



Special Issue Editors' Introduction: "Genomics and the Human Genome Project"

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This special issue of the *Journal of the History of Biology* on "Genomics and the Human Genome Project" emanates from an April 2015 workshop held at the U.S. National Institutes of Health (NIH) entitled "Capturing the History of Genomics." That workshop was organized by the National Human Genome Research Institute's (NHGRI's) History of Genomics Program, which was established in 2012 to ensure that the Institute's assets relevant to the history of genomics and the Human Genome Project are appropriately preserved, described, and made available for scholarly work.

NHGRI increased its attention to the systematic archiving of documents and other materials of historic value following the departure of Dr. Francis Collins, who served as the Institute's Director from 1993 to 2008. As the NHGRI Director, Collins was on the front line of the early development of the field of genomics, including key leadership roles in the Human Genome Project and multiple landmark genomics efforts that followed. As such, Collins's notes, files, and other collected materials contain an inordinate amount of information about the management and execution of the Human Genome Project and other historically significant genomics initiatives, including the role that NHGRI and other key partners (in the United States and abroad) played in funding and guiding these efforts. Many of Collins's docu-

ments illustrate the important roles that the Institute played in facilitating the growth and maturation of the field of genomics as a whole, especially those related to the immediate efforts to capitalize on the reference human genome sequence generated by the Human Genome Project.

The initial steps to assimilate and organize Collins's and other NHGRI materials of historic interest revealed the need for a larger and more structured effort, leading to an expansion of the NHGRI History of Genomics Program in 2014. Charged with capturing, cataloging, and making accessible the rich archival resources held by the Institute that pertained to the history of genomics, the new Program's immediate priorities included preserving the large number of key documents and capturing the views and experiences of NHGRI staff and members of the genomics community through oral histories.

To augment the intense document-digitization and asset-curation efforts as well as the ongoing oral histories, the History of Genomics Program convened a workshop in 2015 that sought feedback about its programmatic emphases, digitization routines, descriptions of archival resources, oral histories, and development of database tools for scholarly collaboration. Additional purposes of the 2015 "Capturing the History of Genomics" workshop included further introducing the scholarly community to the growing archival holdings at NHGRI; fostering future collaborations between the History of Genomics Program and members of that community, particularly in the field of science studies; and seeking input from outside experts about future goals and directions for the then-young Program. Over two days, the assembled scholars and NHGRI staff vigorously discussed the historical, philosophical, and sociological aspects of the Human Genome Project and subsequent major genomics programs and the value of capturing historical details about those efforts.

The workshop included a presentation summarizing the progress of the History of Genomics Program in digitizing, cataloguing, and describing NHGRI's historical holdings from 2012 to 2015. Plans for developing a database to house these holdings were also outlined. The resulting input was helpful for making that database into an archival resource that a sizable and ever-growing community of scholars now use for examining digital files in the NHGRI archive. The workshop also initiated the planning of this special issue of the *Journal of the History of Biology* with then Editor-in-Chief Michael Dietrich.

Reflecting the many discussions at the workshop, the articles in this special issue represent the first body of scholarly work on genomics that

directly draws from the History of Genomics Program's organized archives and oral histories. The articles that follow illustrate the breadth and depth of NHGRI's holdings and of the scientifically, historically, and sociologically relevant issues from the "prehistory" of the Human Genome Project to the ethical and normative consequences of genomics being experienced today. The topics cover a range of pathbreaking efforts in bioinformatics and database development, human genomic variation research, genome technology development, and the profoundly important commitment to open science and free access to genomic data, as exemplified by the "Bermuda Principles" governing genomic data sharing.

In the article "Whose Turn? Chromosome Research and the Study of the Human Genome," Soraya de Chadarevian outlines the importance of historic archives for understanding the roles of hereto underacknowledged researchers and communities in the history of modern biology, including those related to the Human Genome Project. She underscores that such archives, including those held by NHGRI, actually complicate the neat narrative that seamlessly and unproblematically conjoins the discovery of DNA and the generation of the human genome sequence by the Human Genome Project. Chadarevian contends that it is as important to examine the role and contributions of human cytogeneticists in their postwar study of human chromosomes as well as the contributions of molecular biologists to understand the genesis of the tools, methods, and approaches that were instrumental in the Human Genome Project.

By examining the prehistory of the practices, knowledge, and approaches that guided the Human Genome Project, Hallam Stevens describes, in his article "Globalizing Genomics: The Origins of the International Nucleotide Sequence Database Collaboration" how the global dissemination of genome-sequence information predated the Human Genome Project. Focusing on the institutions and practices that allowed genome-sequence data to become globally valuable objects, Stevens traces the long collaborative history among three key databases: GenBank in the United States, the European Molecular Biology Laboratory Databank (EMBL-Bank), and the DNA Database of Japan (DDBJ).

In "More than Moore's Mores: Computers, Genomics, and the Embrace of Innovation," Joseph November discusses how genomics researchers frequently cite "Moore's law"—the idea that computer-processing capacity doubles every 18–24 months—to illustrate the extraordinary advances in DNA-sequencing technologies over the last

quarter century. He argues that NHGRI's references to "Moore's Law" reflect not only the genomics community's appreciation of progress and innovation, but also that of the public. November also cautions that while such an emphasis on progress in DNA-sequencing technology has been good for public relations, it has masked some of the other major innovations from the Human Genome Project and other genomics efforts.

Providing the first in-depth analysis of the story behind the development of the 1996 "Bermuda Principles," which mandated that DNA-sequence data being generated by the Human Genome Project be released daily into the public domain, Kathryn Maxon Jones, Robert Cook-Deegan, and Rachel Ankeny exhaustively detail how such an expansive and, as the authors underscore, "radical" policy became formulated, established, and standardized in "The Bermuda Triangle." Drawing from extensive archival resources as well as oral histories and semi-structured interviews, the authors chart the genesis of one of the Human Genome Project's signature legacies.

Finally, moving beyond the Human Genome Project proper and examining the societal impact of genomic advances through a different lens in "Variations on a Chip: Technologies of Difference in Human Genetics Research," Ramya Rajagopalan and Joan Fujimura describe how microarray technologies have been used to catalog human genomic differences. The authors underscore that interest in individual and group genomic variation has been one of the central areas of interest for 20th century genetics and biology, and they outline the consequential focus in the late 20th century on single-nucleotide polymorphisms (SNPs). They contend that through the focus on these genomic markers and the use of microarray-based technologies, researchers have made specific and heretofore under-examined claims about the nature of individual and group differences at the genomic level.

Now responsible for an enormous treasure trove of archival materials, the NHGRI History of Genomics collection promises to begin a fundamental reorientation of scholarly work on the rapidly expanding field of genomics, now in its fourth decade. Towards that end, we hope this special issue will further promote engagement of the science studies community with NHGRI and the Institute's History of Genomics Program. In addition to focusing on an important topic, each of the contributions in this issue also speaks to many social, historical, and ethical issues in 20th century science and technology, while also illuminating the complex roots of many contemporary scientific practices. Consistently at the forefront of genomics, NHGRI helped to shape the

Human Genome Project and, more recently, has catalyzed the growth and dissemination of genomics. We firmly believe that this special issue continues this tradition by leveraging the historical resources of the Institute, answering important questions, and encouraging new scholarly directions for historical studies.