



National Reference Center for Bioethics Literature  
The Joseph and Rose Kennedy Institute of Ethics  
Box 571212, Georgetown University  
Washington, DC 20057-1212  
888-BIO-ETHX; 202-687-3885; fax: 202-687-6770  
e-mail: [bioethics@georgetown.edu](mailto:bioethics@georgetown.edu)  
<http://bioethics.georgetown.edu/>

## SCOPE

## NOTE

# The Human Genome Project

17

### Table of Contents

- [Background Essay](#)
- [I. General Surveys](#)
- [II. The Debate](#)
- [III. U.S. Project History](#)
- [IV. International Involvement](#)
- [V. The Human Genome Map](#)
- [VI. Ethical, Legal, and Social Issues](#)
- [VII. Position Statements](#)
- [VIII. Additional Information Resources](#)
- [Get a Quick Bib for Recent Literature on this Topic](#)

*First published in December 1991, Scope Note 17 is an annotated bibliography with links to electronic texts and/or sites where possible. It is updated on a periodic basis.*

To commemorate the [50th anniversary](#) of the discovery of DNA's structure, the human genome sequence and the Human Genome Project were declared complete on April 14, 2003. Using the "working draft" announced on February 12, 2001 (see [Science: Human Genome Special Issue](#)), scientists from around the world (the [International Human Genome Sequencing Consortium](#)) put draft fragments in order so that researchers could use the data "out of the box" without additional sequencing. "If you are looking for a disease gene, you can be confident that it exists in one continuous stretch of highly accurate sequence," said Dr. Francis Collins, director of the genome center at the U.S. National Institutes of Health ([III. Wade 2003](#)). The current [vision](#) for using the completed sequence flows along three themes, and includes "grand challenges" such as developing a catalog of genetic variation ([International HapMap Project](#)). ([V. Collins et al. 2003](#)) Online resources such as *Nature* magazine's [OmicsGateway](#) (which includes [A User's Guide to the Human Genome](#)) ([V. Wolfsberg et al. 2002](#)), the U.S. Department of Energy's [DOEgenomes.org](#), and the U.S. National Human Genome Research Institute's [genome.gov](#) help scientists and the general public alike to track the status of these challenges. The ultimate success of the Human Genome Project resides in its celebration of the complexity of biology. "It is a rare and wonderful moment when success teaches us humility...[i]ndeed, of all the benefits that genomics has bequeathed on us, this humility may ultimately prove to have been its greatest contribution." ([V. Keller, 2000](#))

### Background Essay

Scientists throughout the world have embarked upon a long-term biological investigation that promises to revolutionize the decisions people make about their lives and lifestyles, the way doctors practice medicine, how scientists study biology, and the way we think of ourselves as individuals and as a species. It is called the Human Genome Project, and its ultimate goal is to map and determine the chemical sequence of the three billion nucleotide base pairs that comprise the human genome.

These three billion base pairs include an estimated 50,000 to 100,000 genes. The rest of the genome - perhaps 95 percent of it - is nongenic sequences with unknown function, sometimes called "junk." Determining the order and organization of all this material has been likened to tearing six volumes of the Encyclopaedia Britannica into pieces, then trying to put it all back together to read the information ([I, Hall 1990](#)). The effort could be well worth it, many scientists say, because it is expected to yield major insights into many common and complex diseases, including cancer, cardiovascular disease, and Alzheimer's disease ([II, Dulbecco 1986; Koshland 1989](#)).

The Human Genome Project is not without controversy, however ([See II, The Debate](#)). Many scientists fear that funding for it will be diverted from other areas of research, rather than obtained from new funding sources. This has enlivened the debate about the relative value of "big" versus "small" science. Also, the value of undertaking a complete sequencing of the genome has been questioned, especially given the high proportion of nongenic sequences.

Advocates of the effort converted many critics by making two alterations in the original plan. Plans were included to simultaneously determine the nucleotide sequence of the genomes of other organisms; this provides comparisons and points of reference for the human sequence ([III, NIH/DOE 1990](#)). Second, in response to concerns about the high cost of developing technology to sequence the whole genome, the focus moved from large-scale sequencing to mapping the genome, which would hasten the search for disease genes ([III, NIH/DOE 1990](#)).

