

January 6, 2015

Happy New Year! I start 2015 optimistic that the new year will bring great things to the field of genomics.

Since my December issue of *The Genomics Landscape*, the Fiscal Year 2015 budget was passed by a 'cromnibus' bill that funds most federal government agencies (including NIH) through September 30, 2015. The funding bill appropriates just over \$499 million for NHGRI, a 0.3% increase over Fiscal Year 2014, and just over \$30.3 billion for NIH.

Also since my December issue, President Barack Obama <u>visited</u> the NIH to see first-hand the progress being made to fight the Ebola virus outbreak, to thank the NIH staff for their contributions to these efforts, and to reiterate his commitment to biomedical research. For further details about the President's visit, see directorsblog.nih.gov/2014/12/02/presidents-visit-to-nih-highlights-research-on-ebola/#more-3953.

In this month's *The Genomics Landscape*, I describe the next phase of NHGRI's well-known Genome Sequencing Program as well as highlight additional items that I hope will be of interest to you.

Specifically, December's *The Genomics Landscape* features stories about:

- Next Phase: NHGRI's Genome Sequencing Program
- Draft NIH Policy on the Use of a Single IRB for Multi-Site Research
- New Biographical Sketch Format Required for NIH Grant Applications
- Welcoming New NHGRI Advisors
- NIH Intramural Research Program Blog

All the best,



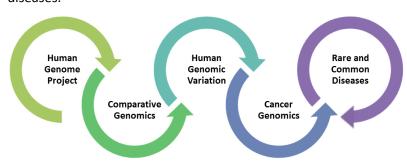
Watch here for current and upcoming locations of the Smithsonian-NHGRI exhibition "Genome: Unlocking Life's Code" as it tours North America!



Next Phase: NHGRI's Genome Sequencing Program

Large-scale genome sequencing has been a central component of NHGRI's Extramural Research Program since the Institute's inception, starting with the Human Genome Project. Considering the time and money it took to generate that first reference human genome sequence (upwards of 8 years and a billion dollars, respectively), I am amazed that we are now on the cusp of being able to generate a human genome sequence in a day and for less than \$1,000. That is simply astonishing!

Riding the waves of the many technical advances in DNA sequencing, the well-established NHGRI Genome Sequencing Program (GSP) has tackled a variety of biomedical research projects. Over the years, its priorities and focus areas have continuously 'morphed' to capitalize on new opportunities—starting with sequencing that first human genome and then moving on to sequence the genomes of multiple organisms for comparative genomics studies, sequencing many human genomes for human genomic variation studies, sequencing many cancer genomes for cancer genomics studies, and, most recently, sequencing human genomes for studies of rare and common diseases.



With the funding period of the current set of GSP grants ending later this year, NHGRI sought to consider the new opportunities for genome sequencing. In traditional NHGRI fashion, we convened a <u>program review meeting</u> in July 2014 that helped to detail several key applications for large-scale genome sequencing going forward, including: (1) determining the genomic contributions to rare and common disease; (2) improving our understanding of how genomic variation influences genome functions; (3) tackling new questions in comparative and evolutionary genomics; (4) assessing the utility of genome sequencing in clinical practice; and (5) understanding genotype-phenotype relationships.

Here, I highlight the first two Requests for Applications (RFAs) resulting from this strategic planning effort. The first RFA is for the establishment of the Centers for Common Disease Genomics (CCDG), which aim to identify genomic variants that contribute to common human diseases, explore a range of common diseases with different underlying genomic architectures, and develop

Draft NIH Policy on the Use of a Single IRB for Multi-Site Research



Recently, NIH issued a draft policy promoting the use of single Institutional Review Boards (IRBs) in multi-site clinical studies. IRBs review the risks and potential benefits of proposed research activities as they relate to the study participants. With clinical research increasingly spanning multiple institutions, it is critical to maintain the appropriate protection of research participants while reducing duplication of effort, mitigating delays in the initiation of research projects, and saving time and money. The draft policy aims to accomplish these objectives. NIH is soliciting comments on the draft policy until January 29. To review the draft policy and submit comments, visit

grants.nih.gov/grants/guide/notice-files/NOT-OD-15-026.html.

New Biographical Sketch Format Required for NIH Grant Applications



NIH is implementing a new format for the biographical sketch (i.e., biosketch) component of grant applications. Applicants with grant and cooperative agreement applications submitted for due dates after January 25 must use this new format. Changes include the extension of the page limit from four to five pages, as well as space for investigators to describe up to five of their most significant contributions to science (along with historical background about their research). The new format allows investigators to better highlight both their own research and their contributions to consortia and collaborations. For more information, see grants.nih.gov/grants/guide/notice-files/NOT-OD-15-032.html.

resources for multiple common disease research communities. The second RFA is for a renewal of the <u>Centers for Mendelian</u> <u>Genomics</u> (CMG), which aim to discover the genomic variants underlying several hundred human disorders, called Mendelian disorders, and to develop and share methods to enhance efficiency for others to discover such variants. Such knowledge would facilitate rapid and accurate diagnosis of these disorders and might lead to new therapeutic approaches.



