



September 3, 2014

A core value of the genomics research community is rapid and broad data sharing. This month, I share details about the new NIH Genomic Data Sharing Policy and its importance to the biomedical research enterprise. I also discuss the new policy during my Director's Report at the beginning of the open session of the National Advisory Council for Human Genome Research (NACHGR) meeting on Monday, September 8, starting at 10:00 AM. To watch the presentation as well as other portions of the NACHGR meeting, visit genome.gov/GenomeTVLive/.

For your reading pleasure, September's *The Genomics Landscape* features stories about:

- [The Natural Evolution of Genomic Data Sharing](#)
- [Genome: Unlocking Life's Code Exhibition Travels North America](#)
- [modENCODE Consortium Publishes Landmark Papers](#)
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Next month, I plan to share my thoughts on a recent release from the Food and Drug Administration (FDA) regarding steps that they are taking to help ensure the reliability of certain diagnostic tests. For more information, see fda.gov/NewsEvents/Newsroom/PressAnnouncements/ucm407321.htm and stay tuned.

All the best,

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

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The Natural Evolution of Genomic Data Sharing

Rapid and broad data sharing has been a hallmark of genomics since the early days of the Human Genome Project (HGP). Today, it is well-appreciated in genomics that the work of individual investigators and large collaborative efforts alike benefits from access to data resources such as ENCODE, 1000 Genomes, and The Cancer Genome Atlas (TCGA). Furthermore, the cumulative benefit realized through the culture of genomic data sharing transcends individual projects, and has been essential to accelerating genomics research across the board.



On August 26, NIH released its new [Genomic Data Sharing \(GDS\) Policy](#), updating a previous policy that focused on genome-wide association study data. The new policy promotes sharing a broader array of large-scale human and non-human genomic data generated for appropriate research purposes when those data are generated through NIH-supported or NIH-conducted research. This new policy represents another step the agency is taking to encourage the culture of data sharing and to extend such data-sharing practices beyond large research projects (e.g., HGP and 1000 Genomes).



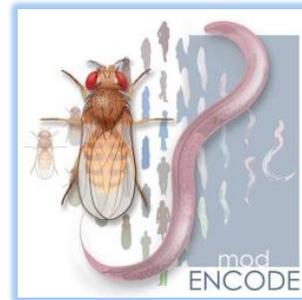
The GDS Policy builds upon an [existing data sharing](#) framework, ensuring responsible and respectful research participant protections and promoting appropriate access to genomic and associated data. In a recent *Nature Genetics* article entitled [“Data Use under the NIH GWAS Data Sharing Policy and Future Directions”](#), NIH leaders detail the experience to date of the two-tiered access structure established by the Genome-Wide Association Studies (GWAS) Policy and implemented through the [database of Genotypes and Phenotypes \(dbGaP\)](#). Under the GWAS policy, more than 2,200 investigators from 41 different countries have received access to dbGaP data from 304 studies and produced more than 900 publications and significant advances. The findings highlight that access to such data

Genome: Unlocking Life's Code Exhibition Travels North America

GENOME || UNLOCKING || LIFE'S || CODE ||

After a little over a year in residence at the Smithsonian's National Museum of Natural History, the exhibition “Genome: Unlocking Life's Code” closed its doors on September 1 and is preparing to tour around North America. Its first stop will be the Reuben H. Fleet Science Center in San Diego, CA, starting later this month. The exhibition will move across North America, ending its journey in Canada in January 2018. For more details and specific destinations on its tour, see genome.gov/27558789.

modENCODE Consortium Publishes Landmark Papers



The recently completed model organism ENCYclopedia Of DNA Elements (modENCODE) Project has published three integrative papers in *Nature* comparing fly, worm, and human genomic data and representing seven years of research. A series of companion papers has been published in *Nature*, *Genome Research*, and *Genome Biology*, with more manuscripts to follow. The goal of modENCODE has been to provide the biological research community with an extensive encyclopedia of genomic functional elements spanning the domains of gene structure, mRNA and ncRNA expression profiling, transcription factor-binding sites, histone modifications and replacement, chromatin structure, DNA replication initiation and timing, and copy number variation in the model organisms *C. elegans* and *D. melanogaster*. The findings offer new insights into various aspects of embryonic development, gene regulation, and other biological processes. To learn more, see nih.gov/news/health/aug2014/nhgri-27.htm.

provides not just an opportunity to accelerate research by virtue of combining large and information-rich datasets (or just enabling additional research questions to be addressed), but also the potential to maximize the public benefit achieved through this increased capacity.

