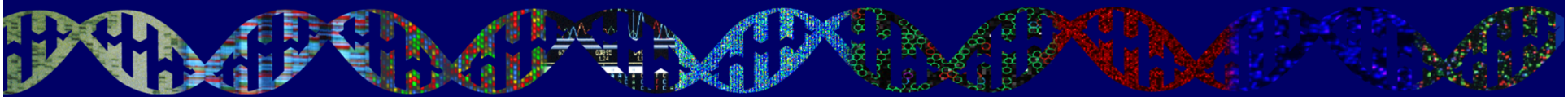
DIRECTOR'S REPORT

National Advisory Council for Human Genome Research

September 2015

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

NIH National Human Genome Research Institute



Director's Report-Related Documents: September 2015

Director's Report

Director's Report

No.	Relevant Documents
1	Mourning the Loss of Elizabeth Thomson
2	Departure of Chief, Policy and Program Analysis Branch
3	New Chief, Communications and Public Liaison Branch
4	**New ASHG-NHGRI Fellows** NHGRI Welcomes 2015 ASHG/NHGRI Education and Public policy fellows Genetics and Public Policy Fellowship Genetics and Education Fellowship
5	New NHGRI Brochure
6	NHGRI Implementation of NIH Genomic Data Sharing Policy
7	Genomics and Health Disparities Lecture Series

genome.gov/DirectorsReport

Document #

Open Session Presentations

- Genomics and Society Working Group

  Lisa Parker

- Workshop Report: Research Directions in Stevens Johnson Syndrome – Toxic Epidermal Necrolysis

  Teri Manolio

Open Session Presentations

- **Genomic Medicine Working Group &**
  Meeting Report: Genomic Medicine 8

  Teri Manolio

- **The Complementarity of Comparative Effectiveness Research and Precision Medicine**

  Joe Selby

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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Mourning the Loss of Elizabeth Thomson

Document 1

Departure of Chief, Policy and Program Analysis Branch

Derek Scholes, Ph.D.

Document 2

New Chief, Communications and Public Liaison Branch

John Ohab, Ph.D.

New ASHG-NHGRI Fellows

Caroline Young, M.S.

Genetics and Public Policy Fellow

Julie Nadel, Ph.D.

Genetics and Education Fellow

Document 4

New NHGRI Brochure

- New brochure features NHGRI's history, organization, core values, and research portfolio across four major scientific areas
- PDF version on genome.gov
- Printed copies available

Document 5

NHGRI Implementation of NIH Genomic Data Sharing Policy


NHGRI Implementation of the NIH Genomic Data Sharing (GDS) Policy

The National Human Genome Research Institute (NHGRI) supports and complies with all National Institutes of Health (NIH) data sharing policies. Information about general NHGRI expectations for implementation of the NIH Genomic Data Sharing (GDS) Policy is provided below.

If applicable, Funding Opportunity Announcements (FOA) will specify additional data sharing expectations for specific programs. Information about the NIH GDS Policy and expectations is available through the NIH GDS Policy website. NHGRI will update this implementation plan as needed to maintain consistency with program priorities, agency policies or any trans-NIH implementation guidance.

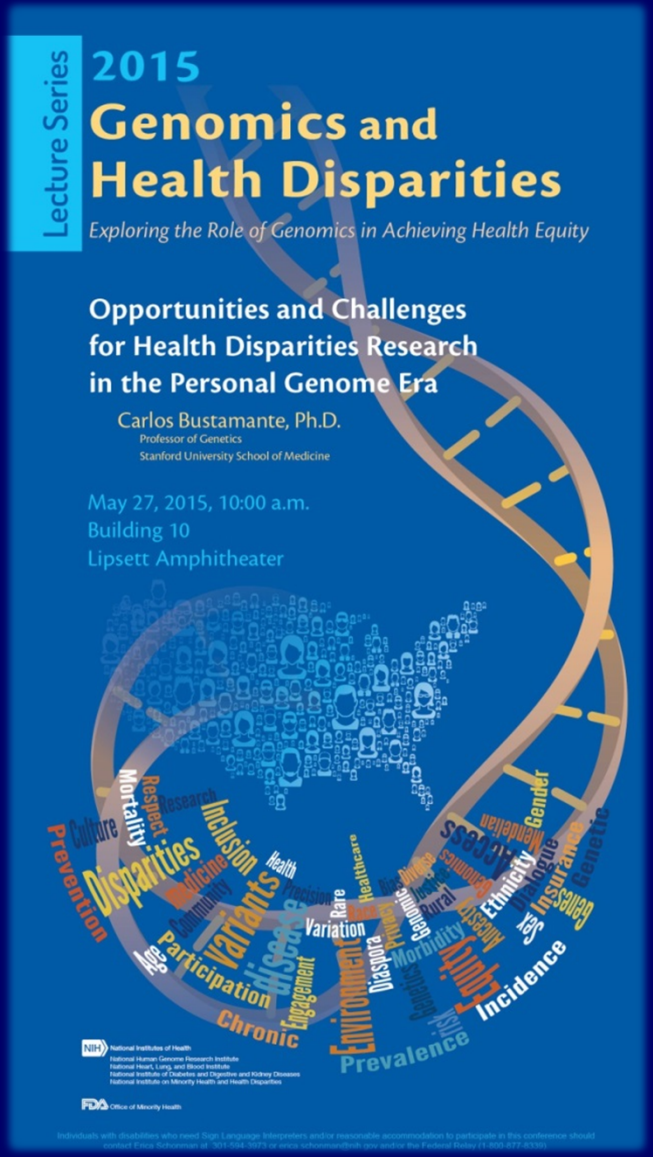
- Overarching Principles and Applicability
- Informed Consent
- Data Sharing Plans
- Exceptions to Data Deposition and Alternative Data Sharing Plans
- Data Submission and Release
- Governance and Contact Information
- Additional Information

NHGRI-specific expectations:

- Same data sharing timelines for human and non-human genomic data on or after January 25, 2016
- Move toward using data sources with explicit consent for future research use and broad data sharing by early 2020

Document 6

Genomics and Health Disparities Lecture Series



Inaugural Lecture

May 27, 2015, 10:00 a.m. - 11:00 a.m.

Opportunities and Challenges for Health Disparities Research in the Personal Genome Era

Carlos Bustamante, Ph.D.

Document 7

NHGRI Roundtable on Inclusion and Engagement of Underrepresented Populations in Genomics Research

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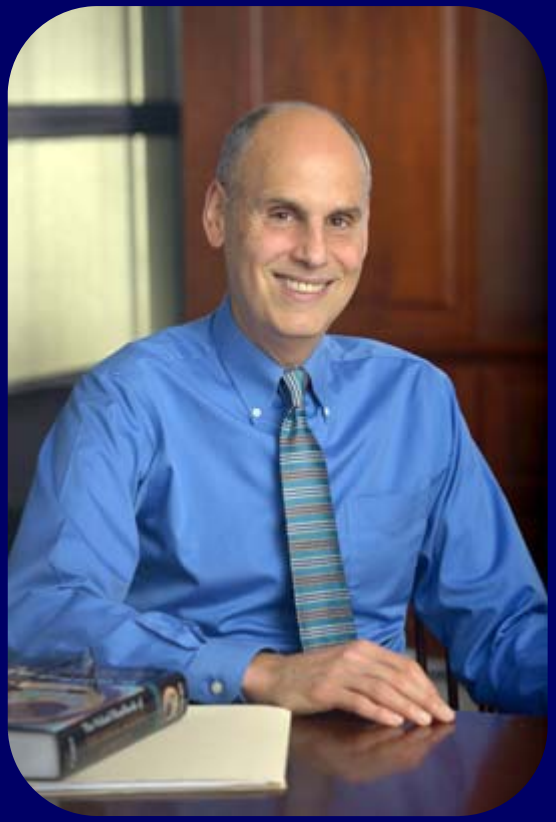
Alan Guttmacher Retires as NICHD Director



Alan Guttmacher, M.D.

Catherine Spong, M.D.

NIH *Eunice Kennedy Shriver* National Institute of Child Health and Human Development

Document 8

New Director, National Institute of Neurological Disorders and Stroke

Walter Koroshetz, M.D.

NIH National Institute of Neurological Disorders and Stroke

Document 9

New Director, NIH Office of Behavioral and Social Sciences Research

Bill Riley, Ph.D.

NIH National Institutes of Health

Document 10

New Director, NCATS Office of Rare Diseases Research

Petra Kaufmann, M.D., M.Sc.

NIH National Center for Advancing Translational Sciences

OFFICE OF RARE DISEASES RESEARCH

Document 11

Acting Director,
NIH Office of AIDS Research

Robert Eisinger, Ph.D.

NIH National Institutes of Health

Document 12

Sally Rockey Steps Down as Director, NIH Office of Extramural Research

Sally Rockey, Ph.D.

Larry Tabak, D.D.S, D.Phil.

