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SCOPE NOTE

Human Genome Project

17

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This annotated bibliography with an introductory essay was first published in the *Kennedy Institute of Ethics Journal*, Vol. 1, No. 4, pp. 347-362, December 1991. It has been updated by Bioethics Research Library at Georgetown staff on a periodic basis through July 2011. These citations provide a representative sample of the literature on the Human Genome Project as an aid for students and researchers who are beginning to explore the topic. Search strategies for numerous databases are supplied at the end of this bibliography to support comprehensive research.

Introduction

The Human Genome Project (HGP), officially spanning the years 1990 to 2003, was an [international](#) research effort to determine the sequence of the three billion bases of DNA that constitute our [blueprint](#)—the human genome. Coordinated by the U.S. National Institutes of Health and the Department of Energy, scientists from China, France, Germany, Japan, the United Kingdom and the United States deposited DNA sequence data in public databases posted on the Internet not only for their own collaborative efforts but also for use by other scientists and the public (I. [Quackenbush 2011](#)). Beyond developing comprehensive genetic databases, the HGP fostered a sustained effort to improve the techniques used to map and sequence genetic material, thereby training a whole generation of scientists in a short period of time and reducing the costs of genetic research (I. [McElheny 2010](#)). While different in scope from previous biomedical research, the Human Genome Project will continue to "...yield a harvest of information that will drive the research enterprise for at least the next 100 years" (IV. [Collins 1991](#)).

History

Unlike “top-down” government-directed endeavors such as the atomic bomb and space programs, the Human Genome Project has been characterized as a grassroots initiative developed and managed by scientists (I. [McElheny 2010](#)). From 1984 through 1986, a series of scientific workshops and conferences doubled as “strategic planning” sessions for this genome project. These opportunities for deliberation and argumentation took place in Alta, Utah (December, 1984); Santa Cruz, California (May, 1985); Santa Fe, New Mexico (March 3-4, 1986); and Cold Spring Harbor, New York (June, 1986). Numerous surveys of this period (I. [Cook-Deegan 1994](#); [McElheny 2010](#); [Shreeve 2004](#); [Sulston 2002](#); [Wickelgren 2002](#)) detail this process which was “controversial from the start” (I. [Roberts 2001](#)). Scientists not only voiced opinions about the scientific merits of various mapping and sequencing techniques, but also expressed ideas about the most advantageous business plans. Participants tackled such issues as whether “small science” projects would be subsumed by the “big science” Human Genome Project; whether genome mapping would ever provide real benefits in the clinic; and whether data derived from publicly-funded research should remain in the public sector.

These surveys of the Human Genome Project also trace the role of government funding in sponsoring research in the United States and around the world. Genome sequencing and mapping research was simultaneously occurring in Europe and Asia as the U.S. National Institutes of Health and Department of Energy began funding the Human Genome Project in the late 1980s. In 1998, J. Craig Venter founded a private company, Celera Genomics, to sequence the human genome "in three years," ahead of the schedule being followed by the publicly-funded teams (I. [Shreeve 2004](#)). Human genome research was at once deeply cooperative and intensely competitive, team-oriented and personality-driven. Exemplifying these contradictions, the announcement of the publication of the human genome draft sequences authored by more than 350 scientists working in over twenty genome centers was brokered by two public figures while they had beer and pizza in the basement of a mutual friend (II. [International Human Genome Sequencing Consortium 2001](#); II. [Venter et al. 2001](#); VIII. [Collins 2006](#)).

The human genome sequence and the Human Genome Project were declared complete on April 14, 2003, the [50th anniversary](#) of the discovery of DNA's structure. The celebration included the publication of a [blueprint](#) for future research projects (V. [Collins et al. 2003](#)). Examples of these sequencing and mapping projects include the [1000 Genomes Project](#), an international collaboration to develop a catalog of human genetic variation; the [Human Microbiome Project \(HMP\)](#), an attempt to understand the diversity of microbial cells that affect metabolism, drug interactions, and susceptibility to disease; and [The Cancer Genome Atlas \(TCGA\)](#), a joint project of the National Cancer Institute and the National Human Genome Research Institute.

To celebrate the 10th anniversary of mapping the human genome, the U.S. National Human Genome Research Institute (NHGRI) published a new [vision](#) for genomic medicine (V. [Green et al. 2011](#)). In addition to setting goals for increased public outreach and education, the document calls for research on the use of genomic information to reduce health disparities and to lower health care costs.

Ethical, Legal and Social Implications (ELSI) of the Human Genome Project

As a “Big Science” initiative, the Human Genome Project (HGP) was associated in the minds of many with the Manhattan Project which developed the atomic bomb. The secrecy required by this military program “...permanently altered the public's unquestioning trust in science and scientists.” (VI. [Wexler 2003](#)). To address these concerns, the Ethical, Legal and Social Implications (ELSI) Joint Working Group of NIH's new Human Genome Institute was formed in 1989 as the first act of James Watson, the Institute's first Director. The ELSI Working Group was allotted 3% of the genome funding (I. [Cook-Deegan 1994](#)). Tensions immediately developed between the “hard science” geneticists and the “soft science” ELSI members, even though many members of the ELSI working group were scientists. In addition to criticism from within the HGP, ELSI working group members found that colleagues in the community felt that they were “...being used as a front to give the illusion that scientists were being responsible” (VI. [Beckwith 2002](#)). Despite criticism, the joint working group established task forces on genetic testing and insurance/employment discrimination; stigmatization; privacy; genetic counseling; genetics and reproduction; genetics and clinical practice; historical analysis of past misuses of genetics; and the influence of commercialization of genetic testing (VI. [Beckwith 2002](#)). In 2004, the U.S. National Human Genome Research Institute created [Centers for Excellence in ELSI Research](#) (CEERs). Current funding for ELSI research is coordinated by these Centers, and teams of researchers focus on complex topics in genomic research.

Concerns with ELSI issues were not limited to the United States. When HUGO, the Human Genome Organization, was formed in 1988 to direct international collaboration in genomic research, one of its founding purposes was to provide guidance on ELSI issues to genome projects around the world (III. [Bodmer 1991](#)). The goal of creating a truly [global approach](#) to ELSI issues continues to be a top priority as researchers anticipate the future challenges raised by genomic research that spans continents and populations (VI. [Kay 2011](#)).

As research published by ELSI grantees gained an audience, other areas of biomedical research, such as neurological enhancement and nanotechnology, began to address ELSI issues as well (VI. [Greely 2006](#)). Current genomic research programs such as the [Human Microbiome Project](#) have a special ELSI task force to address the unique issues raised about informed consent and privacy (VI. [Achenbaum et al. 2011](#)).

The decades of ELSI research helped set the stage for passage of the [Genetic Information Nondiscrimination Act \(GINA\)](#) in May, 2008. As the top item on the U.S. Department of Energy's Human Genome Project [timeline](#), which includes entries for historic meetings and landmark scientific articles, GINA is recognized as the first law of its kind to preemptively prohibit health insurers or employers from denying coverage or employment on the basis of genetic information.

Ten years after the Human Genome Project has been declared complete, improvements in “genetic medicine” — using knowledge about single genes to improve treatment of single-gene disorders — have paved the way for advances in “genomic medicine,” attempts to understand the interactions between genomic and nongenomic factors for the development of “...new diagnostic and therapeutic approaches to common multifactorial conditions” (V. [Feero et al 2010](#)). Open access to the genome databases “...adopted by the Human Genome Project in 1996 and now the norm for other community resource projects, empowers the best brains on the planet to begin work immediately in analyzing the massive amounts of genomic data now being produced” (V. [Collins 2010](#)). This analysis is accompanied by “...a continuing sense of wonder, a continuing need for urgency, a continuing desire to balance ambition with reality, and a continuing responsibility to protect individuals while maximizing the societal benefits of genomics” (V. [Green et al. 2011](#)).

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I. GENERAL SURVEYS

Cook-Deegan, Robert. [The Gene Wars: Science, Politics, and the Human Genome](#). New York: W.W. Norton & Company, 1994. 416 p.

As the author of several influential reports on genome research in the late 1980s, Cook-Deegan provides in rich detail a chronicle of the Human Genome Project up to Francis Collins' acceptance of its NIH directorship in 1993. The book is divided into five sections: the scientific aspects of genome mapping; the “early years” of genome research funded through the Department of Energy; the development of an infrastructure to coordinate individual “gene hunts” through the efforts of the Howard Hughes Medical Institute, the National Academy of Sciences, and the National Institutes of Health;

international partners in genome research; and the public effort to address the ethical, legal, and social implications (ELSI) of mapping the genome

Cooper, Necia Grant, ed. *The Human Genome Project: Deciphering the Blueprint of Heredity*. Mill Valley, CA: University Science Books, 1994. 360 p.

This collection focuses on the scientific concepts involved in constructing gene maps: the classification structure for DNA sequences, the construction of copy DNAs (cDNAs), polymerase chain reactions (PCRs) and sequence-tagged sites, and single-molecule spectroscopy (used for rapid DNA sequencing). Also included are discussions of the history of genetic research, the future of genomics, and electronic publishing of sequence data.

Crow, James F. and Dove, William F., eds. *Perspectives on Genetics: Anecdotal, Historical, and Critical Commentaries, 1987-1998*. Madison: University of Wisconsin Press, 2000. 723 p.

Editors of the longstanding column "Perspectives" in *Genetics*, the journal of the Genetics Society of America, Crow and Dove have culled over 100 columns to produce a cumulative history of genetics research and genome mapping in the United States.

Davies, Kevin. *Cracking the Genome: Inside the Race to Unlock Human DNA*. New York: Free Press, 2001. 310 p.

Davies, the editor of *Nature Genetics*, closely followed the ongoing Human Genome Project for 10 years. He details the finances, the scientific developments, and the "key players" involved in mapping the human genome. Interviews with Francis Collins and J. Craig Venter are included.

Drlica, Karl A. *Double-Edged Sword: The Promises and Risks of the Genetic Revolution*. New York: Addison-Wesley, 1994. 242 p.

The author describes numerous facets of the "genetic revolution", including the forensic use of genetic analysis and DNA tracking for infectious disease control. Drlica pairs a personal case history with each scientific discussion to provide a comprehensive overview of genetic technology.

Gee, Henry. *Jacob's Ladder*. New York: W.W. Norton & Company, 2004. 272 p.

Gee explores the roots of genetic research from antiquity to the present because he believes that "...our view of the long history of biology has been clouded and distorted by the titanic presence of Charles Darwin." The author discusses the works of Johann Wolfgang Goethe and William Bateson, among others, in an attempt to expand the discourse on biology, evolution, and genetics.

Gert, Bernard; Berger, Edward M.; Cahill, George F.; Clouser, K. Danner; Culver, Charles M.; Moeschler, John B.; and Singer, George H.S. *Morality and the New Genetics: A Guide for Students and Health Care Providers*. Sudbury, MA: Jones and Bartlett Publishers, 1996. 242 p.

This textbook is the product of a three-year collaboration by ethicists, scientists, and medical professionals at Dartmouth. It includes an historical overview of genome research, a critique of principlism in the ethical analysis of genetic issues, and a discussion of the psychosocial aspects of "genetic malady".

Keller, Evelyn Fox; and Lloyd, Elisabeth A., eds. *Keywords in Evolutionary Biology*. Cambridge: Harvard University Press, 1992. 414 p.

Containing essays that trace the etymological development of words and expressions such as "gene," "genetic load," "epistasis," "genotype", "progress," "random drift," and "fitness", the editors' stated goal is "...to identify and explicate those terms in evolutionary biology that, though commonly used, are plagued in their usage by multiple concurrent and historically varying meanings."

McElheny, Victor K. *Drawing the Map of Life: Inside the Human Genome Project*. Philadelphia, PA: Basic Books, 2010. 360 p.

Written for both researchers and the general public, this history of the Human Genome Project discusses the scientific and organizational hurdles that were overcome in order to map the genome. Beginning in 1968 with a description of Hamilton O. Smith's experiments using restriction enzymes as "site-specific" scissors for DNA, McElheny describes the scientific discoveries that set the stage for genome mapping and the series of scientific meetings held between 1984 and 1986 that helped develop a consensus among scientists about the direction of the project.