The ideal map would be both *genetic*, locating DNA markers, or signposts, at closely spaced intervals along the chromosomes, and *physical*, indicating the exact distance between these markers ([V, McKusick 1991](#)). ([See VIII, Additional Information Resources](#), for descriptions and access points for genome mapping databases).

## HISTORICAL BACKGROUND OF THE UNITED STATES EFFORT

The first serious discussions about sequencing the entire human genome occurred at a workshop at the University of California at Santa Cruz in 1985 ([III, Sinsheimer 1989](#)). A second workshop, organized by the U.S. Department of Energy (DOE) and held in March 1986, addressed the feasibility of an organized program ([III, DOE 1986](#)). Shortly thereafter, DOE instituted its own genome project ([III, DOE 1987](#)). Reports in early 1988 from both the National Research Council (NRC) of the National Academy of Sciences (NAS) and Congress' Office of Technology Assessment, (OTA) ([III, NRC 1988; OTA 1988](#)) served as catalysts, and in fiscal year 1988, the U.S. Congress officially launched the Human Genome Project by appropriating funds to both the Department of Energy and the National Institutes of Health (NIH).

To avoid potential congressional "meddling" ([III, Roberts 1988](#)), NIH and DOE drafted a memorandum of understanding for interagency coordination in October 1988 ([III, NIH/DOE 1990](#)). The agencies then created both separate and joint committees, and working groups to administer the project. NIH established the Office of Human Genome Research in 1988 (directed by James D. Watson) to plan and coordinate NIH genome activities. That office evolved first into the National Center for Human Genome Research (NCHGR), an independent funding unit with authority to award grants and contracts ([III, Watson 1990](#)), and subsequently into the National Human Genome Research Institute (NHGRI). Institute status at the National Institutes of Health is necessary for NHGRI's director to coordinate genome research with other projects at NIH. ([III, Human Genome News, 1997](#))

## INTERNATIONAL INVOLVEMENT

International involvement has increased steadily over the life of the project. The programs of the countries involved vary, depending on their particular concerns, capabilities and funding. ([IV, McLaren 1991](#)).

The European Community also has played a role in the Human Genome Project. In 1988, the EC introduced a

proposal entitled the "Predictive Medicine Programme." A few EC countries, notably Germany and Denmark, claimed the proposal lacked ethical sensitivity; objections to the possible eugenic implications of the program were especially strong in Germany ([IV, Dickson 1989](#)). The initial proposal was dropped but later modified and adopted in 1990 as the "Human Genome Analysis Programme" ([IV, Dickman and Aldhous 1991](#); [Ferguson-Smith 1991](#)). This program committed substantial resources to the study of ethical issues ([VII, Council of the European Communities 1990](#); see also [VII, Council of Europe](#), 1996).

The need for an organization to coordinate these multiple international efforts quickly became apparent. Thus the Human Genome Organization (HUGO), which has been called the "U.N. for the human genome," was born in the spring of 1988. Composed of a founding council of scientists from seventeen countries, HUGO's goal is to encourage international collaboration through coordination of research, exchange of data and research techniques, training, and debates on the implications of the projects ([IV, Bodmer 1991](#)).

It has been suggested that the Human Genome Project will benefit not only industrialized countries but developing nations as well ([IV, Watson and Cook-Deegan 1990](#); [Allende 1991](#)). In an effort to include these countries in the project and discussions about it, UNESCO assembled an advisory committee in 1988, to examine UNESCO's role in facilitating international dialogue and cooperation ([IV, Grisolia 1991](#); see also [VII, UNESCO 1997](#)).

### **ETHICAL, LEGAL AND SOCIAL ASPECTS**

The Human Genome Project has generated widespread interest in a large spectrum of questions regarding the ethical, legal, and social implications of the existence and use of human genetic sequences. Many groups concur that sequencing the human genome does not raise ethical questions in and of itself ([VII, Position Statements](#)). Questions center on how the information is used and its implications for individuals, society and the human species ([VI, Annas 1990](#)).

It is generally accepted that the questions raised by the accessibility of and increase in information generated by the Human Genome Project are not new ([VI, Macklin 1985](#); [Murray 1991](#)). Rather, the volume, variety, and the ease with which such information can be obtained, and the vast number of people affected, make it imperative that the possible use or misuse of the project's findings be anticipated and addressed ([VII, Position Statements](#)).

The ability to generate any individual's genetic profile raises important questions of privacy, confidentiality, ownership and autonomy. How should information be protected? Who should have access to the information and under what circumstances? What rights, if any, do employers, insurers, and family members have to an individual's genetic information?