NIH National Institutes of Health

Document 13

Thomas Insel to Depart as NIMH Director

Thomas Insel, M.D.

Bruce Cuthbert, Ph.D.

NIH National Institute of Mental Health

Breaking News

FDA Commissioner Nomination

Robert Califf, M.D.

National Library of Medicine Working Group Report

Institutes of Health
Advisory Committee to the Director

National Library of Medicine (NLM) Working Group

FINAL REPORT – JUNE 11, 2015

MEMBERS: Eric Green (co-chair), Harlan Krumholz (co-chair), Russ Altman, Howard Bauchner, Deborah Brooks, Doug Fridsma, Steven Goodman, Eric Horvitz, Trudy MacKay, Alexa McCray, Chris Shaffer, David Van Essen, Joanne Waldstreicher, James Williams, II, Kathy Hudson (ex officio), Lyric Jorgenson (executive secretary) *(titles and affiliations listed in Appendix A)*

The NIH Director charged the
referred to as the Working Gr
NLM remains an internationa
five months of deliberations,
pertaining to NLM's mission a

Vacancy Announcement

Department of Health and Human Services (DHHS)
National Institutes of Health (NIH)

DIRECTOR

National Library of Medicine

With nation-wide responsibility for improving the health and well-being of all Americans, the Department of Health and Human Services (DHHS) oversees the biomedical research programs of the National Institutes of Health (NIH) and those of NIH's research Institutes and Centers (ICs). As the world's largest medical research facility, NIH consists of 27 ICs including the Clinical Center (an on-site research hospital).

Document 14

NIH Strategic Plan

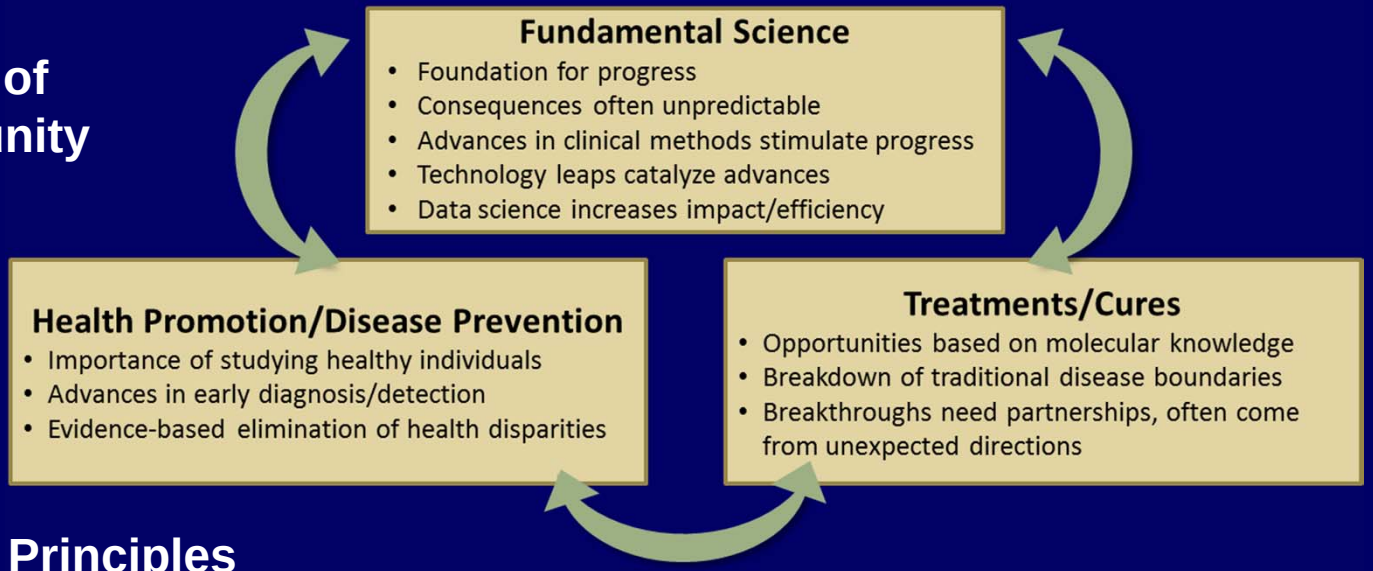
Document 15

NIH Strategic Plan Framework

Overview

- Mission of NIH
- Unique moment of opportunity in biomedical research
- Current NIH-supported research landscape
- Constraints confronting the community in the face of lost purchasing power

Areas of Opportunity



Unifying Principles

Setting Priorities

- Incorporate disease burden as important, but not sole factor
- Foster scientific opportunity; need for nimbleness
- Advance research opportunities presented by rare diseases
- Consider value of permanently eradicating a pandemic

Enhancing Stewardship

- Recruit/retain outstanding research workforce
- Enhance workforce diversity
- Encourage innovation
- Optimize approaches to inform funding decisions
- Enhance impact through partnerships
- Ensure rigor and reproducibility
- Reduce administrative burden
- Employ risk management strategies

Document 15

NIH Rigor and Reproducibility in Grant Applications and Review

Enhancing Reproducibility through Rigor and Transparency

Notice Number: NOT-OD-15-103

Key Dates

Release Date: June 9, 2015

Related Announcements

NOT-NS-11-023
NOT-MH-14-004
NOT-DA-14-007
NOT-OD-15-102

Issued by

National Institutes of Health

Purpose

The National Institutes of He
research findings through in
effect for applications submit

Consideration of Sex as a Biological Variable in NIH-funded Research

Notice Number: NOT-OD-15-102

Key Dates

Release Date: June 9, 2015

Related Announcements

NOT-OD-15-103

Issued by

National Institutes of Health (NIH)

Purpose

The National Institutes of Health (NIH) is committed to improving the health outcomes of men and women through support of rigorous science that advances fundamental knowledge about the nature and behavior of living systems. Sex and gender play a role in how health and disease processes differ across individuals1, and consideration of these factors in research studies informs the development and testing of preventive and therapeutic interventions in both sexes. This notice focuses on NIH's expectation that scientists will account for the possible role of sex as a biological variable in vertebrate animal and human studies. Clarification of these expectations is reflected in plans by NIH's Office of Extramural Research (OER) to update application instructions and review questions; once approved by the Office of Management and Budget (OMB), these updates will take effect for applications submitted for the January 25, 2016, due date and thereafter. Please refer to NOT-OD-15-103 for further consideration of NIH expectations about enhancing reproducibility through rigor and transparency.

Document 16

New Legislation: 21st Century Cures (H.R. 6)

- Passed by the House on July 10
- Boosts NIH research and reforms FDA's regulatory framework to accelerate the discovery, development, and delivery of cures
- Senate continues to work on its "Innovations Bill"

Document 17

NIH Appropriations and Budget

Fiscal Year 2016 Appropriations

Entity	Chamber	Amount	% Change	Difference
NIH	House	$31.3B	3.3% Increase	
	Senate	$32.1B	6.0% Increase	+$900M
NHGRI	House	$515.5M	3.4% Increase	
	Senate	$526.1M	4.4% Increase	+ $10.6M

Document 18

Congressional NIH Caucus

- Announced in May 2015
- Goal: Increase the purchasing power of NIH
- Senate: bipartisan group of 23 senators; co-chaired by Durbin (D-IL) and Graham (R-SC)
- House: bipartisan group of 17 representatives; co-chaired by Higgins (D-NY), King (R-NY), and Delauro (D-CT)

Document 19

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Mourning the Loss of Bill Gelbart

Document 20

Mourning the Loss of Eric Davidson

Document 21

Associate Vice Chancellor for Computational Health Sciences, University of California San Diego

UC San Diego
SCHOOL OF MEDICINE

Jill Mesirov, Ph.D.

Document 22

Inaugural Chair, Stanford Department of Biomedical Data Science

STANFORD
UNIVERSITY

Carlos Bustamante, Ph.D.

New Investigators, Howard Hughes Medical Institute

Job Dekker, Ph.D.

Levi Garraway, M.D., Ph.D.

Pardis Sabeti, D.Phil., M.D.

