



GENETICS AND ETHICS

Few enterprises have been as grand as the Human Genome Project (HGP). According to its leaders, the project is "an ambitious effort to understand hereditary instructions that make each of us unique."¹ It is intended "to find the location of the 30,000 or so human genes and to read the entire genetic script, all three billion bits of information, by the year 2005."

Researchers have announced that, although some chores remain, the task of identifying and sequencing the human genome is substantially complete. The ultimate goal of the HGP is to decode, letter by letter, the exact sequence of all three billion nucleotide base pairs that make up the human genome. Researchers hope this knowledge will lead to new technologies that can, first, identify defective genes, and, second, either neutralize their debilitating qualities or replace them entirely with "good genes." In the future, genetic science may even be able to wipe out some diseases altogether. The HGP's potential benefits seem endless.

In the past, physicians diagnosed an illness as the result of a clinical examination, confirming it through biomedical testing. Those procedures are becoming obsolete, according to the HGP's leaders.

On the horizon is a new era of molecular medicine characterized less by testing symptoms and more by looking to the



Fr. O'Rourke is senior lecturer, Stritch School of Medicine, Loyola University, Chicago.

*Is the
Ministry
Prepared to
Answer
Ethical
Questions
Raised by
The Human
Genome
Project?*

**BY REV. KEVIN D.
O'ROURKE, OP, JCD**

most fundamental causes of diseases. Rapid and more specific diagnostic tests will make possible earlier treatments of countless maladies. Medical researchers will be able to devise novel therapeutic regimens based on new classes of drugs, immunotherapy techniques, avoidance of environmental conditions that trigger disease, and even replacement of defective genes.²

Francis S. Collins, MD, the HGP's director, has long maintained that medicine would make amazing strides as a result of genetic research. Some of the developments Collins predicted are already in motion. They include the following:

- Each year clinical labs in this country perform millions of tests aimed at detecting potential or actual diseases caused by genetic defects.
- Newborn infants are screened for sickle cell anemia, a metabolic illness called phenylketonuria, and congenital thyroid diseases.
- Other tests reveal whether people predisposed by family history to develop cancer have in fact inherited dangerous genetic mutations.

Early detection of such genes allows people to begin therapy before symptoms occur. Genetic therapy for potential diseases discovered through genetic testing may be applied in one of three different ways: through splicing into human cells a healthy gene to displace the defective gene, by administering pharmaceuticals containing altered cells, or by stifling harmful genes by interfering with their protein production.

Still, we know that, although genetic makeup determines human physiology, it contributes only a *predisposition* to human behavior.³ Human personhood "is embodied intelligent freedom, with many levels of human activity most clearly manifest and definable by its maximum activity, the power to integrate these activities."⁴ Collins has borrowed



the words of another intrepid scientist, Copernicus, to suggest that the HGP itself may be part of God's plan: "To know the mighty works of God, to comprehend His wisdom and majesty and power, to appreciate the wonderful working of His laws, surely all this must be a pleasing and acceptable mode of worship to the Most High to whom ignorance cannot be more grateful than knowledge."⁵

GENETIC SCREENING AND STEM CELL THERAPY

Two dramatic applications of genetic knowledge have already made news.

Preimplantation Genetic Screening One news story concerned a six-year-old Colorado child who had been born with Fanconi anemia, a genetically caused disease that normally results in death at an early age.⁶ Her parents sought advice from medical scientists familiar with the illness. The scientists suggested that if the parents were to have a second child without the defective gene, that child's blood might be transfused into their daughter and save her life.

Because both parents had the Fanconi gene, their chances of producing a child who did not have the illness were only 1 in 4. But the scientists were able (before implantation) to examine the newly conceived embryo, select from it a totipotent cell (one capable of generating a complete organism) that did not have the defective gene, and implant that cell in the womb of the mother. (They also destroyed those totipotent cells that did carry the defective gene.)

When the second child was born, blood stem cells from his umbilical cord were transfused into his sister. It is not yet clear whether the procedure, which cost more than \$100,000, will save the girl's life. However, it *is* now clear that preimplantation genetic screening makes it possible to produce children without Fanconi anemia.