Diagnosis of many genetic disorders will be possible before treatment becomes available. How will we resolve dilemmas raised by such a gap? ([VI, Juengst 1991](#)).

In addition, the potential trend towards understanding diseases or traits in genetic terms may raise questions about human nature and our fundamental beliefs about ourselves and our species ([VI, Charlesworth 1990](#); [Lammers and Peters 1990](#); [Murray 1991](#)). Will we view an individual as merely a product of interacting genes? How will we define normalcy, abnormalcy, or disability? ([VI, Annas 1990](#); [Capron 1990](#)).

The recognition of the importance of such questions, and the increased dialogue surrounding them has resulted in an international commitment to and interest in addressing issues of ethical, legal, and social import ([VI, Capron 1991](#)). Many nations and organizations participating in the Human Genome Project have allocated funding and/or assembled committees to address such concerns ([VI, Aldhous 1991](#)). It is likely that these initial efforts will increase as national programs and the number of countries involved expands.

Below is a sample of the literature on social, political, and ethical aspects of the Human Genome Project, and a brief listing of references on human genome mapping.

---

## 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 a 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.

Cook-Deegan, Robert Mullan. Genome Mapping and Sequencing. In *Encyclopedia of Bioethics*, Revised Edition, ed. Warren T. Reich, pp. 1011-20. New York: Simon & Schuster Macmillan, 1995.

This entry reviews the concept of genome mapping, international development of genome research, parallel programs for ethical review of genetic research, forensic uses of genetic markers, and patenting law as it pertains to genetics.

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..." (p. xiii). 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".

Hall, Stephen S. **James Watson and the Search for Biology's 'Holy Grail'**. *Smithsonian* 20(11): 40-49, February 1990.

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

Containing essays which 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."

McVey, Patrician Gail. **Human Genome Project**. In *Encyclopedia of U.S. Biomedical Policy*, Robert H. Blank and Janna C. Merrick, eds., pp. 128-30. Westport, CT: Greenwood Press, 1996.

This synopsis of the founding of the Human Genome Project includes brief descriptions of the biological, political, legal, and scientific components of genome research. The volume also contains entries on related topics such as genetic engineering and screening.

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.

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 prize winner Watson were written over a period of 30 years and include many of the introductory essays included in the Cold Spring Harbor Laboratory's annual reports. 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.

Wexler, Barbara. **The Human Genome Project**. In *Genetics and Genetic Engineering*. Detroit, Michigan: Thomson Gale, 2006. pp. 99-116.

This volume is one of the *Information Plus Reference Series* which is geared to high school and undergraduate students. Background information about the scientists and the techniques they developed is provided. Also included is the history of the Project along with funding issues and the international response. The Haplotype Mapping Project, an international effort to identify genetic variants within a chromosomal area, is described. The chapter contains many diagrams and tables.

## II. THE DEBATE

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.

Keenan, James F. **Genetic Research and the Elusive Body**. In *Embodiment, Morality, and Medicine*, eds. L. Sowle Cahill and M.A. Farley, pp. 59-73. 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 that 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, Thodore 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."

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."

Murphy, Timothy F. **The Genome Project and the Meaning of Difference**. In *Justice and the Human Genome Project*, with Marc A. Lappe, pp. 1-13. 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."

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.

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.

### III. U.S. PROJECT HISTORY

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 survivors of the Hiroshima and Nagasaki bombings, Barnhart, manager of DOE's genome program, explains. This paper traces from DOE's

initial interest to the important 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.

Cantor, Charles. **Orchestrating the Human Genome Project.** *Science* 248(4951): 49-51, 6 April 1990.

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.

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.

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*. Edited by Kathi E. Hanna. Washington, DC: National Academy Press, 1991, pp. 99-175.

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.

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 here collect over 100 of these essays 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. In this work, he details the finances, the scientific steps and the characters involved in mapping the human genome. The author has a Ph.D. in genetics and brings his expertise to the work, including interviews with geneticists Francis Collins and Craig Venter.

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.

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.

National Research Council. *Mapping and Sequencing the Human Genome*. Washington: National Academy Press, 1988.

One of the two landmark U.S. reports that gave momentum to the genome project, this study outlines its goals and recommendations as well as the major issues, including: 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.

[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.