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

hhmi
Howard Hughes Medical Institute

Document 24

New ASHG Position Statements

ASHG POSITION STATEMENT

Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents

Jeffrey R. Botkin,[1,14,*] John W. Belmont,[2,14] Jonathan S. Berg,[3,14] Benjamin E. Berkman,[4,14] Yvonne Bombard,[5,6,14] Ingrid A. Holm,[7,14] Howard P. Levy,[8,14] Kelly E. Ormond,[9,14] Howard M. Saal,[10,14] Nancy B. Spinner,[11,14] Benjamin S. Wilfond,[12,14] and Joseph D. McInerney[13,14]

THE AMERICAN SOCIETY OF HUMAN GENETICS

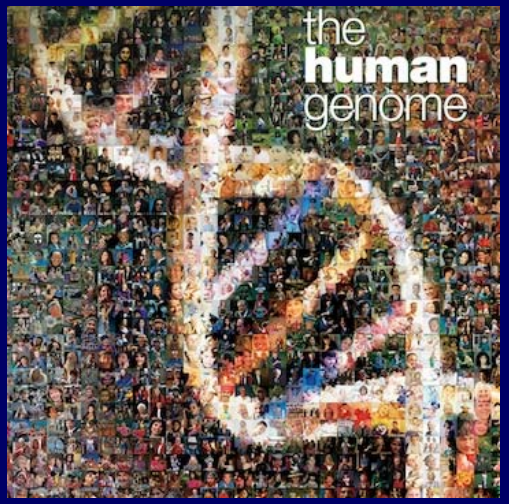
ASHG Statement of Support for Licensure of Genetic Counselors

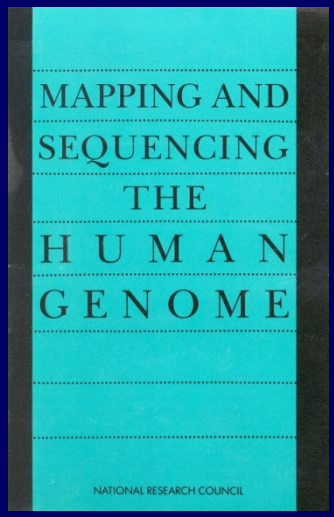
September 10, 2015

The American Society of Human Genetics (ASHG) supports state licensure of certified genetic counselors to help ensure that the public has access to genetic and genomic services provided by qualified health professionals.

Document 25

25th Anniversary of the Launch of the Human Genome Project

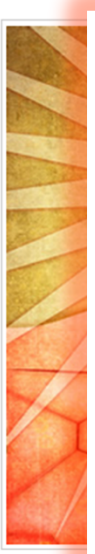




NHGRI Genome Advance of the Month

Scientists discover topical insights into the effects of sun exposure on skin

By Julie Coursen
Scientific Program Analyst, NHGRI



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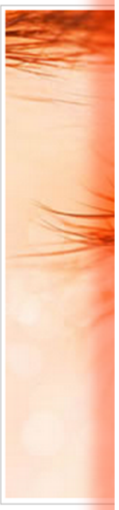
seemed t

Genomics holds promise of treatments for inherited blindness

By Bianca Patel
Scientific Program Analyst, NHGRI



Before insert
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Gene-disease association data could improve drug development

By Brenda Iglesias
Scientific Program Analyst, NHGRI

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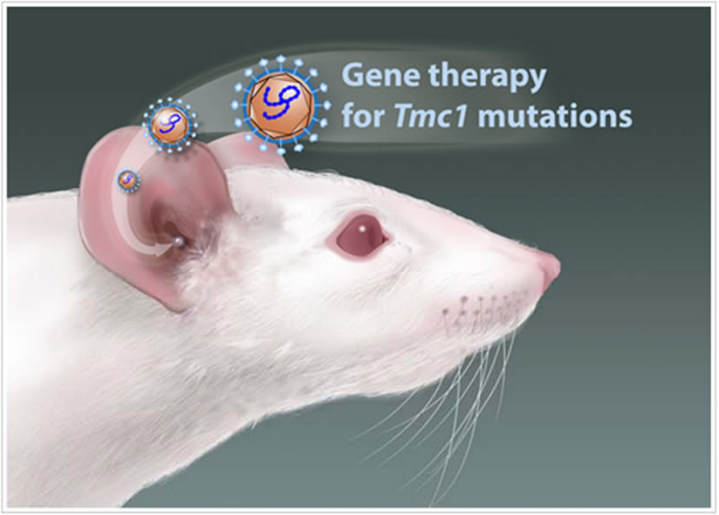
Gene Therapy and the Biological Treatment of Hearing Loss

By Jonathan Lotempio, Jr.
Scientific Program Analyst, NHGRI



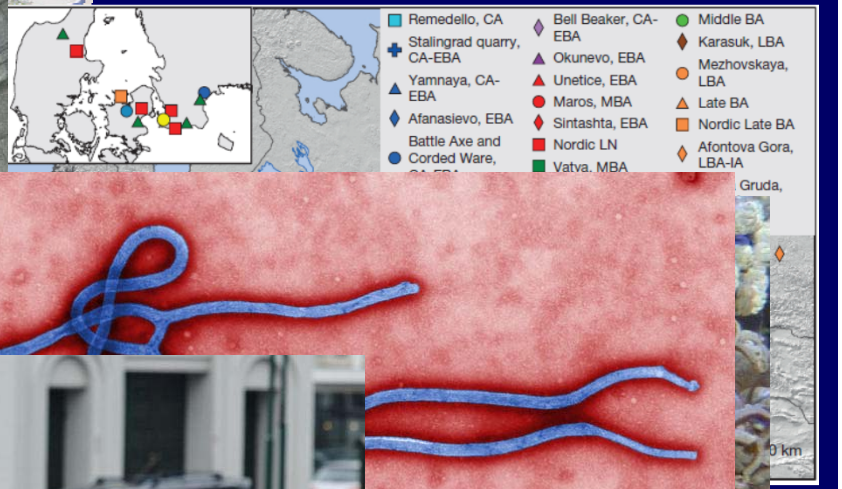
Ludwig von Beethoven suffered from deafness in his mid-twenties. To combat that hearing loss, the composer turned to the advanced technology of 1813 and commissioned his mechanically minded contemporaries to make ear trumpets to funnel and magnify sound.

While ear trumpets were ultimately ineffective, 200 years of progress have provided a much more promising biological alternative. July's Genome Advance of the Month highlights a study in the journal *Science Translational Medicine* that demonstrated gene therapy as an effective way to improve hearing in patients with two genes linked to genetic prelingual deafness, or hearing loss that occurs before a child learns to speak. The study focused on deafness caused by defects in transmembrane channel-like 1 (*TMC1*), which is a gene that codes for a protein that helps convert sounds into electrical signals for the brain to interpret. Defects in the *TMC1* gene are a common cause of genetic deafness, accounting for 4 to 8 percent of cases.

The study's authors, Charles Askew, Ph.D., Jeffery R. Holt, Ph.D. and colleagues at École Polytechnique Fédérale de Lausanne in Switzerland, examined the use of gene therapy to cure hereditary deafness in mice with two types of *Tmc1* mutations. The first type had the *Tmc1* gene deleted, which is a good model for children who have two *TMC1* mutations and experience hearing loss at a very young age. The second type was a specific malfunctioning variant of *TMC1* fittingly known as Beethoven in honor of the 19th century composer. Mice with the Beethoven point mutation closely model the loss of hearing in humans who have an autosomal dominant *TMC1* mutation that leads to progressive hearing loss in the teen years.

Document 26

Genomes In The News…



Document 27

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Extramural Research Program Retreat

Genome Sequencing Program

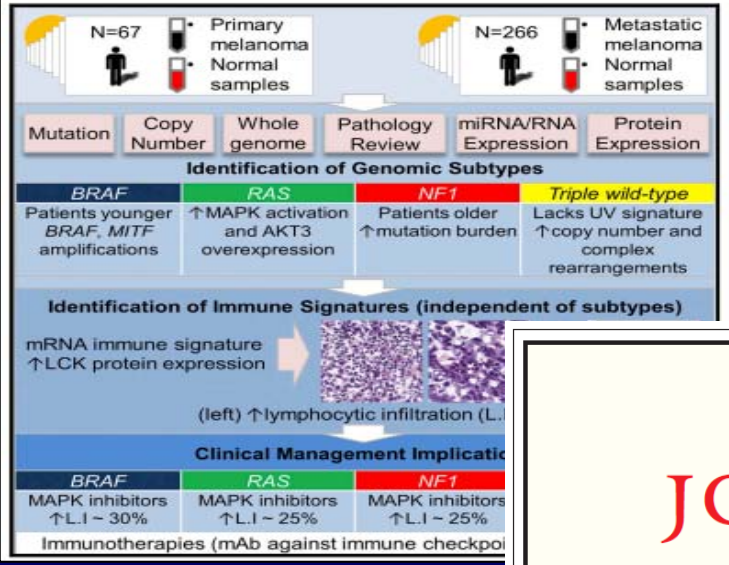
- **Funding Plans Under Consideration**
  - Centers for Common Disease Genomics (UM1)
  - Centers for Mendelian Genomics (UM1)
  - Coordinating Center (U24)
- **Letters of Intent Received**
  - Genome Sequencing Program Analysis Centers (U01)
  - Human & Non-Human Primate Genome Sequences (U24)

Resource

Cell

Genomic Classification of Cutaneous Melanoma

Graphical Abstract



Authors

The Cancer Genome Atlas Network

Correspondence

irwatson@mdanderson.org (I.R.W.), jgershen@mdanderson.org (J.E.G.), lchin@mdanderson.org (L.C.)

