October 4, 2016

Again this year, it came down to the wire for Congress to pass a Continuing Resolution for the federal budget, so as to keep the government open past the end of Fiscal Year 2016 (that is, beyond September 30, 2016). Similar to previous years of late, we are now operating on what can be likened to "bridge funding" for grants – although in this election year, it is not quite clear where the bridge will lead us in terms of our final Fiscal Year 2017 funding level! We will obviously be monitoring this situation very carefully in the coming weeks and months.

October's *The Genomics Landscape* features stories about:

- NHGRI Technology Development Programs Blaze Forward
- New Grants to Examine How Variation in Non-Coding Genomic Regions Affects Genome Function
- NHGRI Turns Docs into Researchers Through the Physician-Scientist Development Program
- Family Health History Tool Workshop
- ASHG-NHGRI Genetics & Public Policy and Genetics & Education Fellows

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!

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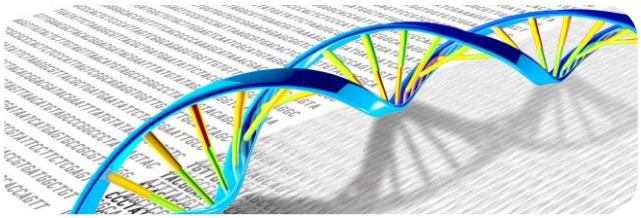
NHGRI Technology Development Programs Blaze Forward

I frequently boast of the accomplishments of the NHGRI Advanced Sequencing Technology Program, often referred to as the $1000 Genome Program. In fact, I am on record as saying that – in my opinion – it is the most successful technology development program in NIH history. As an example, such glowing accolades were heard out of my mouth a couple of weeks ago at the 78th meeting of the National Advisory Council on Human Genome Research.

Simply stated, the $1000 Genome Program has been spectacularly catalytic in advancing the development and refinement of new genome sequencing technologies. The fruits of multiple rounds of grant awards aimed at stimulating technology development in this area have helped to create the various new 'next-generation' DNA sequencing platforms that are now available. The field is in striking distance of truly achieving a '$1000 Genome.' Despite this collective accomplishment, NHGRI is not backing off in supporting genome sequencing technology development.



In 2015, NHGRI paused to take stock of the state of genome analysis technologies (DNA sequencing and other) in order to chart a path forward for future technology development efforts. We solicited input from the research community to guide our strategic thinking, which yielded concrete recommendations to support additional research in two main areas: DNA and RNA sequencing technologies and novel genomic technologies.

After generation of Funding Opportunity Announcements (FOAs), grant application receipt, peer review, and budget deliberations, NHGRI funded the first round of grants for Novel Nucleic Acid Sequencing Technology Development and Novel Genomic Technology Development. Both of these have an emphasis on pushing the genomics field to take big, bold steps.

The Novel Nucleic Acid Sequencing Technology Development program aims to develop new DNA and direct RNA sequencing technologies capable of yielding long read lengths at high accuracy and low cost. Recent awards were made to six investigators for a total of $6.75 million. Of note, this program is now funding technology development for direct RNA

New Grants to Examine How Variation in Non-Coding Genomic Regions Affects Genome Function

NHGRI, in partnership with the National Cancer Institute (NCI), has awarded new grants to support the development of computer modeling and experimental methods for understanding how variation in non-coding portions of the genome can affect genome function and influence disease risk. The grants, awarded through the Non-Coding Variants (NoVa) program, will support research that characterizes variants residing in the portion of the genome that does not encode proteins. We have come to learn that these non-coding regions play an integral role in genome function, both in healthy and diseased states. The five new grants support studies aiming to establish how non-coding regions control gene expression by developing computer-based statistical tools that model how variation in such regions affect the activity of genes. The specific projects will focus on non-coding genomic variation in the context of prostate cancer, autoimmune disorders, and heart disease. For more information, see genome.gov/27566612.

NHGRI Turns Docs into Researchers Through the Physician-Scientist Development Program

With advances in our abilities to sequence human genomes comes great potential to understand better the fundamental bases of human genetic diseases. Such studies require researchers who can dive into the highly technical world of genome science and physicians with clinical insights about genetic disease – but we also need physician-scientists who can do both. To that end, NHGRI has a long-standing commitment to training physicians to carry out independent research focused on medically important problems. The Physician-Scientist Development Program (PSDP) within the

sequencing – a new area for NHGRI. For more information on these awards, see genome.gov/27566794.

The Novel Genomic Technology Development program aims to catalyze investigator-initiated genomic technology development that will advance genomics research within five to seven years. The program hopes to enable a wide swath of genomic technology development, including single-cell methods, transcriptome analysis, and functional genomics. NHGRI hopes to accelerate scientific discovery by challenging researchers to stretch themselves in bringing forward completely novel ways of analyzing genomes and genome function. It is notable that the Novel Genomic Technology Development grants encourage researchers to put together teams of scientists from fields outside of genomics, such as bioengineering, to take on risky projects. Such efforts help to open up lines of communication among different scientific communities. Recent awards were made to seven investigators for a total of $10.4 million.

