# Director's Report-Related Documents: February 2015

<table>
<thead>
<tr>
<th>No.</th>
<th>Relevant Documents</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>New Video Spotlights NHGRI Programs</td>
</tr>
<tr>
<td>2</td>
<td>New NHGRI Executive Officer</td>
</tr>
<tr>
<td>3</td>
<td>New Branch Chiefs, NHGRI Division of Policy, Communications, and Education</td>
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<tr>
<td></td>
<td>New Chief, Genomic Healthcare Branch</td>
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<tr>
<td></td>
<td>New Chief, Education and Community Involvement Branch</td>
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<tr>
<td>4</td>
<td>Changing Role for Vence Bonham</td>
</tr>
<tr>
<td>5</td>
<td>Upcoming NHGRI Recruitment: Division of Genomic Medicine</td>
</tr>
</tbody>
</table>
Open Session Presentations

- Update on the Genomic Medicine Working Group
  
  Teri Manolio

- The Alzheimer’s Disease Sequencing Project
  
  Eric Boerwinkle

Concept Clearance:

- Centers of Excellence in ELSI Research
  
  Joy Boyer
Open Session Presentations

Concept Clearances:

- Genome Sequencing Program Analysis Satellites
- Genome Sequencing Program Coordinating Center
- High-Quality Human and Primate Genomes as Foundational Resources
- Comparative and Evolutionary Genomics

Adam Felsenfeld
Open Session Presentations

- Biennial Report on the Inclusion of Women and Minorities in NHGRI-Supported Research
  - Rongling Li
  - Jacqueline Odgis

- Review of the Statement of Understanding between NACHGR and NHGRI
  - Rudy Pozzatti
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
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
New Video Spotlights NHGRI Programs

National Human Genome Research Institute (NHGRI)

Human Genome Project officially begins, 1990

NHGRI launches ENCODE—Encyclopedia of DNA Elements, 2003
New NHGRI Executive Officer

Ellen Rolfes, M.A.
New Chief, Genomic Healthcare Branch

Bob Wildin, M.D.
Changing Role for Vence Bonham

Vence Bonham, Jr., J.D.
New Chief, Education and Community Involvement Branch

Carla Easter, Ph.D.
Upcoming NHGRI Recruitment

Chief, Communications and Public Liaison Branch

Division of Policy, Communications, and Education

Contact: Dr. Laura Lyman Rodriguez
laura.rodriguez@nih.gov or 301-594-7185
Upcoming NHGRI Recruitment

Medical Officer
Division of Genomic Medicine

Vacancy opening soon:
Feb. 16 – Feb. 25, 2015

Contact: Dr. Teri Manolio
manolio@mail.nih.gov or 301-402-2915
NHGRI Implementation of NIH Genomic Data Sharing Policy

- January 25 is effective date for policy
- Consistent implementation across NHGRI portfolio
- Coordinating with NIH-wide implementation
President Obama Visits NIH
Secretary Burwell Visits NIH
President Obama: A Long-Standing Interest in Genomics

Senator Obama, 2006
A broader context for ‘individualizing’ medical care to advance human health
Today: most medical care based on expected response of the average patient

Tomorrow: medical care based on individual in genomic, environmental, and lifestyle differences that enable more precise ways to prevent and treat disease

How do we get from today to tomorrow?
“And that’s why the budget I send this Congress on Monday will include a new Precision Medicine Initiative that brings America closer to curing diseases like cancer and diabetes, and gives all of us access, potentially, to the personalized information that we need to keep ourselves and our families healthier.”

President Barack Obama
January 30, 2015
January 30, 2015

A New Initiative on Precision Medicine
Francis S. Collins, M.D., Ph.D., and Harold Varmus, M.D.

"Tonight, I'm launching a new Precision Medicine Initiative to bring us closer to curing diseases like cancer and diabetes — and to give all of us access to the personalized information we need to keep ourselves and our families healthier."