In Brief

An integrative analysis of cutaneous melanomas establishes a framework for genomic classification into four subtypes that can guide clinical decision-making

The NEW ENGLAND JOURNAL of MEDICINE

ESTABLISHED IN 1812 JUNE 25, 2015 VOL. 372 NO. 26

Comprehensive, Integrative Genomic Analysis of Diffuse Lower-Grade Gliomas

The Cancer Genome Atlas Research Network*

Document 29

Centers for Mendelian Genomics

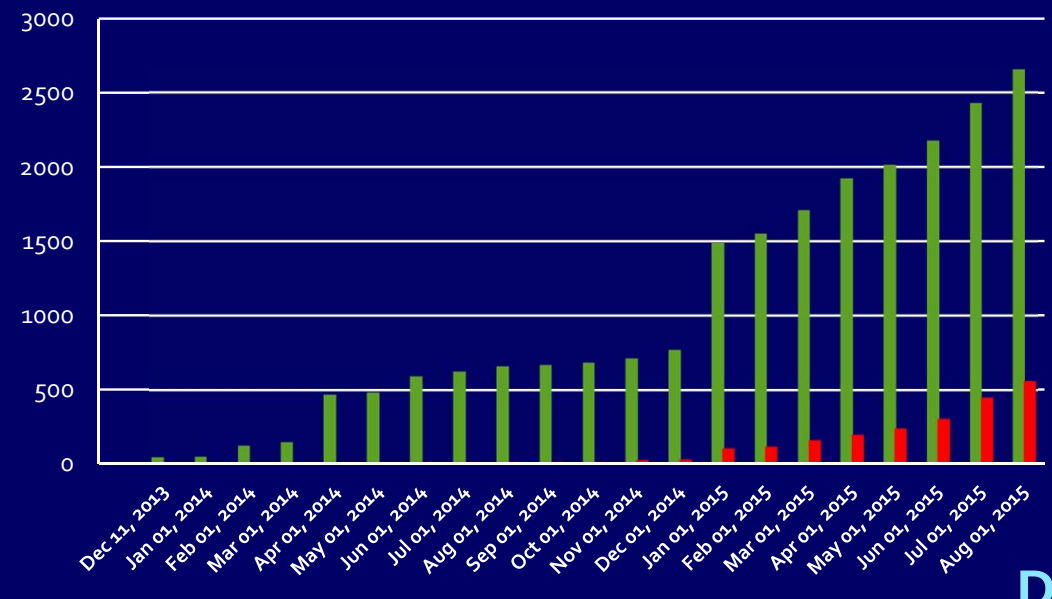
Finding the genes underlying human Mendelian conditions

Discovery

- **>1,500 'causal genes' for Mendelian phenotypes;**
  >590 novel causal genes
- **>195 publications**

GeneMatcher

- **>600 users**
- **43 countries**



2,658 Genes

560 Matches

Centers for Mendelian Genomics

Finding the genes underlying human Mendelian conditions

AJHG

The American Journal of Human Genetics 97, 199–215, August 6, 2015 199

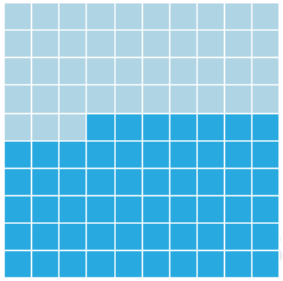
REVIEW

The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities

- **Progress on causal gene discoveries, method improvement, and resource sharing**
- **Remaining discoveries and challenges**

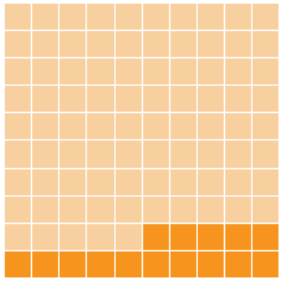
‘Scorecard’

Figure 1.



Mendelian phenotypes associated with ‘causal genes’ - 57% (4,163/7,315)

Figure 2.



Human genes known to underlie Mendelian phenotypes - 15% (2,937/19,580)

Document 30

Clinical Sequencing Exploratory Research Program

- **Enrolled 2,889 adults, 759 children**
- **161 publications, 12 working group publications**

CSER and eMERGE: current and potential state of the display of genetic information in the electronic health record

RECEIVED 12 March 2015
REVISED 30 April 2015
ACCEPTED 12 May 2015

***JAMIA* 2015**

Professionally Responsible Disclosure of Genomic Sequencing Results in Pediatric Practice

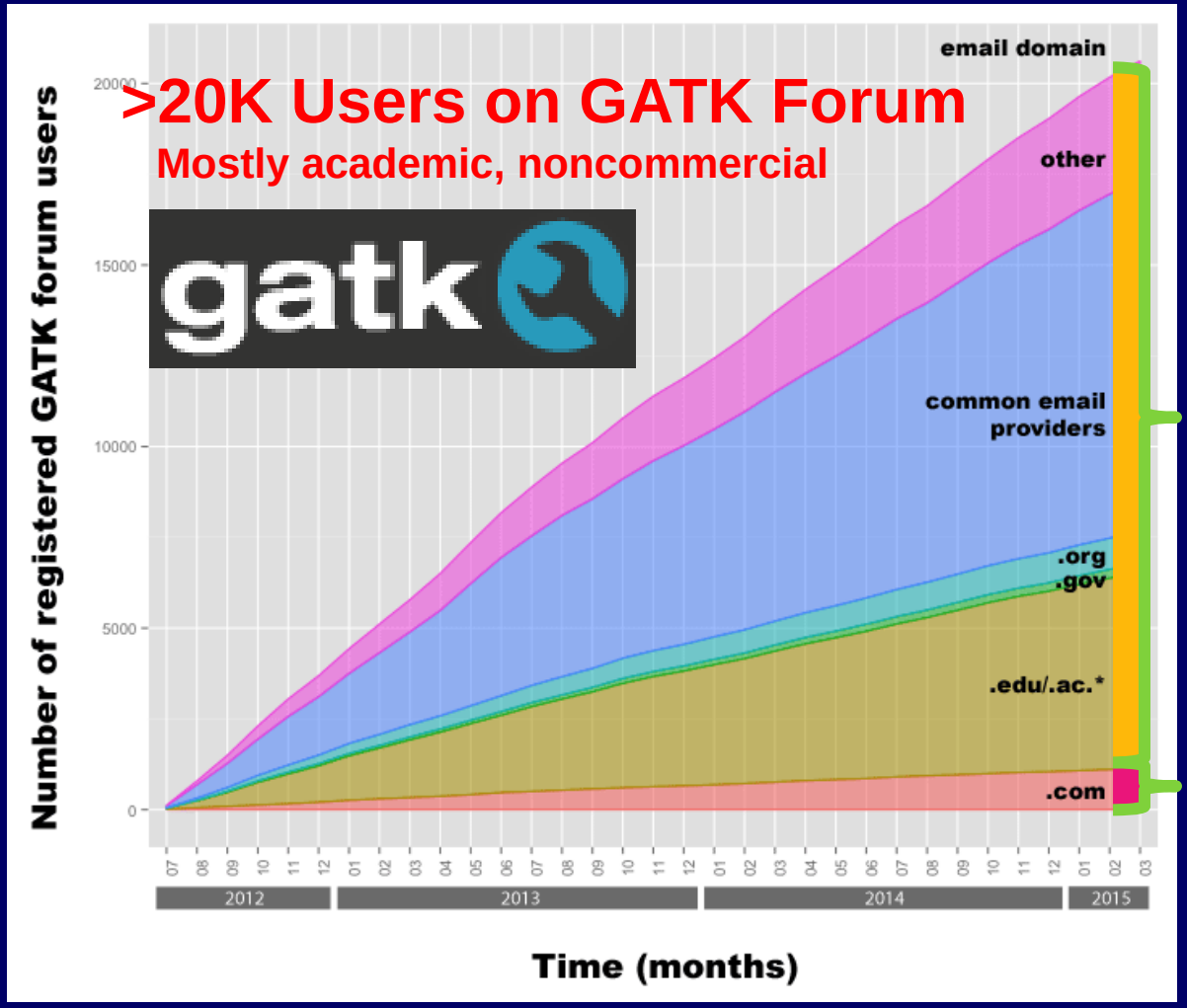
Laurence B. McCullough, PhD[a], Kyle B. Brothers[b], Wendy K. Chung[c], Steven Joffe[d], Barbara A. Koenig[e], Benjamin Wilfond[f], Joon-Ho Yu[f], on behalf of the Clinical Sequencing Exploratory Research (CSER) Consortium Pediatrics Working Group

***Pediatrics*, in press**

Document 31

Genome Sequencing Informatics Tools

iSeqTools



2015 GATK Forum (per month):

- ~650 users
- ~150 discussions
- ~100,000 views

Document 32

Technology Development Program

- **RFAs for Novel Nucleic Acid Sequencing released:**
  - **(R01): RFA-HG-15-032**
  - **(R21): RFA-HG-15-031**
  - **(R43/R44): RFA-HG-15-033**
- **FOAs for remaining concepts to be released soon:**
  - **Genome Technology Development**
    - **(R01, R21, R43/R44)**