Though excited about these recent awards, I am also pleased to report that these are just the first sets of grants. The FOA for Novel Nucleic Acid Sequencing Technology Development is still active; a second set of applications has been received, and there is an upcoming application due date for the third set in June 2017. The FOA for Novel Genome Technology Development is also active, with upcoming applications for a second set due on October 31, 2016.

By continuing support for technology development, NHGRI aims to foster innovations that will continue moving genomics forward. With these two new programs, we are re-upping our commitment to technology development as a core element of NHGRI's genomics research portfolio.

Institute's Intramural Research Program is a mentored, pre-tenure-track training program started in 2003. The goal is to advance the training of physicians committed to pursuing translational research by providing: (1) a highly supportive research environment coupled with strong mentorship, and (2) the opportunity to develop skills necessary for establishing an independent translational genomics research program. The PSDP has been quite successful—so much so, that other NIH institutes have adopted its structure to develop their own analogous programs. More information about the NHGRI PSDP can be found at genome.gov/27565707.

Family Health History Tool Workshop

In June, NHGRI held a conference to explore the development and use of electronic Family Health History Tools (FHHTs). FHHTs are computer programs for collecting and organizing the health histories of a patient's family members; such information can be used to assess risks of hereditary conditions and to inform healthcare decisions, often in concert with a medical professional. At the conference, FHHT developers and vendors, patient advocates, subject matter experts, researchers, information technology specialists, electronic health record system vendors, and representatives from government agencies discussed changes in family health history data use, health information technology capabilities, and new research opportunities. Policy, research, and collaborator efforts to address issues in this important area were also discussed. A report from the conference is available at genome.gov/pages/health/healthcareprovidersinfo/2016-07-05_fhht_meeting_summary.pdf.

Spotlight on the Precision Medicine Initiative (PMI)

nih.gov/precisionmedicine

- In a Q&A with *Nature*, NIH Deputy Director for Science, Outreach, and Policy Dr. Kathy Hudson discusses the challenges of the Precision Medicine Initiative Cohort Program.
- PMI Director Eric Dishman was recently featured in the *NIH Record*: Dishman Follows Instincts to Top PMI Cohort Post.

ASHG-NHGRI Genetics & Public Policy and Genetics & Education Fellows

Each year, NHGRI's Division of Policy, Communications, and Education partners with the American Society of Human Genetics (ASHG) in sponsoring two fellowships – the Genetics and Public Policy Fellowship and the Genetics and Education Fellowship. The Genetics and Public Policy Fellowship provides the fellow an opportunity to gain valuable policy experience at the Institute, at ASHG, and in the U.S. Congress. The Genetics and Education Fellowship provides a genetics professional the opportunity to receive training and experience in preparation for a career in genetics/genomics education. This year's Genetics and Public Policy fellow is Christa Wagner, Ph.D., and the Genetics and Education fellow is Teresa Ramirez, Ph.D. – we welcome both to NHGRI!

On left: Christa Wagner.
On right: Teresa Ramirez.

Genomics Research

Genes Essential to Life Are Enriched for Human Disease Genes, Large-Scale Mouse Phenotyping Study Shows

Of Mice and Men: Study Pinpoints Genes Essential for Life

"Sixth Sense" May Be More than Just a Feeling

Out of Africa: DNA Analysis Points to a Single Major Exodus

New Videos

Seventy-Eighth Meeting: National Advisory Council for Human Genome Research

Ancient DNA and the New Science of the Human Past – David Reich

'Most Challenging Patients' Hope to Get Diagnoses from Medical Experts

Funding Opportunities

Centers of Excellence in Genomic Science (CEGS)

Dissemination and Implementation Research in Health (R21)

Serious Adverse Drug Reaction Research – R01 and R21

Genomics of HIV/AIDS Drug Response and Co-Morbidities

Big Data to Knowledge (BD2K): Enhancing the Efficiency and Effectiveness of Digital Curation for Biomedical Big Data, Community-Based Data and Metadata Standards Efforts, Research Education Curriculum Development: Data Science Overview for Biomedical Scientists, and Enhancing Diversity in Biomedical Data Science

Limited Competition: Additional Sequencing for the Alzheimer's Disease Sequencing Project

Data Integration and Analysis Tools: Accessible Resources for Integration and Analysis of Carbohydrate and Glycoconjugate Data in the Context of Comparable Gene, Protein, and Lipid Data

4D Nucleome Opportunity Pool Initiative Announces: Transformative Collaborative Project Award

NIH/NHGRI News of Interest

Unlocking Life's Code: September 2016 Newsletter

HHS Takes Steps to Provide More Information about Clinical Trials to the Public

Clinical Trials: Sharing of Data and Living Up to Our End of the Bargain

NIH Awards More than $150 Million for Research on Environmental Influences on Child Health

NCI Collaborates with Multiple Myeloma Research Foundation

NIH Common Fund Announces 2016 High-Risk, High-Reward Research Awards

Upcoming Webinar

Gabriella Miller Kids First Pediatric Research Program: Public Webinar – November 18, 2016