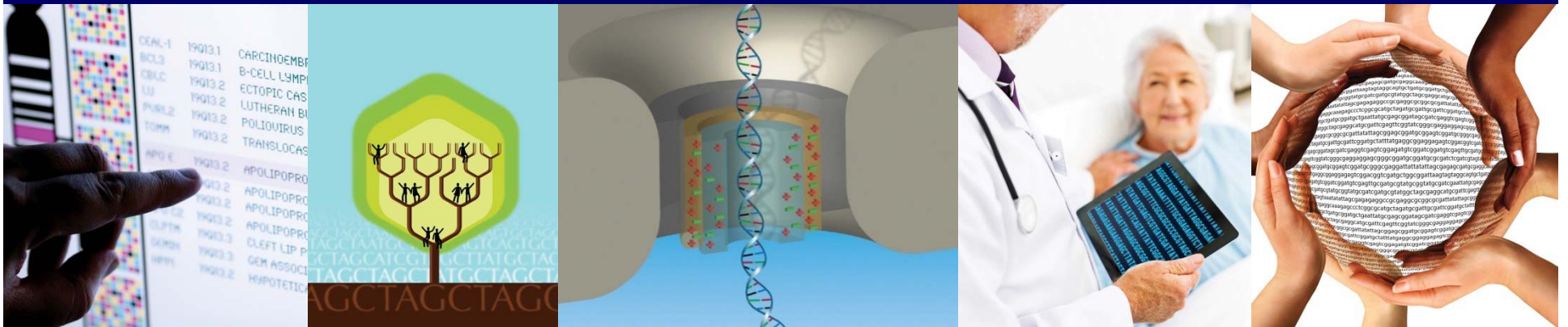


# DIRECTOR'S REPORT

## National Advisory Council for Human Genome Research

May 2016

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



## Director's Report-Related Documents: May 2016

[Director's Report](#) 

[Director's Report](#) 

No.	Relevant Documents
1	<a href="#">Bettie Graham: Beyond Genomics</a>
2	<a href="#">New Director, National Library of Medicine</a>
3	<a href="#">New Director, NIH Environmental Influences on Child Health Outcomes (ECHO) Program</a>
4	<a href="#">New Commissioner, U.S. Food and Drug Administration</a>
5	<a href="#">National Cancer Moonshot Initiative</a>
6	<a href="#">Basic Science: Bedrock of Progress</a>
7	<b>Genetic Research Privacy Protection Act</b> <a href="#">Genetic Research Privacy Protection Act Bill</a> <a href="#">Genetic Research Privacy Protection Act Fact Sheet</a>

[genome.gov/DirectorsReport](http://genome.gov/DirectorsReport)

Document #



# Open Session Presentation

- **Presentation from the New Director,  
National Institute on Minority Health and  
Health Disparities (NIMHD)**

**Eliseo Perez-Stable**

# **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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# Bettie Graham: Beyond Genomics



**Bettie Graham, Ph.D.**  
**(with coaches Ray and Jean Finkleman)**

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# New Director, National Library of Medicine

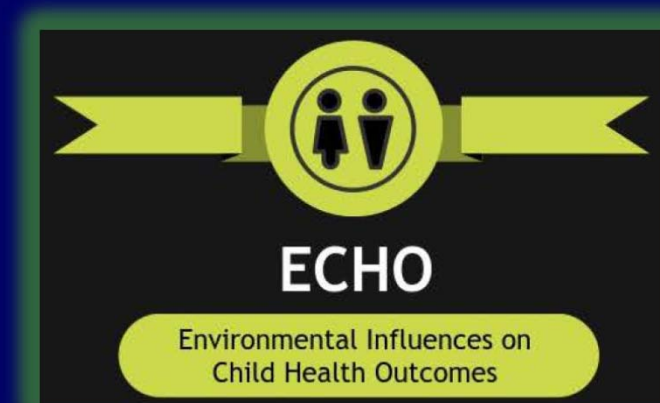


**Patricia Brennan, R.N., Ph.D.**

# New Director, NIH Environmental Influences on Child Health Outcomes (ECHO) Program



**Matthew Gillman, M.D.**



# Colleen Barros Departing as NIH Deputy Director for Management



**Colleen Barros, M.A.**



**John J. McGowan, Ph.D.**

# New Commissioner, U.S. Food and Drug Administration




**Robert Califf, M.D.**






# National Cancer Moonshot Initiative

 **NATIONAL CANCER INSTITUTE**

1-800-4-CANCER   Live Chat   Publications   Dictionary

ABOUT CANCER   CANCER TYPES   **RESEARCH**   GRANTS & TRAINING   NEWS & EVENTS   ABOUT NCI   search

Home > Research > Key Initiatives




**NATIONAL CANCER MOONSHOT INITIATIVE**

Research Areas

Task Force

Blue Ribbon Panel


Get Involved




## National Cancer Moonshot Initiative

During his State of the Union address on January 12, 2016, President Barack Obama announced the establishment of a new [National Cancer Moonshot Initiative](#) to accelerate cancer research. The initiative—led by Vice President Joe Biden—aims to make more therapies available to more patients, while also improving our ability to prevent cancer and detect it at an early stage.

Learn how NCI is supporting this important initiative, which seeks to accelerate progress in prevention, diagnosis, and treatment.

 Get email updates from NCI on the National Cancer Moonshot Initiative



### Research Areas

Information about the cutting-edge research opportunities that will receive additional funding, incentives, and other support to accelerate the rate of progress.



