Planning for the Age of Genetics

A Denver-Based System Makes Ethical, Theological, and Practical Decisions about Genetic Medicine

Genomics, the study of genes that are the units of inheritance, is emerging as the leading science of the 21st century. The genomic revolution—and its impact on the field of medical genetics—has important implications and opportunities for the practice of medicine and the delivery of health care. Genomics will increase identification of heritable genetic characteristics and lead to the development of new technologies, such as genetically customized drugs, for the elimination, treatment, and management of a variety of diseases. Some observers predict that the impact of the genomic revolution will be greater than that of the Copernican revolution and Darwin's theory of evolution combined.

The Human Genome Project (HGP), one of the most significant scientific endeavors of all time, was a 13-year, international effort to read the information encoded in human chromosomes, discover all of the approximate 20,000 to 25,000 human genes and make them available for further study, and determine the complex sequence of the three billion DNA subunits (chemical bases). The sequencing of the human genome was officially completed in April 2003.

Catholic health care organizations should view these advances positively because they present radically new opportunities to relieve human suffering, to preserve and enhance human dignity, and to build up the human family. These developments are further evidence of the role and ability of humans to be co-creators with God in the dynamic unfolding of human life and potential in all creation.

At the same time, genomics takes us into uncharted medical, moral, and legal territory, raising serious ethical challenges and questions. Some in Catholic health care have taken a wait-and-see position with respect to medical genetics because of ethical challenges and unanswered questions associated with advances in the field, or in the belief that the ministry will not be affected by genomics until some time in the distant future. However, if the ministry is to use the new knowledge responsibly and for the good of society as a whole, it must begin now to participate in the unfolding practice of medical genetics and in the discernment of the ethical principles and practices that can guide its use.

Catholic Health Initiatives (CHI), Denver, has determined the approach it will take to medical genetics. At CHI, we have decided to focus on one facet of this field—genetic testing and counseling—and to promote reflection and dialogue on this topic among our leaders at the national and community levels.

The Urgency Behind Ethical Reflection

Genetic testing is a medical intervention with profound implications for a patient's personal and family life, and, in some cases, the life and health of the larger community. Increasingly, medical practitioners and society in general are beginning to view genetic testing and counseling as part of comprehensive, high-quality medical care. Genetic screening of newborns for specific genetic diseases is standard practice. Prenatal genetic testing of pre-embryos, embryos, and fetuses is increasing and becoming progressively more accurate in diagnosing the presence or absence of genetic disease. Although not yet widespread, predictive genetic tests and testing of presymptomatic adults are being offered by specialty clinics and sought by patients and clients. Such tests may determine parents' susceptibility (or that of their children) to specific genetic diseases, as well as their potential for passing serious genetic defects on to future generations.

By Carl Middleton, DMin; and Mary Elise Biegert

Dr. Middleton is vice president, theology and ethics, and Ms. Biegert is director, communications services, Catholic Health Initiatives. Dr. Middleton is based in Denver; Ms. Biegert in Erlanger, KY.
Growth in the field of medical genetics will proceed with or without the participation of Catholic health care providers. However, the spiritual and human values that have historically informed and grounded our Catholic ethical decisions and practices should be an integral part of the experience and ethical discernment of individuals, families, and communities who might benefit from genetic testing. These values and principles are also essential to the ongoing dialogue in the medical and scientific arenas regarding the ethical uses and limits of new genetic knowledge, future research goals, and advances in clinical practice.

In light of these demands, CHI in 2000 formed an Ethics Implementation Advisory Committee to do background study and research. The advisory committee considered it imperative that Catholic health care leaders educate themselves on genomics, reflect on what they learned, and engage in dialogue so that the Catholic ethical perspective would influence the practice of genetic testing in our organization. Although the committee might have chosen other areas of genomics, it decided to focus on genetic testing and counseling because those services have the most applicability for our facilities.

Since genetic testing may soon be required for health care facilities that provide obstetrics services, Catholic organizations must ensure that such testing and counseling are offered in environments that demonstrate respect for the dignity of the whole person—body, mind, and spirit. Just as we who serve the ministry provide support for people who make difficult decisions at the end of life, we must similarly demonstrate compassion and help people make deeply personal and difficult decisions about genetic testing and about the information they receive as the result of that testing. The advisory committee began by asking: What is genetic testing and what impact is it likely to have on CHI facilities?