Stem Cell Therapy Stem cells are pluripotent, capable of developing into any of the 210 types of cell that make up the human body. Researchers are conducting experiments to determine whether stem cells can be manipulated to develop in vitro in the same way they do in a normally developing human embryo. If scientists can develop stem cells outside the body, they may be able to use them in stem cell therapy—replacing defective organs in the human body. "Culture of the cells in the laboratory could be nudged down different developmental pathways to become heart, bone marrow, or pancreatic cells," as one observer has noted.⁷

The stem cells needed for such research are available. Scientists originally harvested the cells from "spare embryos" developed through in vitro fertilization and from recently aborted human embryos, but today they can cultivate them in the laboratory.

The causes
of ill health
are social as
well as
genetic.

Stem cell therapy already seems to be an effective treatment for certain diseases, especially diseases of the autoimmune system. Researchers report that some patients with lupus erythematosus have improved significantly as a result of being transfused with their own blood stem cells.⁸ In this procedure, physicians collect some of the patient's blood stem cells and subject them to intense chemotherapy, after which they transfuse the cells back into the patient. The treated stem cells migrate to the patient's bone marrow, where they repopulate the deficient cells. Researchers hope someday to use patients' own blood stem cells to treat such neurological disorders as Parkinson's disease, Huntington's disease, and Alzheimer's disease; stroke; and problems resulting from spinal cord injuries.

How will health care providers deal with these new developments? Ethicists have discussed a number of issues arising from genetic mapping, especially those involving hiring practices and insurance coverage.⁹ However, three ethical issues sure to affect Catholic health care providers have *not* been much considered. They are:

- Genetic testing and counseling
- Stem cell research and church teaching
- Stem cell research and the desire to prolong life indefinitely

GENETIC TESTING AND COUNSELING

Any person planning to undergo genetic testing should first have counseling so that he or she can make good decisions based on its results. Counseling is vital because most people (including, unfortunately, members of the health care professions) are uninformed about the new genetic science and its developing technology. As a result, myth rather than fact dominates many discussions of genetics. Three facts in particular are often overlooked.

Disease Has Social Roots The causes of ill health are social as well as genetic. As one observer has put it:

Poor health runs in families, but usually this has little to do with genes. The best predictor of health and disease is class. Tuberculosis and lung cancer may soon be diseases of the poor, and even for cancer, the chances of survival are related to income. Wealth and poverty are inherited, and most people who are born poor stay that way. As important as this is to public health, nobody sees this as falling within the province of genetics.¹⁰

Testing Is Not a Crystal Ball Susceptibility to an illness, rather than certainty that one will develop it, is the best that can be expected from genetic



testing. Some scientists were saying a few years ago that research would soon detect the genes that “cause cancer” and find a way to replace them with more dependable substitutes. Today, however, scientists see the development of a disease, especially an inherited one, as a much more complex phenomenon.

Nobody doubts that cystic fibrosis is a single illness. However, most inherited diseases are not due to errors in a single or a few genes: instead they are symptoms of a great constellation of failures. Sometimes a single error is involved in certain cases, but not others: sometimes inherited changes whose individual effect are imperceptible may together produce a disease. . . . Because some conditions have a largely environmental origin in some patients and a mainly genetic one in others, to unravel the causes will not be easy. Even then, it is not clear how useful the information will be.¹¹

A Shortage of Genetic Counselors Exists For a person undergoing genetic testing to respond intelligently and maturely to test results, he or she will need counseling from a professional equipped to consider the ethical implications. Unfortunately, few such professionals are available. The United States currently has only 1,600 certified genetic counselors; no more than 120 new professionals join their ranks each year.¹² Many of these, moreover, are untutored in the Catholic tradition. It is common, for example, for genetic counselors to recommend abortion in cases where amniotic screening suggests that a fetus has a genetic defect. How many Catholic hospitals have qualified genetic counselors in their obstetric-gynecological departments? And even if counseling is provided, how “Catholic” is it?

Because we have so few counselors, the job of explaining the risks involved in genetic screening to patients often falls to physicians. But physicians are not prepared to be adept counselors. A recent study revealed that, although 70 percent of the physicians and other health care professionals surveyed have discussed genetics with their clients, only 10 percent feel confident in their ability to do so.¹³ In another study, nearly a third of the physicians who had received results from a test detecting serious mutations misinterpreted the findings.¹⁴ A concerted effort is under way to teach physicians how to talk empathetically to patients about the results of genetic research and technology.¹⁵ But no one should assume that a physician is necessarily competent to discuss genetic information or to conduct genetic counseling.