# Basic Science: Bedrock of Progress

## Science AAAS

### LETTERS

### Basic science: Bedrock of progress

Francis S. Collins<sup>1,\*</sup>, James M. Anderson<sup>2</sup>, Christopher P. Austin<sup>3</sup>, James F. Battey<sup>4</sup>, Linda S. Birnbaum<sup>5</sup>, Josephine P. Briggs<sup>6</sup>, Janine A. Clayton<sup>7</sup>, Bruce Cuthbert<sup>8</sup>, Robert W. Eisinger<sup>9</sup>, Anthony S. Fauci<sup>10</sup>, John I. Gallin<sup>11</sup>, Gary H. Gibbons<sup>12</sup>, Roger I. Glass<sup>13</sup>, Michael M. Gottesman<sup>14</sup>, Patricia A. Gray<sup>15</sup>, Eric D. Green<sup>16</sup>, Franziska B. Greider<sup>17</sup>, Richard Hodes<sup>18</sup>, Kathy L. Hudson<sup>1</sup>, Betsy Humphreys<sup>19</sup>, Stephen I. Katz<sup>20</sup>, George F. Koob<sup>21</sup>, Walter J. Koroshetz<sup>22</sup>, Michael S. Lauer<sup>23</sup>, Jon R. Lorsch<sup>24</sup>, Douglas R. Lowy<sup>25</sup>, John J. McGowan<sup>26</sup>, David M. Murray<sup>27</sup>, Richard Nakamura<sup>28</sup>, Andrea Norris<sup>29</sup>, Eliseo J. Perez-Stable<sup>30</sup>, Roderic I. Pettigrew<sup>31</sup>, William T. Riley<sup>32</sup>, Griffin P. Rodgers<sup>33</sup>, Paul A. Sieving<sup>34</sup>, Martha J. Somerman<sup>35</sup>, Catherine Y. Spong<sup>36</sup>, Lawrence A. Tabak<sup>1</sup>, Nora D. Volkow<sup>37</sup>, Elizabeth L. Wilder<sup>38</sup>

+ Author Affiliations

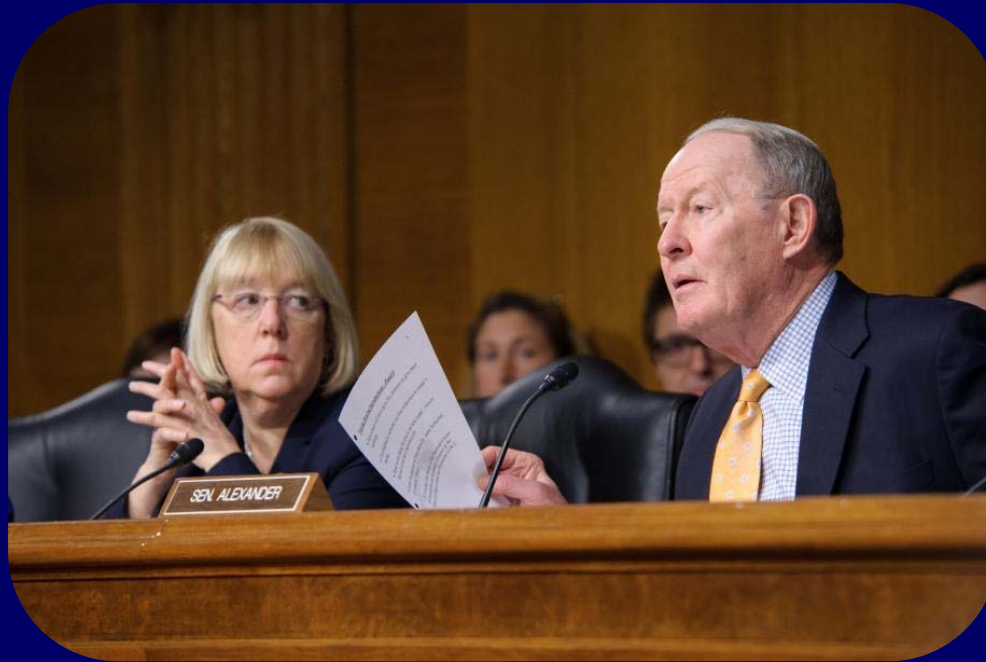
\*Corresponding author. E-mail: collinsf@mail.nih.gov

Science 25 Mar 2016;  
Vol. 351, Issue 6280, pp. 1405  
DOI: 10.1126/science.351.6280.1405-a



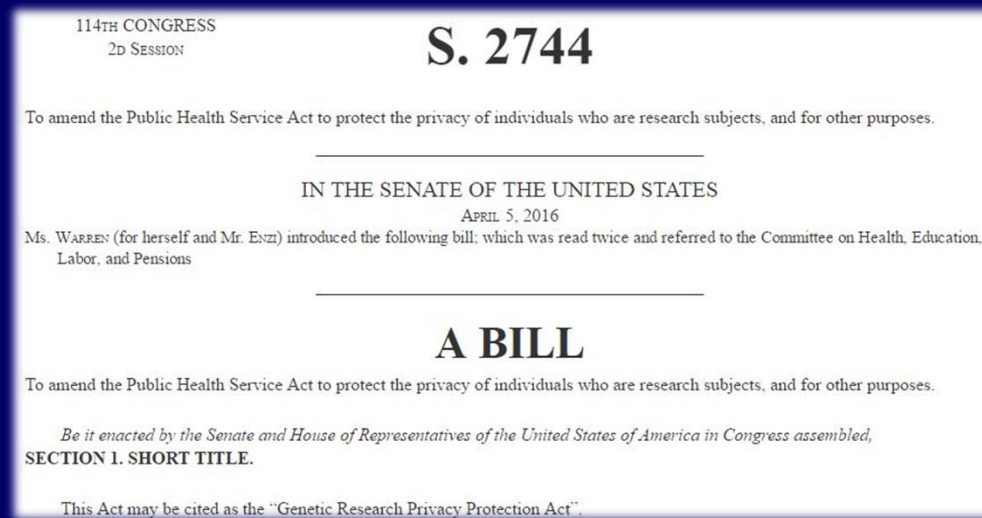
PHOTO: GPHIL DEGGINGER/ALAMY STOCK PHOTO

# **'Cures' Legislation Passes Senate**



- **Senate HELP Committee marked up 19 bills as part of 'Cures' agenda**
- **Proposed funding increases for NIH**

# Genetic Research Privacy Protection Act



- **Senate sponsors: Warren and Enzi**
- **Enhance privacy protections for individuals who contribute re-identifiable information to federally funded research**
- **Mandatory Certificates of Confidentiality**
- **FOIA exemptions**

# NIH Appropriations and Budget

## Fiscal Year 2017 President's Budget

Entity	FY15	FY16	FY17
NIH	\$30.1 B	\$32.3 B	\$33.1 B
NHGRI	\$498.7 M	\$513.2M	\$513.2 M

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# AAAS 2015 Philip Hauge Abelson Prize