Reardon, Jenny. *Race to the Finish: Identity and Governance in an Age of Genomics*. Princeton, NJ: Princeton University Press, 2005. 237 p.

The author presents a history of the Human Genome Diversity Project (HGDP) and the scientific and social issues it encountered. Chapters focus on community consent for population research, the working relationships between HGDP's anthropologists and geneticists, and ethical issues with population genetics.

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

David Galas, then DOE's associate director of health and environmental research, turned around DOE'S genome program, according to Roberts. Under Galas, DOE has broadened its role in the genome project, involving itself more in the science and taking a more interdisciplinary approach.

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.

U.S. Congress. Office of Technology Assessment (OTA). [Federal Technology Transfer and the Human Genome Project](#). Washington, DC: GPO, 1995. 118 p.

Observing that judicial and legislation 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.

U.S. Congress. Office of Technology Assessment (OTA). [Mapping Our Genes: Genome Projects: How Big, How Fast?](#) Washington: U.S. Government Printing Office, 1988.

This second major report, released in 1988, was written in response to U.S. Congressional interest and concern. The report, offers a comprehensive analysis of the U.S. genome project and the surrounding issues, especially organization and resource allocation, in an effort to present Congress with options for U.S. involvement and direction in the project.



U.S. Department of Energy. *Human Genome 1989-90 (Program Report) March 1990*. Springfield, VA: National Technical Information Service, March 1990.

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

U.S. Department of Energy. 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.

U.S. 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.

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.

U.S. 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.

U.S. Department of Energy. 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 benefits. Recommendations from this meeting fueled interest and action on a DOE human genome project.

U.S. 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.

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.

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?"

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 such that it can now be called operationally complete.

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.

Watson, James D. **The Human Genome Project: Past, Present and Future.** *Science* 248(4951): 44-49, 6 April 1990.

The director of the National Center for Human Genome Research, NIH, describes the development of the Human Genome Program, tracing it from the "1973 birth of the recombinant DNA revolution," through the 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.

### **Additional References**

Baltimore, David. **Our Genome Unveiled** *Nature* 2001 February 15; 409(6822): 814-816

Davenport, R. John; Pennisi, Elizabeth; Marshall, Eliot; Roberts, Leslie; and Helmuth, Laura. **A History of the Human Genome Project.** *Science* 2001 February 16; 291(5507): 1195-1200

International Human Genome Mapping Consortium. **A Physical Map of the Human Genome.** *Nature* 2001 February 15; 409(6822): 934-941

Marshall, Eliot. **Genome Teams Adjust to Shotgun Marriage [news].** *Science* 2001 June 15; 292(5524): 1982-1983

### **IV. INTERNATIONAL INVOLVEMENT**

Cavalli-Sforza, Luigi Luca. **The Human Genome Diversity Project.** In *Actes 1995 - Proceedings*, Vol. 2, p. 71-83. International Bioethics Committee of UNESCO, 1995.

Describing the Human Genome Diversity Project (HGDP) as an "international anthropology project that seeks to study the genetic richness of the entire human species," the author provides a history of the HGDP; descriptions of early planning meetings at Stanford (US/CA), Pennsylvania State University (US/PA), Alghero (Sardinia, Italy), and Bethesda (US/MD); and a review of HGDP's methodology and guidelines for DNA banking and participant remuneration. Cavalli-Sforza also responds to critics who suggest that the HGDP is a racist endeavor by saying that "...our discipline, population genetics...has given the best proofs we have that racism is wrong."

Centro de Direito Biomedico, Faculdade de Direito, Universidade de Coimbra, [and] Institut fur Artz und Arzeneimittelrecht Universitat Gottingen. **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, address 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.

Human Genome Organization. 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 recommendations on informed consent, privacy, and oversight of genome research.

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.

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

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.

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

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.

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 flavor 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. Online. Human Genome Organization (HUGO). Available: <http://hugo.gdb.org/bermuda.htm>.

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 act as the coordinating agency for these efforts. Also available from the same URL is a summary of the Second International Strategy Meeting held February 27 - March 2, 1997.

## **Additional References**

Alberts, Bruce, and Klug, Aaron. **"The Human Genome Itself Must Be Freely Available to All Humankind"** [statement of Bruce Alberts, president of the U.S. National Academy of Sciences, and Sir Aaron Klug, president of the Royal Society of London]. *Nature* 2000 March 23; 404(6776): 325

Allende, Jorge E. **A View from the South**. *FASEB Journal* 5(1): 6-7, January 1991.

