



The Genomics Landscape

A monthly update from the NHGRI Director



October 7, 2014

Today marks the end of the first week of Fiscal Year 2015 for the U.S. government. I am pleased to send you this message since it means that the federal government is ‘open for business’ (unlike the circumstance that we were in one year ago!). We are currently operating under a Continuing Resolution that funds the government through December 11, 2014, at Fiscal Year 2014 levels.

In this month’s *The Genomics Landscape*, I describe a recent release from the U.S. Food and Drug Administration (FDA) announcing the steps that they are taking to help ensure the reliability of certain diagnostic tests. See various details below, along with other information items that I hope will be of interest to you.

Specifically, October’s *The Genomics Landscape* features stories about:

- [Laboratory-Developed Tests: Public Comments Sought](#)
- [Small Business Grant Workshop at the 2014 ASHG Annual Meeting](#)
- [NIH Neuroscience Institute Director Retires](#)
- [NHGRI Intramural Faculty Recruitment](#)

Lastly, look for NHGRI at the 2014 [American Society of Human Genetics \(ASHG\) Annual Meeting](#) from October 18 – 22 in San Diego, California. As in previous years, there will be an NHGRI exhibit booth. Stop by to visit with NHGRI staff, including myself, to talk about the Institute and the exciting state of genomics research.

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!

Traveling Exhibition		
	Current	Next
	September 27, 2014 - January 4, 2015	January 22 - April 27, 2015
	Reuben H. Fleet Science Center San Diego, California	The Tech Museum of Innovation San Jose, California
	See unlockinglifescode.org for details	

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Laboratory-Developed Tests: Public Comments Sought



The integration of genomics into medicine is not solely dependent upon research advances – it also depends upon the development and implementation of appropriate policies and laws. For instance, anti-discrimination and privacy laws are important to protect patients from potential harm. Coverage and reimbursement policies are vital to promote patient access to appropriate genomic technologies. Policies fostering the development of electronic health record systems able to capture genomic information are necessary to enable clinical decision support. And there should be appropriate oversight to ensure the accuracy, consistency, and reliability of genomic-based tests that are available to healthcare professionals for clinical care. Recently, the U.S. Food and Drug Administration (FDA) issued draft guidance on the regulation of laboratory-developed tests (LDTs) – tests designed, manufactured, and used within a single laboratory – to address this latter need. While the FDA has always had the authority to regulate such tests, historically, it has not done so. Rather, it has exercised what is called ‘enforcement discretion.’



The FDA announcement comes at a critical transition for genomic medicine. Historically, most genetic/genomic LDTs have been used to identify rare disease-causing mutations, and the risk to a patient of an inaccurate or misinterpreted result was relatively low. Recent advances in technology have led to the use of more and more complex genomic tests for guiding clinical decisions, such as what drug to use to treat cancer. While we celebrate these clinical advances, it is important to ensure the quality and validity of the genomic tests for the sake of patient safety.

The idea of FDA regulation of LDTs is not new. In 2000 and in 2008, advisory committees to the Secretary of the Department of Health

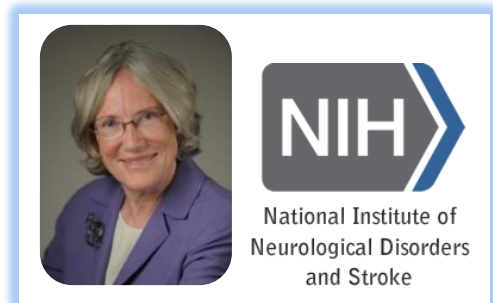
Small Business Grant Workshop at the 2014 ASHG Annual Meeting



NHGRI will hold a workshop at this year’s ASHG Annual Meeting entitled “Building your genomics business with SBIR/STTR support from NHGRI and NIH” to encourage participation in the Institute’s small business grant programs. NHGRI dedicates about \$12 million in small business grants each year, with particular interest in genomic analysis (from nucleic acids to cells), discovery and diagnostics 'omics' technologies, genomics-based algorithms (analysis and informatics), and general genome science. The amount of these funds will be increasing by a Congressionally mandated 0.12% of our total budget annually (or about \$0.6 million per year) in Fiscal Years 2015-2017.

For more information and to register for the October 19 workshop, visit genome.gov/SBIR2014Workshop.

NIH Neuroscience Institute Director Retires



A great friend of NHGRI, Dr. Story Landis, retired last month as Director of the National Institute of Neurological Disorders and Stroke (NINDS), a position she has held since 2003. Story came to NIH in 1995 to be the NINDS Scientific Director, and was promoted 8 years later to become the NINDS Director. Recently, she co-led NIH's

and Human Services recommended that FDA oversight should address all genetic/genomic laboratory tests. In 2010, the FDA announced that it was reviewing the issue and planned to regulate LDTs.