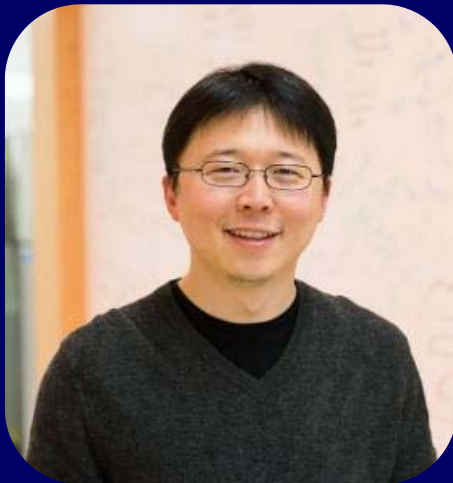
**Eric Lander, Ph.D.**



# Canada Gairdner Awards



**Anthony Fauci, M.D.**



**Feng Zhang, Ph.D.**



**Jennifer Doudna, Ph.D.**



**Emmanuelle Charpentier, Ph.D.**



# Genetics Society of America 2016 George W. Beadle Award



**Susan Celniker, Ph.D.**





# New Fellow, Royal Society



**Gil McVean, Ph.D.**



# **Elected to National Academy of Sciences**

**Adrian Bird**

**Myles Brown**

**Joseph DeRisi**

**Philip Hieter**

**Hopi Hoekstra**

**Robert Kingston**

**Kenneth Kinzler**

**Eugene Koonin**

**Stephen Palumbi**



**NATIONAL ACADEMY  
OF SCIENCES**

# New President, Rockefeller University



**Richard Lifton, M.D., Ph.D.**

# New Chief Scientific Officer, Metabiota



**Eddy Rubin, M.D., Ph.D.**

# Jan Witkowski Steps Down as Director, CSHL Banbury Center



**Jan Witkowski, Ph.D.**



# *MIT Technology Review*

## Breakthrough Technologies for 2016



### Precise Gene Editing in Plants

CRISPR offers an easy, exact way to alter genes to create traits such as disease resistance and drought tolerance.



### DNA App Store

An online store for information about your genes will make it cheap and easy to learn more about your health risks and predispositions.





# Sequencing Human Genome Rated Top Healthcare Advance of Past 40 Years

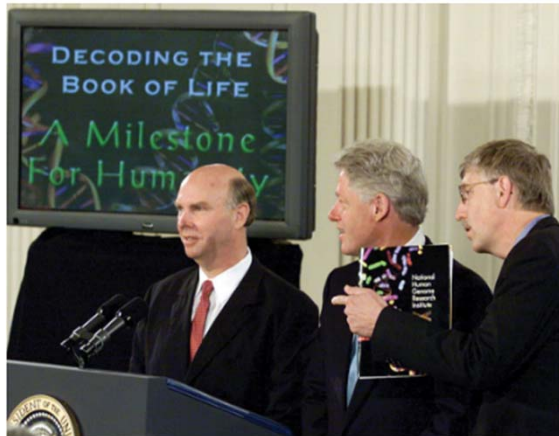
## Forward from 40: The top healthcare milestones of the past 40 years

By Modern Healthcare | March 26, 2016

In celebration of Modern Healthcare's 40th anniversary, readers were asked to choose the top healthcare milestones since the magazine's founding in 1976.

Over 700 respondents to the survey selected their top five picks from each of three categories: healthcare delivery, politics and policy, and science and technology. Read on for the complete list of the 40 most significant developments of the past 40 years.

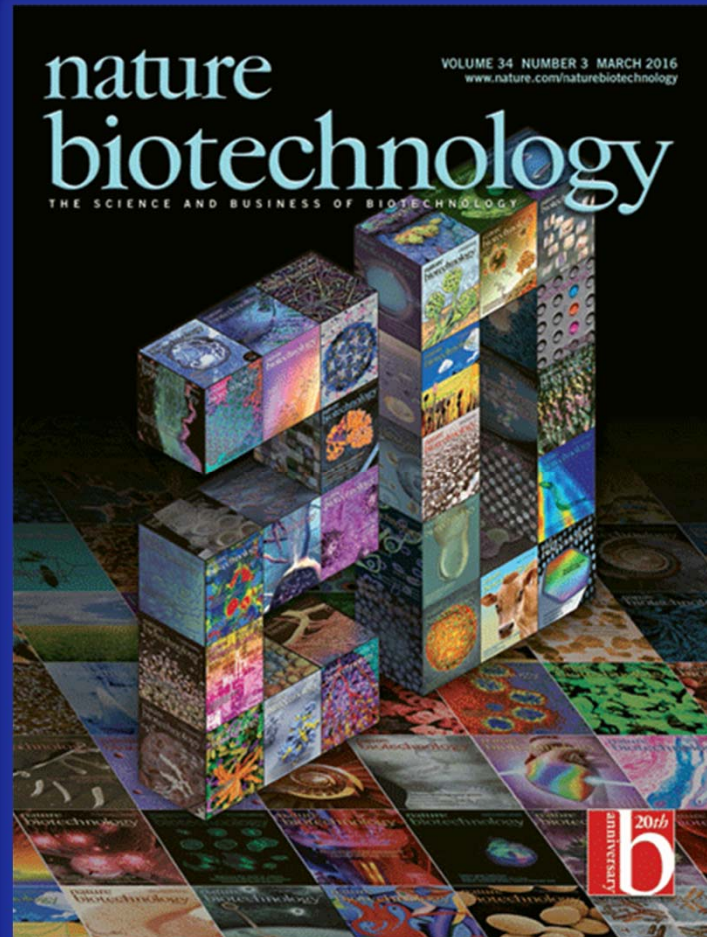
### 1. Sequencing the human genome



In June 2000, President Bill Clinton, flanked by Dr. Francis Collins, then director of the National Human Genome Research Institute at NIH, and Dr. Craig Venter, CEO of Celera Genomics, jointly announce the sequencing of the human genome.

