



February 4, 2014

For this second month of 2014, I hope you enjoy reading about the new trans-NIH Big Data to Knowledge (BD2K) Initiative, the centerpiece of NIH's efforts to address the 'Big Data' problem facing biomedical research. And while parts of the country continue to suffer the chilling effects of a polar vortex, I am relieved to report that Washington, D.C. shows some signs of a thaw with regard to the budget battles. The politicians in our nation's capital have agreed to a federal budget for this year! See details below along with other informational items that I hope will be of interest to you.

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

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All the best,

A handwritten signature in black ink, appearing to read 'Eric'.

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The Big Data to Knowledge (BD2K) Initiative

The topic of 'Big Data' (of all sorts) has become a hot one across the industrial, academic, and non-profit sectors. Recognizing the importance of biomedical Big Data to NIH, a Data and Informatics Working Group of the Advisory Committee to the NIH Director made a set of recommendations in 2012 (acd.od.nih.gov/Data%20and%20Informatics%20Working%20Group%20Report.pdf) that outlined programmatic ways for NIH to address the opportunities and challenges facing all biomedical researchers in accessing, managing, analyzing, and integrating the increasingly large amount of data. On the basis of that report, the Big Data to Knowledge (BD2K) Initiative was conceived.



The mission of BD2K is to enable a quantum leap in the ability of the biomedical research enterprise to maximize the value of the growing volume and complexity of biomedical data. New and powerful technologies, being used by a growing number of investigators, are resulting in the generation of increasingly large amounts of highly complex and diverse data. As a result, biomedical research is becoming increasingly data-intensive and data-driven. However, an insufficient availability of relevant software tools and, frequently, a lack of expertise often limit the ability of individual investigators to locate, analyze, and use these biomedical 'Big Data' to advance their research. BD2K aims to develop new approaches; useful standards; more effective methods, tools, and software; and improved competencies that will enhance the ability of all investigators to use biomedical 'Big Data' more effectively. To this end, BD2K is developing a program of research, implementation, and training in data science and other related fields, all of which are relevant to biomedical research.

BD2K is truly a trans-NIH initiative. All NIH Institutes and Centers, as well as the NIH Common Fund, are contributing to the funding for BD2K, starting with \$24M in Fiscal Year 2014 and increasing to

NHGRI's Fiscal Year 2014 Budget



Good news! The U.S. Congress passed and the President signed a new budget for the federal government on January 16. Previously, the federal government had been funded by a Continuing Resolution, which was scheduled to expire in mid-January. The budget for Fiscal Year 2014 (October 1, 2013 – September 30, 2014) provides \$29.9 billion to NIH. Of that, NHGRI is allocated \$498 million; this reflects a ~3% increase over the post-sequestration level for Fiscal Year 2013. That said, it is still a drop in funding compared to Fiscal Year 2012, when NHGRI's budget was \$513 million. To see NHGRI's budget request for Fiscal Year 2014, go to genome.gov/27553261.

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



NHGRI and the American Society of Human Genetics (ASHG) are now accepting applications for the 2014 Genetics and Public Policy Fellowship and for the new Genetics and Education Fellowship. The Genetics and Public Policy fellowship is designed as a bridge for genetics professionals wishing to transition to a policy career. The inaugural Genetics and Education Fellowship is designed for genetics professionals with an advanced degree who are early in their careers and interested in developing their expertise in national genomic literacy efforts, science education policy, and program development. NHGRI and ASHG will accept applications through April 25. For more information about the fellowships and how to apply, visit genome.gov/27556100.

about \$100M in Fiscal Year 2016. Staff members from almost every NIH Institute and Center are participating in BD2K's planning and implementation. BD2K is initially planned as a seven-year effort, through Fiscal Year 2020; toward the end of this period, there will be a rigorous review of the success of BD2K in meeting the 'Big Data' needs of the NIH-supported research community, with the results of that review determining the longer-term plans for trans-NIH data science activities.

Standing up BD2K has taken the hard work and dedication of many, and has been accomplished so rapidly that the Initiative is now poised to take off. The first set of planning workshops has been completed, more information has been obtained from the research community in the form of responses to Requests for Information, the first Funding Opportunity Announcements (FOAs) have been released (bd2k.nih.gov/funding_opportunities.html#sthash.c1MzcpY7.dpbs), and the first grant applications are under review. Several more funding opportunities are under development and will be released soon. All of this progress with BD2K was accomplished in the face of significant constraints resulting from sequestration and other budgetary uncertainties!

NHGRI has been heavily involved in the early stages of BD2K, and we will continue to be involved going forward. We are passionate about BD2K because of the major 'Big Data' challenges facing genomics and genomic medicine, but the fruits of BD2K will certainly benefit all of NIH and the larger biomedical research community.