Murray, Thomas H.; Rothstein, Mark A.; and Murray, Robert F., eds. *The Human Genome Project and the Future of Health Care*. Bloomington, IN: Indiana University Press, 1996. 248 p.

This collection of essays addresses the potential influence of genome research on access to health care. Chapters focus on such topics as cost-effectiveness in primary care, insurance coverage and discrimination, and equity in organ allocation.

Quackenbush, John. *The Human Genome: The Book of Essential Knowledge*. New York: Imagine, 2011. 189 p.

After providing a brief history of the Human Genome Project (HGP), the author traces the history of molecular biology from Mendel's experiments to the sequencing of the fruit fly. He synthesizes population genetics, personalized medicine, systems biology, and stem cell research, and discusses the future of genomic medicine.

Roberts, Leslie. [Controversial from the Start](https://doi.org/10.1126/science.291.5507.1182a). *Science* 291(5507): 1182-1188, February 16, 2001. doi:10.1126/science.291.5507.1182a

Roberts chronicles the “...hotly contested-- and contentious” race to map the human genome. Focusing on many of the scientists who helped to develop a consensus early on - - Robert Sinsheimer, Walter Gilbert, Charles DeLisi, and James Wyngaarden -- the author describes the competition that later developed between the public sector researchers (twenty centers coordinated by Francis Collins at NIH) and the private sector (a team led by J. Craig Venter of Celera Genomics.)

Shreeve, James. *The Genome War: How Craig Venter Tried to Capture the Code of Life and Save the World*. New York: Alfred A. Knopf, 2004. 403 p.

Shreeve provides a detailed description of Celera Genomics, from its founding in May, 1998 by J. Craig Venter to Venter's resignation from the company in January, 2002. He tracks Celera's progress in sequencing the human genome, describes the interaction between Venter and Francis Collins at NIH's genome center, and provides many anecdotes and opinions about the Human Genome Project from prominent scientists involved in the research.

Sulston, John, and Ferry, Georgina. *The Common Thread: A Story of Science, Politics, Ethics and the Human Genome*. London/New York: Bantam Press, 2002. 310 p.

Nobel Laureate Sulston, head of the Sangar Centre in Cambridge (U.K.), describes the scientific and political controversies of the Human Genome Project from a British perspective. Calling it a “three-ring circus” from the start, he describes the skepticism expressed by fellow scientists when he transitioned from sequencing the nematode worm to the human genome. While the “razzmatazz” accompanying the announcement of the completed draft of the human genome was appropriate, understanding the implications for health and well-being “...will take decades and will encompass all of biology.”

Watson, James D. *A Passion for DNA: Genes, Genomes, and Society*. Cold Spring Harbor, NY: Cold Spring Harbor Laboratory Press, 2000. 250 p.

These 26 essays by Nobel Laureate Watson, many of them written as the introduction for the Cold Spring Harbor Laboratory's annual reports, span 30 years of scientific discoveries. The book is divided into five sections: Autobiographical Flights, Recombinant DNA Controversies, Ethos of Science, War on Cancer, and Societal Implications of the Human Genome Project.

Wickelgren, Ingrid. *The Gene Masters: How a New Breed of Scientific Entrepreneurs Raced for the Biggest Prize in Biology*. New York: Henry Holt and Company, 2002. 291 p.

A journalist covering scientific developments for numerous publications including *The New York Times* and *Science*, Wickelgren tells the story of the Human Genome Project from the vantage points of William Haseltine, a Harvard professor who founded the biopharmaceutical corporation Human Genome Sciences; “cowboy scientist” J. Craig Venter of Celera Genomics; Francis Collins, the first director of the U.S. National Institute of Health's genome institute; and Kari Stefansson, co-founder of deCODE Genetics, the Icelandic genome population database project. She pays special attention to

the formative role played by the media in moving the Human Genome Project forward.

II. Landmark Scientific Articles

This list contains citations to articles frequently referenced in the general literature on genome sequencing. A [comprehensive listing](#) of landmark articles on Human Genome Project milestones is maintained on the U.S. Department of Energy's [Human Genome Project Information](#) site.

Adams, Mark, D.; Kelley, Jenny M.; Gocayne, Jeannine D.; Dubnick, Mark; Polymeropoulos, Mihael H.; Xiao, Hong; Merril, Carl R.; Wu, Andrew; Olde, Bjorn; Moreno, Ruben F.; Kerlavage, Anthony R.; McCombie, W. Richard; and Venter, J. Craig. [Complementary DNA Sequencing: Expressed Sequence Tags and Human Genome Project](#). *Science* 252(5013):1651-6, June 21, 1991.

An automated process for partially sequencing human brain cDNA generated expressed sequence tags (ESTs) representing 337 new genes. Faster and cheaper than complete genomic sequencing, the authors predict that the use of ESTs will facilitate the tagging of all human genes within a few years.

Botstein, David; White, Raymond L.; Skolnick, Mark; and Davis, Ronald W. [Construction of a Genetic Linkage Map in Man Using Restriction Fragment Length Polymorphisms](#). *American Journal of Human Genetics* 32:314-31, May 1980.

The authors propose a new method for finding and organizing markers on human chromosomes. They predict that this form of mapping will delineate pedigrees that can be used for genetic counseling.

Deloukas, Panos; Schuler, Gregory D.; Gyapay, Gabor; Beasley, Ellen M.; Soderlund, Carol; Rodriguez-Tomé, Patricia, et al. [A Physical Map of 30,000 Human Genes](#). *Science*. 282(5389):744-6, October 23, 1998. doi:10.1126/science.282.5389.744

Known as GeneMap'98, this map contained nearly twice the genes as the previous map (1996) and brought the Human Genome Project almost halfway to mapping the entire human genome. The electronic data supplement for this map can be found at: <http://www.ncbi.nlm.nih.gov/genemap99/>

Dib, Colette; Faure, Sabine; Fizames, Cecile; Samson, Delphine; Drouot, Nathalie; Vignal, Alain; et al. [A Comprehensive Genetic Map of the Human Genome Based on 5,264 Microsatellites](#). *Nature* 380: A1-A138, March 14, 1996. doi:10.1038/380152a0

This special issue presents the final version of the Généthon human genetic linkage map on pages A1-A138.

Hudson, Thomas J.; Stein, Lincoln D.; Gerety, Sebastian S.; Ma, Junli; Castle, Andrew B.; Sliva, James, et al. [An STS-based Map of the Human Genome](#). *Science* 270(5244):1945-54, December 22, 1995. doi:10.1126/science.270.5244.1945

The authors proclaim that this map of sequence-tagged sites (STSs) is the first step toward constructing a transcript map of the human genome, which in turn will function as scaffolding for a large-scale sequencing of the human genome.

[The Human Genome](#). *Science* [Special Issue] 291 (5507) February 16, 2001, 1145-1434.

To celebrate the announcement of the "working draft" of the human genome sequence, *Science* magazine dedicated an issue to the various aspects of the project and made it available online at no charge. Sections provide a timeline for the sequencing of various chromosomes, an overview of the process, and instructions for accessing the human genome sequence data maintained in the public domain.

International Human Genome Sequencing Consortium. [Initial Sequencing and Analysis of the Human Genome](#). *Nature* 409(6822):860-921, February 15, 2001. doi:10.1038/35057062

This report on the draft sequencing of 94% of the human genome was compiled by the 20 collaborating organizations from China, France, Germany, Japan, the United Kingdom and the United States. Over 350 individual authors are listed by sequencing center. They describe the process involved in creating the draft sequence, as well as the three-step program needed to finish the map.

Lander, Eric S., and Botstein, David. [Homozygosity Mapping: A Way to Map Human Recessive Traits with the DNA of Inbred Children](#). *Science* 236(4808):1567-70, June 19, 1987.

The authors report on a strategy utilizing mapped restriction fragment length polymorphisms (RFLPs) to map recessive disease genes in children from consanguineous marriages. The method is considered efficient because a RFLP linkage map can be completed using data from less than a dozen children.

Lennon, Greg; Auffray, Charles; Polymeropoulos, Michael; and Bento Soares, Marcelo. [The I.M.A.G.E. Consortium: An Integrated Molecular Analysis of Genomes and Their Expression](#). *Genomics* 33(1):151-2, April 1, 1996. doi:10.1006/geno.1996.0177

The I.M.A.G.E. (Integrated Molecular Analysis of Genome Expression) Consortium was formed in November, 1993, to produce public domain databases of data derived from mapping complementary DNA (cDNA) clones so that scientists could share their work with the least redundancy. The authors review the guidelines for their project, the procedure for adding data to the database, and the composition of the clones from 20 different human cDNA libraries.

McKusick, Victor A. [Current Trends in Mapping Human Genes](#). *FASEB Journal* 5(1):12-20, January 1991.

McKusick, a longstanding expert in the field of genetic disorders and the author of [Mendelian Inheritance in Man](#), presents an historical overview and summary of the status of the human gene mapping. He also explains the methods by which scientists access genetic data and emphasizes the importance of genetic mapping in biology and medicine.

NIH/CEPH Collaborative Mapping Group. [A Comprehensive Genetic Linkage Map of the Human Genome](#). *Science* 258(5079):67-86, October 2, 1992. doi: 10.1126/science.1439770

This international collaboration between scientists at the National Center for Human Genome Research (U.S. National Institutes of Health, Bethesda, Maryland) and the Centre d'Etude du Polymorphisme Humain (CEPH, Paris, France) produced a map for determining disease phenotypes "...even for those [conditions] with limited pedigree resources."

[Omics Gateway](#)

Nature magazine augments the online version of its issue dedicated to the mapping of the human genome (Nature 409 (6822) February 15, 2001) with an archive of research papers on all aspects of genome mapping, a section of news items on developments in the field, and a module devoted to post-genomics (technical discussions of techniques used in genetic research).

Risch, Neil, and Merikangas, Kathleen. [The Future of Genetic Studies of Complex Human Diseases](#). *Science* 273(5281):1516-7, September 13, 1996.

The authors review the statistical requirements and technological strategies needed to undertake genome-wide association studies (GWAS).

Venter, J. Craig; Adams, Mark D.; Myers, Eugene W.; Li, Peter W.; Mural, Richard J.; Sutton, Granger G. et al. [The Sequence of the Human Genome](#). *Science* 291(5507):1304-51, February 16, 2001. doi: 10.1126/science.1058040

Using a whole-genome random shotgun method, the authors describe the creation of a draft sequence of the euchromatic portion of the human genome.

Watson, James D., and Crick, Francis H.C. [Molecular Structure of Nucleic Acids: A Structure for Deoxyribose Nucleic Acid](#). *Nature* 171(4356):737-8, April 25, 1953. doi:10.1038/171737a0

The completion of the Human Genome Project was celebrated at a [scientific symposium](#) held on the 50th anniversary of this landmark publication on the double helix structure of DNA.

Wolfsberg, Tyra G.; Wetterstrand, Kris A.; Guyer, Mark S.; Collins, Francis S.; and Baxevanis, Andreas D. [User's Guide to the Human Genome](#). *Nature Genetics*, September 2002: 32 (Supplement), pp. 1-79.

This supplement is "...an elementary hands-on guide for browsing and analyzing data produced by the International Human Genome Sequencing Consortium." The guide is designed as a workbook geared to answer such questions as "how do you find a specific gene?"

III. The Early Years - 1984-1989

Barnhart, Benjamin J. **The Department of Energy (DOE) Human Genome Initiative**. *Genomics* 5(3): 657-60, October 1989.

DOE's interest in genome mapping developed from its study of genetic damage to the hibakusha ("those affected by the bomb"), survivors of the Hiroshima and Nagasaki bombings. This paper traces discusses DOE's sponsorship of the Santa Fe meeting in 1986, and the subsequent reports encouraging genome research. Also provided is an outline of aims and objectives, communications at various levels, budgets, and expected benefits.