Encyclopedia of DNA Elements (ENCODE)

- 2015 ENCODE Community Users Meeting

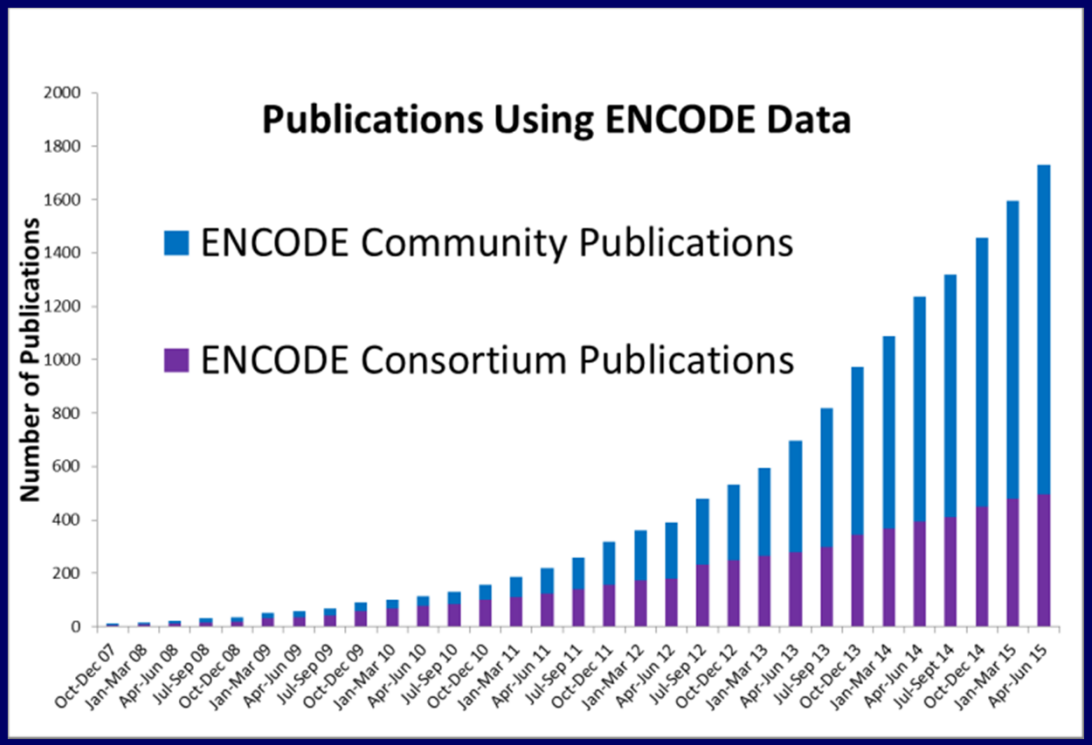
- ENCODE portal infrastructure used by ClinGen

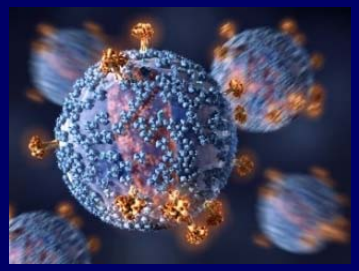
Document 34

Encyclopedia of DNA Elements (ENCODE)

- **Postponement of Functional Genomics Initiatives to September 2016 Council review**

- **ENCODE Publications:**

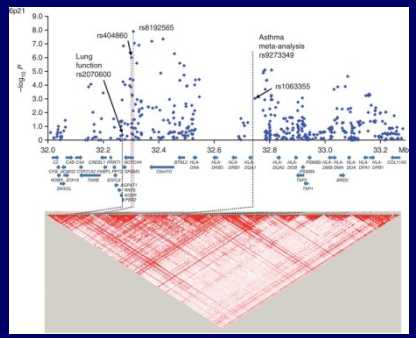


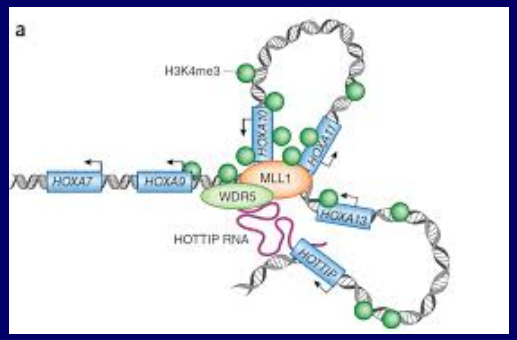


Document 34

New Functional Genomic Variation Grants

- Develop computational approaches to infer causal variants for phenotypes, and assess predictions with experimental data
- 6 grant awards made thus far
- Information on transcripts, TF binding, nucleosomes, enhancers, DNA shape, conservation, & phenotypes
- Studies of autism, cancer, bipolar disorder, type 2 diabetes, & age-related macular degeneration





Document 35

Centers of Excellence in Genomic Science (CEGS) Program

Two new CEGS awards:

- **Harvard Medical School-**
  Center for Genomically Engineered Organs
- **University of Washington-**
  Center for Photogenomics

eMERGE network

ELECTRONIC MEDICAL RECORDS AND GENOMICS

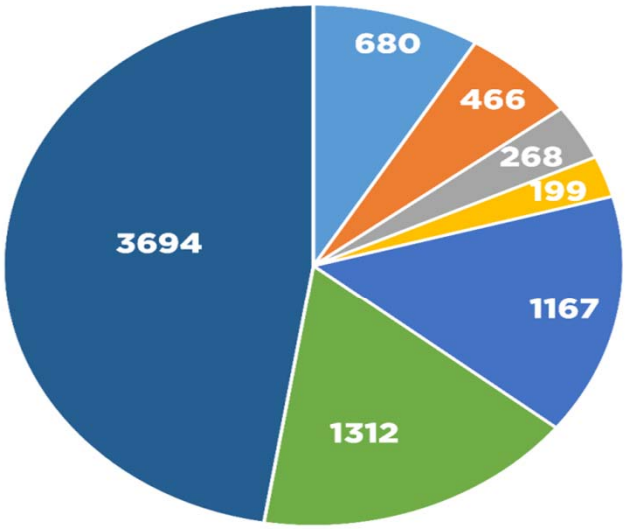
- Site Projects (333)
- Network Projects (198)

= **531 total projects**

Cumulative Citation Count

- 2007-July 2015: **9,298**
- Phase II Publications Only: **8,306**

Citations of eMERGE Publications by Category through April 2015:



- All Network / Foundation: 680(160)
- Privacy: 466(47)
- EHRI: 268(268)
- PGx/Implementation: 199(199)
- CERC/RoR: 1167(260)
- Phenotyping: 1312(436)
- Genomics: 3694(1334)

