

Human Genome Project *Progress*

from the National Center for Human Genome Research, Office of Communications (301) 402-0911

November 1990

Background

NCHGR Bioethics Program Addresses Social and Ethical Implications of Knowledge About Human Genes

If a medical test could tell you whether (or not) you will die young from an incurable, devastating genetic disease, would you take it? What if the same test could also tell you whether (or not) you will pass the disease along to your children? If a medical test could tell you that, because of your genes, working in a chemical plant will give you cancer, would you want to know? Would you want your boss and insurance company to know?

Ethical dilemmas such as these are one product of recent advances in medical technology that now allow doctors and researchers to analyze human genes. Scientists estimate that some 4,000 diseases afflicting humankind are rooted in malfunctioning genes, which are made of the chemical DNA. Genes are also likely to play a role in many other diseases, such as heart disease, some cancers, and some neurologic diseases, where "predisposition" is a key factor.

The new science initiative known as the Human Genome Project seeks to locate all of the nearly 100,000 genes on the 24 different human chromosomes. Eventually, genome project scientists will decode the very language of heredity as they establish the precise order of the 3 billion chemical subunits of human DNA, known as nucleotides.

Understanding the molecular details of human heredity promises to give researchers astounding new opportunities to learn where on chromosomes genes are located, what they look like, and how they work in both health and disease. But as scientific information about human DNA unfolds, so will new opportunities to test people for an ever increasing number of inherited conditions, which may affect their lifespans, their ability to perform certain jobs, or how they handle alcohol or prescription drugs. In fact, tests to detect genetic diseases are likely to be developed far faster than are treatments or cures.

So how will the new availability of detailed and sensitive genetic information affect our personal and family privacy, our medical treatment and insurance coverage, our livelihoods, and even social attitudes toward us? The National Center for Human Genome Research, the organization charged with administering the National Institutes of Health's role in the Human Genome Project, has established a funding program aimed at examining

the ethical, legal, and social questions that may arise as genome project technology makes it increasingly possible for us, and others, to know the secrets locked away in our genes.

NCHGR Bioethics Program

Doctors have used biochemical and genetics methods to test for inherited diseases for decades. Although the ability to obtain health information from human DNA is not a direct outcome of the Human Genome Project, technology developed as part of the project will increase the amount and kind of information obtainable from DNA. These tests can determine if a person is a healthy carrier of a genetic disease or if he or she will become ill with a disease. DNA of carriers contains only one of the two defective gene copies needed to cause a disease. Although carriers are healthy, they can pass the gene on to their children, who may develop the disease if the other parent is also a carrier.

Human genetics research has made great strides over the past several decades in obtaining valuable information about the causes and treatments of genetic diseases. Unfortunately, however, these successes followed a dark period in the early part of this century when the so-called "eugenics" movement took hold in the United States and in some parts of Europe. Eugenics proponents sought to use genetic information to "improve" the human race by discouraging individuals with certain genetic qualities from having children. Because the misguided and ill-founded eugenics policies impinged on human rights and resulted in social stigmatization and discrimination, these past mistakes must be guarded against in the new quest for information about the human genome.

To help ensure that the fruits of the Human Genome Project are used for human good, the NCHGR bioethics research program will strive to anticipate and resolve conflicts between technological advances and personal freedoms. With approximately 3 percent of NCHGR's annual budget earmarked for this research, the center has become the largest federal source of research dollars for bioethics studies. Directed by philosopher Eric Juengst, Ph.D., the NCHGR program will provide financial support to researchers in bioethics, philosophy, law, economics, sociology, health policy, and other disciplines that bear on the impact of genetics research. Research funds will be dispersed in the form of research grants, training grants, contracts, and for supporting workshops, symposia, and commissioned papers.

NCHGR-Department of Energy Joint Bioethics Working Group

To help guide its bioethics research program, NCHGR, along with the Department of Energy's Office of Health and Environmental Research, has established the genome project's Joint Working Group on Ethical, Legal, and Social Issues (ELSI) related to mapping and sequencing the human genome. Composed of experts in law, ethics, psychology, genetics, clinical medicine, and other fields, the aim of the working group is to:

- o anticipate and address the implications for individuals and society of mapping and sequencing the human genome.
- o examine the ethical, legal, and social consequences of mapping and sequencing the human genome.
- o stimulate public discussion of these issues
- o and develop policy options to assure that genetic information is used for the benefit of individuals and society.

The ELSI working group will meet several times a year to help focus and refine research priorities of NCHGR and DOE bioethics programs. The group also plans to help develop public education and outreach programs designed to raise public understanding of the promises and pitfalls of medical genetics and new genetics technologies.

Expertise of the working group members spans the range of issues that may arise as genome project technology proceeds. The group is chaired by Nancy S. Wexler, Ph.D., president of the Hereditary Disease Foundation and clinical psychologist in the department of neurology and psychiatry at the Columbia University College of Physicians and Surgeons. A former member of the Huntington's Disease Commission, Dr. Wexler has worked for many years on Huntington's disease, focusing on genetic analysis of large families affected by the disease and more recently on responses to newly available HD tests. Jonathan R. Beckwith, Ph.D., is a bacterial geneticist at Harvard University medical school. Interested in genetic screening for over a decade, he has raised concerns over research proposals to mount behavioral studies of boys and men with the XYY chromosome makeup and has continued to participate in public discussions about behavioral genetics. Robert Cook-Deegan, M.D.'s, interest in genetics began with his research on Alzheimer's disease. While on an Office for Technology Assessment fellowship, he prepared two reports on human gene therapy and public policy related to the Human Genome Project. Patricia King, J.D., has worked in civil rights law and reproductive law and is interested in the impact of genetic studies on minority groups. She has been a member of two prominent federal bioethics commissions--the National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research, which operated from 1974-1978, and the President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research, which was active from 1980-1983. She also served on the NIH Recombinant DNA Advisory Committee, is a Fellow of the Hastings Center, and co-directs the program in health, law, and ethics at Georgetown University law school. Victor A. McKusick, M.D., has been involved in the study of human genetics for over 40 years. An internationally respected clinician and scientist, Dr. McKusick has compiled the largest compendium of data on human genetic diseases, entitled *Mendelian Inheritance in Man*, which is maintained at the Johns Hopkins University. Robert Murray, M.D., a clinician and researcher at the Howard University college of medicine, has been closely involved in genetic testing and screening programs and their social impacts for well over a decade. He has worked on sickle anemia and thalassemia testing programs and continues to offer genetic counseling to patients. Social psychologist Thomas Murray, Ph.D., is director of the Center for Biomedical Ethics at Case Western Reserve University. He has written extensively about the ethical impact of genetic testing and screening in the workplace. He was recently elected a fellow of the Hastings Center, where he worked for several years in the 1970s.

####

Prepared by
Leslie Fink
Office of Communications
NCHGR