Bitensky, Mark W., Los Alamos National Laboratory. [Letter to Charles DeLisi, Office of Health and Environmental Research, United States Department of Energy](#). Unpublished document: Robert M. Cook-Deegan Human Genome Archive, Bioethics Research Library, April 2, 1986. 5 p.

This letter is a comprehensive report on a genome sequencing workshop sponsored by the Department of Energy's Office of Health and Environmental Research in Santa Fe, New Mexico, on March 3-4, 1986. Characterized as an event of "impassioned esprit," the workshop participants discussed scientific techniques, business models, and funding strategies for the proposed genome project. The involvement of colleagues from Europe and Asia was recommended.

Bodmer, Walter F. [HUGO: The Human Genome Organization](#). *FASEB (Federation of American Societies of Experimental Biology) Journal* 5(1): 73-74, January 1991.

Bodmer, the first president of HUGO, describes its founding in 1989 and provides a description of each HUGO committee, including the Ethical, Legal and Social Issues committee chaired by Victor McKusick.

Cook-Deegan, Robert M. **The Human Genome Project: The Formation of Federal Policies in the United States, 1986-1990. (With commentaries by Paul Berg and Ernest May.)** *In: Biomedical Politics*, pp. 99-175. Edited by Kathi E. Hanna. Washington, DC: National Academy Press, 1991.

Based on interviews, planning documents, and literature reviews, Cook-Deegan surveys the process by which the genome project was conceived, formulated, and approved at

various levels in federal science agencies.

Cook-Deegan, Robert M. [Wizards of the Information Age](#). In his *Gene Wars: Science, Politics, and the Human Genome*. New York: W.W. Norton & Company, 2004, pp. 283-289.

Cook-Deegan chronicles the development of the Genome Database "...for gene mappers...[which] contained information on genetic linkage maps and physical maps" and GenBank which stored information about DNA sequence. These databases pooled data from across genome centers, and "...spawned a new field [where] [c]omputers and mathematical techniques were turned loose on the data to construct theories of biological structure and function." Genome databases continue to be available on the [site](#) of the U.S. National Institute of Health's National Center for Biotechnology Information.

Culliton, Barbara J. **Mapping Terra Incognita (Humani Corporis)**. *Science* 250(4978): 210-12, 12 October 1990.

Culliton provides a brief panorama of the history of genetic mapping, from the first description of the inheritance of color blindness by Horner in 1876, through improvements in methods and techniques, to current approaches.

DeLisi, Charles. **The Human Genome Project**. *American Scientist* 76(5): 488-93, September/October 1988.

DeLisi, director of the DOE's Health and Environmental Research programs during the initiation of the genome project, describes events in the early history of the DOE genome effort. He also provides a general overview including a discussion of the expected benefits, "big science" projects, and a description of mapping techniques.

Gilbert, Walter. [Letter to Robert Edgar, Thimann Labs, University of California, Santa Cruz](#). Unpublished document: Robert M. Cook-Deegan Human Genome Archive, Bioethics Research Library; May 27, 1985; 3 p.

After attending the Santa Cruz proceedings, Nobel Laureate Walter Gilbert sent a letter detailing his ideas for a genome institute to molecular biologist Robert Edgar. Gilbert not only presented his vision for such an institute, but also included suggestions about such practical matters as staff size, space allocation, and potential funding sources. Gilbert envisioned a timetable for sequencing the genome and touted the benefits of such a project for mankind. Robert L. Sinsheimer is copied on the letter.

National Research Council. [Mapping and Sequencing the Human Genome](#). Washington: National Academy Press, 1988. 116 p.

This landmark report gave momentum to the genome project. After outlining its goals and recommendations, it focuses on the issues of genome mapping, sequencing, handling of materials and information, and strategies for implementation and management. Also provided are helpful descriptions of the genome program and definitions of basic genetic concepts.

Roberts, Leslie. **Carving Up the Human Genome**. *Science* 242(4883): 1244-46, December 2, 1988.

Roberts discusses the "Valencia meeting," and the agreement of the participants that the genome project is not a U.S. monopoly, but a project involving everyone. A brief overview is given of genome activities in various countries around the world.

Sinsheimer, Robert L. **The Santa Cruz Workshop, May 1985**. *Genomics* 5(4): 954-56, November 1989.

Robert Sinsheimer, former chancellor of the University of California at Santa Cruz, outlines the thoughts and events that led to the first discussions on sequencing the human genome, and provides an overview of the workshop.

United States. Congress. Office of Technology Assessment (OTA). **Mapping Our Genes: Genome Projects: How Big, How Fast?** Washington: U.S. Government Printing Office, 1988. 218 p.

This second major report, released in 1988, was written in response to U.S. Congressional concerns. The report offers a comprehensive analysis of the issues pertaining to the U.S. genome project—especially its organization and funding requirements— in an effort to present Congress with options for U.S. involvement and direction in the project.

United States. Department of Energy (DOE). **Human Genome 1989-90 (Program Report) March 1990**. Springfield, VA: National Technical Information Service, March 1990. 157 p.

This document is a status report on DOE's Human Genome Program, and includes a background to the initiative and the vision for the next 15 years. Also provided is a timeline of DOE's program development, an overview of management issues, research highlights, and abstracts of current research.

United States. Department of Energy (DOE). Health and Environmental Research Advisory Committee. Subcommittee on the Human Genome. **Report on the Human Genome Initiative for the Office of Health and Environmental Research ("HERAC Report")**. Germantown, MD: Department of Energy, April 1987.

A specially convened subcommittee on the human genome proposal prepared this report for DOE outlining the advantages of sequencing the human genome, and recommending that DOE move rapidly to fund and administer the program.

United States. Department of Energy (DOE). Office of Health and Environmental Research. **Sequencing the Human Genome. Summary Report of the Santa Fe Workshop, March 3-4, 1986**. Los Alamos, NM: Los Alamos National Laboratory, 1986.

Sponsored by DOE, this international workshop assessed the technical feasibility of sequencing the human genome, its costs and its benefits. Recommendations from this meeting fueled interest and action on a DOE human genome project.

Watson, James D.; and Jordan, E. **The Human Genome Program at the National Institutes of Health.** *Genomics* 5(3): 654-56, October 1989.

Watson and Jordan briefly describe the history of the genome program at NIH and the agency's plan to achieve the goals of the initiative. The paper covers the early discussions about the merit of the program and funding, the central coordination of genome programs, and the development of later committees, working groups, and related organizations.

IV. The Human Genome Project – 1990 - 2003

Adams, Mark D., and Venter, J. Craig. [Should Non-Peer-Reviewed Raw DNA Sequence Data Release Be Forced on the Scientific Community?](#) *Science* 274(5287): 534-536, October 25, 1996. doi: 10.1126/science.274.5287.534

While acknowledging that data sharing is an integral part of scientific research, the authors suggest that nightly uploads of "shotgun data" (unannotated DNA sequence data) to public Internet databases creates more problems than it solves. They discuss the merits of electronic publishing, and give examples of misunderstandings that have occurred with the use of non-peer reviewed published data.

Bentley, David R. [Genomic Sequence Information Should Be Released Immediately and Freely in the Public Domain.](#) *Science* 274(5287): 533-534, October 25, 1996. doi: 10.1126/science.274.5287.533

Bentley details the rationale behind the "Bermuda Statement" issued by those attending the first International Strategy Meeting on Human Genome Sequencing [See entry below under: Wellcome Trust]. Proclaiming that genome mapping data should be distributed freely, the author stresses that sharing minimizes duplication of effort by facilitating coordination, and that distributing data freely encourages scientific advances in a free-market economy.

Cantor, Charles. [Orchestrating the Human Genome Project.](#) *Science* 248(4951): 49-51, 6 April 1990. doi: 10.1126/science.2181666

Cantor, principal scientist for the Department of Energy's genome project contends that DOE and NIH are cooperating effectively to develop organizational structures and scientific priorities that will keep the project on schedule and within its budget. He notes that there will be small short-term costs to traditional biology, but that the long-term benefits will be immeasurable.

Centro de Direito Biomédico, Faculdade de Direito, Universidade de Coimbra, [and] Institut für Artz und Arzneimittelrecht Universität Göttingen. *Genome Analysis: Legal Rules - Practical Application.* Coimbra, Portugal: Livaria Almedina, 1994. 447 p.

This compilation of papers presented at a June, 1992 conference sponsored by the Commission of the European Communities at the University of Coimbra, Portugal, addresses legal issues raised by genome mapping. Presentations focus on privacy and patenting rights, health insurance and genetic counseling, forensic uses of DNA, and DNA data banks.

Collins, Francis S. [The Genome Project and Human Health](#). *FASEB (Federation of American Societies of Experimental Biology) Journal* 5(1): 77, January 1991.

Noting that the total cost of identifying the gene for Cystic Fibrosis was \$50 million, Collins maintains that the coordinated efforts of the Human Genome Project (HGP) will be much more cost-effective than a piecemeal approach to sequencing the genome. Recalling that "...the mandate for publicly funded biomedical research is to investigate the cause and treatment of human disease with the maximum intensity, efficiency, and creativity the research community can raise," he holds that the HGP is part of this tradition and predicts that "[i]t will yield a harvest of information that will drive the research enterprise for at least the next 100 years."

Collins, Francis; and Galas, David. [A New Five-Year Plan for the U.S. Human Genome Program](#). *Science* 262(5130): 43-46, October 1, 1993.

This article updates [Understanding Our Genetic Inheritance: The U.S. Human Genome Project: The First Five Years FY 1991-1995](#). Collins and Galas describe the technological improvements in DNA mapping that make it possible for the initial goals for the Human Genome Project to be redefined and extended to 1998. The authors also stress the importance of international collaborations in sequence mapping.

Collins, Francis S.; Morgan, Michael; and Patrinos, Aristides. [The Human Genome Project: Lessons from Large-scale Biology](#). *Science* 300(5617):286-90, April 11, 2003.

Beginning with the observation that "...[t]hinking big comes naturally to many biologists...[but] pursuing biological research on a monumental scale traditionally has not," the authors review the "big science" lessons learned from the Human Genome Project. They discuss the issues involved with keeping the project science-driven while meeting managerial goals, and acknowledge the importance of the five largest mapping centers (the "G5": Sanger Institute (U.K. Wellcome Trust), Joint Genome Institute in Walnut Creek, California (U.S. Department of Energy), and three U.S. National Institutes of Health centers: Baylor College of Medicine, Houston, Texas; Washington University at St. Louis, Missouri; and the Whitehead Institute, Cambridge, Massachusetts) in keeping the Human Genome Project on track.

Dulbecco, Renato. [Human Genome Project: Italian Contribution. Future Directions](#). *Journal of Cellular Physiology* 173(2):140-3, November, 1997. doi: 10.1002/(SICI)1097-4652(199711)173:2<140::AID-JCP11>3.0.CO;2-K

The author reviews Italy's genome mapping program which began in 1987 with a "modest budget," and continued to thrive despite several reorganizations given that "[b]ureaucracy is an Italian specialty." The work of sequencing teams in Milan, Trieste, Toniolo, and Pavia are detailed.

Genetics, Ethics and Human Values. Human Genome Mapping, Genetic Screening and Gene Therapy. Edited by Z. Bankowski and A.M. Capron. Geneva: CIOMS, 1991. 200 p.

This resource contains proceedings from the XXIVth Round Table Conference of the Council for International Organizations of Medical Sciences. The conference proceedings include views of experts from a variety of disciplines and cultures on genome mapping, genetic screening, and genetic therapy. Included also are reports of the various working groups and the [Inuyama Declaration](#).

Goodfellow, P.N. **Human Genome Project: Variation Is Now the Theme.** *Nature* 359(6398):777-8, October 29, 1992. doi:10.1038/359777a0

Goodfellow celebrates the new techniques of molecular biology that made it possible to produce a second-generation linkage map of the human genome, and marvels at the speed of progress made now that "...[m]ap construction is...big science, run by scientist-administrators and factory managers."

