The Human Genome Project

BY REV. JAMES J. McCARTNEY, OSA, PhD

A map of the complete human genome—the focus of research by hundreds of scientists around the globe—will provide significant societal benefits in clinical medicine, gene therapy, and genetic counseling. However, because of the nature of the information acquired and its intimate relationship with living persons, care must be taken in the use of the data generated from the map. Ethical issues raised by advances in genetics include who should have access to genetic information, how they ought to use it, and how such information can be integrated into the practice of medicine.

The Genome Initiative

The Human Genome Project is a global activity coordinated by the Human Genome Organization. Its goals are to develop genetic marker maps of human chromosomes, to improve the technology of sequencing chromosomal DNA, and eventually to develop molecular maps of the approximately 3 billion base pairs of DNA of the complete human genome.

In the United States the National Institutes of Health, a part of the Department of Health and Human Services, has established the National Center for Human Genome Research (NCHGR) as the main coordinating agency for the genome initiative. The Department of Energy, "because of its long-standing interest in monitoring inherited damage caused by exposure to radiation and other environmental hazards,"[1] is also sponsoring grants for human genome research.

NCHGR works with other federal, private, and international organizations to increase our understanding of the genetic aspects of human health and disease, including the prevention and treatment of the 3,000 known inherited disorders and of genetically linked physiological reactions to external pathogens, toxins, and mutagens.[2] Over the next 15 years this commitment of the U.S. government is expected to become the largest investment it has ever made in bioethical research.

Fr. McCartney is associate professor, Department of Philosophy, Villanova University, Villanova, PA, and ethics consultant for the Allegany Health System and the Franciscan Sisters of the Poor Health System.

Implications

Unless one believes that all knowledge is interpretive and thus inherently related to power and control, it is accurate to say that scientific data in the abstract are amoral, since they are neither good nor evil, but rather true or false. Ethical concerns come into play in the acquisition and application of genetic information. With this in mind, NCHGR decided to commit approximately 5 percent of its annual budget to ethical, legal, and social science research related to its goals. To accomplish this objective, in 1990 NCHGR established its Program on Ethical, Legal, and Social Implications (ELSI) under the direction of Eric Juengst, PhD, a philosopher with a concentration in bioethics.

The purpose of ELSI, as stated in NCHGR's five-year plan, is to:

- Address and anticipate the implications for individuals and society of mapping and sequencing the human genome;
- Examine the ethical, legal and social sequelae of mapping and sequencing the human genome;
- Stimulate public discussion of the issues;
- Develop policy options to assure that the information is used for the benefit of the individual and society.[3]

Research Issues

ELSI is interested in three broad areas of research in considering proposals for possible funding. The first deals with professional policy issues, or "issues involved in the integration of new genetic tests into medical practice."[4] This area is primarily concerned with ethical, legal, and social issues involving the acquisition of genetic data, rather than its application, although the latter has some relevance here also.

Specific topics that could be explored under this area include the ethics of genetic testing and screening, benefit-cost ratios of genetic tests, the

ethics of scientific design, legal and ethical dimensions of research involving human subjects, standards of care for the use of genetic tests (i.e., who should receive tests and the results of tests to discover distinct gene-linked diseases), fair reimbursement policies for testing and test-related counseling, and the professional responsibilities of clinicians who perform the tests.[5]

The second area of possible ELSI funding deals with "issues of access to, and use of, genetic test results by third parties, including insurance providers, researchers, and employers"[6]—in other words, issues of access to genetic information by those outside the therapeutic community.

Some possible research topics are the scope and limits of medical privacy and confidentiality, the rights of at-risk persons to receive genetic information about potential spouses and other relevant persons, the amount of genetic information (if any) insurance underwriters should be allowed to access, whether they could exclude people from coverage on the basis of this information, whether genetic disease support groups could allow their registries to be used for research, whether genetic information is a legitimate basis for employment or educational discrimination, and whether military personnel and prisoners may be subject to genetic screening so that DNA "fingerprints" can be used as a form of identification.

The third set of questions of interest to ELSI deals with "issues involved in educating and counseling individuals about genetic test results"[7] or, more broadly, values and attitudes associated with genetic concepts and subsequent education of nonprofessionals about genetic issues. As the director of ELSI says:

> The primary risk that health professionals will face in using genetic tests is the misinterpretation of their findings and the resulting potential for psychological trauma, stigmatization, and discrimination. Sometimes out of ignorance, patients, families, and social institutions already stigmatize genetic disorders and treat those with them unfairly. This risk broadens as the genetic elements of more health problems are uncovered and gene-based tests for susceptibilities and carrier states are developed.[8]

Possible research proposals for this area of study could include the causes of stigmatization and discrimination, educational models to explain the nature and limits of genetic tests, projects "to clarify concepts of genetic susceptibility and draw out the social meaning"[9] for different groups, analysis of how our notions of "the good" and aesthetic value are formed and modified, and the relationship between inherited tendencies and the surrounding environment (nature versus nurture).

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Catholic Involvement

In the spirit of Abbot Gregor Mendel, the Augustinian priest and researcher considered to be the founder of genetic science, Catholic universities and healthcare systems should take the lead in researching the ethical implications of genetics. Many Catholic universities emphasize ethics and healthcare as integral parts of their curriculum, and these organizations seem a natural place for centers of ethics and genetics to be developed. Catholic healthcare systems are often involved in genetic counseling and would be natural leaders in developing grant proposals to study various facets of this activity (as indicated above). Finally, ethics committees at Catholic healthcare facilities should consider the implications of advances in genetics as they develop treatment policies and educational programs.

The Human Genome Project expects to develop fairly accurate molecular maps of the 46 human chromosomes in the next 15 years. During that time, theologians, philosophers, legal scholars, and social scientists must also be working to develop moral and legal "maps" to guide both the acquisition and the application of these important scientific data toward the true betterment of humankind. □

NOTES

1. Eric D. Green and Robert H. Waterston, "The Human Genome Project: Prospects and Implications for Clinical Medicine," *JAMA*, October 9, 1991, p. 1,967.
2. Eric T. Juengst, "The Human Genome and Bioethics," *Kennedy Institute of Ethics Journal*, March 1991, p. 71.
3. U.S. Department of Health and Human Services and U.S. Department of Energy, *Understanding Our Genetic Inheritance; The U.S. Human Genome Project: The First Five Years, FY 1991-1995*, National Technical Information Service, U.S. Department of Commerce, Springfield, VA, 1990.
4. Juengst, p. 72.
5. Juengst, p. 72.
6. Juengst, p. 73.
7. Juengst, p. 72.
8. Juengst, p. 72.
9. Juengst, pp. 72-73.