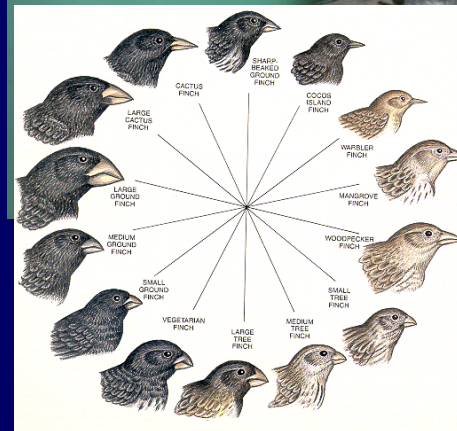
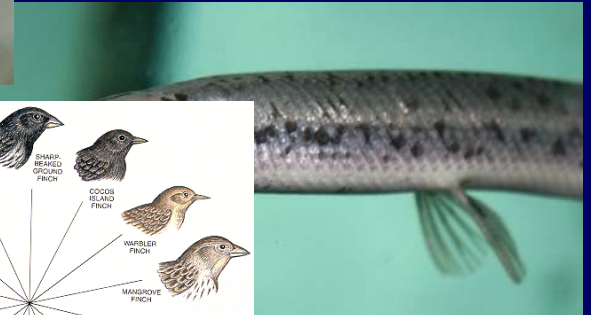


# 20<sup>th</sup> Anniversary Issue of *Nature Biotechnology*





# Genomes In The News...



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# Genome Sequencing Program



## 2016 Consortium Meeting

- Areas of collaboration identified
- Data sharing plans outlined
- Scientific directions discussed



# Technology Development Program



- **Novel Nucleic Acid Sequencing Technology Development**

RFA-HG-15-031 (to 33; R01, R21, and R43/44)

RFA-HG-15-039 (Direct to Phase II R44)

Upcoming due dates: July 14, 2016 and June 15, 2017

- **Novel Genome Technology Development**

PAR-16-14 (to 17; R01, R21, R43/44, and R44)

Upcoming due dates: October 31, 2016 and 2017



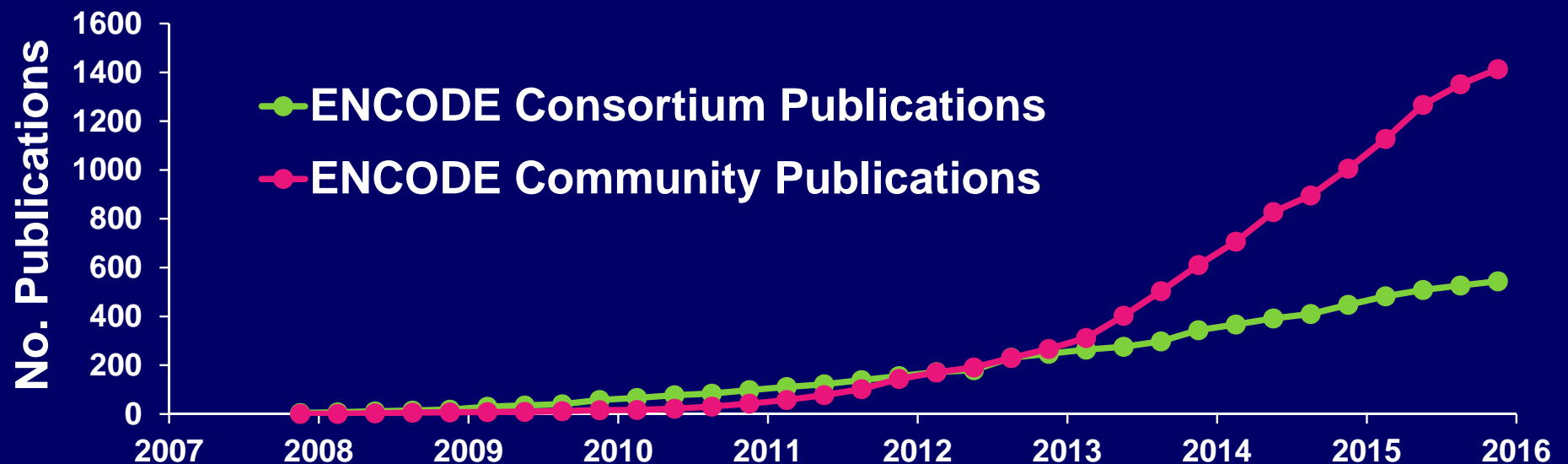
# Encyclopedia of DNA Elements (ENCODE)

- **ENCODE Outreach and Collaboration**

ENCODE 2016: Research Applications and Users Meeting  
June 8-10 (Palo Alto, CA)

Publication in *Trends in Genetics* on “Temporal Dynamics of Collaborative Networks in Large Scientific Consortia”

- **Publications using ENCODE Data**





# Encyclopedia of DNA Elements (ENCODE)

- **Applications have been received for FOAs:**

- Expanding the Encyclopedia of DNA Elements (ENCODE) in the Human and Mouse (RFA-HG-16-002)

- Characterizing the Functional Elements in the Encyclopedia of DNA Elements (ENCODE) Catalog (RFA-HG-16-003)

- Computational Analysis of the Encyclopedia of DNA Elements (ENCODE) Data (RFA-HG-16-004)

- ENCODE Data Coordination Center (RFA-HG-16-005)

- ENCODE Data Analysis Center (RFA-HG-16-006)

- **Key Dates:**

- Scientific Merit Review

- Summer 2016

- Advisory Council Review

- September 2016

- Anticipated Start Date

- February 2017

YOUR HEALTH

**Science**  
In Ne

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Updated February  
Published February

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LOCAL BIZ/TECH

Trans

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**Depress**  
**ancestr**

Originally published February

New research from  
hominids can reduce  
attack – but only

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**ROSEANNE BARR**  
MAY 26

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Los Angeles Times



**Science Now**

Discoveries from the world of science and medicine

f t e

Science / Science Now

**Your Neanderthal inheritance could affect  
your mood, your skin and your smoking habits**



A new study finds Neanderthal DNA influences many physical traits in people of Eurasian heritage. (Michael Smeltzer/Vanderbilt)

Presenting Author	Title	Type
Marc Williams Casey Overby David Crosslin	Practical Implementation of Genomic Sequencing in Healthcare Settings	Panel
Geneviève Galarneau	Usage of Electronic Medical Record Data to Characterize the <i>APOL1</i> Risk Allele Association with Blood Pressure Traits	Podium Presentation
Jacqueline Kirby	CDS-KB: A KnowledgeBase of Lessons Learned for Clinical Decision Support Implementation	Podium Presentation







# Clinical Sequencing Exploratory Research Program

- Enrolled 4,429 adults and 1,148 children
- 231 publications, 16 working group publications

Performance of ACMG-AMP Variant-Interpretation Guidelines among Nine

Lab  
Clinical Sequencing Exploratory Research Consortium: Accelerating  
Evidence-Based