Great Britain. Parliament. House of Commons. Committee on Science and Technology. **Human Genetics: The Science and Its Consequences.** Volume I. Reports and Minutes of Proceedings. 145 p. Human Genetics. Volume II. Memoranda received up to January 31, 1995. 197 p. Human Genetics. Volume III. Memoranda received after January 31, 1995. 58 p.

The report describes the science of human genetics, the genome project and genetic research in the United Kingdom, medical applications of genetics, genetic science and industry, and human rights including discrimination, privacy, employment and insurance, ethical, legal and social issues, and international regulations of human genetics. There is a nine page summary of conclusions and recommendations (Vol. I, pp. xci-xcix); including the establishment of a Human Genetics Commission with statutory powers "to deal with screening and related matters."

Kaushik, Vijay, and Yudin, Boris. **Gene Therapy and the Human Genome Project in Russia.** *Eubios: Journal of Asian and International Bioethics* 7(1): January 1997, 6-7.

This article describes genome research in Russia since 1988, as well as surveys conducted by the Russian National Committee on Bioethics to gather information on ethical issues raised by this research.

Knoppers, Bartha Maria; and Chadwick, Ruth. **[The Human Genome Project: Under an International Ethical Microscope.](#)** *Science* 265(5181): September 30, 1994, 2035-2036. doi: 10.1126/science.8091225

The authors review the areas of "commonalty" that underpin regulation of genomic research: autonomy, privacy, justice, equity, and respect for human dignity. These normative international principles "...signify political will to do more than pay lip service to legitimate public concerns."

Lorentz, Cindy P.; Wieben, Eric D.; Tefferi, Ayalew; Whiteman, David A.; and Dewald, Gordon W. [Primer on Medical Genomics Part I: History of Genetics and Sequencing of the Human Genome](#). *Mayo Clinic Proceedings* 77(8):773-82, August 2002. doi: 10.4065/77.8.773

Starting with the ancient Babylonians, the authors set the stage for the Human Genome Project with a timeline from the history of genetics. They believe that knowing this background will help prepare scientists and the public to meet the challenges inherent in mapping the genome.

McLaren, Diane J. *Human Genome Research: a Review of European and International Contributions*. London: Medical Research Council, 1991. 140 p.

This report was undertaken at the request of the European Science Foundation and Academia Europaea, to provide them with information required to report on human genome research in Europe. A comprehensive review of international activity which surveys: genome research, countries and funding agencies with programs or planning programs, national strategies, future developments, and European contributions.

Megascience, the OECD Forum. *The Global Human Genome Programme*. Paris, France: Organisation for Economic Co-operation and Development, 1995. 75 p.

This report provides details on efforts by the Human Genome Organization (HUGO) and the Centre D'Etude du Polymorphisme Humain (CEPH) to facilitate international cooperation in genetic research. This description is complimented by overviews of the basic science and ethical issues involved in genome mapping.

National Institutes of Health (United States). National Human Genome Research Institute. *The Human Genome Project Progress Report: Fiscal Years 1995-1996*. Bethesda, MD: National Human Genome Research Institute, 1997. 132 p.

After providing background information on the Human Genome Project (HGP), this report goes on to cover progress made in mapping mouse, bacteria, drosophila, and human genomes. Appendices include HGP's policy on patenting DNA sequences, guidance on human subjects issues, and a funding history.

National Institutes of Health (United States). National Human Genome Research Institute. [NHGRI Workshop on DNA Sequence Validation](#) [April 15, 1996].

To prepare for large-scale mapping projects, the National Advisory Council for Human Genome Research met in January, 1996, to address issues of data integrity. Their report addresses validation issues such as which criteria should be used to determine if a cloned fragment represents genomic DNA.

[NCHGR Becomes NIH Institute](#). *Human Genome News* 8(3): 8, January 1997.

In order for genome research to operate under the same legislative authorities as other research being conducted at the National Institutes of Health (NIH), The National Center for Human Genome Research was granted institute status as of January, 1997, and

renamed the National Human Genome Research Institute (NHGRI). This reorganization also enables NHGRI's director to coordinate genome research with other projects at NIH.

Roberts, Leslie. **DOE's Genome Project Comes of Age**. *Science* 252(5005): 498-501, April 26, 1991. doi: 10.1126/science.2020849

The author describes how David Galas, then DOE's associate director of health and environmental research, turned around DOE's genome program. Under Galas, DOE has broadened its role in human genome mapping, taking a more interdisciplinary approach.

United States. Congress. Office of Technology Assessment (OTA). **Federal Technology Transfer and the Human Genome Project**. Washington, DC: GPO, 1995. 118 p.

Observing that judicial and legislative policies from the 1980s "...expressly encouraged moving results from federally supported biomedical research to the marketplace," this report goes on to describe the federal-private sector partnerships that make up the Human Genome Project. Cooperative Research and Development Agreements (CRADAs) are a special focus of this study.

United States. Department of Energy, Office of Energy Research, Office of Health and Environmental Research. ***Human Genome 1991-92 (Program Report)***. June 1992. Washington, DC: U.S. Department of Energy, 1992. 248 p.

Highlights of this report include descriptions of research activities at the Lawrence Berkeley, Lawrence Livermore, and Los Alamos laboratories, project information for both active and completed projects, and an outline of the Human Genome Project's management infrastructure. This report also includes an index of the researchers on each project, and an acronym list.

United States. Department of Energy. Office of Health and Environmental Research. Human Genome Program; and National Institutes of Health (United States). National Center for Human Genome Research. **Understanding Our Genetic Inheritance: The U.S. Human Genome Project: the First Five Years, FY 1991-1995**. Springfield, VA: National Technical Information Service, April 1990. 89 p.

This report describes the plans for the U.S. Human Genome Project and updates plans prepared by the OTA and NRC in 1988. Five-year goals are identified for its six components: mapping and sequencing the human and model organism genomes; data collection and distribution; ethical, legal, and social issues; research training; technology development; and technology transfer.

United States. Department of Health and Human Services. National Institutes of Health. ***National Center for Human Genome Research. National Center for Human Genome Research Annual Report 1990***. Bethesda, MD: U.S. National Institutes of Health, 1990. 83 p.

This annual report outlines the organizational and scientific achievements of the initial year of the Human Genome Project. Included is a brief history of the program, plus

descriptions of progress in major areas of focus, including: mapping; sequencing; informatics; ethical, legal, and social issues; and research training.

Vogel, Friedrich, and Grunwald, Reinhard, eds. *Patenting of Human Genes and Living Organisms*. New York: Springer-Verlag, 1994. 244 p.

This compilation of papers and discussions from the Heidelberg Academy of Sciences workshop on gene patenting (July 1-2, 1993) provides an in-depth analysis of the issues involved in applying patent law to medicine and agriculture. Topics include contemporary European patent law, intellectual property and the life sciences, genetic diversity in developing countries, patenting parts of organisms, and an analysis of intellectual property rights and genetic research from a number of ethical viewpoints.

Wade, Nicholas. **Once Again, Scientists Say Human Genome Is Complete**. *New York Times*, April 15, 2003; p. F1, F4.

Wade describes the scientific process behind validating the human genome data so that it can now be called operationally complete.

Watson, James D. [The Human Genome Project: Past, Present and Future](#). *Science* 248(4951): 44-49, April 6, 1990. doi: 10.1126/science.2181665

As the director of the National Institutes of Health(NIH) National Center for Human Genome Research (NCHGR), Watson describes the development of the Human Genome Program, tracing it from the "1973 birth of the recombinant DNA revolution," through subsequent meetings, to the current status of NIH's involvement in the program. He includes a brief description of international genome initiatives and prospects for future applications.

Watson, James D.; and Cook-Deegan, Robert Mullan. [Origins of the Human Genome Project](#). *Federation of American Societies of Experimental Biology Journal* 5(1): 8-11, January 1991.

While the expected outcome of the Santa Cruz Workshop—the founding of a genomics research institute at the University of California—was not realized, the proceedings did convince Nobel Laureate Walter Gilbert of the importance of genome sequencing. Gilbert began promoting the project at other professional meetings held in 1985 and 1986, thereby building broad-based support in the scientific community for genome mapping. The authors review the other pertinent meetings that helped to build a consensus in the scientific community about the Human Genome Project.

Watson, James D.; and Cook-Deegan, Robert Mullan. [The Human Genome Project and International Health](#). *Journal of the American Medical Association* 263(24): 3322-24, 27 June 1990.

Watson and Cook-Deegan illustrate the inherent international nature of the Human Genome Project and predict its positive impact on disease around the world, especially in developing nations. The authors outline current international genome efforts as well as social implications of the research, emphasizing the international nature of the project

and the need for cooperation and interdependence.

Wellcome Trust (Great Britain). [Summary of Principles Agreed at the International Strategy Meeting on Human Genome Sequencing](#) [Bermuda, 25th-28th February, 1996]. London: Wellcome Trust, February 1996. 2 p.

Referred to as the "Bermuda statement," "...this consensus document stresses that mapping data should be released as soon as it is sequenced, and that this data should remain in the public domain." The participants suggested that HUGO, the Human Genome Organization, act as the coordinating agency for these efforts. A followup meeting, the [Second International Strategy Meeting](#), was held February 27 - March 2, 1997.

V. The Future of Genomics Research

This section contains vision statements, commentary, and links to Web sites on trends and projects in human genome research since the human genome sequence was declared [complete](#) on April 14, 2003.

Auffray, Charles; Caulfield, Timothy; Khoury, Muin J.; Lupski, James R.; Schwab, Matthias; and Veenstra, Timothy. [Genome Medicine: Past, Present and Future](#). *Genome Medicine* 3(1): 6-1 to 6-5, January 31, 2011. doi: 10.1186/gm220

To celebrate *Genome Medicine's* 2nd anniversary, the editors reflect on recent advances in research and what they might mean for medicine and health. One notable breakthrough is the application of cloud computing to low-cost genome sequencing, thus making personalized medicine financially feasible. The expansion of direct-to-consumer genetic testing, however, is seen as premature because an appropriate evidence base to guide their use in clinical practice has not been generated. The application of pharmacogenetics to patient care is being delayed by the lack of continuing medical education courses on the use of biomarkers for predicting drug safety. Finally, studies of the public's perception of genetic risk indicate that individualized preventive health strategies based on genetic testing "...may not pan out as expected."

Collins, Francis. [Has the Revolution Arrived?](#) *Nature* 464(7289): 674-675, April 1, 2010. doi:10.1038/464674a

Collins, current director of the U.S. National Institutes of Health (NIH) and former director of NIH's National Human Genome Research Institute (1993 – 2008), reviews the scientific accomplishments in genomic medicine since the completion of the Human Genome Project. While genome-wide association studies (GWAS) have discovered a number of common DNA variations important to understanding the development of common diseases such as heart disease, diabetes, and cancer, Collins reports that "...the consequences for clinical medicine ...have thus far been modest." Advances in clinical medicine include the development of new cancer drugs, breast cancer chemotherapy protocols, macular degeneration treatments, and drug responses for over a dozen drugs.

Notwithstanding these accomplishments, "...the Human Genome Project has not yet directly affected the health care of most individuals."

Collins, Francis S.; Green, Eric D.; Guttmacher, Alan E.; and Guyer, Mark S. [**A Vision for the Future of Genomics Research: A Blueprint for the Genomic Era**](#). *Nature* 2003 April 24; 422(6934): 835-847.

After providing a brief history of genomics research up through the completion of the human genome sequence, the authors describe future research as flowing along three themes - genomics to biology, genomics to health, and genomics to society - with six crosscutting elements shared among them. "Grand challenges" are associated with each theme; an example from genomics to biology is the International HapMap Project, a catalog of the heritable variation in the human genome.

Feero, W. Gregory; Guttmacher, Alan E.; and Collins, Francis S. [**Genomic Medicine—an Updated Primer**](#). *New England Journal of Medicine* 362(21): 2001-11, May 27, 2010.