The
United States
currently has
only 1,600
certified
genetic
counselors.

STEM CELL RESEARCH AND CHURCH TEACHING

A second ethical issue, especially related to embryo stem cell research, concerns the manner in which such pluripotent cells are obtained.¹⁶ Most are currently derived from two sources:

- Embryos declared “extra” in the process of in vitro fertilization
- Newly aborted embryos¹⁷

In either case, obtaining embryo stem cells in this manner is contrary to the teaching of the church because it involves the destruction of human beings or close cooperation with those who destroy human beings (even if the persons destroyed were in but the initial stage of existence). When scientists first began using fetal tissue in research some years ago (before stem cell research became so prominent), they tried to do it with spontaneously aborted fetuses. But those efforts were not successful.¹⁸ Spontaneously aborted fetuses do not seem to be a viable source of supply for research and therapy involving fetal tissue.

Not all embryo stem cells currently available for research are directly obtained from the remains of human fetuses. Some, called “cultured” stem cells, are developed from those thus obtained and sold to researchers.¹⁹ In other cases, stem cells may be harvested from adults. The latter source is recommended by spokespersons for the Catholic Church because it does not involve the direct destruction of human beings.²⁰ Adult stem cells are effective in some therapeutic procedures, including those mentioned above for lupus erythematosus and other diseases of the autoimmune system. But the prospects for developing adult stem cells as a plentiful source of stem cell research and therapy is not encouraging. According to the National Institutes of Health, “There are some significant limitations to what we may or may not be able to accomplish with them (adult stem cells). First of all, stem cells have not been isolated for all tissues of the body. . . . Secondly, adult stem cells are often present only in minute quantities, are difficult to isolate and purify, and their numbers decrease with age.”²¹

Would it be possible to carry on research or therapy in Catholic health care facilities or university laboratories with stem cells cultured from stem cells originally derived from aborted fetuses? The *Ethical and Religious Directives for Catholic Health Care Services* says that “Catholic health care institutions should not make use of human tissue obtained by direct abortions even for research and therapeutic purposes.”²² This statement (Directive 66) was promulgated at a time when research with fetal cells concentrated on transplanting brain cells for the cure of Parkinson’s disease. Stem cell research had not yet become widespread. It seems, nevertheless,



that the prohibition would apply to any human tissue used for research or therapy if the cells in question were obtained *immediately* from aborted fetuses.

But would Directive 66 prohibit the use of fetal tissue not immediately harvested from direct abortions? Would it apply to embryo stem cells obtained by the culturing of fetal stem cells? Although those using such material would not themselves be involved in the immediate and direct misuse of human fetuses, they would be cooperating with the persons who originally harvested the stem cells. What kind of cooperation would this be? Would it be prohibited because it was too closely associated with the killing of innocent human beings? In other words, would this type of cooperation be formal (intending the evil) or material (not intending the evil but involved in the action in some way)? And if it was material, would it be immediate (involved in the action in an essential way) or mediate (involved in a nonessential way)?²³

A recent document from the Pontifical Academy of Life offers some insight into answering this question. The Pontifical Academy asked: "Is it morally licit to use ES (embryo stem) cells, and the differentiated cells obtained from them, which are supplied by other researchers or are commercially obtainable?"²⁴

The answer is negative because (prescinding from participation—formal or otherwise—in the morally illicit intention of the principal agent) the case in question would entail a proximate material cooperation in the production and manipulation of human embryos on the part of those producing or supplying them.²⁵

The Pontifical Academy's declaration, although not the official teaching of the church, was developed within the tradition of prior church teaching, and thus must be respected. However, two things may be noted concerning this statement. First, the response seems to grant that cooperation with the person originally obtaining the cells need not be formal cooperation (that is, one does not intend the evil of obtaining the cells from aborted fetuses). If it possible that material cooperation is involved, is it immediate or mediate? Immediate cooperation has a different proximate intention (*finis operis*) from that of the principal wrongdoer, but it is *integrally* involved in the sinful act. Mediate cooperation also has a different proximate intention from that of the principal wrongdoer, but is only *accidentally* involved in the sinful act. Distinguishing immediate cooperation from mediate cooperation is often difficult.²⁶ If the cooperation is judged to be immediate, then it could never be justified because it seems the orig-