Laura M.  
Jonatha  
Arezou  
Aleksan  
Natasha  
16 These a  
Robert C. Green, PhD, K  
Barbara A. Bernhardt, Le  
Charlis F. Caga-anan,  
Stephanie M. Fullerton, L  
Michelle Huckaby Lewis,  
Ian D. Krantz, Teri A. Ma  
Deborah A. Nickerson, Je  
Joseph S. Salama, Sara  
David L. Veenstra, Nikhil  
the CSER Consortium

JAMA Oncology

Research

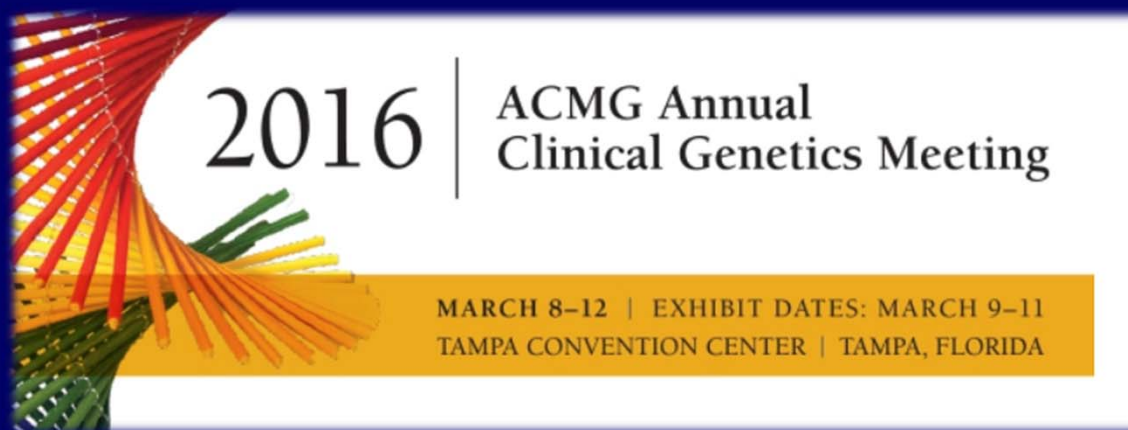
Original Investigation

## Diagnostic Yield of Clinical Tumor and Germline Whole-Exome Sequencing for Children With Solid Tumors

D. Williams Parsons, MD, PhD; Angshumoy Roy, MD, PhD; Yaping Yang, PhD; Tao Wang, PhD; Sarah Scollon, MS, CGC; Katie Bergstrom, MS, CGC; Robin A. Kerstein, BS, MT; Stephanie Gutierrez, BS; Andrea K. Petersen, MD; Abhishek Bavle, MD; Frank Y. Lin, MD; Dolores H. López-Terrada, MD, PhD; Federico A. Monzon, MD; M. John Hicks, MD, PhD, DDS; Karen W. Eldin, MD; Norma M. Quintanilla, MD; Adekunle M. Adesina, MD, PhD; Carrie A. Mohila, MD, PhD; William Whitehead, MD; Andrew Jea, MD; Sanjeev A. Vasudevan, MD; Jed G. Nuchtern, MD; Uma Ramamurthy, PhD; Amy L. McGuire, JD, PhD; Susan G. Hilsenbeck, PhD; Jeffrey G. Reid, PhD; Donna M. Muzny, MSc; David A. Wheeler, PhD; Stacey L. Berg, MD; Murali M. Chintagumpala, MD; Christine M. Eng, MD; Richard A. Gibbs, PhD; Sharon E. Plon, MD, PhD



# Clinical Sequencing Exploratory Research Program



- **CSER2 Request for Applications released**  
(RFA-HG-16-010, -011, -012)
- **Receipt dates:**  
August 5, 2016 for non-AIDS applications  
September 7, 2016 for AIDS applications







# Implementing Genomics In Practice (IGNITE) Network



- 36 publications
- Genomic Medicine Payer Engagement Meeting (August 18, 2016)
- IGNITE and Beyond: The Future of Genomic Medicine Implementation Meeting (August 30, 2016)



- ClinGen's website redesigned to feature curation results, tools, and resources

**GenomeConnect**   
Learn more and enroll in the  
GenomeConnect, ClinGen  
portal.

**Pathogenicity Calculator**  **Registration Required**  
The Calculator  
provides an intuitive interface to enter and store  
ACMG-style evidence tags, generate  
conclusions, examine and share the  
evidence behind each conclusion and identify  
that--if present--may lead to new c

**Data Model Documentation**   
This site documents the  
models created by the Data Model  
Working Group.



- Successful presence at ACMG 2016



# Newborn Sequencing in Genomic Medicine and Public Health (NSIGHT)

## PEDIATRICS

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

JANUARY 2016 • VOLUME 137 • SUPPLEMENT 1

## SPECIAL SUPPLEMENT

Bioethical Issues in Pediatrics: A Series of Supplements to  
*Pediatrics*

Supplement V:

Ethical Issues in Genomic Testing of Children

# Genomic Medicine Working Group

NHGRI Genomic Medicine IX: *Bench to Bedside – Mind the Gaps*

April 19-20, Silver Spring, MD

- Reviewing successful interactions between basic scientists and clinicians
- Identifying ways to enhance communications
- Integrating basic science efforts with clinically important questions



# Computational Genomics and Data Science Program Meetings

## NHGRI Large Genomic Resources Meeting

May 23-24, 2016

- Discuss new organizational models for:
  - Model Organism Databases
  - Gene Ontology Consortium
- Participants:
  - PIs and key personnel
  - External advisors (including Council)
  - NIH staff
- Will report to Council in September 2016





# Computational Genomics and Data Science Program Meetings

## NHGRI Informatics Workshop

Sept 29-30, 2016

- Develop plan to expand and strengthen the NHGRI extramural computational and data science portfolio over the next 5-10 years
- Will report to Council in February 2017



# Research Training and Career Development Program



**2016 NHGRI Annual Trainee Meeting**  
**April 7-8, 2016**

# Research Training and Career Development Program

- **Renewal of existing Data Analysis and Coordinating Center (DACC)**

**PIs: Drs. Treva Rice and Donna Jeffe**

- **Reissue Diversity Action Plan (DAP) Program announcement later this year**
- **New Genomic Medicine and Ethical, Legal, Social Implications (ELSI) T32 training grants**