Bodmer, Walter F. **HUGO: The Human Genome Organization**. *FASEB Journal* 5(1): 73-74, January 1991.

Cross, Kath. **Framing Whiteness: The Human Genome Diversity Project (As Seen on TV)**. *Science as Culture* 2001 September; 10(3): 411-438

Dodson, Michael, and Williamson, Robert. **Indigenous Peoples and the Morality of the Human Genome Diversity Project**. *Journal of Medical Ethics* 1999 April; 25(2): 204-208

Dickman, Steven; and Aldhous, Peter. **Helping Europe Compete in Human Genome Research**. *Nature* 350(6316): 261, 28 March 1991.

Dickson, David. **Genome Project Gets Rough Ride in Europe**. *Science* 243(4891): 599, 3 February 1989.

Ferguson-Smith, M.A. **European Approach to the Human Gene Project**. *FASEB Journal* 5(1): 61-65, January 1991.

Grisolia, Santiago. **UNESCO Program for the Human Genome Project**. *Genomics* 9(2): 404-5, February 1991.

Human Genome Organization [HUGO]. Ethics Committee. **Genetic Benefit Sharing [editorial]**. *Science* 2000 October 6; 290(5489): 49

**Rules of Genome Access [editorial]**. *Nature* 2000 March 23; 404(6776): 317

Salzano, Francisco M. **Global Human Diversity and Ethics**. *Politics and the Life Sciences* 1999 September; 18(2): 330-332

Taylor, Allyn L. **Globalization and Biotechnology: UNESCO and an International Strategy to Advance Human Rights and Public Health**. *American Journal of Law and Medicine* 1999; 25(4): 479-541

## **V. THE HUMAN GENOME MAP**

Collins, Francis S.; Green, Eric D.; Guttacher, 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 HapMap Project, a catalog of the heritable variation in the human 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 genetics, and electronic publishing of sequence data.

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.

Dib, Colette; Faure, Sabine; Fizames, Cecile; Samson, Delphine; Drouot, Nathalie; Vignal, Alain; Millasseau, Philippe; Marc, Sophie; Hazan, Jamile; Seboun, Eric; Lathrop, Mark; Gyapay, Gabor; Morissette, Jean; and Weissenbach, Jean. **A Comprehensive Genetic Map of the Human Genome Based on 5,264 Microsatellites**. *Nature* 380: A1-A138, March 14, 1996.

This special issue presents the final version of the Genethon human genetic linkage map described by the authors in the March 14, 1996 issue of *Nature* on pages 152-154.

Drews, Jurgen. **Genomic Sciences and the Medicine of Tomorrow**. *Nature Biotechnology* 14(11): 1516-1518, November 1996.

Originally given as a talk at the symposium "Genetic Basis of Human Disease" held in Basel, Switzerland, October 1996, this article describes the impact of genome research on drug development. Drews predicts that the identification of complex disease phenotypes for conditions such as hypertension and diabetes will enable pharmaceuticals to produce medicines targeting areas "that are the least represented in the current state of drug development".

Finkel, Elizabeth. **The Post-Genome Era: Medical Promise with Problems**. *The Lancet* 349: 1228, 26 April 1997.

Reviewing the status of the Human Genome Project ten years after its inception, the author describes the use of "expressed sequence tags", or ESTs, for reducing the time needed to identify disease-causing genes from 10 years to 6 weeks. With the rapid progress of gene mapping, Finkel suggests that the next focus for genetic research will be "functional genomics - i.e., how genes work together to make organisms."

**The Human Genome**. *Science* [Special Issue] 291 (5507) February 16, 2001. [Online at:<http://www.sciencemag.org/content/vol291/issue5507/>].

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 makes a portion of it available online at no charge. Sections provide a timeline for the mapping of various genomes, an overview of the process, and instructions on how to use the human genome sequence data maintained in the public domain.

Goldstein, David B., and Cavalleri, Gianpiero L. **Understanding Human Diversity**. *Nature* 437(7063): 1241-1242, October 27, 2005. Available online: <http://www.nature.com/nature/journal/v437/n7063/pdf/4371241a.pdf>

Describing the current state of genomic research "as a sort of awkward adolescence", the authors provide a context for the publication of the first phase of the HapMap's compilation of genetic variation across populations. The report to which they refer, *A Haplotype Map of the Human Genome*, can be accessed online: <http://www.nature.com/nature/journal/v437/n7063/pdf/nature04226.pdf>

Joyce, Christopher. **The Race to Map the Human Genome**. *New Scientist* 113(1550): 35-39, 5 March 1987.