The goal of FDA's proposed oversight is to verify independently that LDTs being used clinically are safe and effective for patient care. Significantly, the draft guidance states that FDA intends to phase in LDT oversight over a number of years based on the risk associated with each test. Since the agency plans to prioritize their oversight, it will continue to practice 'enforcement discretion' for tests deemed to be lower risk, for seldom-performed tests for rare diseases, and for those fulfilling an unmet need.



The proposed guidance would bring oversight of LDTs into alignment with the oversight of those tests marketed and distributed as kits, which also follows a risk-based system. This would bring the same standard to both types of tests. This consistency in regulation is expected to benefit those wishing to commercialize a genetic/genomic test by removing the uncertainty in the current regulatory landscape. The FDA also expects that establishing strong and clear test standards will benefit patients by improving consistency and reliability in testing.

NHGRI supports the efforts of our sister agency to address this important issue for genomic medicine. Ultimately, the success of our collective efforts to improve health through genomics will be dependent upon finding the right balance in regulating genetic/genomic tests to ensure accuracy and clinical relevance while promoting innovation. I encourage you to review the [draft guidance](#) during the public comment period and to share your thoughts with the FDA on how it could be improved.

For further details, visit genome.gov/10002335.

role in the federal brain-mapping project known as the BRAIN Initiative. I have worked closely with Story for many years – first when we were both Scientific Directors and later when we were both Institute Directors. She has truly been a terrific scientific leader and a wonderful colleague and mentor. Dr. Walter Koroshetz will serve as Acting Director while a national search for Dr. Landis' replacement is conducted.

For more information, see ninds.nih.gov/about_ninds/message/message-dr-landis-retirement.htm.

NHGRI Intramural Faculty Recruitment



The NHGRI Intramural Research Program (IRP) recently launched a major recruitment to hire multiple investigators to pursue innovative, independent genomics and genetics research at the NIH. Multiple areas of genomics research are being targeted. Candidates must have a doctoral degree, as well as comprehensive, advanced training and a record of accomplishment. Interested applicants should visit <http://www.genome.gov/27559175> for further details and application instructions.

For more information on the Institute's IRP, see genome.gov/DIR.



<i>Genomics Research</i>	<i>NIH News of Interest</i>	<i>Genome Advance of the Month</i>
<u>Yet Another Genetic Reason to Protect Skin from Sun</u>	<u>U.S. Science Suffering from Booms and Busts in Funding</u>	<u>Investigating the Genomic Origins of the 2014 Ebola Outbreak</u>
<u>DNA Ties Ashkenazi Jews to Group of Just 330 People from Middle Ages</u>	<u>NIH Awards Aim to Improve Understanding of Cell Pathways, Development of New Therapies</u>	<i>New Genomics Videos</i>
<u>Sequencing of Five African Fishes Reveals Diverse Molecular Mechanisms Underlying Evolution</u>	<u>New Human Microbiome Project Marker Paper Highlights Studies on Pregnancy and Preterm Birth, IBD, and Diabetes</u>	<u>Genetics of Complex Disease - Larry Brody</u>
<u>Healthy Humans Make Nice Homes for Viruses</u>	<u>Request for Information (RFI): Consideration of Sex as a Biological Variable in Biomedical Research</u>	<u>NHGRI Advisory Council Meeting, September 8, 2014</u>
<u>Sex in the Sink: Gene-Swapping Bacteria Are Making New Superbugs</u>	<u>White House Declares War on Drug-Resistant Superbugs</u>	<u>Genetic Counseling Training Program Alumni Research Symposium 2014</u>
<u>Experts Provide Much-Needed Policy Analysis for Clinical Integration of Next Generation Sequencing</u>	<u>Statement on USG Policy for Institutional Oversight of Life Sciences Dual Use Research of Concern</u>	<u>NIH Leadership on C-SPAN's Washington Journal</u>
<u>Cancer Genome Sequencing Will Mean More Targeted Therapies</u>	<u>Take a Look at the Science Research Supported by Federal Funders: Launch of RePORTER tool</u>	<i>Funding Opportunity</i>
<u>Tall Tale: Scientists Unravel the Genetics of Human Height</u>	<u>Baylor College of Medicine, Medical College of Wisconsin to conduct DNA sequencing for Undiagnosed Diseases Network</u>	<u>NIH Pathway to Independence Award (K99/R00)</u>
	<u>NIH Awards Initial \$46 Million for BRAIN Initiative Research</u>	<u>4D Nucleome Program First Funding Opportunities</u>
	<u>NIH Common Fund Announces 2014 High-Risk, High-Reward Research Awardees</u>	<i>Genomics News</i>
	<u>Obama Honors Nation's Top Scientists</u>	<u>Genome Magazine Publishes Second Issue</u>

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