— President Barack Obama, State of the Union Address, January 20, 2015

The proposed initiative has two main components: a near-term focus on cancers and a longer-term aim to generate knowledge applicable to the whole range of health and disease. Both components are now within our reach because of advances in basic research, including molecular biology, genomics, and bioinformatics. Furthermore, the initiative
Precision Medicine Initiative: The Vision

- **NEAR TERM**: Cancer as a Model of Precision Medicine
  Leading edge of precision medicine, yet more to learn
  Ramp up current efforts to include more cancer types

- **LONGER TERM**: Expanding the Model to Other Diseases
  Create national research cohort of >1 million volunteers
  Generate knowledge base for precision medicine

- **POLICY CHANGES**: Remove Barriers to Clinical Implementation
  Update federal rules protecting research participants
  Advance FDA oversight of precision medicine products
## Precision Medicine Initiative: Proposed Fiscal Year 2016 Funding

<table>
<thead>
<tr>
<th>Agency</th>
<th>$ Million</th>
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<tbody>
<tr>
<td>National Institutes of Health</td>
<td>$200</td>
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<tr>
<td>Food and Drug Administration</td>
<td>$10</td>
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<tr>
<td>Office of the National Coordinator for Health Information Technology</td>
<td>$5</td>
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<tr>
<td><strong>TOTAL</strong></td>
<td><strong>$215</strong></td>
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</table>
Secretary Burwell Speaks Candidly at NIH
Obama seeks $215 million for personalized medicine effort

Obama to Unveil Research Initiative to Develop Tailored Medical Treatments

Obama Announces $215 Million Precision-Medicine Genetic Plan

Obama's $215 Million DNA Sequencing Project Is A Great Idea

A Path for Precision Medicine

Obama Seeks Millions for 'Precision Medicine'
White House fleshes out Obama’s $215 million plan for precision medicine

Obama Enumerates Precision Medicine Initiative
The President requests $215 million to launch his push for personalized clinical care.

Obama to seek $215 million for precision-medicine plan
Details emerge as White House prepares to release budget request to Congress.

U.S. to Develop DNA Study of One Million People
An Obama initiative seeks to channel a torrent of gene information into treatments for cancer, other diseases.
“This is an incredible area of promise,” said Senator Bill Cassidy, Republican of Louisiana and a gastroenterologist. “There will be bipartisan support.”
Precision Medicine Initiative: The Vision

- **NEAR TERM**: Cancer as a Model of Precision Medicine
  - Leading edge of precision medicine, yet more to learn
  - Ramp up current efforts to include more cancer types

- **LONGER TERM**: Expanding the Model to Other Diseases
  - Create national research cohort of >1 million volunteers
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- **POLICY CHANGES**: Remove Barriers to Clinical Implementation
  - Update federal rules protecting research participants
  - Advance FDA oversight of precision medicine products
National Research Cohort

- >1 million U.S. volunteers
  - Numerous existing cohorts (many funded by NIH)
  - New volunteers
- Participants to share genomic data, lifestyle information, biological samples – all linked to their EHRs
- Provide scientists with a ready platform for myriad new studies to propel understanding of health and disease
- Forge new model for ‘doing science’ that emphasizes engaged participants and open, responsible data sharing with strong privacy protections
National Research Cohort: Components

- **Participants**
  - Existing cohorts
  - New volunteers

- **Data**
  - Medical history
  - Diet and lifestyle
  - Environmental exposures
  - Biological samples

- **Research**
  - Cohort researchers
  - Other qualified researchers

- >1 million diverse participants
- Centralized database
- Scientific community
Building a Large U.S. Cohort for Precision Medicine Research

- NIH workshop to be held February 11-12, 2015
- Representatives from a wide variety of fields
- Major areas of discussion:
  - Cohort identification and participant recruitment
  - Participant engagement, data privacy, and novel ways of returning information to participants
  - Data collection, including mobile technologies
  - Informatics and electronic health records
The Precision Medicine Initiative: Data-Driven Treatments as Unique as Your Own Body
Precision Medicine Initiative