Joyce compares various strategies in developing a physical map of the human genome and briefly discusses some of the technology involved.

Lander, Eric S. **The New Genomics: Global Views of Biology**. *Science* 274(5287): 536-539, October 25, 1996.

Confident that technological advances will ensure that 100 percent of the human genome will be sequenced by 2005, Lander speculates about which scientific tasks in genome research will be tackled thereafter. His "wish list" includes: 1) routine re-sequencing of multi-megabase regions of both human and mouse DNA; 2) systematic identification of all common variants in human genes; 3) rapid *de novo* sequencing of other organisms; 4) simultaneous monitoring of the expression of all genes; 5) generic tools for manipulating cell circuitry; 6) monitoring the level and modification state of all proteins; 7) systematic catalogs of protein interactions; and 8) the identification of all basic protein shapes.

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 author of *Mendelian Inheritance in Man*, presents an historical overview and summary of the status of the human genetic map. He also explains the methods by which scientists incorporate and access genetic map data and emphasizes the importance of genetic mapping in biology and medicine.

**Omics Gateway**. [Online at: <http://www.nature.com/genomics/>].

*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) featuring [\*A User's Guide to the Human Genome\*](#) (a supplement to *Nature Genetics*, September 2002.)

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..."

## VI. ETHICAL, LEGAL, AND SOCIAL ISSUES

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.

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.

Annas, George J. **Mapping the Human Genome and the Meaning of Monster Mythology**. *Emory Law Journal* 39(3): 629-64, Summer 1990.

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 finally suggests strategies for regulation of genetic technology.

Bentley, David R. **Genomic Sequence Information Should Be Released Immediately and Freely in the Public Domain**. *Science* 274(5287): 533-534, October 25, 1996.

Bentley details the rationale behind the "Bermuda Statement" issued by those attending the first International Strategy Meeting on Human Genome Sequencing [See International Involvement: 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.

Caplan, Arthur L. **Mapping Ourselves**. In: *Smart Mice, Not So Smart People*. Lanham, Maryland: Rowman & Littlefield Publishers, 2007. pp. 119-138.

This section of Caplan's book confronts the ethical dilemmas involved in the mapping of the human genome. His ethics of genome mapping include discussions of genetic privacy and genetic discrimination. He argues that public policy has not kept up with advances in genome mapping.

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

Capron describes a meeting of the NIH Ethical, Legal and Social working group held in June, 1991. This meeting gathered scientists, physicians and philosophers from fifteen countries to consider genetic ethics issues of international significance, such as insurance, employment, forensics, and the role of "under-represented" nations. A Global Steering Committee on Ethical and Social Issues in Genome Research was established.

Capron, Alexander Morgan. **Which Ills to Bear?: Reevaluating the 'Threat' of Modern Genetics**. *Emory Law Journal* 39(3): 665-96, Summer 1990.

Capron contends that we have misdirected our fears at the threat of genetic engineering (gene therapy) and the Human Genome Initiative, when we should be more concerned with the issues raised by genetic screening. The author gives special attention to the various types of screening and how it relates to insurability and employability, and suggests regulatory measures along with a general reworking of how we think about normality and abnormality.

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. "The 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" (p.16)

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

Human genome analysis requires that we develop criteria that will enable us to decide which kinds of genetic manipulation should be allowed to enhance life, and which kinds should not be allowed. The author asserts that we must have a clear picture of human nature that should not be derived from genetics alone, in order to make these decisions.

Clayton, Ellen. **Ethical Concerns in HapMap Project**. In: *Protecting Human Subjects* 2006 Spring; (13): 9-11.

This article outlines the ethical concerns in the HapMap Project. It is not possible to identify individual samples, so individual privacy is not at risk. However, samples are labeled by population, and there are concerns that discrimination or stigmatization could occur based on generalizing those haplotype findings to the entire population. Community advisory groups are being used to provide support and community input.