These first two RFAs illustrate how our GSP has naturally evolved over the years to now strongly emphasize the use of genome sequencing for studying human diseases. Consistent with the conclusions of the July 2014 meeting, NHGRI plans to fund other areas involving genome sequencing— watch for a discussion of these potential new initiatives during the Open Session of the National Advisory Council for Human Genome Research meeting at 10:00 AM (Eastern Standard Time) on February 9, 2015 at genome.gov/GenomeTVLive/.

The Centers for Common Disease Genomics (CCDG) and the Centers for Mendelian Genomics (CMG) RFAs are available at grants.nih.gov/grants/guide/rfa-files/RFA-HG-15-001.html and grants.nih.gov/grants/guide/rfa-files/RFA-HG-15-002.htm, respectively. Applications for both RFAs are due April 7, 2015.

I find it striking to compare what the GSP will do over the next 4-5 years compared to what it was doing a decade ago. The fundamental differences in these two time periods truly reflect the monumental progress that the field of genomics has made in capitalizing on its advances in technology and knowledge.

President Barack Obama Visits NIH





On left (photo by Eric Green): President Barack Obama speaking at NIH on December 2, 2014. On right (photo by Ernie Branson): The President shakes hands with the NHGRI Director (who fortunately has very long arms!).

Welcoming New NHGRI Advisors



NHGRI relies extensively on external experts to advise the Institute in setting and achieving its goals for advancing genomics research. I would like to welcome newly nominated/appointed members of our two major advisory groups the National Advisory Council for Human Genome Research (NACHGR; see above photo), which mostly provides advice about our Extramural Research Program, and the Board of Scientific Counselors (BSC), which provides advice about our Intramural Research Program. Carol Bult, Ph.D., Dan Roden, M.D., Val Sheffield, M.D., Ph.D., and Jay Shendure, M.D., Ph.D. have been nominated to become new members of the NACHGR. Mark Daly, Ph.D. has been appointed to the BSC. For more information about the NACHGR, see genome.gov/10000905. For more information about the BSC, see genome.gov/10000911.

NIH Intramural Research Program Blog



Recently, NIH launched a new blog called "I am Intramural," which features commentaries written by scientists in the NIH Intramural Research Program. The first installment reflected a collaborative effort between NIH Director Dr. Francis Collins and NIH Deputy Director for Intramural Research Dr. Michael Gottesman, which was entitled "Science that Changes Lives." It also included some thoughts from Dr. Robin Stanley, L'Oreal USA Women in Science fellowship awardee and current Stadtman Tenure-Track Investigator, on making your scientific career search successful. For more information, visit irp.nih.gov/blog.







Genomics Research

NIH Researchers Link Chromosome Region to Gigantism

Researchers Conduct Comprehensive Genomic Study of Sub-Saharan Africans

<u>Cure Hunters: How Do Scientists Beat</u> <u>Devastating Diseases? By Living with</u> <u>Chronic Failure</u>

Genetic Errors Linked to Aging Underlie Leukemia that Develops after Cancer Treatment

Genes May Play Greater Role In Lou Gehrig's Disease

'Molecular Autopsy' Examines
Unexplained Deaths

Study Finds Genetic Clue to Menopause-Like Condition in Young Women

Ancient in Dog Years: NHGRI/Smithsonian to Sequence What May be North
America's Oldest Dog Relics

<u>California Girl's NIH Visit Includes Happy</u> <u>Excursions Around the Nation's Capital</u>

End of Cancer-Genome Project Prompts Rethink

Genome Advance of the Month

Researchers Examine Supercentenarians'
Genomes for Longevity Key

Upcoming Webcast

Workshop to Explore the Ethical, Legal and Social Implications (ELSI) of Citizen Science

New Genomics Videos

Cool Videos: Know When to Fold Them

NIH News of Interest

NIH Modification to Guidance on Marking Changes in Resubmission Applications

NIH Implementation of the US Government
Policy on Institutional Oversight of Life
Sciences Dual Use Research of Concern

Statement on the National Children's Study

<u>Genome Sequencing: Exploring the</u> Diagnostic Promise

NIH Complementary and Integrative Health Agency Gets New Name

NIH Grants Aim to Decipher the Language of Gene Regulation

Fundina News

Reminder for the Extramural Scientific Community: Implementation of the Genomic Data Sharing Policy Begins January 25, 2015

Funding Opportunities

NIA Coordinating Center for Genetics and Genomics of Alzheimer's Disease

Alzheimer's Disease Sequencing
Project Replication Phase Analysis
Studies

NIH Big Data to Knowledge Initiative Research Education: Massive Open Online Course on Data Management for Biomedical Big Data

NIH Big Data to Knowledge Initiative Research Education: Open Educational Resources for Sharing, Annotating and Curating Biomedical Big Data

NHGRI Establishment of New Training Program in the Ethical, Legal and Social Implications of Genetic and Genomic Research and Participation in PA-14-015 "Ruth L. Kirschstein NRSA Institutional Research Training Grants"

Past editions of *The Genomics Landscape* can be accessed at genome.gov/27541196.

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