For just over a year, I have served as Acting NIH Associate Director for Data Science, a newly created position to lead NIH's efforts in 'Big Data' and data science. Recently, Dr. Phil Bourne was named the first NIH Associate Director for Data Science (nih.gov/news/health/dec2013/od-09.htm). I am delighted to report that Phil will arrive in March, and he is excited to guide NIH's 'Big Data' enterprise, including leading the BD2K Initiative.

BD2K has created a positive buzz in the NIH research community. The BD2K Centers of Excellence FOA resulted in a very robust response, and the first awards will be made this summer. With more FOAs to be issued in the near future, we anticipate a growth in NIH-supported data science research and, eventually, a large benefit to the biomedical research community. To learn more about BD2K, visit bd2k.nih.gov. In these somewhat tumultuous budgetary times for NIH, it is truly gratifying to see this exciting and important effort launched.

Rare Disease Day at NIH



On February 28, NIH will celebrate the seventh annual Rare Disease Day with a day-long celebration and recognition of various rare disease research activities. The event will be held in the Masur Auditorium (Building 10) from 8:30 a.m. to 5:00 p.m. Attendance is free and open to the public. Rare Disease Day was established to raise public awareness about rare diseases, the challenges encountered by those affected, the importance of research to develop diagnostics and treatments, and the impact of these diseases on patients' lives. For further details and to register for this event, visit rarediseases.info.nih.gov/news-and-events/pages/28/rare-disease-day.

NIH and Appistry Partner to Implement Genome Analysis Pipeline for Undiagnosed Diseases Program



The NIH Undiagnosed Diseases Program is teaming up with Appistry, Inc., a provider of cloud-based computing and analytics, to implement a genome-analysis pipeline for diagnosing rare diseases. The pipeline will assemble reference sequence datasets using genomic information from a patient's family in an effort to find the genomic variants involved in the patient's disease, with the goal of leading to a diagnosis. This collaboration is an excellent example of public and private interests working together to achieve advances in genomic medicine. For more information about this partnership, see appistry.com/news-and-events/appistry-nih-partner-implement-genetic-analysis-pipeline.



Genomics News of Interest

Genomic Educational Resources for Pharmacists Added to G2C2 Website genome.gov/27555955

As Genomics Breaches Cancer Walls, Researchers Forge New Treatment Paths genome.gov/27555887

Harvard Medical School to Play Key Role in Undiagnosed Diseases Network genome.gov/27555821

Elephant Shark Genome Decoded news.wustl.edu/news/Pages/26319.aspx

NIH Scientists Map Genetic Changes that Drive Tumors in a Common Pediatric Soft-Tissue Cancer nih.gov/news/health/jan2014/nci-23.htm

Exploring the Complex Genetics of Schizophrenia directorsblog.nih.gov/2014/01/28/exploring-the-complex-genetics-of-schizophrenia/

TCGA Bladder Cancer Study Reveals Potential Drug Targets, Similarities to Several Cancers nih.gov/news/health/jan2014/nhgri-29.htm

Study Expands the Cancer Genomics Universe <http://www.broadinstitute.org/news/5467>

Accelerating Medicines Partnership nih.gov/science/amp/index.htm

New Funding Opportunities

Development of an NIH BD2K Data Discovery Index Coordination Consortium (U24) grants.nih.gov/grants/guide/rfa-files/RFA-HL-14-031.html

NHGRI Participation in PA-14-015: Restructuring of NHGRI Training Programs Focused on Genomic Sciences grants.nih.gov/grants/guide/notice-files/NOT-HG-14-016.html

NHGRI Participation in PA-14-015: Creation of New NHGRI Postdoctoral Training Programs in Genomic Medicine Research grants.nih.gov/grants/guide/notice-files/NOT-HG-14-017.html

NHGRI Participation in PA-14-044: Mentored Research Scientist Development Award grants.nih.gov/grants/guide/notice-files/NOT-HG-14-018.html

NHGRI Participation in PA-14-046: Mentored Clinical Scientist Research Career Development Award grants.nih.gov/grants/guide/notice-files/NOT-HG-14-019.html

Upcoming Webcast

NHGRI Advisory Council Meeting, February 10, 2014 genome.gov/27552683

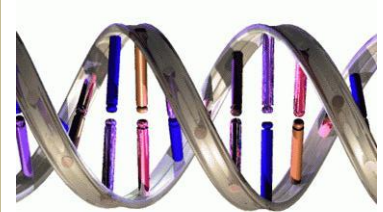
New Genomics Videos

Genomic Medicine VI Meeting youtube.com/playlist?list=PL1ay9ko4A8skrxKjHSIT-Z4DWdf9QpiGu

2013-2014 Genomics in Medicine Lecture Series youtube.com/playlist?list=PL1ay9ko4A8snwEd0X5IOYprkiXHgNlcSZ

NIH Extramural News

Rock Talk-Helping Connect You with the NIH perspective nexus.od.nih.gov/all/category/blog/



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