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# Integrative Human Microbiome Project



- **NIST-NIAID-HMP Workshop: August 2016**
- **Microbiome Interagency Working Group**
- **‘Microbiome Futures’ Meeting: August 2017**



[illegible]

- **First LINCS outreach meeting held in March**
- **First LINCS data challenge just opened**
- **NIH now receiving grant applications from non-LINCS scientists that utilize LINCS resources**

# Protein Capture Reagents Program

- **Grantee Phase:** near completion, one grantee finishing production of monoclonal antibodies
- **Contract Phase:** third party validation of ~500 antibodies out of 1,700+ produced



PROTEIN CAPTURE  
REAGENTS PROGRAM



## Protein Capture Reagents Program

The Common Fund's **Protein Capture Reagents** program is developing new resources and tools to understand the critical role the multitude of cellular proteins play in development, health, and disease. These resources will support a wide-range of research and clinical applications by enabling the isolation and tracking of proteins of interest.

[Full Catalog](#)





# Human Heredity and Health in Africa (H3Africa)



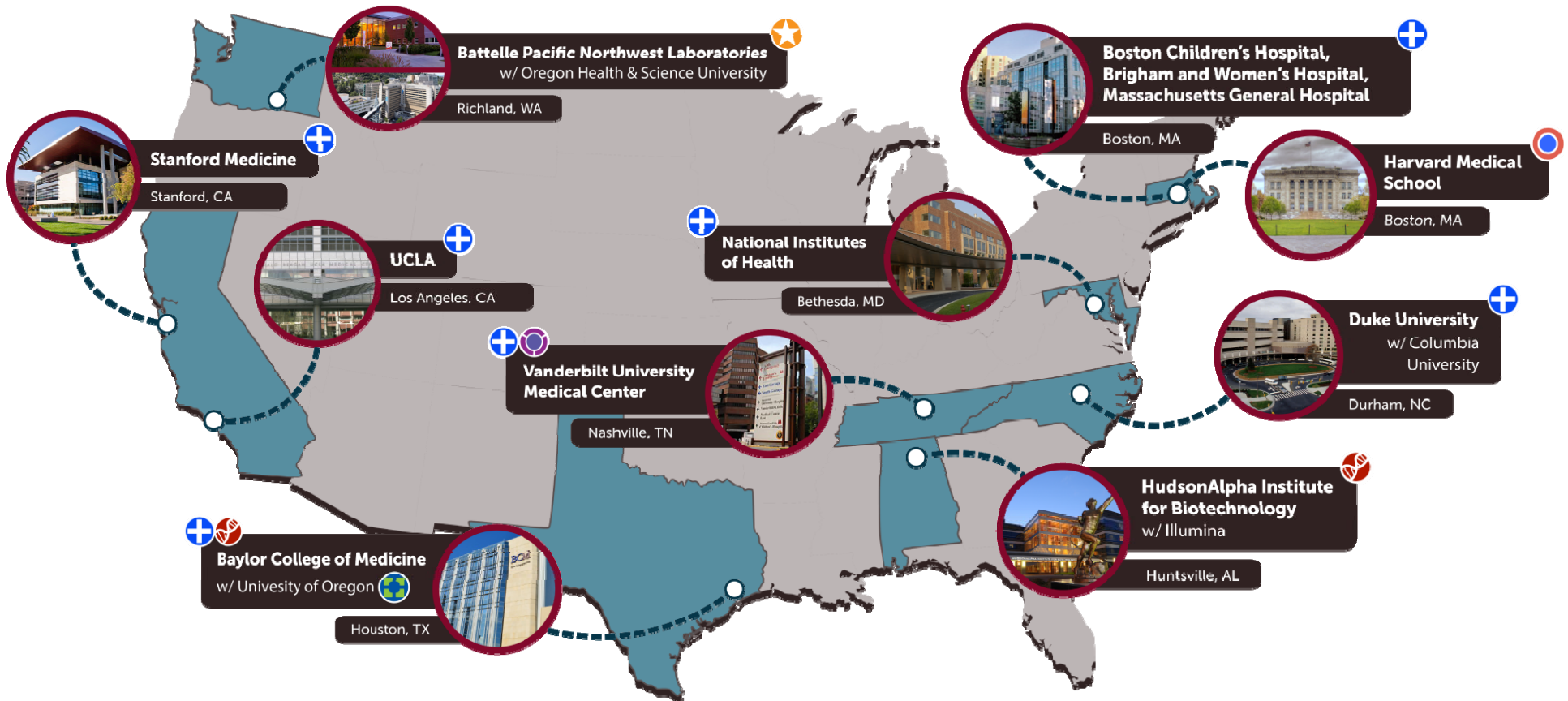
- **Community Engagement Evaluation Workshop**  
March 2016 (Stellenbosch, South Africa)
- **8<sup>th</sup> Consortium Meeting (Dakar, Senegal)**  
Editors Publication Workshop  
Sustainability Panel  
African Society of Human Genetics Meeting
- **Renewed by Common Fund**

# Undiagnosed Diseases Network (UDN)



Undiagnosed  
Diseases Network

UDN Site Locations



**516 Applications**  
**185 Acceptances**

 **APPLY**



**Document 35**

# New Director, Precision Medicine Initiative (PMI) Cohort Program

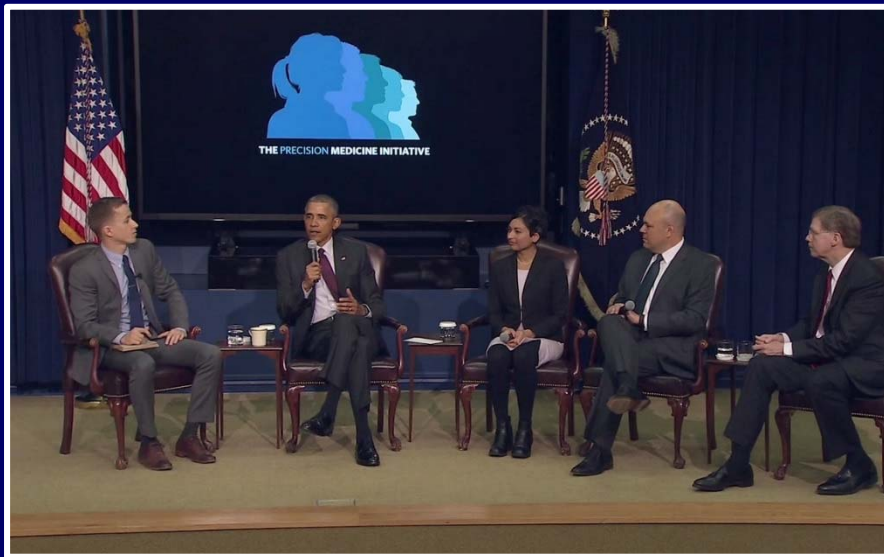


**Eric Dishman**





# Precision Medicine Initiative (PMI) Cohort Program Activities



VANDERBILT  
UNIVERSITY

**verily**

**WONDROS**

**HCM**  
STRATEGISTS

**HRSA**  
Health Resources & Services Administration



Institutional  
Board (IRB)