To illustrate the current "state of the art" for applying genetic advances to clinical medicine, the authors present the case study of a woman who develops breast cancer even though she has tested negative for BRCA1 and BRCA2 mutations. Genetic-expression profiling of the tumor indicates a high risk of recurrence so the patient is prescribed tamoxifen and is cancer-free after five years. This is an example of going beyond "genetic medicine"—the use of single genes to improve the diagnosis and treatment of single-gene disorders—to 'genomic medicine' in which understanding the interactions between the entire genome and nongenomic factors results in new diagnostic and therapeutic approaches to multifactorial conditions such as cancer. The authors conclude with an overview of genetic concepts such as gene regulation and genomic variation.

Green, Eric D.; Guyer, Mark S.; and the National Human Genome Research Institute. [**Charting a Course for Genomic Medicine from Base Pairs to Bedside**](#). *Nature* 470(7333):204-13, February 10, 2011. doi:10.1038/nature09764

After prompting the scientific community to reflect on the current state of genomics and to explore future challenges, the 2011 vision for the Human Genome Project was articulated and organized into five domains: 1) understanding the structure of genomes; 2) understanding the biology of genomes; 3) understanding the biology of disease; 4) advancing the science of medicine; and 5) improving the effectiveness of healthcare. Imperatives for genomic medicine that will capitalize in overlapping advances in these domains include: 1) making genetic tests routine; 2) defining the genetic components of disease; 3) creating a comprehensive view of all cancer genomes; 4) developing genetic information tools for use in the clinic; and 5) sequencing the human microbiome and understanding the role of resident microflora in specific diseases.

Greif, Karen F., and Merz, Jon F. **Big Science: The Human Genome Project and the Public Funding of Science**. In their: *Current Controversies in the Biological Sciences: Case Studies of Policy Challenges from New Technologies*. Cambridge, MA: MIT, 2007: 17-34

Greif and Merz trace the funding streams that supported the Human Genome Project, and posit that without public investment "...the human genome would be "owned" by private companies and not available to all."

Huang, Kenneth G., and Murray, Fiona E. [Entrepreneurial Experiments in Science Policy: Analyzing the Human Genome Project](#). *Research Policy* 39(5): 567-582, June 2010. doi: 10.1016/j.respol.2010.02.004

The authors describe the Human Genome Project as "...a setting for a much richer understanding of the ways in which organizational and institutional choices shape scientific productivity." They posit that government collaboration promotes experimentation by reducing risk and by insuring that data is shared by all project participants. This article includes a detailed history of the Human Genome Project collaborators.

The International HapMap Consortium. [A Haplotype Map of the Human Genome](#). *Nature* 437(7063): 1299-1320, October 27, 2005. doi:10.1038/nature04226

To celebrate the completion of the [International HapMap Project](#) database, the consortium authors (from sixty-three centers) describe its development and its potential use for genome-wide association studies.

Marturano, Antonio. [When Speed Truly Matters, Openness is the Answer](#). *Bioethics* 23(7): 385-393, September 2009. doi: 10.1111/j.1467-8519.2009.01723.x

Marturano describes the influence of alternative open-source methodologies on maintaining the Human Genome Project as "a visionary ideal" focused on the intrinsic goods of scientific research rather than on the limited extrinsic goods of "patent and publish" market forces.

NIH HMP Working Group. [The NIH Human Microbiome Project](#). *Genome Research* 19(12):2317-23, December 2009. doi: 10.1101/gr.096651.109

This article outlines the history and implementation of a program to characterize the human microbiome, a term coined by Joshua Lederberg "...to signify the ecological community of commensal, symbiotic and pathogenic microorganisms that literally share our body space." In addition to reviews of the influential scientific papers in the field, this paper includes descriptions of current demonstration projects and sequencing centers.

Pohlaus, Jennifer Reineke, and Cook-Deegan, Robert M. [Genomics Research: World Survey of Public Funding](#). *BMC Genomics* 9: 472, 2008. doi:10.1186/1471-2164-9-472

The authors surveyed 89 organizations in 26 countries to gather a quantitative estimate of the amount of genomics research funded each year. They found that public funding was US \$2.9 billion annually for the years 2003-2006. While the United States spent the most

money in terms of total dollars, Ireland, Great Britain and Canada spent more of their total research budget on genomics research.

Rabinow, Paul; and Dan-Cohen, Talia. *A Machine to Make a Future: Biotech Chronicles*. Princeton, NJ: Princeton University Press, 2005. 199 p.

Noting that books about human genome mapping usually ‘...tell a story of vivid actors, dramatic events, and significant scientific discoveries’, the authors choose instead to “...present the vicissitudes, strategies, and tactics of an emergent project during a finite period of time, as articulated by the actors themselves.” Interviews were conducted over time with Celera Diagnostics scientists Tom White, Gabriella Dalisay, Kathy Ordonez, Shirley Kwok, Joe Catanese, Paul Billings, Victor Lee, and James Devlin, to “...create an anthropological archive of a biotechnological event (most traces of which have already disappeared, as have the traces of countless others) for others to use later...”

Varmus, Harold. [Ten Years on--The Human Genome and Medicine](#). *New England Journal of Medicine* 362(21):2028-9, May 27, 2010. doi: 10.1056/NEJMe0911933

Varmus, writing as the director of the National Cancer Institute, holds that genomics remains aligned with modern science instead of with modern medicine a decade after the sequencing of the human genome. He supports the use of the term “personalized medicine” because it “...wards off claims that an overreliance on genotypes in medical practice is deterministic” and thus allows for integrating insights from genomic medicine with environmental, social and behavioral factors in the treatment of patients.

Via, Marc; Gignoux, Christopher; and Burchard, Esteban González. [The 1000 Genomes Project: New Opportunities for Research and Social Challenges](#). *Genome Medicine* 2(1):3, January 21, 2010. doi: [10.1186/gm124](#)

The authors describe the sequencing the genomes of individuals from 20 different populations, many of whom are children. A major purpose of the project is to reconstruct the parental chromosomal phase using genetic data from children. These studies will provide more precise data on disease-causing genes than genome-wide association studies.

Web sites:

[The Cancer Genome Atlas](#) [Web site]

The Cancer Genome Atlas portal enables researchers to download and analyze genomic data sets to further understand the molecular basis of cancer. A partnership of the U.S. National Institutes of Health’s National Cancer Institute and the National Human Genome Research Institute, the main goal is to identify key genetic “targets” within cancer cells in order to create therapeutic drugs with more specificity.

[Human Genome at Ten](#) [Web site]

This special issue of Nature contains review articles and commentary on the state of genomic research ten years after the completion of the draft human genome sequence. A video survey of over 20,000 genes is included along with a link to the complete DNA sequence of the human genome.

[Human Microbiome Project](#) [Web site]

The purpose of the Human Microbiome Project (HMP) is to analyze microbes from the nose, mouth, skin, and GI tract to understand the role they play in health and disease. A specific HMP ELSI (Ethical, Legal and Social Implications) program has been established to research issues regarding forensic uses, biodefense applications, and privacy concerns.

[International HapMap Project](#) [Web site]

Focusing on genetic variation and disease through research with single nucleotide polymorphisms (SNPs), the International HapMap Project was a partnership of scientists and funding agencies from sixty-three centers in Canada, China, Japan, Nigeria, the United Kingdom, and the United States. The researchers have created a public database that is used to pinpoint over a hundred regions of the genome associated with common diseases such as coronary artery disease and diabetes.

[1000 Genomes Project](#) [Web site]

The genomes of over 1000 unidentified individuals from around the world are being sequenced by researchers in China, Germany, the United Kingdom and the United States to produce a catalog of human genetic variation.

VI. Ethical, Legal and Social Implications of the Human Genome Project (ELSI)

Achenbaum, Laura S.; McCurdy, Sheryl; Whitney, Simon N.; Slashinski, Melody J.; and McGuire, Amy L. [The Human Microbiome Project: Ethics from the Trenches](#) [Presentation]. International Human Microbiome Congress: Vancouver, British Columbia, Canada, March 9-11, 2011. 18 p.

The authors present an overview of the ethical issues raised by the [Human Microbiome Project](#) (HMP). They review the informed consent document used in their research project, and note that its length and complexity lead to problems with comprehension. Risk analysis is problematic given that multiple sites are simultaneously investigated on research subjects' bodies. Results from a survey of participants and investigators reveal that trust is the key element in the consent process.

Annas, George J. [Mapping the Human Genome and the Meaning of Monster Mythology](#). In: *A Companion to Genethics*,. Pp. 127-143. Edited by Justine Burley and John Harris. Malden,

MA: Blackwell Publishers, 2008 (2007, 2002). Previously published in *Emory Law Journal* 39(3): 629-64, Summer 1990. doi:10.1002/9780470756423.ch10

Law professor Annas states that a project like mapping the human genome needs to confront "mythical dragons with knowledge" and needs to anticipate the "...real monsters: the value conflicts that new knowledge produces." Using literary examples to frame the debate, he presents a brief discussion of the genome project, provides an analysis of three levels of related legal and ethical issues (individual/family, society, and species), and suggests strategies for regulation of genetic technology.

Beckwith, Jon. **Geneticists and the Two Cultures**. In his *Making Genes, Making Waves: A Social Activist in Science*, pp. 191-210. Cambridge, MA: Harvard University Press, 2002.

As a member of the NIH-DOE Joint ELSI Working Group, Beckwith describes the animosity faced by ELSI researchers from geneticists who believed the program to be a "...welfare program for ethicists who only talked but didn't change the world." Referencing C.P. Snow's *Two Cultures*, the author reflects on the continuing conflict between scientists and those in other disciplines. While he documents the success of ELSI in recommending a moratorium on cystic fibrosis carrier screening, he was ultimately disappointed in his ELSI experience and calls for increased cooperation to solve the social dilemmas associated with scientific research.

Capron, Alexander M. [Human Genome Research in an Interdependent World](#). *Kennedy Institute of Ethics Journal* 1(3): 247-51, September 1991.

Capron describes an ELSI meeting held on June 2-4, 1991, which included representatives from fifteen countries including the Soviet Union and Japan. The purpose of the meeting was to identify ELSI issues of international significance, and to determine whether the issue would best be addressed in an international or domestic context. A "Global Steering Committee on Ethical and Social Issues in Genome Research" was established to follow-up on the resolutions passed by conference participants. This committee included representatives from the Joint NIH-DOE ELSI Working Group; UNESCO; Council for International Organizations of Medical Sciences (CIOMS); Council of Europe's CAHBI (Ad Hoc Committee of Experts on Bioethics); European Community's ESLA (Ethical, Social, and Legal Aspects of Genome Research); HUGO; International Association of Bioethics; and the genome mapping projects of the Soviet Union and Japan.

Fletcher, John C., and Wertz, Dorothy C. [Ethics, Law, and Medical Genetics: After the Human Genome Is Mapped](#). *Emory Law Journal* 39(3): 747-809, Summer 1990.

In a survey of 677 international geneticists, the authors asked respondents to rank the significance of ten ethical issues. The study provides guidelines for ethical problems that may arise in medical genetics and for avoiding abuse of genetic knowledge by employers, insurers, or the health care system.

Frankel, Mark S.; and Teich, Albert H., eds. *The Genetic Frontier: Ethics, Law, and Policy*. Washington: American Association for the Advancement of Science (AAAS), 1994. 240 p.

Two of the four sections in this report of a conference sponsored by the AAAS-American Bar Association National Conference of Lawyers and Scientists and the AAAS Committee on Scientific Freedom and Responsibility are concerned with genetic testing. Part II includes privacy and confidentiality issues discussed in three areas by Laurence Tancredi, Alan F. Westin, and Madison Powers. Part IV reports on genetic testing and determination of property rights: one by Gilbert S. Omenn on the scope of patent protection; three papers touching on intellectual property and genetic testing by Kate H. Murashige, Thomas J. White, Joan Overhauser; and Ted Peters, intellectual property and human dignity.

Greely, Henry T. [Neuroethics and ELSI: Similarities and Differences](#). *Minnesota Journal of Law, Science & Technology* 7(2): 599-637, May 2006.