Some genetic research and the technologies resulting from it may make it possible to extend the average life span.

inal act of harvesting the embryo stem cells from aborted fetuses is intrinsically evil.²⁷

The Pontifical Academy's statement maintains that any cooperation "in the production and manipulation of human embryos on the part of those supplying them is proximate material cooperation."²⁸ This statement is not entirely helpful. The academy uses the term "proximate cooperation," but this term is usually used to specify a type of mediate material cooperation. Our concern is not with those who originally salvage or produce embryo stem cells, but rather with those who avail themselves of the opportunity to use cultured stem cells for research or therapy. Could we envision the person using cells developed in culture being so far removed from the original procurement of the cells that he or she would be cooperating with the original harvesting of the cells—but in a manner that could be judged mediate rather than immediate?

Mediate material cooperation is legitimate if there is a sufficient reason for it. This reasoning is based on the principle of double effect. In this case, the good effect would be the research or therapy that could be accomplished through the use of the cells.²⁹ The bad effect, not intended but nevertheless foreseen, is accidental cooperation with the original process. The issue is not a new one. People have asked, for example, about the ethics of using information gathered by physicians working in Nazi concentration camps. Though much of this "science" was worthless, it did seem to include some useful information concerning human reaction to hypothermia. Could such knowledge be used licitly by researchers who had no sympathy for Nazis and no connection with their crimes? The question was hotly debated. The use of cultured embryo stem cells will likely be hotly debated as well.

STEM CELL RESEARCH AND THE DESIRE TO PROLONG LIFE

Some HGP research and the technologies resulting from it may make it possible to extend human life well beyond the average life span.³⁰ Indeed, this phenomenon has already begun. The notion that only a long life is a good life is rampant in our society. Yet a 16th-century theologian, one of the first to consider aging and our moral responsibility to prolong life, maintained that "God does not want us to worry about a long life" and that "directly terminating our life is one thing, but not striving to prolong it is another thing."³¹

According to Catholic teaching, human life is not an absolute good. Charity, or friendship with God, is the only absolute good, so far as human life is concerned. Catholic health care providers are encouraged to foster this mentality, especially



as they care for the dying.³² Death is not the enemy but the gateway to eternal life for those who love God.

But how will a mentality that concentrates on extending human life to 150 or 200 years for those who can afford it influence our desire to serve God and neighbor? Will our culture try to deny that death is a part of life? We are mortal; death is not a regrettable accident. The philosopher Leon Kass has said, "To argue that human life is better without death is to argue human life would be better without being human."³³

It seems entirely possible that health care providers will neglect our nation's more important health priorities: universal health care coverage and compassionate care of the dying. Spiritual values are not strongly present in our health care efforts. The poor and severely debilitated do not receive much attention. Moreover, the health care community could become so concerned about enhancing and extending the life of U.S. citizens that it neglects the more basic health care needs of the rest of the world.

People committed to the stewardship of scarce resources must be concerned about the products and programs emerging from HGP research. Attempts to extend life may well deplete the resources needed for basic health care. The ministry's leaders, our sponsors and bishops, have an obligation to keep our priorities in order so that the extension of human life does not become a primary goal of Catholic health care.

IS THE MINISTRY PREPARED?

In recent times, three significant events—the discovery of atomic fusion, the development of the computer, and the mapping of the human genome—have revolutionized the way we live. Each event has come replete with ethical issues. To date, the human community has not been successful in solving these issues.

The genetic revolution cannot help but have a historic impact on the provision of health care. Are Catholic providers ready for this responsibility? □

People
committed to
the stewardship
of scarce
resources must
be concerned
about the
products and
programs
emerging
from HGP
research.