Clayton, Ellen Wright; Steinberg, Karen K; Khoury, Muin J.; Thomson, Elizabeth; Andrews, Lori; Kahn, Mary Jo Ellis; Kopelman, Loretta M.; and Weiss, Joan O. **Informed Consent for Genetic Research on Stored Tissue Samples.** *JAMA Journal of the American Medical Association* 274(22): 1786-1792, December 13, 1995.

The authors participated in a workshop jointly sponsored by the National Institutes of Health (NIH) and the Centers for Disease Control and Prevention (CDC) in July, 1994, to examine the problems involved in collecting tissue samples for use in genetic research. They make recommendations regarding informed consent and the use of both anonymous and identified samples, disclosure of research results to donors, public health investigations of disease clusters.

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*, eds. Theresa Marteau and Martin Richards, pp. 235-48. 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.

Earley, Charles L., and Strong, Louise C. **Certificates of Confidentiality: A Valuable Tool for Protecting Genetic Data.** *American Journal of Human Genetics* 57(3): 727-731, September 1995.

The authors suggest that certificates of confidentiality developed by the Department of Health and Human Services to protect participants in drug abuse studies also could be valuable for maintaining confidentiality in genetic research.

Eisenberg, Rebecca S. **Patenting the Human Genome.** *Emory Law Journal* 39(3): 721-45, Summer 1990.

Eisenberg relates the basic requirements of patent law (the subject matter must be new, useful, and nonobvious), to the patenting of human DNA sequences. She details the relevant court decisions, defines circumstances under which human DNA sequences might be patentable, and also addresses some pertinent public policy considerations.

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

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](#).

Goldworth, Amnon. **Informed Consent in the Human Genome Enterprise.** *Cambridge Quarterly of Healthcare Ethics* 4(3): 296-303, Summer 1995.

After reviewing the development of informed consent in the United States, Goldworth discusses the advantages of using the subjective standard of disclosure in genetic research because it is "the only one that protects all parties from invalid informed consent in which imparted information is either not absorbed or is not of material interest to the patient."

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*, eds. Ruth E. Gulger, Elizabeth M. Bobby, and Harvey V. Fineberg, pp. 432-457. 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."

Juengst, Eric T. **The Human Genome Project and Bioethics**. *Kennedy Institute of Ethics Journal* 1(1): 71-74, March 1991.

Juengst, director of the Ethical, Legal and Social Implications Program of the National Center for Human Genome Research at NIH describes "the largest public investment in bioethical analysis to date". Citing the most immediate consequence of genome research as the development of diagnostic tests, well before development of therapeutic answers, Juengst explains the need for a bioethics program to deal with the resulting issues.

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.

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 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.

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." (pp. 7-8) 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.

Koski, Cheryl A. **The Human Genome Project: An Examination of its Challenge to the Technological Imperative**. *New Genetics and Society* 2005 December; 24(3): 265-2811.

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."

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

Lappé, Marc. **The Limits of Genetic Inquiry**. *Hastings Center Report* 17(4): 5-10, August 1987.

Lappé, addressing the boom in genetic knowledge, asks what limits, if any, should be imposed on acquisition, who should control it and how it should be used. Lappe stresses caution and the necessity for integrating this knowledge with sensitivity to ethical principles.

Macer, Daryl. **Whose Genome Project?** *Bioethics* 5(3): 183-211, July 1991.

Looking at many angles of the genome project, Macer answers the question posed in the title, contending the genome belongs to everyone. The international nature of the project and its universally applicable results, make it one belonging to humanity, needing all of the views and concerns of humanity to shape and guide it.

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

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 Briody. *Genes, Women, Equality*. New York: Oxford University Press, 2000. 314 p.

The author writes that she gives ethical importance in reverse order of the title with equality first, then women, and then her recent study, genetics. She questions both the benefits and burdens to women offered by current genetic understanding. The 16 chapters in the work discuss genetics combined with topics such as: a feminist view, women in research and clinical genetics, allocation of services, testing for diseases, behavioral genetics, privacy, health insurance, cloning, parenthood, and cultural differences.

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 (see annotation for NCHGR-DOE Guidance on Human Subjects Issues in Large-Scale DNA Sequencing below.)

McGourty, Christine. **Public Debate on Ethics.** *Nature* 342(6250): 603, 7 December 1989.

This article describes proposed activities of the ELSI Working Group designed to address possible problems before they appear. Activities include: public debates and town meetings, focus groups that include professionals from various disciplines, and development of educational materials.