Far too many diseases do not have a proven means of prevention or effective treatments. We must gain better insights into the biology of these diseases to make a difference for the millions of Americans who suffer from them. Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person. While significant advances in precision medicine have been made for select cancers, the practice is not currently in use for most diseases. Many efforts are underway to help make precision medicine the norm rather than the exception. To accelerate the pace, President Obama has now unveiled the Precision Medicine Initiative—a bold new enterprise to revolutionize medicine and generate the scientific evidence needed to move the concept of precision medicine into every day clinical practice.

www.nih.gov/precisionmedicine
New U.S. Surgeon General

Vivek Murthy, M.D.
U.S. FDA Commissioner to Step Down

Margaret Hamburg, M.D.
Retirement of Donald Lindberg

Donald Lindberg, M.D.

Betsy Humphreys, M.L.S.

NIH NLM
Co-Chairs: Eric Green & Harlan Krumholz

Charge:

- Review mission, organization, and programmatic priorities
- Articulate a strategic vision for NLM

Report Due Date:
June 2015 ACD Meeting
Cessation of the National Children’s Study
Renaming of an NIH Center: NCCIH

NCCAM is now NCCIH
New Associate Director for Science Policy, NIH

Carrie Wolinetz, Ph.D.
New Deputy Director, National Institute of General Medical Sciences

Judith Greenberg, Ph.D.
‘CRomnibus’ passed, establishing NHGRI Fiscal Year (FY) 2015 funding

On February 2, President’s Fiscal Year 2016 budget sent to Congress
Director’s Report Outline

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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
Mourning the Loss of Mary Lyon
National Medal of Science and National Medal of Technology and Innovation

Bruce Alberts, Ph.D.

Douglas Lowy, M.D. and John Schiller, Ph.D.
Lasker–Koshland Special Achievement Award in Medical Science

Mary-Claire King, Ph.D.
Awards at 2014 ASHG Annual Meeting

David Valle, M.D.

Gonçalo Abecasis, D.Phil.

Mark Daly, Ph.D.
2015 Breakthrough Prize in Life Sciences
Elected to the Institute of Medicine

Goncalo Abecasis
Todd Golub
Julie Johnson
Harry Orr
Joe Takahashi
New Director, Woods Hole Marine Biological Laboratory

Huntington Willard, Ph.D.
New Executive VP of Global Research and Chief Scientific Officer, Vertex

David Altshuler, M.D., Ph.D.
The Scientist’s Top Ten Innovations 2014

1. DRAGEN Bio-IT Processor • Edico Genome
2. MiSeqDx • Illumina
3. HiSeq X Ten • Illumina
4. IrysChip V2 • BioNano Genomics
5. RainDrop Digital PCR System • RainDance Technologies
Researchers detect cancer precursors in blood DNA before disease develops

By Jacqueline A. Odgis
Scientific Program Analyst, NHGRI

Cost-effective, high throughput technologies used to analyze DNA are uncovering variations in our genetic code. Increasing numbers of these variations, sometimes referred to as mutations, are implicated in disease, including many cancers. With the ability to sequence DNA in the clinic, doctors can more definitively diagnose and predict patients’ personal risks for developing cancer, based on the presence of these variants in their DNA.

For some types of cancer, the damaging variations are inherited. Thus, our parents’ medical history might provide clues to disease risks to look for in ourselves. However, there are other types of changes that increase cancer risk that form in our cells after birth, before symptoms appear.

The question then is, how do we seek out those individuals who may be at higher risk, but have no history of cancer in their family? To help solve this problem, scientists are investigating “pre-cancerous” mutations that might allow us to catch, monitor and possibly treat cancer caught early in otherwise healthy patients.