After tracing the first usages of the expressions “ethical, legal and social implications,” “ELSI,” and “genethics,” Greely reviews the strengths and weaknesses of the Human Genome Project’s (HGP) ELSI program. While he finds that much of the research on ELSI issues funded by the HGP does not apply to the field of neuroethics, the author posits that there are three ways the field benefits from ELSI’s legacy: by creating a generation of interdisciplinary researchers skilled in studying ethical issues; by contributing to the financial stability of bioethics centers; and by expanding the audience for bioethical discussions and analysis.

Hanna, Kathi E. **The Ethical, Legal, and Social Implications Program of the National Center for Human Genome Research: A Missed Opportunity?** In *Society's Choices: Social and Ethical Decision Making in Biomedicine*, pp. 432-457. Edited by Ruth E. Bulger, Elizabeth M. Bobby, and Harvey V. Fineberg. Washington: National Academy Press, 1995.

The author reviews efforts to carry on a public dialogue about the potential social consequences of genome research. Hanna describes James D Watson's creation of the Ethical, Legal, and Social Implications (ELSI) Program as part of the NIH Office of Human Genome Research and the controversy this engendered. She goes on to describe ELSI and its extramural grants program, and suggests that "...because the only citizens with access to the [grant's] process are those schooled in an academic or professional discipline...[ELSI research] is a reductionist process that runs the risk of ignoring the most pressing policy issues...[since it] can in no way guarantee fair representation of all points of view." In addition, ELSI has no mechanism to synthesize the research it funds for distribution to Congress and to agencies involved in policy formulation. Hanna recommends that ELSI develop such a mechanism as well as change its grants procedure to allow for more diversity of opinion. "Otherwise, it will be remembered as a missed opportunity to aggressively address the complex social issues raised by the Human Genome Project."

Human Genome Organization (HUGO). Ethical, Legal, and Social Issues Committee. [Principled Conduct of Genetics Research](#). *Bulletin of Medical Ethics* 121: 10-11, September 1996.

Based on a discussion paper "Ethical Issues in International Collaborative Research on the Human Genome: The HGP and the HGDP" by Bartha Maria Knoppers, Marie Hirtle, and Sebastien Lormeau, the HUGO-ELSI committee makes ten recommendation on informed consent, privacy, and oversight of genome research.

Juengst, Eric T. **Self-Critical Federal Science? The Ethics Experiment within the U.S. Human Genome Project**. *Social Philosophy and Policy* 13(2): 63-95, Summer 1996. doi: 10.1017/S0265052500003460

Based on his experience as Chief of the Ethical, Legal, and Social Implications (ELSI) Branch of the National Center for Human Genome Research from 1990-1994, Juengst discusses critiques of ELSI as a public relations gimmick and as an avenue for "alarmist hype," ELSI's track record as an "un-commission" for policy issues on genetic research, and ways for ELSI to be more "proactive" within a climate of special interests and cost-cutting measures.

Kay, Jane; Meslin, Eric M.; Knoppers, Bartha; and Juengst, Eric T. [Global ELSI: A Research Strategy for Genomics](#) [Position Statement]. [Public Population Project in Genomics](#) [P3G], April 21, 2011.

P3G is an international consortium that provides expertise and resources to the population genomics community. As members of this consortium, the authors propose an international ELSI network to address the specific global ethical issues associated with this research.

Koski, Cheryl A. [The Human Genome Project: An Examination of its Challenge to the Technological Imperative](#). *New Genetics and Society* 2005 December; 24(3): 267-281. doi: 10.1080/14636770500349791

After noting that public debate about genetic research and the technological imperative did not end with the 2003 announcement of the mapping of the human genome, Koski reminds us that "...the general public has been weighing benefits against harms ever since the inception of the HGP in 1990." The author asserts that the interaction of scientists and the public regarding ethical, legal and social issues of genome mapping has resulted in the general public becoming a "...major player in science policy in the United States during the past quarter century."

Macklin, Ruth. **Mapping the Human Genome: Problems of Privacy and Free Choice**. In: *Genetics and the Law III*, pp. 107-14. Edited by Aubrey Milunsky and George J. Annas. New York: Plenum Press, 1985.

Professor Macklin argues that the genome project raises privacy and free choice issues that, although far from solved, have been addressed in the past by scientists, clinicians,

patients, and policymakers in other settings. She discusses points of a President's Commission report entitled [Screening and Counseling for Genetic Conditions](#) (1983) and suggests that we apply similar findings to the human genome, remembering that such precedents should continue to undergo renewed inquiry and debate.

Mahowald, Mary B.; Levinson, Dana; Cassel, Christine; Lemke, Amy; Ober, Carole; Bowman, James; Le Beau, Michelle; Ravin, Amy; and Times, Melissa. [The New Genetics and Women](#). *Milbank Quarterly: A Journal of Public Health and Health Care Policy*. 74(2): 239-283, 1996.

The authors note that "...because of their central role in reproduction and caregiving, [women] are affected not only differently but also more significantly than men by the information emerging from the HGP [Human Genome Project]." They go on to note that only a few of the studies sponsored by HGP on the social implications of genome research address women's issues. To further such research, the authors conduct an extensive literature review on women and genetics, and compile a list of areas deserving further study.

Marshall, Eliot. **Whose Genome Is It, Anyway?** *Science* 273(5283): 1788-1789, September 27, 1996.

As seven large-scale sequencing projects were scheduled to begin, the directors of the research reviewed the composition of the DNA libraries to be used in their studies. It was found that, instead of a "mosaic of DNA from a variety of anonymous sources...[the DNA] appears to come primarily from a limited group of donors: three men and one woman." Further investigation revealed that informed consent had not been obtained from these donors and that their anonymity had not been preserved. This situation prompted the development of ethical guidelines for the construction of DNA libraries: [NCHGR-DOE Guidance on Human Subjects Issues in Large-Scale DNA Sequencing](#).

McGee, Glenn. *The Perfect Baby: A Pragmatic Approach to Genetics*. London: Rowman & Littlefield, 1997. 166 p.

McGee asserts that "[s]cholarship concerning the Human Genome Project has tended, either in condemnation or in endorsement of genetic engineering, to rely on outmoded and poorly thought through notions of 'nature', 'technology', and 'freedom'...[t]he antidote to such categories is a reconstruction of the discussion that takes note of confluences between genetic engineering and other scientific, parental, and social practices." Drawing on the work of American Pragmatists Thomas Dewey and William James, the author proceeds to discuss the hopes and fears surrounding genome research, and proclaims that "in the field of common sense", "the usefulness of our approach to genetics must be tested in the context of ordinary people."

McGuire, Amy L.; Colgrove, James; Whitney, Simon N.; Diaz, Christina M.; Bustillos, Daniel; and Versalovic, James. **Ethical, Legal, and Social Considerations in Conducting the Human Microbiome Project**. *Genome Research* 18(12):1861-1864, December 2008. doi: 10.1101/gr.081653.108

The authors describe the unique ethical challenges raised when considering what constitutes informed consent for the [Human Microbiome Project](#) (HMP) research subjects, and call for the creation of an interdisciplinary advisory group to assist Institutional Review Boards in their review of HMP research.

McLean, Margaret R. **Religion, Ethics, and the Human Genome Project**. In: *Science, Religion, and Society: An Encyclopedia of History, Culture, and Controversy*, pp. 787-794. Edited by Arri Eisen and Gary Laderman. Armonk, NY: M.E. Sharpe, 2007.

This chapter highlights the Ethical, Legal, and Social Implications (ELSI) portion of the Human Genome Project. Seventy six million dollars were provided for this endeavor. The ELSI program is particularly interested in genetic testing and pharmacogenomics. Many questions arise in the course of planning genetic testing. These include: who has access to genetic information, is screening voluntary or mandatory, and how genetic information will be used. Pharmacogenomics is a form of genetic testing that assesses individual response to a given medication.

Murray, Thomas H. [Ethical Issues in Human Genome Research](#). *FASEB Journal* 5(1): 55-60, January 1991.

In this concise description of the major ethical questions surrounding genome research, Murray breaks his discussion into three major areas: uses and misuses of genetic information, genetic manipulation, and challenges to our self-understanding. He concludes that research and scientific findings should not be abandoned, but that we must learn to communicate these findings and their implications effectively.

Nordgren, Anders. **The Human Genome Project: Justification, Promotion, and Access to Results**. In his *Responsible Genetics: The Moral Responsibility of Geneticists for the Consequences of Human Genetics Research*, pp. 91-126. Dordrecht: Kluwer Academic Publishers, 2001.

Nordgren's answers the question "what should responsible geneticists do?" by saying that they should take responsibility for all stages of genetic research: challenging the "geneticization" of medicine beyond "tool-making;" balancing scientific and social values in applying genetic techniques to clinical practice; fighting the misrepresentation of genetics, and protesting the patenting of genes. "Responsible dialogue" is required of geneticists on all these issues, as well as continued vigilance regarding attempts to redefine the Human Genome Project by those who would profit from it.

Oliver, Jill M., and McGuire, Amy L. [Exploring the ELSI Universe: Critical Issues in the Evolution of Human Genomic Research](#). *Genome Medicine* 3(6):38, June 21, 2011.

The authors report on the National Human Genome Research Institute (NHGRI) Ethical, Legal, and Social Implications Research Program 2011 Congress, [Exploring the ELSI Universe](#), held in Chapel Hill, North Carolina (USA) on April 12-14, 2011. ELSI

researchers reported on projects addressing privacy issues with biobanks, and on the potential impact of genomic research on health care disparities.

Rollin, Bernard E. *The Frankenstein Syndrome: Ethical and Social Issues in the Genetic Engineering of Animals*. New York: Cambridge University Press, 1995. 241 p.

Noting that "...science and technology have become whipping boys for social ills," the author finds that it is "...no surprise that the Frankenstein story strikes a socially responsive chord, providing us with a way of articulating our fears and doubts about science and technology...[and that this] myth is either accepted as literal truth or categorically rejected as nonsense, with little thought for the possibilities in between, where the truth surely lies. This dichotomized tendency... blocks... any meaningful attempt to place [genetic research] under... social control or to orchestrate practicable social policies." Rollin attempts to undo this polarization by frankly discussing the possible dangers and potential benefits of genetic research. He also looks at the problems associated with creating transgenic animals in the context of the animal rights movement.

United States. National Institutes of Health [NIH] and Department of Energy [DOE]. Joint Committee to Evaluate the Ethical, Legal, and Social Implications Program (ELSI) of the Human Genome Project. (Committee: Rothstein, Mark A.; Spence, M. Anne; Buffler, Patricia A.; Childress, James F.; Epstein, Charles J.; Hilgartner, Stephen; Knoppers, Bartha Marie; Mackta, Jayne; Olson, Maynard V.; Shine, Kenneth I.; and Walker, Bailus). [Report of the Joint NIH-DOE Committee to Evaluate the Ethical, Legal, and Social Implications Program \(ELSI\) of the Human Genome Project](#). February 27, 1997. 11 p.

The Committee found that the scope of work for the ELSI Working Group was "...so broad and complex as to be confusing," which in turn lead to operating problems. The Committee recommended that the ELSI effort be strengthened by: 1) restructuring the working group to focus on evaluation of ELSI grants; 2) mandating the director of NIH to coordinate dissemination of ELSI information among the Institutes; and 3) establishing an Advisory Committee on Genetics and Public Policy in the Office of the Secretary at Health and Human Services. This committee would be responsible for "...formulating policy to ensure integration of new genetic knowledge into health care standards".

United States. Congress. House. *Human Genome Privacy Act*. H.R. 2405, 102d Congress, 1st Session. By John Conyers. Introduced 24 April 1991.

This landmark bill proposed protecting the individual's right to privacy of one's genetic information, and upholds the individual's right to access personal genome records.

United States. Department of Health and Human Services, National Institutes of Health, National Center for Human Genome Research. [ELSI: A Review of the Ethical, Legal, and Social Implications Research Program and Related Activities \(1990 - 1995\)](#). Bethesda: National Center for Human Genome Research, April, 1996. 17 p.