Tools/Software

PheKB.org **Myresults.org**

SPHINX

InfoButton Project

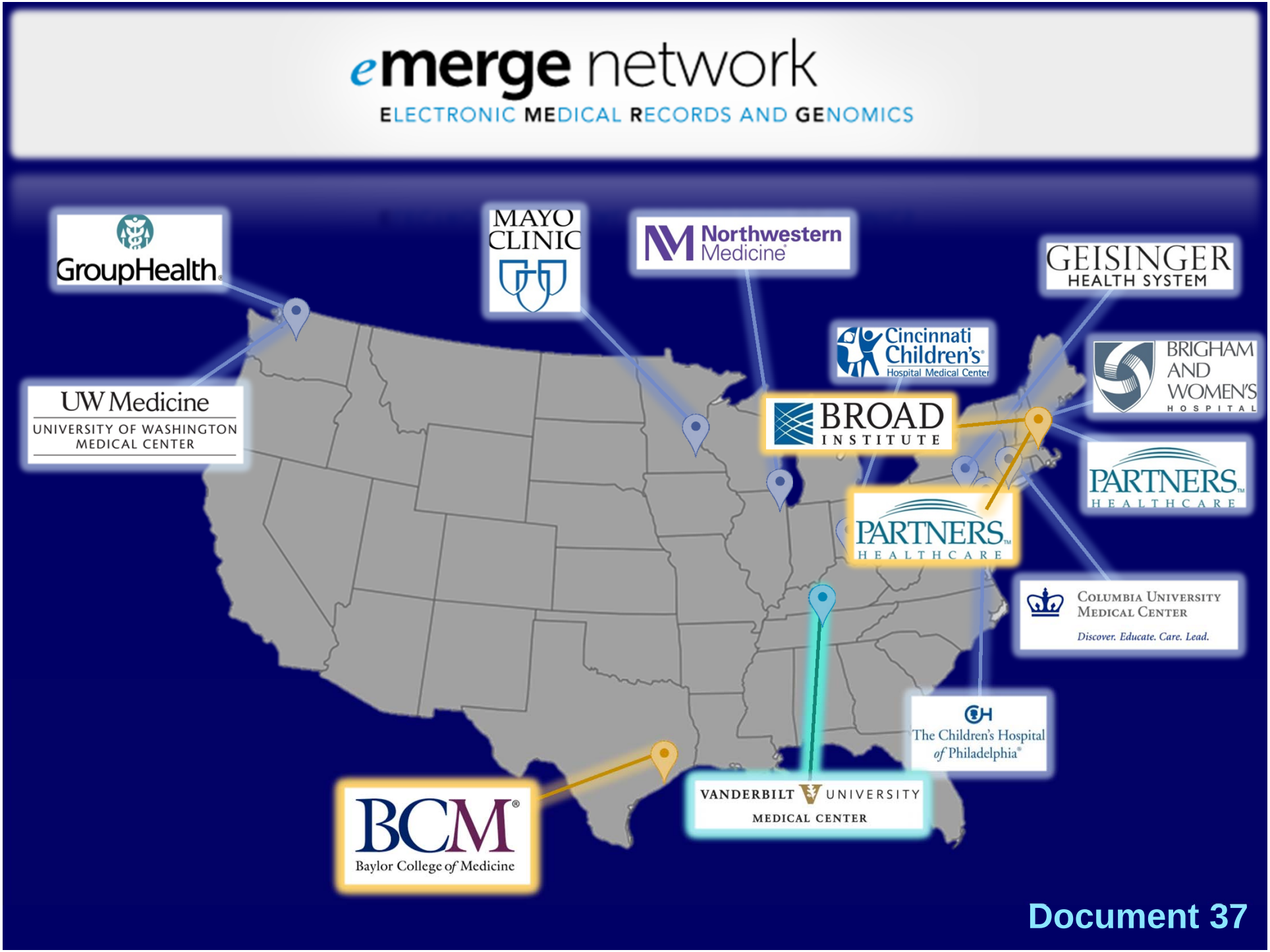
eCDS KB

Phenotyping Tools

Genotyping Tools

Model Consent Language

Document 37


emerge network
ELECTRONIC MEDICAL RECORDS AND GENOMICS
GroupHealth
MAYO CLINIC
Northwestern Medicine
GEISINGER HEALTH SYSTEM
Cincinnati Children's Hospital Medical Center
BRIGHAM AND WOMEN'S HOSPITAL
UW Medicine
UNIVERSITY OF WASHINGTON MEDICAL CENTER
BROAD INSTITUTE
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COLUMBIA UNIVERSITY MEDICAL CENTER
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The Children's Hospital of Philadelphia
VANDERBILT UNIVERSITY MEDICAL CENTER
BCM
Baylor College of Medicine

Document 37

Phenotypes & eXposures (PhenX) Toolkit

80 New Measures

PhenX Toolkit

Protocols in REDCap

REDCap
Research Electronic Data Capture

NHLBI Sickle Cell Disease Meeting

NIH National Heart, Lung, and Blood Institute

August 18-20, 2015
NIH Main Campus

ASHG Invited Session

Pin it

ASHG2015
October 6-10 • Baltimore, MD

Document 38

ClinGen: Sharing Data. Building Knowledge. Improving Care.

ClinGen *NEJM* Marker Paper

Clinical Actionability: ACMG 56 gene list

High visibility at ASHG and NSGC meetings

Genomics and Society Working Group

- In-person meeting held in April 2015
- Update from Dr. Lisa Parker in Open Session

Document 40

Small Business Grants

Small Business	Phase I Proof of principle	Phase II Pre-commercialization
Innovation Research (SBIR)	13	11
Technology Transfer (STTR)	5	1
Total	18	12

Small business funding to increase ~$0.6M/year

Document 41

Genomics Training and Career Development

RFA-HG-15-025: Data Analysis and Coordinating Center (DACC) for Research Training and Career Development Activities

- **Maintain, improve, and/or develop database**
- **Collect and analyze data about trainee career paths**
- **Logistical support for an expanded annual training program meeting**
- **Application due date: October 20, 2015**

Document 42

Director's Report Outline

I. General NHGRI Updates

II. General NIH Updates

III. General Genomics Updates

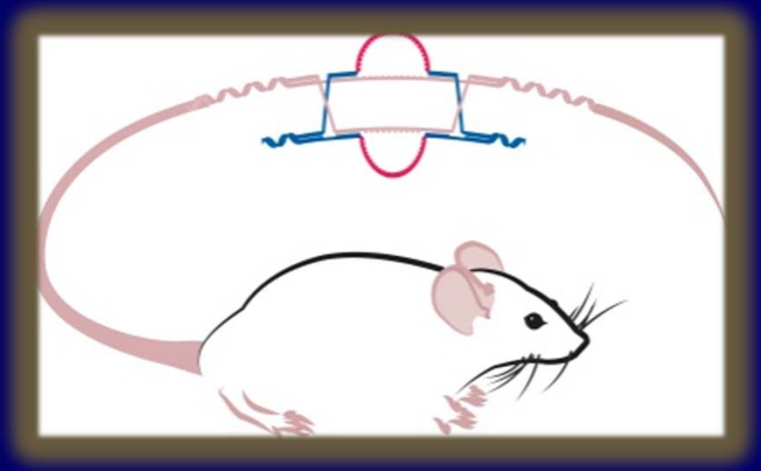
IV. NHGRI Extramural Research Program

V. NIH Common Fund/Trans-NIH

VI. NHGRI Division of Policy, Communications, and Education

VII. NHGRI Intramural Research Program

Knockout Mouse Phenotyping Project (KOMP2)



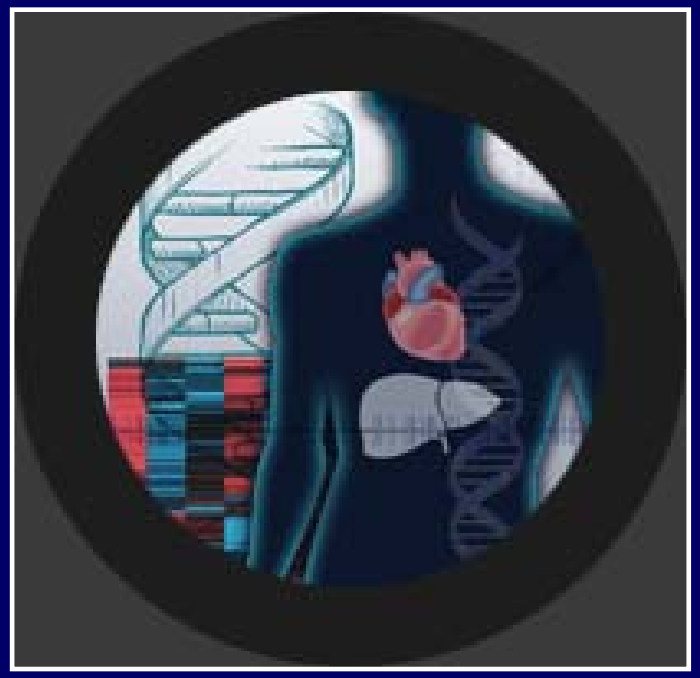
IMPC
International Mouse Phenotyping Consortium

- Common Fund approves 5 year continuation
- 18 Institutes/Centers/Offices contributing funds
- Total funds: ~$100M
- FOAs in process

Recent Progress:

- Public release of data and harmonized metadata
- Multiple internal collaborative projects
- Outreach efforts: 8 collaborations funded jointly by LINCS and other entities (NIAAA, NCCIH, NIEHS, NIMH, NINDS, & BD2K)

Genotype-Tissue Expression (GTEx) Project



- Donor recruitment goal (n=900) completed

- New GTEx portal and version 5 data release

Protein Capture Reagents Program

- ~1100 validated antibodies to 370 human transcription factors; available via data portal
- Program near end of pilot phase, with one grantee finishing production of mouse monoclonals
- JHU subcontractor attended White House Demo Day