The December Genome Advance of the Month explores the use of specific genetic mutations to identify patients at high risk for cancer, even in people without a family history of the disease. The research team, led by Giulio Genovese, Ph.D., of the Broad Institute at MIT and Harvard in Boston, focused on precursors for blood cancers like leukemia, lymphoma and myeloma. Though innovative drug therapies and increasing access to treatments have dramatically improved blood cancer survival, blood cancer remains one of the most common forms of cancer in the United States and worldwide. In 2014, blood cancer accounted for approximately 9.4 percent of the estimated 1,665,540 new cancer cases diagnosed across the country.
Genomes In The News…
Director’s Report Outline

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VII. NHGRI Intramural Research Program
Genome Sequencing Program

- Two RFAs released in mid-December 2014:
  - Centers for Common Disease Genomics (UM1): RFA-HG-15-001

- Applicant information webinar on February 18

- Submission deadline for both RFAs is April 7
Large-Scale Genome Sequencing and Analysis Centers

- 54 new papers in most recent quarter
  - Microbiome
  - Comparative Genomics
  - Inherited Disease
  - Cancer
  - Technology Development
### TCGA Exome Sequencing Progress

![Bar Graph showing TCGA Exome Sequencing Progress](image)

### TCGA Tumor Publication Status

<table>
<thead>
<tr>
<th>Cancer Type</th>
<th>Status</th>
<th>Year</th>
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<tbody>
<tr>
<td>Glioblastoma Multiforme 1</td>
<td>Nature</td>
<td>2008</td>
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<tr>
<td>Ovarian Carcinoma</td>
<td>Nature</td>
<td>2010</td>
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<tr>
<td>Breast Cancer</td>
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<tr>
<td>Colorectal Adenocarcinoma</td>
<td>Nature</td>
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<td>Lung Squamous Cell Ca</td>
<td>Nature</td>
<td>2012</td>
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<tr>
<td>Acute Myeloid Leukemia</td>
<td>NEJM</td>
<td>2013</td>
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<td>Uterine Corpus Endometrial Ca</td>
<td>Nature</td>
<td>2013</td>
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<td>Kidney Renal Clear Cell Ca</td>
<td>Nature</td>
<td>2013</td>
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<tr>
<td>Glioblastoma Multiforme 2</td>
<td>Cell</td>
<td>2013</td>
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<tr>
<td>Bladder Urothelial Carcinoma</td>
<td>Nature</td>
<td>2014</td>
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<tr>
<td>Lung Adenocarcinoma</td>
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<td>Stomach Adenocarcinoma</td>
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<td>Chromophobe Renal Cell Ca</td>
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<td>Papillary Thyroid Carcinoma</td>
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<td>Head and Neck Squamous Ca</td>
<td>Nature</td>
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<tr>
<td>Brain Lower Grade Glioma</td>
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<tr>
<td>Skin Cutaneous Melanoma</td>
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**Document 34**
Discovery

- Discovery of over 600 causal genes for Mendelian conditions
- Over 125 publications

Public Project List

- Downloadable project list
- Annotation – OMIM #, project site, interested in additional cases or not
- About 1000 Mendelian disorders currently listed
Launched in December 2013
Clinical Sequencing Exploratory Research (CSER) Program

- Enrolled 3,058 adults, 707 children; 2,661 germline, 510 tumor sequences

Bennette et al., *Genetics in Medicine*, 2014
Clinical Sequencing Exploratory Research (CSER) Program

Examples of recent impact:

- >130 publications, 9 working group publications
- >200 presentations/posters

ASHG 2014
San Diego, CA October 18-22

ASBH 16th Annual Meeting
October 16-19, 2014
Hilton San Diego Bayfront Hotel • San Diego, CA

INSTITUTE OF MEDICINE
OF THE NATIONAL ACADEMIES
Genome Sequencing Informatics Tools

- iSeqTools provides GKNO pipelines that integrate results from multiple variant or mutation callers

  - Uses graph realignment to make best call
  - IOBIO ‘apps’ for rapid, easy, visual analysis of data
DNA Sequencing Technology Development

- Grantee meeting and public meeting in May 2015

Sequencing Technology Grantees
Planning workshop: “From Genome Function to Biomedical Insight: ENCODE and Beyond”
March 10-11, 2015; NIH Campus

Annual ENCODE Consortium Meeting
March 14-17, 2015; Cold Spring Harbor Laboratory

ENCODE User’s Meeting with hands-on workshops
June 29-July 1, 2015; Bolger Center, Potomac, MD
ENCODE Publications