After outlining the goals of the Ethical, Legal, and Social Implications (ELSI) program, this report focuses on high priority issues for genetic research: privacy of genetic information, integration of genetic technologies into clinical settings, informed consent for genetic research, and genetics education for both health professionals and the public.

United States. National Center for Human Genome Research, and United States. Department of Energy. [*NCHGR-DOE Guidance on Human Subjects Issues in Large-Scale DNA Sequencing: Executive Summary \[and\] Executive Summary of Joint NIH-DOE Human Subjects Guidelines*](#). August 17, 1996.

This document addresses the ethical issues involved in recruiting and protecting tissue donors for genome research. The guidance is divided into six sections: 1) the risks/benefits of genome sequencing; 2) privacy and confidentiality; 3) recruitment of donors for DNA libraries; 4) informed consent for those donating DNA; 5) IRB approval of DNA library construction; and 6) use of existing DNA libraries consisting of samples for which proper informed consent has not been obtained.

Wexler, Nancy. **James D. Watson and the ELSI Years**. In *Inspiring Science: Jim Watson and the Age of DNA*, pp. 403-412. Edited by John R Inglis, Joseph Sambrook, and Jan A. Witkowski. Cold Spring Harbor, NY: Cold Spring Harbor Laboratory Press, 2003.

As the co-chair (with Victor McKusick) of the first Joint NIH-DOE ELSI Working Group, Wexler begins her essay by recalling that James Watson's first act as Director of the Human Genome Project (HGP) at the U.S. National Institutes of Health was to announce that he was setting aside a significant percentage of the HGP budget to address its impact on people and society. Before Watson "...it was unprecedented to include, at the inception of a science project, be it large or small, a simultaneous commitment to investigate and attend to the societal ramifications of this science." While some researchers had "...moral quandaries about accepting ELSI funding—they were concerned that taking ELSI money was tantamount to supporting the science... the funding from the ELSI program was without strings...[c]riticism and suggestions were genuinely welcome."

Yesley, Michael S. [*Bibliography: Ethical, Legal, and Social Implications of the Human Genome Project*](#). Washington, DC: U.S. Department of Energy, Office of Energy Research, 1993. 265 p. doi: 10.2172/10108311

Compiled from Los Alamos National Laboratory databases, this bibliography updates one published the previous year, and is organized into 15 topic areas: behavior, cystic fibrosis, counseling, discrimination, ethics, eugenics, forensics, Huntington's disease, law, patents, privacy, reproduction, screening/diagnosis, sickle cell anemia, and therapy. A [supplement](#) was published in 1994.

Yesley, Michael S. [*What's ELSI Got to Do With It? Bioethics and the Human Genome Project*](#). *New Genetics and Society* 27(1): 1-6, March 2008.

Yesley compares bioethics commissions and extra-mural programs – i.e. programs funded by the executive branch agencies for work done outside the government— regarding the ethical issues involving the Human Genome Project. The more restrictive nature of the extra-mural program has created literature, but not public policy. He recommends instead that Congress allocate part of the ELSI budget to fund a commission.

Zilinskas, Raymond A., and Balint, Peter J., eds. *The Human Genome Project and Minority Communities: Ethical, Social and Political Dilemmas*. Westport, CT: Praeger Publishers, 2001. 144 p.

This collection of presentations from the conference [*The Human Genome Project: Reaching the Minority Communities in Maryland*](#), held at the University of Maryland [Baltimore campus] in June 1997, address "...the divisions between minority groups and the scientific community, particularly in the area of medical and genetic research." The editors suggest that the Human Genome Project "...conducted in accordance with the highest ethical standards," can be particularly helpful to minority communities who "...have much to gain from innovative medical therapies that may result from the study of human genetic

VII. POSITION STATEMENTS

American College of Medical Genetics. Storage of Genetics Materials Committee. [**ACMG Statement: Statement on Storage and Use of Genetic Materials**](#). *American Journal of Human Genetics* 57(6): 1499-1500, December 1995.

Noting that "many health professionals as well as lay people may not appreciate how frequently biological samples are stored and how easily samples that have been stored for an unrelated reason could be used for genetic analysis in the future", the Committee enumerates the issues that must be addressed when obtaining samples for both clinical and research purposes.

American Society of Human Genetics. [**ASHG Report: Statement on Informed Consent for Genetic Research**](#). *American Journal of Human Genetics* 59(2): 471-474, August 1996.

Noting that concepts in bioethics evolve in concert with scientific developments, this American Society of Human Genetics (ASHG) statement "affirms traditional research practices in human genetics and recommends new ones that it believes can provide direction for ongoing developments." The report discusses retrospective studies using existing samples, research with prospectively collected samples, disclosure and informed consent, and disposition of collected samples and test results.

Council for Responsible Genetics. [**Position Paper on Genetic Discrimination**](#). *Issues in Reproductive and Genetic Engineering* 3(3): 287-95, 1990. [Updated, January 2001]

Written to stimulate discussion on the direction of human genetic research, this statement presents a critique of the scientific limitations of research in human genetics, and highlights the adverse social and economic implications of an increase in genetic testing.

Council of Europe, Directorate of Legal Affairs. [Convention for the Protection of Human Rights and the Dignity of the Human Being with Regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine](#). Strasbourg: Council of Europe, 1997. 11 p. *Kennedy Institute of Ethics Journal* 7(3): 277-90, September 1997; a summary is published in the *Bulletin of Medical Ethics* (119): 18-21, June 1996.

Genetic research is the focus of Chapter IV of this declaration, in which genetic discrimination is prohibited, genetic testing is limited to those matters pertaining to health, somatic cell gene therapy is permitted but germ-line gene therapy is prohibited, and preimplantation screening may not be used to select the sex of the child except when serious sex-linked genetic diseases are involved.

Council of the European Communities. **Council Decision Adopting a Specific Research and Technological Development Programme in the Field of Health: Human Genome Analysis (1990 to 1991)**. *Official Journal of the European Communities* L196: 8-14, 26 July 1990.

Adopting a two-year program for the EC in the field of human genome research, this statement sets out the specific recommendations and rules, including the decision to prohibit alteration of germ cells or any stage of embryo development aimed at achieving inheritable modifications.

[The Declaration of Inuyama and Reports of the Working Groups](#). *Human Gene Therapy* 2(2): 123-29, Summer 1991.

The Council for International Organizations of Medical Sciences held its XXIVth Round Table Conference in July 1990, entitled, "Genetics, Ethics and Human Values: Human Genome Mapping, Genetic Screening and Therapy". Outlined in this article are the interdisciplinary and transcultural views of the participating attendees and working groups and their final agreement, the Inuyama Declaration, which validates the project, but warns against misuse of knowledge gained.

European Parliament. European Parliament on Genetics. **(Resolution of 16 March 1989, slightly abridged)**. *Bulletin of Medical Ethics* (57): 8-10, April 1990.

The European Parliament outlines its resolution on ethical and legal problems of genetic engineering. The Council upholds individual rights and the right of the patient, genetic strategies for social problems, and confidentiality and reliability of information. It denounces discrimination by employers and insurers against employees with a predisposition to illness, and calls for protection of genetic data.

Human Genome Organization (HUGO). [HUGO Statement on Patenting of DNA Sequences](#) [January 1995]. Online. Human Genome Organization (HUGO). [Accessed July 26, 2011]

This statement discusses the concerns of scientists that patent law, when applied to genome sequencing, would reward those who map genes but not those who determine biological functions and applications.

NIH-DOE Working Group on Ethical, Legal, and Social Issues (ELSI). [Statement: The Bell Curve](#). *Human Genome News* 7(5): January-March, 1996.

Commenting on the link between genetics and intelligence proposed by The Bell Curve authors Richard Hernstein and Charles Murray, ELSI members stated that "...as geneticists and ethicists associated with the Human Genome Project, we deplore The Bell Curve's misrepresentation of the state of genetic knowledge [in behavioral genetics] and the misuse of genetics to inform social policy."

Short, Elizabeth M. [Proposed ASHG Position on Mapping/Sequencing the Human Genome](#). *American Journal of Human Genetics* 43(1): 101-2, July 1988.

The proposed policy recommendations of the Committee on Mapping and Sequencing the Human Genome of the Board on Basic Biology of the National Academy of Sciences provides suggestions for the project on organizing, establishing goals, and funding.

UNESCO - International Bioethics Committee. [Universal Declaration on the Human Genome and Human Rights](#) [revised draft]. Paris, France: 1997. 3 p. [Also published in the *Bulletin of Medical Ethics* (126): March, 1997, 9-11]

This consensus document sets forth a list of principles intended to protect individual rights as global genome research progresses. The statement references other international instruments pertaining to genome projects, such as the Budapest treaty on micro-organisms and patenting, the Bern and World Trade Organization intellectual property rights agreements, and United Nations Convention on Biological Diversity. The draft was ratified on November 11, 1997.

United States Department of Health and Human Services [NIH] National Human Genome Research Institute; and Department of Energy (DOE). [NIH-DOE Guidelines for Access to Mapping and Sequencing Data and Material Resources](#). August 17, 2007. Last reviewed December 19, 2010.

This guideline is a response to the need a policy for rapid dissemination of genomic data versus the right of researchers to intellectual property rights. The guideline states that a six month period between generation of data and dissemination of the data is the maximum length of time allowable. Grant applicants will be expected to provide information concerning their plan for dissemination.

Workshop on International Cooperation for the Human Genome Project. October 24-26, 1988, Valencia, Spain. **Valencia Declaration on the Human Genome Project**. *Hastings Center Report* 19(4): S19, July/August 1989.

The Declaration, approved by attendants of the Workshop on International Cooperation for the Human Genome Project, Valencia, Spain, asserts that the genome project will have great benefits provided that genetic information is used only to enhance the dignity of the individual. The signers encourage international collaboration, coordination of information, development of compatible database networks, availability to public of

information, and recognition of HUGO as the lead body to promote goals and objectives addressed in their declaration.

World Medical Association. [Declaration on the Human Genome Project](#). In *Handbook of Declarations*. Ferney-Voltaire, France: World Medical Association, 1992, p. 17.S/1.1 - 17.S/1.3. [Rescinded WMA General Assembly, Santiago, 2005]

Stating that "[t]he ethical issues raised by the Human Genome Project are not linked with the technology itself but with its proper use," this declaration sets forth five basic guidelines for genetic research: 1) international sharing of information; 2) equitable access to genetic services; 3) maintenance of privacy; 4) full disclosure of genetic information; and 5) that all genetic screening be done on a voluntary basis.

VIII. Paradigms of Genetics, Disease and Personhood

Baird, Patricia.A. **Genetics and Health Care: A Paradigm Shift**. *Perspectives in Biology and Medicine* 33(2): 203-213, Winter 1990.

Baird notes that, with advances in genetics, our "...view of the determinants of health as being external is too simplistic." As progress is made in genome sequencing, the concept of "genetic individuality" and individual risk will be incorporated into disease prevention programs. "Genetics will increasingly allow us to interfere earlier in the cascade of events leading to overt disease and clinical manifestations. But as a society we will take advantage of this opportunity...only if we are prepared to incorporate a new view of disease with an internal as well as an external cause into our planning for health care systems and programs."

Chapman, Audrey R., ed. *Unprecedented Choices: Religious Ethics at the Frontiers of Genetic Science*. Theology and the Sciences Series. Minneapolis: Fortress Press, 1999. 260 p.

Chapman opens with a general background of genetic developments and then discusses cloning, patenting life, human personhood, sociobiology, and other topics from a religious and theological perspective. She posits that "[t]he genetic revolution offers both a challenge and an opportunity to the religious community: a challenge to apply religious values and frameworks to new and unprecedented issues and an opportunity to help interpret and illuminate significant ethical choices before their members and the broader society."

Charlesworth, Max. **Human Genome Analysis and the Concept of Human Nature**. In: *Human Genetic Information: Science, Law and Ethics*, pp. 180-98. Ciba Foundation Symposium 149. Chichester, England: Wiley, 1990.