**About Genetic Testing**

In May 1997, the National Task Force on Genetic Testing approved the definition of genetic testing as “the analysis of human DNA, RNA, chromosomes, proteins or other gene products to detect disease-related genotypes, mutations, phenotypes, or karyotypes for clinical purposes.”

Genetic testing involves examining a person’s DNA (which is taken from cells in a sample of blood or tissue) for some anomaly indicating a disease or disorder. The anomaly can be relatively large, such as a missing or added piece of chromosome—even an entire chromosome—that is visible under a microscope. Or it can be miniscule, as small as one extra, missing, or altered chemical base. Genes can be over-expressed (too many copies), inactivated, or lost altogether. Sometimes, pieces of chromosomes become switched, or transposed, so that a gene ends up in a location where it is permanently and inappropriately “turned off.”

Initially, genetic testing focused on the next generation. It provided information with which an adult could decide whether or not to have a child and enabled screening of newborns for phenylketonuria* and sickle cell anemia. But testing is focusing increasingly on the current generation—on, that is, whether a person is susceptible to disease. Genetic testing has enabled the medical community to identify many of the diseases for which a person may have a predisposition. Tests have proved helpful in the early detection, diagnosis, prognosis, and treatment of a number of adult-onset diseases and cancers.

Much of the current excitement in genetic testing centers on predictive gene testing—tests that identify people at risk of getting a disease, such as breast or cervical cancer, before any symptoms appear. Tests are now available in research programs for some two dozen such diseases, and as more disease genes are discovered, more genetic tests can be expected. Such dramatic advances will inspire hopes of developing additional approaches to the prevention and cure of disease through such advances as genetically customized drugs and other gene therapies.

**Redefining Health and Disease**

For decades, genetic testing was largely the province of select pediatricians and obstetricians who used genetic test results to diagnose existing disease and abnormalities. Today, this is rapidly changing. Medical practice seems to be moving from a diagnosis model of care, based on symptoms, to a prediction-of-susceptibility model.

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*Phenylketonuria is an inherited metabolic imbalance that can result in mental retardation, organ damage, and problems in pregnancy.
based on genetic information. In the past, medical science believed that external agents such as pathogens or carcinogens caused most diseases. The patient exhibited symptoms that assisted the practitioner to identify the cause of the disease process and prescribe the appropriate treatment. However, with the advance of genetic technologies, medicine is focusing more on genetic defects as the source of disease and disorder.

This shift in focus from external agents to internal gene disorders as a primary cause of disease changes the essential meaning of disease in at least three fundamental ways. Seen genetically, a person can:

- Hypothetically have a disease or disorder without exhibiting symptoms
- Be identified as having a disease or disorder even before being born
- Be a "carrier" of a disease or disorder, without experiencing symptoms oneself, and have the potential to transmit the disease or disorder to others

The definition of disease itself has expanded. Disease is no longer considered to be merely an episode that appears with symptoms that are treated and cured, or as a chronic condition that is managed. As a result of the discoveries in genetics, patients will be diagnosed as potentially having a symptom-less disease.

In the past, physicians and their symptomatic patients worked together to diagnose disease. Today, and in the future, physicians and their asymptomatic patients will attempt to determine how susceptible the patients are for developing various maladies. Because treatments for the vast majority of genetic diseases do not yet exist, in the near term curing the disease could become a secondary consideration.

Steven Jones, PhD, a British scientist, anticipates a huge popular demand for tests for carrier status. Such a groundswell should be resisted, he believes, when the costs of these tests are high and their efficacy is low. "If nothing can be done for a condition, what is the point in diagnosing it?" Jones asks.

To date, it has been easier for scientists to develop tests for susceptibility to disease than to devise effective interventions to prevent manifestations in people who are born affected. (Currently, approximately 1,000 clinical tests are available to screen for single genes that cause specific diseases.) Then, too, susceptibility to an illness—rather than certainty that one will develop it—is the best that can be expected from genetic testing. This complicates the work of genetic counseling practitioners. They must not only make clear to patients the difficult choices involved when faced with genetic abnormalities; they must also try to determine the level of likelihood that patients will actually manifest disease.

Two specialists in the field have examined four uncertainties regarding test results that practitioners will need to communicate to patients.  

