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

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<td><a href="#">Bettie Graham: Beyond Genomics</a></td>
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<td><a href="#">New Director, National Library of Medicine</a></td>
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<td><a href="#">New Director, NIH Environmental Influences on Child Health Outcomes (ECHO) Program</a></td>
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<td><a href="#">New Commissioner, U.S. Food and Drug Administration</a></td>
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<td><a href="#">National Cancer Moonshot Initiative</a></td>
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- [Genetic Research Privacy Protection Act](#)
  - [Genetic Research Privacy Protection Act Bill](#)
  - [Genetic Research Privacy Protection Act Fact Sheet](#)
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.
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.

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


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2 Corresponding author. E-mail: collins@mail.nih.gov

Science 25 May 2018
Vol. 359, issue 6328, pp. 1405
DOI: 10.1126/science.359.6328.1405-a
‘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

<table>
<thead>
<tr>
<th>Entity</th>
<th>FY15</th>
<th>FY16</th>
<th>FY17</th>
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<tr>
<td>NIH</td>
<td>$30.1 B</td>
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<td>NHGRI</td>
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AAAS 2015 Philip Hauge Abelson Prize

Eric Lander, 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
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.
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.
Forward from 40: The top healthcare milestones of the past 40 years

By Modern Healthcare | March 20, 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.
20th 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
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
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
- Anticipated Start Date: February 2017
Your Neanderthal inheritance could affect your mood, your skin and your smoking habits
<table>
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<tr>
<th>Presenting Author</th>
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<tr>
<td>Marc Williams, Casey Overby, David Crosslin</td>
<td>Practical Implementation of Genomic Sequencing in Healthcare Settings</td>
<td>Panel</td>
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<td>Geneviève Galarneau</td>
<td>Usage of Electronic Medical Record Data to Characterize the APOL1 Risk Allele Association with Blood Pressure Traits</td>
<td>Podium Presentation</td>
</tr>
<tr>
<td>Jacqueline Kirby</td>
<td>CDS-KB: A KnowledgeBase of Lessons Learned for Clinical Decision Support Implementation</td>
<td>Podium Presentation</td>
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Clinical Sequencing Exploratory Research Program

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

- Successful presence at ACMG 2016
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
Develop plan to expand and strengthen the NHGRI extramural computational and data science portfolio over the next 5-10 years.

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
Integrative Human Microbiome Project

- NIST-NIAID-HMP Workshop: August 2016
- Microbiome Interagency Working Group
- ‘Microbiome Futures’ Meeting: August 2017
- 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
Human Heredity and Health in Africa (H3Africa)

- Community Engagement Evaluation Workshop
  March 2016 (Stellenbosch, South Africa)
- 8th Consortium Meeting (Dakar, Senegal)
  Editors Publication Workshop
  Sustainability Panel
  African Society of Human Genetics Meeting
- Renewed by Common Fund
New Director, Precision Medicine Initiative (PMI) Cohort Program

Eric Dishman
Precision Medicine Initiative (PMI) Cohort Program Activities
Precision Medicine Initiative (PMI) Cohort Program Components

- Coordinating Center (RFA-PM-16-001)
  - Large HPOs (RFA-PM-16-002)
  - Direct Volunteers (DV) (OT-PM-16-001)
  - FQHCs
  - Participant Technologies Center (RFA-PM-16-003)
  - Biobank (RFA-PM-16-004)
  - DV Health Exams & Specimens
  - Laboratory Analyses

- FOAs Released
- Under development

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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
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
 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
Partnership for Community Outreach and Engagement in Genomics
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Senior Member, International Society for Computational Biology

Andy Baxevanis, Ph.D.
NHGRI Intramural Research Highlights

An electronic atlas of human malformation syndromes in diverse populations

Systematic Evaluation of Sanger Validation of Next-Generation Sequencing Variants

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**
Thanks!

Special Thanks!
Advancing human health through genomics research