Of course, sharing data generated about human research participants must be done in a manner that appropriately protects participant interests and respects the participant agreements for sharing data and health information. Like the GWAS Policy before it, the GDS policy aims to maximize scientific advances and potential public benefit in a manner consistent with research participant informed consent and the provision of appropriate considerations for participant privacy risks. While the new data types covered by the GDS Policy are different from the relatively narrow specifications for GWAS data previously covered, the ethical and scientific issues raised are very similar, and the appropriate principles that should govern responsible data use of various genomic data types are not substantially distinct. Consideration for the broad array of study designs, study populations, and potential consent provisions attached to genomic datasets remains integral to the GDS Policy, as does the agency's commitment to maintain the Policy's data-sharing expectations and participant-protection mechanisms.



The ethical use of genomic data and the public's trust in the systems used to govern that use continue to be of paramount concern for NHGRI and NIH. As such, the Institute, the agency, and I, personally, remain committed to the ongoing development and improvement of our data sharing policies and their oversight, so that biomedical (and genomic) research data are maximally utilized to promote public benefit. For further information on the GDS Policy, visit nih.gov/news/health/aug2014/od-27.htm.



ENCODE Dataset Release, New Data Release Policy, and ENCODE Portal



The Encyclopedia of DNA Elements (ENCODE) Consortium recently released data from the third phase of its program, a continuation of ENCODE and Mouse ENCODE whole-genome data production, with additional resources provided for data processing, analysis, and functional validation. This dataset was also released under a [new rapid release policy](#) by which primary data are released after data validation and processed data are released after quality assessment. Unlike the previous ENCODE production phase, there is no moratorium on external use of these data. All ENCODE data can now be accessed through the new ENCODE portal at encodeproject.org. The new portal serves as the primary and comprehensive source for data and information about ENCODE, with tools for browsing and searching ENCODE data via assays, biological samples, and experimental reagents used.

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



On left: Kate Blizinsky. On right: Elizabeth Tuck.

For many years, NHGRI has partnered with the American Society of Human Genetics (ASHG) in sponsoring a 16-month Genetics & Public Policy Fellowship. The program, started in 2002, provides the fellow an opportunity to gain valuable policy experience at the Institute, at ASHG, and in the U.S. Congress. This year, ASHG and NHGRI have also teamed up to sponsor a new Genetics & Education Fellowship for a genetics professional to receive training and experience in preparation for a career in genetics/genomics education. I am pleased to announce that this year's Genetics & Public Policy fellow is Kate Blizinsky, Ph.D. and this year's Genetics & Education fellow is Elizabeth Tuck, M.A. For details about the fellowships, visit genome.gov/27558850.

<i>NIH News of Interest</i>	<i>Genomics Research</i>	<i>Genome Advance of the Month</i>
<p>NIH Chief Keeps Hopes Afloat</p> <p>Promoting Health, Science, and Public Trust through Laboratory Safety</p> <p>RockTalk: Expanding the Impact of Genomic Data</p>	<p>Single Animal to Human Transmission Event Responsible for 2014 Ebola Outbreak</p> <p>Cancer Study Reveals Powerful New System for Classifying Tumors</p> <p>8,000-Year-Old Mutation Key to Human Life at High Altitudes</p> <p>Test Reliably Detects Inherited Immune Deficiency in Newborns</p> <p>Gut Bacteria May Reveal Colon Cancer</p> <p>Suspect Gene Corrupts Neural Connections</p>	<p>Researchers Examine Disease-causing Mutations in Mitochondrial Genomes</p> <p><i>Upcoming Webcast</i></p> <p>National Advisory Council for Human Genome Research Meeting, September 8, 2014</p> <p><i>New Genomics Videos</i></p> <p>Next-Generation Sequencing Technologies – Elaine Mardis</p> <p>David Rubenstein Interview of Dr. Collins at the Economic Club of Washington, D.C.</p> <p>2014 Genetic Counseling Training Program Alumni Research Symposium</p> <p>Genetics of Complex Disease – Larry Brody</p>
<p><i>NIH Funding News</i></p> <p>NIH Pathway to Independence Award (Parent K99/R00)</p> <p>Building Your Genomics Business with SBIR/STTR Support from NHGRI and the NIH Workshop at ASHG</p> <p>Nominations for the 2015 Lurie Prize in Biomedical Sciences</p> <p>NIH Single Cell Analysis Challenge: Follow That Cell</p>	<p><i>Media Availability</i></p> <p>Rare Kidney Tumor Provides Insights on Role of Metabolic Changes in Cancer</p>	

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