The author asserts that we must have a clear picture of human nature not solely derived from genetics to help us decide which kinds of genetic procedures should be permitted.

Collins, Francis S. **Deciphering God's Instruction Book: The Lessons of the Human Genome**, pp. 109-144. In his *The Language of God: A Scientist Presents Evidence for Belief*. New York: Free Press, 2006.

Describing his experiences with the Human Genome Project both as a scientist and as the director of the U.S. National Institutes of Health genome center, Collins relates that "...as a believer, the uncovering of the human genome sequence held additional significance. This book was written in the DNA language by which God spoke life into being." While materialists claim that mapping the genome proves there is no God, he counters that "[f]reeing God from the burden of special acts of creation does not remove Him as the source of the things that make humanity special... [i]t merely shows us something of how He operates."

Durant, John; Hansen, Anders; and Bauer, Martin. **Public Understanding of the New Genetics**. In *The Troubled Helix: Social and Psychological Implication of the New Human Genetics*, pp. 235-48. Edited by Theresa Marteau and Martin Richards. Cambridge: Cambridge University Press, 1996.

The authors studied public perceptions of the Human Genome Project by analyzing the results of focus group interviews on genome research and references to genetics in the British media. They review similar research conducted in other countries, and situate their findings within the broader context of popularizations of basic science.

Griffiths, Paul E.; and Stotz, Karola. [Genes in the Postgenomic Era](#). *Theoretical Medicine and Bioethics* 27(6): 499-521, 2006. doi: 10.1007/s11017-006-9020-y

The authors present three different concepts of "the gene": instrumental (its role in experimentation and research), nominal (its function as a scientific tool), and post-genomic (the continuing project of understanding the multiple layers of its functioning). Noting that definitions in genomic research are used imprecisely in both the scientific and popular literature, Griffiths and Stotz propose and defend their own definition of "the gene": "...they are ways in which cells utilize available template resources to create biomolecules that are needed in a specific place at a specific time."

Heller, Jan Christian. **Human Genome Research and the Challenge of Contingent Future Persons: Toward an Impersonal Theocentric Approach to Value**. Omaha: Creighton University Press, 1996. 179 p.

The author asserts that the traditional weighing of harms and benefits is not sufficient when considering those who would not be born but for genome research. Heller proposes ways to extend our moral domain to include these "contingent future persons", and discusses the works of Derek Parfit, David Heyd, Richard McCormick, and James Gustafson in this regard.

Juengst, Eric T. **Concepts of Disease after the Human Genome Project**. In *Ethical Issues in Health Care on the Frontiers of the Twenty-First Century*, pp. 127-154. Edited by Stephen

Wear, James J. Bono, Gerald Logue, and Adrienne McEvoy,. Dordrecht: Kluwer Academic Publishers, 2000.

Juengst discusses nineteenth-century concepts of disease that have “carried forward” into discussions of genetics: genetic imperialism (“all disease is genetic disease”); genetic contagionism (genetic diseases have carriers just like infectious diseases); and genetic humoralism (genetic mutation represents an intrinsic imbalance). The author calls for us to go “...beyond simply substituting ‘genes’ for both ‘germs’ and ‘humors’,” and “...shift to a multidimensional understanding of disease causation” to counter “...the determinism with which genetic explanations are now invested.”

Juengst, Eric, and Huss, John. [From Metagenomics to the Metagenome: Conceptual Change and the Rhetoric of Translational Genomic Research](#). *Genomics, Society and Policy* 5(3): 1-19, 2009.

The authors undertake a “close reading” of the [Human Microbiome Project](#) (HMP), and argue that “...the human body should be understood as an ecosystem with multiple ecological niches and habitats in which a variety of cellular species collaborate and compete...” Rising above “the essentialistic promotional metaphors” of the Human Genome Project, the HMP challenges a reductionistic view of genetics and proposes instead “...a post-modern understanding of the individual itself as a pragmatic construction that we project upon a much more complex system.” The authors suggest we develop metaphors of metagenomics to accurately translate the message of microbiomics if we are to truly benefit from “translational genomic research.”

Keller, Evelyn Fox. *The Century of the Gene*. Cambridge, MA: Harvard University Press, 2000. 186 p.

Keller, professor of History and Philosophy of Science at MIT, reminds us that “[f]or almost fifty years, we lulled ourselves into believing that, in discovering the molecular basis of genetic information, we had found the “secret of life”...but now, with the call for functional genomics, we can read at least a tacit acknowledgment of how large the gap between genetic “information” and biological meaning really is.” She calls for an appreciation of the complexity of biology, and advises that “gene talk” - the meanings of words used in discussing genetics - should be scientific and exact in usage.

Keenan, James F. **Genetic Research and the Elusive Body**. In *Embodiment, Morality, and Medicine*, pp. 59-73. Edited by L. Sowle Cahill and M.A. Farley. Boston: Kluwer, 1995.

Keenan claims that by failing to distinguish between the human genome and the human body, scientists dodge the question “After mapping out the nearly 10,000 genes in our code, will we know a human body? Is a body simply the genetic code?” When charged with reductionism, scientists will respond that “the genome” is distinct from “the person” instead of different from “the body,” thus objectifying the latter. The author sees the objectification of the human body through genetics as a moral threat which must be challenged. “In the field of genetics, the discoveries of the human body as relational and as intergenerational further our understanding of the body not as matter or object, but as

disposed to being subject... through genetics we find in the human body the histories of ancestors encoded and the opportunities of our posterity forecasted. Our bodies call us to treat ourselves and our neighbor, then, as sharing identities."

Kent, Theodore C. *Mapping the Human Genome: Reality, Morality, and Deity*. Lanham, MD: University Press of America, 1995. 129 p.

As a psychologist, the author discusses the roles that "harmony" and "struggle" play in our desire to redesign ourselves. Kent also describes how the works of Nietzsche and the life of Christ "both present perceptions of human nature and ways to improve it" that can "help us become aware of some of our options in the Age of the Genome."

Lammers, Ann; and Peters, Ted. [Genethics: Implications of the Human Genome Project](#). *The Christian Century* 107(27): 868-72, October 3, 1990.

With new possibilities arising from the ability to reorder our genetic code, Lammers and Peters concur that we need to look at the relationship between divine and human agency and ask who is responsible for the transformation of the human race. The article points out various ethical issues and raises vital questions to be addressed.

Murphy, Timothy F. **The Genome Project and the Meaning of Difference**. In *Justice and the Human Genome Project*, pp. 1-13. With Marc A. Lappé. Berkeley: University of California Press, 1994.

Murphy asks of genome research: "What is the moral argument to be offered that the suffering of people here and now can be sacrificed to expected benefits in the future?" He goes on to point out that, as "big science", the goal of the Human Genome Project is to consolidate all research into "a single way of representing genetic information." Since "...there are many ways to represent the nature of human beings, and none [is] value neutral...even a genomic characterization is already always determined by our social and conceptual background." Murphy cautions that "given the lessons of history, it is not even clear that we should aspire to the effecting of all things possible...on the contrary...we [should] find what ways there are in the use of research projects...to preserve the lessons of difference."

Shinn, Roger Lincoln. *The New Genetics: Challenges for Science, Faith, and Politics*. Wakefield, RI: Moyer Bell, 1996. 175 p.

Citing Alfred North Whitehead's observation that "every philosophy is tinged with the colouring of some secretive imaginative background, which never emerges explicitly into its trains of reasoning," Shinn proposes "building some speed bumps on the road that prescribers travel...[by] challenging all the contestants in furious ethical controversies." The author addresses such issues as the power of ideology to shape scientific research, the responsibility of religious communities to engage in public policy debates, and the potential for gene therapy to simultaneously heal, enhance, and distort the human body.

Sleeboom, Margaret, ed. *Genomics in Asia: a Clash of Bioethical Interests?* London/New York: Kegan Paul; Distributed in the U.S. by Columbia University Press, 2004. 321 p.

This anthology provides an overview of the diversity of religious, philosophical and cultural attitudes toward genetic research in Asian countries. Chapters focus on topics such as Hindu bioethics and eugenics, Confucian bioethics and genetic intervention, and Islamic attitudes toward cloning.

Strohman, Richard C. **Ancient Genomes, Wise Bodies, Unhealthy People: Limits of a Genetic Paradigm in Biology and Medicine.** *Perspectives in Biology and Medicine* 37(1): 112-145, Autumn 1993.

The author argues that conceiving of genes interacting with the environment as a dualism does not do justice to the complexity of genetic interactions. Rather, the system is one of epigenetic regulation: "...[i]ndividual cells, groups of cells, and whole organisms can absorb information (including mutation) and ...rearrange interactive genetic and metabolic pathways to produce new phenotypes in response to the environment." Strohman exhorts genetic researchers not to focus "...on genetic engineering designed to fit the organism to an increasingly hostile environment, but on environmental engineering designed to refit or return the world to a state consistent with evolved, stable genomic and epigenetic capability."

IX. Search Strategies for the Human Genome Project

These search strategies reflect the status of the respective databases as of April 2011.

Database: [Academic Search Premier](#) (Private Database: EBSCO Industries, Inc.)

Search Strategy:

1st query box: human genome project [*select **Subject Terms** from the right drop-down menu*]

2nd query box: OR human genome project [*select **Author-Supplied Keywords** from the right drop-down menu*]

3rd query box: OR "human genome project" [*select **Title** from the right drop-down menu*]

Database: [ETHXWeb](#) (Public Database: Bioethics Research Library at Georgetown University)

Search Strategy: (15.10[pc] or (*human genome project))

Database: [Google Scholar](#) (Public Database: Google)

Search Strategy:

exact phrase Human Genome Project

at least one legislation or privacy or confidentiality or discrimination or
policy or religion or ethics or ethical or bioethics or bioethical

Database: [JSTOR](#) (Private Database: ITHAKA)

Search Strategy:

1st query box: “human genome project” [*retain full-text*]

2nd query box: AND (legislation or privacy or confidentiality or discrimination or policy
or religion or bioethics or bioethical or ethical or ethics)
[*retain full-text*]

Database: [LexisNexis Academic](#) (Private Database: Reed Elsevier, Inc.)

Search Strategy:

Combined Search “human genome project” [check **Law Reviews**]

Database: [NLM Book Catalog](#) (Public Database: U.S. National Library of Medicine)

Search Strategy: (human genome project [majr] OR human genome project [ti])

Database: [ProQuest](#) (Private Database: Cambridge Information Group)

Search Strategy:

1st query box: (human genome project) [*select **Subject** from the right drop-down menu*]

2nd query box: AND (legislat* or privacy or confident* or policy or religion)

[select **Subject** from the drop-down menu]

3rd query box: OR (bioethic* or ethic*) [retain **Citation and abstract**]

Database: [PubMed](#) (Public Database: U.S. National Library of Medicine)

Search Strategy:

((human genome project [majr] or human genome project [ti]) AND bioethics[sb])

Database: [WorldCat](#) (Public Database: OCLC Online Computer Library Center)

Search Strategy: "human genome project"

Note: for a focused search, select Subject from the drop-down menu.

Human Genome Project was originally authored in 1991 by Sharon J. Durfy, a Postdoctoral Fellow at the Kennedy Institute of Ethics, and Amy E. Grotevant, a Research Assistant at the Bioethics Research Library (BRL) and published in the *Kennedy Institute of Ethics Journal*, Vol. 1, No. 4, pp. 347-362, December, 1991. This publication has been updated periodically by BRL staff members Martina Darragh, Harriet Gray, Susan Poland, and Kathleen Schroeder. Charles "Nat" Norton contributed annotations for materials from the Robert Cook-Deegan Genome Collection at the Kennedy Institute of Ethics. Maddalena Tilli Shiffert, Assistant Professor, Department of Biology, Georgetown University, was a scientific advisor on this project in 2011, when this bibliography was last updated. Also in 2011, Katherine L. Record and Melissa K. Bourne of the Georgetown University Law Center, developed search strategies to access the legal literature; these are incorporated into each Scope Note on genetics, along with strategies for other disciplines designed by BRL staff.

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