NOTES

1. *The Human Genome Project*, National Center for Human Genome Research, National Institutes of Health, 1995.
2. *The Human Genome Project*.
3. See S. Pope, *The Evolution of Altruism and the Order of Love*, Georgetown University Press, Washington, DC, 1994; "Colloquy Explores Genetic Predisposition," *Human Genome News*, July-August, 1995.
4. B. Ashley and K. O'Rourke, *Health Care Ethics*, 4th ed., Georgetown University Press, Washington, DC, 1997, p. 7.
5. F. Collins, "Human Genetics: Where Do We Stand?" *Origins*, November, 1996, p. 468.
6. E. Goodman, "Adam Nash Was Designed to Save His Sister," *St. Louis Post-Dispatch*, October 12, 2000; L. Platt and D. Carlson, "Prenatal Diagnosis, When and How," *New England Journal of Medicine*, August 27, 1992, p. 327.
7. N. Wade, "Scientists Cultivate Cells at Root of Human Life," *New York Times*, November 6, 1998, p. 1.
8. R. Kotulak, "Lupus Therapy Stirs Hope for Patients," *Chicago Tribune*, August 25, 2000, p. 1.
9. See R. Hubbard and R. C. Lewontin, "Pitfalls of Genetic Testing," *New England Journal of Medicine*, May 2, 1996, p. 334; AMA Council Report, *JAMA*, 1991, p. 266; P. Reilly, et al., "Ethical Issues in Genetic Research: Disclosure and Informed Consent," *Nature Genetics*, January, 1997, pp. 16-20.
10. S. Jones, "Genetics in Medicine: Real Promises and Unreal Expectations," Milbank Memorial Fund, New York City, 2000, p. 4.
11. Jones, p. 8.
12. P. Boyle, "Pastoral Counseling Faces Genetics," *The Park Ridge Center Bulletin*, September-October, 2000, p. 13.
13. V. Lapham, "Genetics: Blind Spot in Medical Training," *Genetics in Medicine*, Summer 2000, p. 16.
14. J. Stephenson, "Group Drafts Core Curriculum for What Docs Need to Know about Genetics," *JAMA*, March 11, 1998, p. 735.
15. J. Stephenson.
16. "Stem Cells: A Primer," National Institutes of Health, December 1999, p. 2.
17. "Stem Cells: A Primer."
18. See K. O'Rourke, "Research on Human Embryos: Ethical Perspective," *Ethical Issues in Health Care*, Center for Health Care Ethics, Saint Louis University Medical Center, January 1989; D. Dickson, "Europe Split on Embryo Research," *Science*, November 1988, p. 1,117.
19. See "Human Primordial Stem Cells: A Symposium," *Hastings Center Report*, March-April 1999, p. 34.
20. See R. Doerflinger, "Destructive Stem Cells Research on Human Embryos," April 29, 1999, p. 770.
21. "Stem Cells: A Primer," p. 4; see also "Government Urged to Consider Stem Cell Banks," *The Royal Society of Great Britain*, March 8, 2000.
22. National Conference of Catholic Bishops, *Ethical and Religious Directives for Catholic Health Care Services*, U.S. Catholic Conference, Washington, DC, 1995, p. 24.
23. See B. Merklebach, *Summa Theologiae Moralis*, vol. 1, 8th ed., Desclee de Brouwer, Brussels, 1946, p. 487ff.
24. "Declaration on the Production and the Scientific and Therapeutic Use of Human Embryonic Stem Cells," Pontifical Academy of Life, Vatican City, August 25, 2000.
25. "Declaration on the Production."
26. D. Prummer, *Manuale Theologiae Moralis*, vol. 1, 13th ed., Herder, Rome, 1958, p. 447ff.
27. John Paul II, "Veritatis Splendor," n. 80.81, *Origins*, October 14, 1993.
28. "Declaration on the Production."
29. B. Merklebach.
30. S. Walman, "The Recycled Generation," *New York Times Magazine*, January 20, 2000, p. 30ff.
31. F. de Vittoria, *On Homicide*, J. Doyle, ed., Marquette University Press, 1997.
32. *Ethical and Religious Directives*, General Introduction, p. 5.
33. Quoted in N. Wade, "Arguments over Life and the Need for Death," *New York Times*, March 7, 2000.

JOURNAL OF THE CATHOLIC HEALTH ASSOCIATION OF THE UNITED STATES

www.chausa.org

HEALTH PROGRESS®

Reprinted from *Health Progress*, March-April 2001
Copyright © 2001 by The Catholic Health Association of the United States