COMMUNITY HEALTH CENTER  
**FQHC**  
QUALITY

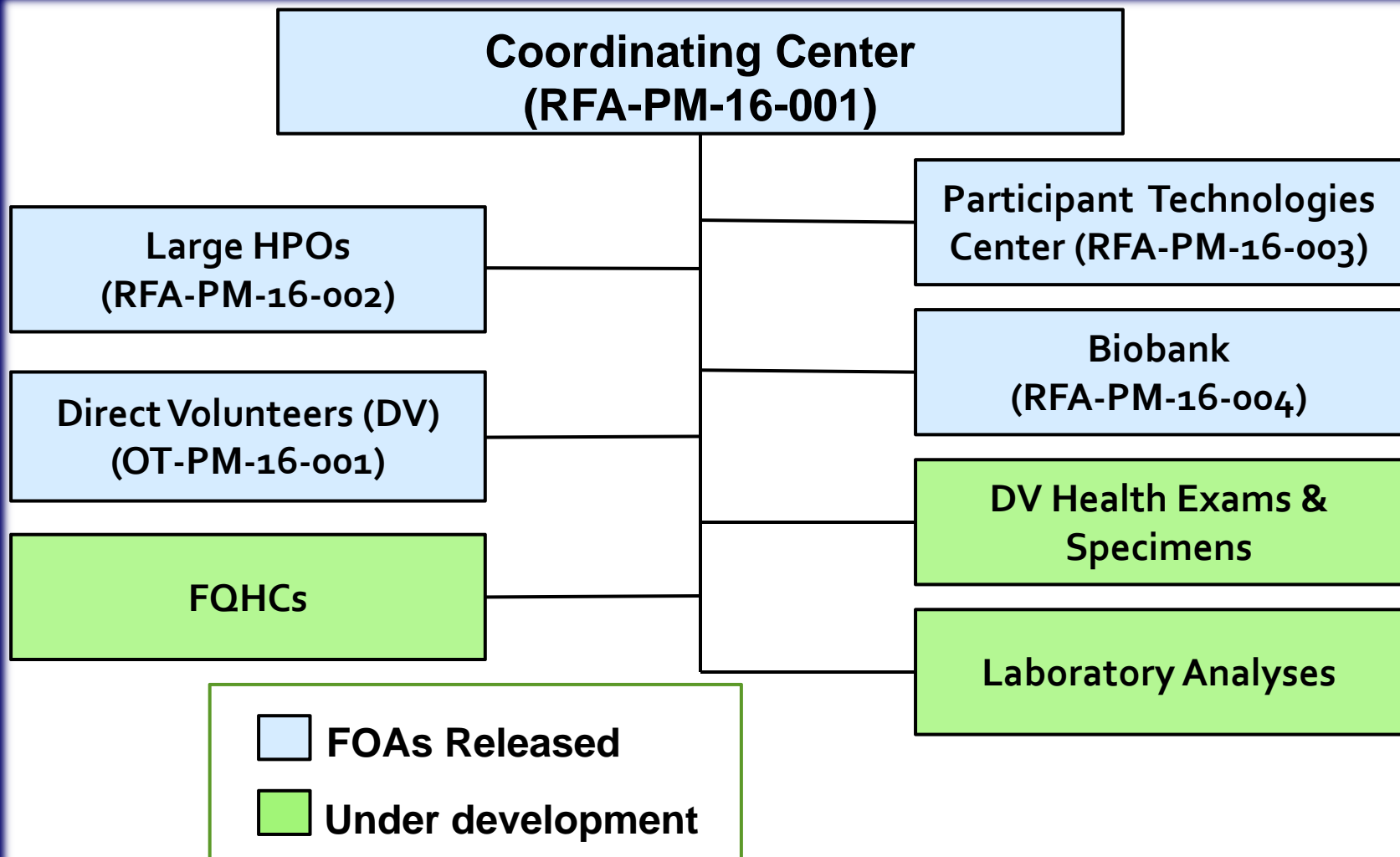


**HARVARD**  
MEDICAL SCHOOL



Document 36

# Precision Medicine Initiative (PMI) Cohort Program Components



# Director's Report Outline

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# Genomic Medicine on Genome.gov



- Central landing page for general audiences
- Search engine optimization
- New URL aliases
- Updated content and improved navigation



# History of Genomics Program

- Oral histories
- “History of Genomics & Molecular Biology” lecture series
- “A Quarter Century after the Human Genome Project's Launch: Lessons Beyond the Base Pairs” seminar series



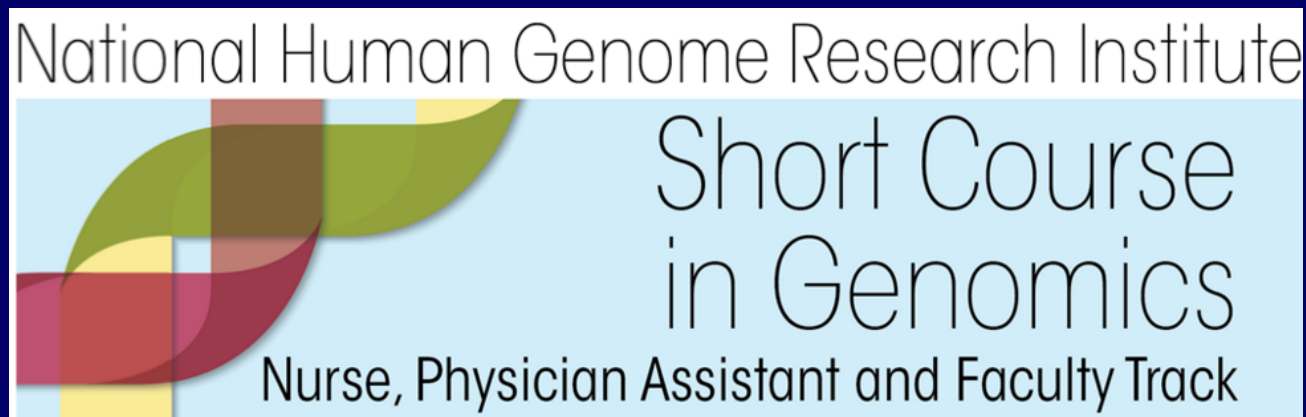
# Investigational Device Exemptions (IDEs) and Genomic Research Workshop



