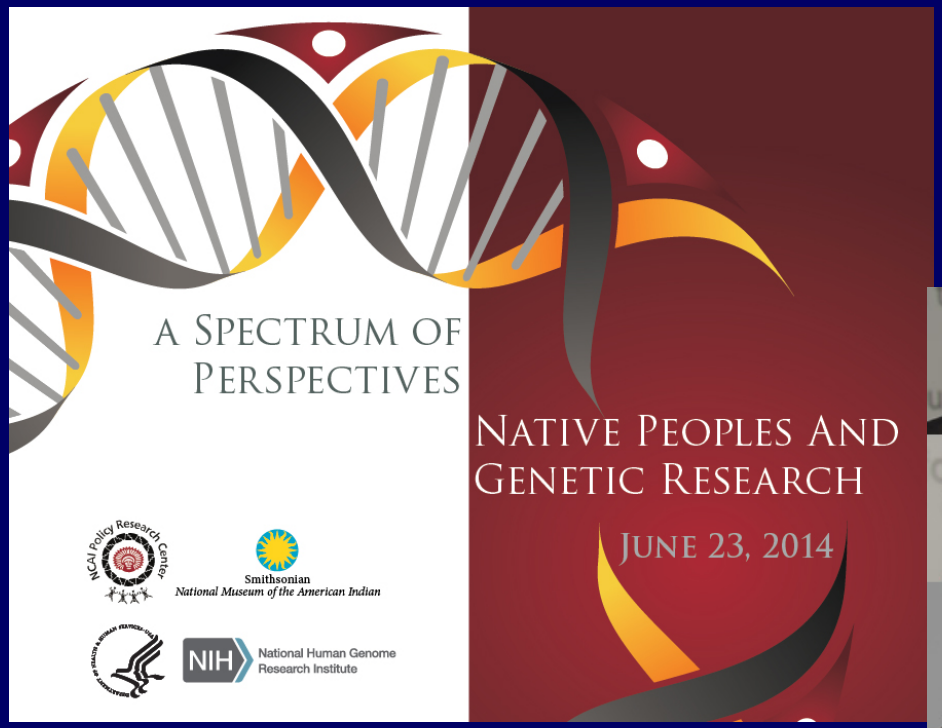
A Spectrum of Perspectives: Native Peoples and Genetic Research



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Brooklyn Community Celebrates Henrietta Lacks

Henrietta Lacks

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2014 NHGRI Summer Workshop in Genomics (Short Course)

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NHGRI Recruitment

Chief

Communications and Public Liaison Branch

Division of Policy, Communications, and Education

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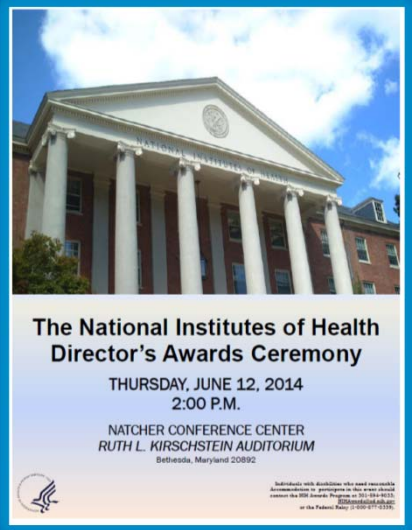
Alan S. Rabson Award for Clinical Care Given to Bill Gahl



NHGRI Intramural Research Highlights

Science Translational Medicine | Science

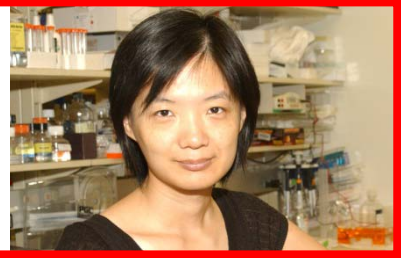
Macrophage Models of Gaucher Disease for Evaluating Disease Pathogenesis and Candidate Drugs

The NEW ENGLAND JOURNAL of MEDICINE

Diagnostic Clinical Genome and Exome Sequencing

PNAS

A Dual Role for Planar Cell Polarity Genes in Ciliated Cells



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The Genomics Landscape

A monthly update from the NHGRI Director

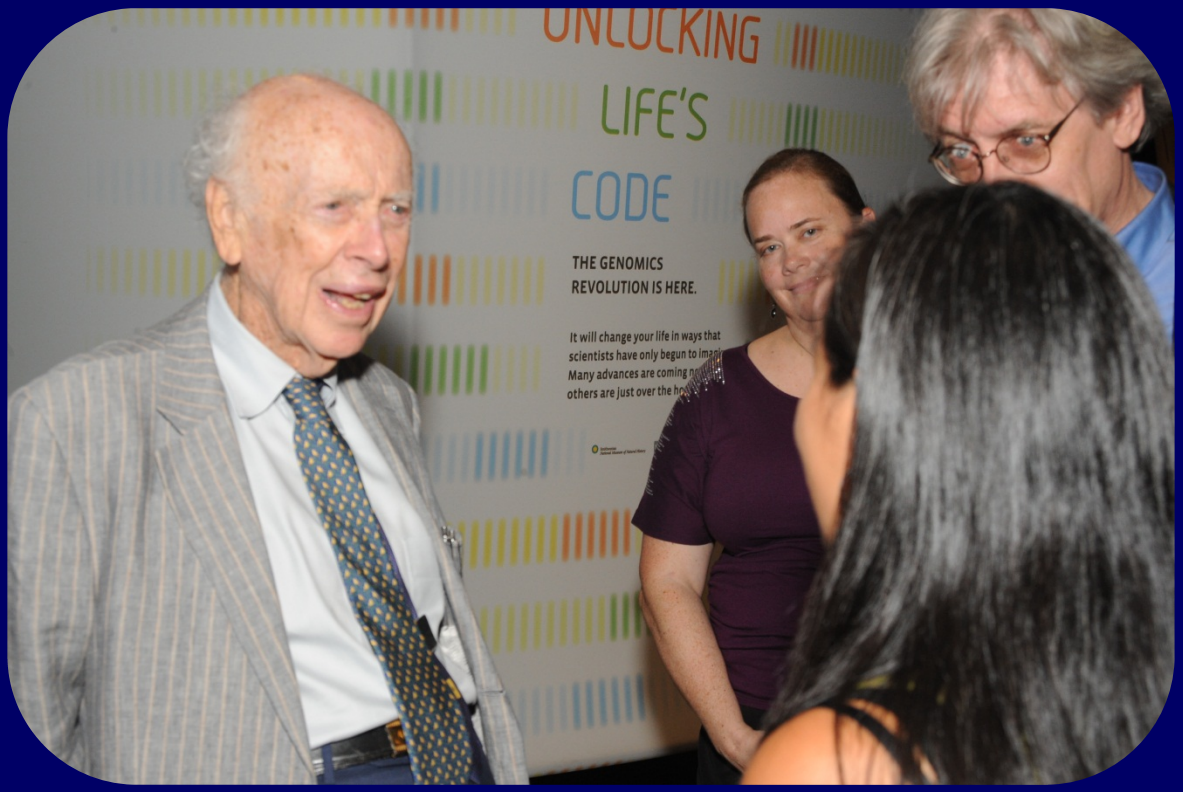
To receive *The Genomics Landscape*, go to list.nih.gov

Search for NHGRILANDSCAPE

Past issues can be accessed at: genome.gov/27527308

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

Thanks!



Special Thanks!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