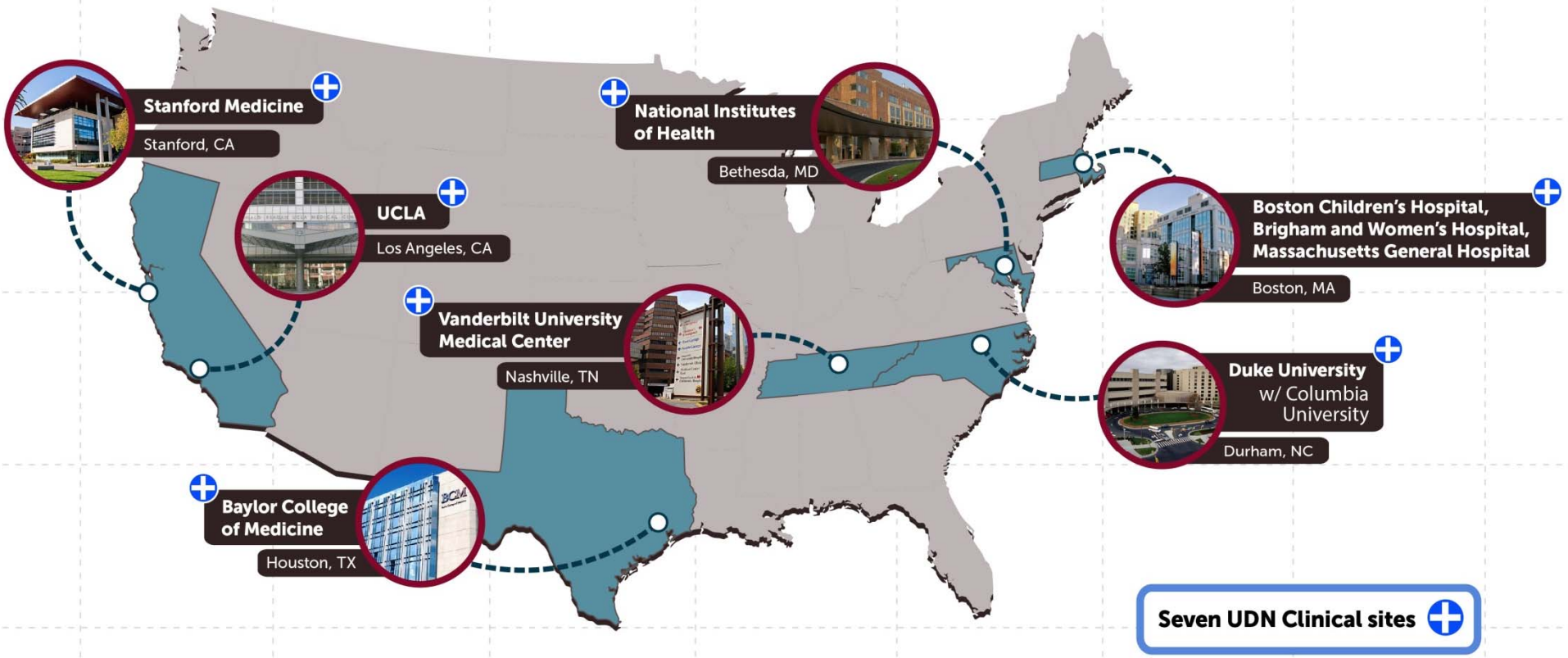
Human Heredity and Health in Africa (H3Africa)

- 7th Consortium Meeting in October (Washington DC)
  - ASHG Session
  - Presentations at NIH
  - NIH-H3Africa Subject Area Meetings
- 2 UH3 Biorepository awards (Nigeria and Uganda)
- Approval for a Phase II of H3Africa

Document 47

Undiagnosed Diseases Network (UDN)

Undiagnosed Diseases Network | Seven UDN Clinical Site Locations



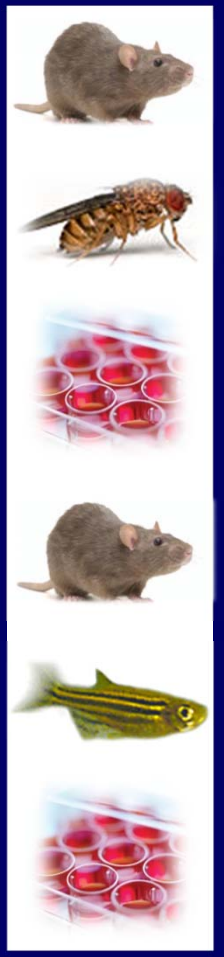
Click 'Apply' button on any UDN webpage to apply!

APPLY

Document 48

Undiagnosed Diseases Network (UDN)

PI Name	Gene	Model	Patient Phenotype
Hall	*BAI2*	astrocytes from mouse	shrinking spinal cord area and limb muscle stiffness
Ye	*YPEL3*	drosophila	reduced myelin, enlarged nerves, abnormal nerve conduction, normal cognition
Smith	*BHLHB9*	knock down mouse neural stem cells and patient iPS neural cells	motor dysfunction, cognitive impairment, low brain volume, peripheral nerve damage
Yu	*CHML*	mouse	developmental regression, nerve damage with brain volume loss
Westerfield	*YPEL3*	zebrafish	reduced myelin, enlarged nerves, eye abnormalities
Ho	*FOXR1*	human fibroblasts, mouse	microcephaly, developmental delay, progressive brain atrophy



Undiagnosed Diseases Network

Gabriella Miller Kids First Pediatric Research Program

- NIH Common Fund Program ($12.6M/year for ten years)
- Clinical and genome sequence data
  - Childhood cancers
  - Structural birth defects
- Fiscal Year 2015
  - PAR 15-259 (X01) seeking samples
  - Up to 6000 samples
- Fiscal Year 2016
  - Creation of data resource
  - Solicitation of additional samples
  - Generation of genome sequence data

Document 49

Big Data to Knowledge (BD2K) Initiative

- Training the next generation of biomedical researchers in data science
- Funding for software projects in targeted research areas
- Funding supplement programs for data sharing, discovery, and interoperability

Precision Medicine Initiative

Health Information | Grants & Funding | News & Events | Research & Training | Institutes at NIH | About NIH

NIH Home > Research & Training

PRECISION MEDICINE INITIATIVE

Precision Medicine Initiative

- Near-term Goals
- Longer-term Goals
- Scale and Scope
- Participation
- PMI Working Group
- Events
- Announcements
- PMI in the News
- Multimedia

Faces of the Precision Medicine Initiative — Dr. Russ Altman

NIH Director's blog: Read precision medicine-related blogs by the NIH Director.

ABOUT THE 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 unveiled the Precision Medicine Initiative (PMI) — 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.

Email Updates

To sign up for updates please enter your e-mail address.

Submit

Related Links

- NEJM Perspective: A New Initiative on Precision Medicine
- White House Precision Medicine Web Page
- White House Fact Sheet: President Obama's Precision Medicine Initiative
- Precision Medicine Initiative and Cancer Research
- Storify: #PMINetwork Twitter Chat
- Storify: The Precision Medicine Initiative Announcement
- Precision Medicine Initiative YouTube Channel

Document 51

Precision Medicine Initiative Workshops

- **Digital Health Data in a Million-Person Precision Medicine Initiative Cohort**
  - **May 28-29, 2015**
  - **Vanderbilt University, Nashville, TN**
- **Participant Engagement and Health Equity**
  - **July 1-2, 2015**
  - **NIH, Bethesda, MD**
- **Mobile and Personal Technologies in Precision Medicine**
  - **July 27-28, 2015**
  - **Intel Corp., Santa Clara, CA**

PRECISION MEDICINE INITIATIVE

Precision Medicine Initiative

- Near-term Goals
- Longer-term Goals
- Scale and Scope
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- PMI Working Group
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- PMI in the News
- Multimedia

The Precision Medicine Initiative: Infographic
View larger (PDF - 163KB)

Events

The following are upcoming and past events held by the NIH to gather input from participant, scientific, and other stakeholder groups as it plans the development of the President's Precision Medicine Initiative and the vision for building the national participant group. Please sign up for updates or check back frequently for additional meeting information.

All public workshops will be videocast live.

UPCOMING EVENTS

- No currently scheduled events

PAST EVENTS

2015

- Mobile and Personal Technologies in Precision Medicine Workshop
  Monday, July 27 - Tuesday, July 28, 2015
  Intel Corp., Santa Clara, Calif.
- Participant Engagement and Health Equity Workshop
  Wednesday, July 1 - Thursday, July 2, 2015
  National Institutes of Health, Bethesda, Md.
- Digital Health Data in a Million-Person Precision Medicine Initiative Cohort
  Thursday, May 28 - Friday, May 29, 2015
  Vanderbilt University, Nashville, Tenn.
- Senate Hearing: Continuing America's Leadership: Realizing the Promise of Precision Medicine for Patients
  Tuesday, May 5, 2015
  NIH Director's Testimony (PDF - 89.9KB)
- Public workshop on unique scientific opportunities for the national research cohort
  Tuesday, April 28 - Wednesday, April 29, 2015
  National Institutes of Health, Bethesda, Md.
- Public Workshop on Building a Precision Medicine Research Cohort
  Wednesday, February 11 - Thursday, February 12, 2015
  National Institutes of Health, Porter Neuroscience Building, Bethesda, Md.

Email Updates

To sign up for updates please enter your e-mail address.