- **Save the Date: Friday, June 10, 2016**
- **To register, email: [NHGRIIDEWorkshop@nih.gov](mailto:NHGRIIDEWorkshop@nih.gov)**

# Short Course in Genomics: Nurse, Physician Assistant, and Faculty Track

- Sponsored by the Genomic Healthcare Branch
- Targets healthcare professionals and their educators
- Review existing curricula and successful models
- Application deadline: May 20, 2016



**August 1-3,  
2016 at NIH**

# National DNA Day 2016

**National DNA DAY 25** Celebrating Genomics Through Awareness

Find Events Plan An Event Celebrate With NHGRI Get Activity Ideas Learn The History Visit Newsroom

Connect with DNA Day

National DNA Day is a unique day when students, teachers and the public can learn more about genetics and genomics! The day commemorates the completion of the Human Genome Project in April 2003, and the discovery of DNA's double helix in 1953. [Subscribe to our newsletter!](#)

**Find Events In Your Area**

The National DNA Day Network has events all over the country. Find an event near you to celebrate DNA Day.

**Chat With Geneticists in the "Ask Me Anything" Series**

Join NHGRI, pgEd, and ASHG for a week-long National DNA Day "Ask Me Anything" (AMA) Series on reddit. /r/Science.

**Join @DNADay16 For a Twitter Chat**

#DNADay16

**Check out our 2016 DNA Day Lecture**

Harry Potter and the Genetics of Wizarding

**National DNA DAY 25** Celebrating Genomics Through Awareness

Find Events Plan An Event Celebrate With NHGRI Get Activity Ideas Learn The History Visit Newsroom

Connect with DNA Day

**Find Events**

National DNA Day Events

**National DNA DAY AMA SERIES**

"Ask Me Anything" (AMA) Series  
On the reddit science community forum  
"/r/Science"

Hosted by ASHG, pgEd, and NHGRI  
Featuring prominent geneticists from a range of research areas.

**Monday, April 18 – Friday, April 22,  
& Monday, April 25**

**National DNA DAY TWITTER CHAT**

**#DNADay16 @DNADay**

**Monday, April 25th, 11:00 a.m.**

Join us for an exciting Twitter chat session celebrating genetics and genomics.

**Harry Potter and the Genetics of Wizarding**

**Eric P. Spana, Ph.D.**  
Assistant Professor of the Practice  
Department of Biology,  
Duke University

**HOT (Human Origins Today) Topic:**  
**Genes and Animal Coloration:**  
**It's Not All Black and White**

**William J. Pavan, Ph.D.**  
Senior Investigator,  
Genetic Disease Research Branch  
National Human Genome Research Institute

**National Museum of Natural History**  
Friday, April 29, 2016 , 4:00 p.m.–5:00 p.m.

**FREE** and open to the public

**NIH** National Human Genome Research Institute



# 2016 USA Science & Engineering Festival



# ***Genome: Unlocking Life's Code***

## **Exhibition Travel Schedule**

**2016**

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

September 30-January 1  
Exploration Place  
Wichita, KS

**2017**

January 28-May 29  
Peoria Riverfront Museum  
Peoria, IL

June 12-September 11  
Health Museum  
Houston, TX



**GENOME**  
**UNLOCKING**  
**LIFE'S**  
**CODE**



# Partnership for Community Outreach and Engagement in Genomics

**Your GENOME & YOU**

Knowing about your genome helps you understand your health and supports your health decisions.

**GENOME** is just a fancy word for all your DNA.

Your GENOME contains all the instructions for you to grow throughout your lifetime. These instructions are passed down from your mother and father.

$\frac{1}{2}$  Father  $\frac{1}{2}$  Mother

**Your GENOME**

Height ↑ Eye color ↓ Disease

These instructions make you unique.

You can play an active role in keeping your genome healthy. You can eat healthy foods. You can exercise. You can avoid things that might cause diseases.

**Your Life**

**Your Environment**

We are learning new things about the human genome every day.

Developed by the National Human Genome Research Institute's Partnership for Community Outreach and Engagement in Genomics. To find out more about genetics and genomics, visit [www.genome.gov](http://www.genome.gov).

**Your GENOME & YOU**

Your GENOME contains all the instructions for you to grow throughout your lifetime. These instructions are passed down from your mother and father.

$\frac{1}{2}$   $\frac{1}{2}$

You can play an active role in keeping your genome healthy. You can eat healthy foods. You can exercise. You can avoid things that might cause diseases.

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**Your Environment**

We are learning new things about the human genome every day.

# 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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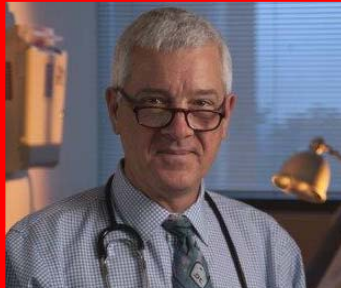
**Andy Baxevanis, Ph.D.**



# NHGRI Intramural Research Highlights

**Genetics  
inMedicine** Official Journal of the American College of Medical Genetics and Genomics

**An electronic atlas of human malformation syndromes in diverse populations**



**Clinical  
Chemistry**

**Systematic Evaluation of Sanger Validation of Next-Generation Sequencing Variants**

**AJHG**

**Biallelic *BRCA2* Mutations Shape the Somatic Mutational Landscape of Aggressive Prostate Tumors**





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

Search for **NHGRILANDSCAPE**

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





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**Thanks!**



**Special Thanks!**



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