**Uncertainty about Who Will Develop Disease** For most practitioners, training in genetics has involved a single gene model. This model was useful in understanding a few conditions, such as cystic fibrosis, hemoglobinopathies (blood disorders), and phenylketonuria. Today, this simple model breaks down for most common genetic diseases because a "positive" genetic test result does not definitively forecast the associated clinical condition. Therefore, there is uncertainty as to whether a person may actually develop the disease.

**Uncertainty about How Much the Risk Has Been Overstated** Statements such as "You have at most a 75 percent risk of developing colon cancer" need to be interpreted carefully. Quantifying the probability of disease is made more difficult by multiple genetic alterations and multiple modifiers. For example, at least four genes are associated with nonpolyposis colon cancer, and one of the genes associated with breast cancer (BRCA1) has more than 500 mutations. With such variability, there is concern regarding the possibility of overstating the potential risks.

**Uncertainty Regarding the Right Time to Learn the Information** A unique aspect of genetic testing for chronic disease is that genetic abnormalities can be detected long before they express themselves in visible properties. Because a genetic abnormality can often be detected decades before it is manifested as illness, test timing becomes an issue. What is the right time or right age to test?

**Uncertainty about the Benefits of Early Intervention** A physician can—in discussing the long lead times in the development of such chronic conditions as breast, thyroid, prostate, and colon cancers—send confusing signals to a patient. How, for example, should the patient respond if the physician says,
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"You have at most a 75 percent risk of developing colon cancer if you live to age 65, and we are not sure that actions based on this information will be helpful or improve your life". The truth is that the patient's potential for acquiring the disease will require continuous surveillance. The question remains: Will early intervention be effective?

Now that the HGP is completed, scientists anticipate that the estimated 20,000 to 25,000 genes will become possible sites for disease susceptibility identification. When this occurs, the function of diagnosis and the labeling of disease states will change significantly.

Using genetic test results, rather than symptoms, to predict or diagnose disease could radically alter how society interprets “normal” and “health.” The terms normality and abnormality may come to be defined not by health professionals, but, rather, by the biotechnology industry, as it discovers which conditions and disabilities can be identified and treated.

However, despite the uncertainties and ambiguities that remain with respect to genetic testing, there will be no putting the genie back in the bottle. The question before us was and is: What should be CHI’s response to the issues surrounding genetic testing and counseling?

GENETIC TESTING AND CHI

CHI’s facilities are becoming actively involved in helping to determine and direct our organization’s response to genetic testing and counseling. A number of opportunities for participation exist at the national and local community levels. CHI and its facilities are beginning to engage in dialogue about these issues with scientists, physicians, counselors, and patients in the communities we serve. We will share our expertise and work to assist key stakeholders in understanding the ethical issues, promises, and pitfalls involved.

One of the great challenges will be enabling people to make decisions about whether or not to engage in testing and helping them to respond to genetic information from a balanced perspective—one that includes realistic expectations, hope for medical breakthroughs, and attention to ethical demands.

Recent advances in the field of medical genetics have profound implications for health care and our healing ministry. Therefore, it is ethically appropriate for Catholic health care facilities with the necessary competence and expertise to provide genetic testing and counseling services within certain ethical parameters and cautions.

As for facilities that lack this expertise, we believe it will be important for them to provide continuing medical education on genetic testing for physicians, thereby preparing them for current and future medical realities. Genetic education should include, in addition to scientific and medical information, the theological and ethical principles that form a foundation and perspective for reflection and action concerning genetic testing issues.

A THEOLOGICAL PERSPECTIVE

The Catholic Church has a long-standing commitment to carry on the healing ministry of Jesus. The Ethical and Religious Directives for Catholic Health Care Services emphasize healing as a primary purpose of a Catholic health facility. “The Church has always sought to embody our Savior’s concern for the sick,” the directives’ authors note. “The gospel accounts of Jesus’ ministry draw special attention to his acts of healing. . . . In faithful imitation of Jesus Christ, the Church has served the sick, suffering, and dying in various ways throughout history.”

Catholic health care is called to touch people at their deepest level of experience and seeks to effect physical, mental, and spiritual healing. The church rightly views all involved in Catholic health care as ministers and agents of God’s healing and presence.

In regard to genetic testing, the church’s guidance and teachings have been limited because testing is a relatively new scientific technology. The U.S. bishops and Pope John Paul II have affirmed genetic testing as ethical when it is “an extension of sound medical practice.” As “an extension of sound medical practice,” genetic testing must aid critical medical diagnosis, enhance therapeutic intervention, and facilitate healing.