McLean, Margaret R. **Religion, Ethics, and the Human Genome Project.** In: *Science, Religion, and Society: An*

*Encyclopedia of History, Culture, and Controversy*. Edited by Arri Eisen and Gary Laderman. Armonk, NY: M.E. Sharpe, 2007, pp. 787-794.

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.

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."

Sleeboom, Margaret, ed. ***Genomics in Asia: a Clash of Bioethical Interests?*** London/New York: Kegal 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.

Topol, Eric J., Murray, Sarah S., and Frazier, Kelly A. **The Genomics Gold Rush**. *Journal of the American Medical Association* 298(2): 218-221, 11 July 2007.

This commentary article describes the recent discoveries in the genomics of complex traits. Haplotype maps (portions of DNA that are inherited as a unit) have shown that most of the base pair variations are inherited via 250,000 to 500,000 of the haplotypes. It is also now possible to analyze base pair variants called single nucleotide polymorphisms (SNPs) by rapid genotyping. The authors discuss the patenting of genes and the commercialization of gene markers. Molecular functional aspects of DNA variants appear to be more crucial than 'anatomic' roadmaps.

United States. National Institutes of Health [NIH] and Department of Energy. 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.

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".

U.S. 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.

U.S. 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.

U.S. National Center for Human Genome Research, and U.S. 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.

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.

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.

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 genetics."

## **Additional References**

Aldhous, P. **Who Needs a Genome Ethics Treaty?** *Nature* 351(6327): 507, 13 June 1991.

Hamel, Ron. **Genetics and Ethics - Issues and Implications of the Human Genome Project**. *Health Progress* 2001 March-April; 82(2): 22-23

Mahowald, Mary B.; McKusick, Victor A.; Scheuerle, Angela S., and Aspinwall, Timothy J., eds. ***Genetics in the Clinic: Clinical, Ethical, and Social Implications for Primary Care***. St. Louis: Mosby, 2001. 304 p.

Rothman, Barbara Katz. *The Book of Life: A Personal and Ethical Guide to Race, Normality, and the Implications of the Human Genome Project*. Boston: Beacon Press, 2001. 272 p.

## 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.

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. Draft 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 Bioethics](#). Strasbourg: Council of Europe, 1996. 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 germline 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). .

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.

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] and Department of Energy, National Human Genome Research Institute **NIH-DOE Guidelines for Access to Mapping and Sequencing Data and Material Resources**. <http://www.genome.gov/10000925>, August 17, 2007

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.

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. ADDITIONAL INFORMATION RESOURCES

[Bioethics Resources on the Web: Genetics Snapshot](#) provides links to directories, news sources, and teaching resources in genetics and ethics.

**National Information Resource on Ethics & Human Genetics** provides free bibliographic search assistance on topics relating to ethical and public policy issues in medicine and biomedical research. Searches are available upon request from the National Reference Center for Bioethics Literature (800-633-ETHX or 202-687-3885).

Email: [bioethics@georgetown.edu](mailto:bioethics@georgetown.edu)

The **National Reference Center for Bioethics Literature (NRCBL) Archives**, Georgetown University holds a collection of materials assembled by Robert M. Cook-Deegan, M.D. while preparing The *Gene Wars: Science, Politics and the Human Genome* (Norton, 1994) (See I. General Surveys). These materials, which are not indexed, include correspondence, memoranda, press reports, and transcripts of meetings and interviews relating to the Human Genome Project. To use these materials, contact NRCBL's archivist at 800-633-3849 or [bioethics@georgetown.edu](mailto:bioethics@georgetown.edu).

---

*The Human Genome Project* was first prepared for publication in 1991 by Sharon Durfy, a Postdoctoral Fellow at the Kennedy Institute during 1991, and Amy E. Grotevant, a former Research Assistant at the NRCBL. It continues to be updated by NRCBL reference staff members Martina Darragh, Harriet Gray, Anita Nolen, Susan Poland, and Kathleen Schroeder.

The National Reference Center for Bioethics Literature, Kennedy Institute of Ethics, Georgetown University is supported in part by contract NO1-LM-4-3532 with the National Library of Medicine, National Institutes of Health, and grant P41 HG01115 from the National Human Genome Research Institute, National Institutes of Health. Materials on this site are copyrighted and made available to individual researchers. Any redistribution or commercial use requires written permission of the [Institute](#).

---

*Last updated: October 2007*

**Return to [NRC Home Page](#)**