Mouse ENCODE integrative and companion papers published in late 2014
Genomics of Gene Regulation Project

- Goal: To learn how to derive predictive gene regulatory networks starting from genomic data
- Five awards issued in January 2015
- Biological systems include immune system, skin, and nuclear hormone receptor response
Centers of Excellence in Genomic Science (CEGS) Program

Two new CEGS awards:

- Stanford University, Center for Personal Dynamic Regulomes
- Harvard Medical School, Neuropsychiatric Genome-Scale and RDoC Individualized Domains (N-GRID)
GAIN Data Access Committee

- GAIN DAC retired in late 2014 and all GAIN datasets transferred to relevant NIH DACs
- Nearly 1,700 project requests from 878 investigators over the course of 8 years

Cumulative Project Requests

<table>
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<th>Year</th>
<th>Project Requests</th>
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<td>2013</td>
<td>700</td>
</tr>
<tr>
<td>2014</td>
<td>800</td>
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Beginning early March 2015, the GWAS Catalog infrastructure will be migrating to the European Bioinformatics Institute (EMBL-EBI). Content prior to this time will continue to be available on this NHGRI web page, but additional content updates will be limited. We will provide updates here as they are available; interested users can sign up for email updates at gwas-announce@ebi.ac.uk.
AMIA 2014 Distinguished Paper & Homer R. Warner Awards

Development and validation of an electronic phenotyping algorithm for chronic kidney disease

SOEMPI: A Secure Open Enterprise Master Patient Index Software Toolkit for Private Record Linkage
Csaba Toth, Elizabeth A. Durham, Murat Kantarcioğlu, Yuan Xue, Bradley Malin
- NIMH suicide and PTSD measures released in December 2014
- New NHLBI funding for sickle cell disease
- PhenX protocols released in REDCap
PAGE Multi-Ethnic Genotyping (MEGA) Array

- Custom Content
- 36,000 Multiethnic Exomes
- ClinVar, OMIM
- 1000 Genomes Project
- Existing
Standardizing clinical assessment of genomic variants and their deposition into ClinVar

GenomeConnect: ClinGen’s Patient Portal

ClinGen-DECIPHER Public Meeting in May 2015
Newborn Sequencing In Genomic Medicine and Public Health (NSIGHT)
Genomics and Society Working Group

- Next in-person meeting in April 2015
- Outgoing members: Tim Caulfield, Jeff Long, Andrea Patenaude, and David Williams
- New members: Chanita Hughes-Halbert, Barbara Bernhardt, and David Veenstra
- Change in leadership:

Pamela Sankar, Ph.D.
University of Pennsylvania

Lisa Parker, Ph.D.
University of Pittsburgh
ELSI Research Program

- New ELSI NRSA Institutional Training Grant (T32) mechanism

- Centers of Excellence in ELSI Research (CEER):
  
  First Regional CEER Networking Meeting
  February 23-24, 2015

  Annual CEER Investigator Meeting
  March 16-18, 2015

  New CEER RFA Concept Clearance
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Human Microbiome Project (HMP)

- HMP Phase 2: ‘integrative HMP’ (iHMP)
- iHMP marker paper (open access)
- iHMP Data Coordination Center
- 2nd iHMP Consortium Meeting
  June 2015
  Bethesda
- 5th International Human Microbiome Consortium Congress
  March 2015
  Luxembourg
Knockout Mouse Phenotyping Project (KOMP2)

- International Mouse Phenotyping Consortium annual meeting in November
- Planning for renewal of KOMP2 program
- Sexual dimorphism evident in mutant phenotypes in the KOMP2 database
- Six investigator-initiated LINCS Data and Signature Generation Centers
- One NIH-initiated BD2K- LINCS Data Coordination and Integration Center
- First trans-LINCS meeting in October
- Data/tools release milestones for all centers developed
- Collaborative data science research opportunity announcement published
H3Africa

- 5th Consortium Meeting in November (Tanzania)
  - Grant Writing Workshop
  - Study Coordinators Session
  - Sickle Cell Disease Workshop