Church teachings acknowledge the growth of the genetic sciences and their potential for significant benefits for humankind. While these scientific developments hold promise, they also pose potential ethical problems. In the Christian theological tradition, Catholic health care leaders and practitioners will find principles for a more careful assessment of the rapidly growing practice of genetic testing and counseling. Three ethical principles, central to the Catholic theological tradition, form a framework for reflection and ethical analysis:

The Dignity of Each Human Person: A theological understanding of human dignity begins with a general recognition of the goodness of God’s creation. Humans, along with all living creatures, were called into being and are sustained by God’s goodness. Therefore, each and every human being is inherently valuable. Nothing is required to earn
that respect because God loves every person.

The Catholic theological tradition affirms that from the beginning, humans are "created in God's image and likeness." The Catholic theological tradition affirms that from the beginning, humans are "created in God's image and likeness." We arc created by God and vested with creativity to share in the transforming work of God's ongoing creation. Being cocreators with God requires us to be responsible, especially when it comes to the use of scientific technology such as genetic testing. Responsible use of genetic testing must affirm human dignity, support human life, and protect a person's right of privacy, confidentiality, and informed consent.

The Common Good The "common good" is a theological principle that serves as a compass that helps one navigate difficult moral and social issues. Human beings are essentially social beings called to live and work together in society. People, therefore, owe each other realistic efforts to attain the common good. The common good has been defined as "the complex of spiritual, social and material conditions needed in society for a human person to achieve integral human dignity." The common good is understood as the sum total of all the conditions of social life that enable all to flourish. According to this vision, it is each person's responsibility to work for his or her own welfare as well as that of others. The common good also requires that one make personal sacrifices for the good of the whole community. Genetic testing services, therefore, must benefit not only those who are able to pay for them; they must also contribute to enhancing the common good of all in society.

A Call to Justice All people are created unique, precious, and sacred, and are called to live in community. We are created "good." We are stewards of God's creation. We are created in community and therefore have responsibilities of relationship. This is the basic meaning of justice.

Contemporary secular definitions of justice focus on giving to each other his or her due. This vision of justice is seen as defining the boundaries among individuals and safeguarding their rights.

The biblical and theological concept of justice is concerned with providing the "glue" that holds society together, which is care for one's neighbor. Justice requires being faithful in relationships that are manifested in caring and demonstrating compassion to others, especially the disenfranchised, the vulnerable, and the poor. Because justice is an essential component of the Gospel of Jesus, as cited in the Beatitudes (Mt. 5:1-12), Catholic health care strives to create and sustain right relationships both within the ministry and with those served by the ministry.

Justice requires us to be faithful to our relational responsibilities to God, self, and all human beings as well as to all creation. Therefore, we should strive to make the benefits of genetic testing services available to those who are at greatest risk and have the greatest need.

These three principles—enhancing human dignity, focusing on the common good, and promoting justice—serve as a framework and provide a foundation for evaluating the ethics of genetic testing. When a Catholic health care facility considers specific genetic testing and counseling interventions, it should address four questions:

- Will the genetic testing procedure affirm our commitment to healing by aiding in medical diagnosis and furthering therapeutic intervention?
- Does the genetic testing or counseling intervention enhance the dignity and contribute to the development of the human person by safeguarding privacy and confidentiality and providing informed consent?
- Will the provision of genetic testing and counseling services enhance the common good of the local community or become another medical commodity supplied to patients?
- Will genetic testing and counseling be accessible and available to those who are most at risk and have the greatest need?

CHI has made these traditional theological principles foundational for its efforts to respond to the emerging field of medical genetics in general, and genetic testing and counseling in particular. We now examine these efforts in several phases.

CHI's Preparatory Phase

In 2000, CHI's advisory committee made three important recommendations. It urged the system to:

- Develop educational materials and resources that would assist CHI's facilities in providing genetic testing education programs for senior leadership, medical staffs, and ethics committees
- Develop ethical guidelines for the provision of genetic testing and counseling services when developing policies and procedures for the implementation of these services
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- Form a multidisciplinary task force to consider the medical, legal, ethical, and risk management issues surrounding the provision of genetic testing services (an existing ethics committee could serve this purpose).