Submit

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- Precision Medicine Initiative YouTube Channel

Document 51

Report on Precision Medicine Initiative Cohort Program

The Precision Medicine Initiative Cohort Program – Building a Research Foundation for 21st Century Medicine

Precision Medicine Initiative (PMI) Working Group Report to the Advisory Committee to the Director, NIH

September 17, 2015

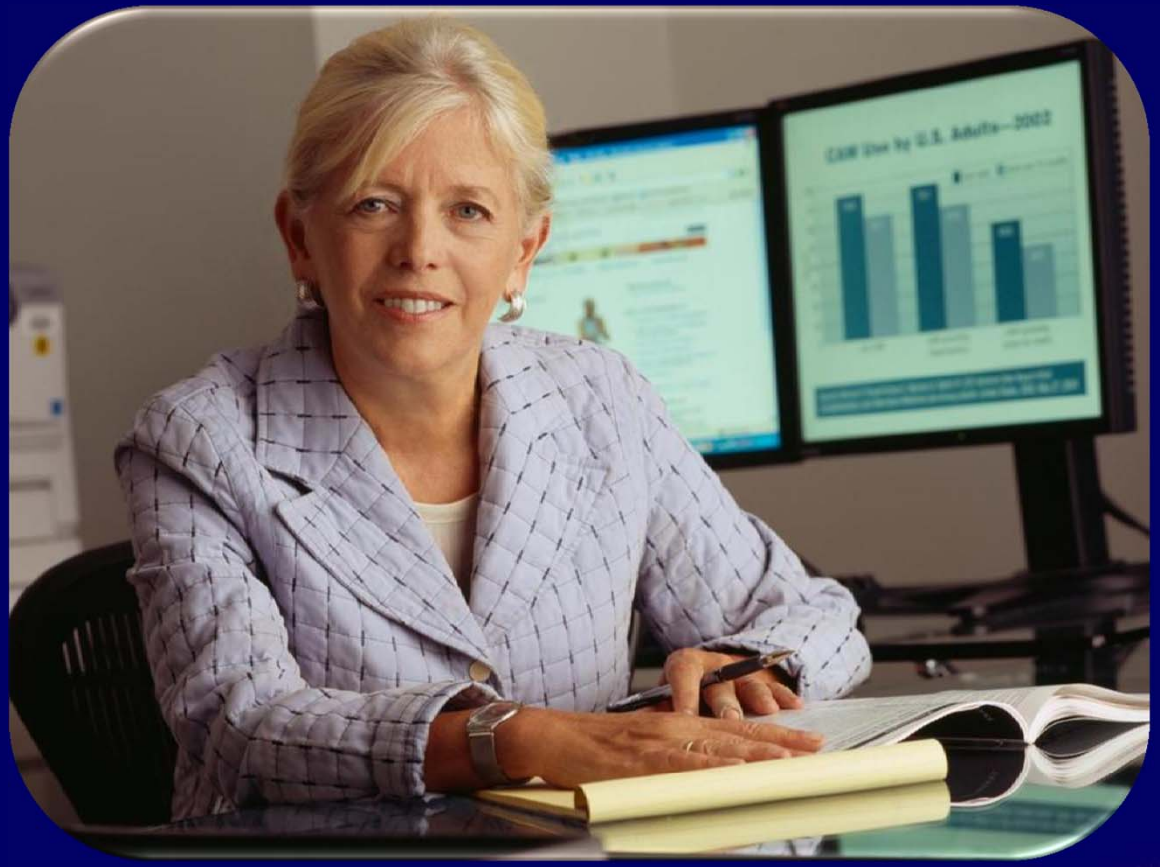
For Immediate Release: Thursday, September 17, 2015

NIH framework points the way forward for building national, large-scale research cohort, a key component of the President's Precision Medicine Initiative

The National Institutes of Health Advisory Committee to the Director (ACD) today presented to NIH Director Francis S. Collins, M.D., Ph.D., a detailed design framework for building a national research participant group, called a cohort, of 1 million or more Americans to expand our knowledge and practice of precision medicine. Dr. Collins embraced the design recommendations made by the ACD, noting the need to remain nimble and adaptable as the Initiative progresses. He also thanked the Committee for their recommendations on policy issues and welcomed the opportunity to review them. NIH plans to move quickly to build the infrastructure so that participants can begin enrolling in the cohort in 2016, with a goal of enrolling at least 1 million participants in three to four years.

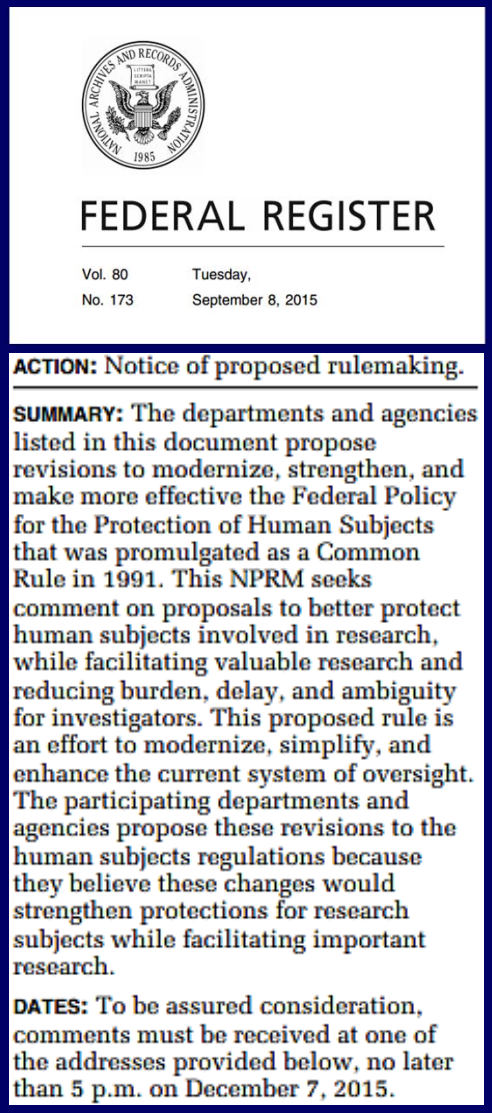
Document 51

Interim Director, Precision Medicine Initiative Cohort Program



Josie Briggs, M.D.

Notice of Proposed Rulemaking for Revisions to the 'Common Rule'



FEDERAL REGISTER

Vol. 80 No. 173 Tuesday, September 8, 2015

ACTION: Notice of proposed rulemaking.

SUMMARY: The departments and agencies listed in this document propose revisions to modernize, strengthen, and make more effective the Federal Policy for the Protection of Human Subjects that was promulgated as a Common Rule in 1991. This NPRM seeks comment on proposals to better protect human subjects involved in research, while facilitating valuable research and reducing burden, delay, and ambiguity for investigators. This proposed rule is an effort to modernize, simplify, and enhance the current system of oversight. The participating departments and agencies propose these revisions to the human subjects regulations because they believe these changes would strengthen protections for research subjects while facilitating important research.

DATES: To be assured consideration, comments must be received at one of the addresses provided below, no later than 5 p.m. on December 7, 2015.

- Improve informed consent
- Calibrate review to study risks
- Require informed consent for use of biospecimens
- New data security and information protection standards
- Streamline IRB review
- Extend protection to all research at federally-funded sites

Document 52

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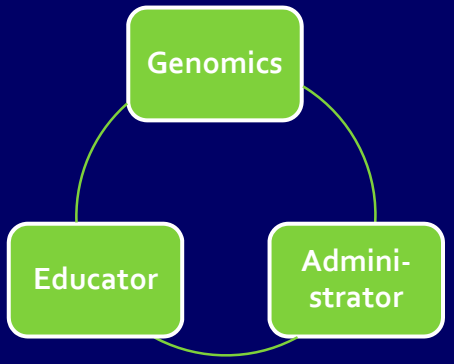
VII. NHGRI Intramural Research Program

Method for Introducing a New Competency

Toolkit to Facilitate Integration of Genomics

MINC
A METHOD FOR INTRODUCING A NEW COMPETENCY
GENOMICS



- **Promote genomic practice**
- **Capture expertise and processes**
- **Collect resources**

Document 53

Genetic and Rare Diseases (GARD) Information Center Videos

Document 54

NHGRI Social Media

livestream

Genome: Unlocking Life's Code

Exhibition Travel Schedule

2015

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

2016

January 28-April 25
Discovery World
Milwaukee, WI

May 21-September 5
Natural History Museum of Utah
Salt Lake City, UT

September 30-January 1
Exploration Place
Wichita, KS

GENOME UNLOCKING LIFE'S CODE

Genome: Unlocking Life's Code

eNewsletter

Document 56

Precision Medicine Event in St. Louis






Document 56

2015 NHGRI Summer Workshop in Genomics: The 'Short Course'

High School Teachers

Nursing Faculty

Document 57

Partnership for Community Outreach and Engagement in Genomics

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Mourning the Loss of Rep. Louis Stokes

Document 58

American College of Rheumatology Distinguished Fellow Award

Pravitt Gourh, M.D.

Document 59

NHGRI Intramural Research Highlights

CSH PRESS GENOME RESEARCH

The use of CRISPR/Cas9 as a genome-editing tool in various model organisms has radically changed targeted mutagenesis

AJHG The American Journal of Human Genetics

Individualized Iterative Phenotyping for Genome-wide Analysis of Loss-of-Function Mutations

Genetics inMedicine

Official Journal of the American College of Medical Genetics and Genomics

A critical reappraisal of dietary practices in methylmalonic acidemia raises concerns about the safety of medical foods. Part 1: isolated methylmalonic acidemias

Document 60

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!