- Supplement for whole-genome sequencing
- Progress on developing custom genotyping chip
- Supplement for research ethics training
- 6th Consortium Meeting in May (Zambia)
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.
<table>
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<tr>
<th>PI Name</th>
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<th>Patient Phenotype</th>
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<tr>
<td>Worley</td>
<td>FRMPD4</td>
<td>mouse, human male cohort</td>
<td>developmental delay and regression, seizures</td>
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<td>Lin</td>
<td>HK1, SUSD4, CCDC89, and BAI2</td>
<td>zebrafish</td>
<td>neurodegeneration and spastic tetraplegia like syndrome</td>
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<td>recessive disease phenotypes</td>
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<td>ATP1A3, SYNE1, SNAP29, ARHGAP22, KIF4B and XRN1</td>
<td>patient primary fibroblasts, drosophila</td>
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<td>zebrafish</td>
<td>epilepsy</td>
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Big Data to Knowledge (BD2K) Initiative

Funded BD2K Elements

- Centers of Excellence, Data Discovery Index Coordination Consortium, Short Courses, Open Educational Resources, Mentored Career Development
  - Joint kickoff meeting in November

Recently Reviewed Elements

- Targeted Software Development
- Institutional Pre-Doctoral Training Programs
Future BD2K Efforts

- The Commons
- NIH Standards Information Resource
- Database Sustainability
- Training Coordination Center
- Diversity
iDASH center and collaborators develop solutions to problems in data sharing and privacy protection

2nd iDASH privacy challenge in March 2015

Challenge 1: Homomorphic encryption

Challenge 2: Secure multiparty computing for secure genomic data analysis across institutions
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Inter-Society Coordinating Committee for Practitioner Education in Genomics

- Physicians and Dentists
- Pharmacists and Nurses
- Genetic Counselors

- In-person meeting in November 2014
- New working groups:
  - Physician-patient Communications
  - Innovative Approaches to Education
Physician Resources Now on G2C2

Search for Genetics & Genomics Resources to Use in Your Classroom or in Your Practice
Find websites, books, articles and more - enhance your class content with peer-reviewed resources.

NEW!
Newborn Screening Saves Lives Reauthorization Act of 2014
Clinical Trials Policy Update

- New definition of “clinical trial”
- Reporting requirements
- Centralized Institutional Review Boards (IRBs)
FDA Lab-Developed Test (LDT) Guidance

JAMA
The Journal of the American Medical Association

Genetic Testing and FDA Regulation
Overregulation Threatens the Emergence of Genomic Medicine
James P. Evans, MD, PhD; Michael S. Watson, PhD

Should the FDA regulate laboratory-developed diagnostic tests? — No.

In November 2014, the US Food and Drug Administration (FDA) revealed its intent to regulate thousands of medical diagnostic tests being performed in as many as 13,000 clinical laboratories throughout the United States, focusing especially on genomic medicine. Although the FDA is well intentioned, the current plan for regulation is unnecessary and, if carried out, could result in the closure of many laboratories, undermine innovation, and potentially limit patient choice. Moreover, the proposed regulation, if unchanged, is likely to lead to thousands of laboratory submissions to the FDA, for which its own staffing capacity is tenuous at best. If implemented, the requirements may have the unintended effect of derailing the long-awaited emergence of genomic medicine.

The last several years have seen substantial expansion in genetic testing, resulting from advances in technology that allow rapid and accurate sequencing of large fractions of an individual's DNA. Such advances have begun to inform patient care in spheres ranging from carrier screening and the diagnosis of birth defects to individualized diagnosis and treatment of cancer. These developments have occurred in the span of just a few years, in large part because of the nimbleness of relatively small clinical and academic laboratories that can quickly respond to new medical findings and patient needs by rapidly and safely developing and improving laboratory-developed tests. The resulting landscape is one of vibrant competition in which laboratories that offer genetic testing now compete on the basis of quality, service, innovation, and cost.

