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Embargoed for release until
1 p.m. EDT, Wednesday, August 7, 2013

Access to HeLa cell genome data restored following agreement: Genome of HeLa cell line first published by *G3: Genes|Genomes|Genetics* now released to scientists

BETHESDA, MD -- The first study to sequence and analyze the entire genome of a HeLa cell line, along with access to its sequence data, has been published today (Wednesday, August 7) in its final version, by G3: Genes|Genomes|Genetics, an open-access, scientific journal of the Genetics Society of America.

The article, "The Genomic and Transcriptomic Landscape of a HeLa Cell Line," by Landry et al., was authored by scientists at the European Molecular Biology Laboratory (EMBL) in Heidelberg, Germany, and was published in an early online version March 11, 2013.

Genomic data from the HeLa cell line are also being released with the final version of the paper as a result of discussions between leaders of the National Institutes of Health (NIH) and relatives of Henrietta Lacks, from whose cervical tumor the original HeLa cell line was derived prior to her death in 1951. The genomic data will now be available to scientists via the NCBI's Database of Genotypes and Phenotypes (dbGaP).

In direct response to the concerns of the Lacks family that the privacy of their genetic information might be affected by the availability of the HeLa genome sequence data published in *G3*, the EMBL scientists voluntarily removed the HeLa cell line sequence data from public access, and offered to work with the family towards a mutually acceptable solution. NIH Director Francis S. Collins, M.D., Ph.D., and NIH Deputy Director for Science, Outreach, and Policy Kathy L. Hudson, Ph.D., met several times with representatives of the Lacks family, and came to a mutual understanding to allow biomedical researchers controlled access to the data.

Since their isolation in 1951, HeLa cells have been the most widely used human cell line in research. They have become a valuable resource for biologists, enabling momentous scientific breakthroughs including the development of the polio vaccine the Nobel Prize winning studies defining the role of telomerase in aging, and research on the causative role of human papillomavirus (HPV) in some types of cervical cancer. The latter discovery spurred the development of an HPV vaccine to prevent certain types of cervical cancer from occurring. In fact, Ms. Lacks' death was caused by cervical cancer.

The advent of genomics and rapid sequencing techniques has seen HeLa cells used in numerous large-scale studies of gene function and expression. Yet, "these studies using HeLa cells had to rely on information from the 'reference' sequence produced by the Human Genome Project, even though there was evidence that the genomes of HeLa cell lines were probably quite different," said Lars Steinmetz, Ph.D., who led the *G3* study.

Dr. Steinmetz and his team found the genome of the HeLa cell line that they sequenced differs dramatically from a normal human genome sequence. These differences include widespread sequence variation, extra copies of genes, and massive, complex rearrangements.

Because the dataset now will be available to the scientific community, researchers will be able to account for these differences when designing and interpreting experiments using this HeLa cell line. The genomic particularities of HeLa cells relate to their origin from an aggressive cancer and subsequent cultivation in laboratories for decades, both of which cause considerable genomic alterations.

"Understanding the unique nature of the HeLa genome is important for guiding future studies with these cells," explained Brenda Andrews, Ph.D., editor-in-chief of *G3*. "This paper and the genomic data it includes provide an important resource for the scientific community, especially with the increasing number of studies that require genome sequence information for accurate design and interpretation."

"We are very happy that *G3* is able to publish these findings, and to make this dataset available to the scientific community," says Adam Fagen, Ph.D., executive director of the Genetics Society of America. "The Lacks family has taken an important step towards ensuring HeLa cells continue to catalyze important advances in biomedical research."

Genomics studies like those commonly conducted with HeLa cells play an instrumental role in revealing how variation in genome sequence and function can lead to disease. "Progress in genomic research has already begun to transform modern medicine," said Tracey DePellegrin, executive editor of *G3*, which, like its sister journal GENETICS also published by Genetics Society of America, promotes full data sharing and dissemination for scientific reseachers, "and this progress is contingent on scientists being able to access the genomic sequences, now available through dbGaP.

"As soon as they learned about the Lacks family's concerns, the authors of the *G3* paper voluntarily removed the sequence data from public access. We supported our authors' response, in particular as a resolution to the issues became actively discussed by NIH officials and the Lacks family," she added.

The early publication of the *G3* study sparked dialogue among scientists, bioethicists and the public concerning the handling of genomic data. Such conversations become critical as technological advances make personal genome sequencing increasingly affordable and commonplace. Indeed, GSA is initiating discussions with other scientists, ethicists and members of the public to refine strategies to manage genomic data in socially conscious ways that foster progress in scientific research.

"Although NIH played an essential role in the discussions with the Lacks family about the use of HeLa cells, we all need to think about how we approach issues that arise as science moves forward, balancing privacy concerns with advances in research, and the ways policy can be updated to reflect these complexities," added Dr. Fagen. "Everyone – including scientists, the public, policymakers, our health care system, and research funders – has a stake in the outcome."

Titles and authors of the Perspectives:

"Building Trust in 21st Century Genomics"
Michael J. Szego, Ph.D., M.HSc. (clinical ethicist), Janet A. Buchanan, Ph.D., and
Stephen W. Scherer, Ph.D., all of The Centre for Applied Genomics, The Hospital for Sick Children, Toronto, Canada.

"Policy Uncertainty, Sequencing and Cell Lines"
Timothy Caulfield, B.Sc., LL.B., LL.M., of Health Law Institute, Faculty of Law, University of Alberta, Edmonton, Alberta Canada; and Amy McGuire, J.D., Ph.D., of Center for Medical Ethics and Health Policy, Baylor College of Medicine, Houston, Texas.

"From Tissues to Genomes"
Bartha M. Knoppers, Ph.D., Director, Centre of Genomics and Policy at McGill University, Montreal, Canada.

ABOUT *G3*|*Genes*|*Genomes*|*Genetics*:

G3: Genes|*Genomes*|*Genetics* publishes high-quality, valuable findings, regardless of perceived impact. G3 publishes research that generates useful genetic and genomic information such as genome maps, single gene studies, QTL studies, mutant screens and advances in methods and technology, novel mutant collections, genome-wide association studies (GWAS) including gene expression, SNP and CNV studies; exome sequences related to a specific disease but lacking functional follow-up, personal exome and genome sequencing case, disease and population reports, and more.

Conceived by the Genetics Society of America, with its first issue published June 2011, *G3* is fully open access. *G3* uses a Creative Commons license that allows the most free use of the data, which anyone can download, analyze, mine and reuse, provided that the authors of the article receive credit. GSA believes that rapid dissemination of useful data is the necessary foundation for analysis that leads to mechanistic insights. It is our hope is that this strategy will spawn new discovery.

ABOUT GSA:

Founded in 1931, the Genetics Society of America (GSA) is the professional membership organization for scientific researchers, educators, bioengineers, bioinformaticians and others interested in the field of genetics. Its nearly 5,000 members work to advance knowledge in the basic mechanisms of inheritance, from the molecular to the population level. The GSA is dedicated to promoting research in genetics and to facilitating communication among geneticists worldwide through its conferences, including the biennial conference on "Model Organisms to Human Biology," an interdisciplinary meeting on current and cutting edge topics in genetics research, as well as annual and biennial meetings that focus on the genetics of particular organisms, including C. elegans, Drosophila, fungi, mice, yeast, and zebrafish. GSA publishes *GENETICS*, a leading journal in the field since 1916, and *G3: Genes|Genomes|Genetics*, an open-access journal launched in 2011. For more information about GSA, please visit www.genetics-gsa.org. Also follow GSA on Facebook at facebook.com/GeneticsGSA and on Twitter @GeneticsGSA.

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