In the spring of 2001, CHI developed a packet of materials, *The Healing Ministry in the Age of Genetics*, to foster reflection, education, and discussion regarding the issues of genetic testing and counseling. For this packet, Carl Middleton and Kevin Hanley, PhD, an ethics consultant and member of the advisory committee, prepared a background paper, much of which has formed the basis of this article. Susan Anthony, CHI’s director, mission integration and program development, developed additional materials for the packet. These materials included a paper, “Ethical Guidelines for the Provision of Genetic Testing and Counseling Services”; scientific primers that contain basic information regarding genetics; a facilitator’s guide; and a slide presentation. The ethics committees of Centura Health and Penrose-St. Francis Health Services, Colorado Springs, CO, and Good Samaritan Health Systems, Kearney, NE, piloted the materials and provided valuable feedback and suggestions.*

**THE GENETICS ADVISORY COMMITTEE**

In the summer of 2002, CHI formed a Genetics Advisory Committee. This is a multidisciplinary task force whose 23 members include a medical geneticist; a genetic counselor; mission leaders and ethicists; various clinicians, including physician specialists and representatives of nursing, oncology, laboratory, pharmacy, and clinical quality; and leaders from the areas of advocacy, human resources, finance, legal, risk management, strategy, and communications.

To date, the Genetics Advisory Committee has sponsored a CHI leadership education summit and developed a CHI genetic testing and counseling policy. The summit, held in San Antonio in January 2004, brought together 68 CHI leaders (of both the national organization and local facilities), in order to:

- Educate them about genetics
- Encourage them to become catalysts and agents for change throughout the system
- Elicit feedback from them regarding a future direction for genetic services
- Develop ownership and commitment among them vis-à-vis integrating genetic services within CHI

Kevin E. Lofton, CHI’s president and CEO, who has made innovation a high priority for the system, opened the summit. He encouraged participants to take a leadership role in this emerging new science. Other speakers were Jeffrey G. Shaw, program director and senior genetic counselor, Centura Health and Penrose-St. Francis Health Services; Judith Pratt Rossiter, MD, director, St. Joseph Perinatal Center, St. Joseph Medical Center, Towson, MD; Arthur A. Serpick, MD, an oncologist and vice president, medical affairs, St. Joseph Medical Center; Jesse Adams, MD, a cardiologist at the Jewish Heart and Lung Institute, Louisville, KY; Ron Hamel, PhD, senior director for ethics, CHA; and Middleton.

Following these presentations, the participants discussed possible “next steps” in genetic medicine for CHI. They recommended that the system:

- Develop a strategic focus around genetics, including assessing financial impact
- Develop system education and resources regarding genetic testing and counseling
- Assess current services
- Research the possibility of offering genetic testing and counseling as an employee benefit

The advisory committee met in April 2004 to develop an action plan, including a business-case analysis and recommendations for systemwide education, to submit to CHI’s national leaders.

**THE AGE OF GENETICS**

Catholic health care facilities have a unique opportunity to participate in dialogue and apply our spiritual values and ethical principles to genetic testing issues. It is through such ethical reflection and discourse that we will come to a new understanding of our healing ministry in the age of genetics.

**NOTES**


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Committee to address the perinatal care needs of all uninsured low-income women, including recent immigrants, in the greater Richmond area. Both BSR’s Community Health Services and the CVHPA are represented on this committee, as are numerous other providers and interested organizations (including many of the original steering committee members) that deal with perinatal needs. The committee is currently working with the Richmond OB/GYN Society’s physicians and other providers to establish a network of perinatal care services to provide comprehensive obstetrical care, including medical care, diagnostic testing, transportation services, and social supports.

This comprehensive health planning approach has been a catalyst in identifying and meeting immigrant health care needs. BSR’s leadership of and support for the initiative is widely recognized in greater Richmond as an important contribution to the improvement of the health of the communities the system serves. Moreover, the report’s data and findings have been extensively used by community organizations, health care providers, state government agencies, and others for both planning and resource development.

The complete report can be found at www.bonsecours.com/news/events/pdf/Richmond%20Health%20Planning%20Report.pdf and www.cvha.org/Richmond%20Immigrants%20Health%20Planning%20Report.pdf. For additional information, or for assistance in conducting an assessment in your own community, contact Eletta Hansen, Bon Secours Richmond Health System’s Office of Community Health Services (804-287-7343; ehansen@bsbhs.com), or Karen Cameron, Central Virginia Health Planning Agency (804-233-6206; kcameron@cvhpa.org).