The FDA now proposes to regulate laboratory-developed tests as “medical devices,” mandating that laboratories be treated as manufacturers that must meet formal FDA manufacturer requirements for each test developed—a costly and time-consuming process. However, this approach has little valid or even apparent justification. The FDA has failed to cite more than a few anecdotal examples of patient harm to justify its proposed actions. During a congressional hearing on the draft regulation, the director of the

FDA Regulation of Laboratory-Developed Diagnostic Tests
Protect the Public, Advance the Science
Joshua Sharfstein, MD

Should the FDA regulate laboratory-developed diagnostic tests? — Yes.

In April 2014, the Centers for Disease Control and Prevention and the US Food and Drug Administration (FDA) published a warning in Morbidity and Mortality Weekly Report about a commercially available test for Lyme infection. The test returned the result “culture positive,” when in fact the procedure was far more complex than a routine culture. There were serious concerns about false-positive results caused by laboratory contamination, leading to “the potential for misdiagnosis.”

The questionable assay was a laboratory-developed test, meaning an “in vitro diagnostic test that is designed, manufactured, and used within a single laboratory.” Laboratory-developed tests exist in a regulatory crevice. Because of its broad statutory authority over products “intended for use in the diagnosis of disease or other conditions,” the FDA considers laboratory-developed tests under its jurisdiction. Yet for many years, the FDA has taken the position that there were too few of these tests, and that they were of sufficiently low risk, to merit oversight. As a result, tests “designed, manufactured, and used within a single laboratory” are not subject to the standards for quality and validity applicable to other diagnostic tests, such as those made by medical device manufacturers.

Recently, however, the FDA has expressed concern with the proliferation of laboratory-developed tests, their marketing, and their potential to mislead physicians and patients and undermine clinical care. On July 31, 2014, the agency notified Congress that the agency would shortly release a draft guidance document containing a framework for the application of agency standards for quality, safety, and validity to laboratory-developed tests. On September 30, the agency posted the draft guidance document to its website and opened a comment period lasting until February 4, 2015. The agency will hold a public meeting on the topic on January 8 and 9, 2015, at the National Institutes of Health in Bethesda, Maryland.
FDA Workshop on Oversight of ‘Next-Generation’ Genome Sequencing

Public Workshop: Optimizing FDA’s Regulatory Oversight of Next-Generation Sequencing Diagnostic Tests
February 20, 2015
Informed Consent Resource for Genomics

- Discussion of topics essential to genomics research
- Sample language and consent forms
- Relevant regulations and policies
- Email: informedconsent@mail.nih.gov
Genome: Unlocking Life’s Code Exhibition

Military Family Day

Military Family Day at the Science Center in San Diego’s Balboa Park

Sunday, November 9, 10:00 am-6:00 pm

- Free admission to the Fleet Center, hands-on activities, and resource fair
- Lunch will be provided for the first 500 registrants


Sponsored by:

Questions? Please email Kat Brown at SDMilitaryFamilyDay@gmail.com, or call (858) 800-1400

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Genome: Unlocking Life’s Code Exhibition

Travel Schedule

2015

January 22-April 27:
The Tech Museum of Innovation
San Jose, CA

May 15-September 10
The Saint Louis Science Center
St. Louis, MO

October 2-January 3
Oregon Museum of Science and Industry
Portland, OR

2016

January 18-April 25
Discovery World Milwaukee
Milwaukee, WI
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
2014 CHANEL-CERIES Research Award

Julie Segre, Ph.D.
2014 Rare Voice Award

William A. Gahl, M.D., Ph.D.
2014 NSGC Leadership Award

Natalie Weissberger Paul National Achievement Award

Barbara B. Biesecker, M.S., Ph.D., C.G.C.
2014 South African Medical Research Council Scientific Merit Award

Charles Rotimi, Ph.D.
2014 AJHG C.W. Cotterman Award

Shurjo Sen, Ph.D.
NHGRI Intramural Research Highlights

The African Genome Variation Project shapes medical genetics in Africa

Biogeography and individuality shape function in the human skin metagenome

Vector design influences hepatic genotoxicity after adeno-associated virus gene therapy
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genome.gov/27527308
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Special Thanks!